

HGNC_Symbol	Variant1FullName	Variant2FullName	Inheritance	AutoCaSc	Zygosity	Origin	Number_Candidates_In_Family	DiseaseGroup_LeadingSymptom	HPO_Main_Terms
GLS	NM_001256310.1:c.695dupp.(Asp232Glufs*2)		AR_homo	12.4	homo	maternal&paternal	1	NDD + Epilepsy	Seizures, Status epilepticus, Infantile onset, Infantile spasms, Epileptic encephalopathy
DGKZ	NM_001199266.1:c.3227C>Gp.(Thr1076Arg)	NM_001199266.1:c.3326A>Gp.(Gln1109Arg)	AR_comphet	3.7	comphet	maternal&paternal	1	NDD + Epilepsy	Epileptic encephalopathy, Seizures, Failure to thrive, Hypoplasia of the corpus callosum, Hypsarrhythmia, Infantile onset, muscular hypotonia,
DUT	XM_005254212.1:c.218T>Cp.(Val73Ala)		AD_denovo	7.6	het	de novo	1	NDD + Epilepsy	Retrognathia, Myoclonus, EEG abnormality, Infantile encephalopathy, Epileptic encephalopathy
GLS	NM_001256310.1:c.815G>Ap.(Arg272Lys)	NM_001256310.1:c.241C>Tp.(Gln81*)	AR_comphet	10.7	comphet	maternal&paternal	1	NDD + Epilepsy	Microcephaly, Seizures, Status epilepticus, CNS demyelination, EEG with burst suppression, Peripheral demyelination, Epileptic encephalopathy
PLXNB3	NM_001163257.1:c.4343C>Ap.(Thr1448Asn)		AD_denovo	7.1	het	de novo	2	NDD	Hydrocephalus, Intellectual disability, hypotonia, Global developmental delay, Atria septal defect, Patent ductus arteriosus,Transposition of the great arteries with ventricular septal defect
GBP5	NM_001134486.2:c.154T>Cp.(Ser52Pro)	NM_001134486.2:c.502_505dupp.(Ser169*)	AR_comphet	5.0	comphet	maternal&paternal	2	NDD	Hydrocephalus, Intellectual disability, hypotonia, Global developmental delay, Atria septal defect, Patent ductus arteriosus,Transposition of the great arteries with ventricular septal defect
GRIN3B	NM_138690.1:c.1811C>Tp.(Thr604Met)	NM_138690.1:c.2114A>Cp.(Tyr705Ser)	AR_comphet	6.1	comphet	maternal&paternal	1	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Hypsarrhythmia, Infantile onset,
CLSTN1	NM_001009566.1:c.1844C>Tp.(Thr615Met)		AR_homo	8.1	homo	maternal&paternal	2	NDD	Cataract, Peters anomaly, Autism, Global developmental delay
CASP9	NM_001229.4:c.868+5G>Cp.?		AR_homo	8.8	homo	maternal&paternal	2	NDD	Cataract, Peters anomaly, Autism, Global developmental delay
PUM2	NM_015317.1:c.1595G>Ap.(Ser532Asn)		AD_denovo	7.6	het	de novo	1	NDD + Epilepsy	Seizures, Global developmental delay, Hypsarrhythmia,
CARMIL1	XM_005249221.1:c.3617C>Tp.(Ser1206Leu)	XM_005249221.1:c.2659G>Ap.(Glu887Lys)	AR_comphet	3.5	comphet	maternal&paternal	1	NDD + Epilepsy	Microcephaly, Delayed puberty, Abnormality of skin pigmentation, Seizures, Agenesis of corpus callosum, Growth delay, Intellectual disability, Limb hypertonía, Scoliosis, Chorioretinal lacunae, Muscular hypotonia of the trunk, Infantile axial hypotonia, Infantile spasms, Small hand
SMCR8	NM_144775.2:c.2404C>Tp.(Arg802Cys)		AD_denovo	4.7	het	de novo	2	NDD	Microcephaly, Epicanthus, Intellectual disability, Global developmental delay, Plagiocephaly, Abnormal facial shape, Wide nasal base
FRMPD3	XM_042978.8:c.3538C>Tp.(Arg1180Trp)		XL	3.3	hemi	maternal	2	NDD	Microcephaly, Epicanthus, Intellectual disability, Global developmental delay, Plagiocephaly, Abnormal facial shape, Wide nasal base
PUM1	NM_001020658.1:c.3439C>Tp.(Arg1147Trp)		AD_denovo	9.5	het	de novo	2	NDD	Global developmental delay, Microcephaly, Cryptorchidism, Ptosis, Short stature, Short phalanx of finger, Frontal hirsutism, Arachnoid cyst
BAIAP3	NM_001199096.1:c.892G>Tp.(Gly298Trp)		AD_denovo	5.9	het	de novo	2	NDD	Global developmental delay, Microcephaly, Cryptorchidism, Ptosis, Short stature, Short phalanx of finger, Frontal hirsutism, Arachnoid cyst
PSMB3	NM_002795.2:c.424T>Cp.(Cys142Arg)		AD_denovo	4.7	het	de novo	1	NDD	Trismus, Arthrogryposis multiplex congenita, Vesicoureteral reflux, Abnormality of the kidney, abnormal facial shape, Global developmental delay
VPS4A	NM_013245.2:c.291T>Gp.(Ser97Arg)		AD_denovo	7.3	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Epileptic encephalopathy
TAB3	NM_152787.3:c.1952A>Gp.(Gln651Arg)		XL	3.9	hemi	maternal	2	NDD + Epilepsy	Intellectual disability, Seizures, Epileptic encephalopathy
PPP1R37	NM_019121.1:c.509C>Tp.(Ser170Phe)		AD_denovo	6.0	het	de novo	2	NDD	Bilateral cryptorchidism, Short stature, Epileptic encephalopathy, Microcephaly
AQP6	NM_001652.3:c.146C>Tp.(Pro49Leu)		AD_denovo	5.2	het	de novo	2	NDD	Bilateral cryptorchidism, Short stature, Epileptic encephalopathy, Microcephaly
IRAK1	NM_001025242.1:c.609T>Gp.(Cys203Trp)		AD_denovo	6.1	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Generalized myoclonic seizures, Infantile onset
MED22	NM_133640.4:c.397_399delp.(Glu133del)		AR_homo	5.6	homo	maternal&paternal	1	NDD + Epilepsy	Seizures, Global developmental delay, Microcephaly, Hearing impairment, Visual impairment, Intellectual disability,
GTPBP2	NM_019096.3:c.1191C>Ap.(Asn397Lys)		AD_denovo	7.1	het	de novo	1	NDD	Tall stature, Macrocephaly, Retrognathia, High forehead, Low-set ears, Global developmental delay

NCOA2	NM_006540.2:c.1454T>Cp.(Met485Thr)	NM_006540.2:c.3509T>Cp.(Met1170Thr)	AR_comphet	6.1	comphet	maternal&paternal	1	NDD	Intellectual disability, Seizures, Encephalopathy, Cerebral atrophy, Intellectual disability, profound, EEG abnormality, Intellectual disability, severe, Cognitive impairment
SPEN	NM_015001.2:c.8092A>Gp.(Asn2698Asp)		AD_denovo	8.0	het	de novo	2	NDD	Microcephaly, Underdeveloped nasal alae, Strabismus, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Postnatal microcephaly
BOK	NM_032515.4:c.356C>Tp.(Thr119Met)		AD_denovo	5.6	het	de novo	2	NDD	Microcephaly, Underdeveloped nasal alae, Strabismus, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Postnatal microcephaly
ENOX2	NM_006375.2:c.148A>Gp.(Met50Val)		XL	3.3	hemi	maternal	2	NDD	Microcephaly, Underdeveloped nasal alae, Strabismus, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Postnatal microcephaly
CHD5	NM_015557.2:c.5003-5G>Ap.?	NM_015557.2:c.5249C>Tp.(Thr1750Met)	AR_comphet	5.3	comphet	maternal&paternal	3	NDD	Autism, Intellectual disability, Global developmental delay
HDAC4	NM_006037.3:c.1663G>Ap.(Gly555Ser)		AD_inherited	6.7	het	maternal	3	NDD	Autism, Intellectual disability, Global developmental delay
SLC10A3	NM_001142391.1:c.1160C>Tp.(Thr387Met)		XL	5.0	hemi	maternal	3	NDD	Autism, Intellectual disability, Global developmental delay
WBP1	NM_012477.3:c.25G>Ap.(Gly9Ser)		AD_denovo	3.8	het	de novo	1	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Atonic seizures
DUX4L4	NM_001177376.2:c.880C>Tp.(Gln294*)		AD_denovo	7.0	het	de novo	1	NDD	Microcephaly, Intellectual disability, Global developmental delay, Short stature
TEX44	NM_152614.2:c.1146C>Gp.(His382Gln)		AD_denovo	4.9	het	de novo	1	NDD	mild global developmental delay, delayed speech and language development
ASIC1	NM_001095.3:c.363-2A>Gp.?		AD_denovo	10.2	het	de novo	3	NDD + Epilepsy	Seizures, Abnormal social behavior, Epileptic encephalopathy
FAM168B	NM_001009993.2:c.452G>Ap.(Gly151Glu)		AD_denovo	6.5	het	de novo	3	NDD + Epilepsy	Seizures, Abnormal social behavior, Epileptic encephalopathy
COL20A1	NM_020882.2:c.3402+5C>Tp.?	NM_020882.2:c.1662C>Tp.(=)	AR_comphet	2.7	comphet	maternal&paternal	3	NDD + Epilepsy	Seizures, Abnormal social behavior, Epileptic encephalopathy
SPEN	NM_015001.2:c.3968T>Gp.(Met1323Arg)		AD_denovo	8.1	het	de novo	3	NDD + Epilepsy	mild global developmental delay, seizures, heterotopia, oral cleft, tall stature, obesity
CSMD1	NM_033225.5:c.7327A>Gp.(Ile2443Val)	NM_033225.5:c.8444A>Cp.(Glu2815Ala)	AR_comphet	5.3	comphet	maternal&paternal	3	NDD + Epilepsy	mild global developmental delay, seizures, heterotopia, oral cleft, tall stature, obesity
CENPV	NM_181716.2:c.75_92delp.(Ala26_Ala31del)		AD_denovo	5.4	het	de novo	3	NDD + Epilepsy	mild global developmental delay, seizures, heterotopia, oral cleft, tall stature, obesity
CACNB4	NM_000726.3:c.848C>Tp.(Ser283Leu)		AD_denovo	9.5	het	de novo	5	NDD + Epilepsy	Seizures, Global developmental delay, Generalized clonic seizures
KLHL17	NM_198317.2:c.1568C>Tp.(Ala523Val)		AR_homo	5.2	homo	maternal&paternal	5	NDD + Epilepsy	Seizures, Global developmental delay, Generalized clonic seizures
POLR2A	NM_000937.4:c.4808G>Ap.(Arg1603His)	NM_000937.4:c.778G>Ap.(Val260Met)	AR_comphet	6.1	comphet	maternal&paternal	5	NDD + Epilepsy	Seizures, Global developmental delay, Generalized clonic seizures
PNMA3	NM_013364.4:c.82G>Ap.(Glu28Lys)		XL	2.9	hemi	maternal	5	NDD + Epilepsy	Seizures, Global developmental delay, Generalized clonic seizures
ZNF12	NM_006956.2:c.670T>Cp.(Ser224Pro)	NM_006956.2:c.1438G>Ap.(Val480Ile)	AR_comphet	3.1	comphet	maternal&paternal	5	NDD + Epilepsy	Seizures, Global developmental delay, Generalized clonic seizures
CASKIN1	NM_020764.3:c.4103G>Ap.(Ser1368Asn)		AR_homo	7.8	homo	maternal&paternal	3	NDD	global developmental delay, absent speech, gait disturbance, EEG abnormality, decreased body weight
CELSR2	NM_001408.2:c.4706C>Tp.(Pro1569Leu)	NM_001408.2:c.8629G>Ap.(Gly2877Ser)	AR_comphet	7.6	comphet	maternal&paternal	3	NDD	global developmental delay, absent speech, gait disturbance, EEG abnormality, decreased body weight

FAT3	NM_001008781.2:c.3669+7G>Ap.?	NM_001008781.2:c.12922G>Cp.(Asp4308His)	AR_comphet	4.5	comphet	maternal&paternal	3	NDD + Epilepsy	Autism, Seizures, Global developmental delay, Motor delay, Absent speech, Epileptic encephalopathy
MADD	NM_001135943.1:c.1037T>Cp.(Leu346Pro)		AR_homo	9.6	homo	maternal&paternal	3	NDD	global developmental delay, absent speech, gait disturbance, EEG abnormality, decreased body weight
CHMP7	NM_152272.3:c.214C>Ap.(Leu72Met)		AD_denovo	6.0	het	de novo	3	NDD + Epilepsy	Autism, Seizures, Global developmental delay, Motor delay, Absent speech, Epileptic encephalopathy
ANKFY1	NM_001257999.1:c.1966G>Ap.(Ala656Thr)		AR_homo	5.5	homo	maternal&paternal	3	NDD + Epilepsy	Autism, Seizures, Global developmental delay, Motor delay, Absent speech, Epileptic encephalopathy
LUC7L	NM_018032.3:c.614G>Ap.(Arg205His)		AD_denovo	5.9	het	de novo	2	NDD + Epilepsy	Seizures, Global developmental delay
PRDX4	NM_006406.1:c.724G>Ap.(Gly242Arg)		XL	5.5	hemi	maternal	2	NDD + Epilepsy	Seizures, Global developmental delay
DIS3	NM_001128226.1:c.1486A>Gp.(Arg496Gly)	NM_001128226.1:c.2785T>Cp.(*929Glnext*14)	AR_comphet	7.1	comphet	maternal&paternal	2	NDD	Microcephaly, Intellectual disability, Global developmental delay, Abnormality of body weight, Increased body weight,
CAMTA2	NM_001171166.1:c.2639A>Gp.(Asp880Gly)		AR_homo	4.6	homo	maternal&paternal	2	NDD + Epilepsy	Seizures, Status epilepticus, Hypsarrhythmia,
FAT1	NM_005245.3:c.2137A>Gp.(Ile713Val)	NM_005245.3:c.9440T>Gp.(Val3147Gly)	AR_comphet	5.6	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Status epilepticus, Hypsarrhythmia,
STAM	NM_003473.3:c.119G>Cp.(Arg40Pro)		AD_denovo	8.1	het	de novo	1	NDD	Short stature, Ataxia, Cataract, Microphthalmia, Microcephaly, Nystagmus, Global developmental delay
GAL3ST3	NM_033036.2:c.39G>Cp.(Lys13Asn)		AD_denovo	5.0	het	de novo	3	NDD + Epilepsy	seizures, focal seizures
SDK1	NM_152744.3:c.1295G>Cp.(Gly432Ala)	NM_152744.3:c.3802C>Tp.(Arg1268Trp)	AR_comphet	4.7	comphet	maternal&paternal	3	NDD + Epilepsy	seizures, focal seizures
ZNF503	NM_032772.4:c.69_71dup, p.(Gly27dup)	NM_032772.4:c.1105G>Tp.(Gly369Cys)	AR_comphet	4.1	comphet	maternal&paternal	3	NDD + Epilepsy	seizures, focal seizures
TOB1	NM_001243877.1:c.888_907delTAACCTCAGTCTCTCCAGTinsGGGp.(Leu296Leufs*4)		AD_denovo	9.9	het	de novo	1	NDD	Cerebral calcification, Seizures, Congenital cataract, Autistic behavior, Obesity, Global developmental delay
GPKOW	NM_015698.4:c.1334G>Ap.(Arg445Gln)		XL	3.4	hemi	maternal	1	NDD	Autism, Global developmental delay
MACF1	NM_012090.5:c.1531C>Tp.(Arg511Cys)	NM_012090.5:c.3465G>Ap.(=)	AR_comphet	6.3	comphet	maternal&paternal	1	NDD + Epilepsy	global developmental delay, seizures,
TAAR2	NM_001033080.1:c.113G>Tp.(Arg38Ile)		AD_denovo	4.4	het	de novo	3	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Global developmental delay, Abnormality of the caudate nucleus, Infantile onset, Attention deficit hyperactivity disorder
MORF4L2	NM_001142418.1:c.287A>Gp.(Gln96Arg)		XL	4.8	hemi	maternal	3	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Global developmental delay, Abnormality of the caudate nucleus, Infantile onset, Attention deficit hyperactivity disorder
SLC35B3	NM_001142540.1:c.1135C>Tp.(Pro379Ser)	NM_001142540.1:c.1069G>Cp.(Gly357Arg)	AR_comphet	3.5	comphet	maternal&paternal	3	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Global developmental delay, Abnormality of the caudate nucleus, Infantile onset, Attention deficit hyperactivity disorder
URB2	NM_014777.2:c.1949delp.(Gly650Valfs*2)		AD_denovo	5.8	het	de novo	2	NDD + Epilepsy	Seizures, Myoclonic absences, developmental delay

OGDHL	NM_001143996.1:c.489G>Cp.(Trp163Cys)	NM_001143996.1:c.1315C>Tp.(Arg439Cys)	AR_comphet	4.7	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Myoclonic absences, developmental delay
SNX27	NM_030918.5:c.913G>Ap.(Ala305Thr)	NM_030918.5:c.69_71dup, p.(Gly25dup)	AR_comphet	6.7	comphet	maternal&paternal	1	NDD	Microcephaly, Hirsutism, Intellectual disability, Global developmental delay, Short stature
DOC2B	NM_003585.4:c.898G>Ap.(Gly300Ser)		AD_denovo	6.9	het	de novo	3	NDD + Epilepsy	global developmental delay, encephalopathy, seizures, infantile onset
ANKFN1	NM_153228.2:c.1052A>Gp.(Asn351Ser)		AD_denovo	6.4	het	de novo	3	NDD + Epilepsy	global developmental delay, encephalopathy, seizures, infantile onset
POU4F2	NM_004575.2:c.417C>Ap.(Asp139Glu)	NM_004575.2:c.180_200delp.(Gly62_Gly68del)	AR_comphet	3.8	comphet	maternal&paternal	3	NDD + Epilepsy	global developmental delay, encephalopathy, seizures, infantile onset
C11ORF95	NM_001144936.1:c.1592T>Cp.(Val531Ala)		AR_homo	4.9	homo	maternal&paternal	2	NDD + Epilepsy	global developmental delay, seizures, hypoplasia of the corpus callosum
SCUBE2	NM_001170690.1:c.68C>Tp.(Pro23Leu)		AD_denovo	4.9	het	de novo	2	NDD + Epilepsy	global developmental delay, seizures, hypoplasia of the corpus callosum
NINL	NM_025176.4:c.277+2T>Cp.?		AR_homo	9.4	homo	maternal&paternal	4	NDD	Intellectual disability, Global developmental delay
CTSB	NM_001908.3:c.444C>Tp.(=)		AR_homo	5.7	homo	maternal&paternal	4	NDD	Intellectual disability, Global developmental delay
CNOT1	NM_001265612.1:c.6727A>Gp.(Met2243Val)		AR_homo	7.8	homo	maternal&paternal	2	NDD	Intellectual disability, Global developmental delay
B4GALNT3	NM_173593.3:c.1798G>Ap.(Glu600Lys)	NM_173593.3:c.1640C>Tp.(Pro547Leu)	AR_comphet	3.5	comphet	maternal&paternal	4	NDD	Intellectual disability, Global developmental delay
SRPX	NM_001170750.1:c.1270A>Tp.(Thr424Ser)		XL	3.9	hemi	maternal	4	NDD	Intellectual disability, Global developmental delay
NPTX1	NM_002522.3:c.970G>Ap.(Gly324Arg)		AD_denovo	6.7	het	de novo	2	NDD	Spastic tetraparesis, Optic atrophy, Periventricular leukomalacia, Microcephaly, Global developmental delay
H2BC4	NM_003526.2:c.154G>Tp.(Asp52Tyr)		AD_denovo	5.4	het	de novo	2	NDD	Spastic tetraparesis, Optic atrophy, Periventricular leukomalacia, Microcephaly, Global developmental delay
FRY	NM_023037.2:c.4688G>Cp.(Ser1563Thr)		AD_denovo	7.5	het	de novo	1	NDD	global developmental delay, intellectual disability, epileptic seizures, microcephaly, Dandy-Walker malformation, Polymicrogyria, syndactyly, partial duplication of thumb phalanx
MICAL1	NM_001159291.1:c.571+1G>Tp.?	NM_001159291.1:c.2724-8C>Tp.?	AR_comphet	3.8	comphet	maternal&paternal	3	NDD + Epilepsy	Specific learning disability, Absence seizures, Cortical dysplasia, EEG with continuous slow activity, Seizures
SPATA31A3	NM_001083124.1:c.3206C>Tp.(Ser1069Phe)		AD_denovo	3.6	het	de novo	3	NDD + Epilepsy	Specific learning disability, Absence seizures, Cortical dysplasia, EEG with continuous slow activity, Seizures
ATP2B4	NM_001001396.2:c.2819A>Gp.(Lys940Arg)		AR_homo	5.3	homo	maternal&paternal	3	NDD + Epilepsy	Specific learning disability, Absence seizures, Cortical dysplasia, EEG with continuous slow activity, Seizures
EGR3	NM_001199880.1:c.477C>Ap.(Tyr159*)		AD_denovo	10.1	het	de novo	1	NDD	Intellectual disability, learning disability
FREM3	NM_001168235.1:c.728delp.(Glu243Glyfs*25)	NM_001168235.1:c.5401C>Tp.(Leu1801Phe)	AR_comphet	5.3	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Encephalopathy, Focal seizures, Encephalitis
PLXNA1	NM_032242.3:c.2690G>Ap.(Arg897His)	NM_032242.3:c.1045G>Cp.(Val349Leu)	AR_comphet	4.3	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Encephalopathy, Focal seizures, Encephalitis

DPP9	ENST00000262960:c.842G>C p.Arg281Pro		AD_denovo	A	het	de novo	1	other	(+) Splenomegaly,(+) Pancytopenia,(+) Congenital thrombocytopenia,(+) Immunodeficiency,(+) Bone marrow hypocellularity,(+) Hemophagocytosis,(+) Lymphocytosis
SPTBN5	NM_016642.3:c.5680G>Tp.(Glu1894*)		AR_homo	8.2	homo	maternal&paternal	2	NDD	intellectual disability
HOOK2	NM_001100176.1:c.1718-6C>Tp.?		AR_homo	4.4	homo	maternal&paternal	2	NDD	intellectual disability
ZKSCAN3	NM_001242894.1:c.253A>Tp.(Ile85Phe)		AD_denovo	5.0	het	de novo	3	NDD	Hypothyroidism, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Short stature, Abnormal social behavior
DAGLA	ENST00000257215:c.2613dup p.Ser872GlnfsTer6		AD_denovo	A	het	de novo	1	Neuro	abnormality of eye movement, ataxia
KALRN	NM_001024660.3:c.5980C>Gp.(Leu1994Val)	NM_001024660.3:c.2171C>Tp.(Ser724Leu)	AR_comphet	6.9	comphet	maternal&paternal	3	NDD	Hypothyroidism, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Short stature, Abnormal social behavior
SFXN3	NM_030971.3:c.785G>Ap.(Arg262His)	NM_030971.3:c.640delp.(Ala214Glnfs*9)	AR_comphet	4.9	comphet	maternal&paternal	3	NDD	Hypothyroidism, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Short stature, Abnormal social behavior
AP1G1	XM_005255821.1:c.468G>Ap.(=)		AD_denovo	6.9	het	de novo	1	NDD + Epilepsy	Seizures, Epileptic encephalopathy
NLRX1	NM_024618.2:c.428C>Tp.(Pro143Leu)		AD_denovo	4.7	het	de novo	1	NDD	Ptosis, Muscular hypotonia, Global developmental delay, Abnormal facial shape, Short stature, Feeding difficulties, Thick hair
L3MBTL1	NM_015478.6:c.478T>Ap.(Ser160Thr)		AR_homo	7.2	homo	maternal&paternal	3	NDD	Agitation, Aggressive behavior, Delayed speech and language development, Intellectual disability
MIA3	NM_198551.2:c.3981+3A>Gp.?		AR_homo	7.0	homo	maternal&paternal	3	NDD	Agitation, Aggressive behavior, Delayed speech and language development, Intellectual disability
GCN1	NM_006836.1:c.7082G>Ap.(Arg2361Gln)		AD_denovo	7.7	het	de novo	3	Neuro	Seizures, Hypoglycemia, Myopathy, Focal seizures, Ichthyosis, EEG with focal epileptiform discharges
GRIA1	NM_000827.3:c.81C>Ap.(=)		AR_homo	8.2	homo	maternal&paternal	3	Neuro	Seizures, Hypoglycemia, Myopathy, Focal seizures, Ichthyosis, EEG with focal epileptiform discharges
DEPTOR	NM_022783.2:c.496A>Gp.(Met166Val)	NM_022783.2:c.426-5C>Tp.?	AR_comphet	4.0	comphet	maternal&paternal	3	NDD + Epilepsy	Seizures, Hypoglycemia, Myopathy, Focal seizures, Ichthyosis, EEG with focal epileptiform discharges
SPTAN1	NM_001130438.2:c.2612delp.(Lys871Serfs*5)		AD_denovo	13.4	het	de novo	4	NDD + Epilepsy	Intellectual disability,Global developmental delay, Motor delay, Developmental regression
FHDC1	NM_033393.2:c.568C>Tp.(Arg190Trp)		AD_denovo	4.8	het	de novo	1	NDD	Hypertension, Intellectual disability,mild, Obesity, Abnormality of the pulmonary valve, I Hyperlipidemia, Childhood-onset truncal obesity
RASGRP1	NM_001128602.1:c.1487C>Gp.(Ser496*)		AD_denovo	9.8	het	de novo	3	NDD + Epilepsy	global developmental delay, encephalopathy, seizures
CNTNAP4	NM_033401.3:c.3353G>Cp.(Gly1118Ala)		AD_denovo	8.3	het	de novo	3	NDD + Epilepsy	global developmental delay, encephalopathy, seizures
ZNF708	NM_021269.2:c.443T>Ap.(Val148Asp)	NM_021269.2:c.1013G>Ap.(Cys338Tyr)	AR_comphet	2.3	comphet	maternal&paternal	3	NDD + Epilepsy	global developmental delay, encephalopathy, seizures
MCM7	NM_001278595.1:c.1147A>Cp.(Met383Leu)		AD_denovo	7.7	het	de novo	1	NDD + Epilepsy	Intellectual disability, Seizures, IGlobal developmental delay, Infantile onset, epileptic encephalopathy
DRG1	NM_004147.3:c.43-1G>Tp.?		AD_denovo	5.9	het	de novo	3	NDD + Epilepsy	Autism, Intellectual disability, Seizures, Global developmental delay, Poor speech, Focal seizures
ANK2	NM_001148.4:c.1288-1G>Ap.?		AD_denovo	12.4	het	de novo	3	NDD + Epilepsy	benign epilepsy
KMT2E	NM_018682.3:c.3554C>Gp.(Ser1185*)		AD_denovo	12.4	het	de novo	1	NDD	Intellectual disability, Seizures, EEG with spike-wave complexes, EEG with continuous slow activity,
DGKZ	NM_001199266.1:c.132_134delp.(Ser45del)	NM_001199266.1:c.16G>Cp.(Gly6Arg)	AR_comphet	4.4	comphet	maternal&paternal	3	NDD + Epilepsy	Autism, Intellectual disability, Seizures, Global developmental delay, Poor speech, Focal seizures
ARHGEF7	NM_001113511.2:c.17A>Cp.(Gln6Pro)		AD_denovo	7.9	het	de novo	3	NDD	global developmental delay, intellectual disability

CUX1	NM_001202543.1:c.3783_3784dup, p.(Leu1262Argfs*10)		AD_denovo	12.1	het	de novo	1	NDD	Macrocephaly, Umbilical hernia, Chronic constipation, Inguinal hernia, Delayed speech and language development, mild global developmental delay
SEMA3B	NM_001005914.2:c.952C>Tp.(His318Tyr)	NM_001005914.2:c.728T>Cp.(Phe243Ser)	AR_comphet	3.6	comphet	maternal& paternal	3	NDD	global developmental delay, intellectual disability
ETV5	NM_004454.2:c.232+1G>Ap.?		AD_denovo	10.0	het	de novo	4	NDD	global developmental delay, intellectual disability, generalized hypotonia,
DGKK	NM_001013742.3:c.689T>Gp.(Phe230Cys)		XL	2.0	hemi	maternal	4	NDD	global developmental delay, intellectual disability, generalized hypotonia,
ANK2	ENST00000357077.4:c.10768G>T p.Glu3590Ter		AD_denovo	11.9	het	de novo	1	Epilepsy	Focal myoclonic seizure
MDN1	NM_014611.2:c.2965-3T>Cp.?	NM_014611.2:c.9524A>Cp.(His3175Pro)	AR_comphet	4.4	comphet	maternal& paternal	4	NDD	global developmental delay, intellectual disability, generalized hypotonia,
CASS4	NM_001164114.1:c.1576G>Ap.(Val526Ile)	NM_001164114.1:c.1421G>Tp.(Arg474Leu)	AR_comphet	3.1	comphet	maternal& paternal	4	NDD	global developmental delay, intellectual disability, generalized hypotonia,
EXD3	NM_017820.4:c.859G>Ap.(Asp287Asn)	NM_017820.4:c.1831-2A>Gp.?	AR_comphet	6.7	comphet	maternal& paternal	2	NDD + Epilepsy	seizures, peripheral axonal neuropathy, motor delay, gait disturbance, EEG with focal epilepti-form discharges
FAM83G	NM_001039999.2:c.1133G>Ap.(Gly378Asp)	NM_001039999.2:c.2179G>Ap.(Val727Ile)	AR_comphet	2.6	comphet	maternal& paternal	3	NDD	Coloboma, Iris coloboma, mild Intellectual disability, mild Global developmental delay
CFAP54	XM_001715090.5:c.2257A>Gp.(Met753Val)	XM_001715090.5:c.2057G>Ap.(Arg686Lys)	AR_comphet	3.4	comphet	maternal& paternal	3	NDD	Coloboma, Iris coloboma, mild Intellectual disability, mild Global developmental delay
GRIN3B	NM_138690.1:c.1090_1091delp.(Met364Valfs*5)	NM_138690.1:c.1936A>Gp.(Met646Val)	AR_comphet	7.2	comphet	maternal& paternal	1	NDD	Intellectual disability, Abnormal facial shape, Myoclonus
EIF5B	NM_015904.3:c.3607C>Tp.(Gln1203*)		AD_denovo	10.1	het	de novo	1	NDD	Macrocephaly, Autism, Intellectual disability, Absent speech, Intellectual disability, severe
PTP4A1	NM_003463.4:c.8G>Ap.(Arg3Gln)		AD_denovo	5.3	het	de novo	1	NDD	mental retardation, autism
POLR1B	NM_001137604.1:c.2893G>Ap.(Val965Ile)		AD_denovo	6.5	het	de novo	3	NDD	Seizures, Pachygyria, Delayed CNS myelination, Heterotopia, Periventricular gray matter heterotopia, Intracranial cystic lesion, Abnormality of brain morphology
HIST1H4B	NM_003544.2:c.158A>Gp.(Glu53Gly)		AD_denovo	4.2	het	de novo	3	NDD	Seizures, Pachygyria, Delayed CNS myelination, Heterotopia, Periventricular gray matter heterotopia, Intracranial cystic lesion, Abnormality of brain morphology
BAHCC1	NM_001080519.2:c.4691+5C>G		AD_denovo	3.0	het	de novo	3	NDD	Seizures, Pachygyria, Delayed CNS myelination, Heterotopia, Periventricular gray matter heterotopia, Intracranial cystic lesion, Abnormality of brain morphology
PHACTR1	NM_001242648.2:c.1156G>Ap.(Glu386Lys)		AD_denovo	7.4	het	de novo	2	NDD	Global developmental delay, Intellectual disability, mild
KDM5A	NM_001042603.2:c.4048C>Tp.(Arg1350*)		AD_denovo	11.5	het	de novo	1	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Global developmental delay, Focal clonic seizures, Focal seizures with impairment of consciousness or awareness, Intellectual disability, severe, Focal motor seizures, Focal tonic seizures
DBP	NM_001352.4:c.511G>Tp.(Ala171Ser)		AD_denovo	6.1	het	de novo	2	NDD	Global developmental delay, Intellectual disability, mild
TANC2	NM_025185.3:c.4405delp.(Arg1469Glyfs*6)		AD_denovo	11.4	het	de novo	1	NDD + Epilepsy	Seizures, Global developmental delay, Encephalopathy, Epileptic encephalopathy
STC1	NM_003155.2:c.693_697delp.(Glu232Glyfs*12)		AD_denovo	6.7	het	de novo	1	NDD	mild global developmental delay, expressive speech disorder, obesity since age three years
KANK4	NM_181712.4:c.1849C>Tp.(Gln617*)		AD_denovo	4.6	het	de novo	1	NDD	Retinal coloboma, Seizures, Intellectual disability, mild, Global developmental delay, Motor delay, Hypoplasia of the retina, Intracranial cystic lesion, Mild global developmental delay, Infantile spasms
LCTL	NM_207338.3:c.692_693dup		AD_denovo	5.7	het	de novo	2	NDD + Epilepsy	epileptic encephalopathy, seizures

PABPC1	NM_002568.3:c.1691A>Gp.(Glu564Gly)		AD_denovo	11.0	het	de novo	1	NDD + Epilepsy	global developmental delay, seizures, visual impairment, bicuspid aortic valve
KLHL6	NM_130446.2:c.1061C>Ap.(Pro354Gln)		AR_homo	4.9	homo	maternal&paternal	2	NDD + Epilepsy	epileptic encephalopathy, seizures
MAPK8IP3	NM_001040439.1:c.1556G>Ap.(Arg519Gln)		AD_denovo	10.9	het	de novo	2	NDD	Microcephaly, Intellectual disability, Global developmental delay, Abnormality of body weight, Increased body weight,
RORB			AD_denovo	10.9	het	de novo	2	NDD	Hearing impairment, Hypermetropia, Nystagmus, Delayed speech and language development, Intellectual disability, Global developmental delay, Motor delay, Generalized tonic-clonic seizures, Short stature, Decreased body weight, Simple febrile seizures
GRIN3B	NM_138690.2:c.2114A>Gp.(Tyr705Cys)	NM_138690.2:c.2314G>Ap.(Gly772Ser)	AR_comphet	6.0	comphet	maternal&paternal	1	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Intellectual disability, progressive, Focal seizures, EEG with focal slow activity, Intellectual disability, severe, Focal motor seizures, EEG with focal epileptiform discharges, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Neurodevelopmental abnormality, Cognitive impairment
DNAJC7	NM_001144766.2:c.941C>Tp.(Ala314Val)		AD_denovo	6.4	het	de novo	1	NDD + Epilepsy	Seizures, Global developmental delay, Generalized seizures, Hypsarrhythmia, Epileptic spasms
ACTL6B	NM_016188.4:c.1027G>Ap.(Gly343Arg)		AD_denovo	10.7	het	de novo	1	NDD	Muscular hypotonia, Abnormality of mouth shape, Stereotypical hand wringing, Microcephaly, Global developmental delay
KIRREL2	NM_032123.6:c.1275delp.(Pro425Profs*41)		AD_inherited	6.0	het	paternal	1	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Absence seizures, Generalized myoclonic seizures, Episodic vomiting, Epileptic spasms, Myoclonic atonic seizures, Epileptic encephalopathy
CACNA1C	NM_199460.3:c.496T>Cp.(Phe166Leu)		AD_denovo	10.7	het	de novo	2	NDD + Epilepsy	epilepsy with absences and generalized tonic-clonic seizures, severe intellectual disability with autistic traits, low blood pressure, obstipation, normal MRI 2008
EIF3B	NM_001037283.1:c.28C>Ap.(Pro10Thr)		AD_denovo	7.0	het	de novo	1	NDD + Epilepsy	Absence seizures, EEG abnormality, Febrile seizures, Eyelid myoclonias, Childhood onset
HIST1H3H	NM_003536.2:c.397G>Tp.(Gly133Cys)		AD_denovo	4.6	het	de novo	1	NDD + Epilepsy	Global developmental delay, Hypsarrhythmia, Inability to walk, Epileptic spasms, Infantile spasms
FBP2	NM_003837.3:c.128A>Gp.(Lys43Arg)		AD_denovo	6.2	het	de novo	1	NDD	Macrocephaly, Delayed speech and language development, Global developmental delay, Motor delay, Frontal bossing, Delayed gross motor development, Delayed fine motor development, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Profound global developmental delay
TAB2	NM_015093.5:c.1448delp.(Pro483Leufs*16)		AD_denovo	10.6	het	de novo	3	NDD	Hypotelorism, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Intellectual disability, moderate, Single median maxillary incisor, Agenesis of permanent teeth, Abnormality of dental morphology, Reduced number of teeth, Intellectual disability, severe
AASDH	NM_181806.3:c.2908-5_2908-4insGTTp.?	NM_181806.3:c.3220dup, p.(Leu1074Profs*10)	AR_comphet	5.6	comphet	maternal&paternal	3	NDD + Epilepsy	Narrow mouth, Uplanted palpebral fissure, Delayed speech and language development, Intellectual disability, Global developmental delay, Pachygyria, Lissencephaly, Absent speech, Dysphagia, Polymicrogyria, Status epilepticus, Gliosis, Intellectual disability, moderate, Cerebellar malformation, Poor speech, Abnormality of the cerebral white matter, Excessive salivation, Focal white matter lesions, Focal seizures, Multifocal epileptiform discharges, Intellectual disability, severe, Epileptic spasms, EEG with focal epileptiform discharges, Cerebral white matter atrophy, Cerebral white matter agenesis, Oral-pharyngeal dysphagia
CAST	deletionexon16		AD_denovo	9.0	het	de novo	3	NDD + Epilepsy	Narrow mouth, Uplanted palpebral fissure, Delayed speech and language development, Intellectual disability, Global developmental delay, Pachygyria, Lissencephaly, Absent speech, Dysphagia, Polymicrogyria, Status epilepticus, Gliosis, Intellectual disability, moderate, Cerebellar malformation, Poor speech, Abnormality of the cerebral white matter, Excessive salivation, Focal white matter lesions, Focal seizures, Multifocal epileptiform discharges, Intellectual disability, severe, Epileptic spasms, EEG with focal epileptiform discharges, Cerebral white matter atrophy, Cerebral white matter agenesis, Oral-pharyngeal dysphagia
E2F4	NM_001950.3:c.947_958delp.(Ser316_Ser319del)		AD_denovo	6.6	het	de novo	2	NDD	Cleft palate, Intellectual disability, Intellectual disability, mild, Global developmental delay, Absent speech, Atria septal defect, Abnormal facial shape, Intellectual disability, moderate, Short stature, Intellectual disability, severe
C1orf228	NM_001145636.1:c.979C>Tp.(Arg327Cys)		AD_denovo	4.6	het	de novo	2	NDD	Cleft palate, Intellectual disability, Intellectual disability, mild, Global developmental delay, Absent speech, Atria septal defect, Abnormal facial shape, Intellectual disability, moderate, Short stature, Intellectual disability, severe

NSD2	NM_001042424.2:c.3295G>Ap.(Glu1099Lys)		AD_denovo	10.3	het	de novo	1	NDD	Cryptorchidism, Renal dysplasia, Phenotypic abnormality, Nephrocalcinosis, Delayed speech and language development, Global developmental delay, Motor delay, Cholestasis, Patent ductus arteriosus, Splenomegaly, Pyloric stenosis, Splenic cyst
KDM6B	NM_001080424.1:c.1130C>Tp.(Ala377Val)		AR_homo	8.4	homo	maternal&paternal	3	NDD + Epilepsy	Nystagmus, Horizontal nystagmus, Seizures, Global developmental delay, Absent speech, Cardiomyopathy, Vacuolated lymphocytes, Abnormal facial shape, Gait ataxia, Absence seizures, EEG abnormality, Myoclonic atonic seizures, Epileptic encephalopathy
ZNF664	NM_001204298.1:c.691G>Ap.(Glu231Lys)		AD_denovo	4.8	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Hypsarrhythmia, Epileptic spasms, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Profound global developmental delay, Cognitive impairment, Epileptic encephalopathy
NIT1	NM_001185092.1:c.244_256delp.(Phe83Hisfs*63)	NM_001185092.1:c.302T>Cp.(Leu101Pro)	AR_comphet	6.2	comphet	maternal&paternal	2	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Hypsarrhythmia, Epileptic spasms, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Profound global developmental delay, Cognitive impairment, Epileptic encephalopathy
LPIN2	NM_014646.2:c.2537A>Gp.(Asn846Ser)		AD_denovo	7.2	het	de novo	3	NDD + Epilepsy	Nystagmus, Horizontal nystagmus, Seizures, Global developmental delay, Absent speech, Cardiomyopathy, Vacuolated lymphocytes, Abnormal facial shape, Gait ataxia, Absence seizures, EEG abnormality, Myoclonic atonic seizures, Epileptic encephalopathy
GBP2	NM_004120.4:c.576_578delp.(Glu192_Pro193delinsAsp)	NM_004120.4:c.412G>Ap.(Ala138Thr)	AR_comphet	2.6	comphet	maternal&paternal	3	NDD + Epilepsy	Nystagmus, Horizontal nystagmus, Seizures, Global developmental delay, Absent speech, Cardiomyopathy, Vacuolated lymphocytes, Abnormal facial shape, Gait ataxia, Absence seizures, EEG abnormality, Myoclonic atonic seizures, Epileptic encephalopathy
MAPKAPK2	NM_004759.4:c.445C>Tp.(Arg149*)		AD_denovo	9.3	het	de novo	1	NDD + Epilepsy	Cryptorchidism, Hypospadias, Microcephaly, Visual impairment, Visual field defect, Intellectual disability, Muscular hypotonia, Global developmental delay, Plagiocephaly, Oligohydramnios, Intellectual disability, severe, Epileptic spasms, Moderate global developmental delay, Severe global developmental delay, Profound global developmental delay
MOXD1	NM_015529.3:c.350A>Gp.(His117Arg)		AR_homo	6.0	homo	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Intellectual disability, profound, Intellectual disability, moderate, Febrile seizures, Intellectual disability, borderline, Intellectual disability, severe, Focal tonic seizures
TLK2	NM_001112707.1:c.667A>Tp.(Met223Leu)		AD_unknown	4.8	het	unknown	3	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Intellectual disability, profound, Intellectual disability, moderate, Febrile seizures, Intellectual disability, borderline, Intellectual disability, severe, Focal tonic seizures
TTLL6	NM_001130918.1:c.2129G>Tp.(Ser710Ile)		AD_unknown	1.0	het	unknown	3	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Intellectual disability, profound, Intellectual disability, moderate, Febrile seizures, Intellectual disability, borderline, Intellectual disability, severe, Focal tonic seizures
CPXM2	NM_198148.2:c.170_172delp.(Phe57del)		AD_unknown	2.0	het	unknown	3	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Intellectual disability, profound, Intellectual disability, moderate, Febrile seizures, Intellectual disability, borderline, Intellectual disability, severe, Focal tonic seizures
KDM5B	NM_006618.4:c.1286T>Gp.(Ile429Ser)		AD_denovo	10.1	het	de novo	1	NDD + Epilepsy	Renal duplication, Hydrocephalus, Autism, Hypertrichosis, Intellectual disability, Seizures, Global developmental delay, Agenesis of corpus callosum, Abnormal facial shape, Intellectual disability, moderate, Impaired pain sensation, Intellectual disability, severe, Colpocephaly, Cognitive impairment, Septo-optic dysplasia
NIPAL3	NM_020448.4:c.205G>Ap.(Ala69Thr)	NM_020448.4:c.163-8G>Ap.?	AR_comphet	3.7	comphet	maternal&paternal	2	NDD	Hearing impairment, Sensorineural hearing impairment, Delayed speech and language development, Precocious puberty, Muscular hypotonia, Global developmental delay, Absent speech, Poor speech, High-frequency hearing impairment, Muscular hypotonia of the trunk

PLEKHG4B	NM_052909.3:c.461G>Tp.(Cys154Phe)	NM_052909.3:c.3124G>Ap.(Asp1042Asn)	AR_comphet	2.8	comphet	maternal&paternal	2	NDD	Hearing impairment, Sensorineural hearing impairment, Delayed speech and language development, Precocious puberty, Muscular hypotonia, Global developmental delay, Absent speech, Poor speech, High-frequency hearing impairment, Muscular hypotonia of the trunk
ZIK1	NM_001010879.3:c.924delp.(Ser308Serfs*203)		AR_homo	7.8	homo	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, Generalized seizures, Febrile seizures, Focal seizures
ZNF331	NM_001079906.1:c.281G>Ap.(Arg94His)		AR_homo	3.8	homo	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, Generalized seizures, Febrile seizures, Focal seizures
UBE3C	NM_014671.2:c.485G>Cp.(Ser162Thr)	NM_014671.2:c.871G>Ap.(Val291Ile)	AR_comphet	4.9	comphet	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, Generalized seizures, Febrile seizures, Focal seizures
SGF29	NM_138414.2:c.733T>Cp.(Tyr245His)		AD_denovo	6.7	het	de novo	1	NDD	Microcephaly, Abnormality of the outer ear, Protruding ear, Abnormality of the ear, Hypotelorism, Autistic behavior, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Talipes equinovarus, Hypoplasia of the corpus callosum, Intellectual disability, profound, Intellectual disability, moderate, Short stature, Intellectual disability, severe, Clinodactyly
HSPD1	NM_002156.4:c.1394_1406delp.(Ile465Lysfs*9)		AD_denovo	12.8	het	de novo	1	Neuro	Hypogonadotrophic hypogonadism, Tall stature, Psychosis, Depression, Psychotic episodes, Dementia, Overgrowth, Neurodegeneration, Bipolar affective disorder, Brain atrophy
PLCB3	NM_000932.2:c.1792G>Cp.(Glu598Gln)		AD_denovo	B	het	de novo	1	Renal/urology	Failure to thrive, Growth delay, Omphalocele, Double outlet right ventricle
STARD9	NM_020759.2:c.1649A>Gp.(Asn550Ser)	NM_020759.2:c.10380C>Gp.(His3460Gln)	AR_comphet	3.9	comphet	maternal&paternal	1	NDD + Epilepsy	Global developmental delay, Absence seizures, Intellectual disability, moderate, Progressive truncal ataxia, Epileptic spasms, Myoclonic absences, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Infantile spasms
GABRA3	NM_000808.3:c.931+5G>Ap.?		XL	7.3	hemi	maternal	2	NDD + Epilepsy	Microcephaly, Agitation, Intellectual disability, Intellectual disability, mild, Global developmental delay, Constipation, Intellectual disability, moderate, EEG abnormality, Intellectual disability, borderline, Attention deficit hyperactivity disorder, Epileptic spasms, Anteverted ears
ELMOD2	NM_153702.3:c.580C>Tp.(Arg194Cys)		AD_denovo	5.3	het	de novo	2	NDD + Epilepsy	Microcephaly, Agitation, Intellectual disability, Constipation, moderate, Attention deficit hyperactivity disorder, Epileptic spasms, Anteverted ears
NR2F6	NM_005234.3:c.1051G>Ap.(Gly351Arg)		AD_denovo	5.49	het	de novo	1	NDD	Microcephaly, Global developmental delay, Generalized hypotonia, Neonatal hypotonia, Failure to thrive, Severe failure to thrive, Failure to thrive in infancy, Ventricular septal defect, Abnormal cardiac septum morphology, Overlapping toe, Neonatal onset, Short stature, Muscular hypotonia of the trunk, Infantile muscular hypotonia, Abnormal ventricular septum morphology, Gerbode ventricular septal defect, Inlet ventricular septal defect, Muscular ventricular septal defect, Subarterial ventricular septal defect, Perimembranous ventricular septal defect, Restrictive ventricular septal defect, Abnormality of cardiovascular system morphology, Ventricular septal aneurysm, Muscular ventricular septal aneurysm
TMEM199	NM_152464.2:c.5C>Tp.(Ala2Val)		AD_denovo	5.3	het	de novo	2	NDD + Epilepsy	Microcephaly, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Abnormal facial shape, Status epilepticus, Intellectual disability, moderate, Infantile muscular hypotonia, Intellectual disability, severe, Epileptic spasms, Cognitive impairment
NCAPH	NM_001281710.1:c.563-4T>Gp.?	NM_001281710.1:c.667G>Ap.(Glu223Lys)	AR_comphet	5.2	comphet	maternal&paternal	2	NDD + Epilepsy	Microcephaly, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Abnormal facial shape, Status epilepticus, Intellectual disability, moderate, Infantile muscular hypotonia, Intellectual disability, severe, Epileptic spasms, Cognitive impairment
DOK2	NM_003974.3:c.1007C>Ap.(Thr336Asn)	NM_003974.3:c.602G>Ap.(Arg201His)	AR_comphet	C	comphet	maternal&paternal	1	Immunology	Hemolytic anemia, Fever, Abnormal thrombosis, Vasculitis, Intermittent thrombocytopenia, Congenital blindness, Colon perforation

EID2	NM_153232.3:c.161C>Tp.(Pro54Leu)	NM_153232.3:c.148G>Cp.(Ala50Pro)	AR_comphet	2.7	comphet	maternal&paternal	1	NDD + Epilepsy	Seizures, Abnormality of the corpus callosum, Agenesis of corpus callosum, Cerebellar hypoplasia, Neonatal hypoglycemia, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Generalized seizures, Focal clonic seizures, Heterotopia, Neonatal onset, Pontocerebellar atrophy, Periventricular gray matter heterotopia, Focal seizures, Epileptic spasms, Focal tonic seizures, Epileptic encephalopathy
ETS2	NM_001256295.1:c.148A>Tp.(Lys50*)		AD_unknown	2.9	het	unknown	1	NDD + Epilepsy	Global developmental delay, congenital bilateral perisylvic syndrome, EEG abnormalities, progressive tonic dystonia, perceptual disturbance, vertical gaze palsy
UNC13C	NM_001080534.2:c.283C>Tp.(Arg95*)		AR_homo	8.3	homo	maternal&paternal	1	NDD	Global developmental delay, microcephaly, Ehlers-Danlos-Syndrom (CHST1 positive)
GRIA4	NM_000829.3:c.2090G>Cp.(Arg697Pro)		AD_denovo	10.1	het	de novo	1	NDD	Delayed speech and language development, Intellectual disability, Poor speech
NRIP1	NM_003489.3:c.2077G>Tp.(Gly693Cys)		AD_denovo	5.9	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Generalized tonic clonic seizures, Focal clonic seizures, Intellectual disability, moderate, Focal seizures with impairment of consciousness or awareness, Intellectual disability, borderline, Generalized tonic seizures, Symptomatic seizures, Focal tonic seizures, Cognitive impairment
DDX42	NM_007372.3:c.221+1G>Ap.?		AD_denovo	10.2	het	de novo	1	NDD + Epilepsy	Epilepsy, optic atrophy, diabetes insipidus and hypothyroidism
ZCRB1	NM_033114.3:c.78G>Cp.(Leu26Phe)		AD_denovo	5.4	het	de novo	3	NDD + Epilepsy	Refractory epilepsy, epileptic encephalopathy, psychomotor retardation, microcephaly, short stature, facial dysmorphisms, nephrocalcinosis
DNAJC10	NM_001271581.1:c.1671+1G>Tp.?		AD_denovo	7.2	het	de novo	3	NDD + Epilepsy	Refractory epilepsy, epileptic encephalopathy, psychomotor retardation, microcephaly, short stature, facial dysmorphisms, nephrocalcinosis
CD200	NM_001004196.3:c.161C>Ap.(Thr54Lys)		AD_denovo	5.8	het	de novo	3	NDD + Epilepsy	Refractory epilepsy, epileptic encephalopathy, psychomotor retardation, microcephaly, short stature, facial dysmorphisms, nephrocalcinosis
SPHK2	NM_001204158.2:c.1534G>Tp.(Val512Leu)		AR_homo	5.0	homo	maternal&paternal	1	NDD + Epilepsy	Perinatal cerebral infarction, global developmental delay, motor and speech delay, microcephaly, epilepsy, short stature, combined heart failure (DORV, VSD, ASD, valvular pulmonary stenosis)
CELSR3	NM_001407.2:c.8254T>Cp.(Phe2752Leu)	NM_001407.2:c.79T>Cp.(Phe27Leu)	AR_comphet	6.1	comphet	maternal&paternal	1	NDD + Epilepsy	No language development, microcephaly (-2,2 SD), short stature (<3P), EEG abnormalities, epilepsy, delayed myelination in cMRT; Trio-Exom-Analysis unremarkable (05,12,2016)
GEMIN4	NM_015721.2:c.1580A>Gp.(Asn527Ser)	NM_015721.2:c.1415_1416delp.(Pro472Argfs*23)	AR_comphet	6.8	comphet	maternal&paternal	2	NDD + Epilepsy	Microcephaly, Intellectual disability, Global developmental delay, Abnormal heart morphology, Ventricular septal defect, Status epilepticus, Intellectual disability, moderate, Short stature, Atrioventricular canal defect, Intellectual disability, borderline, Intellectual disability, severe, Epileptic spasms, Epileptiform EEG discharges, Abnormality of cardiovascular system morphology
ZNF319	NM_020807.2:c.1294G>Cp.(Val432Leu)		AD_denovo	5.2	het	de novo	2	NDD + Epilepsy	Microcephaly, Intellectual disability, Global developmental delay, Abnormal heart morphology, Ventricular septal defect, Status epilepticus, Intellectual disability, moderate, Short stature, Atrioventricular canal defect, Intellectual disability, borderline, Intellectual disability, severe, Epileptic spasms, Epileptiform EEG discharges, Abnormality of cardiovascular system morphology
RENBP	NM_002910.5:c.695G>Ap.(Gly232Glu)		XL	5.5	hemi	maternal	1	NDD + Epilepsy	Epilepsy, susceptibility to fall
GABBR1	NM_001470.3:c.1190C>Tp.(Ala397Val)		AD_denovo	10.1	het	de novo	2	NDD	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Global developmental delay, Motor delay, Encopresis
NARS1	NM_004539.3:c.1600C>Tp.(Arg534*)		AD_denovo	7.6	het	de novo	2	NDD + Epilepsy	GDD, ataxia, muscular hypotonia, microcephaly, EEG abnormalities, brain volume reduction
INTS1	NM_001080453.2:c.6248T>Gp.(Phe2083Cys)	NM_001080453.2:c.5272A>Gp.(Ile1758Val)	AR_comphet	5.5	comphet	maternal&paternal	2	NDD + Epilepsy	GDD, ataxia, muscular hypotonia, microcephaly, EEG abnormalities, brain volume reduction
STARD9	NM_020759.2:c.12652C>Tp.(His4218Tyr)	NM_020759.2:c.13445C>Tp.(Ser4482Phe)	AR_comphet	4.2	comphet	maternal&paternal	1	NDD + Epilepsy	mild ID, generalized epilepsy
IQCH	NM_001031715.2:c.2552T>Cp.(Leu851Pro)		AD_denovo	5.2	het	de novo	1	NDD	GDD, bland-white-garland-syndrome, facial dysmorphisms, cleft palate, sudden cardiac arrest at the age of 3 months, hemi spastic
ZC3H4	NM_015168.1:c.54_71dup, p.(Pro19_Pro24dup)		AD_denovo	6.5	het	de novo	1	NDD + Epilepsy	global developmental delay, faocal epilepsy, truncal ataxia
GIPC1	NM_005716.3:c.718C>Tp.(Arg240*)		AD_denovo	6.3	het	de novo	1	NDD	Intellectual disability, V,a, epilepsy, failure to thrive, short stature, microcephaly, hypotonia, obstipation, strabismus, not able to walk, no language

EIF2AK2	NM_001135651.2:c.1210T>Cp.(Tyr404His)		AD_denovo	7.1	het	de novo	1	NDD + Epilepsy	West syndrome with salaam spasms, hypsarrythmia in EEG, age-appropriate development
MADD	NM_001135943.1:c.2284G>Tp.(Ala762Ser)		AR_homo	9.0	homo	maternal&paternal	1	NDD + Epilepsy	epileptic encephalopathy, therapy-resistant seizures, pain in legs
CSMD3	NM_052900.2:c.9581A>Cp.(Gln3194Pro)	NM_052900.2:c.7073G>Ap.(Arg2358Gln)	AR_comphet	5.1	comphet	maternal&paternal	1	NDD + Epilepsy	one tonic spasm, developmental delay, 20-30 headdrops per day, hypsarrythmia
MTMR9	NM_015458.3:c.220A>Gp.(Lys74Glu)		AD_denovo	6.2	het	de novo	2	NDD	Developmental delay, club foot, short stature, microcephaly, deafness
WDFY3	NM_014991.4:c.749A>Gp.(Asn250Ser)		AD_denovo	10.0	het	de novo	2	NDD + Epilepsy	seizures, peripheral axonal neuropathy, motor delay, gait disturbance, EEG with focal epilepti-form discharges
BBX	NM_001142568.2:c.2524C>Gp.(Arg842Gly)		AR_homo	7.3	homo	maternal&paternal	2	NDD	mild ID, facial dysmorphisms, Missing anterior teeth in the lower jaw, Vierfingerfurche, dry skin, narrow mouth, behavioral abnormalities
SNX6	NM_021249.4:c.586C>Tp.(Arg196*)		AR_homo	5.6	homo	maternal&paternal	2	NDD	mild ID, facial dysmorphisms, Missing anterior teeth in the lower jaw, simian crease, dry skin, narrow mouth, behavioral abnormalities
CAPS2	NM_032606.3:c.525+1G>Ap.?		AD_denovo	5.7	het	de novo	1	NDD + Epilepsy	mental retardation, epilepsy, no speech development, deafness, short stature
APLN	NM_014499.3:c.416T>Cp.(Phe139Ser)		XL	4.9	hemi	maternal	1	NDD	developmental delay, speech delay, motor delay, aggressive behaviour, selfharming behaviour, no ID (IQ98)
SLTM	NM_001013843.2:c.2595G>Ap.(Trp865*)		AD_denovo	9.2	het	de novo	1	Neuro	Muscular hypotonia, Hypometric saccades, Chorea, Mild conductive hearing impairment, Constipation, Scapular winging, Gait ataxia, Truncal ataxia, Motor delay
POU3F3	NM_006236.2:c.1220G>Tp.(Arg407Leu)		AD_unknown	7.5	het	unknown	1	NDD	GDD (first words with 27mo, first steps with >30mo), microcephaly, EEG abnormalities, borad-based gait, strabism, myopia, facial dysmorphism
PPM1G	NM_177983.2:c.1579T>Cp.(Ser527Pro)		AR_homo	4.2	homo	maternal&paternal	2	NDD + Metabolis	GDD, adipositas, unbalanced glucosetolerance, massive hyperinsulinamia, muscular hypotonia,
ERN2	NM_033266.3:c.2489C>Tp.(Pro830Leu)		AR_homo	4.4	homo	maternal&paternal	2	NDD	GDD, adipositas, unbalanced glucosetolerance, massive hyperinsulinamia, muscular hypotonia,
PCSK1N	NM_013271.4:c.491T>Cp.(Leu164Pro)		XL	6.0	hemi	maternal	1	NDD + Epilepsy	familiar epilepsy, speech delay, ADHS
NCOR2	NM_001077261.3:c.7241C>Tp.(Ala2414Val)	NM_001077261.3:c.1520_1528dup, p.(Gln507_Gln509dup)	AR_comphet	6.1	comphet	maternal&paternal	1	NDD + Epilepsy	Balkenagenesie, Polymikrogyrie, Plexuszysten, retinale Auffälligkeiten
PCDHA11	NM_018902.4:c.88C>Tp.(Gln30*)		AD_denovo	5.2	het	de novo	2	NDD + Epilepsy	epilepsy with Lennox-Gastaut syndrome and complex retardation
PAK1	NM_001128620.1:c.1409T>Gp.(Leu470Arg)		AD_denovo	9.9	het	de novo	1	ndd	Macrocephaly, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Focal clonic seizures, Focal seizures, Intellectual disability, severe, Epileptic spasms, Focal motor seizures, Focal tonic seizures, Cognitive impairment
DHRS7	NM_016029.3:c.475A>Gp.(Ile159Val)		AD_denovo	5.3	het	de novo	2	NDD + Epilepsy	epilepsy with Lennox-Gastaut syndrome and complex retardation
EXOC4	NM_001037126.1:c.472-6T>Cp.?	NM_001037126.1:c.860C>Tp.(Ala287Val)	AR_comphet	5.6	comphet	maternal&paternal	1	NDD + Epilepsy	early onset epilepsy, mild developmental delay, decreased glucose in liquor, behavarioal abnormalities
ANKRD17	NM_032217.4:c.3751_3754delp.(Arg1252Thrfs*6)		AD_denovo	9.9	het	de novo	1	NDD	Cryptorchidism, Microcephaly, Strabismus, Hypermetropia, Behavioral abnormality, Stereotypy, Global developmental delay, Small for gestational age, Short stature
ANKRD17	NM_032217.4:c.5360_5363delp.(Gln1787Argfs*5)		AD_denovo	9.9	het	de novo	1	NDD	Coarse facial features, Muscular hypotonia, Global developmental delay, Motor delay, Hypertonia, Pes planus, Gait ataxia, Limb hypertonia, Muscular hypotonia of the trunk
ZMYM2	NM_001190964.2:c.2338C>Tp.(Arg780*)		AD_denovo	9.9	het	de novo	1	NDD + Epilepsy	Seizures, Focal seizures, Generalized myoclonic seizures, Focal myoclonic seizures, Segmental myoclonic seizures, Falls, Frequent falls
PKP4	NM_001005476.2:c.744_745delp.(Val250Aspfs*110)		AD_denovo	6.8	het	de novo	1	NDD	Microcephaly, Global developmental delay, Global brain atrophy
PTK2B	NM_004103.4:c.1057C>Tp.(Arg353Trp)		AD_inherited	5.0	het	paternal	1	NDD + Epilepsy	Absence seizures, familiar

DST	NM_001144769.2:c.11762G>Ap.(Arg3921Gln)	NM_001144769.2:c.227C>Ap.(Ala76Glu)	AR_comphet	B	comphet	maternal&paternal	1	Metabolism	hypotrophes Frühgeborenes, Mikrozephalie, Leberversagen, Cholestase, Herzinsuffizienz, Gastrointestinale Blutung, Hypoglykämie, intraventrikuläre Blutung
GABRA2	NM_000807.3:c.438delp.(Arg147Glufs*12)		AD_denovo	A	het	de novo	1	Growth, Skeletal	short stature, muscular hypotonia, micropenis, acromely, hydronephrosis, congenital GH-deficiency, hypogonadism
RGL4	NM_153615.1:c.101C>Tp.(Thr34Met)		AD_denovo	3.5	het	de novo	2	NDD + Epilepsy	Autism, Intellectual disability, Intellectual disability, mild, Global developmental delay, Mental deterioration, Intellectual disability, profound, Generalized seizures, Intellectual disability, moderate, Intellectual disability, borderline, Focal seizures, Multifocal epileptiform discharges, Intellectual disability, severe, Epileptic spasms, Epileptiform EEG discharges, EEG with focal epileptiform discharges, Polymorphic focal epileptiform discharges, EEG with generalized epileptiform discharges, Cognitive impairment, Epileptic encephalopathy
DOCK7	NM_001271999.1:c.2932C>Tp.(Arg978Cys)	NM_001271999.1:c.2464G>Tp.(Ala822Ser)	AR_comphet	6.2	comphet	maternal&paternal	1	NDD	Global developmental delay, postnatal growth retardation, facial dysmorphisms with ante-verted nares, smooth philtrum, narrow upper lip and dysplastic ears
STMN3	NM_015894.3:c.19+8C>Ap.?		AD_denovo	3.7	het	de novo	1	NDD + Epilepsy	moderate ID, focal epilepsy, brain atrophy, stair fall
SYTL5	NM_001163334.1:c.2118T>Ap.(Gly706=)		AD_denovo	1.8	hemi	de novo	1	NDD + Epilepsy	GDD, syndromal mental retardation, no walking, no speech, epilepsy, dysmorphism, microcephaly, calcium salt in bones decreased
TRIM47	NM_033452.2:c.433C>Tp.(Leu145Phe)		AD_denovo	7.0	het	de novo	1	NDD + Epilepsy	Structural focal epilepsy with secondary generalized tonic-clonic seizures, first seizures with 3 month, right frontal polymicrogyria, small visceral cranium, right convex thoracic scoliosis, moderate mental retardation with behavioral abnormalities, absent speech, latent left-sided pareses
RFX3	NM_001282116.1:c.115C>Tp.(Gln39*)		AD_denovo	9.8	het	de novo	1	NDD	(+) Global developmental delay,(+) Poor coordination,(+) Large for gestational age,(+) Overgrowth,(+) Tall stature,(+) Ataxia,(+) Muscular hypotonia,(+) Behavioral abnormality
DNAJC17	NM_018163.2:c.273G>Tp.(Glu91Asp)		AD_denovo	5.2	het	de novo	1	NDD + Epilepsy	Hearing abnormality, Hearing impairment, Sensorineural hearing impairment, Strabismus, Psychosis, Osteoporosis, Intellectual disability, Seizures, Intellectual disability, mild, Mental deterioration, Generalized tonic-clonic seizures, EEG abnormality, Kyphosis, Type II diabetes mellitus, Intellectual disability, progressive, Intellectual disability, borderline, Severe hearing impairment, Cognitive impairment
ARAP2	NM_015230.3:c.4037G>Ap.(Arg1346Gln)	NM_015230.3:c.2257G>Cp.(Glu753Gln)	AR_comphet	3.2	comphet	maternal&paternal	1	NDD	N/A
BHLHE41	NM_030762.2:c.1222G>Cp.(Ala408Pro)		AD_denovo	5.4	het	de novo	1	NDD + Epilepsy	Seizures, Febrile seizures, Childhood onset, Epileptic encephalopathy
SLC29A1	NM_001078174.1:c.766+5G>Ap.?	NM_001078174.1:c.1357C>Tp.(Arg453Trp)	AR_comphet	5.2	comphet	maternal&paternal	2	NDD + Epilepsy	Autism, Intellectual disability, Intellectual disability, mild, Global developmental delay, Mental deterioration, Intellectual disability, profound, Generalized seizures, Intellectual disability, moderate, Intellectual disability, borderline, Focal seizures, Multifocal epileptiform discharges, Intellectual disability, severe, Epileptic spasms, Epileptiform EEG discharges, EEG with focal epileptiform discharges, Polymorphic focal epileptiform discharges, EEG with generalized epileptiform discharges, Cognitive impairment, Epileptic encephalopathy
GPKOW	NM_015698.5:c.511A>Gp.(Met171Val)		XL	6.1	hemi	maternal	1	NDD + Epilepsy	ID, focal epilepsy, motor delay, speech delay, autism, behavioral abnormalities
CNP	NM_033133.4:c.44A>Gp.(Lys15Arg)		AR_homo	8.9	homo	maternal&paternal	1	NDD	schwere Intelligenzminderung, spricht nur Einzelworte, eingeschränktes Sprachverständnis, körperlich gesund
MBD2	NM_003927.4:c.107G>Tp.(Gly36Val)		AD_denovo	7.3	het	de novo	1	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Focal seizures, Atonic seizures, Intellectual disability, severe, Epileptic spasms, Epileptic encephalopathy
MEGF11	NM_032445.2:c.3080T>Gp.(Leu1027Arg)	NM_032445.2:c.254G>Cp.(Arg85Thr)	AR_comphet	4.4	comphet	maternal&paternal	1	NDD + Epilepsy	Seizures, Prolonged QT interval, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Recurrent infections, Focal seizures

ANKRD33B	NM_001164440.1:c.784G>Ap.(Glu262Lys)	NM_001164440.1:c.1421A>Cp.(Glu474Ala)	AR_comphet	3.3	comphet	maternal&paternal	1	NDD	Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Intellectual disability, moderate, Intellectual disability, severe
SEL1L	NM_001244984.1:c.149C>Tp.(Thr50Ile)		AD_denovo	5.5	het	de novo	1	NDD + Epilepsy	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Intellectual disability, profound, Intellectual disability, moderate, Intellectual disability, severe
TNRC18	NM_001080495.2:c.7518dup, p.(Ala2507Argfs*44)		AD_denovo	7.0	het	de novo	1	NDD	Microcephaly, Intellectual disability, Global developmental delay, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Profound global developmental delay
DOCK7	NM_001271999.1:c.2977T>Cp.(Trp993Arg)	NM_001271999.1:c.708delp.(Phe236Leufs*13)	AR_comphet	8.0	comphet	maternal&paternal	3	NDD + Epilepsy	Microcephaly, Strabismus, Ptosis, Hypermetropia, Nystagmus, Behavioral abnormality, Autism, Stereotypy, Seizures, Global developmental delay, Absent speech, Abnormal facial shape, Hypoplasia of the corpus callosum, Intellectual disability, profound, Intellectual disability, moderate, Poor speech, Short stature, Intellectual disability, severe, Epileptic spasms, Epileptic encephalopathy
ZNF219	NM_001101672.1:c.673_678delp.(Ala225_Pro226del)		AD_denovo	5.5	het	de novo	3	NDD + Epilepsy	Microcephaly, Strabismus, Ptosis, Hypermetropia, Nystagmus, Behavioral abnormality, Autism, Stereotypy, Seizures, Global developmental delay, Absent speech, Abnormal facial shape, Hypoplasia of the corpus callosum, Intellectual disability, profound, Intellectual disability, moderate, Poor speech, Short stature, Intellectual disability, severe, Epileptic spasms, Epileptic encephalopathy
DCDC1	NM_181807.3:c.515G>Ap.(Arg172Lys)		AD_denovo	4.6	het	de novo	3	NDD + Epilepsy	Microcephaly, Strabismus, Ptosis, Hypermetropia, Nystagmus, Behavioral abnormality, Autism, Stereotypy, Seizures, Global developmental delay, Absent speech, Abnormal facial shape, Hypoplasia of the corpus callosum, Intellectual disability, profound, Intellectual disability, moderate, Poor speech, Short stature, Intellectual disability, severe, Epileptic spasms, Epileptic encephalopathy
ZSWIM5	NM_020883.1:c.2369G>Ap.(Arg790His)	NM_020883.1:c.1915C>Tp.(Pro639Ser)	AR_comphet	3.4	comphet	maternal&paternal	1	NDD + Epilepsy	Microcephaly, Hearing impairment, Myopia, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Febrile seizures
APOLD1	NM_001130415.1:c.755_756delp.(Glu252Valfs*20)		AR_homo	7.6	homo	maternal&paternal	4	NDD + Epilepsy	Intellectual disability, Epileptic spasms, Seizures, Focal seizures with impairment of consciousness or awareness, Focal motor seizures, Abnormality of brain morphology, Neuroepithelial neoplasm
AVPR1A	NM_000706.4:c.164T>Ap.(Ile55Asn)		AR_homo	7.9	homo	maternal&paternal	4	NDD + Epilepsy	Intellectual disability, Epileptic spasms, Seizures, Focal seizures with impairment of consciousness or awareness, Focal motor seizures, Abnormality of brain morphology, Neuroepithelial neoplasm
HELZ2	NM_001037335.2:c.6691+4C>Tp.?		AR_homo	5.5	homo	maternal&paternal	4	NDD + Epilepsy	Intellectual disability, Epileptic spasms, Seizures, Focal seizures with impairment of consciousness or awareness, Focal motor seizures, Abnormality of brain morphology, Neuroepithelial neoplasm
NUSAP1	NM_001243142.1:c.808C>Tp.(Arg270Cys)		AR_homo	5.2	homo	maternal&paternal	4	NDD + Epilepsy	Intellectual disability, Epileptic spasms, Seizures, Focal seizures with impairment of consciousness or awareness, Focal motor seizures, Abnormality of brain morphology, Neuroepithelial neoplasm
FRYL	NM_015030.1:c.1224delp.(Lys409Argfs*15)		AD_denovo	9.7	het	de novo	1	NDD	Cleft palate, Microcephaly, Dental crowding, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Abnormal facial shape, Mild short stature, Decreased body weight, facial dysmorphism, low-set ears, down-slanting palpebral fissures, hearing loss, eating disorder, Borderline personality disorder.
STARD7	NM_020151.3:c.64C>Tp.(Leu22Phe)		AD_denovo	4.8	het	de novo	1	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Absence seizures
PIGC	NM_002642.3:c.422C>Tp.(Thr141Ile)	NM_002642.3:c.138C>Ap.(Tyr46*)	AR_comphet	7.6	comphet	maternal&paternal	2	NDD + Epilepsy	Macrocephaly, Hypotelorism, Intellectual disability, Seizures, Intellectual disability, mild, Obesity, Large for gestational age, Dilated cardiomyopathy, Absence seizures, Focal seizures with impairment of consciousness or awareness, Diffuse cerebellar atrophy
PHRF1	NM_020901.3:c.1451+3G>Ap.?	NM_020901.3:c.3544A>Gp.(Lys1182Glu)	AR_comphet	3.6	comphet	maternal&paternal	2	NDD + Epilepsy	Macrocephaly, Hypotelorism, Intellectual disability, Seizures, Intellectual disability, mild, Obesity, Large for gestational age, Dilated cardiomyopathy, Absence seizures, Focal seizures with impairment of consciousness or awareness, Diffuse cerebellar atrophy

SYNJ2	NM_003898.3:c.107C>Ap.(Ala36Asp)		AD_denovo	6.8	het	de novo	1	NDD + Epilepsy	Restlessness, Intellectual disability, Hypsarrhythmia, Epileptic spasms, Infantile spasms, I
KCNK9	NM_001282534.1:c.391C>Tp.(Arg131Cys)		AD_denovo	9.5	het	de novo	1	NDD	muscular hypotonia, tongue fasciculation, motor developmental delay
ANO4	NM_178826.3:c.868G>Ap.(Ala290Thr)		AR_homo	5.2	homo	maternal&paternal	2	NDD + Epilepsy	Seizures, Focal seizures, Multifocal epileptiform discharges, EEG with focal epileptiform discharges, Polymorphic focal epileptiform discharges, EEG with focal spikes
GNAI1	NM_002069.5:c.143C>Ap.(Thr48Lys)		AD_denovo	9.5	het	de novo	2	NDD + Epilepsy	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Absent speech, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Generalized seizures, Focal clonic seizures, Intellectual disability, moderate, Focal seizures with impairment of consciousness or awareness, Poor speech, Focal seizures, Intellectual disability, severe, Epileptic spasms, Focal motor seizures, Focal tonic seizures, Abnormality of movement, Cognitive impairment
TAF7L	NM_001168474.1:c.1100A>Gp.(Gln367Arg)		XL	4.0	hemi	maternal	2	NDD + Epilepsy	Seizures, Focal seizures, Multifocal epileptiform discharges, EEG with focal epileptiform discharges, Polymorphic focal epileptiform discharges, EEG with focal spikes
ZC3H12B	NM_001010888.3:c.899A>Gp.(Asn300Ser)		XL	5.2	hemi	maternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Ataxia
NUCB2	NM_005013.2:c.88_91delp.(Asp30Argfs*15)		AD_denovo	6.2	het	de novo	3	NDD + Epilepsy	Intellectual disability, Seizures, Ataxia
NCOA7	NM_001122842.2:c.2660+2T>Ap.?		AD_denovo	5.7	het	de novo	1	NDD	Behavioral abnormality, Delayed speech and language development, Global developmental delay
BSN	NM_003458.3:c.11163C>Gp.(Ser3721Arg)		AD_denovo	8.7	het	de novo	2	NDD + Epilepsy	Seizures, Hemiplegia, Developmental stagnation, Epileptic encephalopathy, Global developmental delay, Abnormality of movement, Progressive extrapyramidal movement disorder, Reduced consciousness/confusion, Epileptiform EEG discharges
DNAH17	NM_173628.3:c.11678-7_11678-3delp.?	NM_173628.3:c.9998C>Tp.(Ser3333Leu)	AR_comphet	3.9	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Hemiplegia, Developmental stagnation, Epileptic encephalopathy, Global developmental delay, Abnormality of movement, Progressive extrapyramidal movement disorder, Reduced consciousness/confusion, Epileptiform EEG discharges
ASTN2	NM_014010.4:c.1013A>Gp.(Lys338Arg)	NM_014010.4:c.872A>Tp.(Asp291Val)	AR_comphet	6.2	comphet	maternal&paternal	1	NDD + Epilepsy	Ptosis, Seizures, Epileptic encephalopathy
EZH2	NM_001203247.1:c.2197G>Ap.(Ala733Thr)		AD_denovo	9.8	het	de novo	3	NDD	Microcephaly, Delayed speech and language development, Global developmental delay, Abnormal facial shape, Severe short stature, Short stature, Intellectual disability
TCP11	NM_001093728.2:c.1440T>Ap.(Phe480Leu)		AD_denovo	4.4	het	de novo	3	NDD	Microcephaly, Delayed speech and language development, Global developmental delay, Abnormal facial shape, Severe short stature, Short stature, Intellectual disability
TNMD	NM_022144.2:c.145G>Tp.(Gly49Trp)		AD_denovo	4.6	het	de novo	3	NDD	Microcephaly, Delayed speech and language development, Global developmental delay, Abnormal facial shape, Severe short stature, Short stature, Intellectual disability
NRXN2	NM_015080.3:c.4484delp.(Phe1495Serfs*71)		AD_unknown	10.6	het	unknown	1	NDD	Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Abnormal facial shape, Intellectual disability, moderate
SLIT1	NM_003061.2:c.4378C>Tp.(Arg1460Trp)		AR_homo	6.8	homo	maternal&paternal	1	NDD	Intellectual disability, movement disorder
KLHDC9	NM_152366.4:c.886+1G>Cp.?		AR_homo	8.2	homo	maternal&paternal	1	NDD + Epilepsy	Seizures, Global developmental delay, Generalized hypotonia, Abnormality of the cerebellum, Abnormal facial shape, Generalized seizures
CASKIN1	NM_020764.3 :c.3091C>Tp.(Arg1031Cys)		AR_homo	6.0	homo	maternal&paternal	1	NDD	Speech delay, mild intellectual disability, non-verbal IQ of 57, no prominent facial dysmorphism, stereotypic behavior
TMEM121B	NM_031890.3:c.254T>Cp.(Val85Ala)		AD_denovo	4.5	het	de novo	2	NDD	Hydrocephalus, Hand polydactyly, Postaxial hand polydactyly, Megalencephaly, Foot polydactyly, Postaxial foot polydactyly, Polymicrogyria, Polymicrogyria, anterior to posterior gradient, Severe hydrocephalus, Unilateral polymicrogyria, Frontoparietal polymicrogyria, Polydactyly, Perisylvian polymicrogyria, Postaxial polydactyly
FAM13B	NM_016603.3:c.2203G>Ap.(Val735Ile)	NM_016603.3:c.110A>Gp.(Glu37Gly)	AR_comphet	4.7	comphet	maternal&paternal	2	NDD	Hydrocephalus, Hand polydactyly, Postaxial hand polydactyly, Megalencephaly, Foot polydactyly, Postaxial foot polydactyly, Polymicrogyria, Polymicrogyria, anterior to posterior gradient, Severe hydrocephalus, Unilateral polymicrogyria, Frontoparietal polymicrogyria, Polydactyly, Perisylvian polymicrogyria, Postaxial polydactyly

MDGA2	NM_001113498.2:c.794T>Ap.(Val265Asp)		AD_denovo	7.7	het	de novo	3	NDD	Aggressive behavior, Autistic behavior, Short attention span, Intellectual disability, mild, Global developmental delay, Specific learning disability, Leukemia, Acute leukemia, Short stature, Lymphoid leukemia, Chronic leukemia, Intellectual disability, borderline, Abnormal aggressive, impulsive or violent behavior, Attention deficit hyperactivity disorder, Myeloid leukemia
ACAD10	NM_001136538.1:c.1670C>Gp.(Pro557Arg)	NM_001136538.1:c.3230A>Gp.(His1077Arg)	AR_comphet	5.4	comphet	maternal&paternal	3	NDD	Aggressive behavior, Autistic behavior, Short attention span, Intellectual disability, mild, Global developmental delay, Specific learning disability, Leukemia, Acute leukemia, Short stature, Lymphoid leukemia, Chronic leukemia, Intellectual disability, borderline, Abnormal aggressive, impulsive or violent behavior, Attention deficit hyperactivity disorder, Myeloid leukemia
MAGEA10	NM_001011543.2:c.125C>Tp.(Thr42Ile)		XL	4.9	hemi	maternal	3	NDD	Aggressive behavior, Autistic behavior, Short attention span, Intellectual disability, mild, Global developmental delay, Specific learning disability, Leukemia, Acute leukemia, Short stature, Lymphoid leukemia, Chronic leukemia, Intellectual disability, borderline, Abnormal aggressive, impulsive or violent behavior, Attention deficit hyperactivity disorder, Myeloid leukemia
STPG2	NM_174952.2:c.1128T>Ap.(Ser376Arg)	NM_174952.2:c.431G>Ap.(Gly144Asp)	AR_comphet	3.8	comphet	maternal&paternal	1	NDD + Epilepsy	Delayed speech and language development, Seizures, Focal seizures, Multifocal epileptiform discharges, EEG with focal epileptiform discharges, Epileptic encephalopathy
CACNA1B	NM_000718.3:c.1442C>Tp.(Ala481Val)		AD_denovo	11.0	het	de novo	2	NDD + Epilepsy	Seizures, Global developmental delay, Hypoglycorrhachia, Hyperglycorrhachia
DGKQ	NM_001347.3:c.1736A>Tp.(His579Leu)	NM_001347.3:c.1408C>Tp.(Arg470Trp)	AR_comphet	4.8	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Global developmental delay, Hypoglycorrhachia, Hyperglycorrhachia
TACC2	NM_206862.3:c.65_66insCCTCp.(Gln23Leufs*22)	NM_206862.3:c.7801C>Tp.(Pro2601Ser)	AR_comphet	5.4	comphet	maternal&paternal	3	NDD	hydrocephalus, intellectual disability, spastic paraplegia, global developmental delay, agenesis of corpus callosum, spastic paraparesis, intellectual disability, severe, cerebral palsy
GET4	NM_015949.2:c.491A>Gp.(Tyr164Cys)		AR_homo	5.9	homo	maternal&paternal	3	NDD	hydrocephalus, intellectual disability, spastic paraplegia, global developmental delay, agenesis of corpus callosum, spastic paraparesis, intellectual disability, severe, cerebral palsy
CDCA2	NM_152562.3:c.922A>Gp.(Arg308Gly)	NM_152562.3:c.1634C>Tp.(Thr545Ile)	AR_comphet	3.4	comphet	maternal&paternal	3	NDD	hydrocephalus, intellectual disability, spastic paraplegia, global developmental delay, agenesis of corpus callosum, spastic paraparesis, intellectual disability, severe, cerebral palsy
DZIP3	NM_014648.3:c.209C>Tp.(Pro70Leu)		AD_denovo	6.6	het	de novo	1	NDD	Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Poor speech, Cognitive impairment
MTCL1	NM_015210.3:c.604A>Gp.(Thr202Ala)	NM_015210.3:c.1607T>Cp.(Ile536Thr)	AR_comphet	4.7	comphet	maternal&paternal	1	NDD + Epilepsy	Microcephaly, Hearing impairment, Intellectual disability, Seizures, Intellectual disability, mild, Spasticity, Global developmental delay, Polyhydramnios, Intellectual disability, profound, Intellectual disability, moderate, Respiratory failure, Short stature, Intellectual disability, progressive, Intellectual disability, borderline, Intellectual disability, severe, Gait imbalance, Gait disturbance, Abnormality of movement, Severe short stature
MPP3	NM_001330233.1:c.742C>Tp.(Arg248Cys)		AR_homo	4.7	homo	maternal&paternal	2	NDD	Macrocephaly, Delayed speech and language development, Global developmental delay, Obesity
VGF	NM_003378.3:c.1318G>Ap.(Glu440Lys)		AR_homo	6.3	homo	maternal&paternal	4	NDD	Macrocephaly, Delayed speech and language development, Global developmental delay, Obesity
ZNF182	NM_001178099.1:c.1319C>Tp.(Thr440Met)		XL	4.7	hemi	maternal	4	NDD	Macrocephaly, Delayed speech and language development, Global developmental delay, Obesity
CASB	NM_007220.3:c.352_354dup, p.(Gly118dup)		XL	4.4	hemi	maternal	4	NDD	Macrocephaly, Delayed speech and language development, Global developmental delay, Obesity
TNK2	NM_001010938.1:c.278T>Gp.(Leu93Arg)		AD_denovo	7.6	het	de novo	2	NDD	Myopia, Nystagmus, Stereotypy, Delayed speech and language development, Intellectual disability, Motor delay, Absent speech, Abnormality of the foot, Intellectual disability, profound, Difficulty walking, Poor speech, Equinus calcaneus, Vertical nystagmus, Intellectual disability, severe, Severe global developmental delay, Psychomotor retardation

PSD3	NM_015310.3:c.3092A>Gp.(Glu1031Gly)	NM_015310.3:c.2929-3C>Tp.?	AR_comphet	5.8	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Focal seizures, Intellectual disability, severe
ARMC3	NM_173081.4:c.1346G>Ap.(Arg449His)		AR_homo	3.4	homo	maternal&paternal	2	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Focal seizures, Intellectual disability, severe
SRGAP3	NM_014850.3:c.2227+6_2227+9delp.?		AD_denovo	C	het	de novo	1	Fehlbildungen	Premature birth, Esophageal atresia, Spina bifida, Total anomalous pulmonary venous return
CSMD1	NM_033225.5:c.3641T>Cp.(Leu1214Pro)		AD_denovo	7.7	het	de novo	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Intellectual disability, severe, Epileptiform EEG discharges, Neurodevelopmental delay, Epileptic encephalopathy, Myoclonic absences, EMG: myotonic discharges, Generalized tonic-clonic seizures
MFAP1	NM_005926.2:c.88T>Cp.(Ser30Pro)		AD_denovo	6.8	het	de novo	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Intellectual disability, severe, Epileptiform EEG discharges, Neurodevelopmental delay, Epileptic encephalopathy, Myoclonic absences, EMG: myotonic discharges, Generalized tonic-clonic seizures
DPY19L4	NM_181787.2:c.1256C>Tp.(Ser419Phe)	NM_181787.2:c.1870C>Tp.(Arg624*)	AR_comphet	3.5	comphet	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Intellectual disability, severe, Epileptiform EEG discharges, Neurodevelopmental delay, Epileptic encephalopathy, Myoclonic absences, EMG: myotonic discharges, Generalized tonic-clonic seizures
AP3B2	NM_001278512.1:c.2879A>Gp.(Asn960Ser)	NM_001278512.1:c.2662G>Ap.(Glu888Lys)	AR_comphet	8.3	comphet	maternal&paternal	4	NDD + Epilepsy	Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Developmental regression, Intellectual disability, progressive, Intellectual disability, borderline, Intellectual disability, severe, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Delayed social development, Profound global developmental delay, Neurodevelopmental delay, Cognitive impairment
EIF3B	NM_001037283.1:c.2120G>Ap.(Arg707Gln)		AD_denovo	7.6	het	de novo	4	NDD + Epilepsy	Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Developmental regression, Intellectual disability, progressive, Intellectual disability, borderline, Intellectual disability, severe, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Delayed social development, Profound global developmental delay, Neurodevelopmental delay, Cognitive impairment
GABRE	NM_004961.3:c.41T>Cp.(Leu14Ser)		XL	5.2	hemi	maternal	1	NDD	Strabismus, Myopia, Autistic behavior, Anxiety, Hyperactivity, Intellectual disability, Muscular hypotonia, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Abnormal fear/anxiety-related behavior
PRRG3	NM_024082.3:c.572C>Tp.(Pro191Leu)		XL	3.5	hemi	maternal	4	NDD + Epilepsy	Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Developmental regression, Intellectual disability, progressive, Intellectual disability, borderline, Intellectual disability, severe, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Delayed social development, Profound global developmental delay, Neurodevelopmental delay, Cognitive impairment
USP20	NM_001008563.4:c.582delp.(Lys194Asnfs*46)		AR_homo	8.0	homo	maternal&paternal	3	NDD	Micropenis, Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Expressive language delay, Delayed fine motor development, Intellectual disability, severe
FAM171A1	NM_001010924.1:c.2435C>Tp.(Ala812Val)		AR_homo	4.8	homo	maternal&paternal	3	NDD	Micropenis, Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Expressive language delay, Delayed fine motor development, Intellectual disability, severe

LCN15	NM_203347.1:c.399C>Ap.(Ser133Arg)		AR_homo	3.8	homo	maternal&paternal	3	NDD	Micropenis, Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Expressive language delay, Delayed fine motor development, Intellectual disability, severe
ADAM11	NM_002390.5:c.98G>Tp.(Trp33Leu)		AD_denovo	6.6	het	de novo	1	NDD	Strabismus, Hypermetropia, Delayed speech and language development, Intellectual disability, Seizures, Global developmental delay, Absent speech, Absence seizures, Febrile seizures, Receptive language delay
HMGXB3	NM_014983.2:c.2026C>Tp.(Pro676Ser)		AD_denovo	6.1	het	de novo	1	NDD	Delayed speech and language development, Intellectual disability, Global developmental delay, Expressive language delay
RAB11FIP2	NM_001330167.1:c.1334T>Cp.(Met445Thr)		AD_denovo	5.9	het	de novo	1	NDD	kombinierte Entwicklungsverzögerung/Lernbehinderung (IQ=69), leichtes Übergewicht, faziale Dysmorphie, kurze Finger, Brachycephalus, CA und FRAX unauffällig, Array: Dup1q31.1 mat, Dup11q14.1 mat
KDM2B	NM_032590.4:c.2345C>Tp.(Ser782Leu)		AR_homo	7.1	homo	maternal&paternal	9	NDD	Urinary incontinence, Microcephaly, Micrognathia, Strabismus, Aggressive behavior, Inappropriate laughter, Paroxysmal bursts of laughter, Poor eye contact, Seizures, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Absent speech, Global brain atrophy, Sleep disturbance, Excessive salivation, Infantile muscular hypotonia
UNC5D	NM_080872.3: c.977A>Gp.(His326Arg)		AR_homo	6.5	homo	maternal&paternal	9	NDD	Urinary incontinence, Microcephaly, Micrognathia, Strabismus, Aggressive behavior, Inappropriate laughter, Paroxysmal bursts of laughter, Poor eye contact, Seizures, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Absent speech, Global brain atrophy, Sleep disturbance, Excessive salivation, Infantile muscular hypotonia
RNF10	NM_001330474.1:c.850C>Tp.(His284Tyr)		AR_homo	5.8	homo	maternal&paternal	9	NDD	Urinary incontinence, Microcephaly, Micrognathia, Strabismus, Aggressive behavior, Inappropriate laughter, Paroxysmal bursts of laughter, Poor eye contact, Seizures, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Absent speech, Global brain atrophy, Sleep disturbance, Excessive salivation, Infantile muscular hypotonia
PCLO	NM_033026.5 :c.13206G>T p.(Gln4402His)	NM_033026.5:c.1297G>Ap.(Ala433Thr)	AR_comphet	5.9	comphet	maternal&paternal	9	NDD	Urinary incontinence, Microcephaly, Micrognathia, Strabismus, Aggressive behavior, Inappropriate laughter, Paroxysmal bursts of laughter, Poor eye contact, Seizures, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Absent speech, Global brain atrophy, Sleep disturbance, Excessive salivation, Infantile muscular hypotonia
DOCK1	NM_001380.4:c.4546A>Gp.(Ser1516Gly)		AR_homo	6.3	homo	maternal&paternal	9	NDD	Urinary incontinence, Microcephaly, Micrognathia, Strabismus, Aggressive behavior, Inappropriate laughter, Paroxysmal bursts of laughter, Poor eye contact, Seizures, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Absent speech, Global brain atrophy, Sleep disturbance, Excessive salivation, Infantile muscular hypotonia
SF3B2	NM_006842.2:c.76G>Ap.(Ala26Thr)		AR_homo	5.1	homo	maternal&paternal	9	NDD	Urinary incontinence, Microcephaly, Micrognathia, Strabismus, Aggressive behavior, Inappropriate laughter, Paroxysmal bursts of laughter, Poor eye contact, Seizures, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Absent speech, Global brain atrophy, Sleep disturbance, Excessive salivation, Infantile muscular hypotonia
PCDHA9	NM_031857.1:c.1134_1135delCGinsTTp.(Ala379Ser)		AR_homo	4.6	homo	maternal&paternal	9	NDD	Urinary incontinence, Microcephaly, Micrognathia, Strabismus, Aggressive behavior, Inappropriate laughter, Paroxysmal bursts of laughter, Poor eye contact, Seizures, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Absent speech, Global brain atrophy, Sleep disturbance, Excessive salivation, Infantile muscular hypotonia
MRPL15	NM_014175.3:c.743C>Tp.(Thr248Ile)		AR_homo	6.0	homo	maternal&paternal	9	NDD	Urinary incontinence, Microcephaly, Micrognathia, Strabismus, Aggressive behavior, Inappropriate laughter, Paroxysmal bursts of laughter, Poor eye contact, Seizures, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Absent speech, Global brain atrophy, Sleep disturbance, Excessive salivation, Infantile muscular hypotonia
TCP11	NM_001093728.2:c.256A>Gp.(Lys86Glu)		AR_homo	3.4	homo	maternal&paternal	9	NDD	Urinary incontinence, Microcephaly, Micrognathia, Strabismus, Aggressive behavior, Inappropriate laughter, Paroxysmal bursts of laughter, Poor eye contact, Seizures, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Absent speech, Global brain atrophy, Sleep disturbance, Excessive salivation, Infantile muscular hypotonia

KDM2A	NM_012308.2:c.956G>Ap.(Arg319Gln)		AD_denovo	9.4	het	de novo	3	NDD + Epilepsy	Narrow mouth, Upslanted palpebral fissure, Delayed speech and language development, Intellectual disability, Global developmental delay, Pachygyria, Lissencephaly, Absent speech, Dysphagia, Polymicrogyria, Status epilepticus, Gliosis, Intellectual disability, moderate, Cerebellar malformation, Poor speech, Abnormality of the cerebral white matter, Excessive salivation, Focal white matter lesions, Focal seizures, Multifocal epileptiform discharges, Intellectual disability, severe, Epileptic spasms, EEG with focal epileptiform discharges, Cerebral white matter atrophy, Cerebral white matter agenesis, Oral-pharyngeal dysphagia
MARCHF6	NM_005885.3:c.1108T>Cp.(Tyr370His)	NM_005885.3:c.1897-3C>Tp.?	AR_comphet	4.3	comphet	maternal&paternal	1	NDD + Epilepsy	global development delay, seizures, microcephaly, autism, single transverse palmar crease, broad palm, abnormal fracial shape
RSRC2	NM_023012.5:c.603-8T>Cp.?		AD_denovo	4.0	het	de novo	3	NDD + Epilepsy	Behavioral abnormality, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Generalized tonic-clonic seizures, Absence seizures, Generalized myoclonic seizures, Intellectual disability, moderate, Generalized tonic seizures, Atonic seizures, Cognitive impairment
WDR59	NM_030581.3:c.2326G>Tp.(Val776Leu)	NM_030581.3:DelExons19-25	AR_comphet	4.4	comphet	maternal&paternal	3	NDD + Epilepsy	Behavioral abnormality, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Generalized tonic-clonic seizures, Absence seizures, Generalized myoclonic seizures, Intellectual disability, moderate, Generalized tonic seizures, Atonic seizures, Cognitive impairment
WWC3	NM_015691.3:c.2935C>Tp.(Arg979Trp)		XL	4.2	hemi	maternal	3	NDD + Epilepsy	Behavioral abnormality, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Generalized tonic-clonic seizures, Absence seizures, Generalized myoclonic seizures, Intellectual disability, moderate, Generalized tonic seizures, Atonic seizures, Cognitive impairment
ZMYM2	NM_001190964.2:c.2881G>Cp.(Glu961Gln)		AD_denovo	9.0	het	de novo	2	NDD + Epilepsy	Seizures, Global developmental delay, Episodic ataxia
OPCML	NM_001012393.2:c.175delp.(Val59Trpfs*4)		AD_denovo	7.1	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Generalized tonic-clonic seizures, Focal clonic seizures, Intellectual disability, moderate, Focal seizures with impairment of consciousness or awareness, Intellectual disability, borderline, Generalized tonic seizures, Symptomatic seizures, Focal tonic seizures, Cognitive impairment
PRKCA	NM_002737.2:c.64C>Tp.(Arg22Cys)		AD_denovo	8.6	het	de novo	4	NDD	Tall stature, Strabismus, Esotropia, Precocious puberty, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Curly hair, Scoliosis
SRRT	NM_015908.5:c.437C>Tp.(Pro146Leu)		AD_denovo	8.4	het	de novo	4	NDD	Tall stature, Strabismus, Esotropia, Precocious puberty, Intellectual disability, Muscular hypotonia, Intellectual disability, mild, Global developmental delay, Motor delay, Curly hair, Woolly hair, Intellectual disability, moderate, Scoliosis, Infantile muscular hypotonia, Precocious puberty in females, Proportionate tall stature, Cognitive impairment
KALRN	NM_001024660.4:c.4026-8T>Cp.?	NM_001024660.4:c.5369A>Gp.(Gln1790Arg)	AR_comphet	6.3	comphet	maternal&paternal	4	NDD	Tall stature, Strabismus, Esotropia, Precocious puberty, Intellectual disability, Muscular hypotonia, Intellectual disability, mild, Global developmental delay, Motor delay, Curly hair, Woolly hair, Intellectual disability, moderate, Scoliosis, Infantile muscular hypotonia, Precocious puberty in females, Proportionate tall stature, Cognitive impairment
TRMT1	NM_001136035.2:c.1964G>Ap.(Gly655Glu)		AD_denovo	8.0	het	de novo	4	NDD	Tall stature, Strabismus, Esotropia, Precocious puberty, Intellectual disability, Muscular hypotonia, Intellectual disability, mild, Global developmental delay, Motor delay, Curly hair, Woolly hair, Intellectual disability, moderate, Scoliosis, Infantile muscular hypotonia, Precocious puberty in females, Proportionate tall stature, Cognitive impairment
SLC2A8	NM_014580.4:c.1150G>Ap.(Gly384Ser)	NM_014580.4:c.1239C>Gp.(Cys413Trp)	AR_comphet	4.5	comphet		2	NDD + Epilepsy	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Absent speech, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Generalized seizures, Focal clonic seizures, Intellectual disability, moderate, Focal seizures with impairment of consciousness or awareness, Poor speech, Focal seizures, Intellectual disability, severe, Epileptic spasms, Focal motor seizures, Focal tonic seizures, Abnormality of movement, Cognitive impairment
GDF11	NM_005811.4:c.955dup, p.(Thr319Asnfs*5)		AD_denovo	8.9	het	de novo	3	NDD + Epilepsy	Delayed speech and language development, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Absence seizure, Typical absence seizure, Early onset absence seizures
STX1A	NM_004603.3:c.284-1G>Ap.?		AR_homo	12.9	homo	maternal&paternal	1	NDD	severe ID, decreased fetal movements, muscular hypotonia

TRAK2	NM_015049.2:c.1210G>Ap.(Val404Ile)		AD_denovo	6.9	het	de novo	1	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Intellectual disability, profound, Intellectual disability, moderate, Multifocal epileptiform discharges, Intellectual disability, severe, Epileptiform EEG discharges, Cognitive impairment, Epileptic encephalopathy
TENM1	NM_001163278.1:c.5977A>Tp.(Thr1993Ser)		XL	5.7	hemi	maternal	1	NDD	Delayed speech and language development, Intellectual disability, Global developmental delay
ACTR5	NM_024855.3:c.958G>Tp.(Asp320Tyr)		AR_homo	6.9	homo	maternal& paternal	3	NDD + Epilepsy	Delayed speech and language development, Seizures, Global developmental delay, Motor delay, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Febrile seizures, Postnatal microcephaly
TENM3	NM_001080477.3:c.2221G>Ap.(Glu741Lys)		AD_denovo	7.8	het	de novo	1	NDD	Autism, Autistic behavior, Intellectual disability, Global developmental delay, Intellectual disability, severe, no speech
ZMYM4	NM_005095.2:c.1300A>Gp.(Thr434Ala)		AD_denovo	6.6	het	de novo	1	NDD + Epilepsy	Seizures, Global developmental delay, Generalized tonic-clonic seizures with focal onset, Focal seizures, Epileptic encephalopathy
GALNT2	NM_004481.4:c.865C>Tp.Gln289*		AR_homo	9.4	homo	maternal& paternal	1	NDD + Epilepsy	very severe ID, seizures, autism, aggressive behavior, feeding problems in infancy, short stature, constipation, strabismus, inguinal hernia
MAGI2	NM_012301.3:c.3780C>Ap.Asp1260Glu		AR_homo	8.9	homo	maternal& paternal	1	NDD	mild ID, hypermetropia
SLC44A1	NM_080546.4:c.377_380delGTGAp.Ser126fs		AR_homo	9.1	homo	maternal& paternal	1	NDD	mild ID, macrocephaly, acanthosis nigricans, accessory mamilla, muscular hypotonia, frontotemporal cerebral atrophy
TRAP1	NM_016292.2:c.1941-1G>Ap.?		AR_homo	10.0	homo	maternal& paternal	1	NDD	moderate ID, mental deterioration, autism, self-mutilation, muscular hypotonia, nystagmus, leukodystrophy
CCAR2	NM_021174.5:c.2484C>Ap.Tyr828*		AR_homo	9.7	homo	maternal& paternal	2	NDD	moderate ID, small for gestational age, short stature
CLMN	NM_024734.3:c.730C>Tp.Arg244*		AR_homo	7.7	homo	maternal& paternal	1	NDD	moderate ID, muscular hypotonia, gait disturbance, EEG abnormalities, cerebral atrophy
ENO2	NM_001975.2:c.710C>Tp.Thr237Met		AR_homo	8.6	homo	maternal& paternal	1	NDD	mild ID, small for gestational age, short stature, microcephaly
AMZ2	NM_001033569.1:c.25C>Tp.Gln9*		AR_homo	7.4	homo	maternal& paternal	2	NDD	mild ID, muscular hypotonia, microcephaly, hypospadias, megalocornea, cerebral atrophy
ICE2/NARG2	NM_024611.5:c.2764G>Tp.Gly922*		AR_homo	9.4	homo	maternal& paternal	1	NDD + Epilepsy	mild ID, deafness, febrile seizures, EEG abnormalities, atrial septal defect
FAM234B	NM_020853.1:c.1009C>Tp.Gln337*		AR_homo	8.2	homo	maternal& paternal	1	NDD + Epilepsy	mild ID, seizures, obesity, delayed puberty
SEC23IP	NM_007190.3:c.2101G>Tp.Glu701*		AR_homo	8.5	homo	maternal& paternal	1	NDD	severe ID, feeding problems in infancy, microcephaly, non-midline cleft of the upper lip, 1-2 and 3-4 toe syndactyly, broad toes, mirror image duplication of toes, craniosynostosis, scaphocephaly, hypoplastic corpus callosum, holoprosencephaly, lissencephaly, leukodystrophy, central diabetes insipidus
SV2C	NM_014979.3:c.533G>Cp.Ser178Thr		AR_homo	7.0	homo	maternal& paternal	1	NDD	moderate ID, microcephaly, short stature
PPFIA1	NM_003626.3:c.1070A>Gp.His357Arg		AR_homo	7.9	homo	maternal& paternal	1	NDD	very severe ID, muscular hypotonia, spasticity, resting tremor, abnormality of the thorax, seizures, cerebral atrophy
LRRIQ3	NM_001105659.1:c.968C>Ap.Ser323*		AR_homo	7.1	homo	maternal& paternal	2	NDD	mild ID
INIP	NM_021218.2:c.266delCp.Ala89fs		AR_homo	9.2	homo	maternal& paternal	1	NDD + Epilepsy	mild ID, febrile seizures, recurrent infections, carious teeth, microcephaly, muscular hypotonia, ataxia, myopia
GTF3C3	NM_012086.4:c.1436A>Gp.Tyr479Cys		AR_homo	8.0	homo	maternal& paternal	1	NDD + Epilepsy	mild ID, seizures, recurrent infections, constipation, abnormalities of the face, postaxial hexadactyly, ataxia, radioulnar synostosis, ventricular septal defect, EEG abnormalities
MBNL3	NM_018388.3:c.279delTp.Ala94fs		AR_homo	9.0	hemi	maternal& paternal	1	NDD	moderate ID, autism
OGDHL	NM_018245.2:c.2606G>Ap.Arg869Gln		AR_homo	7.2	homo	maternal& paternal	2	NDD	moderate ID, small for gestational age, short stature
CACNA2D1	NM_000722.3:c.1514C>Tp.Thr505Ile		AR_homo	8.7	homo	maternal& paternal	1	NDD	severe ID, muscular hypotonia, stereotypical motor behaviors, inguinal hernia, omphalocele
TMEM132D	NM_133448.2:c.1489A>Gp.Lys497Glu		AR_homo	6.2	homo	maternal& paternal	2	NDD	mild ID
HACL1	NM_012260.3:c.1246C>Gp.His416Asp		AR_homo	7.2	homo	maternal& paternal	1	NDD	severe ID, muscular hypotonia, low-set ears, bifid uvula, cryptorchidism, aplasia cutis congenita, unilateral renal agenesis, cardiac malformation, increased creatine kinase
SPOUT1	NM_016390.3:c.1058C>Tp.Thr353Met		AR_homo	6.6	homo	maternal& paternal	1	NDD + Epilepsy	profound ID, seizures, microcephaly, short stature, limb hypertonia, bruxism

SMURF2	NM_022739.3:c.1921A>Gp.Thr641Ala		AR_homo	8.2	homo	maternal&paternal	2	NDD	mild ID, muscular hypotonia, microcephaly, hypospadias, megalocornea, cerebral atrophy
GRAMD1B	NM_001286563.1:c.586C>Tp.Arg196Trp		AR_homo	7.2	homo	maternal&paternal	1	NDD	moderate ID
PPRC1	NM_015062.4:c.1825C>Tp.Pro609Ser		AR_homo	6.2	homo	maternal&paternal	1	NDD + Epilepsy	severe ID, seizures, cerebral atrophy, leukodystrophy, macular degeneration, abnormality of the retina
BDH1	NM_004051.4:c.668G>Ap.Arg223His		AR_homo	7.2	homo	maternal&paternal	1	NDD + Epilepsy	very severe ID, seizures, muscular hypotonia, limb hypertonia, spasticity, short stature, microcephaly, leukodystrophy
CHD1L	NM_004284.4:c.1175G>Ap.Arg392His		AR_homo	9.0	homo	maternal&paternal	1	NDD	mild ID, microcephaly, muscular hypotonia, rigidity, ataxia, intention tremor, hypopigmented macules, EEG abnormalities
ATP2C2	NM_001286527.2:c.2636A>Gp.Asp879Gly		AR_homo	7.8	homo	maternal&paternal	1	NDD	severe ID, muscular hypotonia of the trunk, spastic paraparesis, preaxial polydactyly, abnormality of muscle fibers, colpocephaly, cerebellar hypoplasia, hypoplasia of the corpus callosum
PARD6A	NM_016948.2:c.934C>Tp.Arg312*		AD_denovo	6.2	het	de novo	1	NDD	mild ID, stereotypical motor behaviors, muscular hypotonia, strabismus, EEG abnormalities
HMG20A	NM_001304504.1:c.694C>Gp.Arg232Gly		AR_homo	6.6	homo	maternal&paternal	1	NDD + Epilepsy	moderate ID, seizures
TSPAN18	NM_130783.4:c.275T>Cp.Leu92Pro		AR_homo	6.4	homo	maternal&paternal	1	NDD	severe ID, deafness
CEP76	NM_024899.3:c.302T>Cp.Ile101Thr		AR_homo	7.6	homo	maternal&paternal	1	NDD	moderate ID, muscular hypotonia, short stature, microcephaly
ADIPOR1	NM_001290553.1:c.644T>Cp.Leu215Pro		AR_homo	6.9	homo	maternal&paternal	1	NDD	very severe ID, EEG abnormalities, microcephaly
TMEM147	NM_032635.3:c.344+5G>Ap.?		AR_homo	5.7	homo	maternal&paternal	1	NDD	very severe ID, impaired vision, joint contractures
GCC2	NM_181453.3:c.3982C>Tp.His1328Tyr		AR_homo	7.6	homo	maternal&paternal	1	NDD	ID, short stature, elbow contractures, wrist contractures, axillar pterygium, abnormalities of the face, deafness, abnormality of thrombocytes
SKIDA1	NM_207371.3:c.2600C>Tp.Ala867Val		AR_homo	6.5	homo	maternal&paternal	1	NDD	severe ID, small for gestational age, strabismus, short stature
LRCH3	NM_032773.3:c.761A>Gp.Gln254Arg		AR_homo	5.8	homo	maternal&paternal	1	NDD + Epilepsy	severe ID, seizures, muscular hypotonia, cardiac malformation, cerebral atrophy
RXRB	NM_001270401.1:c.1091C>Tp.Pro364Leu		AR_homo	7.1	homo	maternal&paternal	1	NDD	very severe ID, short stature, microcephaly
BTN2A2	NM_001197237.1:c.386G>Ap.Cys129Tyr		AR_homo	6.3	homo	maternal&paternal	1	NDD	very severe ID, muscular hypotonia, constipation
LENG8	NM_052925.3:c.2147G>Ap.Arg716Gln		AR_homo	6.2	homo	maternal&paternal	1	NDD	severe ID, mental deterioration, sleep disturbances, behavioral abnormality, hyperpigmented macules, EEG abnormalities
FNDC3A	NM_001079673.1:c.1186G>Ap.Asp396Asn		AR_homo	7.1	homo	maternal&paternal	1	NDD + Epilepsy	severe ID, seizures, muscular hypotonia, short stature
KCTD18	NM_001321547.1:c.875C>Tp.Ser292Leu		AR_homo	5.5	homo	maternal&paternal	1	NDD	moderate ID, short stature, microcephaly, dislocated hips
EIF4A2	NM_001967.3:c.109_111delGATp.Asp37del		AR_homo	7.6	homo	maternal&paternal	1	NDD	mild ID, muscular hypotonia, tremor
RSRC2	NM_023012.5:c.1271T>Gp.(Phe424Cys)		AD_denovo	6.1	het	de novo	2	NDD	Global developmental delay, Microcephaly, Agenesis of corpus callosum, Failure to thrive, Growth delay, EEG abnormality, Abnormal cry
TTBK1	NM_032538.2:c.3116_3118delp.(Thr1039del)		AD_inherited	4.4	het	paternal	2	NDD	Tall stature, Behavioral abnormality, Short attention span, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Attention deficit hyperactivity disorder, Cognitive impairment
SNF8	NM_007241.3:c.572G>Ap.(Gly191Asp)	NM_007241.3:c.236C>Tp.(Pro79Leu)	AR_comphet	5.0	comphet	maternal&paternal	2	NDD	Global developmental delay, Microcephaly, Agenesis of corpus callosum, Failure to thrive, Growth delay, EEG abnormality, Abnormal cry
ARL13A	NM_001162491.1:c.349G>Cp.(Asp117His)		XL	3.3	hemi	maternal	1	NDD	Intellectual disability, Global developmental delay, Hemiplegia/hemiparesis
TMEM94	NM_001321148.1:c.2906G>Ap.(Arg969Gln)	NM_001321148.1:c.2978T>Cp.(Met993Thr)	AR_comphet	6.2	comphet	maternal&paternal	1	NDD + Epilepsy	Seizures, Global developmental delay, Focal seizures, Retinoblastoma

AFDN	NM_001207008.1:c.436A>Gp.(Lys146Glu)		AD_inherited	6.0	het	paternal	2	NDD	Tall stature, Behavioral abnormality, Short attention span, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Attention deficit hyperactivity disorder, Cognitive impairment
GEMIN5	NM_015465.4:c.1627A>Gp.(Ser543Gly)	NM_015465.4:c.851G>Ap.(Arg284His)	AR_comphet	5.3	comphet	maternal&paternal	2	NDD	Cryptorchidism, Microcephaly, Global developmental delay, Motor delay, Growth delay, Intrauterine growth retardation
ARFGEF3	NM_020340.4:c.421-4A>Gp.?	NM_020340.4:c.2003C>Tp.(Ala668Val)	AR_comphet	5.0	comphet	maternal&paternal	2	Neuro	Abnormality of the corpus callosum, Agenesis of corpus callosum, Talipes equinovarus, Polymicrogyria, Myelomeningocele, Brainstem dysplasia, Dysplastic corpus callosum, Periventricular gray matter heterotopia
COL19A1	NM_001858.5:c.1843G>Ap.(Gly615Ser)		AR_homo	4.7	homo	maternal&paternal	3	ndd	Microcephaly, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe
SLC25A35	NM_001320870.1:c.194G>Ap.(Gly65Asp)		AR_homo	4.7	homo	maternal&paternal	3	ndd	Microcephaly, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe
GRIK5	NM_001301030.1:c.818C>Ap.(Ser273Tyr)	NM_001301030.1:c.1745G>Ap.(Arg582His)	AR_comphet	8.6	comphet	maternal&paternal	2	NDD + Epilepsy	Strabismus, Single umbilical artery, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Spastic tetraparesis, Absent speech, Generalized myoclonic seizures, Polymicrogyria, Tetraparesis, Intellectual disability, moderate, EEG abnormality, Sleep disturbance, Myoclonic spasms, Unilateral polymicrogyria, Frontoparietal polymicrogyria, Generalized tonic seizures, Epileptic spasms, Focal myoclonic seizures, EEG with generalized spikes, Perisylvian polymicrogyria, Tetraplegia/tetraparesis, Cognitive impairment, Maternal seizures, Abnormal eating behavior, Exodeviation, Segmental myoclonic seizures
SLC25A43	NM_145305.2:c.224C>Tp.(Ala75Val)		XL	6.3	hemi	maternal	2	NDD	Cryptorchidism, Microcephaly, Global developmental delay, Motor delay, Growth delay, Intrauterine growth retardation
TRIM9	NM_015163.5:c.1117G>Ap.(Val373Met)		AD_denovo	8.3	het	de novo	1	NDD + Epilepsy	Intellectual disability, Seizures, Muscular hypotonia, Global developmental delay, Mental deterioration, Pes cavus, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Generalized seizures,Leukodystrophy, Abnormality of the cerebral white matter, Infantile spasms
TAOK1	NM_020791.2:c.332C>Tp.(Ser111Phe)		AD_denovo	8.8	het	de novo	1	NDD	Dysmorphic syndrome, cleft lip and palate, failure to thrive, macrocephaly, muscular hypotonia, developmental delay
LAMA5	NM_005560.4:c.6659G>Tp.(Arg2220Leu)	NM_005560.4:c.1246C>Gp.(Pro416Ala)	AR_comphet	5.3	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Global developmental delay, Episodic ataxia
AGO2	NM_001164623.1:c.602G>Tp.(Gly201Val)		AD_denovo	8.8	het	de novo	1	NDD + Epilepsy	Intellectual disability, Global developmental delay, Motor delay, Gait disturbance, Absent speech, Bicuspid aortic valve, Patent foramen ovale, Atrioventricular block, Intellectual disability, moderate, Poor speech, Obstructive sleep apnea, Short stature, Sleep apnea, Intellectual disability, severe, Epileptic spasms, Epileptic encephalopathy
CDH20	NM_031891.3:c.958G>Cp.(Asp320His)		AD_denovo	6.5	het	de novo	5	NDD + Epilepsy	Tall stature, Autism, Precocious puberty, Intellectual disability, Seizures, Global developmental delay, Focal seizures with impairment of consciousness or awareness, Focal seizures, Precocious puberty in males, Increased serum insulin-like growth factor 1
FUNDC1	NM_173794.3:c.154A>Gp.(Thr52Ala)		XL	6.8	hemi	maternal	5	NDD + Epilepsy	Tall stature, Autism, Precocious puberty, Intellectual disability, Seizures, Global developmental delay, Focal seizures with impairment of consciousness or awareness, Focal seizures, Precocious puberty in males, Increased serum insulin-like growth factor 1
DDB1	NM_001923.4:c.563G>Ap.(Arg188Gln)		AD_denovo	8.8	het	de novo	1	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Status epilepticus, Intellectual disability, severe, Epileptiform EEG discharges, EEG with focal sharp slow waves, EEG with generalized sharp slow waves, EEG with occipital sharp slow waves, EEG with parietal sharp slow waves, EEG with temporal sharp slow waves, EEG with frontal sharp slow waves, EEG with central sharp slow waves, EEG with occipital sharp waves, EEG with parietal sharp waves
SPSB1	NM_025106.3:c.572T>Cp.(Ile191Thr)		AD_denovo	6.5	het	de novo	5	NDD + Epilepsy	Tall stature, Autism, Precocious puberty, Intellectual disability, Seizures, Global developmental delay, Focal seizures with impairment of consciousness or awareness, Focal seizures, Precocious puberty in males, Increased serum insulin-like growth factor 1

PRSS41	NM_001135086.1:c.30_41dup, p.(Leu11_Ala14dup)		AR_homo	3.0	homo	maternal& paternal	5	NDD + Epilepsy	Tall stature, Autism, Precocious puberty, Intellectual disability, Seizures, Global developmental delay, Focal seizures with impairment of consciousness or awareness, Focal seizures, Precocious puberty in males, Increased serum insulin-like growth factor 1
RNF44	NM_014901.4:c.802-8T>Gp.?		AD_denovo	4.9	het	de novo	5	NDD + Epilepsy	Tall stature, Autism, Precocious puberty, Intellectual disability, Seizures, Global developmental delay, Focal seizures with impairment of consciousness or awareness, Focal seizures, Precocious puberty in males, Increased serum insulin-like growth factor 1
MINPP1	NM_004897.4:c.75_94delp.(Leu27Argfs*39)		AR_homo	9.2	homo	maternal& paternal	2	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Ataxia, Global developmental delay, Gait ataxia, Olivopontocerebellar atrophy, Short stature, Pontocerebellar atrophy, Olivopontocerebellar hypoplasia, Cognitive impairment
LAMA5	NM_005560.4:c.10753G>Tp.(Asp3585Tyr)	NM_005560.4:c.1390G>Ap.(Gly464Ser)	AR_comphet	5.7	comphet	maternal& paternal	1	NDD + Epilepsy	Abnormality of the head, Microcephaly, Seizures, Postnatal microcephaly, Loss of consciousness, Atonic seizures
CRYBG1	NM_001624.3:c.4489G>Ap.(Val149Ile)		AD_denovo	6.1	het	de novo	1	NDD	Hearing impairment, Prelingual sensorineural hearing impairment, Conductive hearing impairment, Hypermetropia, Nystagmus, Horizontal nystagmus, Intellectual disability, Motor delay, Growth delay, Generalized tonic-clonic seizures, Mild short stature, Proportionate short stature, Decreased body weight, High hypermetropia, Simple febrile seizures
GRK3	NM_005160.3:c.916G>Tp.(Glu306*)		AD_inherited	6.1	het	maternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Neurodevelopmental delay, Cognitive impairment, Epileptic encephalopathy
TENM1	NM_001163278.1:c.757A>Gp.(Asn253Asp)		XL	5.5	het	maternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Neurodevelopmental delay, Cognitive impairment, Epileptic encephalopathy
DNAJC27	NM_016544.2:c.422delp.(His141Leufs*4)		AD_inherited	5.7	het	maternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Mild global developmental delay, Moderate global developmental delay, Severe global developmental delay, Neurodevelopmental delay, Cognitive impairment, Epileptic encephalopathy
GUCY2F	NM_001522.2:c.1445C>Gp.(Ser482Cys)		AR_homo	5.1	homo	maternal& paternal	2	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Ataxia, Global developmental delay, Gait ataxia, Olivopontocerebellar atrophy, Short stature, Pontocerebellar atrophy, Olivopontocerebellar hypoplasia, Cognitive impairment
ANKRD30B	NM_001145029.1:c.1795G>Tp.(Glu599*)		AR_homo	6.2	homo	maternal& paternal	1	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Global developmental delay
UNC5A	NM_133369.2:c.578C>Ap.(Ser193Tyr)	NM_133369.2:c.267C>Gp.(Ile89Met)	AR_comphet	4.7	comphet	maternal& paternal	1	NDD + Epilepsy	Hypermetropia, Intellectual disability, Seizures, Global developmental delay, Absence seizure, Intellectual disability, severe, Moderate global developmental delay, Severe global developmental delay, Profound global developmental delay, Cognitive impairment
PIKFYVE	NM_015040.3:c.1319A>Gp.(Gln440Arg)		AR_homo	9.0	homo	maternal& paternal	4	NDD	Microcephaly, Autistic behavior, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe
GEMIN5	NM_015465.4:c.3340C>Gp.(Leu1114Val)	NM_015465.4:c.2504A>Gp.(Lys835Arg)	AR_comphet	6.6	comphet	maternal& paternal	3	NDD + Epilepsy	Delayed speech and language development, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Absence seizure, Typical absence seizure, Early onset absence seizures
VPS54	NM_016516.2:c.701C>Tp.(Ala234Val)		AR_homo	8.2	homo	maternal& paternal	4	NDD	Microcephaly, Autistic behavior, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe
BCAS1	NM_003657.3:c.1720C>Tp.(Pro574Ser)		AR_homo	6.6	homo	maternal& paternal	4	NDD	Microcephaly, Autistic behavior, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe
LRIG3	NM_153377.4:c.979G>Ap.(Asp327Asn)		AD_denovo	6.7	het	de novo	1	NDD	Global developmental delay, Absent speech, Myelomeningocele

COPS2	NM_001143887.1:c.37G>Ap.(Glu13Lys)		AD_denovo	8.6	het	de novo	3	NDD + Epilepsy	Delayed speech and language development, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Absence seizure, Typical absence seizure, Early onset absence seizures
ATP2B1	NM_001001323.1:c.1376A>Gp.(His459Arg)		AD_denovo	8.8	het	de novo	3	NDD + Epilepsy	Autism, Intellectual disability, Seizures, Global developmental delay, Poor speech, Focal seizures
CD99L2	NM_001242614.1:c.541G>Cp.(Gly181Arg)		XL	3.9	hemi	maternal	1	NDD + Epilepsy	Tall stature, Glaucoma, Growth hormone excess, Intellectual disability, Seizures, Global developmental delay, Obesity, Mitral regurgitation, Abnormal facial shape, Progeroid facial appearance, Focal-onset seizure
RHEB	NM_005614.3:c.47C>Tp.(Ser16Phe)		AD_denovo	7.9	het	de novo	1	NDD + Epilepsy	Tall stature, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, normal MRI
PSMC5	NM_002805.5:c.587delp.(Lys196Argfs*29)		AD_inherited	8.4	het	maternal	2	NDD	Microcephaly, Intellectual disability, Muscular hypotonia, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Intellectual disability, moderate, Scoliosis, Short stature, Cognitive impairment
NOVA2	NM_002516.3:c.1267G>Cp.(Gly423Arg)		AD_inherited	5.5	het	maternal	2	NDD	Microcephaly, Intellectual disability, Muscular hypotonia, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Intellectual disability, moderate, Scoliosis, Short stature, Cognitive impairment
PTPRN2	Del(NM_002847.4)-7-157873875-158384503		AD_denovo	6.7	het	de novo	1	NDD	Behavioral abnormality, Autism, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Intellectual disability, moderate, Poor speech, Intellectual disability, borderline
LCN1	NM_001252618.1:c.305A>Gp.(His102Arg)		AD_denovo	3.4	het	de novo	1	NDD	Tall stature, delayed speech and language development, neuroblastoma
ORC3	NM_181837.2:c.419A>Gp.(Asp140Gly)		AR_homo	6.7	homo	maternal&paternal	4	NDD	Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Severe muscular hypotonia, Muscular hypotonia of the trunk, Infantile muscular hypotonia, Intellectual disability, severe, Central hypotonia, Cognitive impairment
SRRM4	NM_194286.3:c.560G>Ap.(Arg187His)	NM_194286.3:c.140C>Tp.(Pro47Leu)	AR_comphet	5.1	comphet	maternal&paternal	1	NDD	Microcephaly, Brachydactyly, Syndactyly, Intellectual disability, Intellectual disability, mild, Motor delay, Hypertonia, Toe syndactyly, Intellectual disability, moderate, 2-3 toe syndactyly, Feeding difficulties, Cognitive impairment, Impaired feeding ability
ALDH8A1	NM_022568.3:c.160G>Tp.(Ala54Ser)		AD_denovo	5.6	het	de novo	1	NDD	Macrocephaly, Global developmental delay, Hepatosplenomegaly, Hypertriglyceridemia, Hepatomegaly, Recurrent infections
PLCH2	NM_014638.3:c.595C>Tp.(His199Tyr)		AD_inherited	2.5	het	paternal	2	NDD	Intellectual disability
FEN1	NM_004111.5:c.140G>Ap.(Arg47His)		AR_homo	6.5	homo	maternal&paternal	1	NDD + Epilepsy	Seizures, Focal impaired awareness seizure, Spherocytosis, Arrhythmia
CX3CR1	NM_001171174.1:c.756delp.(Cys253Alafs*12)		AD_inherited	6.0	het	maternal	1	Neuro	Familial predisposition, Migraine, EEG abnormality, Episodic hemiplegia, Left hemiplegia
TMEM151B	NM_001137560.1:c.1319T>Ap.(Val440Asp)		AD_denovo	6.3	het	de novo	1	NDD + Epilepsy	Cleft soft palate, Hydrocephalus, Abnormality of the inner ear, Hearing impairment, Iris coloboma, Delayed speech and language development, Macular coloboma, Intellectual disability, Seizures, Global developmental delay, Agenesis of corpus callosum, Dandy-Walker malformation, Abnormal ear morphology
FAM214B	NM_001317991.1:c.588delp.(Ile196Metfs*115)		AD_inherited	6.5	het	paternal	2	NDD	Intellectual disability
SENP3	NM_015670.5:c.713C>Ap.(Ser238*)		AD_denovo	8.7	het	de novo	3	NDD + Epilepsy	epilepsy with absences and generalized tonic-clonic seizures, severe intellectual disability with autistic traits, low blood pressure, obstipation, normal MRI 2008
BDP1	NM_018429.2:c.6847G>Tp.(Glu2283*)		AR_homo	9.4	homo	maternal&paternal	3	NDD + Epilepsy	Delayed speech and language development, Seizures, Global developmental delay, Motor delay, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Febrile seizures, Postnatal microcephaly, suspected myopia
CHD5	NM_015557.2:c.776C>Gp.(Ser259Cys)	NM_015557.2:c.3650C>Tp.(Thr1217Ile)	AR_comphet	5.8	comphet	maternal&paternal	2	NDD	Delayed speech and language development, Intellectual disability
DENND4B	NM_014856.2:c.319G>Ap.(Val107Met)	NM_014856.2:c.941G>Ap.(Ser314Asn)	AR_comphet	4.3	comphet	maternal&paternal	2	NDD	Delayed speech and language development, Intellectual disability

RHOT2	NM_138769.2:c.586T>Gp.(Ser196Ala)	NM_138769.2:c.1201C>Tp.(Arg401Cys)	AR_comphet	4.9	comphet	maternal&paternal	1	NDD + Epilepsy	spastic tetraparesis, generalized tonic-clonic seizures, microcephaly, polymicrogyria, periventricular gliosis and cysts, global developmental delay
CAPN9	NM_006615.2:c.1591G>Ap.(Ala531Thr)	NM_006615.2:c.1273-1_1287delp.(Cys425Glufs*262)	AR_comphet	7.4	comphet	maternal&paternal	1	NDD	Global developmental delay, Motor delay, Polyneuropathy, Hip dysplasia, Coxa valga, Kyphosis
KCNJ4	NM_020452.3:c.1745G>Ap.(Arg582Gln)		AD_denovo	6.4	het	de novo	2	NDD	Intellectual disability, Intellectual disability, mild, Intellectual disability, moderate, Increased body weight, Increased adipose tissue
DIP2A	NM_015151.3:c.410C>Tp.(Ser137Leu)	NM_015151.3:c.2476G>Ap.(Ala826Thr)	AR_comphet	6.0	comphet	maternal&paternal	2	NDD	Intellectual disability, Intellectual disability, mild, Intellectual disability, moderate, Increased body weight, Increased adipose tissue
AKAP13	NM_006738.5:c.742C>Tp.(Arg248*)		AD_denovo	9.9	het	de novo	1	NDD + Epilepsy	Seizures, Global developmental delay, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Generalized tonic seizures, Epileptic encephalopathy
AKAP17A	NM_005088.2:c.1328T>Cp.(Leu443Pro)		AD_denovo	4.9	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Generalized tonic-clonic seizures, Focal clonic seizures, Intellectual disability, moderate, Increased body weight, Focal-onset seizure, Increased adipose tissue, Generalized tonic seizures, Focal myoclonic seizures, Focal tonic seizures
UTP11	NM_016037.3:c.230A>Gp.(Asp77Gly)		AD_denovo	5.2	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Generalized tonic-clonic seizures, Focal clonic seizures, Intellectual disability, moderate, Increased body weight, Focal-onset seizure, Increased adipose tissue, Generalized tonic seizures, Focal myoclonic seizures, Focal tonic seizures
GPSM3	NM_001276501.1:c.318G>Cp.(Gln106His)		AD_denovo	4.7	het	de novo	2	Neuro	Microcephaly, Edema, Agenesis of corpus callosum, Abnormal cerebellum morphology, Cerebellar hypoplasia, Growth abnormality, Growth delay, Intrauterine growth retardation, Hypoplasia of the corpus callosum, Polymicrogyria, Abnormality of neuronal migration, Gray matter heterotopias, Gray matter heterotopia, Spontaneous abortion, Periventricular heterotopia, White matter neuronal heterotopia, Aplasia/Hypoplasia of the cerebellum, Fetal onset, Small cerebellar cortex
EMC9	NM_016049.3:c.158A>Tp.(His53Leu)		AD_denovo	5.0	het	de novo	2	Neuro	Microcephaly, Edema, Agenesis of corpus callosum, Abnormal cerebellum morphology, Cerebellar hypoplasia, Growth abnormality, Growth delay, Intrauterine growth retardation, Hypoplasia of the corpus callosum, Polymicrogyria, Abnormality of neuronal migration, Gray matter heterotopias, Gray matter heterotopia, Spontaneous abortion, Periventricular heterotopia, White matter neuronal heterotopia, Aplasia/Hypoplasia of the cerebellum, Fetal onset, Small cerebellar cortex
SLC4A7	NM_001321103.1:c.249_252delp.(Lys83Asnfs*62)		AR_homo	8.2	homo	maternal&paternal	4	NDD	Adducted thumb, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Pyloric stenosis, Ventriculomegaly, Intellectual disability, moderate, Infantile muscular hypotonia, Feeding difficulties, Cognitive impairment, Impaired feeding ability
SCRN1	NM_001145514.1:c.1106A>Gp.(Lys369Arg)		AR_homo	5.9	homo	maternal&paternal	4	NDD	Adducted thumb, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Pyloric stenosis, Ventriculomegaly, Intellectual disability, moderate, Infantile muscular hypotonia, Feeding difficulties, Cognitive impairment, Impaired feeding ability, VUS in COLQ (31.07.2019)
COL20A1	NM_020882.2:c.3614-8C>Tp.?		AD_denovo	3.9	het	de novo	4	NDD	Adducted thumb, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Pyloric stenosis, Ventriculomegaly, Intellectual disability, moderate, Infantile muscular hypotonia, Feeding difficulties, Cognitive impairment, Impaired feeding ability, VUS in COLQ (31.07.2019)
WTAP	NM_001270531.1:c.463A>Gp.(Lys155Glu)		AD_denovo	6.9	het	de novo	1	NDD	Microcephaly, Hyperactivity, Global developmental delay, dystrophy

GPR161	NM_001267609.1:c.1550dup, p.(Gly518Argfs*44)		AD_denovo	7.1	het	de novo	2	NDD	Hypertelorism, Low-set ears, Brachydactyly, Intellectual disability, Global developmental delay, Hypoplasia of the corpus callosum, Elevated serum creatinine, Moderate global developmental delay
TENM2	NM_001122679.1:c.4082A>Gp.(Tyr1361Cys)	NM_001122679.1:c.7924G>Ap.(Val2642Met)	AR_comphet	5.0	comphet	maternal&paternal	2	NDD	Hypertelorism, Low-set ears, Brachydactyly, Intellectual disability, Global developmental delay, Hypoplasia of the corpus callosum, Elevated serum creatinine, Moderate global developmental delay
H3-3A	NM_002107.4:c.250C>Gp.(Arg84Gly)		AD_denovo	9.8	het	de novo	2	NDD + Epilepsy	Stereotypy, Delayed speech and language development, Global developmental delay, Motor delay, Delayed gross motor development, EEG abnormality, Delayed fine motor development
CHURC1	NM_145165.3:c.349_350insGp.(Leu117Argfs*15)	NM_145165.3:c.400delp.(Arg134Aspfs*3)	AR_comphet	7.8	comphet	maternal&paternal	2	NDD + Epilepsy	Tall stature, Macrocephaly, Delayed speech and language development, Enuresis, Seizures, Global developmental delay, Obesity, Rett syndrome
RGL1	NM_015149.4:c.737C>Gp.(Ser246Cys)		AD_denovo	6.4	het	de novo	2	NDD + Epilepsy	Tall stature, Macrocephaly, Delayed speech and language development, Enuresis, Seizures, Global developmental delay, Obesity, Rett syndrome
USF3	NM_001009899.3:c.1750C>Tp.(Gln584*)		AD_denovo	8.6	het	de novo	1	NDD	muscular hypotonia, developmental delay, normal cMRI, left retinal coloboma
EFHC1	NM_018100.3:c.323delp.(Pro108Leufs*13)		AR_homo	9.9	homo	maternal&paternal	3	NDD	Microcephaly, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe
WWP2	NM_001270453.1:c.491A>Cp.(Glu164Ala)	NM_001270453.1:c.166G>Cp.(Ala56Pro)	AR_comphet	4.6	comphet	maternal&paternal	2	NDD + Epilepsy	Strabismus, Single umbilical artery, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Spastic tetraparesis, Absent speech, Generalized myoclonic seizures, Polymicrogyria, Tetraparesis, Intellectual disability, moderate, EEG abnormality, Sleep disturbance, Myoclonic spasms, Unilateral polymicrogyria, Frontoparietal polymicrogyria, Generalized tonic seizures, Epileptic spasms, Focal myoclonic seizures, EEG with generalized spikes, Perisylvian polymicrogyria, Tetraplegia/tetraparesis, Cognitive impairment, Maternal seizures, Abnormal eating behavior, Exodeviation, Segmental myoclonic seizures
CTBP2	NM_022802.2:c.1192dup, p.(Arg398Profs*68)		AD_inherited	7.9	het	maternal	3	NDD + Epilepsy	Intellectual disability, epilepsy with generalized tonic-clonic seizures, short attention span
SLIT3	NM_003062.3:c.2818C>Tp.(Arg940Cys)		AD_inherited	5.1	het	maternal	3	NDD + Epilepsy	Intellectual disability, epilepsy with generalized tonic-clonic seizures, short attention span
CHKA	NM_001277.2:c.1021T>Cp.(Phe341Leu)	NM_001277.2:c.14dup, p.(Cys6Leufs*19)	AR_comphet	7.0	comphet	maternal&paternal	1	NDD + Epilepsy	severe psychomotor retardation, central movement disorder with preference for right-sided extremities, epilepsy with epileptic spasms, microcephaly, tendency to self-harm
CLCC1	NM_001048210.2:c.1324C>Tp.(Leu442Phe)		AD_inherited	4.0	het	maternal	3	NDD + Epilepsy	Intellectual disability, epilepsy with generalized tonic-clonic seizures, short attention span
ABCA2	NM_001606.4:c.2261T>Cp.(Phe754Ser)		AD_denovo	9.0	het	de novo	3	NDD + Epilepsy	epilepsy, febrile seizures
SF3A3	NM_006802.3:c.1408C>Tp.(Arg470*)		AD_denovo	8.9	het	de novo	3	NDD + Epilepsy	epilepsy with febrile seizures and dyscognitive seizures
NLE1	NM_018096.4:c.593A>Gp.(His198Arg)		AD_denovo	6.7	het	de novo	3	NDD + Epilepsy	epilepsy, febrile seizures
FRYL	NM_015030.1:c.3851T>Gp.(Leu1284Arg)		AR_homo	6.3	homo	maternal&paternal	1	NDD	Cryptorchidism, Hydroureter, Cleft palate, Cleft soft palate, Global developmental delay, Absent septum pellucidum, Polyhydramnios, Premature birth, Abnormal facial shape, Ventriculomegaly, Severe short stature, Short stature, Frontal cortical atrophy, Temporal cortical atrophy, Bilateral cryptorchidism, Moderately short stature, Brain atrophy
ADAMTSL1	NM_001040272.5:c.1316A>Gp.(Lys439Arg)		AD_denovo	5.8	het	de novo	1	NDD + Epilepsy	Global developmental delay, dystonic movements, abnormal EEG, epilepsy, microcephaly, clinodactyly of the 5th finger, pectus excavatum
STARD9	NM_020759.2:c.4624C>Ap.(Leu1542Met)	NM_020759.2:c.1655G>Tp.(Arg552Leu)	AR_comphet	3.3	comphet	maternal&paternal	2	NDD	Seizures, Generalized tonic-clonic seizures, Myoclonic atonic seizures, Epileptic encephalopathy
CRIM1	NM_016441.2:c.2867C>Tp.(Ala956Val)	NM_016441.2:c.1658+4C>Tp.?	AR_comphet	3.5	comphet	maternal&paternal	2	Neuro	Dystonia, Flexion contracture, Difficulty walking, Limb dystonia, Progressive inability to walk, Ankle flexion contracture, Loss of ability to walk in first decade, Inability to walk by childhood/adolescence, Loss of ability to walk, Generalized dystonia

PASK	NM_001252120.1:c.307G>Ap.(Gly103Ser)	NM_001252120.1:c.2713C>Tp.(Arg905Trp)	AR_comphet	4.1	comphet	maternal&paternal	2	Neuro	Dystonia, Flexion contracture, Difficulty walking, Limb dystonia, Progressive inability to walk, Ankle flexion contracture, Loss of ability to walk in first decade, Inability to walk by childhood/adolescence, Loss of ability to walk, Generalized dystonia
RASGEF1A	NM_001282862.1:c.346-3delp.?		AD_denovo	C	het	de novo	1	Fehlbildungen	Meningocele, Hypoplastic nasal bridge, Increased nuchal translucency, Short fetal femur length
ARL8B	NM_018184.2:c.286A>Tp.(Ile96Leu)		AD_denovo	6.3	het	de novo	2	Neuro	Abnormality of the corpus callosum, Agenesis of corpus callosum, Talipes equinovarus, Polymicrogyria, Myelomeningocele, Brainstem dysplasia, Dysplastic corpus callosum, Periventricular gray matter heterotopia
ITPKA	NM_002220.2:c.1093G>Ap.(Gly365Arg)		AR_homo	5.6	homo	maternal&paternal	2	NDD + Epilepsy	Macrocephaly, Seizures, Global developmental delay, Epileptic encephalopathy
UNC13A	NM_001080421.2:c.2786G>Ap.(Gly929Glu)		AD_denovo	8.4	het	de novo	3	NDD	Agitation, Aggressive behavior, Delayed speech and language development, Intellectual disability
OS9	NM_006812.3:c.1181A>Tp.(Glu394Val)		AR_homo	6.7	homo	maternal&paternal	2	NDD + Epilepsy	Macrocephaly, Seizures, Global developmental delay, Epileptic encephalopathy
DMAP1	NM_001034023.1:c.581G>Ap.(Arg194Gln)	NM_001034023.1:c.670C>Tp.(Arg224*)	AR_comphet	6.8	comphet	maternal&paternal	2	NDD	Microcephaly, Intellectual disability, Global developmental delay, Abnormal facial shape, Cognitive impairment
RAPGEF3	NM_001098531.2:c.2312A>Cp.(His771Pro)		AD_denovo	6.6	het	de novo	2	NDD	Microcephaly, Intellectual disability, Global developmental delay, Abnormal facial shape, Cognitive impairment
STT3B	NM_178862.2:c.777+4A>Cp.?		AD_denovo	7.7	het	de novo	3	NDD	Hypotelorism, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Intellectual disability, moderate, Single median maxillary incisor, Agenesis of permanent teeth, Abnormality of dental morphology, Reduced number of teeth, Intellectual disability, severe
ARSH	NM_001011719.1:c.339A>Gp.(Ile113Met)		AD_denovo	5.9	het	de novo	3	NDD	Hypotelorism, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Intellectual disability, moderate, Single median maxillary incisor, Agenesis of permanent teeth, Abnormality of dental morphology, Reduced number of teeth, Intellectual disability, severe
KANSL2	NM_017822.3:c.880C>Tp.(His294Tyr)		AD_unknown	3.7	het	unknown	1	NDD	Intellectual disability, Intellectual disability, moderate, Intellectual disability, severe
ZFP91	NM_053023.4:c.172C>Tp.(Arg58Trp)		AD_denovo	5.2	het	de novo	3	NDD + Epilepsy	neonatal epileptic encephalopathy
VPS52	NM_022553.5:c.10G>Ap.(Ala4Thr)	NM_022553.5:c.92C>Tp.(Ala31Val)	AR_comphet	5.4	comphet	maternal&paternal	3	NDD + Epilepsy	neonatal epileptic encephalopathy
EVI5L	NM_001159944.2:c.841delp.(Leu281fs)		AD_denovo	9.2	het	de novo	2	NDD + Epilepsy	Pallister Hall syndrome with hand and foot polydactyly, cleft palate, seizures, global developmental delay, 4-5 finger syndactyly, hamartoma
HCK	NM_002110.3:c.1547C>Ap.(Thr516Lys)		AD_denovo	6.1	het	de novo	2	NDD + Epilepsy	Pallister Hall syndrome with hand and foot polydactyly, cleft palate, seizures, global developmental delay, 4-5 finger syndactyly, hamartoma
ZFYVE16	NM_014733.4:c.2570C>Tp.(Pro857Leu)		AR_homo	5.5	homo	maternal&paternal	2	NDD + Epilepsy	Global developmental delay, febrile seizures, muscular hypotonia
NARS1	NM_004539.3:c.1067A>Cp.(Asp356Ala)	NM_004539.3:c.1564C>Tp.(Arg522*)	AR_comphet	7.2	comphet	maternal&paternal	2	NDD + Epilepsy	Global developmental delay, febrile seizures, muscular hypotonia
RHOQ	NM_012249.3:c.359G>Ap.(Gly120Glu)		AD_denovo	7.3	het	de novo	1	NDD	Delayed speech and language development, Intellectual disability, Abnormality of the kidney (horseshoe kidney), Anal atresia, Jejunal atresia, Ileal atresia, Premature Abnormality of the pubic hair
MYRIP	NM_001284423.1:c.383G>Ap.(Arg128His)	NM_001284423.1:c.86G>Ap.(Arg29His)	AR_comphet	4.9	comphet	maternal&paternal	3	NDD	Severe global developmental delay, sleep disturbance, behavioral abnormalities, flat face
AATF	NM_012138.3:c.695-3C>Ap.?		AD_denovo	3.5	het	de novo	3	NDD	Severe global developmental delay, sleep disturbance, behavioral abnormalities, flat face

ZNF280D	NM_001288588.1:c.2532C>Gp.(His844Gln)		AD_denovo	4.5	het	de novo	1	NDD + Epilepsy	Mild intellectual disability, hydrocephalus, abnormality of brain morphology including cortical dysplasia, focal impaired awareness seizure, generalized tonic-clonic seizures with focal onset
PCNX2	NM_014801.3:c.3846C>Ap.(Asp1282Glu)		AD_denovo	5.5	het	de novo	2	NDD	Visual loss, Optic nerve hypoplasia, Global developmental delay, Hypoglycemia, Abnormality of optic chiasm morphology
DNAH14	NM_001373.1:c.13384G>Ap.(Ala4462Thr)	NM_001373.1:c.6100C>Tp.(Arg2034*)	AR_comphet	4.3	comphet	maternal&paternal	2	NDD	Visual loss, Optic nerve hypoplasia, Global developmental delay, Hypoglycemia, Abnormality of optic chiasm morphology
FAM199X	NM_207318.3:c.961T>Ap.(Ser321Thr)		XL	5.6	hemi	maternal	2	NDD + Epilepsy	Aggressive behavior, Delayed speech and language development, Seizures, Global developmental delay, Absent speech, EEG abnormality, Focal impaired awareness seizure, Focal-onset seizure
NEU4	NM_001167599.2:c.1396T>Cp.(Cys466Arg)	NM_001167599.2:c.407G>Ap.(Arg136His)	AR_comphet	3.6	comphet	maternal&paternal	2	NDD + Epilepsy	Focal impaired awareness seizures since 10/2016, EEG abnormality, delayed speech and language development, aggressive behavior
ARL14EP	NM_152316.2:c.707G>Ap.(Arg236His)		AD_denovo	8.2	het	de novo	2	NDD + Epilepsy	Intellectual disability, Absent speech, Hyperreflexia, Generalized tonic-clonic seizures, Absence seizure, EEG abnormality, Excessive salivation, Brain atrophy, Epileptic encephalopathy
PSMB4	NM_002796.2:c.226G>Ap.(Gly76Ser)		AD_denovo	7.7	het	de novo	2	NDD + Epilepsy	Intellectual disability, Absent speech, Hyperreflexia, Generalized tonic-clonic seizures, Absence seizure, EEG abnormality, Excessive salivation, Brain atrophy, Epileptic encephalopathy
SHANK1	NM_016148.3:c.5324G>Tp.(Gly1775Val)		AR_homo	B	homo	maternal&paternal	3	Fehlbildungen	Meningo- or exencephalocele occipital, right radius aplasia with shortened ulna, death in utero
SYT3	NM_001160328.1:c.401C>Gp.(Ala134Gly)		AR_homo	C	homo	maternal&paternal	3	Fehlbildungen	Meningo- or exencephalocele occipital, right radius aplasia with shortened ulna, death in utero
RPS6KC1	NM_012424.4:c.2633G>Ap.(Gly878Glu)		AR_homo	C	homo	maternal&paternal	3	Fehlbildungen	Meningo- or exencephalocele occipital, right radius aplasia with shortened ulna, death in utero
TACC2	NM_206862.3:c.1407G>Cp.(Glu469Asp)	NM_206862.3:c.1242G>Cp.(Glu414Asp)	AR_comphet	3.6	comphet	maternal&paternal	2	NDD	Macrocephaly, hypoplasia of the corpus callosum, suspected developmental delay,
SETD1B	NM_015048.1:c.1743C>Gp.(Asp581Glu)	NM_015048.1:c.2999G>Ap.(Arg1000Gln)	AR_comphet	5.9	comphet	maternal&paternal	2	NDD	Macrocephaly, hypoplasia of the corpus callosum, suspected developmental delay, right hemiparesis
DUSP16	NM_030640.2:c.1091C>Ap.(Pro364His)	NM_030640.2:c.183C>Gp.(Asp61Glu)	AR_comphet	3.8	comphet	maternal&paternal	1	NDD + Epilepsy	Microcephaly, Delayed speech and language development, Seizures, Global developmental delay, Recurrent infections, Infantile onset, Postnatal microcephaly
MCIDAS	NM_001190787.1:c.487C>Tp.(Arg163Trp)		AD_denovo	6.2	het	de novo	2	NDD + Epilepsy	Stereotypy, Delayed speech and language development, Global developmental delay, Motor delay, Delayed gross motor development, EEG abnormality, Delayed fine motor development
POLD1	NM_001308632.1:c.1657G>Ap.(Val553Ile)		AR_homo	8.1	homo	maternal&paternal	1	NDD	Global developmental delay
FAT2	NM_001447.2:c.9524T>Cp.(Leu3175Pro)		AR_homo	7.4	homo	maternal&paternal	4	NDD	Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Severe muscular hypotonia, Muscular hypotonia of the trunk, Infantile muscular hypotonia, Intellectual disability, severe, Central hypotonia, Cognitive impairment
PRDM13	NM_021620.3:c.994G>Ap.(Gly332Arg)		AR_homo	4.7	homo	maternal&paternal	4	NDD	Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Severe muscular hypotonia, Muscular hypotonia of the trunk, Infantile muscular hypotonia, Intellectual disability, severe, Central hypotonia, Cognitive impairment

RASSF10	NM_001080521.2:c.816C>Gp.(Tyr272*)		AD_denovo	3.7	het	de novo	2	NDD	Cleft palate, Hydrocephalus, Microcephaly, Retinopathy, Intellectual disability, Global developmental delay, Pes planus, Short stature, Pes valgus, Cognitive impairment, Cleft hard palate
GPATCH2	NM_018040.3:c.1167-8C>Tp.?		AD_denovo	3.3	het	de novo	2	NDD	Cleft palate, Hydrocephalus, Microcephaly, Retinopathy, Intellectual disability, Global developmental delay, Pes planus, Short stature, Pes valgus, Cognitive impairment, Cleft hard palate
ATP1A3	NM_001256214.1:c.6+3A>Gp.?		AR_homo	10.5	homo	maternal&paternal	4	NDD	Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Severe muscular hypotonia, Muscular hypotonia of the trunk, Infantile muscular hypotonia, Intellectual disability, severe, Central hypotonia, Cognitive impairment
CLTCL1	NM_007098.3:c.1820A>Gp.(His607Arg)	NM_007098.3:c.2791A>Gp.(Ile931Val)	AR_comphet	C	comphet	maternal&paternal	1	congenital heart defects	Motor delay, Ventricular septal defect, Tracheal stenosis, Abnormal trachea morphology, Pulmonary artery atresia, Pulmonary valve atresia, Abnormality of cardiovascular system morphology
RAB5B	NM_001252036.1:c.115C>Gp.(Arg39Gly)		AD_denovo	7.1	het	de novo	3	NDD + Epilepsy	Hydrocephalus, Intellectual disability, Seizures, Global developmental delay, EEG abnormality, Aqueductal stenosis
ARHGAP4	NM_001666.4:c.301C>Tp.(His101Tyr)		XL	5.2	hemi	maternal	3	NDD + Epilepsy	Hydrocephalus, Intellectual disability, Seizures, Global developmental delay, EEG abnormality, Aqueductal stenosis
CBLL2	NM_152577.3:c.701A>Gp.(Lys234Arg)		XL	4.1	hemi	maternal	3	NDD + Epilepsy	Hydrocephalus, Intellectual disability, Seizures, Global developmental delay, EEG abnormality, Aqueductal stenosis
LRRK2	NM_198578.3:c.3634C>Tp.(Pro1212Ser)	NM_198578.3:c.137C>Tp.(Thr46Met)	AR_comphet	7.8	comphet	maternal&paternal	3	NDD	Behavioral abnormality, Intellectual disability, Gait ataxia
ZDHC14	NM_024630.2:c.1441G>Ap.(Gly481Ser)		AD_denovo	4.4	het	de novo	3	NDD	Behavioral abnormality, Intellectual disability, Gait ataxia
NIF3L1	NM_001136039.2:c.131C>Tp.(Ser44Leu)	NM_001136039.2:c.347C>Gp.(Ala116Gly)	AR_comphet	3.8	comphet	maternal&paternal	3	NDD	Behavioral abnormality, Intellectual disability, Gait ataxia
MAP3K15	NM_001001671.3:c.2037dup, p.(Ile680Hisfs*9)		XL	5.3	hemi	maternal	5	NDD + Epilepsy	Seizures, Generalized myoclonic seizures
HMCN2	NM_001291815.1:c.4444G>Tp.(Gly1482*)	NM_001291815.1:c.489+1G>Cp.?	AR_comphet	3.6	comphet	maternal&paternal	5	NDD + Epilepsy	Seizures, Generalized myoclonic seizures
RPTOR	NM_020761.2:c.3533G>Ap.(Arg1178His)	NM_020761.2:c.503A>Gp.(Asn168Ser)	AR_comphet	6.9	comphet	maternal&paternal	5	NDD + Epilepsy	Seizures, Generalized myoclonic seizures
SLC30A5	NM_022902.4:c.832_836delp.(Ile278Phefs*33)		AR_homo	A	homo	maternal&paternal	1	Fehlbildungen	Cardiomyopathy, Hydrops fetalis, Noncompaction cardiomyopath
ZNF761	NM_001008401.3:c.2085_2086delp.(Cys695Trpfs*5)		AD_denovo	3.9	het	de novo	5	NDD + Epilepsy	Seizures, Generalized myoclonic seizures
HAGH	NM_005326.4:c.355_357dup, p.(Ser119dup)	NM_005326.4:c.578A>Gp.(Tyr193Cys)	AR_comphet	4.0	comphet	maternal&paternal	5	NDD + Epilepsy	Seizures, Generalized myoclonic seizures
POLR3A	NM_007055.3:c.1096A>Gp.(Arg366Gly)		AD_denovo	9.9	het	de novo	1	NDD + Epilepsy	Seizures, Focal impaired awareness seizure, Hypothalamic hamartoma, Focal-onset seizure, Epileptic spasms, Langerhans cell histiocytosis
PLCG1	NM_002660.2:c.3196C>Gp.(Pro1066Ala)		AD_unknown	6.8	het	unknown	1	NDD + Epilepsy	Focal epilepsy and mild intellectual disability, depression, global developmental delay, EEG abnormality, short stature, Obesity, mild ataxia on physical examination, cerebellar atrophy

EPHA4	NM_004438.4:c.2105T>Cp.(Met702Thr)		AD_unknown	6.9	het	unknown	1	NDD + Epilepsy	Sensorineural hearing impairment, Strabismus, Abnormality of the hand, Intellectual disability, Seizures, Intellectual disability, mild, Abnormality of neuronal migration, Gray matter heterotopias, Gray matter heterotopia, Intellectual disability, moderate, EEG abnormality, Kyphosis, Clinodactyly of the 5th finger, Periventricular heterotopia, Focal-onset seizure, Focal sensory seizure, EEG with focal epileptiform discharges, EEG with focal spikes, Abnormality of brain morphology
AATK	NM_001080395.2:c.2915G>Tp.(Gly972Val)		AR_homo	4.9	homo	maternal&paternal	2	NDD	Delayed speech and language development, Global developmental delay, Ventriculomegaly, Developmental regression, Expressive language delay, Severe expressive language delay, Progressive language deterioration, Receptive language delay, Moderate expressive language delay, Mild expressive language delay, Mild receptive language delay, Moderate receptive language delay, Severe receptive language delay
ARHGAP6	NM_013427.2:c.1586T>Cp.(Val529Ala)		XL	4.0	hemi	maternal	2	NDD	Delayed speech and language development, Global developmental delay, Ventriculomegaly, Developmental regression, Expressive language delay, Severe expressive language delay, Progressive language deterioration, Receptive language delay, Moderate expressive language delay, Mild expressive language delay, Mild receptive language delay, Moderate receptive language delay, Severe receptive language delay
UNC13A	NM_001080421.2:c.3215+1G>Cp.?		AD_unknown	10.3	het	unknown	3	NDD	Single transverse palmar crease, Syndactyly, Intellectual disability, Obesity, Toe syndactyly, Status epilepticus, Abnormality of body weight, Bipolar affective disorder, Increased adipose tissue, Epileptic spasms, Schizophrenia, Mania
LMX1A	NM_001174069.1:c.517dup, p.(Ser173Lysfs*15)		AD_unknown	8.0	het	unknown	3	NDD	Single transverse palmar crease, Syndactyly, Intellectual disability, Obesity, Toe syndactyly, Status epilepticus, Abnormality of body weight, Bipolar affective disorder, Increased adipose tissue, Epileptic spasms, Schizophrenia, Mania
INO80	NM_017553.2:c.1294G>Ap.(Gly432Arg)		AD_unknown	5.5	het	unknown	3	NDD	Single transverse palmar crease, Syndactyly, Intellectual disability, Obesity, Toe syndactyly, Status epilepticus, Abnormality of body weight, Bipolar affective disorder, Increased adipose tissue, Epileptic spasms, Schizophrenia, Mania
ZNF652	NM_001145365.1:c.65T>Cp.(Met22Thr)		AD_unknown	2.5	het	unknown	1	NDD + Epilepsy	Intellectual disability, Seizures, Generalized tonic-clonic seizures, Absence seizure, Generalized myoclonic seizures, Generalized-onset seizure, Short stature, Spontaneous abortion, Almond-shaped palpebral fissure, Atonic seizures, Recurrent spontaneous abortion
TANC2	NM_025185.3:c.3397G>Tp.(Gly1133*)		AD_unknown	8.2	het	unknown	1	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Generalized myoclonic seizures, Status epilepticus, Intellectual disability, moderate, Epileptic spasms, Cognitive impairment, Epileptic encephalopathy
TANC1	NM_001145909.1:c.2395G>Ap.(Asp799Asn)		AD_unknown	4.2	het	unknown	4	NDD	Cleft palate, Cleft upper lip, Microphthalmia, Global developmental delay
COL20A1	NM_020882.2:c.3467G>Tp.(Gly1156Val)	NM_020882.2:c.807C>Ap.(Asn269Lys)	AR_comphet	4.0	comphet	maternal&paternal	4	NDD	Cleft palate, Cleft upper lip, Microphthalmia, Global developmental delay
ATP6V0A1	NM_001130020.1:c.53A>Tp.(Gln18Leu)		AD_denovo	8.1	het	de novo	1	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Absence seizures, Generalized myoclonic seizures, Status epilepticus, Focal clonic seizures, Focal seizures without impairment of consciousness or awareness, EEG abnormality, Focal seizures with impairment of consciousness or awareness, Hypsarrhythmia, Infantile onset, Focal seizures, Epileptic spasms, Symptomatic seizures, Typical absence seizures, Focal motor seizures, Focal tonic seizures, Infantile spasms, Epileptic encephalopathy, Multifocal seizures
C16orf70	NM_001320540.1:c.1050+1G>Ap.?		AD_denovo	9.1	het	de novo	2	NDD	Global developmental delay, short stature, talipes equinovarus
EPHB2	NM_001309193.1:c.2858T>Cp.(Ile953Thr)		AD_denovo	8.0	het	de novo	1	NDD + Epilepsy	epilepsy, global developmental delay, dysmorphic facial features
BIRC6	NM_016252.3:c.8570C>Gp.(Ser2857Cys)	NM_016252.3:c.12796G>Ap.(Val4266Met)	AR_comphet	4.4	comphet	maternal&paternal	1	NDD + Epilepsy	Delayed speech and language development, Seizures, Familial predisposition, Poor speech, Infantile onset, Infantile spasms
WDR3	NM_006784.2:c.989+1G>Cp.?	NM_006784.2:c.1555T>Ap.(Ser519Thr)	AR_comphet	5.4	comphet	maternal&paternal	4	NDD	Cleft palate, Cleft upper lip, Microphthalmia, Global developmental delay

FGF2	NM_002006.4:c.498C>Gp.(Tyr166*)		AD_unknown	7.6	het	unknown	1	NDD + Epilepsy	Hydrocephalus, Intellectual disability, Hemiparesis, Cerebellar atrophy, Focal clonic seizures, Febrile seizures, Hemiplegia/hemiparesis, Periventricular leukomalacia, Focal motor seizure, Focal autonomic seizure, Focal myoclonic seizures, Focal tonic seizures
CNTN6	NM_014461.3:c.275A>Tp.(Asp92Val)	NM_014461.3:c.2573G>Ap.(Ser858Asn)	AR_comphet	5.5	comphet	maternal&paternal	4	NDD	Cleft palate, Cleft upper lip, Microphthalmia, Global developmental delay
FAM131A	NM_144635.4:c.838C>Gp.(Leu280Val)		AR_homo	B	homo	maternal&paternal	1	Muscle	neuromuscular scoliosis, muscular hypotonia, intermittent exotropia
TMEM92	NM_001168215.1:c.212G>Cp.(Cys71Ser)		AD_denovo	B	het	de novo	1	Fehlbildungen	inguinal hernia, cryptorchidism, hypospadias, renal cyst, hemangioma, bicuspid aortic valve, coarctation of aorta, anal atresia, choroid plexus cyst, abnormal vertebral morphology, multiple renal cysts
RORB	NM_006914.3:c.235+1_235+2insTp.?		AD_unknown	8.4	het	unknown	2	NDD + Epilepsy	Male hypogonadism, Hypogonadotrophic hypogonadism, Hypogonadism, Thick upper lip vermilion, Delayed puberty, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Myoclonus, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Intellectual disability, moderate, Intellectual disability, borderline, Focal tonic seizures, Thick vermilion border
RORB	NM_006914.3:c.208T>Cp.(Cys70Arg)		AD_inherited	6.7	het	maternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Mental deterioration, Absence seizure, Generalized myoclonic seizures, Status epilepticus, Intellectual disability, profound, Intellectual disability, moderate, Intellectual disability, severe, Epileptic spasms, Myoclonic absence, Cognitive impairment, Epileptic encephalopathy
NEURL4	NM_032442.2:c.4345C>Gp.(Pro1449Ala)	NM_032442.2:c.2944G>Ap.(Glu982Lys)	AR_comphet	4.5	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Atonic seizures, Focal tonic seizures, Myoclonic atonic seizures
MEIOC	NM_001145080.2:c.2180G>Ap.(Gly727Asp)	NM_001145080.2:c.2622A>Tp.(Arg874Ser)	AR_comphet	2.2	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Atonic seizures, Focal tonic seizures, Myoclonic atonic seizures
XYLB	NM_005108.3:c.1101G>Ap.(Met367Ile)		AD_denovo	5.0	het	de novo	1	NDD	Single transverse palmar crease, Intellectual disability, Muscular hypotonia, Global developmental delay, Gait disturbance, Absent speech, Supernumerary nipple, Lumbar scoliosis, Abnormal social behavior, Cognitive impairment, Self-injurious behavior
ATP2B3	NM_001001344.2:c.3530C>Tp.(Pro1177Leu)		XL	6.0	hemi	maternal	2	NDD	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Global developmental delay, Motor delay, Encopresis
GRIK3	NM_000831.3:c.1531-5T>Gp.?		AD_unknown	4.1	het	unknown	1	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Generalized tonic clonic seizures, Intellectual disability, moderate, Focal-onset seizure, Pituitary hypothyroidism, Intellectual disability, severe, Focal tonic seizures, Arrhythmia, Abnormality of brain morphology, Abnormality of cardiovascular system morphology
BSN	NM_003458.3:c.9919A>Gp.(Ser3307Gly)		AR_homo	6.7	homo	maternal&paternal	2	NDD + Epilepsy	Male hypogonadism, Hypogonadotrophic hypogonadism, Hypogonadism, Thick upper lip vermilion, Delayed puberty, Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Myoclonus, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Intellectual disability, moderate, Intellectual disability, borderline, Focal tonic seizures, Thick vermilion border
PUM2	NM_015317.2:c.2216delp.(His739Leufs*10)		AD_unknown	10.9	het	unknown	1	NDD + Epilepsy	Strabismus, Intellectual disability, Seizures, Global developmental delay, Generalized tonic-clonic seizures, Global brain atrophy, EEG abnormality, Developmental regression, Short stature, Brain atrophy, Cognitive impairment
HMG20A	NM_001304504.1:c.237+5G>Tp.?		AR_homo	5.0	homo	maternal&paternal	1	NDD + Epilepsy	Macroglossia, Mandibular prognathia, Thick eyebrow, Intellectual disability, Seizures, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Kyphoscoliosis, Genu valgum, Excessive salivation, Protruding tongue, Intellectual disability, severe, Thick hair, Rigors

KIF5C	NM_004522.2:c.2385dup, p.(Gln796Alafs*19)		AD_unknown	9.9	het	unknown	1	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Mental deterioration, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Focal clonic seizures, Intellectual disability, moderate, Focal impaired awareness seizure, Intellectual disability, borderline, Focal-onset seizure, Focal motor seizure, Focal tonic seizures, Generalized clonic seizures, Abnormal morphology of the hippocampus, Focal seizures, afebril
RORB	NM_006914.3:c.777G>Ap.(Trp259*)		AD_unknown	8.1	het	unknown	1	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe, Epileptic spasms
RNF13	NM_007282.4:c.(409+1_410-1)_(500+1_501-1)delp.?		AD_denovo	6.2	het	de novo	1	NDD + Epilepsy	Microcephaly, Seizures, Global developmental delay, Hepatosplenomegaly, Anemia, Hypoplasia of the corpus callosum, Polymicrogyria, Abnormality of midbrain morphology, Elevated hepatic transaminase, Elevated gamma-glutamyltransferase activity
PRKCE	NM_005400.2:c.1186C>Tp.(Arg396Trp)		AD_denovo	8.2	het	de novo	1	NDD + Epilepsy	Seizures, Focal-onset seizure
SSBP2	NM_001256732.2:c.566C>Tp.(Pro189Leu)		AD_unknown	3.4	het	unknown	3	NDD + Epilepsy	Delayed speech and language development, Intellectual disability, Seizures, Intellectual disability, mild, Dystonia, Gliosis, Focal clonic seizures, EEG abnormality, Poor speech, Focal-onset seizure, Dyslexia, Focal motor seizure, Focal tonic seizures, Abnormality of movement, Cognitive impairment
ZNF319	NM_020807.2:c.654_655delp.(Arg219Alafs*2)		AD_unknown	5.3	het	unknown	3	NDD + Epilepsy	Delayed speech and language development, Intellectual disability, Seizures, Intellectual disability, mild, Dystonia, Gliosis, Focal clonic seizures, EEG abnormality, Poor speech, Focal-onset seizure, Dyslexia, Focal motor seizure, Focal tonic seizures, Abnormality of movement, Cognitive impairment
GLRA4	NM_001024452.2:c.39_41delp.(Leu14del)		XL	5.5	hemi	unknown	2	NDD + Epilepsy	Thin upper lip vermillion, Turricephaly, Synophrys, Acne, Intellectual disability, Seizures, Mental deterioration, Spastic tetraparesis, Absent speech, Flexion contracture, Cerebral atrophy, Nail dysplasia, Focal clonic seizures, Tetraparesis, Spastic paraparesis, Paraparesis, Tetraplegia, Neonatal respiratory distress, High, narrow palate, Elbow flexion contracture, Limb joint contracture, Skeletal muscle atrophy, Limb muscle weakness, Short stature, Focal-onset seizure, Limb undergrowth, Paraplegia/paraparesis, Intellectual disability, severe, Focal motor seizure, Focal tonic seizures, Tetraplegia/tetraparesis
FAM47C	NM_001013736.2:c.1699_1770delp.(Met567_Ar g590del)		XL	2.1	hemi	unknown	2	NDD + Epilepsy	Thin upper lip vermillion, Turricephaly, Synophrys, Acne, Intellectual disability, Seizures, Mental deterioration, Spastic tetraparesis, Absent speech, Flexion contracture, Cerebral atrophy, Nail dysplasia, Focal clonic seizures, Tetraparesis, Spastic paraparesis, Paraparesis, Tetraplegia, Neonatal respiratory distress, High, narrow palate, Elbow flexion contracture, Limb joint contracture, Skeletal muscle atrophy, Limb muscle weakness, Short stature, Focal-onset seizure, Limb undergrowth, Paraplegia/paraparesis, Intellectual disability, severe, Focal motor seizure, Focal tonic seizures, Tetraplegia/tetraparesis
SIPA1L1	NM_015556.2:c.3321_3322delp.(Arg1107Serfs*1 1)		AD_unknown	7.5	het	unknown	2	NDD + Epilepsy	Macrotia, Myopia, Intellectual disability, Seizures, Intellectual disability, mild, Cerebellar atrophy, Tremor, Cerebral atrophy, Broad-based gait, Intellectual disability, profound, Intellectual disability, moderate, Hand tremor, Focal-onset seizure, Epileptic spasms, Interictal epileptiform activity, Limb tremor, Epileptic encephalopathy, Long ear
SRSF11	NM_004768.3:c.1178delp.(Arg393Hisfs*26)		AD_unknown	8.6	het	unknown	2	NDD + Epilepsy	Macrotia, Myopia, Intellectual disability, Seizures, Intellectual disability, mild, Cerebellar atrophy, Tremor, Cerebral atrophy, Broad-based gait, Intellectual disability, profound, Intellectual disability, moderate, Hand tremor, Focal-onset seizure, Epileptic spasms, Interictal epileptiform activity, Limb tremor, Epileptic encephalopathy, Long ear
BDP1	NM_018429.2:c.4813A>Gp.(Arg1605Gly)		AD_denovo	6.3	het	de novo	3	NDD + Epilepsy	Intellectual disability, Seizures, Status epilepticus
NUP188	NM_015354.2:c.17G>Cp.(Gly6Ala)	NM_015354.2:c.2917C>Tp.(Arg973*)	AR_comphet	5.5	comphet	maternal& paternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Status epilepticus
TMEM151A	NM_153266.3:c.76-1G>Cp.?		AD_denovo	6.8	het	de novo	3	NDD + Epilepsy	Intellectual disability, Seizures, Status epilepticus
NFATC1	NM_001278669.1:c.2249_2251delCCTinsTCGp.(P ro750_Cys751delinsLeuGly)		AD_denovo	6.3	het	de novo	1	NDD + Epilepsy	Seizures, Global developmental delay, Abnormal cerebellum morphology, Abnormality of the basal ganglia, Increased serum lactate, Abnormality of midbrain morphology, Lacticaciduria, Epileptic spasms, Abnormality of brain morphology

ABCA2	NM_001606.4:c.801_802delTGinsGTp.(Val268Phe)		AR_homo	7.3	homo	maternal&paternal	4	NDD	Adducted thumb, Intellectual disability, Intellectual disability, mild, Global developmental delay, Abnormal facial shape, Pyloric stenosis, Ventriculomegaly, Intellectual disability, moderate, Infantile muscular hypotonia, Feeding difficulties, Cognitive impairment, Impaired feeding ability, VUS in COLQ (31.07.2019)
BIRC6	NM_016252.3:c.10735A>Gp.(Met3579Val)		AR_homo	6.4	homo	maternal&paternal	2	NDD + Epilepsy	Microcephaly, Visual impairment, Intellectual disability, Seizures, Global developmental delay, Motor delay, Encephalopathy, Generalized tonic-clonic seizures
PPM1L	NM_139245.3:c.237G>Cp.(Glu79Asp)		AR_homo	4.4	homo	maternal&paternal	2	NDD + Epilepsy	Microcephaly, Visual impairment, Intellectual disability, Seizures, Global developmental delay, Motor delay, Encephalopathy, Generalized tonic-clonic seizures
RGMA	NM_001166283.1:c.748G>Cp.(Ala250Pro)		AD_denovo	6.9	het	de novo	2	NDD	Spasticity, Global developmental delay, Motor delay, Cerebral palsy, Abnormality of movement, Dyskinesia
ANXA6	NM_001155.4:c.1670C>Tp.(Pro557Leu)	NM_001155.4:c.319-6_319-5delCCinsTGp.?	AR_comphet	4.0	comphet	maternal&paternal	2	NDD	Spasticity, Global developmental delay, Motor delay, Cerebral palsy, Abnormality of movement, Dyskinesia
NRDE2	NM_017970.3:c.441delp.(Arg148Alafs*11)		AR_homo	8.5	homo	maternal&paternal	1	NDD	Intellectual disability, seizures, global developmental delay, encephalopathy infantile spasms
INTS7	NM_015434.3:c.2240G>Tp.(Arg747Ile)		AD_denovo	6.0	het	de novo	1	NDD	Microcephaly, Intrauterine growth retardation, Abnormal facial shape, Basal ganglia calcification, Cerebral calcification, Congenital intracerebral calcification
SF3A1	NM_005877.5:c.310G>Ap.(Gly104Arg)		AD_denovo	7.3	het	de novo	1	NDD + Epilepsy	Seizures, Global developmental delay, Abnormality of movement, Epileptic encephalopathy
SLC16A10	NM_018593.4:c.626G>Ap.(Gly209Asp)		AD_denovo	6.6	het	de novo	1	NDD + Epilepsy	Microcephaly, Behavioral abnormality, Seizures, Global developmental delay, Absence seizure, Generalized-onset seizure, Myoclonic atonic seizures
MROH2B	NM_173489.4:c.3685delp.(Asp1229Thrfs*15)		AD_denovo	5.0	het	de novo	1	NDD + Epilepsy	Seizures, Encephalopathy, Absence seizure, Generalized-onset seizure
PRDX2	NM_005809.5:c.153C>Ap.(Cys51*)		AD_denovo	7.3	het	de novo	1	NDD + Epilepsy	Seizures, absent septum pellucidum, paroxysmal dyskinesia, dyskinesia
SLC5A7	NM_021815.4:c.178+1G>Cp.?		AD_inherited	7.8	het	maternal	1	Neuro	Ataxia, spastic paraplegia, muscle weakness, hyperreflexia, pes cavus, myalgia, limb muscle weakness, paraplegia
ZNF341	NM_032819.4:c.2260C>Tp.(Arg754Cys)		AD_denovo	4.3	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Specific learning disability, Absence seizure, Generalized-onset seizure, Intellectual disability, borderline, Attention deficit hyperactivity disorder
KCNK7	NM_033347.1:c.681C>Gp.(His227Gln)		AD_denovo	4.7	het	de novo	1	NDD + Epilepsy	Seizures, Generalized tonic-clonic seizures, Generalized myoclonic seizures
ZZEF1	NM_015113.3:c.1580C>Tp.(Pro527Leu)		AD_denovo	5.9	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Intellectual disability, mild, Specific learning disability, Absence seizure, Generalized-onset seizure, Intellectual disability, borderline, Attention deficit hyperactivity disorder
MTMR3	NM_021090.3:c.848A>Gp.(Asn283Ser)	NM_021090.3:c.1088G>Ap.(Arg363Gln)	AR_comphet	4.3	comphet	maternal&paternal	1	NDD + Epilepsy	Delayed speech and language development, Seizures, Global developmental delay, Focal impaired awareness seizure, Cortical dysplasia, Focal-onset seizure, Complex febrile seizures, Abnormal morphology of the hippocampus
INPP5F	NM_014937.3:c.3172_3174delp.(Ser1058del)	NM_014937.3:c.3144_3149delp.(Leu1049_Glu1050del)	AR_comphet	4.1	comphet	maternal&paternal	1	NDD + Epilepsy	Global developmental delay, Epileptic spasms
HCN2	NM_001194.3:c.1120A>Cp.(Met374Leu)		AD_denovo	8.9	het	de novo	1	NDD + Epilepsy	Microcephaly, delayed speech and language development, intellectual disability, global developmental delay, motor delay, generalized-onset seizure, epileptic spasms, cognitive impairment
DHX36	NM_020865.2:c.800_802delp.(Ile267del)		AD_denovo	5.9	het	de novo	1	NDD	Short attention span, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Delayed gross motor development, Attention deficit hyperactivity disorder, Delayed fine motor development
DOCK3	NM_004947.4:c.1175G>Ap.(Arg392Gln)	NM_004947.4:c.3740T>Cp.(Met1247Thr)	AR_comphet	9.3	comphet	maternal&paternal	1	NDD + Epilepsy	Seizures, Global developmental delay

SEZ6L2	NM_001243332.1:c.1084G>Ap.(Val362Met)	NM_001243332.1:c.85C>Tp.(Pro29Ser)	AR_comphet	6.1	comphet	maternal&paternal	1	NDD + Epilepsy	Seizures, status epilepticus, focal-onset seizure, EEG with spike-wave complexes, epilepsy not completely under control, cognitive deficiency, intellectual disability
NOP58	NM_015934.4:c.1018C>Gp.(Leu340Val)		AD_denovo	7.0	het	de novo	1	NDD + Epilepsy	Autism, Intellectual disability, Status epilepticus, Focal-onset seizure, Hippocampal atrophy
SLITRK4	NM_001184749.2:c.2435T>Cp.(Phe812Ser)		XL	5.1	hemi	maternal	1	NDD	Myopia, Autism, Autistic behavior, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Dysarthria, Global developmental delay, Delayed gross motor development, Intellectual disability, moderate, Delayed fine motor development, High myopia
PGBD2	NM_170725.2:c.607A>Cp.(Thr203Pro)		AD_denovo	4.0	het	de novo	2	NDD	Autism, Autistic behavior, Intellectual disability, Global developmental delay, Situs inversus totalis, Abnormal facial shape, Asthma, Recurrent respiratory infections, Short stature, Respiratory tract infection
ZNF81	NM_007137.3:c.476A>Gp.(Lys159Arg)		XL	6.5	hemi	maternal	2	NDD	Autism, Autistic behavior, Intellectual disability, Global developmental delay, Situs inversus totalis, Abnormal facial shape, Asthma, Recurrent respiratory infections, Short stature, Respiratory tract infection
ZFYVE26	NM_015346.3:c.5779T>Ap.(Tyr1927Asn)		AD_denovo	10.3	het	de novo	1	NDD	Global developmental delay, Absent speech, Proportionate short stature, Short stature
FAT3	NM_001008781.2:c.1367C>Tp.(Ala456Val)	NM_001008781.2:c.11012G>Tp.(Arg3671Leu)	AR_comphet	5.1	comphet	maternal&paternal	1	NDD + Epilepsy	strukturelle und therapierefraktäre Epilepsie (ESES/CSWS), zervikale Syringomyelie, Intelligenzminderung, Verhaltensauffälligkeiten, Z.n. IVH Grad IV (intraventrikuläre Hämorrhagie) in 2. Lebenswoche, cMRT-Auffälligkeiten
PKN3	NM_013355.4:c.137A>Cp.(Asp46Ala)		AD_denovo	5.0	het	de novo	1	NDD + Epilepsy	Generalisierte Epilepsie mit febrilen Anfällen seit dem 3. LJ
DACH2	NM_053281.3:c.1519G>Tp.(Val507Phe)		XL	3.4	hemi	maternal	2	NDD	Macrocephaly, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, profound, Intellectual disability, moderate, Intellectual disability, severe, Cognitive impairment
GABRE	NM_004961.3:c.319G>Tp.(Gly107Cys)		XL	4.9	hemi	maternal	2	NDD	Macrocephaly, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, profound, Intellectual disability, moderate, Intellectual disability, severe, Cognitive impairment
ARRB2	NM_001257328.1:c.684+1G>Cp.?		AD_denovo	10.2	het	de novo	1	NDD + Epilepsy	autism-spectre disorder, focal- onset epilepsy
DBF4B	NM_145663.2:c.902G>Tp.(Cys301Phe)		AR_homo	6.3	homo	maternal&paternal	4	NDD	Microcephaly, Autistic behavior, Intellectual disability, Intellectual disability, mild, Global developmental delay, Intellectual disability, moderate, Poor speech, Intellectual disability, severe
TBC1D9B	NM_198868.2:c.583G>Tp.(Ala195Ser)		AD_denovo	5.6	het	de novo	1	Neuro	Abnormality of the optic nerve, Optic atrophy, Polyneuropathy, Encephalopathy, Leukoencephalopathy, Leukodystrophy, Tetraplegia
CASP9	NM_001229.4:c.631-6T>Cp.?	NM_001229.4:c.710A>Cp.(His237Pro)	AR_comphet	6.5	comphet	maternal&paternal	3	NDD	Renal agenesis, Abnormal cornea morphology, Aniridia, Microphthalmia, Global developmental delay
TNPO3	NM_012470.3:c.2541dup, p.(Tyr848Leufs*8)		AD_denovo	6.9	het	de novo	3	NDD	Renal agenesis, Abnormal cornea morphology, Aniridia, Microphthalmia, Global developmental delay
SLC23A1	NM_152685.3:c.1105A>Gp.(Ile369Val)	NM_152685.3:c.1063C>Ap.(Pro355Thr)	AR_comphet	4.5	comphet	maternal&paternal	3	NDD	Renal agenesis, Abnormal cornea morphology, Aniridia, Microphthalmia, Global developmental delay
DGKK	NM_001013742.3:c.1247A>Tp.(His416Leu)		XL	1.0	hemi	maternal	4	NDD	Micropenis, Intellectual disability, Intellectual disability, mild, Global developmental delay
SMARCA1	NM_003069.4:c.34G>Ap.(Val12Met)		XL	6.5	hemi	maternal	4	NDD	Micropenis, Intellectual disability, Intellectual disability, mild, Global developmental delay
PON1	NM_000446.5:c.717G>Cp.(Glu239Asp)		AD_denovo	5.3	het	de novo	4	NDD	Micropenis, Intellectual disability, Intellectual disability, mild, Global developmental delay

KCNN2	NM_021614.3:c.1082A>Gp.(Tyr361Cys)		AD_denovo	7.8	het	de novo	2	NDD	Myopia, Nystagmus, Stereotypy, Delayed speech and language development, Intellectual disability, Motor delay, Absent speech, Abnormality of the foot, Intellectual disability, profound, Difficulty walking, Poor speech, Equinus calcaneus, Vertical nystagmus, Intellectual disability, severe, Severe global developmental delay, Pschomotor retardation
CAND2	NM_001162499.1:c.2591C>Tp.(Ala864Val)		AD_denovo	4.8	het	de novo	4	NDD	Micropenis, Intellectual disability, Intellectual disability, mild, Global developmental delay
MARVELD3	NM_001017967.3:c.1168G>Ap.(Gly390Ser)		AD_denovo	5.2	het	de novo	1	NDD	Autistic behavior, Intellectual disability, Global developmental delay, Obesity, Polyphagia, Developmental stagnation, Retractable testis, Cognitive impairment
SLC32A1	NM_080552.2:c.787G>Ap.(Val263Met)		AD_denovo	7.8	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizures, Generalized myoclonic seizures, Infantile onset
CLCN3	NM_173872.3:c.336_339delp.(Lys112Asnfs*6)		AR_homo	11.1	homo	maternal&paternal	2	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Global developmental delay, Abnormal corpus callosum morphology, Agenesis of corpus callosum, Generalized tonic-clonic seizures, Hypoplasia of the corpus callosum, Generalized myoclonic seizures, Generalized-onset seizure, Atonic seizures, Epileptic spasms
ANKRD6	NM_001242809.1:c.1667C>Tp.(Pro556Leu)		AD_denovo	5.1	het	de novo	1	NDD	Dandy-Walker malformation, Omphalocele, Occipital encephalocele, Meningocele
MORC4	NM_024657.4:c.1382A>Gp.(Tyr461Cys)		XL	5.1	hemi	maternal	2	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizures, Global developmental delay, Abnormal corpus callosum morphology, Agenesis of corpus callosum, Generalized tonic-clonic seizures, Hypoplasia of the corpus callosum, Generalized myoclonic seizures, Generalized-onset seizure, Atonic seizures, Epileptic spasms
PAM	NM_001319943.1:c.1670C>Gp.(Ser557Trp)		AR_homo	6.5	homo	maternal&paternal	2	NDD	Strabismus, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Generalized hypotonia, Intellectual disability, moderate, Intellectual disability, severe
MYO9B	NM_001130065.1:c.248C>Tp.(Ser83Leu)	NM_001130065.1:c.5020G>Ap.(Val1674Met)	AR_comphet	4.5	comphet	maternal&paternal	2	NDD	Strabismus, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Generalized hypotonia, Intellectual disability, moderate, Intellectual disability, severe
CSNK1A1	NM_001025105.2:c.686G>Ap.(Arg229Gln)		AD_denovo	7.7	het	de novo	3	NDD	Microcephaly, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia
HEPH	NM_138737.4:c.812_814delp.(Pro271del)		XL	3.9	hemi	maternal	3	NDD	Microcephaly, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia
DNHD1	NM_144666.2:c.2758A>Gp.(Ser920Gly)	NM_144666.2:c.2546G>Ap.(Arg849Gln)	AR_comphet	3.7	comphet	maternal&paternal	3	NDD	Microcephaly, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia
RADIL	NM_018059.4:c.1450C>Tp.(Gln484*)		AR_homo	7.5	homo	maternal&paternal	1	NDD	recurrent hypoglycemia, microcephaly, hypopituitarism
PHACTR3	NM_001199505.1:c.17G>Tp.(Gly6Val)		AD_denovo	5.5	het	de novo	1	NDD	Intellectual disability, Global developmental delay
PTBP1	NM_002819.4:c.144A>Tp.(Lys48Asn)		AD_denovo	8.3	het	de novo	2	NDD	Cleft palate, Cleft soft palate, Thickened nuchal skin fold, Intellectual disability, Global developmental delay, Small for gestational age, Short stature, Cleft hard palate
TNR	NM_003285.2:c.3659C>Tp.(Ser1220Phe)	NM_003285.2:c.496A>Gp.(Thr166Ala)	AR_comphet	5.2	comphet	maternal&paternal	2	NDD	Cleft palate, Cleft soft palate, Thickened nuchal skin fold, Intellectual disability, Global developmental delay, Small for gestational age, Short stature, Cleft hard palate
MAB21L4	NM_001085437.2:c.755A>Gp.(Tyr252Cys)		AD_denovo	3.8	het	de novo	1	NDD	Abnormality of dental enamel, Autistic behavior, Delayed speech and language development, Global developmental delay, Motor delay, Sleep disturbance, Poor coordination
NAV2	NM_001244963.1:c.2486C>Tp.(Pro829Leu)	NM_001244963.1:c.7137+3G>Ap.?	AR_comphet	5.4	comphet	maternal&paternal	1	NDD	Astigmatism, Hypermetropia, Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Dandy-Walker malformation, Cerebellar hypoplasia, Delayed gross motor development, Enlarged cisterna magna, Scoliosis, High hypermetropia, Intellectual disability, severe, Mild global developmental delay, Cognitive impairment, Hernia, Mild hypermetropia

MED14	NM_004229.3:c.3657T>Gp.(His1219Gln)		XL	4.0	hemi	maternal	1	NDD + Epilepsy	Autistic behavior, Intellectual disability, Seizures, Global developmental delay, Intellectual disability, severe, Severe global developmental delay, Epileptic encephalopathy
MYRIP	NM_001284423.1:c.1525G>Ap.(Asp509Asn)	NM_001284423.1:c.2419C>Tp.(Pro807Ser)	AR_comphet	4.3	comphet	maternal&paternal	1	NDD + Epilepsy	Hearing impairment, Delayed speech and language development, Atopic dermatitis, Intellectual disability, Seizures, Motor delay, Pachygyria, Lissencephaly, Bradykinesia, Dysdiadochokinesis, Orofacial dyskinesia, Poor speech, Scoliosis, Aspiration, Thoracic scoliosis, Thoracolumbar scoliosis, Lumbar scoliosis, Allergy
ZNF692	NM_001136036.2:c.70C>Gp.(Gln24Glu)		AD_denovo	5.4	het	de novo	2	NDD + Epilepsy	Seizures, Global developmental delay, Generalized-onset seizure, Periventricular leukomalacia
FAT1	NM_005245.3:c.11017G>Cp.(Val3673Leu)	NM_005245.3:c.6079C>Tp.(Arg2027Cys)	AR_comphet	6.0	comphet	maternal&paternal	2	NDD + Epilepsy	Seizures, Global developmental delay, Generalized-onset seizure, Periventricular leukomalacia
PAPOLG	NM_022894.3:c.533C>Gp.(Ser178*)		AD_denovo	9.2	het	de novo	4	NDD + Epilepsy	Seizures, Generalized-onset seizure
SCN11A	NM_014139.2:c.95C>Tp.(Ala32Val)	NM_014139.2:c.2821G>Ap.(Glu941Lys)	AR_comphet	6.1	comphet	maternal&paternal	4	NDD + Epilepsy	Seizures, Generalized-onset seizure
HSD17B6	NM_003725.3:c.440G>Ap.(Ser147Asn)		AD_denovo	6.0	het	de novo	4	NDD + Epilepsy	Seizures, Generalized-onset seizure
XDH	NM_000379.3:c.2559G>Cp.(Lys853Asn)		AD_denovo	6.3	het	de novo	4	NDD + Epilepsy	Seizures, Generalized-onset seizure
FYTTD1	NM_032288.6:c.755G>Cp.(Arg252Pro)		AD_denovo	6.5	het	de novo	1	NDD	Microcephaly, Nystagmus, Impaired social interactions, Intellectual disability, Muscular hypotonia, Global developmental delay, EEG abnormality
ARMCX1	NM_016608.1:c.520dup, p.(Arg174Profs*3)		XL	6.6	hemi	maternal	3	NDD + Epilepsy	Autistic behavior, Delayed speech and language development, Intellectual disability, Seizures, Global developmental delay, EEG abnormality, Poor fine motor coordination, Delayed social development, Cognitive impairment
ARFGEF	NM_020340.4:c.787G>Ap.(Ala263Thr)		AD_denovo	7.0	het	de novo	3	NDD + Epilepsy	Autistic behavior, Delayed speech and language development, Intellectual disability, Seizures, Global developmental delay, EEG abnormality, Poor fine motor coordination, Delayed social development, Cognitive impairment
DMRT3	NM_021240.3:c.917C>Tp.(Ala306Val)		AD_denovo	5.4	het	de novo	3	NDD + Epilepsy	Autistic behavior, Delayed speech and language development, Intellectual disability, Seizures, Global developmental delay, EEG abnormality, Poor fine motor coordination, Delayed social development, Cognitive impairment
CFAP74	NM_001304360.1:c.3409delp.(Gln1137Argfs*37)		AD_denovo	6.0	het	de novo	1	NDD + Epilepsy	Delayed speech and language development, Intellectual disability, Seizures, Global developmental delay, Absence seizure, Generalized-onset seizure, EEG abnormality, Developmental regression, Poor speech
H1-10	NM_006026.3:c.80C>Tp.(Ser27Leu)		AR_homo	3.5	homo	maternal&paternal	2	NDD	Retinal dystrophy, Microphthalmia, Delayed speech and language development, Global developmental delay, Poor speech, Vitreoretinopathy, Congenital blindness
DNHD1	NM_144666.2:c.3410G>Ap.(Arg1137Gln)	NM_144666.2:c.2450A>Cp.(His817Pro)	AR_comphet	3.6	comphet	maternal&paternal	2	NDD	Retinal dystrophy, Microphthalmia, Delayed speech and language development, Global developmental delay, Poor speech, Vitreoretinopathy, Congenital blindness
MRO	NM_001127176.1:c.550T>Ap.(Phe184Ile)		AR_homo	6.3	homo	maternal&paternal	1	NDD + Epilepsy	Absent speech, Obesity, Intellectual disability, severe, Epilepsy
RIC8B	NM_001330145.1:c.399G>Cp.(Gln133His)		AD_denovo	6.1	het	de novo	3	Neuro	Sudden spastic of lower extremitiesa and bowel incontinence at the age of 43 years
SLC25A14	NM_001282197.1:c.124G>Cp.(Val42Leu)		XL	5.7	hemi	maternal	3	Neuro	Sudden spastic of lower extremitiesa and bowel incontinence at the age of 43 years
TRPC7	NM_020389.2:c.1577A>Gp.(Tyr526Cys)		AR_homo	3.7	homo	maternal&paternal	3	Neuro	Sudden spastic of lower extremitiesa and bowel incontinence at the age of 43 years
PAPSS1	NM_005443.4:c.1672G>Ap.(Val558Ile)		AR_homo	5.3	homo	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Cerebellar vermis atrophy, Cognitive impairment
PDE4DIP	NM_001198834.3:c.5842A>Gp.(Lys1948Glu)	NM_001198834.3:c.4063C>Tp.(Arg1355*)	AR_comphet	6.2	comphet	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Cerebellar vermis atrophy, Cognitive impairment
TAF5	NM_006951.4:c.479C>Tp.(Ala160Val)		AR_homo	4.9	homo	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, Seizures, Global developmental delay, Cerebellar vermis atrophy, Cognitive impairment

JPH4	NM_001146028.1:c.953_956delp.(Gly318Alafs*53)		AD_denovo	10.2	het	de novo	1	NDD	Microcephaly, Autism, Intellectual disability, Muscular hypotonia, Global developmental delay
HSPA8	NM_006597.5:c.98A>Gp.(Gln33Arg)		AD_denovo	8.9	het	de novo	1	NDD + Epilepsy	seizures, focal seizures, myoclonic seizures
BAZ1B	NM_032408.3:c.461G>Ap.(Gly154Asp)		AD_denovo	9.7	het	de novo	1	NDD + Epilepsy	absence epilepsy, EEG abnormality
ADARB2	NM_018702.3:c.1570G>Ap.(Glu524Lys)	NM_018702.3:c.914G>Ap.(Ser305Asn)	AR_comphet	6.2	comphet	maternal&paternal	2	NDD	Microcephaly, Hearing impairment, Autism, Intellectual disability, Spasticity, Global developmental delay, Cerebral calcification
DISP1	NM_032890.3:c.1357A>Cp.(Met453Leu)	NM_032890.3:c.3233G>Ap.(Arg1078His)	AR_comphet	7.3	comphet	maternal&paternal	2	NDD	Cleft palate, Panhypopituitarism, Intellectual disability, Patent ductus arteriosus, Facial cleft, Scoliosis, Short stature, Median cleft lip and palate
UNC79	NM_020818.4:c.3857-691A>Gp.(=)	NM_020818.4:c.1547C>Tp.(Ser516Leu)	AR_comphet	5.3	comphet	maternal&paternal	2	NDD	Cleft palate, Panhypopituitarism, Intellectual disability, Patent ductus arteriosus, Facial cleft, Scoliosis, Short stature, Median cleft lip and palate
GABRG1	NM_173536.3:c.487A>Gp.(Thr163Ala)		AD_denovo	7.7	het	de novo	2	NDD	Strabismus, Autism, Ataxia, Specific learning disability, Gait ataxia, Language impairment, Pain insensitivity, Abnormality of movement, Motor tics, Dyskinesia, Exodeviation
ARFGEF3	NM_020340.4:c.5123+2T>Cp.?		AD_inherited	7.3	het	maternal	2	NDD	Strabismus, Autism, Ataxia, Specific learning disability, Gait ataxia, Language impairment, Pain insensitivity, Abnormality of movement, Motor tics, Dyskinesia, Exodeviation
ZHX1	NM_001017926.2:c.179A>Gp.(Asn60Ser)	NM_001017926.2:c.962C>Tp.(Ala321Val)	AR_comphet	3.4	comphet	maternal&paternal	2	NDD + Epilepsy	Hearing impairment, Visual impairment, Nystagmus, Seizures, Abnormality of the cerebrospinal fluid, Epileptic spasms, Abnormal CSF glucose level
CEMIP2	NM_013390.2:c.2648G>Ap.(Ser883Asn)	NM_013390.2:c.1204+6C>Tp.?	AR_comphet	2.8	comphet	maternal&paternal	2	NDD + Epilepsy	Hearing impairment, Visual impairment, Nystagmus, Seizures, Abnormality of the cerebrospinal fluid, Epileptic spasms, Abnormal CSF glucose level
PRPF6	NM_012469.3:c.67C>Tp.(Arg23Trp)		AD_denovo	9.1	het	de novo	1	NDD	Visual impairment, Intellectual disability, Growth delay, Mildly reduced visual acuity, Feeding difficulties
SOX7	NM_031439.3:c.723G>Ap.(Pro241=)		AD_denovo	4.0	het	de novo	2	NDD	Microcephaly, Hearing impairment, Autism, Intellectual disability, Spasticity, Global developmental delay, Cerebral calcification
KCTD16	NM_020768.3:c.1231T>Cp.(Phe411Leu)		AD_denovo	6.2	het	de novo	3	NDD + Epilepsy	Therapy-resistant epilepsy since the age of two, Epileptic encephalopathy
MST1	NM_020998.3:c.1603C>Gp.(Arg535Gly)		AD_denovo	5.5	het	de novo	2	Neuro	Migraine, Migraine with aura, Migraine without aura, Cortical dysplasia, Frontoparietal cortical dysplasia
AKAP13	NM_006738.5:c.914A>Gp.(Gln305Arg)	NM_006738.5:c.8228A>Cp.(Lys2743Thr)	AR_comphet	5.4	comphet	maternal&paternal	2	Neuro	Migraine, Migraine with aura, Migraine without aura, Cortical dysplasia, Frontoparietal cortical dysplasia
ABCC12	NM_033226.2:c.796G>Ap.(Gly266Arg)	NM_033226.2:c.442delp.(Ile148Serfs*20)	AR_comphet	5.7	comphet	maternal&paternal	3	NDD + Epilepsy	Therapy-resistant epilepsy since the age of two, Epileptic encephalopathy
LRCH2	NM_020871.3:c.2141A>Gp.(Asn714Ser)		XL	4.1	hemi	maternal	3	NDD + Epilepsy	Therapy-resistant epilepsy since the age of two, Epileptic encephalopathy
WARS1	NM_173701.1:c.397C>Tp.(Arg133Cys)		AR_homo	8.7	homo	maternal&paternal	2	NDD	Microcephaly, Delayed speech and language development, Intellectual disability, Global developmental delay, Absent speech, Intellectual disability, profound, Intellectual disability, moderate, Poor speech, Inability to walk, Melanoma, Intellectual disability, severe
CSTF2	NM_001306206.1:c.724G>Ap.(Ala242Thr)		XL	5.6	hemi	maternal	2	NDD	Microcephaly, Delayed speech and language development, Intellectual disability, Global developmental delay, Absent speech, Intellectual disability, profound, Intellectual disability, moderate, Poor speech, Inability to walk, Melanoma, Intellectual disability, severe

TANK	NM_001199135.1:c.1012T>Cp.(Tyr338His)		AD_denovo	6.2	het	de novo	1	NDD + Epilepsy	Restlessness, Single transverse palmar crease, Seizures, Global developmental delay, Abnormal corpus callosum morphology, Abnormality of neuronal migration, Abnormality of the periventricular white matter, Infantile spasms
TTC28	NM_001145418.1:c.3020A>Gp.(Tyr1007Cys)		AD_unknown	6.6	het	unknown	1	NDD	Tall stature, Macrocephaly, Autistic behavior, Delayed speech and language development, Intellectual disability, Global developmental delay, Obesity, Abnormal social behavior
TNN	NM_022093.1:c.1949A>Tp.(Tyr650Phe)	NM_022093.1:c.2852T>Gp.(Val951Gly)	AR_comphet	4.5	comphet	maternal& paternal	1	NDD + Epilepsy	infantile spasms since 6 months of age, conspicuous odor, crying phases, failure to thrive
TKT	NM_001135055.2:c.1751T>Cp.(Val584Ala)		AD_denovo	8.5	het	de novo	3	NDD	Global developmental delay, Motor delay
RASAL2	NM_004841.3:c.433G>Tp.(Glu145*)		AD_denovo	7.9	het	de novo	3	NDD	Global developmental delay, Motor delay
HSPB7	NM_014424.4:c.202C>Tp.(Arg68Cys)		AD_denovo	5.1	het	de novo	3	NDD	Global developmental delay, Motor delay
GNL3L	NM_001184819.1:c.884T>Ap.(Leu295Gln)		XL	3.1	hemi	maternal	1	NDD	Global developmental delay with delayed speech and language development and a suspected autism spectre disorder, makrosomia
SETD1B	NM_015048.1:c.3074G>Ap.(Arg1025Gln)	NM_015048.1:c.4354C>Tp.(Arg1452Cys)	AR_comphet	6.3	comphet	maternal& paternal	4	NDD	Delayed speech and language development, Neurological speech impairment, Language impairment, Poor speech
TFE3	NM_006521.5:c.566A>Gp.(Tyr189Cys)		AD_denovo	7.5	hemi	de novo	1	NDD + Epilepsy	Microcephaly, Myopia, Delayed speech and language development, Abnormality of the thumb, Intellectual disability, Seizures, Intellectual disability, mild, Spasticity, Global developmental delay, Mental deterioration, Motor delay, Absent speech, Hip dysplasia, Obesity, Small for gestational age, Short nail, Broad nail, Abnormal facial shape, Generalized tonic-clonic seizures, Generalized myoclonic seizures, Intellectual disability, profound, Hepatomegaly, Intellectual disability, moderate, EEG abnormality, Poor speech, Mild short stature, Short stature, Increased body weight, Precocious puberty in males, Moderately short stature, Generalized tonic seizures, Intellectual disability, severe, Epileptic spasms, Myoclonic atonic seizures, Broad thumb, Cerebral palsy, Cognitive impairment
HYDIN	NM_001270974.2:c.6271A>Cp.(Ile2091Leu)		AD_denovo	6.9	het	de novo	4	NDD	Delayed speech and language development, Neurological speech impairment, Language impairment, Poor speech
MMS22L	NM_198468.2:c.2679+1G>Ap.?	NM_198468.2:c.268A>Gp.(Arg90Gly)	AR_comphet	5.4	comphet	maternal& paternal	4	NDD	Delayed speech and language development, Neurological speech impairment, Language impairment, Poor speech
CHD6	NM_032221.4:c.1678C>Ap.(Gln560Lys)	NM_032221.4:c.2224A>Gp.(Arg742Gly)	AR_comphet	6.6	comphet	maternal& paternal	4	NDD	Delayed speech and language development, Neurological speech impairment, Language impairment, Poor speech
ZNF804A	NM_194250.1:c.1049delp.(Gly350Valfs*7)		AR_homo	11.2	homo	maternal& paternal	1	NDD	High palate, Aggressive behavior, Autistic behavior, Intellectual disability, Global developmental delay, Hepatosplenomegaly, Protuberant abdomen, Abnormal facial shape, Muscular hypotonia of the trunk, Infantile muscular hypotonia, Low levels of vitamin D, Self-injurious behavior, Decreased serum iron
GIPR	NM_000164.3:c.784C>Gp.(Leu262Val)	NM_000164.3:c.393G>Tp.(Arg131Ser)	AR_comphet	4.0	comphet	maternal& paternal	1	NDD	Absent speech, Obesity, Intellectual disability, severe
TENM2	NM_001122679.1:c.3881C>Gp.(Ser1294Cys)		AD_unknown	4.5	het	unknown	1	NDD + Epilepsy	atonic-astatic seizures and mild intellectual disability
KCTD8	NM_198353.2:c.82G>Cp.(Ala28Pro)		AD_denovo	5.4	het	de novo	1	NDD	Regressive global developmental delay with intellectual disability, attention deficit disorder, dysplasia of the corpus callosum, obesity grade 1
TIAM2	NM_012454.3:c.4679_4681dup, p.(Asn1560_Leu1561insHis)		AR_homo	5.7	homo	maternal& paternal	4	NDD	Intellectual disability, microcephaly, scoliosis, short stature and hip dysplasia
CASZ1	NM_001079843.2:c.4004G>Ap.(Arg1335His)		AR_homo	5.9	homo	maternal& paternal	4	NDD	Intellectual disability, microcephaly, scoliosis, short stature and hip dysplasia

PLEKHB1	NM_021200.2:c.164A>Cp.(His55Pro)		AR_homo	6.7	homo	maternal&paternal	4	NDD	Intellectual disability, microcephaly, scoliosis, short stature and hip dysplasia
BARX2	NM_003658.4:c.386G>Ap.(Arg129Gln)		AR_homo	6.7	homo	maternal&paternal	4	NDD	Intellectual disability, microcephaly, scoliosis, short stature and hip dysplasia
SKOR2	NM_001278063.1:c.2752+1G>Tp.?		AR_homo	9.0	homo	unknown	3	NDD	Short stature, microcephaly, mild intellectual disability, hyperopia
FMNL3	NM_175736.4:c.2575C>Tp.(Arg859Trp)		AR_homo	6.1	homo	unknown	3	NDD	Short stature, microcephaly, mild intellectual disability, hyperopia
ARHGEF10L	NM_018125.3:c.354_355delCCinsTTp.(Arg119Trp)		AR_homo	6.0	homo	maternal&paternal	1	NDD + Epilepsy	Seizures, Ataxia, Spasticity, Focal clonic seizures, Myoclonic spasms, Generalized dystonia, Focal-onset seizure, Focal myoclonic seizures
SPTB	NM_001024858.2:c.610G>Ap.(Asp204Asn)	NM_001024858.2:c.5063A>Gp.(Asn1688Ser)	AR_comphet	5.2	comphet	maternal&paternal	1	NDD	Global developmental delay, Leukopenia, Leukemia, Acute lymphoblastic leukemia
USP13	NM_003940.2:c.2498+1G>Ap.?		AD_denovo	6.4	het	de novo	1	NDD	Renal dysplasia, Polycystic kidney dysplasia, Synophrys, Global developmental delay
SNX8	NM_013321.3:c.922C>Tp.(Gln308*)		AD_denovo	B	het	de novo	2	Growth, Skeletal	Growth delay, short stature, intrauterine growth retardation, Silver-Russell-like appearance
ZNF449	NM_152695.5:c.1394G>Ap.(Cys465Tyr)		AD_denovo	B	het	de novo	2	Growth, Skeletal	Growth delay, short stature, intrauterine growth retardation, Silver-Russell-like appearance
MAGED1	NM_001005332.1:c.640A>Gp.(Thr214Ala)		XL	4.7	hemi	maternal	1	NDD	Early onset autism
SLC38A1	NM_001278390.1:c.529A>Gp.(Ile177Val)		AD_unknown	5.5	het	unknown	1	Neuro	Seizure, Tremor, Hand tremor, Nevus, Focal-onset seizure, Abnormality of brain morphology
ZSCAN10	NM_032805.2:c.1436C>Ap.(Ser479Tyr)	NM_032805.2:c.2245G>Tp.(Ala749Ser)	AR_comphet	3.5	comphet	maternal&paternal	3	NDD + Epilepsy	Seizure, Global developmental delay, Gait ataxia, Bilateral tonic-clonic seizure, Unsteady gait, Focal-onset seizure, Cognitive impairment, Mild malformation of cortical development
FLYWCH1	NM_001308068.1:c.2112-3T>Gp.?	NM_001308068.1:c.1111A>Tp.(Ser371Cys)	AR_comphet	6.0	comphet	maternal&paternal	3	NDD + Epilepsy	Seizure, Global developmental delay, Gait ataxia, Bilateral tonic-clonic seizure, Unsteady gait, Focal-onset seizure, Cognitive impairment, Mild malformation of cortical development
HEPHL1	NM_001098672.1:c.1097G>Ap.(Cys366Tyr)		AD_denovo	C	het	de novo	2	Connective Tissue	Syncope, Joint hypermobility, Recurrent fractures, Chronic pain, Dysesthesia
COG6	NM_020751.2:c.1209T>Gp.(Ile403Met)		AD_denovo	B	het	de novo	2	Connective Tissue	Syncope, Joint hypermobility, Recurrent fractures, Chronic pain, Dysesthesia
ZBTB34	NM_001099270.1:c.18delp.(Phe6Leufs*14)		AD_denovo	8.2	het	de novo	1	NDD + Epilepsy	Delayed speech and language development, Global developmental delay, Focal-onset seizure, Childhood onset
PODN	NM_001199080.2:c.559-1G>Cp.?		AD_denovo	B	het	de novo	2	Growth, Skeletal	Joint hypermobility, Asymmetry of the thorax, Scoliosis
GORAB	NM_152281.2:c.383T>Cp.(Ile128Thr)		AD_denovo	C	het	de novo	2	Growth, Skeletal	Joint hypermobility, Asymmetry of the thorax, Scoliosis
GIT2	NM_057169.4:c.699T>Gp.(Tyr233*)		AD_denovo	A	het	de novo	1	Growth, Skeletal	Failure to thrive, Small for gestational age, Short stature, Decreased body weight, Attention deficit hyperactivity disorder, Focal-onset seizure, Abnormal growth hormone level
NDST1	NM_001543.4:c.2468G>Ap.(Gly823Glu)		AD_denovo	8.6	het	de novo	1	NDD + Epilepsy	Focal seizures with cyanosis, sec. generalizing, EEG highly pathological, so far no cMRI examination has been carried out
TTC3	NM_001320703.1:c.3970G>Ap.(Glu1324Lys)		AD_denovo	5.2	het	de novo	1	NDD	Abnormality of the kidney, Global developmental delay, Hip dysplasia, Short stature
ASXL2	NM_018263.4:c.1894C>Gp.(His632Asp)		AD_denovo	8.0	het	de novo	2	NDD	Seizures, Generalized tonic-clonic seizures, Myoclonic atonic seizures, Epileptic encephalopathy
TBCCD1	NM_001134415.1:c.1392T>Gp.(Cys464Trp)		AD_denovo	B	het	de novo	3	Metabons	Ketotic hypoglycemia
MRM3	NM_018146.3:c.173C>Gp.(Pro58Arg)		AD_denovo	B	het	de novo	3	Metabons	Ketotic hypoglycemia
PACIN3	NM_001184974.1:c.604-3C>Gp.?		AD_denovo	B	het	de novo	3	Metabons	Ketotic hypoglycemia
MDN1	NM_014611.2:c.13276C>Gp.(Leu4426Val)		AD_denovo	6.8	het	de novo	2	NDD + Epilepsy	Microcephaly, Seizure, Dystonia, Cerebral palsy, Abnormality of movement, Epileptic encephalopathy
MAP7D1	NM_018067.4:c.1225G>Tp.(Ala409Ser)		AR_homo	3.5	homo	maternal&paternal	1	NDD + Epilepsy	Infantile febrile seizures and tonic-clonic seizures with aura, despite current treatment with valproate, seizures continue
CPLX1	NM_006651.3:c.250dup, p.(Ala84Glyfs*256)		AD_unknown	9.3	het	unknown	3	NDD + Epilepsy	Global developmental delay and obsessive-compulsive behavior, seizures
HEATR1	NM_018072.5:c.3949-26_3954delp.(Asp1317Valfs*827)		AD_unknown	6.9	het	unknown	3	NDD + Epilepsy	Global developmental delay and obsessive-compulsive behavior, seizures

HS6ST2	NM_001077188.1:c.853T>Gp.(Trp285Gly)		XL	5.6	hemi	maternal	1	NDD	global developmental delay, focal epilepsy, absent speech, Delayed gross motor development, Tetraparesis, Facial palsy
USP4	NM_003363.3:c.1748A>Gp.(Tyr583Cys)		AR_homo	A	homo	maternal&paternal	2	Metabolism	Myalgia, Hyperlipoproteinemia, Increased erythrocyte protoporphyrin concentration, Angioedema
DNHD1	NM_144666.2:c.7549C>Tp.(Arg2517Cys)	NM_144666.2:c.2104-4T>Ap.?	AR_comphet	4.2	comphet	maternal&paternal	2	NDD + Epilepsy	Microcephaly, Seizure, Dystonia, Cerebral palsy, Abnormality of movement, Epileptic encephalopathy
FBN3	NM_032447.4:c.7780G>Ap.(Val2594Ile)	NM_032447.4:c.1135C>Tp.(Arg379*)	AR_comphet	C	comphet	maternal&paternal	3	Metabolism	Obesity, Increased adipose tissue, Glioma, Class III obesity, Overweight, Brain neoplasm
SDR42E1	NM_145168.2:c.4G>Ap.(Asp2Asn)		AR_homo	C	homo	maternal&paternal	3	Metabolism	Obesity, Increased adipose tissue, Glioma, Class III obesity, Overweight, Brain neoplasm
FADS1	NM_013402.4:c.247G>Tp.(Ala83Ser)		AD_denovo	A	het	de novo	2	other	Anemia, Fever, Recurrent fever, Refractory anemia
TPR	NM_003292.2:c.1038A>Gp.(Ile346Met)	NM_003292.2:c.2380T>Ap.(Ser794Thr)	AR_comphet	C	comphet	maternal&paternal	2	other	Anemia, Fever, Recurrent fever, Refractory anemia
ZNF449	NM_152695.5:c.961A>Tp.(Lys321*)		AD_denovo	6.6	hemi	de novo	1	NDD + Epilepsy	Hypothyroidism, Primary hypothyroidism, Congenital hypothyroidism, Seizure, Generalized-onset seizure, Atonic seizure, Focal emotional seizure with laughing, Clonic seizure
DOHH	NM_001145165.1:c.446C>Gp.(Pro149Arg)	NM_001145165.1:c.224T>Gp.(Val75Gly)	AR_comphet	6.8	comphet	maternal&paternal	1	NDD + Epilepsy	Global developmental delay, Epilepsy since the age of 3 with tonic-clonic seizures, EEG abnormalities, pain insensitivity
ABCB10	NM_012089.2:c.833_838delp.(Asp278_Thr279deleted)		AD_denovo	4.8	het	de novo	1	NDD	Renal duplication, Global developmental delay, Annular pancreas, Esophageal atresia, Duodenal atresia, Tracheoesophageal fistula, Short stature, Partially duplicated kidney, Anorectal anomaly, Duodenal stenosis, Rectovestibular fistula
DLGAP1	NM_004746.3:c.1018C>Tp.(Arg340*)		AD_denovo	11.8	het	de novo	4	NDD	Renal duplication, Autism, Autistic behavior, Delayed speech and language development, Intellectual disability, Poor speech
DAAM2	NM_001201427.1:c.1339C>Gp.(Gln447Glu)	NM_001201427.1:c.1745C>Ap.(Pro582His)	AR_comphet	4.7	comphet	maternal&paternal	4	NDD	Renal duplication, Autism, Autistic behavior, Delayed speech and language development, Intellectual disability, Poor speech
SIGLEC9	NM_001198558.1:c.682G>Ap.(Val228Ile)		AD_denovo	3.7	het	de novo	4	NDD	Renal duplication, Autism, Autistic behavior, Delayed speech and language development, Intellectual disability, Poor speech
CDH13	NM_001220488.1:c.2228G>Ap.(Arg743His)	NM_001220488.1:c.1505C>Tp.(Ser502Phe)	AR_comphet	5.2	comphet	maternal&paternal	4	NDD	Renal duplication, Autism, Autistic behavior, Delayed speech and language development, Intellectual disability, Poor speech
ASIC1	NM_020039.3:c.1116T>Ap.(Tyr372*)		AD_unknown	6.1	het	unknown	1	NDD + Epilepsy	Behavioral abnormality, Autistic behavior, Delayed speech and language development, Seizure, Pyloric stenosis, Attention deficit hyperactivity disorder
SSPOP	NM_198455.2:c.1280T>Cp.(Met427Thr)	NM_198455.2:c.1997G>Ap.(Arg666His)	AR_comphet	2.6	comphet	maternal&paternal	1	NDD + Epilepsy	Global developmental delay, Incoordination, Poor coordination, Focal-onset seizure, Epileptic encephalopathy
PKHD1L1	NM_177531.4:c.5194C>Tp.(Pro1732Ser)	NM_177531.4:c.8005C>Tp.(Gln2669*)	AR_comphet	5.0	comphet	maternal&paternal	2	NDD + Epilepsy	Seizure, Status epilepticus, EEG abnormality, Focal impaired awareness seizure, Focal-onset seizure, EEG with focal spike waves

LAMA5	NM_005560.4:c.8632G>Ap.(Val2878Ile)	NM_005560.4:c.6578G>Ap.(Arg2193His)	AR_comphet	5.4	comphet	maternal&paternal	2	NDD + Epilepsy	Seizure, Status epilepticus, EEG abnormality, Focal impaired awareness seizure, Focal-onset seizure, EEG with focal spike waves
PRICKLE1	NM_153026.2:c.128A>Gp.(Glu43Gly)		AD_denovo	8.7	het	de novo	1	NDD + Epilepsy	Global developmental delay with a decreased and autistic spectrum disorder characteristics, attends a special school, MRI and EEG inconspicuous
CCDC66	NM_001141947.1:c.847_848delp.(Glu283Serfs*3)		AR_homo	8.0	homo	maternal&paternal	3	NDD + Epilepsy	Global developmental delay with delayed speech, astatic attacks, absence epilepsy and EEG abnormalities
CHMP3	NM_016079.3:c.220G>Ap.(Val74Met)		AD_denovo	5.9	het	de novo	3	NDD + Epilepsy	Global developmental delay with delayed speech, astatic attacks, absence epilepsy and EEG abnormalities
RGPD8	NM_001164463.1:c.3225G>Tp.(Gln1075His)		AD_denovo	4.6	het	de novo	3	NDD + Epilepsy	Global developmental delay with delayed speech, astatic attacks, absence epilepsy and EEG abnormalities
NUMBL	NM_004756.4:c.1193C>Ap.(Pro398His)		AR_homo	5.4	homo	maternal&paternal	2	NDD	Long palpebral fissure, Prominent fingertip pads, Intellectual disability, Large fleshy ears
ATP13A4	NM_032279.3:c.826G>Ap.(Glu276Lys)		AR_homo	6.0	homo	maternal&paternal	2	NDD	Long palpebral fissure, Prominent fingertip pads, Intellectual disability, Large fleshy ears
UBR5	NM_015902.5:c.3682C>Tp.(Pro1228Ser)		AD_denovo	8.9	het	de novo	1	NDD + Epilepsy	Epilepsy associated with fever or infection, tonic-clonic seizures, mild mental retardation, macrocephaly and sleep EEG with sharp slow waves
NPTN	NM_012428.3:c.1025C>Tp.(Pro342Leu)		AD_denovo	8.7	het	de novo	2	NDD	Autism, Delayed speech and language development, Intellectual disability, Global developmental delay, Diarrhea, Macrocephaly, Partial Epilepsy
LRRC2	NM_024512.4:c.412A>Gp.(Thr138Ala)	NM_024512.4:c.14T>Cp.(Val5Ala)	AR_comphet	2.8	comphet	maternal&paternal	2	NDD	Autism, Delayed speech and language development, Intellectual disability, Global developmental delay, Diarrhea, Macrocephaly, Partial Epilepsy
USP8	NM_001128610.2:c.2658+2_2658+3insAAGAp.?	NM_001128610.2:c.2371A>Gp.(Ile791Val)	AR_comphet	5.9	comphet	maternal&paternal	1	neuro	Spasticity, Intention tremor, Vertigo, Dyskinesia
VPS51	NM_013265.3:c.1777A>Gp.(Lys593Glu)		AD_denovo	6.7	het	de novo	3	NDD + Epilepsy	Epilepsy with generalized tonic-clonic seizures, ED 10/2019, microcephaly
RNF144A	NM_014746.4:c.428G>Cp.(Cys143Ser)		AR_homo	5.2	homo	maternal	3	NDD + Epilepsy	Epilepsy with generalized tonic-clonic seizures, ED 10/2019, microcephaly
SCN7A	NM_002976.3:c.2932A>Gp.(Ile978Val)		AR_homo	5.8	homo	maternal	3	NDD + Epilepsy	Epilepsy with generalized tonic-clonic seizures, ED 10/2019, microcephaly
UTP18	NM_016001.2:c.1503+1G>Ap.?		AD_denovo	6.2	het	de novo	2	NDD + Epilepsy	Epilepsy (post- brain haemorrhage condition), intelligence impairment, autism, seizures, premature birth
GCNA	NM_052957.4:c.673C>Ap.(Pro225Thr)		AD_denovo	4.1	het	de novo	2	NDD + Epilepsy	Epilepsy (post- brain haemorrhage condition), intelligence impairment, autism, seizures, premature birth
RYR3	NM_001036.4:c.2770A>Gp.(Thr924Ala)	NM_001036.4:c.11246-5C>Gp.?	AR_comphet	6.1	comphet	maternal&paternal	1	Neuro	Torticollis, Ataxia, Dysarthria, Dystonia, Slurred speech, Gait ataxia, Limb ataxia, Truncal ataxia, Episodic ataxia, Apraxia, Limb dystonia, Focal dystonia, Gait apraxia, Oromandibular dystonia
MTCH1	NM_001271641.1:c.2T>Ap.0?		AR_homo	6.6	homo	maternal&paternal	4	NDD	Deeply set eye, Intellectual disability, Failure to thrive, Narrow palpebral fissure
KCNG4	NM_172347.2:c.1022C>Tp.(Ala341Val)		AR_homo	5.8	homo	maternal&paternal	4	NDD	Deeply set eye, Intellectual disability, Failure to thrive, Narrow palpebral fissure
KIAA1107	NM_015237.3:c.299C>Tp.(Thr100Ile)		AR_homo	3.9	homo	maternal&paternal	4	NDD	Deeply set eye, Intellectual disability, Failure to thrive, Narrow palpebral fissure
CRYBG3	NM_153605.3:c.8492G>Ap.(Arg2831His)		AR_homo	4.8	homo	maternal&paternal	4	NDD	Deeply set eye, Intellectual disability, Failure to thrive, Narrow palpebral fissure
PDE4DIP	NM_001198834.3:c.6862A>Cp.(Lys2288Gln)	NM_001198834.3:c.6043A>Gp.(Ile2015Val)	AR_comphet	6.2	comphet	maternal&paternal	1	NDD	Intellectual disability, Global developmental delay, Motor delay, Failure to thrive, Increased serum lactate, Infantile muscular hypotonia, Delayed myelination, Alaninuria
TCEAL3	NM_001006933.1:c.585C>Gp.(His195Gln)		XL	3.7	hemi	maternal	1	NDD	Global developmental delay, Gait ataxia, Infantile muscular hypotonia
PLEKHM3	NM_001080475.2:c.2219G>Ap.(Arg740Lys)		AD_denovo	4.4	het	de novo	2	Neuro	Gait disturbance, Dystonia, Progressive spastic paraplegia, Paraplegia, Leg dystonia

GPX4	NM_001039848.3:c.587+5G>Ap.?	NM_001039848.3:c.475G>Tp.(Gly159Cys)	AR_comphet	5.8	comphet	maternal&paternal	2	Neuro	Gait disturbance, Dystonia, Progressive spastic paraplegia, Paraplegia, Leg dystonia
KLHDC4	NM_017566.3:c.908T>Cp.(Met303Thr)	NM_017566.3:c.529C>Tp.(Arg177Trp)	AR_comphet	3.6	comphet	maternal&paternal	1	NDD + Epilepsy	Neurodevelopmental delay, Global developmental delay, Infantile spasms, Seizure, Epileptic spasm, Abnormal nervous system physiology, Neonatal seizure
TDRD9	NM_153046.2:c.2273C>Tp.(Pro758Leu)		AR_homo	4.2	homo	maternal&paternal	2	NDD	Autism, Hypertrichosis, Intellectual disability, Global developmental delay, Absent speech, Mutism
PRSS35	NM_001170423.1:c.1231G>Tp.(Ala411Ser)	NM_001170423.1:c.632G>Ap.(Ser211Asn)	AR_comphet	2.6	comphet	maternal&paternal	2	NDD	Autism, Hypertrichosis, Intellectual disability, Global developmental delay, Absent speech, Mutism
TEC	NM_003215.2:c.1526G>Tp.(Gly509Val)		AD_inherited	C	het	maternal	2	immunology	recurrent purulent abscess of the groin
RAB11FIP4	NM_032932.5:c.1562G>Ap.(Gly521Asp)		AD_inherited	C	het	maternal	2	immunology	recurrent purulent abscess of the groin
ITSN1	NM_003024.2:c.1690T>Cp.(Ser564Pro)		AD_unknown	6.6	het	unknown	5	NDD + Epilepsy	Macrocephaly, Aggressive behavior, Delayed speech and language development, Enuresis, Intellectual disability, Seizure, Global developmental delay, Intellectual disability, moderate, Febrile seizure (within the age range of 3 months to 6 years), Polyphagia, Increased adipose tissue, Feeding difficulties, Cognitive impairment, Overweight
DYNC111	NM_004411.4:c.1421C>Gp.(Ala474Gly)		AD_unknown	4.5	het	unknown	5	NDD + Epilepsy	Macrocephaly, Aggressive behavior, Delayed speech and language development, Enuresis, Intellectual disability, Seizure, Global developmental delay, Intellectual disability, moderate, Febrile seizure (within the age range of 3 months to 6 years), Polyphagia, Increased adipose tissue, Feeding difficulties, Cognitive impairment, Overweight
TMEM63A	NM_014698.2:c.1423T>Cp.(Phe475Leu)		AD_unknown	3.5	het	unknown	5	NDD + Epilepsy	Macrocephaly, Aggressive behavior, Delayed speech and language development, Enuresis, Intellectual disability, Seizure, Global developmental delay, Intellectual disability, moderate, Febrile seizure (within the age range of 3 months to 6 years), Polyphagia, Increased adipose tissue, Feeding difficulties, Cognitive impairment, Overweight
SLC22A23	NM_015482.1:c.1076A>Gp.(Tyr359Cys)		AD_unknown	2.8	het	unknown	5	NDD + Epilepsy	Macrocephaly, Aggressive behavior, Delayed speech and language development, Enuresis, Intellectual disability, Seizure, Global developmental delay, Intellectual disability, moderate, Febrile seizure (within the age range of 3 months to 6 years), Polyphagia, Increased adipose tissue, Feeding difficulties, Cognitive impairment, Overweight
MTR	NM_000254.2:c.2812A>Gp.(Ser938Gly)		AD_unknown	6.2	het	unknown	5	NDD + Epilepsy	Macrocephaly, Aggressive behavior, Delayed speech and language development, Enuresis, Intellectual disability, Seizure, Global developmental delay, Intellectual disability, moderate, Febrile seizure (within the age range of 3 months to 6 years), Polyphagia, Increased adipose tissue, Feeding difficulties, Cognitive impairment, Overweight
HNRNPM	NM_005968.4:c.23C>Tp.(Ala8Val)		AR_homo	5.0	homo	maternal&paternal	2	NDD + Epilepsy	generalized epilepsy with nocturnal tonic-clonic seizures (onset in the 2nd year of life), mild intellectual impairment
LRRC7	NM_001330635.1:c.2143C>Tp.(Gln715*)		AD_unknown	7.5	het	unknown, not maternal	1	NDD	Intellectual disability, Global developmental delay, Overweight
DUSP9	NM_001318503.1:c.745G>Ap.(Asp249Asn)		XL	3.8	hemi	maternal	2	NDD + Epilepsy	generalized epilepsy with nocturnal tonic-clonic seizures (onset in the 2nd year of life), mild intellectual impairment
SLC4A2	NM_003040.3:c.2507T>Cp.(Ile836Thr)		AD_denovo	7.0	het	de novo	1	NDD + Epilepsy	Global developmental delay with intelligence impairment and speech delay; epilepsy with tonic-clonic seizures and atypical absences (pseudo-Lennox); short stature; hypercholesterinemia
UTP14A	NM_006649.3:c.124A>Gp.(Lys42Glu)		XL	4.1	hemi	maternal	2	NDD + Epilepsy	Epileptic encephalopathy, Seizure since the age of 11
SMURF1	NM_020429.2:c.1390C>Tp.(Gln464*)		AD_denovo	9.6	het	de novo	2	NDD	Premature infant (32 weeks, 1600g), maldescensus testis bilateral, plagiocephalus, central motor coordination and movement disorder with dystonic movements, trunk muscular hypotension, delayed development, MRI: subependymal left heterotopia, steep tentorium, small posterior fossa, compressed 4th ventricle, flattened skull on the right

SPHK2	NM_001204159.2:c.1774delp.(His592Thrfs*19)		AD_denovo	6.8	het	de novo	2	NDD	Premature infant (32 weeks, 1600g), maldescensus testis bilateral, plagiocephalus, central motor coordination and movement disorder with dystonic movements, trunk muscular hypotension, delayed development, MRI: subependymal left heterotopia, steep tentorium, small posterior fossa, compressed 4th ventricle, flattened skull on the right
SF3A1	NM_005877.5:c.709C>Tp.(Arg237*)		AD_inherited	6.5	het	maternal	1	NDD	Neurodevelopmental delay, Bifid uvula, Global developmental delay, Atrial septal defect, Hypoglycemia, Abnormal blood glucose concentration, Mild global developmental delay, Moderate global developmental delay, Abnormal glycosylation
C7orf26	NM_024067.3:c.575_576insTp.(Ser193Ilefs*3)		AD_denovo	A	het	de novo	2	Immunology	Abnormal facial shape, Alopecia areata, Recurrent infections, Antinuclear antibody positivity, Paroxysmal nocturnal hemoglobinuria, Prolonged neonatal jaundice
POLR2C	NM_032940.2:c.109delp.(Val37Serfs*8)		AD_denovo	B	het	de novo	2	Immunology	Abnormal facial shape, Alopecia areata, Recurrent infections, Antinuclear antibody positivity, Paroxysmal nocturnal hemoglobinuria, Prolonged neonatal jaundice
PLXNA1	NM_032242.3:c.475T>Cp.(Tyr159His)		AD_unknown	4.6	het	unknown	1	NDD + Epilepsy	Developemntal regression with intellectual impairment and behavioral problems; Refractory epilepsy with tonic-clonic seizures, myoclonic seizures, status epilepticus, absence epilepsy and febrile seizures with an onset at the age of 1.5 years,
ZNF182	NM_001178099.1:c.181A>Gp.(Ser61Gly)		XL	4.8	hemi	maternal	1	NDD + Epilepsy	Seizure, Nocturnal seizures
AHCTF1	NM_015446.4:c.5018_5019insGGp.(Ile1673Metfs*4)		AD_denovo	A	het	de novo	3	Growth, Skeletal	Panhypopituitarism, Proportionate short stature, Short stature, Anterior pituitary hypoplasia, Ectopic posterior pituitary, Posterior pituitary hypoplasia
EFHC2	NM_025184.3:c.975A>Cp.(Leu325=)		XL	C	hemi	maternal	3	Growth, Skeletal	Panhypopituitarism, Proportionate short stature, Short stature, Anterior pituitary hypoplasia, Ectopic posterior pituitary, Posterior pituitary hypoplasia
TDRD6	NM_001010870.2:c.1895A>Gp.(His632Arg)	NM_001010870.2:c.2566G>Ap.(Asp856Asn)	AR_comphet	C	comphet	maternal&paternal	3	Growth, Skeletal	Panhypopituitarism, Proportionate short stature, Short stature, Anterior pituitary hypoplasia, Ectopic posterior pituitary, Posterior pituitary hypoplasia
SBNO2	NM_001100122.1:c.1960G>Tp.(Val654Leu)		AD_denovo	4.7	het	de novo	2	NDD + Epilepsy	Epileptic encephalopathy, intrauterine hydrocephalus (gestational week 24), epilepsy
APBA1	NM_001163.3:c.521T>Cp.(Leu174Pro)		AD_denovo	8.4	het	de novo	2	NDD + Epilepsy	Epileptic encephalopathy, intrauterine hydrocephalus (gestational week 24), epilepsy
NRXN3	NM_001330195.1:c.115C>Tp.(Arg39Cys)		AR_homo	8.9	homo	maternal&paternal	3	NDD	Global developmental delay and severe motor delay, cerebral palsy (GMFCS level 5), hypoplasia of the corpus callosum
MCTP2	NM_018349.3:c.409G>Ap.(Gly137Ser)	NM_018349.3:c.1889C>Tp.(Pro630Leu)	AR_comphet	3.5	comphet	maternal&paternal	1	NDD	Global developmental delay, macrocephaly and makrosomia, muscular hypotonia and ischemic stroke at four months of age
BMP4	NM_001202.5:c.172G>Cp.(Glu58Gln)		AR_homo	8.4	homo	maternal&paternal	6	NDD	Intellectual disability, Dystonia, Tremor, Limb dystonia, Cognitive impairment, Leg dystonia, Arm dystonia
NIF3L1	NM_001136039.2:c.860C>Tp.(Thr287Ile)		AR_homo	5.4	homo	maternal&paternal	6	NDD	Intellectual disability, Dystonia, Tremor, Limb dystonia, Cognitive impairment, Leg dystonia, Arm dystonia
MYOF	NM_013451.3:c.3511C>Tp.(Arg1171Trp)	NM_013451.3:c.4268delp.(Pro1423Hisfs*21)	AR_comphet	5.7	comphet	maternal&paternal	6	NDD	Intellectual disability, Dystonia, Tremor, Limb dystonia, Cognitive impairment, Leg dystonia, Arm dystonia
DLC1	NM_182643.2:c.609A>Cp.(Lys203Asn)		AR_homo	5.4	homo	maternal&paternal	6	NDD	Intellectual disability, Dystonia, Tremor, Limb dystonia, Cognitive impairment, Leg dystonia, Arm dystonia
RNF213	NM_001256071.2:c.9611A>Gp.(His3204Arg)		AD_denovo	6.8	het	de novo	6	NDD	Intellectual disability, Dystonia, Tremor, Limb dystonia, Cognitive impairment, Leg dystonia, Arm dystonia
PARBP	NM_001319988.1:c.62G>Tp.(Arg21Leu)		AD_denovo	B	het	de novo	2	Metabolism	Hypertension, Irregular menstruation, Obesity, Hyperuricemia, Red hair, Narcolepsy, Increased blood pressure, Adipositas since the age of five to eight years
SFRP5	NM_003015.3:c.872T>Cp.(Met291Thr)	NM_003015.3:c.861_863delp.(Lys287del)	AR_comphet	C	comphet	maternal&paternal	2	Metabolism	Hypertension, Irregular menstruation, Obesity, Hyperuricemia, Red hair, Narcolepsy, Increased blood pressure, Adipositas since the age of five to eight years
C6orf136	NM_001161376.1:c.478G>Tp.(Ala160Ser)	NM_001161376.1:c.430C>Tp.(Arg144Trp)	AR_comphet	B	comphet	maternal&paternal	1	Metabolism	Hypoglycemia, Neonatal hypoglycemia

SPART	NM_001142294.1:c.1655T>Gp.(Val552Gly)	NM_001142294.1:c.848C>Tp.(Ser283Phe)	AR_comphet	9.4	comphet	maternal&paternal	6	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizure, Abnormality of brain morphology, Abnormality of forebrain morphology
NUSAP1	NM_016359.4:c.1213C>Tp.(Gln405*)		AD_denovo	6.1	het	de novo	6	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizure, Abnormality of brain morphology, Abnormality of forebrain morphology
PDE4DIP	NM_001198834.3:c.3733G>Ap.(Ala1245Thr)	NM_001198834.3:c.1229_1231delAATinsTAGp.(Glu410_Leu411delinsValVal)	AR_comphet	6.7	comphet	maternal&paternal	6	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizure, Abnormality of brain morphology, Abnormality of forebrain morphology
ZNF611	NM_001161499.1:c.1904C>Tp.(Ser635Leu)		AR_homo	4.1	homo	maternal&paternal	6	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizure, Abnormality of brain morphology, Abnormality of forebrain morphology
KIR3DL3	NM_153443.4:c.1053G>Ap.(Lys351=)		AR_homo	4.0	homo	maternal&paternal	6	NDD + Epilepsy	Microcephaly, Intellectual disability, Seizure, Abnormality of brain morphology, Abnormality of forebrain morphology
TMEM181	NM_020823.1:c.448delp.(Ser150Profs*18)	NM_020823.1:c.1781C>Tp.(Pro594Leu)	AR_comphet	B	comphet	maternal&paternal	1	Fehlbildungen	Growth delay, Intrauterine growth retardation
HSP90AA1	NM_001017963.2:c.626G>Ap.(Arg209Gln)		AR_homo	7.7	homo	maternal&paternal	3	NDD	Global developmental delay and severe motor delay, cerebral palsy (GMFCS level 5), hypoplasia of the corpus callosum
HMGN5	NM_030763.2:c.268-3C>Tp.?		XL	2.5	hemi	maternal	3	NDD	Global developmental delay and severe motor delay, cerebral palsy (GMFCS level 5), hypoplasia of the corpus callosum
FAM200A	NM_145111.3:c.1702C>Tp.(Gln568*)		AR_homo	6.4	homo	maternal&paternal	5	NDD + Epilepsy	Intellectual disability, Seizure
POLL	NM_001174084.1:c.1255C>Tp.(Arg419*)		AR_homo	8.3	homo	maternal&paternal	5	NDD + Epilepsy	Intellectual disability, Seizure
PYROXD2	NM_032709.2:c.1062+2T>Gp.?		AR_homo	6.8	homo	maternal&paternal	5	NDD + Epilepsy	Intellectual disability, Seizure
TRPV1	NM_018727.5:c.896C>Tp.(Thr299Met)		AR_homo	6.5	homo	maternal&paternal	5	NDD + Epilepsy	Intellectual disability, Seizure
LRCH3	NM_032773.3:c.256C>Tp.(Arg86Trp)		AR_homo	4.4	homo	maternal&paternal	5	NDD + Epilepsy	Intellectual disability, Seizure
CHRD	NM_001304472.1:c.2491C>Gp.(Pro831Ala)		AD_denovo	8.0	het	de novo	3	NDD	Microcephaly, Intellectual disability, Ataxia, Intellectual disability, mild, Global developmental delay, Spastic gait, Gait ataxia, Intention tremor, Spastic ataxia
URB1	NM_014825.2:c.5312A>Tp.(Tyr1771Phe)	NM_014825.2:c.3362G>Ap.(Arg1121His)	AR_comphet	4.2	comphet	maternal&paternal	3	NDD	Microcephaly, Intellectual disability, Ataxia, Intellectual disability, mild, Global developmental delay, Spastic gait, Gait ataxia, Intention tremor, Spastic ataxia
HELZ2	NM_001037335.2:c.7693C>Tp.(Arg2565Cys)	NM_001037335.2:c.1750C>Tp.(Arg584Trp)	AR_comphet	4.3	comphet	maternal&paternal	3	NDD	Microcephaly, Intellectual disability, Ataxia, Intellectual disability, mild, Global developmental delay, Spastic gait, Gait ataxia, Intention tremor, Spastic ataxia
CYFIP1	NM_001324119.1:c.2817C>Gp.(Tyr939*)		AD_unknown	1.5	het	unknown	5	NDD	Syndactyly, Global developmental delay, Hypoplastic left heart, Aortic valve atresia, Congenital malformation of the left heart
TMEM94	NM_001321148.1:c.1976A>Cp.(Gln659Pro)		AD_unknown	5.3	het	unknown	5	NDD	Syndactyly, Global developmental delay, Hypoplastic left heart, Aortic valve atresia, Congenital malformation of the left heart
CLASP1	NM_015282.2:c.4142T>Ap.(Phe1381Tyr)		AD_unknown	5.8	het	unknown	5	NDD	Syndactyly, Global developmental delay, Hypoplastic left heart, Aortic valve atresia, Congenital malformation of the left heart
EIF4ENIF1	NM_001164501.1:c.1588C>Tp.(Leu530Phe)		AD_unknown	4.0	het	unknown	5	NDD	Syndactyly, Global developmental delay, Hypoplastic left heart, Aortic valve atresia, Congenital malformation of the left heart

IGF2R	NM_000876.3:c.1312G>Ap.(Ala438Thr)	NM_000876.3:c.5506G>Ap.(Val1836Ile)	AR_comphet	6.3	comphet?	unknown	5	NDD	Syndactyly, Global developmental delay, Hypoplastic left heart, Aortic valve atresia, Congenital malformation of the left heart
BTBD6	NM_033271.2:c.223C>Tp.(Leu75Phe)	NM_033271.2:c.835G>Ap.(Ala279Thr)	AR_comphet	5.1	comphet	maternal&paternal	1	NDD	Developmental disorder
ANXA3	NM_005139.2:c.541-2A>Gp.?		AD_inherited	5.1	het	paternal	1	Neuro	+) Sleep disturbance, Restless legs, Insomnia
SLITRK2	NM_001144003.2:c.265G>Tp.(Val89Leu)		XL	6.3	hemi	maternal	1	NDD + Epilepsy	Behavioral abnormality, Intellectual disability, Seizure, Global developmental delay, Short stature, Focal-onset seizure, Focal motor seizure, Generalized-onset motor seizure, Focal-onset epileptic spasm
PPP3CC	NM_001243975.1:c.323G>Ap.(Arg108His)		AD_inherited	4.0	het	maternal	4	NDD + Epilepsy	Episodic ataxia, EEG abnormality
ANKS1A	NM_015245.2:c.2269C>Tp.(Arg757Cys)		AD_inherited	4.1	het	maternal	4	NDD + Epilepsy	Episodic ataxia, EEG abnormality
RNF20	NM_019592.6:c.2783G>Ap.(Arg928His)		AD_denovo	A	het	de novo	2	Metabolism	Obesity
HECTD1	NM_015382.3:c.6068G>Tp.(Gly2023Val)		AR_homo	B	homo	maternal&paternal	2	Metabolism	Obesity
PTOV1	NM_017432.4:c.842dup, p.(Pro282Alafs*79)		AD_denovo	8.1	het	de novo	1	NDD + Epilepsy	Intellectual disability, Seizure, Global developmental delay, bilateral toni-clonic seizure, atonic seizure
ACTN2	NM_001103.3:c.1108-2A>Tp.?		AD_unknown	A	het	unknown	1	Muscle	Tetraparesis and muscle weakness since age of 51 years
LRRK1	NM_024652.5:c.5615C>Gp.(Ser1872Cys)		AD_denovo	6.9	het	de novo	4	NDD + Epilepsy	Episodic ataxia, EEG abnormality
USP19	NM_001199161.1:c.2012C>Ap.(Ser671Tyr)		AD_denovo	6.5	het	de novo	4	NDD + Epilepsy	Episodic ataxia, EEG abnormality
EP400	NM_015409.4:c.4277+1G>Tp.?		AD_unknown	7.5	het	unknown	4	NDD	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Abnormal facial shape, Delayed gross motor development, Abnormal muscle tone, Infantile muscular hypotonia, Delayed fine motor development, Neurodevelopmental delay
WNK2	NM_006648.3:c.5229G>Tp.(Lys1743Asn)		AD_unknown	3.5	het	unknown	4	NDD	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Abnormal facial shape, Delayed gross motor development, Abnormal muscle tone, Infantile muscular hypotonia, Delayed fine motor development, Neurodevelopmental delay
PHC2	NM_198040.2:c.604G>Ap.(Ala202Thr)		AD_unknown	5.0	het	unknown	4	NDD	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Abnormal facial shape, Delayed gross motor development, Abnormal muscle tone, Infantile muscular hypotonia, Delayed fine motor development, Neurodevelopmental delay
CREB5	NM_182898.3:c.302T>Ap.(Met101Lys)		AD_unknown	3.7	het	unknown	4	NDD	Behavioral abnormality, Delayed speech and language development, Intellectual disability, Muscular hypotonia, Global developmental delay, Motor delay, Generalized hypotonia, Abnormal facial shape, Delayed gross motor development, Abnormal muscle tone, Infantile muscular hypotonia, Delayed fine motor development, Neurodevelopmental delay
ATP6AP1	NM_001183.5:c.3G>Tp.0?		XL	8.3	hemi	unknown	5	NDD	Syndactyly, Global developmental delay, Hypoplastic left heart, Aortic valve atresia, Congenital malformation of the left heart
LAMP2	NM_001122606.1:c.731C>Gp.(Thr244Ser)		XL	B	hemi	maternal	2	Metabolism	Myalgia, Hyperlipoproteinemia, Increased erythrocyte protoporphyrin concentration, Angioedema
PMM1	NM_002676.2:c.416C>Tp.(Ser139Leu)		AD_denovo	6.8	het	de novo	2	NDD + Epilepsy	Epileptic encephalopathy, Seizure since the age of 11
EIF2S2	NM_003908.4:c.692G>Ap.(Arg231His)		AD_denovo	8.4	het	de novo	2	NDD	Inguinal hernia, Intellectual disability, Global developmental delay, Small for gestational age, Penile hypospadias, Short stature, Decreased body weight
ZBTB45	NM_001316978.1:c.655G>Ap.(Asp219Asn)		AD_denovo	5.3	het	de novo	2	NDD	Inguinal hernia, Intellectual disability, Global developmental delay, Small for gestational age, Penile hypospadias, Short stature, Decreased body weight
CXorf21	NM_025159.2:c.774A>Cp.(Arg258Ser)		XL	4.5	hemi	maternal	1	NDD + Epilepsy	Leukenzephalopathie, keine Sprache, muskuläre Hypertonie, schwere motorische und sprachlich-mentale Entwicklungsverzögerung, therapierefraktäre Krampfanfälle
OSBPL9	NM_148909.3:c.413_422delp.(Ser138Ilefs*16)		AD_unknown	6.2	het	unknown	1	NDD + Epilepsy	Seizure, Generalized non-motor (absence) seizure, Generalized myoclonic seizure, Atypical absence seizure, Eyelid myoclonia seizure, Myoclonic seizure
FHOD3	NM_001281740.2:c.1836-2A>Gp.?		AD_unknown	5.5	het	unknown	1	NDD + Epilepsy	Entwicklungsstörung, Epilepsie (Absencen, Grand-mal-Anfälle)

ARHGEF28	NM_001080479.2:c.548T>Gp.(Leu183Trp)		AD_denovo	7.0	het	de novo	1	NDD	Abnormality of body height, Tall stature, Epicanthus, Abnormality of the nasal bridge, Abnormal eyelid morphology, Widely spaced teeth, Behavioral abnormality, Restlessness, Low frustration tolerance, Delayed speech and language development, Global developmental delay, Large for gestational age, Abnormal facial shape, Increased body weight, Depressed nasal bridge
ASB13	NM_024701.3:c.404T>Gp.(Leu135Arg)		AD_unknown	C	het	unknown	1	Immunology	Immunodeficiency, Autoimmunity, Decreased antibody level in blood, Combined immunodeficiency, Abnormal immunoglobulin level, Abnormality of immune system physiology
NOL4	NM_003787.4:c.1A>Cp.0?		AD_inherited	5.6	het	maternal	1	NDD	Microcephaly, Aggressive behavior, Intellectual disability, Intellectual disability, mild, Abnormal aggressive, impulsive or violent behavior
TMEM232	NM_001039763.3:c.884A>Gp.(Gln295Arg)		AD_denovo	5.6	het	de novo	1	NDD + Epilepsy	epilepsy, movement disorder, syncope, myoclonia, pathological waking EEG, cystic, malformation of the right kidney
NCOA1	NM_003743.4:c.3457C>Tp.(Gln1153*)		AD_unknown	8.3	het	unknown	2	NDD	Intellectual disability, Global developmental delay, Obesity, Abnormal heart morphology, Truncal obesity, Kyphosis, Attention deficit hyperactivity disorder
ARHGAP35	NM_004491.4:c.597_600delp.(Thr200Serfs*18)		AD_denovo	A	het	de novo	1	Immunology	Splenomegaly, Lymphopenia, recurrent infections, immunodeficiency, decreased circulating IGA, increased circulating IgM level
APLNR	NM_005161.4:c.952C>Tp.(Gln318*)		AD_denovo	7.4	het	de novo	1	NDD	Coarse facial features, Hearing impairment, Delayed speech and language development, Intellectual disability, Global developmental delay, Absent speech, Failure to thrive, Premature birth, Atrial septal defect, Hypoglycemia, Abnormal facial shape, Expressive language delay, Decreased circulating IgA level,(-) Recurrent upper respiratory tract infections, Decreased body weight,(-) Intestinal obstruction, Decreased circulating IgG2 level, Respiratory tract infection, Psychomotor retardation, Decreased body mass index, Decreased circulating IgG subclass level, Decreased circulating IgG1 level
RBFOX1	NM_001308117.1:c.445C>Gp.(Pro149Ala)	NM_001142333.1:c.1069G>Cp.(Ala357Pro)	AR_comphet	7.1	comphet	maternal&paternal	2	NDD	global muscular hypotension with axial weakness, facial dysmorphia, indicated high palate, broad neck, muscle relief decreasing distally, hypersalivation, no secure free sitting
PCID2	NM_001258212.1:c.835G>Ap.(Asp279Asn)	NM_001258212.1:c.35A>Gp.(Gln12Arg)	AR_comphet	4.2	comphet	maternal&paternal	2	NDD	global muscular hypotension with axial weakness, facial dysmorphia, indicated high palate, broad neck, muscle relief decreasing distally, hypersalivation, no secure free sitting
KPNA1	NM_002264.3:c.1015G>Ap.(Ala339Thr)		AD_denovo	7.4	het	de novo	1	NDD	Global developmental delay and speech delay, microcephaly
GNAI1	NM_002069.5:c.430C>Tp.(Arg144*)		AD_unknown	8.5	het	unknown	1	NDD	Intellectual disability, Muscular hypotonia Global developmental delay, Bilateral ptosis, Ventricular septal defect, Abnormal facial shape, Migraine, Neurodevelopmental delay, Neurodevelopmental abnormality
UBR2	NM_015255.2:c.2462+2T>Cp.?		AD_unknown	7.2	het	unknown	1	NDD	Delayed speech and language development, Intellectual disability, Intellectual disability, mild, Obesity, Expressive language delay, Abnormality of body weight, Increased body weight, Increased adipose tissue, Neurodevelopmental delay, Overweight
DLG5	NM_004747.3:c.453dupp.(Gln152Serfs*26)		AD_unknown	7.6	het	unknown	1	NDD	Intellectual disability, Seizure, Intellectual disability, mild, Encephalopathy, Bilateral tonic-clonic seizure, Gliosis, Febrile seizure (within the age range of 3 months to 6 years), Aplasia/Hypoplasia involving the central nervous system, Abnormal nervous system physiology, Epileptic encephalopathy, Motor seizure
DSCAML1	NM_020693.3:c.1322C>Tp.(Ser441Phe)		AD_inherited	4.4	het	maternal	1	NDD + Epilepsy	Intellectual disability, Seizure, Intellectual disability, mild, Pachygyria, Polymicrogyria, Generalized-onset seizure, Abnormal cortical gyration, Focal-onset seizure, Bilateral perisylvian polymicrogyria, Focal polymicrogyria
PSMB6	NM_002798.2:c.238T>Cp.(Ser80Pro)		AR_homo	6.5	homo	maternal&paternal	3	NDD	(+) Microcephaly,(+) Delayed speech and language development,(+) Global developmental delay,(+) Motor delay,(+) Joint hypermobility,(+) Abnormal facial shape,(+) Chronic diarrhea,(+) Abnormal pattern of respiration,(+) Short stature,(+) Infantile muscular hypotonia,(+) Severe global developmental delay,(+) Feeding difficulties
ZC3H3	NM_015117.2:c.1595C>Tp.(Thr532Ile)		AD_denovo	5.3	het	de novo	3	NDD	(+) Microcephaly,(+) Delayed speech and language development,(+) Global developmental delay,(+) Motor delay,(+) Joint hypermobility,(+) Abnormal facial shape,(+) Chronic diarrhea,(+) Abnormal pattern of respiration,(+) Short stature,(+) Infantile muscular hypotonia,(+) Severe global developmental delay,(+) Feeding difficulties

RNF167	NM_015528.2:c.793C>Gp.(Arg265Gly)		AR_homo	4.6	homo	maternal&paternal	3	NDD	(+) Microcephaly,(+) Delayed speech and language development,(+) Global developmental delay,(+) Motor delay,(+) Joint hypermobility,(+) Abnormal facial shape,(+) Chronic diarrhea,(+) Abnormal pattern of respiration,(+) Short stature,(+) Infantile muscular hypotonia,(+) Severe global developmental delay,(+) Feeding difficulties
NKX3-1	NM_006167.3:c.491C>Tp.(Thr164Met)	NM_006167.3:c.113G>Ap.(Gly38Asp)	AR_comphet	4.3	comphet	maternal&paternal	1	NDD + Epilepsy	Intelligenzminderung, Epilepsie, Schwerhörigkeit
TENM2	NM_001122679.1:c.7970C>Tp.(Thr2657Met)		AD_denovo	6.7	het	de novo	3	NDD	Global developmental delay, intellectual impairment, absent speech
SRCIN1	NM_025248.2:c.40C>Tp.(Pro14Ser)		AR_homo	3.4	homo	maternal&paternal	3	NDD	Global developmental delay, intellectual impairment, absent speech
FAT2	NM_001447.2:c.5000T>Gp.(Val1667Gly)		AR_homo	5.1	homo	maternal&paternal	3	NDD	Global developmental delay, intellectual impairment, absent speech
BTBD3	NM_181443.3:c.1502C>Ap.(Pro501Gln)		AD_unknown	4.7	het	unknown	2	NDD	Intellectual disability, Global developmental delay, Obesity, Abnormal heart morphology, Truncal obesity, Kyphosis, Attention deficit hyperactivity disorder
ATP13A5	NM_198505.2:c.1949A>Gp.(Tyr650Cys)	NM_198505.2:c.1241dup, p.(Tyr415Leufs*72)	AR_unknown	6.8	comphet?	unknown	1	NDD	epilepsy, ID, cerebral palsy, EEG and MRI abnormalities
DOCK4	NM_014705.3:c.2945C>Tp.(Thr982Ile)		AD_denovo	8.7	het	de novo	1	NDD + Epilepsy	tonic-clonic seizures, intelligence impairment, tremor, ataxia
RPS6KA6	NM_001330512.1:c.2113-3T>Gp.?		XL	4.8	hemi	maternal	2	NDD + Epilepsy	nocturnal focal seizures, onset on the age of 4, microcephaly, developmental delay, intelligence impairment (IQ=81), ADHS
SPATA8	NM_173499.4:c.28C>Tp.(Gln10*)		AD_denovo	4.1	het	de novo	2	NDD + Epilepsy	nocturnal focal seizures, onset on the age of 4, microcephaly, developmental delay, intelligence impairment (IQ=81), ADHS
CNOT9	ENST00000273064:c.680G>A p.Arg227His		AD_denovo	7.0	het	de novo	1	NDD	global developmental delay, mild intellectual disability, hypotonia, anxiety, impulsivity, decreased head circumference, microretrognathia, hypertelorism, high, narrow palate single, transverse palmar crease, everted upper lip vermilion, wide nasal base, slender build, supernumerary nipple, 2-4 toe syndactyly
TANC1	NM_001145909.1:c.10G>Cp.(Ala4Pro)	NM_001145909.1:c.1007G>Ap.(Arg336Gln)	AR_comphet	5.3	comphet	maternal&paternal	3	NDD	global developmental delay, absent speech, muscular hypotonia, autism spectrum disorder, microcephaly
RHBDF1	NM_022450.3:c.1082G>Ap.(Arg361His)		AD_denovo	5.6	het	de novo	3	NDD	global developmental delay, absent speech, muscular hypotonia, autism spectrum disorder, microcephaly
FOXO4	NM_005938.3:c.43A>Tp.(Ile15Phe)		XL	4.9	hemi	maternal	3	NDD	global developmental delay, absent speech, muscular hypotonia, autism spectrum disorder, microcephaly
CMIP	NM_198390.2:c.42del,p.(Gln15Argfs*36)		AD_inherited	8.0	het	paternal	1	NDD + Epilepsy	Intellectual disability, seizures
MKRN1	NM_013446.4:c.340C>Tp.(Gln114*)		AD_denovo	8.3	het	de novo	2	NDD	Intellectual disability, mild, Global developmental delay, Mild global developmental delay, Short stature, Talipes equinovarus
SLC25A5	NM_001152.4:c.616A>Gp.(Lys206Glu)		XL	6.6	hemi	maternal	2	NDD	Mild global developmental delay, Cleft palate, Hearing impairment, Ventricular septal defect, Patent ductus arteriosus, Pulmonic stenosis, Hypospadias, Enlarged vestibular aqueduct, Pes cavus, Scoliosis
PCSK6	NM_002570.4:c.412C>Ap.(Leu138Ile)	NM_002570.4:c.2232A>Tp.(Arg744Ser)	AR_comphet	4.4	comphet	maternal&paternal	2	NDD	Mild global developmental delay, Cleft palate, Hearing impairment, Ventricular septal defect, Patent ductus arteriosus, Pulmonic stenosis, Hypospadias, Enlarged vestibular aqueduct, Pes cavus, Scoliosis
ECPAS	NM_001080398.1:c.1464G>Ap.(Glu488=)		AD_unknown	2.5	het	unknown	2	NDD	Intellectual disability, severe, Severe global developmental delay, Seizure, Focal-onset seizure, Motor seizure, Cerebral palsy (GMFCS V), Microcephaly, Feeding difficulties no speech
RXRB	NM_001270401.1:c.1472C>Ap.(Ala491Asp)		AD_unknown	4.3	het	unknown	2	NDD	Intellectual disability, severe, Severe global developmental delay, Seizure, Focal-onset seizure, Motor seizure, Cerebral palsy (GMFCS V), Microcephaly, Feeding difficulties no speech

WDR13	NM_001347217.2:c.194G>Ap.(Arg65His)		XL	5.7	hemi	unknown	1	NDD + Epilepsy	Global developmental delay, EEG abnormality, Carious teeth, Finger clinodactyly, Decreased head circumference, Intellectual disability
HNRNPH3	NM_001322434.1:c.436+5G>Ap.?		AD_unknown	2.0	het	unknown	1	NDD	(+) Focal-onset seizure,(+) Hemimegalencephaly,(+) Spastic hemiparesis,(+) Moderate global developmental delay,(+) Microcephaly,(+) Intellectual disability, moderate
EPHB3	NM_004443.4:c.1711G>Ap.(Val571Met)		AR_homo	4.9	homo	maternal&paternal	1	NDD	Intellectual disability, Seizures (onset at age of 14 years), Sleep disturbance, Hypotonic cerebral palsy
SV2B	NM_001167580.2:c.895C>Tp.(Gln299*)		AD_unknown	6.4	het	unknown	2	NDD + Epilepsy	Seizure, Focal impaired awareness motor seizure, Generalized-onset seizure, Generalized-onset motor seizure, Bilateral tonic-clonic seizure with generalized onset,(+) Subcortical band heterotopia, Neurodevelopmental abnormality, Specific learning disability Intellectual disability, mild
HOXD4	NM_014621.2:c.111C>Ap.(Tyr37*)		AD_unknown	4.0	het	unknown	2	NDD + Epilepsy	Seizure, Focal impaired awareness motor seizure, Generalized-onset seizure, Generalized-onset motor seizure, Bilateral tonic-clonic seizure with generalized onset,(+) Subcortical band heterotopia, Neurodevelopmental abnormality, Specific learning disability Intellectual disability, mild
PIK3AP1	NM_152309.3:c.601A>Tp.(Lys201*)		AD_unknown	5.5	het	unknown	1	NDD + Epilepsy	Epileptic encephalopathy, Cognitive impairment, Microcephaly, Short stature, Febrile seizure
GRM2	NM_000839.3:c.2462C>Tp.(Pro821Leu)		AD_unknown	4.7	het	unknown	1	NDD + Epilepsy	Atypical absence seizure, Multifocal seizures, Focal impaired awareness seizure, Bilateral tonic-clonic seizure with focal onset, Intellectual disability, mild, Intellectual disability, moderate, Ventricular extrasystoles
TBC1D25	NM_001348262.1:c.143G>Cp.(Gly48Ala)		XL	3.0	hemi	maternal	2	NDD	developmental delay, cerebral paresis, dystrophy
TTC28	NM_001145418.1:c.5009A>Tp.(His1670Leu)	NM_001145418.1:c.4237G>Ap.(Gly1413Ser)	AR_comphet	C	comphet	maternal&paternal	1	Growth, Skeletal	Trigonocephaly, Abnormality of calvarial morphology
FNDC3A	NM_001079673.2:c.760+1G>Tp.?		AD_unknown	6.7	het	unknown	1	NDD + Epilepsy	(+) Moderate global developmental delay,(+) Intellectual disability, mild,(+) Aggressive behavior,(+) Pes valgus,(+) Abnormality of the palmar creases,(+) Brachydactyly,(+) Long face,(+) Frontal bossing,(+) Hypertelorism,(+) Epicanthus,(+) Sparse lateral eyebrow,(+) Hypoplastic philtrum,(+) Focal-onset seizure,(+) Cerebral white matter hypoplasia
STRN3	NM_001083893.2:c.542+2T>Gp.?		AD_inherited	B	het	paternal	2	Growth, Skeletal	Short stature (151cm), Short for gestational age
ATG9A	NM_001077198.3:c.2398C>Tp.(His800Tyr)		AD_inherited	C	het	paternal	2	Growth, Skeletal	Short stature (151cm), Short for gestational age
ZNF142	NM_003442.5:c.44_45delAGp.Glu15Valfs*25		AR_homo	8.6	homo	maternal&paternal	1	NDD	NDD, microcephaly
ARHGEF6	NM_004840.2:c.257A>Cp.Ap86Ala		XL	8.2	hemi	unknown	1	NDD	NDD
FRMD5	NM_032892.5:c.1045A>Cp.(Ser349Arg)		AD_denovo	6.4	het	de novo	1	NDD	Global developmental delay, Expressive language delay, Receptive language delay, Gait ataxia, Muscular hypotonia, Microcephaly, Cerebral atrophy, Abnormality of eye movement
CCNT2	NM_058241.3:c.370-3_370-2insAp.?		AD_denovo	C	het	de novo	1	Immunology	Increased circulating IgG4 level, Neutropenia, Papule, Folliculitis, Lymphangitis, Pustule, Immunodeficiency
MRTFA	NM_001318139.2:c.800delp.(Lys267Argfs*2)		AD_denovo	9.8	het	de novo	3	NDD + Epilepsy	Suspected vitamin-B6-dependent epilepsy, Generalized-onset epileptic spasm, Atypical absence seizure, EEG abnormality
LRFN4	NM_024036.5:c.473G>C,p.(Arg158Pro)	NM_024036.5:c.853C>T,p.(Arg285Cys)	AR_comphet	4.9	comphet	maternal&paternal	2	NDD	schwere Entwicklungsverzögerung, spastische Cerebralparese, Dystrophie, MRT unauf-fällig, EEG auffällig
PACS1	NM_018026.4:c.445-17_445-7delp.?		AD_denovo	6.6	het	de novo	3	NDD + Epilepsy	Suspected vitamin-B6-dependent epilepsy, Generalized-onset epileptic spasm, Atypical absence seizure, EEG abnormality
ZNF518A	NM_001278524.2:c.3520G>Tp.(Val1174Leu)		AR_homo	2.3	homo	maternal&paternal	3	NDD + Epilepsy	Suspected vitamin-B6-dependent epilepsy, Generalized-onset epileptic spasm, Atypical absence seizure, EEG abnormality
BTAFL1	NM_003972.3:c.2662G>Ap.(Glu888Lys)		AD_unknown	5.0	het	unknown	1	NDD + Epilepsy	(+) Global developmental delay,(+) Absent speech,(+) Seizure,(+) Intellectual disability
ADCY7	NM_001114.5:c.2866C>Tp.(Arg956Trp)		AD_denovo	6.6	het	de novo	3	Neuro	Multifocal cerebral white matter abnormalities, Leukoencephalopathy, Migraine, Abnormal cerebellum morphology, Gait disturbance, Gait imbalance

TTLL4	NM_014640.5:c.2401C>Gp.(Leu801Val)	NM_014640.5:c.2692G>Ap.(Glu898Lys)	AR_comphet	4.1	comphet	maternal&paternal	3	Neuro	Multifocal cerebral white matter abnormalities, Leukoencephalopathy, Migraine, Abnormal cerebellum morphology, Gait disturbance, Gait imbalance
FBN3	NM_032447.5:c.6184G>Ap.(Ala2062Thr)	NM_032447.5:c.4370A>Gp.(Asn1457Ser)	AR_comphet	3.5	comphet	maternal&paternal	3	Neuro	Multifocal cerebral white matter abnormalities, Leukoencephalopathy, Migraine, Abnormal cerebellum morphology, Gait disturbance, Gait imbalance
HIRA	NM_003325.4:c.194A>Gp.(Gln65Arg)		AD_inherited	C	het	maternal	2	Renal/urinary	Non-midline cleft lip and palate
RGMB	NM_001012761.3:c.863C>Tp.(Thr288Ile)		AD_inherited	C	het	maternal	2	Renal/urinary	Non-midline cleft lip and palate
STARD8	NM_001142503.2:c.2248C>Ap.(Leu750Ile)		XL	4.0	hemi	maternal	1	NDD + Epilepsy	EEG with burst suppression, Epileptic encephalopathy, Global developmental delay, Intellectual disability, Seizure
KDR	NM_002253.3:c.3161_3162insAAp.(Tyr1054*)		AD_unknown	B	het	unknown	1	Congenital heart defect	Abnormal aortic morphology, Abdominal aortic aneurysm, Descending thoracic aorta aneurysm, Cerebral arterial thrombosis
FAM199X	NM_207318.4:c.932T>Gp.(Met311Arg)		AD_denovo	C	hemi	de novo	2	Connective Tissue	Recurrent fractures, Patellar dislocation, Recurrent infections, Migraine, Asthma
LIMD1	NM_014240.3:c.1669C>Tp.(His557Tyr)	NM_014240.3:c.1532C>Tp.(Ala511Val)	AR_comphet	A	comphet	maternal&paternal	2	Connective Tissue	Recurrent fractures, Patellar dislocation, Recurrent infections, Migraine, Asthma
FBXW7	NM_033632.3:c.23_24delp.(Val8Glyfs*14)		AD_unknown	B	het	unknown	1	other	(+) Brain neoplasm,(+) Ewing sarcoma
ATR	NM_001184.4:c.2419G>Ap.(Gly807Arg)		AR_homo	8.8	homo	maternal&paternal	3	NDD + Epilepsy	Global developmental delay, Microcephaly, Seizures
CDK12	NM_016507.4:c.4237C>Tp.(His1413Tyr)		AR_homo	7.2	homo	maternal&paternal	3	NDD + Epilepsy	Global developmental delay, Microcephaly, Seizures
SLC18B1	NM_052831.3:c.821G>Tp.(Gly274Val)	NM_052831.3:c.654T>Ap.(Asn218Lys)	AR_comphet	3.7	comphet	maternal&paternal	3	NDD + Epilepsy	Global developmental delay, Microcephaly, Seizures
ZFYVE9	NM_004799.3:c.3220C>Ap.(Leu1074Met)	NM_004799.3:c.4124A>Tp.(Tyr1375Phe)	AR_comphet	5.3	comphet	maternal&paternal	2	NDD + Epilepsy	Neonatal hypoglycemia, Seizure, Global developmental delay
LANCL3	NM_001170331.2:c.1037G>Ap.(Ser346Asn)		XL	3.2	hemi	maternal	2	NDD + Epilepsy	Neonatal hypoglycemia, Seizure, Global developmental delay
LOXL4	NM_032211.6:c.396C>Ap.(Cys132*)		AD_unknown	C	het	unknown	1	Growth, Skeletal	(+) Small for gestational age,(+) Mild short stature,(+) Attention deficit hyperactivity disorder,(+) Delayed skeletal maturation,(+) Intrauterine growth retardation,(+) Mild intrauterine growth retardation
NKTR	NM_005385.4:c.3076delp.(Glu1026Argfs*26)		AD_denovo	10.4	het	de novo	1	NDD + Epilepsy	Myoclonic spasms, Seizure, EEG abnormality
DPYSL2	ENST00000311151.5:c.1544C>T p.Pro515Leu		AD_unknown	7.2	het	unknown	2	NDD	Cognitive impairment, Global developmental delay, Tall stature, Obesity
DGCR2	ENST00000263196.7:c.998T>C p.Leu333Pro		AD_unknown	4.6	het	unknown	2	NDD	Cognitive impairment, Global developmental delay, Tall stature, Obesity
KIF5B	NM_004521.3:c.135_136dupp.(Tyr46Phefs*67)		AD_unknown	B	het	unknown	1	Fehlbildungen	Macroductyly, Upper limb asymmetry, Hemihypertrophy of upper limb, Hyperextensible thumb
NRCAM	NM_001193582.1:c.3362C>Gp.(Pro1121Arg)		AD_unknown	6.3	het	unknown	1	NDD + Epilepsy	Hypospadias, Microcephaly, Atypical absence seizure, Bilateral tonic-clonic seizure, Intellectual disability, Premature birth, Patent ductus arteriosus, Hearing impairment
PSMB10	NM_002801.4:c.56+1G>Ap.?		AR_homo	8.5	homo	unknown	1	NDD	(+) Global developmental delay,(+) Intellectual disability, borderline,(+) Intellectual disability, mild,(+) Short stature,(+) Microcephaly,(+) Bird-like facies
TOPAZ1	NM_001145030.1:c.481A>Tp.(Ser161Cys)		AD_denovo	4.6	het	de novo	1	NDD + Epilepsy	Focal-onset seizure, Focal sensory seizure
ARHGEF38	NM_001242729.2:c.1363_1365delACGinsGCAp.(Thr455Ala)	NM_001242729.2:c.2122G>Ap.(Asp708Asn)	AR_comphet	C	comphet	maternal&paternal	1	Metabolism	Diabetes insipidus, Central diabetes insipidus, Panhypopituitarism, Short stature, Proportionate short stature
ATP8B4	NM_024837.3:c.2698-2A>Gp.?		AD_denovo	5.2	het	de novo	2	NDD + Epilepsy	mild global developmental delay, febrile seizure (within the age range of 3 months to 6 years)

MYO5B	NM_001080467.2:c.1624C>Tp.(Arg542Cys)		AD_denovo	6.3	het	de novo	2	NDD + Epilepsy	mild global developmental delay, febrile seizure (within the age range of 3 months to 6 years)
PTPRT	NM_133170.4:c.3039+1G>Ap.?		AD_unknown	B	het	unknown	2	Leukodystrophy	(+) Cerebral vasculitis,(+) Ischemic stroke,(+) Moyamoya disease,(+) Leukoencephalopathy
XPOT	NM_007235.6:c.1516_1517delp.(Val506Cysfs*2)		AD_unknown	7.7	het	unknown	2	Neuro	(+) Cerebral vasculitis,(+) Ischemic stroke,(+) Moyamoya disease,(+) Leukoencephalopathy
TMEM35B	NM_001195156.1:c.289+2delp.?		AR_homo	A	homo	unknown	1	other	+) Elevated serum alanine aminotransferase,(+) Elevated serum aspartate aminotransferase,(+) Abnormality of the liver,(+) Splenomegaly,(-) Wilson disease,(-) Niemann-Pick disease type D
HSPH1	NM_006644.4:c.515delp.(Asn172Metfs*3)		AD_unknown	6.5	het	unknown	2	NDD + Epilepsy	(+) Seizure,(+) Global developmental delay,(+) Stereotypical hand wringing,(+) Muscular hypotonia
ZBTB21	NM_001098402.2:c.2088delp.(Lys696Asnfs*5)		AD_unknown	6.1	het	unknown	2	NDD + Epilepsy	(+) Seizure,(+) Global developmental delay,(+) Stereotypical hand wringing,(+) Muscular hypotonia
SVEP1	NM_153366.4:c.6371T>Cp.(Ile2124Thr)		AR_homo	5.5	homo	maternal&paternal	3	NDD + Epilepsy	atypic absence seizure, startle-induced seizure, attention deficit hyperactivity disorder, seizure
ALS2CL	NM_147129.5:c.1109+5G>Ap.?		AD_denovo	5.0	het	de novo	3	NDD + Epilepsy	atypic absence seizure, startle-induced seizure, attention deficit hyperactivity disorder, seizure
CENPI	NM_006733.3:c.652C>Tp.(Arg218Cys)		XL	5.8	hemi	maternal	3	NDD + Epilepsy	atypic absence seizure, startle-induced seizure, attention deficit hyperactivity disorder, seizure
FAT3	NM_001008781.2:c.763C>Gp.(His255Asp)	NM_001008781.2:c.11140A>Gp.(Lys3714Glu)	AR_comphet	5.4	comphet	maternal&paternal	1	NDD + Epilepsy	Atypical absence seizure, Myoclonic seizure, Epileptic encephalopathy, Myoclonus, EEG abnormality, Hyperammonemia, Abnormal vitamin B12 level, normal development
USP34	NM_014709.4:c.7561G>Cp.(Val2521Leu)	NM_014709.4:c.4229C>Tp.(Ala1410Val)	AR_comphet	5.5	comphet	maternal&paternal	1	NDD	(+) Intellectual disability,(+) Hyperactivity,(+) Autistic behavior
ANKDD1A	NM_182703.5:c.1470G>Cp.(Arg490Ser)		AD_denovo	5.4	het	de novo	1	NDD	(+) Delayed speech and language development,(+) Diminished ability to concentrate,(+) Cognitive impairment,(+) Hearing impairment
KLHL29	NM_052920.2:c.797C>Tp.(Pro266Leu)		AD_denovo	4.1	het	de novo	1	Neuro	Behavioral abnormality, Frontotemporal dementia
ACTR1A	NM_005736.3:c.715G>Cp.(Ala239Pro)		AD_unknown	4.6	het	unknown	1	NDD + Epilepsy	Generalized-onset motor seizure, Spastic tetraplegia, Intellectual disability, severe, Cataract, Pes planus
ZCCHC14	NM_015144.2:c.52C>Tp.(Gln18*)		AD_denovo	8.5	het	de novo	1	NDD	motor delay, proximal muscle weakness, makrozephalia, epicanthus med., frontal blossing
SEZ6L2	NM_001243332.1:c.910A>Gp.(Thr304Ala)		AD_inherited	5.4	het	maternal	3	NDD	Autism (Asperger), Autistic behavior, Depressivity, Macrocephaly
GLRA2	NM_002063.4:c.1334G>Ap.(Arg445Gln)		XL	7.5	hemi	maternal	1	NDD + Epilepsy	(+) Tonic seizure,(+) Bilateral tonic-clonic seizure with generalized onset,(+) Intellectual disability,(+) Global developmental delay,(+) Cognitive impairment
POU2F1	NM_002697.4:c.318G>Cp.(Gln106His)		AD_inherited	3.8	het	paternal	3	NDD	Autism (Asperger), Autistic behavior, Depressivity, Macrocephaly
SEMA4C	NM_017789.4:c.2077_2078delGAinsTTp.(Glu693Leu)	NM_017789.4:c.517+3G>Ap.?	AR_comphet	4.4	comphet	maternal&paternal	1	NDD + Epilepsy	At the age of 7-8 months tonic stiffnesses for a few seconds every few weeks, later on big-ger seizures, MRI without findings, no motor delay, increased levels of serum lactate, glutaric aciduria
POU3F2	NM_005604.4:c.664C>Tp.(Pro222Ser)		AD_inherited	5.4	het	paternal	1	Neuro	Leukodystrophy, Leukoencephalopathy, Attention deficit hyperactivity disorder, Neurological speech impairment, Neonatal asphyxia, Gait disturbance
MAST3	NM_015016.2:c.3367C>Tp.(Arg1123*)		AD_unknown	5.5	het	unknown	1	NDD + Epilepsy	Abnormal morphology of the limbic system,Seizure, Focal-onset seizure, Focal impaired awareness motor seizure, Bilateral tonic-clonic seizure with focal onset, Global developmental delay, Mild global developmental delay, Intellectual disability, Intellectual disability, mild, EEG with focal slow activity
PHLPP1	NM_194449.3:c.3756-2A>Gp.?		AD_denovo	10.2	het	de novo	3	NDD + Epilepsy	therapy-resistant epilepsy
SRRM4	NM_194286.3:c.1295C>Tp.(Ser432Phe)	NM_194286.3:c.1172G>Ap.(Arg391His)	AR_comphet	5.5	comphet	maternal&paternal	3	NDD + Epilepsy	therapy-resistant epilepsy

CANX	NM_001024649.1:c.143A>Tp.(Asp48Val)	NM_001024649.1:c.1102G>Ap.(Val368Ile)	AR_comphet	7.4	comphet	maternal&paternal	3	NDD + Epilepsy	therapy-resistant epilepsy
H2AC8	NM_021052.2:c.107G>Ap.(Arg36His)		AD_denovo	4.5	het	de novo	1	NDD	(+) Arachnoid cyst,(+) Headache,(+) Hallucinations,(+) Visual hallucinations,(+) Auditory hallucinations,(+) Delayed speech and language development,(+) Global developmental delay,(+) Intellectual disability,(+) Obesity
TMEM61	NM_182532.2:c.101G>Cp.(Cys34Ser)	NM_182532.2:c.583G>Ap.(Ala195Thr)	AR_comphet	C	comphet	maternal&paternal	2	Wachstum, Skelett	Hypoterlorism, Trigenocephaly
TRPC5	NM_012471.2:c.280G>Ap.(Val94Met)		XL	7.2	hemi	maternal	2	NDD	(+) Global developmental delay,(+) Hyperactivity,(+) Delayed speech and language development,(+) Hypertelorism,(+) Depressed nasal ridge,(+) Low-set ears,(+) Muscular hypotonia, lateral fallende Lidachsen
HIVEP1	NM_002114.3:c.4588T>Cp.(Ser1530Pro)	NM_002114.3:c.1916T>Cp.(Val639Ala)	AR_comphet	3.8	comphet	maternal&paternal	2	NDD	(+) Global developmental delay,(+) Hyperactivity,(+) Delayed speech and language development,(+) Hypertelorism,(+) Depressed nasal ridge,(+) Low-set ears,(+) Muscular hypotonia, lateral fallende Lidachsen
ZNF384	NM_001135734.2:c.459delp.(Gly154Alafs*15)		AD_denovo	9.1	het	de novo	2	NDD	(+) Global developmental delay,(+) Scotoma,(+) Intellectual disability, mild,(+) Intellectual disability, borderline,(+) Myopia,(+) Depressivity,(+) Anxiety,(+) Motor delay,(+) Retinal atrophy
SLC25A6	NM_001636.3:c.239G>Ap.(Arg80His)		AD_denovo	7.2	het	de novo	2	NDD	(+) Global developmental delay,(+) Scotoma,(+) Intellectual disability, mild,(+) Intellectual disability, borderline,(+) Myopia,(+) Depressivity,(+) Anxiety,(+) Motor delay,(+) Retinal atrophy
NIN	NM_020921.3:c.4760A>Cp.(Gln1587Pro)	NM_020921.3:c.446C>Tp.(Thr149Met)	AR_comphet	C	comphet	maternal&paternal	2	Wachstum, Skelett	Hypoterlorism, Trigenocephaly
ZDHC2	NM_016353.5:c.47_52delp.(Arg16_Val17del)		AD_denovo	5.2	het	de novo	2	NDD + Epilepsy	(+) Myoclonic seizure,(+) EEG with spike-wave complexes, suspected focal cortical dysplasia frontal right
KALRN	NM_001024660.4:c.3534G>Tp.(Arg1178Ser)	NM_001024660.4:c.5176+21733A>Gp.(=)	AR_comphet	7.0	comphet	maternal&paternal	2	NDD + Epilepsy	(+) Myoclonic seizure,(+) EEG with spike-wave complexes, suspected focal cortical dysplasia frontal right
TRHDE	NM_013381.2:c.1050_1052delTGTTinsGGGp.(Val351Gly)		AD_denovo	B	het	de novo	1	Wachstum, Skelett	+) Arthrogryposis multiplex congenita,(+) Plagiocephaly,(+) Congenital finger flexion contractures,(+) Wrist flexion contracture,(+) Elbow flexion contracture,(+) Shoulder flexion contracture,(+) Adducted thumb,(+) Respiratory failure
PTPRS	NM_002850.3:c.4810G>Ap.(Ala1604Thr)	NM_002850.3:c.4453G>Ap.(Ala1485Thr)	AR_comphet	5.8	comphet	maternal&paternal	1	NDD	(+) Short stature,(+) Global developmental delay,(+) Intellectual disability,(+) Microcephaly
ABCB5	NM_001163941.1:c.2867_2867+1delp.(Ile956Lysfs*43)		AD_denovo	7.7	het	de novo	1	NDD	(+) Mild global developmental delay,(+) Muscular hypotonia
RASA2	NM_006506.3:c.1591-2A>Gp.?		AD_denovo	8.0	het	de novo	1	NDD	(+) Periventricular leukomalacia,(+) Global developmental delay,(+) Cerebral palsy,(+) Elevated hepatic transaminase,(+) Muscular hypotonia,(+) Small for gestational age
GRAMD1C	NM_017577.4:c.168C>Ap.(Ser56Arg)	NM_017577.4:c.557A>Gp.(Glu186Gly)	AR_comphet	3.7	comphet	maternal&paternal	2	NDD + Epilepsy	(+) Complex febrile seizure,(+) Simple febrile seizure,(+) Seizure,(-) Motor delay,(-) Intellectual disability

STARD9	NM_020759.2:c.4693A>Gp.(Ser1565Gly)	NM_020759.2:c.5795A>Gp.(Asn1932Ser)	AR_comphet	3.7	comphet	maternal&paternal	2	NDD + Epilepsy	(+) Complex febrile seizure,(+) Simple febrile seizure,(+) Seizure,(-) Motor delay,(-) Intellectual disability
NLRP5	NM_153447.4:c.1846_1849delp.(Lys616Glyfs*17)		AR_homo	8.0	homo	maternal&paternal	2	NDD + Epilepsy	(+) Dravet syndrome,(+) Seizure,(+) Myoclonic seizure,(+) Myoclonic absence seizure,(+) Global developmental delay,(+) Intellectual disability
CCDC136	NM_022742.4:c.1018C>Tp.(Arg340Trp)	NM_022742.4:c.1079G>Ap.(Ser360Asn)	AR_comphet	4.9	comphet	maternal&paternal	2	NDD + Epilepsy	(+) Intellectual disability,(+) Arthrogryposis multiplex congenita,(+) Polymicrogyria,(+) Seizure
MDN1	NM_014611.3:c.11732G>Cp.(Ser3911Thr)		AD_denovo	7.4	het	de novo	1	NDD	Global developmental delay, Delayed gross motor development, Macrocephaly, Patent foramen ovale
SUPV3L1	NM_003171.4:c.1931G>Ap.(Arg644Gln)	NM_003171.4:c.2358C>Gp.(Asp786Glu)	AR_comphet	5.6	comphet	maternal&paternal	1	NDD + Epilepsy	(+) Global developmental delay,(+) Focal-onset seizure,(+) Abnormality of the nasal alae,(+) Poor eye contact
RYR2	NM_001035.3:c.6202C>Tp.(Arg2068*)		AD_denovo	11.5	het	de novo	2	NDD + Epilepsy	(+) Dravet syndrome,(+) Seizure,(+) Myoclonic seizure,(+) Myoclonic absence seizure,(+) Global developmental delay,(+) Intellectual disability
RHBDL1	NM_001318733.1:c.1127C>Ap.(Ala376Glu)		AD_denovo	5.6	het	de novo	1	NDD + Epilepsy	Focal-onset seizure, Seizure, Encephalopathy, Focal cortical dysplasia
ATP6AP2	NM_005765.3:c.858G>Ap.(Ala286=)		AD_denovo	8.0	het	de novo	1	NDD	(+) Moderate global developmental delay,(+) Muscular hypotonia,(+) Dysgenesis of the hippocampus,(+) Aggressive behavior,(+) Impulsivity,(+) Low frustration tolerance,(+) Pes planus,(+) Synophrys,(-) Seizure,(-) Ataxia
DNAH3	NM_017539.2:c.7420A>Tp.(Lys2474*)	NM_017539.2:c.5287G>Ap.(Val1763Met)	AR_comphet	5.8	comphet	maternal&paternal	1	NDD	(+) Global developmental delay,(+) Delayed speech and language development,(+) Autistic behavior,(+) Hearing impairment,(+) Developmental regression
PCDH11X	NM_032968.4:c.1688A>Gp.(Gln563Arg)		XL	5.9	hemi	maternal	1	NDD + Epilepsy	(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Short attention span,(+) Specific learning disability,(+) Generalized non-motor (absence) seizure,(+) Headache,(+) Recurrent infections
PNCK	NM_001135740.1:c.643C>Gp.(Leu215Val)		XL	4.6	hemi	maternal	1	NDD	(+) Neurodevelopmental delay,(+) Mild expressive language delay,(+) Morphological central nervous system abnormality,(+) Hydromyelia,(+) Achilles tendon contracture,(+) Testicular torsion,(+) Syringomyelia,(+) Sleep disturbance,(+) Limited hip extension,(+) Spastic paraplegia,(+) Motor delay
ZBTB45	NM_001316978.2:c.976G>Ap.(Gly326Arg)		AR_homo	4.0	homo	maternal&paternal	2	NDD + Epilepsy	(+) Focal-onset seizure,(+) Brain imaging abnormality
NOMO1	NM_014287.4:c.2173G>Ap.(Gly725Ser)		AR_homo	4.4	homo	maternal&paternal	2	NDD + Epilepsy	(+) Focal-onset seizure,(+) Brain imaging abnormality
PLXNA3	NM_017514.5:c.1015C>Gp.(Leu339Val)		XL	6.1	hemi	maternal	2	NDD + Epilepsy	(+) Infantile encephalopathy,(+) Microcephaly,(+) Short stature,(+) Muscular hypotonia,(+) Micropenis,(+) Global developmental delay,(+) Abnormal facial shape,(+) Cerebral ischemia,(+) Focal-onset seizure,(+) Epicanthus,(+) Decreased body weight,(+) Oxycephaly,(+) Hypospadias,(+) Cryptorchidism
SMYD5	NM_006062.3:c.100A>Gp.(Lys34Glu)	NM_006062.3:c.833G>Ap.(Arg278His)	AR_comphet	4.2	comphet	maternal&paternal	2	NDD + Epilepsy	(+) Infantile encephalopathy,(+) Microcephaly,(+) Short stature,(+) Muscular hypotonia,(+) Micropenis,(+) Global developmental delay,(+) Abnormal facial shape,(+) Cerebral ischemia,(+) Focal-onset seizure,(+) Epicanthus,(+) Decreased body weight,(+) Oxycephaly,(+) Hypospadias,(+) Cryptorchidism
GIGYF1	NM_022574.4:c.1778A>Tp.(Asp593Val)		AD_denovo	B	het	de novo	2	Wachstum, Skelett	(+) Cleft soft palate,(+) Cleft hard palate
MAP3K6	NM_004672.4:c.3789-5C>Tp.?	NM_004672.4:c.1733T>Ap.(Val578Asp)	AR_comphet	C	comphet	maternal&paternal	2	Wachstum, Skelett	(+) Cleft soft palate,(+) Cleft hard palate
MAGIX	NM_024859.3:c.851C>Tp.(Pro284Leu)		XL	3.0	hemi	maternal	2	NDD	(+) Abnormal macular morphology,(+) Subretinal deposits,(+) Motor delay,(+) Global developmental delay,(+) Attention deficit hyperactivity disorder

ZNF283	NM_181845.1:c.1927G>Tp.(Val643Phe)	NM_181845.1:c.1342C>Ap.(Gln448Lys)	AR_comphet	2.2	comphet	maternal&paternal	2	NDD	(+) Abnormal macular morphology,(+) Subretinal deposits,(+) Motor delay,(+) Global developmental delay,(+) Attention deficit hyperactivity disorder
TMEM143	NM_018273.3:c.1022T>Cp.(Met341Thr)		AD_denovo	4.4	het	de novo	3	NDD + Epilepsy	(+) Focal tonic seizure,(+) EEG with focal sharp waves,(+) Nocturnal seizures,(-) Brain imaging abnormality
FAM214B	NM_001317991.1:c.1012C>Gp.(Pro338Ala)		AR_homo	5.6	homo	maternal&paternal	3	NDD + Epilepsy	(+) Focal tonic seizure,(+) EEG with focal sharp waves,(+) Nocturnal seizures,(-) Brain imaging abnormality
STX4	NM_004604.4:c.118_120delp.(Glu40del)		AR_homo	5.6	homo	maternal&paternal	3	NDD + Epilepsy	(+) Focal tonic seizure,(+) EEG with focal sharp waves,(+) Nocturnal seizures,(-) Brain imaging abnormality
SEMA5A	NM_003966.3:c.2123C>Tp.(Thr708Met)		AR_homo	8.3	homo	maternal&paternal	6	NDD	(+) Severe global developmental delay,(+) Intellectual disability,(+) Feeding difficulties,(+) Muscular hypotonia
ATP6V0A1	NM_001130021.3:c.2219G>Ap.(Arg740Gln)		AD_unknown	5.3	het	unknown	1	NDD + Epilepsy	(+) Seizure,(+) Large for gestational age,(+) Microcephaly,(+) Global developmental delay,(+) Muscular hypotonia
ADGRD2	NM_001161808.1:c.1068C>Ap.(Cys356*)		AD_denovo	5.0	het	de novo	1	NDD	(+) Global developmental delay,(+) Motor delay,(+) Neonatal asphyxia,(+) Neonatal seizure,(+) Hypertonia,(+) Dysphagia,(+) Tongue fasciculations,(+) Microcephaly,(+) Infantile encephalopathy
AHNAK	NM_001620.2:c.11743G>Ap.(Asp3915Asn)		AR_homo	6.4	homo	maternal&paternal	3	NDD	(+) Global developmental delay,(+) Motor delay,(+) Cleft palate,(+) Cleft lip,(+) Cerebellar hypoplasia
DHRS3	NM_004753.6:c.730G>Cp.(Glu244Gln)		AR_homo	5.6	homo	maternal&paternal	3	NDD	(+) Global developmental delay,(+) Motor delay,(+) Cleft palate,(+) Cleft lip,(+) Cerebellar hypoplasia
TRPM2	NM_003307.3:c.2392G>Tp.(Val798Phe)		AR_homo	5.6	homo	maternal&paternal	3	NDD	(+) Global developmental delay,(+) Motor delay,(+) Cleft palate,(+) Cleft lip,(+) Cerebellar hypoplasia
MAGEA10	NM_001011543.2:c.229G>Tp.(Asp77Tyr)		XL	C	hemi	maternal	2	Wachstum, Skelett	(+) Trigonocephaly
OAS3	NM_006187.3:c.101G>Ap.(Gly34Asp)	NM_006187.3:c.1443C>Ap.(Asn481Lys)	AR_comphet	C	comphet	maternal&paternal	1	Wachstum, Skelett	(+) Trigonocephaly
POLR3E	NM_018119.3:c.437A>Gp.(Asp146Gly)		AD_denovo	A	het	de novo	2	Stoffwechsel	(+) Low levels of vitamin A,(+) Low levels of vitamin D,(+) Leukopenia,(+) Thrombocytopenia,(+) Hepatosplenomegaly,(+) Portal vein thrombosis
TENM2	NM_001122679.1:c.3262A>Tp.(Ile1088Phe)	NM_001122679.1:c.6169C>Tp.(Arg2057Trp)	AR_comphet	C	comphet	maternal&paternal	2	Stoffwechsel	(+) Low levels of vitamin A,(+) Low levels of vitamin D,(+) Leukopenia,(+) Thrombocytopenia,(+) Hepatosplenomegaly,(+) Portal vein thrombosis
ZFHX3	NM_006885.3:c.5449G>Tp.(Val1817Leu)	NM_006885.3:c.2321C>Tp.(Ala774Val)	AR_comphet	5.4	comphet	maternal&paternal	2	NDD + Epilepsy	(+) Intellectual disability,(+) Seizure,(+) Polymicrogyria,(+) Arthrogryposis multiplex congenita
PTPRH	NM_002842.4:c.1324G>Ap.(Ala442Thr)	NM_002842.4:c.683G>Ap.(Trp228*)	AR_comphet	B	comphet	maternal&paternal	1	Auge	(+) Optic neuropathy,(+) Amblyopia,(+) Nystagmus,(+) Strabismus,(+) Mixed astigmatism,(+) Protanomaly
BTBD18	ENST00000422652.1:c.1236dup, p.Arg413*		AD_denovo	A	het	de novo	2	rennendungs	Cleft palate, renal agnesia left
PLEKHB2	ENST00000409158.1:c.83C>T p.Ser28Leu		AR_homo	C	homo	maternal&paternal	2	Fehlbildungen	(+) Cleft lip,(+) Cleft palate,(+) Unilateral renal agenesis
HDAC6	ENST00000334136.5:c.3248G>A p.Gly1083Asp		XL	C	hemi	maternal	2	Wachstum, Skelett	Trigonocephaly, Abnormality of calvarial morphology
ZBTB12	ENST00000375527.2:c.583G>A p.Glu195Lys		AD_denovo	5.0	het	de novo	2	NDD + Epilepsy	(+) Seizure,(+) Global developmental delay,(+) Intellectual disability
ADI1	ENST00000327435.6:c.214G>A p.Asp72Asn	ENST00000327435.6:c.166C>T p.Arg56*	AR_comphet	4.9	comphet	maternal&paternal	2	NDD + Epilepsy	(+) Seizure,(+) Global developmental delay,(+) Intellectual disability
PPP2R5C	ENST00000422945.2:c.1341A>T p.Lys447Asn		AD_unknown	5.5	het	unknown	1	NDD + Epilepsy	(+) Seizure,(+) Global developmental delay,(+) Hemimegalencephaly

FAM171A2	ENST00000293443.7:c.1170del p.Glu391Argfs*67		AR_homo	8.2	homo	maternal&paternal	2	NDD	(+) Intellectual disability,(+) Microcephaly
JMJD1C	ENST00000399262.2:c.1372G>A p.Glu458Lys		AR_homo	7.6	homo	maternal&paternal	2	NDD	(+) Intellectual disability,(+) Microcephaly
RC3H2	ENST00000373670.1:c.382C>A p.Arg128Ser		AD_unknown	4.1	het	unknown	1	NDD + epilepsy	(+) Focal tonic seizure,(+) Focal myoclonic seizure,(+) Atypical absence seizure,(+) Intellectual disability, mild
PHF20	ENST00000374012.3:c.1300A>G p.Lys434Glu		AD_unknown	3.6	het	unknown	2	NDD	(+) Microcephaly,(+) Plagiocephaly,(+) Ventricular septal defect,(+) Short palpebral fissure,(+) Smooth philtrum,(+) Thin upper lip vermillion,(+) Short stature,(+) Absent speech,(+) Motor delay
FAT3	ENST00000298047.6:c.5027A>G p.Tyr1676Cys	ENST00000298047.6:c.10393A>G p.Ile3465Val	AR_comphet	4.7	comphet?	unknown	2	NDD	(+) Microcephaly,(+) Plagiocephaly,(+) Ventricular septal defect,(+) Short palpebral fissure,(+) Smooth philtrum,(+) Thin upper lip vermillion,(+) Short stature,(+) Absent speech,(+) Motor delay
NEFM	ENST00000221166.5:c.446C>G p.Ala149Gly		AD_denovo	7.1	het	de novo	1	NDD + epilepsy	(+) Global developmental delay,(+) Intellectual disability,(+) Behavioral abnormality,(+) Short stature,(+) Focal motor seizure,(+) Focal-onset seizure,(+) Bilateral tonic-clonic seizure with focal onset
PTPN21	ENST00000556564.1:c.1675C>T p.Arg559Trp	ENST00000556564.1:c.2269A>T p.Ile757Phe	AR_comphet	2.5	comphet	maternal&paternal	2	epilepsy	Seizure, abnormality of metabolism, epileptic encephalopathy
AWAT1	ENST00000374521.3:c.273C>G p.Asp91Glu		XL	3.2	hemi	maternal	3	epilepsy	intellectual disability, focal onset seizure, cortical dysplasia, brain atrophy
FAM171A1	ENST00000378116.4:c.364T>C p.Ser122Pro	ENST00000378116.4:c.1418A>G p.Glu473Gly	AR_comphet	3.4	comphet	maternal&paternal	3	epilepsy	intellectual disability, focal onset seizure, cortical dysplasia, brain atrophy
ZNRF4	ENST00000222033.4:c.1135C>G p.His379Asp		AD_denovo	4.5	het	de novo	3	epilepsy	intellectual disability, focal onset seizure, cortical dysplasia, brain atrophy
DCBLD1	ENST00000296955.8:c.1178G>A p.Arg393Gln		AR_homo	4.8	homo	maternal&paternal	6	NDD	Severe global developmental delay, Feeding difficulties, Muscular hypotonia
NCOA7	ENST00000368357.3:c.1396G>A p.Ala466Thr		AR_homo	3.3	homo	maternal&paternal	6	NDD	Severe global developmental delay, Feeding difficulties, Muscular hypotonia
SLC27A4	ENST00000300456.4:c.1462+5_1462+9del None		AR_homo	4.9	homo	maternal&paternal	6	NDD	Severe global developmental delay, Feeding difficulties, Muscular hypotonia
MTUS2	ENST00000431530.3:c.2752C>T p.Arg918Trp		AR_homo	4.3	homo	maternal&paternal	6	NDD	Severe global developmental delay, Feeding difficulties, Muscular hypotonia
STXBP4	ENST00000376352.2:c.866G>C p.Cys289Ser		AR_homo	3.9	homo	maternal&paternal	6	NDD	Severe global developmental delay, Feeding difficulties, Muscular hypotonia
GRIPAP1	ENST00000376441.1:c.1007A>G p.Asn336Ser		XL	5.9	hemi	maternal	2	NDD	(+) Intellectual disability,(+) Global developmental delay,(+) Abnormality of movement,(+) Dystonia,(+) Spasticity
H1FOO	ENST00000324382.2:c.863C>T p.Ala288Val		AD_denovo	4.2	het	de novo	2	NDD	(+) Intellectual disability,(+) Global developmental delay,(+) Abnormality of movement,(+) Dystonia,(+) Spasticity
NKPD1	ENST00000317951.4:c.1076A>G p.Tyr359Cys		AD_denovo	5.4	het	de novo	1	NDD	Caudal regression syndrome, Currarino Triad, Global developmental delay
HTR4	ENST00000360693.3:c.721C>T p.Gln241*		AD_unknown	6.8	het	unknown	2	NDD + epilepsy	Intellectual disability,(+) Atypical absence seizure,(+) Generalized tonic seizure,(+) Generalized-onset epileptic spasm,(+) Myoclonus,(+) Generalized atonic seizure,(+) Bilateral tonic-clonic seizure with generalized onset
NSD3	ENST00000317025.8:c.3725G>A p.Arg1242Gln		AD_unknown	5.7	het	unknown	2	NDD + epilepsy	Intellectual disability,(+) Atypical absence seizure,(+) Generalized tonic seizure,(+) Generalized-onset epileptic spasm,(+) Myoclonus,(+) Generalized atonic seizure,(+) Bilateral tonic-clonic seizure with generalized onset
ARHGEF2	ENST00000361247.4:c.355C>T p.Arg119Trp	ENST00000361247.4:c.415C>T p.Arg139Cys	AR_comphet	7.1	comphet	maternal&paternal	1	NDD + muscle	(+) Muscular hypotonia, (+) Increased serum lactate, (+) Motor delay, (+) Strabismus, (+) Reduced visual acuity, (+) Visual impairment

SHANK1	ENST00000293441.1:c.4932C>G p.Asp1644Glu		AD_unknown	7.1	het	unknown	1	NDD + Epilepsy	Typical absence seizure,(+) Myoclonic seizure,(+) Bilateral tonic-clonic seizure,(+) Intellectual disability, mild,(+) Intellectual disability, borderline,(+) EEG with spike-wave complexes (2.5-3.5 Hz)
NCKAP1	ENST00000360982.2:c.3366_3369del p.Tyr1122*		AD_denovo	11.7	het	de novo	1	NDD + Epilepsy	(+) Epicanthus,(+) Narrow face,(+) Anteverted nares,(+) High palate,(+) Global developmental delay,(+) Focal-onset seizure
NRXN3	NM_001330195.2(NRXN3):c.3985C>T		AD_inherited	8.4	het	maternal	1	NDD + Epilepsy	(+) Generalized non-motor (absence) seizure,(+) Attention deficit hyperactivity disorder,(+) Talipes cavus equinovarus,(+) Global developmental delay,(+) Low-frequency hearing loss
AFF3	ENST00000356421.2:c.3181G>A p.Val1061Ile	ENST00000356421.2:c.3632G>A p.Arg1211Gln	AR_comphet	4.9	comphet	maternal& paternal	1	NDD + epilepsy	(+) Epileptic encephalopathy,(+) Agenesis of corpus callosum,(+) Abnormal cortical gyration, (+) Hypomyelination
CPSF4	ENST00000292476.5:c.655C>T p.Pro219Ser		AD_denovo	7.0	het	de novo	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
PCDH1	ENST00000287008.3:c.3698G>A p.Arg1233His		AR_homo	5.0	homo	maternal& paternal	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
ADNP2	ENST00000262198.4:c.422T>G p.Ile141Ser		AR_homo	6.0	homo	maternal& paternal	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
HTR3B	ENST00000260191.2:c.550G>A p.Asp184Asn		AR_homo	5.3	homo	maternal& paternal	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
PPFIBP1	NM_177444.3:c.1197+1G>A, p.?		AR_homo	8.2	homo	maternal& paternal	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
ARHGEF12	ENST00000397843.2:c.3460_3462del p.Asn1154del		AR_homo	5.4	homo	maternal& paternal	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
ASUN	ENST00000261191.7:c.341G>A p.Arg114Gln		AR_homo	4.2	homo	maternal& paternal	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
TNRC18	ENST00000430969.1:c.4261_4262delinsGG p.Leu1421Gly		AR_homo	5.4	homo	maternal& paternal	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
CDC25C	ENST00000323760.6:c.1129T>C p.Cys377Arg		AR_homo	6.1	homo	maternal& paternal	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
HEXIM2			AR_homo	7.0	homo	maternal& paternal	10	NDD + Epilepsy	(+) Microcephaly,(+) Intellectual disability,(+) Cognitive impairment,(+) Seizure
KANK1	ENST00000382303.1:c.3733G>A p.Gly1245Arg	ENST00000382303.1:c.1652G>A p.Cys551Tyr	AR_comphet	6.3	comphet	maternal& paternal	3	NDD + Epilepsy	(+) Global developmental delay,(+) Infantile spasms,(+) Generalized-onset seizure,(+) Hearing impairment,(+) Epileptic encephalopathy
DRP2	ENST00000395209.3:c.575A>C p.Gln192Pro		XL	4.6	hemi	maternal	3	NDD + Epilepsy	(+) Global developmental delay,(+) Infantile spasms,(+) Generalized-onset seizure,(+) Hearing impairment,(+) Epileptic encephalopathy
RNF113A	ENST00000371442.2:c.265_270del p.Glu89_Glu90del		XL	5.0	hemi	maternal	3	NDD + Epilepsy	(+) Global developmental delay,(+) Infantile spasms,(+) Generalized-onset seizure,(+) Hearing impairment,(+) Epileptic encephalopathy
TSSC1	ENST00000382125.4:c.514G>A p.Val172Met		AD_denovo	5.2	het	de novo	3	Muskel	Motor delay, Muscular hypotonia, Skeletal muscle atrophy
RFX7	NM_022841.5 :c.3083C>T p.(Pro1028Leu)		AD_denovo	6.7	het	de novo	1	NDD + Epilepsy	Congenital cataract, Optic nerve hypoplasia, Delayed speech and language development, Intellectual disability, Seizures, Apnea, Generalized myoclonic seizures, Abnormality of the basal ganglia, Delayed CNS myelination, Sleep disturbance, Focal seizures with impairment of consciousness or awareness, Abnormality of brain morphology, Abnormal myelination, Delayed myelination, Infantile spasms, Abnormality of movement
NKTR	ENST00000232978.8:c.2511_2514del p.Gln838Lysfs*23		AD_denovo	10.2	het	de novo	3	Muskel	Motor delay, Muscular hypotonia, Skeletal muscle atrophy
DRP2	ENST00000395209.3:c.2438C>T p.Ala813Val		XL	4.9	hemi	maternal	3	Muskel	Motor delay, Muscular hypotonia, Skeletal muscle atrophy
KCNRG	ENST00000312942.1:c.394dup, p.Thr132Asnfs*3		AR_homo	8.0	homo	maternal& paternal	1	NDD	(+) Global developmental delay,(+) Cognitive impairment,(+) Autism,(+) Autistic behavior
ERVMER34-1	ENST00000443173.1:c.936A>T p.Lys312Asn		AD_denovo	B	het	de novo	1	other	(+) Intrauterine growth retardation,(+) Oligohydramnios
CELSR3	ENST00000164024.4:c.5751+1G>C None		AR_homo	11.2	homo	maternal& paternal	1		(+) Focal-onset seizure,(+) Generalized-onset seizure,(+) Global developmental delay,(+) Dystonia,(+) Cerebral white matter agenesis,(+) Microcephaly

ITGAM	ENST00000544665.3:c.2923C>T p.Pro975Ser		AD_denovo	6.5	het	de novo	1	NDD + Wachstum	Failure to thrive, Short stature, Feeding difficulties, Hepatomegaly, Atrial septal defect, Ab-dominal distention, Global developmental delay, Congenital microcephaly, Plagiocephaly, Dysmorphic facial features
ALDH3B2	ENST00000349015.3:c.505G>A p.Val169Ile	ENST00000349015.3:c.635G>A p.Arg212Gln	AR_comphet	C	comphet	maternal&paternal	1	congenital heart defects	Unbalanced atrioventricular canal defect, Anomalous pulmonary venous return, Congenital malformation of the great arteries, Bradycardia
NME4	ENST00000219479.2:c.1A>T p.Met1?		AR_homo	7.78	homo	unknown	1	NDD + Epilepsy	Moderate intellectual disability, delayed speech and language development, absence seizure, focal impaired awareness motor seizure, bilateral tonic-clonic seizure with generalized onset, muscular hypotonia, joint laxity, abnormal facial shape, temporal lobe sclerosis right (Hippocampectomy 01/2005), hypogonadotropic hypogonadism
YWHAB	ENST00000372839.3:c.637T>C p.Tyr213His		AD_denovo	7.2	het	de novo	1	NDD + Epilepsy	(+) Seizure,(+) Global developmental delay
DNAH6	ENST00000237449:c.11360G>A p.Gly3787Asp		AD_inherited	5.0	het	maternal	1	Epilepsy	(+) Generalized-onset seizure,(+) Focal motor seizure,(+) EEG abnormality,(+) Mild short stature,(+) Microcephaly,(+) Decreased glucose-6-phosphate dehydrogenase level in blood
CGB1	ENST00000301407.7:c.290T>C p.Val97Ala	ENST00000301407.7:c.401A>G p.Gln134Arg	AR_comphet	2.33	comphet	maternal&paternal	1	NDD + epilepsy	(+) Ataxia,(+) Intellectual disability,(+) Myoclonic spasms,(+) Epileptic spasm,(+) Seizure,(-) Abnormality of the face
ALS2CL	ENST00000318962.4:c.893C>T p.Ala298Val	ENST00000318962.4:c.2704G>A p.Glu902Lys	AR_comphet	3.3	comphet	maternal&paternal	1	epilepsy	(+) Myoclonic seizure,(+) Generalized myoclonic-tonic-clonic seizure,(+) Ataxia,(+) Suicidal ideation
WDFY4	ENST00000325239.5:c.3175+2del None		AD_unknown	6.2	het	unknown	1	NDD	(+)Global developmental delay,(+) Delayed speech and language development,(+) Muscular hypotonia,(+) Anal atresia,(+) Perineal fistula,(+) Atrial septal defect,(+) Dextrocardia,(+) Hearing impairment,(+) Unilateral ptosis,(+) Posterior plagiocephaly,(+) Scoliosis,(+) Low-set ears,(+) Retrognathia,(+) Abnormality of the philtrum,(+) Bilateral single transverse palmar creases,(+) Abnormality of toe
GRIK3	ENST00000373091.3:c.176C>T p.Ala59Val		AD_unknown	5.44	het	unknown	1	NDD	(+) Global developmental delay,(+) Ataxia,(+) Muscular hypotonia,(+) Macrocephaly,(+) Tall stature,(+) Obesity
CHD8	ENST00000399982.2:c.4418G>T p.Arg1473Leu		AD_unknown	6.61	het	unknown	1	NDD + Epilepsy	(+) Tonic seizure,(+) Intellectual disability, severe,(+) Kyphoscoliosis,(+) Hyperlordosis,(+) Focal polymicrogyria,(+) Frontoparietal polymicrogyria,(+) Global brain atrophy,(+) EEG with focal epileptiform discharges,(+) Bilateral tonic-clonic seizure,(+) Absent speech
EHMT2	ENST00000375537.4:c.912_914del p.Glu323del	ENST00000375537.4:c.1509G>A p.Ala503=	AR_comphet	6.33	comphet	maternal&paternal	1	NDD	(+) Intellectual disability,(+) Global developmental delay,(+) Microcephaly,(+) Behavioral abnormality,(+) 2-3 toe syndactyly
FADS1	ENST00000350997.7:c.238G>A p.Asp80Asn		AR_homo	B	homo	maternal&paternal	4	NDD	(+) Double outlet right ventricle,(+) Pulmonic stenosis,(+) Failure to thrive,(+) Frontal hirsutism,(+) Low-set ears,(+) Narrow face,(+) Hearing impairment
RCOR2	ENST00000301459.4:c.1376C>T p.Thr459Met		AR_homo	B	homo	maternal&paternal	4	NDD	(+) Double outlet right ventricle,(+) Pulmonic stenosis,(+) Failure to thrive,(+) Frontal hirsutism,(+) Low-set ears,(+) Narrow face,(+) Hearing impairment
SRGAP1	ENST00000355086.3:c.1421A>G p.Glu474Gly	ENST00000355086.3:c.1217G>A p.Arg406His	AR_comphet	4.8	comphet	maternal&paternal	1	Epilepsy	Generalized-onset seizure, Bilateral tonic-clonic seizure, Focal-onset seizure, EEG with spike-wave complexes

GAL3ST4	ENST00000360039.4:c.1207_1208insC p.Leu403Profs*10		AR_homo	8.0	homo	maternal&paternal	1	NDD	(+) Profound global developmental delay,(+) Muscular hypotonia,(+) Abnormality of the Achilles tendon,(+) Abnormal foot morphology,(+) Increased lactate dehydrogenase level,(+) Increased serum lactate,(+) Delayed CNS myelination,(+) Hypoplasia of the corpus callosum,(+) Abnormal macular morphology,(-) Abnormal facial shape
PER1	ENST00000317276.4:c.694G>C p.Val232Leu	ENST00000317276.4:c.3373G>A p.Val1125Met	AR_comphet	C	comphet	maternal&paternal	4	NDD	(+) Double outlet right ventricle,(+) Pulmonic stenosis,(+) Failure to thrive,(+) Frontal hirsutism,(+) Low-set ears,(+) Narrow face,(+) Hearing impairment
HECTD1	ENST00000399332.1:c.5140C>T p.Arg1714Cys	ENST00000399332.1:c.6725C>T p.Thr2242Met	AR_comphet	C	comphet	maternal&paternal	4	NDD	(+) Double outlet right ventricle,(+) Pulmonic stenosis,(+) Failure to thrive,(+) Frontal hirsutism,(+) Low-set ears,(+) Narrow face,(+) Hearing impairment
TNRC18	ENST00000430969.1:c.690G>T p.Glu230Asp	ENST00000430969.1:c.5525C>T p.Ala1842Val	AR_comphet	4.4	comphet	maternal&paternal	3	Neuro	Leukodystrophy, Leukoencephalopathy, Strabismus (normal development)
NCOR1	ENST00000268712.3:c.3360G>C p.Glu1120Asp	ENST00000268712.3:c.5240G>A p.Arg1747Gln	AR_comphet	5.7	comphet	maternal&paternal	3	Neuro	Leukodystrophy, Leukoencephalopathy, Strabismus (normal development)
TMEM205	ENST00000354882.5:c.326G>A p.Arg109His		AR_homo	3.8	homo	maternal&paternal	3	Neuro	Leukodystrophy, Leukoencephalopathy, Strabismus (normal development)
CROCC	ENST00000375541.5:c.5585G>A p.Arg1862Gln	ENST00000375541.5:c.736G>C p.Ala246Pro	AR_comphet	4.8	comphet?	unknown	4	NDD + Epilepsy	Bilateral tonic-clonic seizure with focal onset, Hypothyroidism, Hepatosplenomegaly, Intellectual disability, Global developmental delay, EEG abnormality, EEG with focal sharp waves, Cranial hyperostosis, Poor speech
USP21	ENST00000368002.3:c.935G>A p.Arg312Gln	ENST00000368002.3:c.112C>T p.Arg38Cys	AR_comphet	2.9	comphet?	unknown	4	NDD + Epilepsy	Bilateral tonic-clonic seizure with focal onset, Hypothyroidism, Hepatosplenomegaly, Intellectual disability, Global developmental delay, EEG abnormality, EEG with focal sharp waves, Cranial hyperostosis, Poor speech
KIAA1407	ENST00000295878.3:c.89A>C p.Lys30Thr	ENST00000295878.3:c.1035dup, p.Lys346Glufs*7	AR_comphet	4.3	comphet?	unknown	4	NDD + Epilepsy	Bilateral tonic-clonic seizure with focal onset, Hypothyroidism, Hepatosplenomegaly, Intellectual disability, Global developmental delay, EEG abnormality, EEG with focal sharp waves, Cranial hyperostosis, Poor speech
RBM19	ENST00000545145.2:c.520T>G p.Ser174Ala	ENST00000545145.2:c.1247A>G p.Glu416Gly	AR_comphet	4.6	comphet?	unknown	4	NDD + Epilepsy	Bilateral tonic-clonic seizure with focal onset, Hypothyroidism, Hepatosplenomegaly, Intellectual disability, Global developmental delay, EEG abnormality, EEG with focal sharp waves, Cranial hyperostosis, Poor speech
TRIM14	ENST00000341469.2:c.1104C>A p.Asp368Glu		AD_inherited	B	het	maternal	1	immunology	(+) Recurrent infections,(+) Sepsis,(+) Affected mother
SCAF8	ENST00000367186.4:c.119dup, p.Leu41Profs*14		AD_unknown	6.06	het	unknown	1	NDD + epilepsy	(+) Intellectual disability, severe,(+) Severe global developmental delay,(+) Bilateral tonic-clonic seizure with focal onset,(+) Cataract,(+) Abnormality of the kidney,(+) EEG abnormality

PRKRIR	ENST00000260045.3:c.2274_2275delinsCT p.Glu759*		AD_unknown	B	het	unknown	1	Muskel	Maligne Hyperthermie
TRANK1	ENST00000429976.2:c.4634A>G p.Asn1545Ser		AR_homo	B	homo	maternal&paternal	4	other	Precocious puberty, Tremor, Hypertrichosis, Hirsutism, Increased head circumference, Increased body weight, Acne
MAP7D1	ENST00000373151.2:c.2003A>C p.Glu668Ala		AR_homo	B	homo	maternal&paternal	4	other	Precocious puberty, Tremor, Hypertrichosis, Hirsutism, Increased head circumference, Increased body weight, Acne
NME6	ENST00000421967.1:c.548A>T p.His183Leu		AR_homo	B	homo	maternal&paternal	4	other	Precocious puberty, Tremor, Hypertrichosis, Hirsutism, Increased head circumference, Increased body weight, Acne
PHC3	ENST00000495893.2:c.959A>G p.His320Arg		AR_homo	B	homo	maternal&paternal	4	other	Precocious puberty, Tremor, Hypertrichosis, Hirsutism, Increased head circumference, Increased body weight, Acne
GPR124	ENST00000412232.2:c.1579C>T p.Leu527Phe		AD_denovo	5.9	het	de novo	1	NDD + epilepsy	Intellectual disability, moderate, Global developmental delay, Focal-onset seizure, Generalized-onset seizure, Abnormality of brain morphology in MRI , Muscle weakness of the right side of the body
TIMP1	ENST00000218388:c.224T>C p.Leu75Ser		XL	4.33	hemi	maternal	2	NDD	Mental retardation
SEMA4B	ENST00000411539:c.1044-8C>T None	ENST00000411539:c.2320G>A p.Gly774Ser	AR_comphet	3.78	comphet	maternal&paternal	2	NDD	Mental retardation
GOLGA2	ENST00000421699:c.2414del p.Met805Argfs*18		AD_unknown	8.8	het	unknown	1	NDD	Intellectual disability, Abnormal facial shape
ATP13A3	ENST00000439040.5:c.2638A>T p.(Met880Leu)		AD_unknown	C	het	unknown	1	Wachstum, Skelett	(+) Mild short stature
SMARCA1	ENST00000371122:c.2402A>G p.Glu801Gly		XL	7.67	hemi	unknown	1	NDD + Epilepsy	Intellectual disability, severe,(+) Severe global developmental delay,(+) EEG abnormality,(+) Generalized tonic seizure,(+) Bilateral tonic-clonic seizure with generalized onset,(+) Status epilepticus,(+) Spastic tetraparesis,(+) Bilateral talipes equinovarus,(+) Pilomatrixoma
SPRED3	ENST00000338502:c.1210C>T p.Arg404Cys		AD_denovo	5.4	het	de novo	2	NDD + epilepsy	(+) Atonic seizure,(+) Generalized clonic seizure,(+) Generalized tonic seizure,(+) Intellectual disability, mild,(+) Gastroesophageal reflux,(+) Postnatal microcephaly
PIPOX	ENST00000323372.4:c.28G>T p.Ala10Ser	ENST00000323372.4:c.514G>A p.Gly172Arg	AR_comphet	4.3	comphet	maternal&paternal	2	NDD + epilepsy	(+) Atonic seizure,(+) Generalized clonic seizure,(+) Generalized tonic seizure,(+) Intellectual disability, mild,(+) Gastroesophageal reflux,(+) Postnatal microcephaly
CCDC180	ENST00000375202:c.820C>T p.Arg274*	ENST00000375202:c.4179+5G>C None	AR_comphet	3.8	comphet?	unknown	1	NDD	Global developmental delay, Aggressive behavior
NSF	ENST00000398238:c.2218C>A p.Pro740Thr		AD_unknown	6.09	het	unknown	1	NDD + Epilepsy	myoklonische Anfälle, komplexe Partialanfälle sekundärer Generalisierung, V.a. Absencen, schwere Intelligenzminderung, Entwicklungsstörung keine Kontaktaufnahme, Strabismus divergens, Nystagmus, Okulomotoriusparese, beginnende Cerebralparese, muskuläre Hypotonie, Optikusatrophy bei Netzhautdystrophie, komplexe Hirnfehlbildungen: Aphasie des Nucleus caudatus und Potamen rechts, Hypoplasie des Balkens, Polygyrie, höhergradige Atrophie der linken Kleinhirnhemisphäre
ITPK1	ENST00000267615:c.899_900insGA p.Gly301Lysfs*6		AD_unknown	6.1	het	unknown	1	Epilepsy	fokale Epilepsie refraktär auf Levetiracetam und Valproat, bislang unauffällige Entwicklung
EIF5B	ENST00000289371:c.1360del p.Ile454Tyrf*5		AD_unknown	6.8	het	unknown	2	NDD + Epilepsy	(+) Intellectual disability, severe,(+) Stereotypical hand wringing,(+) Self-injurious behavior,(+) Obsessive-compulsive behavior,(+) Seizure,(+) Scoliosis
MARK2	ENST00000402010:c.1934+1G>A None		AD_unknown	7.6	het	unknown	2	NDD + Epilepsy	(+) Intellectual disability, severe,(+) Stereotypical hand wringing,(+) Self-injurious behavior,(+) Obsessive-compulsive behavior,(+) Seizure,(+) Scoliosis
NRCAM	ENST00000379028:c.2738G>A p.Gly913Asp	ENST00000379028:c.2491C>A p.Pro831Thr	AR_comphet	7.8	comphet?	unknown	1	NDD + Epilepsy	(+) Intellectual disability,(+) Global developmental delay,(+) Seizure,(+) Motor delay,(+) EEG abnormality,(+) Poor coordination,(+) Delayed speech and language development,(+) Cafe-au-lait spot,(+) Autism

TBC1D7	ENST00000606214:c.728T>C p.Leu243Ser		AR_homo	7.3	homo	maternal&paternal	2	NDD + epilepsy	Entwicklungsverzögerung, atone Anfälle, sporadisches Lennox-Gastaut-Syndrom, Intelligenzminderung, ASD
STRAP	ENST00000419869:c.41C>T p.Thr14Met		AD_denovo	7.01	het	de novo	2	NDD + epilepsy	Entwicklungsverzögerung, atone Anfälle, sporadisches Lennox-Gastaut-Syndrom, Intelligenzminderung, ASD
NYAP1	NM_173564.4:c.2426T>G		AD_denovo	6.2	het	de novo	2	NDD + Epilepsy	(+) Moderate global developmental delay,(+) Muscular hypotonia,(+) Molar tooth sign on MRI,(+) Developmental cataract,(+) Febrile seizure (within the age range of 3 months to 6 years)
ITSN1	ENST00000381318:c.3997T>C p.Cys1333Arg		AD_unknown	C	het	unknown	1	Neuro	(+) Semantic dementia,(+) Frontotemporal dementia,(+) Tremor
SLITRK4	ENST00000381779:c.1282C>T p.Arg428Cys		XL	4.83	hemi	maternal	2	NDD + Epilepsy	(+) Focal-onset seizure,(+) Dandy-Walker malformation,(+) Overgrowth,(+) Global developmental delay
MOSPD2	ENST00000380492:c.1427G>A p.Arg476His		XL	3.9	hemi	maternal	2	NDD + Epilepsy	(+) Focal-onset seizure,(+) Dandy-Walker malformation,(+) Overgrowth,(+) Global developmental delay
GTF3A	ENST00000381140:c.55G>A p.Ala19Thr		AD_denovo	5.35	het	de novo	2	NDD + Epilepsy	West-Syndrom, developmental delay, Neuroblastom
HCN2	ENST00000251287:c.2156_2164dup, p.Pro719_Pro721dup		AD_unknown	C	het	unknown	1	Neuro	(+) Multifocal cerebral white matter abnormalities,(+) Abnormality of the periventricular white matter
NUDT21	ENST00000300291:c.187A>G p.Arg63Gly		AD_unknown	3.9	het	unknown	1	NDD	(+) Global developmental delay,(+) Short stature,(+) Microcephaly,(+) Failure to thrive,(+) Short toe,(+) Abnormality of the face
AOX1	ENST00000374700:c.2024T>C p.Val675Ala	ENST00000374700:c.3478G>A p.Glu1160Lys	AR_comphet	3.62	comphet?	unknown	2	NDD + Epilepsy	West-Syndrom, developmental delay, Neuroblastom
EZH1	ENST00000428826:c.1691A>G p.Lys564Arg		AD_inherited	C	het	maternal	3	Immunology	(+) Abnormal oral mucosa morphology,(+) Aphthous ulcer,(+) Recurrent aphthous stomatitis,(+) Abnormal blistering of the skin,(+) Oral mucosal blisters,(+) Genital blistering,(+) Arthralgia,(+) Nausea,(+) Episodic fatigue,(+) Vertigo
INTS2	ENST00000444766:c.650A>T p.Asn217Ile		AD_inherited	C	het	maternal	3	Immunology	(+) Abnormal oral mucosa morphology,(+) Aphthous ulcer,(+) Recurrent aphthous stomatitis,(+) Abnormal blistering of the skin,(+) Oral mucosal blisters,(+) Genital blistering,(+) Arthralgia,(+) Nausea,(+) Episodic fatigue,(+) Vertigo
LMTK3	ENST00000270238:c.1460C>T p.Pro487Leu		AD_inherited	C	het	maternal	3	Immunology	(+) Abnormal oral mucosa morphology,(+) Aphthous ulcer,(+) Recurrent aphthous stomatitis,(+) Abnormal blistering of the skin,(+) Oral mucosal blisters,(+) Genital blistering,(+) Arthralgia,(+) Nausea,(+) Episodic fatigue,(+) Vertigo
FAM184B	ENST00000265018:c.2750T>C p.Leu917Pro	ENST00000265018:c.1634G>T p.Gly545Val	AR_comphet	3.54	comphet	maternal&paternal	2	Epilepsy	(+) EEG abnormality,(+) Generalized-onset seizure,(+) Autism,(+) Mild global developmental delay
TACC2	ENST00000369005:c.6763G>T p.Asp2255Tyr	ENST00000369005:c.7316G>A p.Arg2439Gln	AR_comphet	3.73	comphet	maternal&paternal	2	Epilepsy	(+) EEG abnormality,(+) Generalized-onset seizure,(+) Autism,(+) Mild global developmental delay
POTEE	ENST00000356920:c.795C>G p.Ile265Met	ENST00000356920:c.1672A>G p.Asn558Asp	AR_comphet	2.64	comphet	maternal&paternal	1	NDD	(+) Global developmental delay,(+) Intellectual disability,(+) Cataract, facial dysmorphism,(+) Behavioral abnormality, known micro duplication 16p13.11 (in 2 of 3 sisters)
PTPRD	ENST00000381196:c.3988G>A p.Gly1330Ser	ENST00000381196:c.1372G>A p.Asp458Asn	AR_comphet	C	comphet	maternal&paternal	3	Fehlbildungen	hypotrophes Neugeborenes (Gewicht 5P, Länge 1P, Kopf 50P, 1z), Plagiozephalus DD Brachyzephalus, präaxiale Polydaktylie Typ 1 mit biphalangaealem Daumen rechts, V.a. bikuspidale Aortenklappe, Harntransprotströrung I-II° rechts und I° links, Neugeborenen-Hörscreening auffällig, Rektumstenose (Stoma), V.a. VACTERL-Assoziation (4/7 Symptomen), Körpermaße zur Vorstellung: Gewicht 52P, Größe 23P, Kopfumfang 10P

WDR5	ENST00000358625:c.620A>G p.Lys207Arg		AD_denovo	B	het	de novo	3	Fehlbildungen	(+) Abnormality of the urinary system,(+) Brachycephaly,(+) Hearing abnormality,(+) Preaxial hand polydactyly,(+) Plagiocephaly,(+) Bicuspid aortic valve,(+) Anal stenosis hypotrophes Neugeborenes (Gewicht 5P, Länge 1P, Kopf 50P, 1z), Plagiozephalus DD Brachyzephalus, präaxiale Polydaktylie Typ 1 mit biphalangealem Daumen rechts, V.a. bikuspidale Aortenklappe, Harntransprotströrung I-II° rechts und I° links, Neugeborenen-Hörscreening auffällig, Rektumstenose (Stoma), V.a. VACTERL-Assoziation (4/7 Symptomen), Körpermaße zur Vorstellung: Gewicht 52P, Größe 23P, Kopfumfang 10P
CKAP5	ENST00000529230:c.3056G>C p.Cys1019Ser		AD_unknown	5.78	het	unknown	2	NDD	(+) Moderate global developmental delay,(+) Macrocephaly,(+) Muscular hypotonia,(+) Strabismus,(+) Midface retrusion,(+) Hand apraxia,(+) Large fontanelles
DST	ENST00000370788		AD_denovo	8.5	het	de novo	2	NDD	(+) Moderate global developmental delay,(+) Muscular hypotonia,(+) Molar tooth sign on MRI,(+) Developmental cataract,(+) Febrile seizure (within the age range of 3 months to 6 years)
PHF14	ENST00000403050:c.541del p.Lys182Asnfs*19		AD_unknown	B	het	unknown	1	Immunology	(+) Autoimmunity,(+) Hepatitis,(+) Recurrent fractures,(+) Allergy,(+) Abnormality of the face,(+) Unerupted tooth,(+) Recurrent infections
URGCP	ENST00000453200:c.2192del p.Gln731Argfs*20		AD_unknown	4.7	het	unknown	1	NDD + Epilepsy	(+) Intellectual disability, severe,(+) Severe global developmental delay,(+) Bilateral tonic-clonic seizure,(+) Abnormality of movement,(+) Short stature,(+) Failure to thrive,(+) Ventriculomegaly,(+) Hypoplasia of the corpus callosum,(+) Cerebellar hypoplasia,(+) Strabismus,(+) Microcephaly
PLCG1	ENST00000373272:c.1687C>T p.His563Tyr		AD_unknown	4.45	het	unknown	1	NDD	(+) Joint hypermobility,(+) Pes cavus,(+) Poor gross motor coordination,(+) Delayed speech and language development,(+) Myopia,(+) Global developmental delay
SUDS3	ENST00000543473:c.557G>A p.Arg186Gln		AD_denovo	5.83		de novo	1	NDD + epilepsy	(+) Global developmental delay,(+) Motor delay,(+) Muscular hypotonia,(+) Frontal bossing,(+) Depressed nasal bridge,(+) Anteverted nares,(+) Hypertelorism,(+) Epicanthus,(+) Bifid uvula,(+) Sacral dimple,(+) Prominent fingertip pads,(+) Atopic dermatitis,(+) Hypermetropia,(+) Strabismus
KIF20B	ENST00000371728:c.2035_2037del p.Ile679del		AR_homo	6.25	homo	maternal&paternal	3	epilepsy	focal onset seizures
RXFP2	ENST00000298386:c.1594C>T p.Arg532*	ENST00000298386:c.1600G>A p.Gly534Arg	AR_comphet	4.49	comphet	maternal&paternal	3	epilepsy	focal onset seizures
LRRC3C	ENST00000377924:c.244C>T p.Arg82Cys	ENST00000377924:c.769C>T p.Arg257Trp	AR_comphet	3.06	comphet	maternal&paternal	3	epilepsy	focal onset seizures
MXRA5	ENST00000217939:c.4176G>C p.Gln1392His		XL	2.97	hemi	maternal	1	NDD + epilepsy	(+) Infantile spasms,(+) Focal impaired awareness seizure,(+) Delayed speech and language development,(+) Intellectual disability, severe,(+) Microcephaly,(+) Esophageal atresia,(+) Spasticity; known maternal inherited microdeletion in region 13q14
G2E3	ENST00000206595:c.419_420del p.Glu140Valfs*12		AD_unknown	5.1	het	unknown	1	NDD	(+) Global developmental delay,(+) Stereotypy,(+) Autism,(+) Poor speech,(+) Muscular hypotonia
AHNAK	ENST00000378024:c.15413C>T p.Ala5138Val	ENST00000378024:c.725C>T p.Ser242Leu	AR_comphet	4.1	comphet	maternal&paternal	2	NDD + epilepsy	(+) Mild global developmental delay,(+) Delayed speech and language development,(+) Behavioral abnormality,(+) Delayed social development,(+) Abnormal facial shape,(+) Focal-onset seizure
FRMPD3	ENST00000276185:c.1379G>C p.Ser460Thr		XL	3.3	hemi	maternal	2	NDD + epilepsy	(+) Mild global developmental delay,(+) Delayed speech and language development,(+) Behavioral abnormality,(+) Delayed social development,(+) Abnormal facial shape,(+) Focal-onset seizure
UBE4B	ENST00000343090:c.2754G>A p.Met918Ile		AD_unknown	4.4	het	unknown	1	epilepsy	(+) Focal atonic seizure
EVPL	ENST00000301607:c.505G>C p.Gly169Arg		AR_homo	3.9	homo	maternal&paternal	1	epilepsy	Focal seizures since the age of ten, genetically proved Ehlers Danlos syndrome
PPP4C	ENST00000279387:c.214G>A p.Val72Ile		AD_unknown	C	het	unknown	1	immunology	(+) Immunodeficiency
REPS2	ENST00000357277:c.1930C>T p.Arg644*		XL	6.9	hemi	maternal	4	NDD	Dystonia,(+) Intellectual disability,(+) Developmental regression
UBR4	ENST00000375254:c.5500A>G p.Ser1834Gly		AD_denovo	9.3	het	de novo	4	NDD	Dystonia,(+) Intellectual disability,(+) Developmental regression

CCDC155	ENST00000447857:c.1214del p.Ile405Thrfs*40		AR_homo	8.08	homo	maternal&paternal	4	NDD	Dystonia,(+) Intellectual disability,(+) Developmental regression
NR2E1	ENST00000368986:c.1154T>C p.Ile385Thr		AD_denovo	8.6	het	de novo	2	NDD	(+) Global developmental delay,(+) Intellectual disability, mild,(+) Sleep disturbance,(+) Short toe,(+) Periauricular sinus and cyst
RLF	ENST00000372771:c.5215_5216del p.Val1739Lysfs*12		AD_denovo	9.8	het	de novo	2	NDD	(+) Global developmental delay,(+) Intellectual disability, mild,(+) Sleep disturbance,(+) Short toe,(+) Periauricular sinus and cyst
SCRIB	ENST00000356994:c.4896A>T p.Glu1632Asp	ENST00000356994:c.2224A>G p.Ile742Val	AR_comphet	C	comphet	maternal&paternal	1	Wachstum, Skelett	(+) Scaphocephaly
SEC14L5	ENST00000251170:c.1368G>C p.Gln456His		AD_denovo	B	het	de novo	1	Wachstum, Skelett	(+) Craniosynostosis,(+) Trigonocephaly, no neurodevelopmental delay
SPTBN5	ENST00000320955:c.9170G>A p.Arg3057Gln		AR_homo	5.1	homo	maternal&paternal	5	Neuro	(-) Abnormality of brain morphology,(+) Lower limb spasticity
ZSCAN29	ENST00000396976:c.1298G>A p.Arg433Gln		AR_homo	3.8	homo	maternal&paternal	5	Neuro	(-) Abnormality of brain morphology,(+) Lower limb spasticity
SSFA2	ENST00000431877:c.1060_1062del p.Ser354del		AR_homo	4.4	homo	maternal&paternal	5	Neuro	(-) Abnormality of brain morphology,(+) Lower limb spasticity
ERBB2IP	ENST00000506030:c.472G>A p.Gly158Ser	ENST00000506030:c.2473C>T p.His825Tyr	AR_comphet	5.6	comphet	maternal&paternal	5	Neuro	(-) Abnormality of brain morphology,(+) Lower limb spasticity
CORO1C	ENST00000261401:c.318+1G>C None		AD_denovo	9.0	het	de novo	1	NDD + epilepsy	(+) Seizure,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Mild global developmental delay,(+) EEG abnormality
CDC42BPB	ENST00000361246:c.4610C>G p.Pro1537Arg	ENST00000361246:c.1439A>G p.Asn480Ser	AR_comphet	5.2	comphet	maternal&paternal	2	epilepsy	(+) Focal-onset seizure,(+) Febrile seizure (within the age range of 3 months to 6 years),(-) Global developmental delay
RGAG1	ENST00000465301:c.1193T>C p.Met398Thr		XL	2.7	hemi	maternal	2	epilepsy	(+) Focal-onset seizure,(+) Febrile seizure (within the age range of 3 months to 6 years),(-) Global developmental delay
PPP3CB	ENSP00000378306:c.2758T>C p.Cys920Arg		AD_unknown	C	het	unknown	1	Wachstum, Skelett	(+) Fused cervical vertebrae,(+) Sprengel anomaly,(+) Scoliosis,(+) Short stature,(+) Abnormal facial shape
BIRC6	ENST00000421745:c.9946T>G p.Phe3316Val		AD_unknown	4.0	het	unknown	1		(+) Hypopituitarism,(+) Optic atrophy,(+) Septo-optic dysplasia,(+) Cerebellar hypoplasia,(+) Hypoplasia of the corpus callosum,(+) Hypoglycemia,(+) Patellar hypoplasia
PTBP3	ENST00000458258:c.207del p.Arg70Glufs*15		AD_unknown	4.7	het	unknown	1	NDD	(+) Mild global developmental delay,(+) Short stature
FAT3	ENST00000298047:c.4430C>T p.Thr1477Met	ENST00000298047:c.10819G>A p.Ala3607Thr	AR_comphet	4.4	comphet	maternal&paternal	2	epilepsy	(+) Focal-onset seizure,(+) Generalized-onset seizure,(+) Migraine with aura,(+) Scoliosis,(+) Patent foramen ovale,(+) Mitral valve prolapse,(+) Mitral regurgitation
MFS9	ENST00000258436:c.391A>G p.Asn131Asp	1011840-2011844	AR_homo	4.3	homo	unknown	5	NDD	(+) Microcephaly,(+) Abnormality of the face,(+) Hypertelorism,(+) Global developmental delay,(+) Severe expressive language delay
KANK4	ENST00000317477:c.2587A>G p.Met863Val	ENST00000371153:c.1957C>T p.Arg653Cys	AR_comphet	3.0	comphet	maternal&paternal	2	epilepsy	(+) Focal-onset seizure,(+) Generalized-onset seizure,(+) Migraine with aura,(+) Scoliosis,(+) Patent foramen ovale,(+) Mitral valve prolapse,(+) Mitral regurgitation
PMEL	ENST00000449260:c.727C>T p.Gln243*		AR_homo	7.3	homo	unknown	3	NDD + epilepsy	(+) Autism,(+) Generalized tonic seizure,(+) Intellectual disability, severe,(+) Parietal cortical atrophy,(+) Occipital cortical atrophy,(+) Bilateral tonic-clonic seizure with generalized onset

ATOH8	ENST00000306279:c.124A>G p.Thr42Ala		AR_homo	3.2	homo	unknown	5	NDD	(+) Microcephaly,(+) Abnormality of the face,(+) Hypertelorism,(+) Global developmental delay,(+) Severe expressive language delay
ROCK1	ENST00000399799:c.4019A>T p.Gln1340Leu		AD_unknown	4.1	het	unknown	3	NDD + epilepsy	(+) Autism,(+) Generalized tonic seizure,(+) Intellectual disability, severe,(+) Parietal cortical atrophy,(+) Occipital cortical atrophy,(+) Bilateral tonic-clonic seizure with generalized onset
ABHD14B	ENST00000483233:c.536G>A p.Arg179Gln	ENST00000483233:c.250A>G p.Ile84Val	AR_comphet	3.1	comphet	maternal& paternal	3	NDD	(Global developmental delay),(+) Dysphagia,(+) Infantile muscular hypotonia
RC3H2	ENST00000373670:c.2386A>G p.Thr796Ala	ENST00000373670:c.1124A>G p.Glu375Gly	AR_comphet	3.6	comphet	maternal& paternal	3	NDD	(Global developmental delay),(+) Dysphagia,(+) Infantile muscular hypotonia
DGKD	ENST00000264057:c.1793C>T p.Ala598Val		AD_unknown	2.6	het	unknown	3	NDD + epilepsy	(+) Autism,(+) Generalized tonic seizure,(+) Intellectual disability, severe,(+) Parietal cortical atrophy,(+) Occipital cortical atrophy,(+) Bilateral tonic-clonic seizure with generalized onset
GABRE	ENST00000370328:c.572T>C p.Ile191Thr		XL	5.3	hemi	maternal	3	NDD	(Global developmental delay),(+) Dysphagia,(+) Infantile muscular hypotonia
RIMS4	ENST00000541604:c.240-3C>G None		AD_unknown	2.0	het	unknown	2	NDD	(+) Delayed speech and language development,(+) Motor delay,(+) Severe expressive language delay
RRN3	ENST00000198767:c.1267A>G p.Lys423Glu		AD_denovo	5.67	het	de novo	2	NDD	Inguinal hernia,(+) Retinal coloboma,(+) Microphthalmia,(+) Optic nerve hypoplasia,(+) Delayed speech and language development,(+) Pectus excavatum of inferior sternum,(+) Hypopigmented skin patches,(+) Hypotonia,(+) Intrauterine growth retardation,(+) Abnormal facial shape,(+) Congenital nystagmus,(+) Mild global developmental delay
CHD9	ENST00000566029:c.7279A>T p.Ile2427Phe		AD_unknown	3.9	het	unknown	2	NDD	(+) Delayed speech and language development,(+) Motor delay,(+) Severe expressive language delay
PDZD4	ENST00000164640:c.1782_1784delinsAGG p.Glu595Gly		XL	4.9	hemi	maternal	2	NDD	Inguinal hernia,(+) Retinal coloboma,(+) Microphthalmia,(+) Optic nerve hypoplasia,(+) Delayed speech and language development,(+) Pectus excavatum of inferior sternum,(+) Hypopigmented skin patches,(+) Hypotonia,(+) Intrauterine growth retardation,(+) Abnormal facial shape,(+) Congenital nystagmus,(+) Mild global developmental delay
PIAS2	ENST00000585916:c.376del p.Thr126Leufs*23		AD_unknown	5.55	het	unknown	1	NDD, Wachstum	(+) developmenal dealy (IQ 68) (+) puberty praecox (+) recurrent infections with fever
PRR32	ENST00000371125:c.751C>T p.Arg251*		AR_homo	3.5	homo	unknown	8	NDD + epilepsy	(+) Microcephaly,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Abnormal cardiac MRI
ZNF701	ENST00000540331:c.842del p.Phe281Serfs*16		AR_homo	4.0	homo	unknown	8	NDD + epilepsy	(+) Microcephaly,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Abnormal cardiac MRI
HMCN1	ENST00000271588:c.15935C>G p.Pro5312Arg		AR_homo	2.6	homo	unknown	8	NDD + epilepsy	(+) Microcephaly,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Abnormal cardiac MRI
FAM78A	ENST00000372271:c.496G>A p.Val166Ile		AR_homo	1.7	homo	unknown	8	NDD + epilepsy	(+) Microcephaly,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Abnormal cardiac MRI
DMBT1	ENST00000368909:c.4562G>A p.Arg1521Gln		AR_homo	1.4	homo	unknown	8	NDD + epilepsy	(+) Microcephaly,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Abnormal cardiac MRI
KCTD15	ENST00000430256:c.521C>T p.Thr174Met		AR_homo	3.0	homo	unknown	8	NDD + epilepsy	(+) Microcephaly,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Abnormal cardiac MRI
NECTIN2	ENST00000252483:c.1601G>A p.Arg534Gln		AR_homo	2.2	homo	unknown	8	NDD + epilepsy	(+) Microcephaly,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Abnormal cardiac MRI
UBC	ENST00000541272:c.277-14_502del		AR_homo	7.2	homo	unknown	8	NDD + epilepsy	(+) Microcephaly,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Abnormal cardiac MRI
LRP8	ENST00000306052:c.100G>T p.Ala34Ser		AD_unknown	5.0	het	unknown	1	NDD	(+) Autism,(+) Delayed speech and language development,(+) Hyperactivity,(+) Intellectual disability, borderline

IMPDH2	ENST00000326739:c.687_689del p.Lys229del		AD_denovo	7.1	het	de novo	1	Fehlbildungen	(-) Cleft palate,(+) Microcephaly,(+) Webbed neck,(+) Cutis laxa,(+) Dandy-Walker malformation,(+) Joint laxity,(+) Intrauterine growth retardation,(+) Small for gestational age,(+) Premature birth,(+) Ventricular septal defect,(+) Dextrocardia,(+) Hypoplasia of the corpus callosum,(+) Scoliosis,(+) Reduced subcutaneous adipose tissue,(+) Persistent patent ductus venosus,(+) Abnormality of the scalp hair,(+) Palpebral edema,(+) Long fingers
CAPZB	ENST00000375142:c.*11-2A>G None		AD_unknown	7.0	het	unknown	1	NDD + Hypotonie	(+) Abnormality of upper lip,(+) Epicanthus,(+) High forehead,(+) Single transverse palmar crease,(+) Hypotonia,(+) Motor delay,(+) Failure to thrive
SUPT5H	ENST00000599117:c.2377C>T p.Leu793Phe		AD_unknown	3.7	het	maternal	1	NDD + epilepsy	(+) Abnormality of the face,(+) Hypotonia,(+) Focal-onset seizure,(+) Intellectual disability, severe,(+) Muscular ventricular septal defect
DOCK4	ENST00000437633:c.593G>C p.Ser198Thr		AD_unknown	5.5	het	unknown	2	Epilepsy	(+) Typical absence seizure,(+) Bilateral tonic-clonic seizure with generalized onset
TLN1	ENST00000314888:c.580C>T p.Arg194Trp		AD_unknown	4.55	het	unknown	1	NDD + Epilepsy	(+) Dolichocephaly,(+) Intellectual disability,(+) Global developmental delay,(+) Sagittal craniosynostosis,(+) Bilateral superior vena cava
WDR24	ENST00000293883:c.2005G>T p.Asp669Tyr		AD_unknown	C	het	unknown	6	Neuro	(+) Spastic gait,(+) Babinski sign,(+) Impaired distal tactile sensation,(+) Abnormal pyramidal sign,(+) Multifocal hyperintensity of cerebral white matter on MRI
MYCBP2	ENST00000544440:c.7277A>G p.Gln2426Arg		AD_unknown	C	het	unknown	3	Neuro	(+) Spastic gait,(+) Babinski sign,(+) Impaired distal tactile sensation,(+) Abnormal pyramidal sign,(+) Multifocal hyperintensity of cerebral white matter on MRI
NAA35	ENST00000361671:c.686A>G p.Gln229Arg		AD_unknown	C	het	unknown	3	Neuro	(+) Spastic gait,(+) Babinski sign,(+) Impaired distal tactile sensation,(+) Abnormal pyramidal sign,(+) Multifocal hyperintensity of cerebral white matter on MRI
PLXNA4	ENST00000359827:c.1246T>A p.Ser416Thr		AD_unknown	4.3	het	unknown	2	Epilepsy	(+) EEG abnormality,(+) Focal motor seizure,(+) Eating-induced seizure,(+) Somatosensory-induced seizure,(+) Generalized-onset motor seizure
KIAA1239	ENST00000309447:c.280G>A p.Asp94Asn		AD_unknown	3.3	het	unknown	2	Epilepsy	(+) EEG abnormality,(+) Focal motor seizure,(+) Eating-induced seizure,(+) Somatosensory-induced seizure,(+) Generalized-onset motor seizure
RNF20	ENST00000389120:c.716T>C p.Leu239Pro		AD_inherited	5.5	het	paternal	2	NDD + epilepsy	(+) Seizure,(+) Abnormal facial shape,(+) Severe muscular hypotonia,(+) Severe global developmental delay
XPO1	ENST00000401558:c.431A>G p.Lys144Arg		AD_inherited	5.4	het	paternal	2	NDD + epilepsy	(+) Seizure,(+) Abnormal facial shape,(+) Severe muscular hypotonia,(+) Severe global developmental delay
PSPC1	ENST00000338910:c.92C>T p.Pro31Leu		AD_unknown	3.5	het	unknown	2	Epilepsy	(+) Myoclonic absence seizure
NFATC3	ENST00000346183:c.1774+1G>A None		AD_unknown	6.2	het	unknown	2	Epilepsy	(+) Myoclonic absence seizure
WNK2	ENST00000297954:c.3381del p.Lys1127Asnfs*23		AD_unknown	5.7	het	unknown	1	NDD	(+) Microcephaly,(+) Pectus excavatum,(+) Hypotonia,(+) Global developmental delay,(+) Pes planus,(+) Pes valgus
SRRM2	ENST00000301740:c.1585C>T p.Gln529*		AD_unknown	8.4	het	unknown	1	NDD	(-) Abnormality of the face,(+) Behavioral abnormality,(+) Short attention span,(+) Low frustration tolerance,(+) Delayed speech and language development,(+) Global developmental delay,(+) Obesity
SCAF11	ENST00000369367:c.1146del p.Lys382Asnfs*5		AD_unknown	6.2	het	unknown	3	NDD	(+) Abnormality of the face,(+) Ataxia,(+) Global developmental delay,(+) EEG abnormality,(+) Poor speech
UNC79	ENST00000256339:c.3515T>C p.Met1172Thr		AR_homo	7.3	homo	unknown	3	NDD	(+) Abnormality of the face,(+) Ataxia,(+) Global developmental delay,(+) EEG abnormality,(+) Poor speech
ATP13A1	ENST00000357324:c.2699C>T p.Pro900Leu		AD_unknown	4.1	het	unknown	3	NDD	(+) Abnormality of the face,(+) Ataxia,(+) Global developmental delay,(+) EEG abnormality,(+) Poor speech
PCSK5	ENST00000545128:c.1024G>A p.Gly342Arg		AD_unknown	4.8	het	unknown	2	NDD	(+) Hypotonia,(+) Global developmental delay,(+) Failure to thrive in infancy,(+) Slender build,(+) Intellectual disability, moderate,(+) Abnormality of dental morphology,(+) Mild intrauterine growth retardation,(+) Broad hallux,(+) Long toe,(+) Primary microcephaly,(+) Long fingers
TMEM132D	ENST00000422113:c.79+1G>A None		AD_unknown	5.8	het	unknown	2	NDD	(+) Hypotonia,(+) Global developmental delay,(+) Failure to thrive in infancy,(+) Slender build,(+) Intellectual disability, moderate,(+) Abnormality of dental morphology,(+) Mild intrauterine growth retardation,(+) Broad hallux,(+) Long toe,(+) Primary microcephaly,(+) Long fingers
HSPA4	ENST00000304858:c.792dup p.Arg265Thrfs*7		AD_unknown	8.7	het	unknown	2	NDD	(+) Tall stature,(+) Autism,(+) Autistic behavior,(+) Hypotonia,(+) Global developmental delay
UBR2	ENST000003728899: c.1532T>C p.Leu511Pro	ENST000003728899: c.5026G>A p.Val1676Ile	AR_comphet	5.6	comphet	maternal&paternal	2	NDD	(+) Hypertelorism,(+) Abnormal eyebrow morphology,(+) Triphalangeal thumb,(+) Intellectual disability,(+) Global developmental delay
NPIB5	ENST00000424340:c.1505C>T p.Pro502Leu		AD_denovo	4.3	het	de novo	2	NDD	(+) Hypertelorism,(+) Abnormal eyebrow morphology,(+) Triphalangeal thumb,(+) Intellectual disability,(+) Global developmental delay

KDM1A	ENST00000400181:c.1894C>T p.Arg632Cys		AD_denovo	9.65	het	de novo	1	NDD + epilepsy	(+) Seizure,(+) Hypsarrhythmia,(+) Moderate global developmental delay,(+) Epileptic encephalopathy
CUL2	ENST00000537177:c.1043dup p.Asn348Lysfs*21		AD_unknown	7.0	het	maternal	3	NDD	(+) Epicanthus,(+) Round face,(+) Short philtrum,(+) Short chin,(+) Large hands,(+) Prominent fingertip pads,(+) Hypotonia,(+) Specific learning disability,(+) Joint hypermobility,(+) Obesity,(+) Pes planus,(+) Impaired continence,(+) Lip hyperpigmentation
TRIM9	ENST00000298355:c.386C>T p.Pro129Leu		AD_unknown	5.0	het	unknown	3	NDD	(+) Epicanthus,(+) Round face,(+) Short philtrum,(+) Short chin,(+) Large hands,(+) Prominent fingertip pads,(+) Hypotonia,(+) Specific learning disability,(+) Joint hypermobility,(+) Obesity,(+) Pes planus,(+) Impaired continence,(+) Lip hyperpigmentation
PHF21A	ENST00000418153:c.882A>G p.Ile294Met		AD_unknown	6.4	het	unknown	3	NDD	(+) Epicanthus,(+) Round face,(+) Short philtrum,(+) Short chin,(+) Large hands,(+) Prominent fingertip pads,(+) Hypotonia,(+) Specific learning disability,(+) Joint hypermobility,(+) Obesity,(+) Pes planus,(+) Impaired continence,(+) Lip hyperpigmentation
SRRM2	ENST00000301740:c.6777_6778del p.Arg2260Asnfs*26		AD_denovo	10.7	het	de novo	1	NDD	(+) Seizure,(+) Status epilepticus,(+) Complex febrile seizure
SPTBN1	ENST00000356805:c.2047T>C p.Phe683Leu		AD_unknown	5.2	het	unknown	2	NDD	(+) Autism,(+) Hypotonia,(+) Global developmental delay,(+) Absent speech
XIRP2	ENST00000409195:c.3288G>A p.Trp1096*	ENST00000409195:c.6515T>C p.Val2172Ala	AR_comphet	5.4	comphet?	unknown	2	NDD	(+) Autism,(+) Hypotonia,(+) Global developmental delay,(+) Absent speech
STARD9	ENST00000290607:c.8609C>T p.Thr2870Ile		AD_denovo	5.4	het	de novo	1	Fehlbildungen	(+) Intrauterine growth retardation,(+) Abnormality of ductus venosus blood flow,(+) Abnormality of umbilical vein blood flow
RHOT1	ENST00000358365:c.517_538del p.Leu173Argfs*2		AD_unknown	7.43	het	unknown	1	NDD + epilepsy	(+) Delayed speech and language development,(+) Dystonia,(+) Migraine,(+) Hemiplegia,(+) Hemiplegia/hemiparesis
FLRT2	ENST00000330753:c.1102C>T p.Pro368Ser	ENST00000330753:c.1766G>A p.Cys589Tyr	AR_comphet	B	comphet	maternal&paternal	2	Muskel	(+) Generalized hypotonia,(+) Motor axonal neuropathy,(+) Progressive distal muscle weakness
KCND2	ENST00000331113:c.107A>G p.Lys36Arg		AD_unknown	5.76	het	unknown	1	NDD + epilepsy	(+) Hydrocephalus,(+) Intellectual disability,(+) Myoclonic seizure
ARCN1	ENST00000264028:c.134A>G p.Gln45Arg		AD_unknown	6.7	het	unknown	1	NDD	(+) Global developmental delay
BAZ1A	ENST00000360310:c.1252A>G p.Thr418Ala		AD_unknown	4.91	het	unknown	1	NDD	(+) Abnormality of the face,(+) Autism,(+) Seizure,(+) Mild global developmental delay
PDCL	ENST00000259467:c.203G>A p.Arg68His		AR_homo	5.57	homo	maternal&paternal	4	NDD + epilepsy	(+) Microcephaly,(+) Cerebellar hypoplasia,(+) Cerebral atrophy,(+) Hypoplasia of the corpus callosum,(+) Status epilepticus,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Delayed myelination
FAM78A	ENST00000372271:c.496G>A p.Val166Ile		AR_homo	4.16	homo	maternal&paternal	4	NDD + epilepsy	(+) Microcephaly,(+) Cerebellar hypoplasia,(+) Cerebral atrophy,(+) Hypoplasia of the corpus callosum,(+) Status epilepticus,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Delayed myelination
DUSP8	ENST00000397374:c.36T>G p.Asp12Glu		AR_homo	3.3	homo	maternal&paternal	4	NDD + epilepsy	(+) Microcephaly,(+) Cerebellar hypoplasia,(+) Cerebral atrophy,(+) Hypoplasia of the corpus callosum,(+) Status epilepticus,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Delayed myelination
GRIK1	ENST00000399907:c.10G>T p.Gly4Cys		AR_homo	7.77	homo	maternal&paternal	4	NDD + epilepsy	(+) Microcephaly,(+) Cerebellar hypoplasia,(+) Cerebral atrophy,(+) Hypoplasia of the corpus callosum,(+) Status epilepticus,(+) Generalized-onset seizure,(+) Moderate global developmental delay,(+) Delayed myelination
ADGRB2	ENST00000373658:c.1203C>A p.Cys401*		AD_unknown	5.89	het	unknown	1	NDD	(+) Behavioral abnormality,(+) Delayed speech and language development,(+) Severe global developmental delay
ITPR3	ENST00000374316:c.143C>G p.Pro48Arg	ENST00000447857:c.1214del p.Ile405Thrfs*40	AD_unknown	6.3	het	unknown	2	NDD	(+) Short attention span,(+) Dyslexia,(+) Abnormal social behavior,(+) Abnormal emotion/affect behavior
LPHN3	ENST00000514591:c.4292A>G p.His1431Arg		AD_unknown	5.5	het	unknown	2	NDD	(+) Short attention span,(+) Dyslexia,(+) Abnormal social behavior,(+) Abnormal emotion/affect behavior

BZRAP1	ENST00000343736:c.5540G>A p.Ser1847Asn	ENST00000343736:c.4348G>T p.Gly1450Cys	AR_comphet	5.1	comphet?	unknown	1	NDD + epilepsy	(+) Autism,(+) Delayed speech and language development,(+) Bilateral tonic-clonic seizure,(+) Mild global developmental delay
ARHGDIB	ENST00000228945:c.239C>T p.Pro80Leu		AR_homo	4.22	homo	maternal&paternal	5	Neuro	(-) Abnormality of brain morphology,(+) Lower limb spasticity
NAP1L1	ENST00000261182:c.1058_1059+1dup		AD_unknown	6.3	het	unknown	2	Epilepsy + ASD	(+) Tall stature,(+) Autistic behavior,(+) Short attention span,(+) Delayed speech and language development,(+) Generalized non-motor (absence) seizure,(+) Diminished ability to concentrate
HTR3E	ENST00000440596:c.1031T>C p.Leu344Pro		AD_denovo	4.4	het	de novo	3	NDD	(+) Intellectual disability,(+) Cortical dysplasia,(+) Focal-onset seizure
XIRP2	ENST00000409195:c.5646G>A p.Trp1882*	ENST00000409043:c.*1158G>A p.Gly810Glu	AR_comphet	5.1	comphet	maternal&paternal	3	NDD	(+) Intellectual disability,(+) Cortical dysplasia,(+) Focal-onset seizure
OGFR	ENST00000290291:c.398+7T>G None	ENST00000290291:c.1108G>A p.Gly370Arg	AR_comphet	2.3	comphet	maternal&paternal	3	NDD	(+) Intellectual disability,(+) Cortical dysplasia,(+) Focal-onset seizure
KCP	ENST00000476647:n.4653C>T None	ENST00000476647:n.1049+2T>G None	AR_comphet	B	comphet	maternal&paternal	3	Fehlbildungen	hypotrophes Neugeborenes (Gewicht 5P, Länge 1P, Kopf 50P, 1z), Plagiozephalus DD Brachyzephalus, präaxiale Polydaktylie Typ 1 mit biphalangaealem Daumen rechts, V.a. bikuspidale Aortenklappe, Harntransprotströrung I-II° rechts und I° links, Neugeborenen-Hörscreening auffällig, Rektumstenose (Stoma), V.a. VACTERL-Assoziation (4/7 Symptomen), Körpermaße zur Vorstellung: Gewicht 52P, Größe 23P, Kopfumfang 10P
KCNG2	ENST00000316249:c.11G>A p.Trp4*		AD_unknown	4.8	het	unknown	1	NDD	(+) Obsessive-compulsive behavior,(+) Global developmental delay,(+) Obesity,(+) Postural instability,(+) Sleep disturbance,(+) Highly arched eyebrow,(+) Polyphagia,(+) Poor fine motor coordination,(+) Dyslexia
CDC42BPG	ENST00000342711:c.1289G>A p.Ser430Asn		AD_denovo	4.7	het	de novo	1	Epilepsy	(+) Bilateral tonic-clonic seizure with focal onset,(+) Focal motor seizure,(+) Autonomic epileptic aura
TOP2B	ENST00000435706:c.3360A>T p.Gln1120His		AD_unknown	5.4	het	unknown	1	NDD	(+) Delayed speech and language development,(+) Episodic hemiplegia
PBRM1	ENST00000394830:c.233G>A p.Arg78Gln		AD_unknown	4.5	het	unknown	1	NDD	(+) Autism,(+) Intellectual disability,(+) Seizure,(+) Scoliosis,(+) Severe global developmental delay
HDAC1	ENST00000373548:c.1322A>G p.Lys441Arg		AD_unknown	5.4	het	unknown	1	NDD + epilepsy	(+) Intellectual disability,(+) Focal-onset seizure,(+) Myoclonic absence seizure,(+) Moderate global developmental delay,(+) Mild malformation of cortical development
HUWE1	ENST00000342160:c.12115C>T p.Pro4039Ser		XL	B	hemi	maternal	1	Fehlbildungen	(+) Renal insufficiency,(+) Aortic valve stenosis,(+) Respiratory insufficiency,(+) Hyperechogenic kidneys,(+) Elevated C-reactive protein level
PRKCB	ENST00000303531:c.1810G>C p.Asp604His		AD_unknown	5.0	het	unknown	1	NDD	(+) Microcephaly,(+) Short stature,(+) Moderate global developmental delay
SIPA1L1	ENST00000555818:c.5402T>C p.Ile1801Thr		AD_unknown	4.33	het	unknown	1	NDD + epilepsy	(+) Cleft palate,(+) Seizure,(+) Ataxia,(+) Spasticity,(+) Short stature,(+) Severe global developmental delay,(+) Cleft lip
ANKRD28	ENST00000399451:c.3065C>G p.Pro1022Arg		AD_denovo	4.9	het	de novo	4	NDD	(+) Wide mouth,(+) Coarse facial features,(+) Autism,(+) Intellectual disability,(+) Moderate global developmental delay
USP39	ENST00000323701:c.1498A>C p.Ile500Leu		AD_denovo	5.9	het	de novo	4	NDD	(+) Wide mouth,(+) Coarse facial features,(+) Autism,(+) Intellectual disability,(+) Moderate global developmental delay
CAPN8	ENST00000366872:c.34C>T p.Arg12Trp		AR_homo	3.6	homo	maternal&paternal	4	NDD	(+) Wide mouth,(+) Coarse facial features,(+) Autism,(+) Intellectual disability,(+) Moderate global developmental delay
SLC44A2	ENST00000335757:c.1060G>A p.Val354Met	ENST00000335757:c.1061T>C p.Val354Ala	AR_comphet	3.7	comphet	maternal&paternal	4	NDD	(+) Wide mouth,(+) Coarse facial features,(+) Autism,(+) Intellectual disability,(+) Moderate global developmental delay
NCKAP1	ENST00000360982:c.1138G>T p.Ala380Ser		AD_unknown	6.0	het	unknown	1	NDD + epilepsy	(+) Microcephaly,(+) Behavioral abnormality,(+) Seizure,(+) Moderate global developmental delay,(+) Dissociative reaction
PITPNM2	ENST00000320201:c.643+2T>C None		AD_unknown	5.94	het	unknown	1	NDD + epilepsy	(+) Coarse facial features,(+) Aggressive behavior,(+) Seizure,(+) Obesity,(+) Moderate global developmental delay

ASTN1	ENST00000361833:c.3622C>T p.Arg1208*		AD_denovo	A	het	de novo	1	Neuro	(+) Depression,(+) Headache,(+) Progressive neurologic deterioration,(+) Nonprogressive cerebellar ataxia,(+) Anti-Yo antibody
EP400	ENST00000389561:c.2665C>T p.Gln889*		AD_unknown	6.5	het	unknown	2	NDD	(+) Global developmental delay,(+) Agenesis of corpus callosum
ZBTB10	ENST00000430430:c.2203C>T p.Arg735*		AD_unknown	5.0	het	unknown	2	NDD	(+) Global developmental delay,(+) Agenesis of corpus callosum
UBR2	ENST00000372899:c.4319G>A p.Gly1440Glu		AD_denovo	7.4	het	de novo	1	NDD	(+) Abnormal lip morphology,(+) Thick lower lip vermilion,(+) Open mouth,(+) Coarse facial features,(+) Intellectual disability,(+) Global developmental delay,(+) Abnormal facial shape,(+) Thick vermilion border
ST3GAL2	ENST00000393640:c.420del p.Tyr141Thrfs*37		AD_unknown	5.6	het	unknown	1	NDD + epilepsy	(+) Seizure,(+) Neonatal hypoglycemia,(+) Generalized non-motor (absence) seizure,(-) EEG abnormality,(+) Proportionate short stature,(-) Abnormal cardiac MRI
CPSF3	ENST00000238112:c.1147C>A p.Pro383Thr		AD_unknown	C	het	unknown	1	immunology	(+) Episodic abdominal pain,(+) Periodic fever
ANXA11	ENST00000438331:c.1403A>G p.Asp468Gly		AD_denovo	4.86	het	de novo	2	NDD	(+) Behavioral abnormality,(+) Dementia,(+) Intellectual disability, mild,(+) Motor delay,(+) Neurological speech impairment,(+) Global brain atrophy,(+) Sleep disturbance,(+) Encephalitis,(+) Pica
MRPL42	ENST00000549982:c.143A>G p.Glu48Gly		AD_denovo	4.9	het	de novo	2	NDD	(+) Behavioral abnormality,(+) Dementia,(+) Intellectual disability, mild,(+) Motor delay,(+) Neurological speech impairment,(+) Global brain atrophy,(+) Sleep disturbance,(+) Encephalitis,(+) Pica
ARFGEF1	ENST00000262215:c.1028-2A>T None		AD_unknown	7.9	het	unknown	1	NDD + epilepsy	(+) Intellectual disability,(+) Focal-onset seizure
HSPA4	ENST00000304858:c.1450G>C p.Val484Leu		AR_homo	7.8	homo	unknown	3	NDD	(+) Intellectual disability,(+) Hypotonia,(+) Mild global developmental delay,(+) Abnormal ear morphology
GPR84	ENST00000551809:c.895del p.Gln299Serfs*19		AR_homo	8.4	homo	unknown	3	NDD	(+) Intellectual disability,(+) Hypotonia,(+) Mild global developmental delay,(+) Abnormal ear morphology
MYO1A	ENST00000442789:c.2827del p.Val943Cysfs*15		AR_homo	8.6	homo	unknown	3	NDD	(+) Intellectual disability,(+) Hypotonia,(+) Mild global developmental delay,(+) Abnormal ear morphology
TMEM131L	ENST00000409959:c.1226G>A p.Trp409*		AD_unknown	5.3	het	unknown	1	NDD	(+) Torticollis,(+) Nystagmus,(+) Behavioral abnormality,(+) Intellectual disability,(+) Global developmental delay,(+) Scoliosis,(+) Abducens palsy
AGAP2	ENST00000257897:c.52C>T p.Arg18*		AD_unknown	7.1	het	unknown	1	neuro	(+) Episodic ataxia
KCNG1	ENST00000371571:c.59C>T p.Ser20Leu		AD_unknown	3.46	het	unknown	1	NDD + epilepsy	(+) Epileptic encephalopathy
TLN2	ENST00000561311:c.4308_4309del p.Cys1436Trpfs*17		AD_unknown	6.3	het	unknown	2	NDD + epilepsy	(+) Focal-onset seizure,(+) EEG with focal epileptiform discharges,(+) EEG with generalized epileptiform discharges,(+) Mild global developmental delay
MCMBP	ENST00000360003:c.1110A>G p.Ile370Met		AD_denovo	4.7	het	de novo	1	NDD	(+) Trigonocephaly,(+) Hypertelorism,(+) Upslanted palpebral fissure,(+) Autism,(+) Delayed speech and language development,(+) Hypotonia,(+) Clinodactyly of the 5th finger,(+) Moderate global developmental delay,(+) Epicanthus palpebralis
SYMPK	ENST00000245934:c.226-7_226-2del None		AD_unknown	B	het	unknown	1	Muskel	(+) Motor delay,(+) Muscle weakness,(+) Lower limb muscle weakness,(+) Infantile muscular hypotonia
CHD1L	ENST00000369258:c.1086-2A>G None		AD_unknown	6.7	het	unknown	1	epilepsy	(+) Generalized non-motor (absence) seizure
DENR	ENST00000280557:c.426_429del p.Glu143Hisfs*15		AD_unknown	5.9	het	unknown	1	NDD + epilepsy	(+) Open mouth,(+) Abnormality of the face,(+) Hypomimic face,(+) Intellectual disability,(+) Spastic diplegia,(+) Aphasia,(+) Focal-onset seizure,(+) Severe global developmental delay,(+) Happy demeanor
PTBP1	ENST00000356948:c.8+2T>G		AD_unknown	8.3	het	inherited	1	Epilepsy	(+) Hydrocephalus,(+) Macrocephaly,(+) Headache,(+) Focal-onset seizure,(+) Episodic hemiplegia
PTPRN	ENST00000295718:c.1237A>G p.Thr413Ala		AD_denovo	5.8	het	de novo	1	NDD	(+) Epicanthus,(+) Depressed nasal ridge,(+) Upslanted palpebral fissure,(-) Intellectual disability,(+) Hypotonia,(+) Motor delay,(+) Expressive language delay,(+) Aplastic/hypoplastic toenail,(+) Oligodactyly,(+) Clinodactyly
WEE1	ENST00000450114:c.848G>A p.Arg283Lys		AD_unknown	4.0	het	unknown	1	NDD	(+) Low-set, posteriorly rotated ears,(+) Abnormality of skin pigmentation,(+) Specific learning disability,(+) Mutism,(+) Intellectual disability, borderline,(+) Mild global developmental delay
LRRC37A2	ENST00000576629:c.4967C>G p.Pro1656Arg		AD_denovo	4.2	het	de novo	2	NDD	(+) Autism,(+) Intellectual disability,(+) Global developmental delay
PLXND1	ENST00000324093:c.5657C>T p.Pro1886Leu	ENST00000324093:c.2668G>A p.Ala890Thr	AR_comphet	5.6	comphet	maternal&paternal	2	NDD	(+) Autism,(+) Intellectual disability,(+) Global developmental delay
ABLIM1	ENST00000277895:c.688G>A p.Gly230Arg		AD_denovo	6.9	het	de novo	2	Wachstum, Skelett	(+) Abnormal thumb morphology,(+) Preaxial hand polydactyly,(+) Vertebral segmentation defect,(+) Pilonidal sinus,(+) Muscular ventricular septal defect,(+) Perimembranous ventricular septal defect

MYO7B	ENST00000428314:c.2349C>G p.Phe783Leu	ENST00000428314:c.6250-1G>A None	AR_comphet	4.2	comphet	maternal&paternal	2	Wachstum, Skelett	(+) Abnormal thumb morphology,(+) Preaxial hand polydactyly,(+) Vertebral segmentation defect,(+) Pilonidal sinus,(+) Muscular ventricular septal defect,(+) Perimembranous ventricular septal defect
FASTKD3	ENST00000264669:c.1634C>T p.Thr545Ile		AD_denovo	5.4	het	de novo	2	NDD + epilepsy	(+) Hemangioma,(+) Seizure,(+) Global developmental delay,(+) Abnormal facial shape,(+) Spastic paraparesis,(+) Abnormality of brain morphology,(+) Cerebral palsy
TIMM8A	ENST00000372902:c.62A>G p.His21Arg		AD_denovo	7.27	het	de novo	2	NDD + epilepsy	(+) Hemangioma,(+) Seizure,(+) Global developmental delay,(+) Abnormal facial shape,(+) Spastic paraparesis,(+) Abnormality of brain morphology,(+) Cerebral palsy
ARPC4	ENST00000397256:c.331C>T p.Arg111Cys		AD_denovo	6.4	het	de novo	1	NDD	(+) Microcephaly,(+) Hypotonia,(+) Global developmental delay
GSG1L	ENST00000447459:c.184A>G p.Asn62Asp		AD_denovo	4.6	het	de novo	1	NDD + epilepsy	(+) Focal clonic seizure,(+) Dyslexia,(+) Mild global developmental delay,(+) Focal impaired awareness tonic seizure
DIP2C	ENST00000280886:c.2216C>T p.Ala739Val		AD_unknown	4.8	het	unknown	1	NDD	Moderate global developmental delay
BTBD18	ENST00000422652:c.1398del p.Tyr467Metfs*45		AD_unknown	B	het	unknown	1	other	Hypotonia,(+) Vocal cord paralysis,(+) Dyspnea
DHX8	ENST00000262415:c.1239A>T p.Lys413Asn		AD_denovo	B	het	de novo	1	Stoffwechsel	at the time of testing at 4 months of age: premature birth, (+) Inguinal hernia,(+) Jaundice,(+) Cholestasis,(+) Organic aciduria,(+) Hyperbilirubinemia,(+) Elevated circulating alanine aminotransferase concentration at age 2 years: good development
PLXNC1	ENST00000258526:c.3505A>C p.Asn1169His		AD_unknown	3.1	het	unknown	1	NDD	(+) Tall stature,(+) Polyuria,(+) Autism,(+) Hyperactivity,(+) Global developmental delay,(+) Obesity,(+) Polydipsia
HMX3	ENST00000357878:c.1031C>A p.Ser344*		AD_unknown	5.9	het	unknown	2	NDD	(+) Autism,(+) Delayed speech and language development,(+) Absent speech,(+) Sleep-wake cycle disturbance,(+) Toe walking
TAOK2	ENST00000308893:c.2811dup p.Cys938Leufs*56		AD_unknown	7.2	het	unknown	2	NDD	(+) Autism,(+) Delayed speech and language development,(+) Absent speech,(+) Sleep-wake cycle disturbance,(+) Toe walking
LRP8	ENST00000306052:c.497-1G>C None		AD_unknown	8.5	het	unknown	1	NDD	(+) Autism,(+) Delayed speech and language development,(+) Developmental regression,(+) Mild global developmental delay
STAM	ENST00000377524:c.265del p.Ser89Alafs*6		AD_unknown	7.7	het	unknown	1	epilepsy	(-) Intellectual disability,(+) Focal-onset seizure
RBBP7	ENST00000380084:c.89_99del p.His30Profs*15		AD_unknown	7.1	het	unknown	2	epilepsy	(+)atypical absence seizure
NAP1L2	ENST00000373517:c.700G>T p.Glu234*		AD_unknown	5.0	het	unknown	1	epilepsy	(+) focal myoclonic seizure (+) generalzied tonic-clonic seizure with focal onset
MAGEB5	ENST00000602297:c.770dup p.Tyr257*		AR_homo	4.0	homo	unknown	1	epilepsy	(+) Absence seizures
HDAC3	ENST00000305264:c.1076G>A p.Arg359His		AD_unknown	5.7	het	unknown	2	NDD	(+) Autistic behavior,(+) Moderate global developmental delay
OTOP1	ENST00000296358:c.803A>G p.Tyr268Cys		AR_homo	5.3	homo	unknown	2	NDD	(+) Autistic behavior,(+) Moderate global developmental delay
UNC13A	ENST00000519716:c.1597-4_1597-3delinsAA None		AD_denovo	7.1	het	de novo	1	NDD + epilepsy	(+) Hydrocephalus,(+) Decreased response to growth hormone stimulation test,(+) Seizure,(+) Cerebral hemorrhage,(+) Premature birth,(+) Intellectual disability, moderate,(+) Scoliosis,(+) Lymphoma,(+) Immunodeficiency,(+) Short stature,(+) Moderate global developmental delay
LAMTOR1	ENST00000278671:c.3G>T p.Met1?		AD_unknown	5.9	het	unknown	2	NDD	(+) Delayed puberty,(+) Obesity,(+) Moderate global developmental delay
SUSD4	ENST00000343846:c.26A>G p.Asn9Ser		AD_denovo	4.8	het	de novo	6	NDD	(+) Autism,(+) Intellectual disability,(+) Global developmental delay
CWC22	ENST00000410053:c.1633C>T p.Arg545*		AD_denovo	7.9	het	de novo	6	NDD	(+) Autism,(+) Intellectual disability,(+) Global developmental delay
PTPRN	ENST00000295718:c.2766C p.Ile922Met	ENST00000295718:c.2766C>G p.Ile922Met	AR_comphet	4.1	comphet	maternal&paternal	6	NDD	(+) Autism,(+) Intellectual disability,(+) Global developmental delay
KIAA0947	ENST00000296564:c.1718C>T p.Thr573Ile	ENST00000296564:c.6464A>G p.His2155Arg	AR_comphet	3.6	comphet	maternal&paternal	6	NDD	(+) Autism,(+) Intellectual disability,(+) Global developmental delay
RGS20	ENST00000276500:c.113C>A p.Pro38His	ENST00000276500:c.154G>A p.Gly52Arg	AR_comphet	2.9	comphet	maternal&paternal	6	NDD	(+) Autism,(+) Intellectual disability,(+) Global developmental delay

CASKIN1	ENST00000343516:c.1709T>C p.Ile570Thr	ENST00000343516:c.246C>T p.Gly82=	AR_comphet	5.5	comphet	maternal&paternal	6	NDD	(+) Autism,(+) Intellectual disability,(+) Global developmental delay
TENT4A	ENST00000230859:c.398C>G p.(Ser133Cys)		AR_homo	4.8	homo	unknown	7	NDD	(+) Delayed speech and language development,(+) Intellectual disability,(+) Global developmental delay,(+) Absent speech,(+) Asymmetric ventricles
RASSF10	ENST00000340901:c.899A>C p.(Glu300Ala)		AR_homo	3.4	homo	maternal&paternal	7	NDD	(+) Delayed speech and language development,(+) Intellectual disability,(+) Global developmental delay,(+) Absent speech,(+) Asymmetric ventricles
KLHL36	ENST00000564996:c.169G>C p.Val57Leu		AR_homo	4.2	homo	maternal&paternal	7	NDD	(+) Delayed speech and language development,(+) Intellectual disability,(+) Global developmental delay,(+) Absent speech,(+) Asymmetric ventricles
KIAA0100	ENST00000528896:c.5345G>A p.Gly1782Glu		AR_homo	5.4	homo	maternal&paternal	7	NDD	(+) Delayed speech and language development,(+) Intellectual disability,(+) Global developmental delay,(+) Absent speech,(+) Asymmetric ventricles
CPD	ENST00000225719:c.691G>A p.Ala231Thr		AR_homo	4.5	homo	maternal&paternal	7	NDD	(+) Delayed speech and language development,(+) Intellectual disability,(+) Global developmental delay,(+) Absent speech,(+) Asymmetric ventricles
SLFN13	ENST00000285013:c.2666C>A p.Ala889Glu		AR_homo	4.4	homo	maternal&paternal	7	NDD	(+) Delayed speech and language development,(+) Intellectual disability,(+) Global developmental delay,(+) Absent speech,(+) Asymmetric ventricles
MICALL2	ENST00000297508:c.1336G>A p.Asp446Asn	ENST00000297508:c.1987C>T p.Arg663Cys	AR_comphet	3.5	comphet	maternal&paternal	7	NDD	(+) Delayed speech and language development,(+) Intellectual disability,(+) Global developmental delay,(+) Absent speech,(+) Asymmetric ventricles
SF3A2	ENST00000221494:c.1354G>T p.Glu452*		AD_unknown	5.0	het	unknown	1	epilepsy	(+) Bilateral tonic-clonic seizure,(+) Generalized-onset seizure
LRP1B	ENST00000389484:c.7366G>A p.Val2456Ile		AR_homo	5.9	homo	maternal&paternal	1	NDD + epilepsy	(+) Hypermetropia,(+) Autism,(+) Intellectual disability,(+) Seizure
MAP4K4	ENST00000347699:c.123+2T>C None		AD_unknown	7.3	het	unknown	2	NDD	(+) Delayed speech and language development,(+) Global developmental delay,(+) Motor tics,(+) Phonic tics
TFDP2	ENST00000489671:c.44_47del p.Val15Glu fs*4		AD_unknown	5.2	het	unknown	2	NDD	(+) Delayed speech and language development,(+) Global developmental delay,(+) Motor tics,(+) Phonic tics
RBL2	ENST00000262133:c.3G>T p.Met1?		AD_unknown	7.3	het	unknown	1	NDD	(+) Strabismus,(+) Autistic behavior,(+) Hypotonia,(+) High myopia,(+) Mild global developmental delay
BAI3	ENST00000370598:c.1516C>T p.Arg506*		AD_unknown	6.0	het	unknown	1	NDD	(+) Intellectual disability,(+) Moderate global developmental delay, large ears, synophris, downslanted palprebal fissures
MINK1	ENST00000355280:c.3199C>T p.His1067Tyr		AD_unknown	4.2	het	unknown	1	NDD + epilepsy	(+) Psychosis,(+) Intellectual disability,(+) Focal tonic seizure,(+) Focal hyperkinetic seizure,(+) Focal cortical dysplasia
PDS5A	ENST00000303538:c.1231C>T p.Arg411Trp		AD_denovo	8.1	het	de novo	1	Wachstum, Skelett	(+) Retrognathia,(+) Epicanthus,(+) Protruding ear,(+) Hypotonia,(+) Short stature
GPHN	ENST00000478722:c.1332_1346del p.His445_Ser449del		AD_unknown	6.4	het	unknown	1	NDD + epilepsy	(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Mild global developmental delay,(+) Bilateral tonic-clonic seizure with generalized onset
PLXNB2	ENST00000449103:c.5455C>A p.Gln1819Lys		AD_unknown	5.1	het	unknown	1	NDD + epilepsy	(+) Microcephaly,(+) Abnormality of the face,(+) Behavioral abnormality,(+) Intellectual disability, mild
XPO7	ENST00000252512:c.1994G>A p.Arg665Gln		AD_unknown	3.6	het	unknown	1	NDD + epilepsy	(+) Intellectual disability,(+) Hemiplegia,(+) Elevated circulating creatine kinase concentration,(+) Severe global developmental delay,(+) Infantile spasms,(+) Eyelid laxity
ACTN1	ENST00000394419:c.1870C>T p.Arg624*		AD_unknown	6.6	het	unknown	2	NDD	(+) Delayed puberty,(+) Obesity,(+) Moderate global developmental delay
TCF7L2	ENST00000543371:c.407C>T p.Ala136Val		AD_denovo	8.5	het	de novo	1	NDD + epilepsy	(+) Hypotonia,(+) Motor delay,(+) Dystonia,(+) Generalized-onset seizure,(+) Severe global developmental delay
BTAF1	ENST00000265990:c.4437T>A p.Ser1479Arg		AD_unknown	4.7	het	unknown	1	NDD + epilepsy	(+) Intellectual disability, mild,(+) Bilateral tonic-clonic seizure,(+) Focal myoclonic seizure,(+) Mild global developmental delay
ZNF827	ENST00000379448:c.292C>T p.Gln98*		AD_unknown	5.6	het	unknown	1	NDD	(+) Microcephaly,(+) Global developmental delay,(+) Short stature
MRP63	ENST00000309594:c.-5-2A>G None		AR_homo	8.9	homo	unknown	2		(+) Generalized-onset seizure
SMG1	ENST00000446231:c.5213A>T p.Asp1738Val		AD_unknown	5.8	het	unknown	2		(+) Generalized-onset seizure
CLUH	ENST00000570628:c.1654A>T p.Lys552*		AD_unknown	B	het	unknown	2	Metabolism	(+) Tall stature,(+) Precocious puberty,(+) Obesity,(+) Hypertriglyceridemia,(+) Accelerated skeletal maturation
SEMA3F	ENST00000002829:c.1093G>A p.Val365Met		AD_denovo	7.6	het	de novo	4	Muskel	Motor delay, Muscular hypotonia, Skeletal muscle atrophy
ADCY9	ENST00000294016:c.2727C>G p.Tyr909*		AD_unknown	5.8	het	unknown	1	NDD + epilepsy	(+) Intellectual disability,(+) Seizure,(+) Dystonia,(+) Severe global developmental delay
RAB11FIP3	ENST00000262305:c.1116-2A>G None		AD_unknown	6.0	het	unknown	1	epilepsy	Generalized non-motor (absence) seizure

GPC1	ENST00000264039:c.1268+4G>A None		AD_denovo	5.2	het	de novo	3	NDD	Trigonocephaly, Epicanthus,Hypertelorism,Short chin, Retinal coloboma, Astigmatism, Hypermetropia, Iris coloboma, Motor delay, Patent foramen ovale, EEG abnormality, Depressed nasal bridge, Vertical nystagmus, Perimembranous ventricular septal defect, Anisometropia
SGK223	ENST00000520004:c.3247del p.Gln1083Argfs*52		AD_denovo	5.0	het	de novo	3	NDD	Trigonocephaly, Epicanthus,Hypertelorism,Short chin, Retinal coloboma, Astigmatism, Hypermetropia, Iris coloboma, Motor delay, Patent foramen ovale, EEG abnormality, Depressed nasal bridge, Vertical nystagmus, Perimembranous ventricular septal defect, Anisometropia
CHAF1A	ENST00000301280:c.829G>T p.Glu277*		AD_denovo	10.1	het	de novo	3	NDD	Trigonocephaly, Epicanthus,Hypertelorism,Short chin, Retinal coloboma, Astigmatism, Hypermetropia, Iris coloboma, Motor delay, Patent foramen ovale, EEG abnormality, Depressed nasal bridge, Vertical nystagmus, Perimembranous ventricular septal defect, Anisometropia
TSC22D4	ENST00000300181:c.1A>G p.Met1?		AD_unknown	5.1	het	unknown	1	NDD	(+) Abnormality of the face,(+) Intellectual disability,(+) Short stature,(+) Moderate global developmental delay,(+) Primary microcephaly
TUBA1B	ENST00000336023:c.362G>A p.Arg121Gln		AD_unknown	4.9	het	unknown	3	NDD + epilepsy	(+) Intellectual disability,(+) Generalized non-motor (absence) seizure,(+) Moderate global developmental delay
CHD9	ENST00000566029:c.4967G>C p.Ser1656Thr		AD_unknown	4.4	het	unknown	3	NDD + epilepsy	(+) Intellectual disability,(+) Generalized non-motor (absence) seizure,(+) Moderate global developmental delay
XKR3	ENST00000331428:c.614T>A p.Leu205*		AR_homo	7.0	het	unknown	3	NDD + epilepsy	(+) Intellectual disability,(+) Generalized non-motor (absence) seizure,(+) Moderate global developmental delay
CUL2	ENST00000537177:c.571G>C p.Val191Leu		AD_unknown	4.4	het	unknown	1	epilepsy	Focal-onset seizure
DAPK1	ENST00000408954:c.2980G>A p.Asp994Asn		AD_unknown	3.7	het	unknown	1	NDD	(+) Intellectual disability,(+) Obesity
TRA2B	ENST00000453386:c.151A>G p.Arg51Gly		AD_unknown	5.0	het	unknown	1	NDD	(+) Psychosis,(+) Intellectual disability, mild
TRA2B	ENST00000453386:c.151A>G p.Arg51Gly		AD_unknown	5.0	het	unknown	1	NDD	(+) Downslanted palpebral fissures,(+) Autism,(+) Global developmental delay
RALGPS1	ENST00000259351:c.1544C>A p.Pro515His		AD_unknown	3.2	het	unknown	2	NDD + epilepsy	(+) Aggressive behavior,(+) Focal clonic seizure,(+) Expressive language delay,(+) Focal tonic seizure,(+) Severe global developmental delay,(+) Focal atonic seizure,(+) Impulsivity
TRA2B	ENST00000453386:c.266_280del p.Asp90_Tyr94del		AD_denovo	6.9	het	de novo	1	NDD + epilepsy	(+) Microcephaly,(+) Delayed speech and language development,(+) Hypotonia,(+) Status epilepticus,(+) Generalized tonic seizure,(+) Atonic seizure
UBE2Q1	ENST00000292211:c.946C>G p.Leu316Val		AD_unknown	3.3	het	unknown	1	NDD + epilepsy	(+) Intellectual disability, mild,(+) Generalized myoclonic-atonic seizure
CHD9	ENST00000566029:c.7499_7501del p.Gly2500del		AD_unknown	3.5	het	unknown	4	NDD	(+) Hearing impairment,(+) Obesity,(+) Mild global developmental delay
FBXL19	ENST00000380310:c.431G>C p.Arg144Pro		AD_unknown	3.0	het	unknown	4	NDD	(+) Hearing impairment,(+) Obesity,(+) Mild global developmental delay
BRPF3	ENST00000357641:c.2228A>C p.Glu743Ala		AD_unknown	4.0	het	unknown	4	NDD	(+) Hearing impairment,(+) Obesity,(+) Mild global developmental delay
GMPPB	ENST00000321599:c.764_765delinsTT p.Thr255Ile		AD_unknown	4.3	het	unknown	4	NDD	(+) Hearing impairment,(+) Obesity,(+) Mild global developmental delay
TNRC6A	ENST00000395799:c.4677_4680del p.Trp1559Cysfs*30		AD_unknown	7.5	het	unknown	1	NDD	(+) Autism,(+) Impaired social interactions,(+) Obesity,(+) Moderate global developmental delay
SEC24A	ENST00000398844:c.1642A>G p.Thr548Ala		AD_denovo	5.9	het	de novo	2	NDD	microcephaly, congenital diaphragmatic hernia, pectus excavatum of inferior sternum, motor delay, failure to thrive in infancy, patent ductus arteriosus mild global developmental delay
CUL1	ENST00000325222:c.2137G>A p.Ala713Thr		AD_denovo	8.2	het	de novo	2	NDD	microcephaly, congenital diaphragmatic hernia, pectus excavatum of inferior sternum, mo-tor delay, failure to thrive in infancy, patent ductus arteriosus, mild global developmental delay
CLOCK	ENST00000309964:c.1599dup p.Thr534Aspfs*55		AD_unknown	7.9	het	unknown	1	NDD	(-) Microcephaly,(+) Delayed speech and language development,(-) Seizure,(+) Global developmental delay,(+) Motor delay,(+) Muscular hypotonia of the trunk
ZNF611	ENST00000543227:c.1319C>T p.Ser440Phe		AD_denovo	3.5	het	de novo	1	NDD	Aggressive behavior, Global developmental delay, Developmental regression, Self-injurious behavior
RAB11A	ENST00000261890:c.335A>G p.His112Arg		AD_denovo	9.8	het	de novo	1	NDD + epilepsy	(+) Coarse facial features,(+) Delayed speech and language development,(+) Intellectual disability, mild,(+) EEG abnormality,(+) Precocious puberty in females,(+) Delayed fine motor development,(+) Primary microcephaly
CT47B1	ENST00000371311:c.622C>T p.Pro208Ser		AD_denovo	4.2	het	de novo	1	NDD + epilepsy	osteopenia, intellectual disability, seizure, global developmental delay
SNW1	ENST00000261531:c.182_187del p.Gly61_Gly62del		AD_denovo	5.9	het	de novo	2	NDD + epilepsy	microcephaly, visual impairment, delayed speech and language development, anemia, bilateral tonic-clonic seizure, abnormal cortical gyration, hip dislocation, thoracolumbar scoliosis, focal-onset seizure, intellectual disability, severe, cerebral palsy
ZNF768	ENST00000380412:c.1511A>G p.His504Arg		AD_denovo	5.4	het	de novo	2	NDD + epilepsy	microcephaly, visual impairment, delayed speech and language development, anemia, bilateral tonic-clonic seizure, abnormal cortical gyration, hip dislocation, thoracolumbar scoliosis, focal-onset seizure, intellectual disability, severe, cerebral palsy

TNPO1	ENST00000337273:c.2438G>C p.Arg813Thr		AD_unknown	C	het	unknown	1	Wachstum, Skelett	(+) Renal duplication,(+) Cleft palate,(+) Abnormality of the ribs,(+) Glandular hypospadias,(+) Atopic dermatitis,(+) Premature birth,(+) Neutropenia,(+) Scoliosis,(+) Cleft lip
WDR13	ENST00000218056:c.194G>A p.Arg65His		XL	5.2	hemi	maternal	3	NDD	(+) Hearing impairment,(+) Abnormality of refraction,(+) Seizure,(+) Hypotonia,(+) Global developmental delay,(+) Motor delay,(+) Holoprosencephaly,(+) Failure to thrive,(+) Muscular dystrophy,(+) Abnormality of temperature regulation,(+) Secondary microcephaly,(+) Bilateral cryptorchidism
RBM10	ENST00000377604:c.308G>A p.Arg103Gln		XL	7.0	hemi	maternal	3	NDD	(+) Hearing impairment,(+) Abnormality of refraction,(+) Seizure,(+) Hypotonia,(+) Global developmental delay,(+) Motor delay,(+) Holoprosencephaly,(+) Failure to thrive,(+) Muscular dystrophy,(+) Abnormality of temperature regulation,(+) Secondary microcephaly,(+) Bilateral cryptorchidism
CCAR2	ENST00000308511:c.2627G>C p.Arg876Pro		AD_denovo	6.1	het	de novo	3	NDD	(+) Hearing impairment,(+) Abnormality of refraction,(+) Seizure,(+) Hypotonia,(+) Global developmental delay,(+) Motor delay,(+) Holoprosencephaly,(+) Failure to thrive,(+) Muscular dystrophy,(+) Abnormality of temperature regulation,(+) Secondary microcephaly,(+) Bilateral cryptorchidism
DBN1	ENST00000292385:c.1333_1334insGCCACGGAGATCC p.Ala445Glyfs*13		AD_unknown	7.9	het	unknown	1	NDD	(+) Obesity,(+) Intellectual disability, borderline
INTS6	ENST00000420668:c.498C>G p.Tyr166*		AD_denovo	9.9	het	de novo	1	NDD	(+) Global developmental delay,(+) Motor delay,(+) Agenesis of corpus callosum,(+) Morphological central nervous system abnormality,(+) Cerebellar dysplasia,(+) Muscular hypotonia of the trunk,(+) Schizencephaly,(+) Abnormal nervous system morphology,(+) Abnormal subarachnoid space morphology,(+) Interhemispheric cyst,(+) Paroxysmal tonic upgaze
TSPAN18	ENST00000340160:c.275T>C p.Leu92Pro		AR_homo	5.2	homo	unknown	2	NDD	(+) Behavioral abnormality,(+) Global developmental delay,(+) Intellectual disability, borderline
NOVA2	ENST00000263257:c.571A>G p.Lys191Glu		AD_unknown	4.8	het	unknown	2	NDD	(+) Behavioral abnormality,(+) Global developmental delay,(+) Intellectual disability, borderline
SLC17A7	ENST00000221485:c.170T>C p.Phe57Ser		AD_unknown	7.5	het	unknown	1	epilepsy + ataxia	(+) Generalized myoclonic seizure,(+) Episodic ataxia,(+) Generalized tonic seizure,(+) Generalized clonic seizure
NSD1	ENST00000347982:c.5468C>T p.Thr1823Met		AD_unknown	B	het	unknown	2	Obesity	(+) Tall stature,(+) Precocious puberty,(+) Obesity,(+) Hypertriglyceridemia,(+) Accelerated skeletal maturation
DUSP26	ENST00000256261:c.56G>T p.Arg19Leu		AD_denovo	6.1	het	de novo	2	epilepsy	epilepsy with focal and generalized components, microcephaly, MRI unremarkable, psychosomatic development unremarkable, seizure-free under Sultiam, EEG with rolando-focus and generalization tendency
B4GALNT4	ENST00000329962:c.2232C>G p.Asn744Lys		AD_denovo	5.3	het	de novo	2	epilepsy	epilepsy with focal and generalized components, microcephaly, MRI unremarkable, psychosomatic development unremarkable, seizure-free under Sultiam, EEG with rolando-focus and generalization tendency
ARMCX4	ENST00000423738:c.2150A>G p.Gln717Arg		XL	4.0	hemi	unknown	1	NDD + epilepsy	(+) Intellectual disability,(+) Seizure,(+) Global developmental delay
DENND1C	ENST00000381480:c.1241C>T p.Ala414Val		AD_denovo	B	het	de novo	1	Wachstum, Skelett	bei U3 auffällige Kopfform festgestellt, Sagittalnahtsynostose, keine neurologischen Auffälligkeiten
GPN1	ENST00000264718:c.982T>A p.Ser328Thr		AD_denovo	4.8	het	de novo	2	epilepsy	bilateral tonic-clonic seizure with generalized onset
TNKS2	ENST00000371627:c.1901A>G p.Asp634Gly		AD_denovo	7.1	het	de novo	2	epilepsy	bilateral tonic-clonic seizure with generalized onset
PITRM1	ENST00000224949:c.2263C>T p.(Arg755Trp)		AD_denovo	B	het	de novo	1	congenital heart defects	(+) Dilated cardiomyopathy,(+) Abnormal left ventricle morphology,(+) Primum atrial septal defect,(+) Multiple muscular ventricular septal defects
DPP6	ENST00000332007:c.1075A>C p.Lys359Gln		AD_denovo	9.4	het	de novo	1	epilepsy	(+) Abnormality of the pinna,(+) Generalized non-motor (absence) seizure,(+) Focal clonic seizure,(+) 2-3 toe syndactyly,(+) Focal tonic seizure
ZFP36	ENST00000248673:c.708del p.Gly237Alafs*129		AD_unknown	5.8	het	unknown	1	Epilepsy	(+) Strabismus,(-) Global developmental delay,(+) Generalized non-motor (absence) seizure,(+) Status epilepticus,(+) Focal-onset seizure,(+) EEG with focal spikes,(+) EEG with focal spike waves
ITGB1	ENST00000302278:c.1844G>A p.Cys615Tyr		AD_denovo	A	het	de novo	1	Leukodystrophy	(+) Gliosis,(+) Cerebral ischemia,(+) Cerebral vasculitis,(+) Perivascular spaces,(+) Arterial stenosis
ZFYVE9	ENST00000287727:c.3217C>T p.Arg1073Cys		AD_denovo	6.5	het	de novo	1	epilepsy	bilateral tonic-clonic seizure, myoclonic seizure, epileptic encephalopathy
PAXBP1	ENST00000290178:c.437C>A p.Ser146*		AD_inherited	5.9	het	paternal	1	Neuro	(+) Macrocephaly,(+) Seizure,(+) Global developmental delay,(+) Leukoencephalopathy
CCNL1	ENST00000295926:c.1134-2A>C None		AD_unknown	5.5	het	unknown	1	NDD	(+) Microcephaly,(+) Mild global developmental delay
RBBP9	ENST00000337227:c.136G>A p.Asp46Asn		AR_homo	4.4	homo	maternal& paternal	3	NDD + epilepsy	aggressive behavior, global developmental delay, bilateral tonic-clonic seizure, expressive language delay, atonic seizure, impulsivity
TERF1	ENST00000518874:c.319G>A p.Asp107Asn		AR_homo	6.4	homo	maternal& paternal	3	NDD + epilepsy	aggressive behavior, global developmental delay, bilateral tonic-clonic seizure, expressive language delay, atonic seizure, impulsivity
CILP2	ENST00000291495:c.2162T>A p.Ile721Asn		AR_homo	4.8	homo	maternal& paternal	4	NDD + epilepsy	aggressive behavior, global developmental delay, bilateral tonic-clonic seizure, expressive language delay, atonic seizure, impulsivity

PUS7L	ENST00000344862:c.749A>C p.Asn250Thr		AD_denovo	4.6	het	de novo	3	NDD + epilepsy	aggressive behavior, global developmental delay, bilateral tonic-clonic seizure, expressive language delay, atonic seizure, impulsivity
SH3RF3	ENST00000309415:c.221A>G p.Gln74Arg		AD_denovo	5.8	het	de novo	1	Epilepsy	seizure, two suspected episodes of seizures
LAMB2	ENST00000305544:None None		AD_unknown	C	het	unknown	2	Stoffwechs	obesity, insuline resistance, hyperuricemia
PTPN1	ENST00000371621:c.794A>G p.Asp265Gly		AD_unknown	B	het	unknown	2	Stoffwechs	obesity, insuline resistance, hyperuricemia
KALRN	ENST00000291478:c.1714_1715del p.Val572Serfs*14		AD_unknown	B	het	unknown	2	Stoffwechs el	(+) Hypertension,(+) Insulin resistance,(+) Striae distensae,(+) Slender finger,(+) Overgrowth,(+) Pes planus,(+) Asthma,(+) Hyperuricemia,(+) Hypertriglyceridemia,(+) Genu valgum,(+) Hyperglycemia,(+) Abnormal oral glucose tolerance,(+) Accelerated skeletal maturation,(+) Class II obesity
WHSC1L1	ENST00000316985:c.1603A>C p.Ile535Leu		AD_unknown	C	het	unknown	2	Stoffwechs el	(+) Hypertension,(+) Insulin resistance,(+) Striae distensae,(+) Slender finger,(+) Overgrowth,(+) Pes planus,(+) Asthma,(+) Hyperuricemia,(+) Hypertriglyceridemia,(+) Genu valgum,(+) Hyperglycemia,(+) Abnormal oral glucose tolerance,(+) Accelerated skeletal maturation,(+) Class II obesity
TM9SF4	ENST00000217315:c.1366C>T p.Arg456*		AD_unknown	5.0	het	unknown	2	NDD + epilepsy	(+) Narrow forehead,(+) Short neck,(+) Strabismus,(+) Aggressive behavior,(+) Intellectual disability,(+) Plagiocephaly,(+) Short stature,(+) Focal-onset seizure,(+) Short phalanx of finger,(+) Small hand
LEF1	ENST00000265165:c.695C>G p.Ser232*		AD_unknown	6.9	het	unknown	2	NDD + epilepsy	(+) Narrow forehead,(+) Short neck,(+) Strabismus,(+) Aggressive behavior,(+) Intellectual disability,(+) Plagiocephaly,(+) Short stature,(+) Focal-onset seizure,(+) Short phalanx of finger,(+) Small hand
ACLY	ENST00000352035:c.1587_1596del p.Met529Ilefs*18	ENST00000352035:c.616+4A>T None	AR_comphet	9.1	comphet	maternal& paternal	1	NDD	epicanthus, upslanted palpebral fissure, hypotelorism, hyperactivity, global developmental delay, absent speech, primary microcephaly
MYCBP2	ENST00000357337:c.7210G>A p.Val2404Ile		AD_unknown	5.5	het	unknown	1	NDD	(+) Tall stature,(+) Synophrys,(+) Autistic behavior,(+) Expressive language delay,(+) Increased body weight
FRY	ENST00000380250:c.3235G>A p.Glu1079Lys		AD_unknown	4.6	het	unknown	1	NDD	(+) Muscular hypotonia of the trunk,(+) Moderate global developmental delay
SH3BP4	ENST00000344528:c.119-2A>G None		AR_homo	8.6	homo	unknown	1	NDD	(+) Hypospadias,(+) Buphthalmos,(+) Developmental glaucoma,(+) Atrial septal defect,(+) Short stature,(+) Bilateral cryptorchidism,(+) Moderate global developmental delay
INPP5D	ENST00000359570:c.3440G>C p.Arg1147Pro		AD_unknown	4.3	het	unknown	1	NDD	(+) Brachycephaly,(+) Triangular face,(+) High forehead,(+) Low-set ears,(+) Congenital strabismus,(+) Downslanted palpebral fissures,(+) Hypermetropia,(+) Hypotelorism,(+) Sacral dimple,(+) Hypotonia,(+) Premature birth,(+) Frontal bossing,(+) Intestinal obstruction,(+) Depressed nasal bridge,(+) Moderate global developmental delay,(+) Midface retrusion
PROX1	ENST00000261454:c.1394A>C p.His465Pro		AD_unknown	5.5	het	unknown	3	NDD + epilepsy	(+) Autism,(+) Stereotypy,(+) Intellectual disability,(+) Global developmental delay,(+) Bilateral tonic-clonic seizure,(+) Hypoplasia of the corpus callosum,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Hyperventilation,(+) Thoracolumbar scoliosis,(+) Generalized tonic seizure,(+) Generalized atonic seizure,(+) Epileptic encephalopathy
U2SURP	ENST00000397933:c.842T>A p.Val281Asp		AD_unknown	5.2	het	unknown	3	NDD + epilepsy	(+) Autism,(+) Stereotypy,(+) Intellectual disability,(+) Global developmental delay,(+) Bilateral tonic-clonic seizure,(+) Hypoplasia of the corpus callosum,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Hyperventilation,(+) Thoracolumbar scoliosis,(+) Generalized tonic seizure,(+) Generalized atonic seizure,(+) Epileptic encephalopathy
UNC5A	ENST00000261961:c.995C>T p.Thr332Ile		AD_unknown	5.6	het	unknown	3	NDD + epilepsy	(+) Autism,(+) Stereotypy,(+) Intellectual disability,(+) Global developmental delay,(+) Bilateral tonic-clonic seizure,(+) Hypoplasia of the corpus callosum,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Hyperventilation,(+) Thoracolumbar scoliosis,(+) Generalized tonic seizure,(+) Generalized atonic seizure,(+) Epileptic encephalopathy
OTUD4	ENST00000296579:None None		AR_homo	4.3	homo	unknown	1	Neuro	(+) Oculomotor apraxia,(+) Spastic tetraparesis,(+) Dysphagia,(+) Cerebral atrophy,(+) Anarthria,(+) Peripheral neuropathy,(+) Peripheral demyelination,(+) Speech apraxia,(+) Cognitive impairment
PTAFR	ENST00000305392:c.736G>A p.Val246Met		AD_denovo	5.2	het	de novo	2	NDD	delayed speech and language development, intellectual disability, mild, EEG abnormality, poor fine motor coordination, decreased head circumference
COBL	ENST00000265136:c.735_737del p.Lys247del		AD_denovo	5.5	het	de novo	2	NDD	delayed speech and language development, intellectual disability, mild, EEG abnormality, poor fine motor coordination, decreased head circumference
UBR4	NM_020765.3(UBR4):c.13049T>C		AD_unknown	7.5	het	unknown	1	NDD	(+) Hydrocephalus,(+) Spasticity,(+) Focal-onset seizure,(+) Mild global developmental delay

CLEC18C	ENST00000314151:c.208C>T p.Arg70Trp		AD_denovo	4.7	het	de novo	1	NDD	delayed speech and language development (first words with 20 month, so far no simple sentences), motor delay (walking with over 18 month)
PLEKHA7	ENST00000355661:c.2203C>T p.Gln735*		AR_homo	8.5	homo	maternal&paternal	1	NDD	myopia, seizure (doubtful), intellectual disability (borderline, IQ 84), mild global developmental delay, hearing impairment
DPYSL2	ENST00000311151:c.1562C>T p.Thr521Met		AD_unknown	7.3	het	unknown	2	NDD	(+) Hypotonia,(+) Motor delay,(+) Elevated circulating creatine kinase concentration
CXXC1	ENST00000285106:c.171C>G p.Ile57Met		AD_unknown	6.0	het	unknown	2	NDD	(+) Hypotonia,(+) Motor delay,(+) Elevated circulating creatine kinase concentration
DNAH12	ENST00000351747:c.5656A>T p.Lys1886*		AD_denovo	5.4	het	de novo	1	Epilepsy	macrocephaly, behavioral abnormality, affect spasms with 14 month until third year of life, focal-onset seizure since the age of four
CNTRL	ENST00000373855:c.1187A>G p.Asn396Ser	ENST00000373855:c.3160G>C p.Gly1054Arg	AR_comphet	5.2	comphet?	unknown	1	NDD + epilepsy	(+) Intellectual disability, mild,(+) Global developmental delay,(+) Focal-onset seizure
ELOB	ENST00000262306:c.245-2_251del None		AD_denovo	6.6	het	de novo	1	NDD	(+) Intellectual disability,(+) Global developmental delay,(+) Headache,(+) Dyscalculia,(+) Dyslexia,(+) Abnormality of movement
CCZ1B	ENST00000316731:c.1106+1G>A None		AR_homo	8.1	homo	unknown	1	NDD	(+) Abnormality of the dentition,(+) Hypoplasia of the maxilla,(+) Abnormal cornea morphology,(+) Oligodontia,(+) Delayed speech and language development,(+) Ectodermal dysplasia,(+) Poor wound healing,(+) Absent distal phalanges,(+) Decreased corneal reflex
FAM71C	ENST00000329257:c.1272+6290C>G Non		AD_denovo	5.9	het	de novo	2	NDD	hypothyroidism, motor delay with hypotonia, congenital ptosis, removal phacomatous choriostoma right lower eyelid
NXPE4	ENST00000375478:c.437C>A p.Ala146Glu		AD_denovo	3.9	het	de novo	2	NDD	hypothyroidism, motor delay with hypotonia, congenital ptosis, removal phacomatous choriostoma right lower eyelid
NRXN2	ENST00000265459:c.3457C>T p.Pro1153Ser		AD_unknown	8.7	het	unknown	1	NDD + epilepsy	brachycephaly, microcephaly, epicanthus, hypertelorism, global developmental delay, absent speech, bilateral tonic-clonic seizure, hair-pulling, self-injurious behavior
SOCS7	ENST00000331159:c.1453C>T p.Gln485*		AD_unknown	5.8	het	unknown	1	NDD	(+) Delayed speech and language development,(+) Global developmental delay,(+) Hypoglycemia
RNPS1	ENST00000301730:c.128C>G p.Ser43*		AD_denovo	10.2	het	de novo	2	NDD	initial global developmental delay- now on the mend, intrauterine growth retardation (length -3.43 SD, weight -3.45 SD until birth), primary microcephaly (-4.2 SD), turricephaly, epicanthus, proptosis right side, temporary hyperinsulinemia, sacral dimple, umbilical hernia, broad thumb, wide nasal base, preaxial polydactyly
UBR4	ENST00000375254:c.12665G>A p.Ser422Asn	ENST00000375254:c.12379T>G p.Phe4127Val	AR_comphet	6.7	comphet	maternal&paternal	2	NDD	initial global developmental delay- now on the mend, intrauterine growth retardation (length -3.43 SD, weight -3.45 SD until birth), primary microcephaly (-4.2 SD), turricephaly, epicanthus, proptosis right side, temporary hyperinsulinemia, sacral dimple, umbilical hernia, broad thumb, wide nasal base, preaxial polydactyly
DENND1A	ENST00000373618:c.452_454del p.Asn151del		AD_denovo	4.3	het	de novo	3	Epilepsy	since several years suspected focal-onset seizure DD parasomnia, episodic visual impairment and vomiting, suspected migraine, since 2020 poor fine motor coordination, episodic ataxia, fatigue
DHX34	ENST00000328771:c.1715C>T p.Ala572Val	ENST00000328771:c.3190C>T p.Arg1064*	AR_comphet	5.4	comphet	maternal&paternal	3	Epilepsy	since several years suspected focal-onset seizure DD parasomnia, episodic visual impairment and vomiting, suspected migraine, since 2020 poor fine motor coordination, episodic ataxia, fatigue
CACNA2D1	ENST00000356860:c.2950G>A p.Asp984Asn	ENST00000356860:c.2804C>G p.Thr935Ser	AR_comphet	5.7	comphet	maternal&paternal	3	Epilepsy	since several years suspected focal-onset seizure DD parasomnia, episodic visual impairment and vomiting, suspected migraine, since 2020 poor fine motor coordination, episodic ataxia, fatigue
TP53BP1	seq[GRCh37] 15q15.2q15.3(43378488x2,43398090_43785291x1,43803137x2)		AD_unknown	8.5	het	unknown	1	Epilepsy	Bilateral tonic-clonic seizure with focal onset

PCDHGA12	ENST00000252085:c.211_218del p.Arg71Alafs*40	ENST00000252085:c.334G>A p.Asp112Asn	AR_comphet	4.5	comphet	maternal&paternal	1	NDD + epilepsy	precocious puberty, intellectual disability, seizure, Arnold-Chiari malformation, myelomeningocele
DPYSL3	ENST00000343218:c.571C>T p.Gln191*		AD_unknown	A	het	unknown	1	stroke, epilepsy	(+) Fasting hypoglycemia,(+) Ketotic hypoglycemia
ABHD3	ENST00000289119:c.293dup p.Ile99Hisfs*12		AD_denovo	5.6	het	de novo	2	NDD + epilepsy	delayed speech and language development, global developmental delay, motor delay, seizure-free since 03/2020, abnormal facial shape, ventriculomegaly, hypoplasia of the corpus callosum, feeding difficulties
TPR	ENST00000367478:c.6626G>A p.Arg2209Gln	ENST00000367478:c.3358G>A p.Ala1120Thr	AR_comphet	6.2	comphet	maternal&paternal	2	NDD + epilepsy	delayed speech and language development, global developmental delay, motor delay, seizure-free since 03/2020, abnormal facial shape, ventriculomegaly, hypoplasia of the corpus callosum, feeding difficulties
PDE4D	ENST00000340635:c.809-1G>C None		AD_unknown	A	het	unknown	1	Fehlbildungen	(+) Pulmonic stenosis,(+) Transposition of the great arteries,(+) Delayed gross motor development (very mild),(+) Perimembranous ventricular septal defect
BICRA	ENST00000396720:c.3390C>G p.Tyr1130*		AD_denovo	8.2	het	de novo	2	NDD	delayed speech and language development, global developmental delay, EEG abnormality, no seizures, periventricular leukomalacia of both lateral ventricles, stereotypy (turn of the head), decreased head circumference
PIK3C3	ENST00000262039:c.1916A>G p.Asp639Gly		AD_denovo	8.2	het	de novo	2	NDD	delayed speech and language development, global developmental delay, EEG abnormality, no seizures, periventricular leukomalacia of both lateral ventricles, stereotypy (turn of the head), decreased head circumference
NAA35	ENST00000361671:c.1702_1705del p.Lys568Phefs*4		AD_unknown	6.6	het	unknown	1	NDD	(+) Autism,(+) Intellectual disability, mild,(+) Disproportionate tall stature,(+) Scoliosis,(+) Skeletal muscle atrophy
YTHDC1	ENST00000344157:c.2171G>A p.Arg724Gln		AD_denovo	6.0	het	de novo	1	NDD + epilepsy	seizure, global developmental delay
TRAPPC1	ENST00000303731:c.293A>C p.His98Pro	ENST00000303731:c.215A>G p.His72Arg	AR_comphet	6.3	comphet	maternal&denovo	1	NDD + epilepsy	seizure since the age of 13 month, global developmental delay since the age of three month, progressive brain atrophy, secondary microcephaly
RSBN1L	ENST00000334955:c.250G>C p.Ala84Pro		AD_denovo	4.6	het	de novo	6	Epilepsy	seizure, suspected tuberous sclerosis, cortical tubers in MRI, ash-leaf spot
HIC1	ENST00000263073:c.545C>A, p.(Thr182Lys)		AD_denovo	6.9	het	de novo	6	Epilepsy	seizure, suspected tuberous sclerosis, cortical tubers in MRI, ash-leaf spot
EMILIN1	ENST00000260598:c.1370G>C, p.(Cys457Ser)		AR_homo	4.9	homo	maternal&paternal	6	Epilepsy	seizure, suspected tuberous sclerosis, cortical tubers in MRI, ash-leaf spot
CKAP5	ENST00000312055:c.2915C>G p.Thr972Ser		AR_homo	7.4	homo	maternal&paternal	6	Epilepsy	seizure, suspected tuberous sclerosis, cortical tubers in MRI, ashleaf spot
AHNAK	ENST00000378024:c.342+11553G>A p.Gly3656Asp	ENST00000378024:c.342+11132G>A p.Asp3516Asn	AR_comphet	4.0	comphet	maternal&paternal	6	Epilepsy	seizure, suspected tuberous sclerosis, cortical tubers in MRI, ash-leaf spot
ZNF106	ENST00000263805:c.1370G>C p.Cys457Ser	ENST00000263805:c.2776A>G p.Arg926Gly	AR_comphet	4.1	comphet	maternal&paternal	6	Epilepsy	seizure, suspected tuberous sclerosis, cortical tubers in MRI, ash-leaf spot
ZMYM4	ENST00000314607:c.1414T>G p.Phe472Val		AD_unknown	4.5	het	not maternal	1	NDD + Auge	(+) Retrognathia,(+) Astigmatism,(+) Hypermetropia,(+) Retinal dystrophy,(+) Optic atrophy,(+) Horizontal nystagmus,(+) Delayed speech and language development,(+) Global developmental delay,(+) Pes planus,(+) Supernumerary nipple,(+) Scapular winging,(+) Reduced visual acuity
ESPL1	ENST00000257934:c.4922+5G>A None		AD_unknown	A	het	unknown	1	Auge	(+) Strabismus,(+) Hypermetropia,(+) Amblyopia,(+) Depression,(+) Visual field defect,(+) Headache,(+) Borderline personality disorder,(+) Abnormal retinal nerve fiber layer morphology,(+) Abnormal eating behavior

SLC41A2	ENST00000258538:c.880+2T>C None		AD_unknown	6.4	het	unknown	1	NDD	Aarskog-Scott-Syndrom
KIAA1244	ENST00000251691:c.4984C>T p.Arg1662*		AD_unknown	6.5	het	unknown	1	NDD	(+) Hypospadias,(+) Single transverse palmar crease,(+) Moderate global developmental delay
CROCC	ENST00000375541:c.1992-3C>T None	ENST00000375541:c.3544C>T p.Arg1182Cys	AR_comphet	4.8	comphet	maternal&paternal	2	NDD + epilepsy	(+) Hypotonia,(+) Generalized-onset seizure,(+) Hypothalamic hamartoma,(+) Focal-onset seizure,(+) Moderate global developmental delay,(+) Abnormality of brain morphology
ZNF275	ENST00000370251:c.21_22del p.Leu9Phefs*30		XL	6.2	hemi	maternal	2	NDD + epilepsy	(+) Hypotonia,(+) Generalized-onset seizure,(+) Hypothalamic hamartoma,(+) Focal-onset seizure,(+) Moderate global developmental delay,(+) Abnormality of brain morphology
LMTK2	ENST00000297293:c.2792C>A p.Ser931*		AD_inherited	5.7	het	paternal	3	NDD	(+) Moderate global developmental delay
ASAP2	ENST00000281419:c.346-2A>G None		AD_inherited	6.8	het	maternal	3	NDD	(+) Moderate global developmental delay
SLC2A5	ENST00000377414:c.475C>T p.Arg159Trp		AR_homo	4.3	homo	maternal&paternal	3	NDD + epilepsy	global developmental delay, motor delay, absent speech, generalized-onset seizure, hypotonia alternating with increased muscle tone, high palate, trigonocephaly, epicanthus, ptosis, synophrys, frontal bossing, bifid tongue, wide nasal base, pulmonary artery stenosis, coronal craniosynostosis (cranioplastic 12/2018)
EXOSC10	ENST00000304457:c.191G>A p.Arg64Gln		AR_homo	6.3	homo	maternal&paternal	3	NDD + epilepsy	global developmental delay, motor delay, absent speech, generalized-onset seizure, hypotonia alternating with increased muscle tone, high palate, trigonocephaly, epicanthus, ptosis, synophrys, frontal bossing, bifid tongue, wide nasal base, pulmonary artery stenosis, coronal craniosynostosis (cranioplastic 12/2018)
TMEM66	ENST00000256255:c.890C>T p.Pro297Leu		AR_homo	5.0	homo	maternal&paternal	3	NDD + epilepsy	global developmental delay, motor delay, absent speech, generalized-onset seizure, hypotonia alternating with increased muscle tone, high palate, trigonocephaly, epicanthus, ptosis, synophrys, frontal bossing, bifid tongue, wide nasal base, pulmonary artery stenosis, coronal craniosynostosis (cranioplastic 12/2018)
PRDM2	ENST00000235372:c.4641del p.Ser1548Profs*16		AD_unknown	7.3	het	unknown	1	NDD + epilepsy	(+) Short attention span,(+) Hypotonia,(+) Generalized-onset seizure,(+) Focal-onset seizure,(+) Moderate global developmental delay,(+) Abnormal social behavior,(+) Abnormal emotion/affect behavior
TULP4	ENST00000367094:c.3439C>T, p.(Pro1147Ser)		AR_homo	6.0	homo	maternal&paternal	5	NDD + epilepsy	autistic behavior, intellectual disability, mild, global developmental delay, absent speech, failure to thrive, bilateral tonic-clonic seizure, expressive language delay,abnormality of the urinary system
FUT11	ENST00000339365:c.638A>G, p.(Tyr213Cys)		AR_homo	6.9	homo	maternal&paternal	5	NDD + epilepsy	autistic behavior, intellectual disability, mild, global developmental delay, absent speech, failure to thrive, bilateral tonic-clonic seizure, expressive language delay,abnormality of the urinary system
MAP4K2	ENST00000312049:c.286G>T, p.(Gly96Cys)		AR_homo	5.3	homo	maternal&paternal	5	NDD + epilepsy	autistic behavior, intellectual disability, mild, global developmental delay, absent speech, failure to thrive, bilateral tonic-clonic seizure, expressive language delay,abnormality of the urinary system
FOLR2	ENST00000298229:c.257T>C, p.(Met86Thr)		AR_homo	6.4	homo	maternal&paternal	5	NDD + epilepsy	autistic behavior, intellectual disability, mild, global developmental delay, absent speech, failure to thrive, bilateral tonic-clonic seizure, expressive language delay,abnormality of the urinary system
RTDR1	ENST00000216036:c.115G>A p.Asp39Asn		AR_homo	4.5	homo	maternal&paternal	5	NDD + epilepsy	autistic behavior, intellectual disability, mild, global developmental delay, absent speech, failure to thrive, bilateral tonic-clonic seizure, expressive language delay,abnormality of the urinary system
PRICKLE1	seq[GRCh37] 12q12(41463887x2,41464388_43527312x1,43747962x2)		AD_denovo	12.0	het	de novo	2	NDD + epilepsy	transient postnatal growth retardation, microcephaly in U5, percentiles currently back in normal range, language delay improving since tympanic tube, pectus excavatum, pulmonic stenosis, suspected atonic seizure (EEG 06/2020 unremarkable)
YAF2	seq[GRCh37] 12q12(41463887x2,41464388_43527312x1,43747962x2)		AD_denovo	7.6	het	de novo	2	NDD + epilepsy	transient postnatal growth retardation, microcephaly in U5, percentiles currently back in normal range, language delay improving since tympanic tube, pectus excavatum, pulmonic stenosis, suspected atonic seizure (EEG 06/2020 unremarkable)
CELF3	NM_007185.7:c.82G>A		AD_inherited	5.8	het	maternal	1	NDD	intellectual disability, behavioural abnormality, abnormality of the face
RC3H2	ENST00000335387:c.1A>G p.Met1?		AD_denovo	8.7	het	de novo	1	NDD	intellectual disability, developmental delay, generalized dystonia
B3GALT2	ENST00000367434:c.429del p.Glu144Lysfs*10		AD_unknown	7.1	het	unknown	1	NDD	(+) Brachycephaly,(+) Microcephaly,(+) Retrognathia,(+) Low-set ears,(+) Macrotia,(+) Motor delay,(+) Lacrimal duct stenosis,(+) Abnormal ossification of the pubic bone,(+) Severe hearing impairment,(+) Arachnoid cyst
SKIDA1	ENST00000444772:c.2427G>A p.Trp809*		AD_denovo	8.4	het	de novo	2	NDD + epilepsy	intellectual disability, seizure, MRI: heterotopia and abnormal cortical gyration
GPC5	ENST00000377067:c.647G>A p.Gly216Glu		AD_denovo	5.8	het	de novo	2	NDD + epilepsy	intellectual disability, seizure, MRI: heterotopia and abnormal cortical gyration

DGKZ	ENST00000318201:c.2274-2A>G None		AD_denovo	6.2	het	de novo	1	NDD + epilepsy	focal epilepsy of left hemisphere with complex focal seizures (suspected state after limbic encephalitis), development unremarkable until first status epilepticus at the age of five, de-velopmental stagnation at onset of seizures: delayed speech and language development, motor delay, intellectual disability (current cognitive abilities of a five to seven year old)
DUSP16	ENST00000228862:c.1290C>G p.Tyr430*		AD_denovo	4.3	het	de novo	2	NDD	global developmental delay, delayed speech and language development
ARHGAP39	ENST00000276826:c.472C>T p.Arg158Trp		AD_denovo	6.5	het	de novo	2	NDD	global developmental delay, delayed speech and language development
NAV3	ENST00000536525:c.1529G>A p.Ser510Asn	ENST00000536525:c.4158G>C p.Leu1386Phe	AR_comphet	4.1	comphet	maternal&paternal	1	NDD	moderate global developmental delay, intellectual disability, tall stature, coarse facial features, autism, hypotonia, obesity, high pitched voice
SLC4A3	ENST00000273063:c.218-7C>A None		AD_denovo	5.7	het	de novo	5	NDD	behavioral abnormality, autism, delayed speech and language development, intellectual disability, global developmental delay
TMTC2	ENST00000321196:c.211C>T p.Leu71Phe		AD_denovo	5.4	het	de novo	5	NDD	behavioral abnormality, autism, delayed speech and language development, intellectual disability, global developmental delay
NUAK1	ENST00000261402:c.1730T>G p.Val577Gly		AD_denovo	5.7	het	de novo	5	NDD	behavioral abnormality, autism, delayed speech and language development, intellectual disability, global developmental delay
KCTD3	ENST00000259154:c.1723A>G p.Met575Val	ENST00000259154:c.1732A>G p.Lys578Glu	AR_comphet	6.0	comphet	maternal&paternal	5	NDD	behavioral abnormality, autism, delayed speech and language development, intellectual disability, global developmental delay
FRY	ENST00000380250:c.2173C>A p.His725Asn	ENST00000380250:c.5509T>C p.Phe1837Leu	AR_comphet	5.1	comphet	maternal&paternal	5	NDD	behavioral abnormality, autism, delayed speech and language development, intellectual disability, global developmental delay
CYFIP2	ENST00000442283:c.543A>G p.Leu181=		AR_homo	8.1	homo	maternal&paternal	1	NDD + epilepsy	(+) Renal insufficiency,(+) Hypothyroidism,(+) Cerebellar atrophy,(+) Spastic tetraparesis,(+) Dehydration,(+) Aphasia,(+) Hypernatremia,(+) Type II diabetes mellitus,(+) Generalized clonic seizure,(+) Severe global developmental delay,(+) Aspiration pneumonia,(+) Refractory status epilepticus,(+) Psychogenic non-epileptic seizure
MTF2	ENST00000370298:c.712del p.Met238Cysfs*30		AD_denovo	9.8	het	de novo	1	Fehlbildung en	(+) Agenesis of corpus callosum,(+) Double outlet right ventricle with non-committed ventricular septal defect and pulmonary stenosis
LCLAT1	ENST00000309052:c.247C>T p.Arg83Cys	ENST00000309052:c.398G>A p.Trp133*	AR_comphet	4.3	comphet	maternal&paternal	2	NDD	moderate global developmental delay, large fontanel, macrocephaly, strabismus, hypoto-nia, midface retrusion, hand apraxia
ZC3H4	ENST00000253048:c.92C>G p.Pro31Arg		AD_denovo	6.7	het	de novo	1	NDD + epilepsy	intellectual disability, attention deficit hyperactivity disorder, seizure (partly precipitated by fever)
MTMR4	ENST00000323456:c.862+5G>A None		AD_unknown	6.7	het	unknown	1	NDD	(+) Microcephaly,(+) Global developmental delay,(+) Absent toenail,(+) Abnormal scalp morphology,(+) Aplasia/Hypoplasia of the 3rd finger
PDS5B	ENST00000315596:c.4308+6T>C None		AD_inherited	C	het	unknown	2	removing	(+) Trigonocephaly,(+) Craniosynostosis
SRRM2	ENST00000301740:c.5653C>T p.Arg1885*		AD_denovo	10.0	het	de novo	1	NDD + epilepsy	microcephaly, motor delay, orofacial hypotonia, failure to thrive, generalized-onset seizure
EP400	ENST00000330386:c.2681_2682del p.Asp894Valfs*11	ENST00000375254:c.10422_10425del p.Thr3475Argfs*4	AD_unknown	8.3	het	maternal	1	NDD + epilepsy	(+) Intellectual disability, mild,(+) Syncope

NEO1	ENST00000339362:c.244C>G p.Pro82Ala	ENST00000339362:c.2863A>G p.Thr955Ala	AR_comphet	4.9	comphet	maternal&paternal	3	NDD + epilepsy	(+) Strabismus,(+) Delayed speech and language development,(+) Global developmental delay,(+) Motor delay,(+) Gait ataxia,(+) Generalized-onset seizure,(+) Recurrent respiratory infections,(+) Secondary microcephaly,(+) Intracranial cystic lesion,(+) Complex febrile seizure,(+) Neonatal seizure,(+) Abnormality of movement
UBR4	ENST00000375254:c.12157C>T p.Leu4053Phe	ENST00000375254:c.10422_10425del p.Thr3475Argfs*4	AR_comphet	8.7	comphet	maternal&paternal	3	NDD + epilepsy	(+) Strabismus,(+) Delayed speech and language development,(+) Global developmental delay,(+) Motor delay,(+) Gait ataxia,(+) Generalized-onset seizure,(+) Recurrent respiratory infections,(+) Secondary microcephaly,(+) Intracranial cystic lesion,(+) Complex febrile seizure,(+) Neonatal seizure,(+) Abnormality of movement
EML6	ENST00000356458:c.298_299del p.Leu100Serfs*2		AD_denovo	6.4	het	de novo	1	Epilepsy	hydrocephalus, seizure, aqueductal stenosis, suspected Pallister-Hall-syndrome
ECHDC3	ENST00000379215:c.382del p.Cys128Valfs*5		AR_homo	8.0	homo	maternal&paternal	2	NDD	delayed speech and language development, retrognathia, astigmatism, hypermetropia, macular degeneration, pes planus, genu valgum, lumbar hyperlordosis, abnormality of macular pigmentation, large earlobe, nasal polyposis
PTPRT	ENST00000356100:c.1115C>T p.Pro372Leu		AR_homo	5.1	homo	maternal&paternal	2	NDD	delayed speech and language development, retrognathia, astigmatism, hypermetropia, macular degeneration, pes planus, genu valgum, lumbar hyperlordosis, abnormality of macular pigmentation, large earlobe, nasal polyposis
PAK7	ENST00000353224:c.133G>A p.Asp45Asn		AD_unknown	4.6	het	unknown	1	Epilepsy	(+) Myoclonic seizure,(+) Schizophrenia
CAD	ENST00000264705:c.3157C>T p.Arg1053Trp		AD_denovo	8.0	het	de novo	2	Fehlbildungen	(+) Microcephaly,(+) Downslanted palpebral fissures,(+) Ptosis,(+) Abnormal facial shape,(+) Proportionate short stature,(+) Infantile muscular hypotonia,(+) Clinodactyly
TAF4	ENST00000252996:c.637C>A p.Pro213Thr		AD_denovo	6.3	het	de novo	2	Fehlbildungen	(+) Microcephaly,(+) Downslanted palpebral fissures,(+) Ptosis,(+) Abnormal facial shape,(+) Proportionate short stature,(+) Infantile muscular hypotonia,(+) Clinodactyly
ADARB1	ENST00000348831:c.1299dup p.Phe434Valfs*2		AR_homo	11.3	homo	maternal&paternal	1	NDD + epilepsy	Hearing impairment,(+) Visual impairment,(+) Intellectual disability,(+) Hypotonia,(+) Global developmental delay,(+) Infantile muscular hypotonia,(+) Moderate global developmental delay,(+) Abnormal myelination,(+) Delayed myelination,(+) Epileptic encephalopathy
BRD4	ENST00000263377:c.2876C>T p.Pro959Leu	ENST00000263377:c.3810G>C p.Glu1270Asp	AR_comphet	6.5	comphet	maternal&paternal	1	NDD	microcephaly, intellectual disability, short stature, 01/2018 miscarriage in early pregnancy, 05/2019 miscarriage with growth retardation
TRPM2	ENST00000300481:c.2478+2T>C None		AD_unknown	6.5	het	unknown	2	NDD + epilepsy	severe global developmental delay, focal-onset seizure, microcephaly, optic atrophy, auto-ggressive behavior, ataxia, hypotonia, spasticity, scoliosis, hypokalemia, pontocerebellar atrophy, state after removal of bathing trunk naevus
RNF19A	ENST00000341084:c.1156A>G p.Met386Val		AD_unknown	4.7	het	unknown	2	NDD + epilepsy	severe global developmental delay, focal-onset seizure, microcephaly, optic atrophy, auto-ggressive behavior, ataxia, hypotonia, spasticity, scoliosis, hypokalemia, pontocerebellar atrophy, state after removal of bathing trunk naevus
ITGA9	ENST00000264741:c.2561A>C p.Asn854Thr		AR_homo	5.5	homo	maternal&paternal	1	NDD	aggressive behavior, anxiety, hypertension, intellectual disability, global developmental delay, obesity, sleep disturbance, impulsivity, gliosis (most likely unrelated to symptoms)
SYTL2	ENST00000316356:c.2180G>A p.Arg727His		AR_homo	4.1	homo	maternal&paternal	1	NDD	aggressive behavior, anxiety, hypertension, intellectual disability, global developmental delay, obesity, sleep disturbance, impulsivity, gliosis (most likely unrelated to symptoms)
CNTNAP5	ENST00000431078:c.383C>A p.Thr128Asn	ENST00000431078:c.2218G>C p.Asp740His	AR_comphet	5.8	comphet	maternal&paternal	1	NDD	aggressive behavior, anxiety, hypertension, intellectual disability, global developmental delay, obesity, sleep disturbance, impulsivity, gliosis (most likely unrelated to symptoms)

UBR4	ENST00000375254:c.7629+7G>T None	ENST00000375254:c.1918A>C p.Ser640Arg	AR_comphet	6.0	comphet	maternal&paternal	1	NDD	aggressive behavior, anxiety, hypertension, intellectual disability, global developmental delay, obesity, sleep disturbance, impulsivity, gliosis (most likely unrelated to symptoms)
UBAP2L	ENST00000271877:c.3003+1G>A None		AD_unknown	6.0	het	unknown	1	Neuro	(+) Sensory neuropathy,(+) Chronic pain,(+) Dyspepsia
FRY	ENST00000380250:c.6463C>G p.His2155Asp	ENST00000380250:c.7177A>G p.Thr2393Ala	AR_comphet	5.8	comphet	maternal&paternal	1	NDD	expressive language delay, poor fine motor coordination, attention deficit hyperactivity disorder,downslanted palpebral fissures
PTPRD	ENST00000356435:c.2368C>T p.Leu790Phe		AD_denovo	8.8	het	de novo	2	Epilepsy	generalized absence seizure, diminished ability to concentrate, tall stature, autistic behavi-or
ACLY	ENST00000352035:c.949_950insTATG p.Tyr317Leufs*2		AD_unknown	7.1	het	unknown	1	NDD + epilepsy	(+) Microcephaly,(+) Autism,(+) Hypotonia,(+) Generalized-onset seizure,(+) Short stature,(+) Intellectual disability, severe,(+) Severe global developmental delay
USP19	ENST00000398888:c.3304G>A p.Val1102Ile		AD_denovo	5.4	het	de novo	3	NDD	hearing impairment, delayed speech and language development, global developmental delay, plagiocephaly, hyperphenylalaninemia
TULP3	ENST00000397132:None None		AR_homo	6.5	homo	maternal&paternal	3	NDD	hearing impairment, delayed speech and language development, global developmental delay, plagiocephaly, hyperphenylalaninemia
GRIK4	ENST00000438375:c.1060-7T>C None		AR_homo	6.3	homo	maternal&paternal	3	NDD	hearing impairment, delayed speech and language development, global developmental delay, plagiocephaly, hyperphenylalaninemia
DENND4A	ENST00000431932:c.1981C>T p.Arg661*		AD_unknown	6.0	het	unknown	1	Epilepsy	(+) Anxiety,(+) Bilateral tonic-clonic seizure,(+) Generalized non-motor (absence) seizure
SGIP1	ENST00000237247:c.74+1G>A None		AD_unknown	8.1	het	unknown	1	Phenylketonuria	(+) Macrocephaly,(+) Cystic hygroma,(+) Glutaric aciduria
PAK6	ENST00000260404:c.545C>T p.Pro182Leu		AR_homo	6.1	homo	maternal&paternal	1	NDD + epilepsy	severe global developmental delay, microcephaly, hypotonia, plagiocephaly, dysphagia, cerebral atrophy, lower limb spasticity, bilateral tonic-clonic seizure, abnormal cerebral ventricle morphology, status epilepticus, abnormal visual fixation
TAF1C	ENST00000567759:c.1570C>T p.Gln524*	ENST00000567759:c.313C>T p.Arg105Cys	AR_comphet	6.4	comphet	maternal&paternal	1	NDD + epilepsy	West-Syndrome, global developmental delay
PSMB5	ENST00000361611:c.733C>T p.Arg245*		AD_unknown	7.2	het	unknown	1	NDD	(+) Delayed speech and language development,(+) Intellectual disability, borderline
PDS5A	ENST00000303538:c.3244G>A p.Ala1082Thr		AD_unknown	6.2	het	unknown	1	NDD + epilepsy	Severe intellectual disability, generalized-onset seizure, cataract
ZNF561	ENST00000302851:c.634C>G p.Leu212Val		AD_denovo	3.9	het	de novo	1	NDD	(+) Ataxia,(+) Syncope,(+) Ventricular septal defect,(+) Bicuspid aortic valve,(+) Hypoglycemia,(+) Hypoplastic left heart,(+) Abnormal ventricular septum morphology,(+) Mild global developmental delay,(+) Mitral atresia
DLGAP3	ENST00000235180:c.2469T>A p.Tyr823*		AD_unknown	7.9	het	de novo	1	NDD	(+) Microcephaly,(+) Abnormality of the face,(+) Hypotonia,(+) Global developmental delay
GABRG3	ENST00000333743:c.1299dup p.Ile434Tyrfs*24		AD_unknown	7.8	het	unknown	1	NDD	(+) Microcephaly,(+) Autism,(+) Moderate global developmental delay
DLGAP2			AD_denovo		het	de novo	1	NDD + epilepsy	
JAKMIP1	ENST00000282924:c.457C>T p.Gln153*		AD_inherited	8.9	het	paternal	1	Epilepsy	(+) Failure to thrive,(+) Generalized non-motor (absence) seizure,(+) Leukoencephalopathy
SCP2	ENST00000371514:c.693+1G>T None	ENST00000371514:c.1483C>T p.Gln495*	AR_comphet	9.9	comphet	maternal&paternal	1	Epilepsy	(+) Generalized-onset seizure,(+) Attention deficit hyperactivity disorder,(+) Focal-onset seizure,(+) Receptive language delay
TBC1D9B	ENST00000356834:c.3047C>T p.Thr1016Met	ENST00000356834:c.2910_2911del p.Gly971Argfs*29	AR_comphet	5.6	comphet	maternal&paternal	1	NDD + epilepsy	intellectual disability, mild, bilateral tonic-clonic seizure, myoclonic seizure, behavioral ab-normality, delayed speech and language development, ataxia, hypotonia, abnormality of joint mobility

UPF1	ENST00000262803:c.2248G>A p.Val750Met		AD_denovo	8.2	het	de novo	4	NDD + epilepsy	profound global developmental delay, focal-onset seizure, progressive microcephaly, hypo-tonia, tetraparesis, hypoplasia of the corpus callosum, ventriculomegaly, CNS hypomyelination, blindness, strabismus, sensorineural hearing impairment
DECR1	ENST00000220764:c.760C>A p.Pro254Thr		AR_homo	7.5	homo	maternal&paternal	4	NDD + epilepsy	profound global developmental delay, focal-onset seizure, progressive microcephaly, hypo-tonia, tetraparesis, hypoplasia of the corpus callosum, ventriculomegaly, CNS hypomyelination, blindness, strabismus, sensorineural hearing impairment
SPHKAP	ENST00000344657:c.4885G>A p.Val1629Met		AR_homo	5.2	homo	maternal&paternal	4	NDD + epilepsy	profound global developmental delay, focal-onset seizure, progressive microcephaly, hypo-tonia, tetraparesis, hypoplasia of the corpus callosum, ventriculomegaly, CNS hypomyelination, blindness, strabismus, sensorineural hearing impairment
NCKIPSD	ENST00000294129:c.1897C>T p.His633Tyr	ENST00000294129:c.734C>A p.Pro245His	AR_comphet	5.3	comphet	maternal&paternal	4	NDD + epilepsy	profound global developmental delay, focal-onset seizure, progressive microcephaly, hypo-tonia, tetraparesis, hypoplasia of the corpus callosum, ventriculomegaly, CNS hypomyelination, blindness, strabismus, sensorineural hearing impairment
USP11	ENST00000218348:c.1953G>C p.Glu651Asp		XL	5.0	hemi	maternal	1	NDD	global developmental delay, behavioral abnormality, EEG abnormality, cortical dysplasia, coarctation of aorta
FRY	ENST00000380250:c.5145G>C p.Glu1715Asp		AD_unknown	5.4	het	unknown	1	NDD	global developmental delay, absent speech, autistic behavior, intellectual disability
FOXO3	ENST00000343882:c.478del p.Leu160Cysfs*6		AD_denovo	10.1	het	de novo	2	NDD	(+) Autistic behavior,(+) Delayed speech and language development,(+) Global developmental delay,(+) Premature birth
NRXN2	ENST00000265459:c.4907C>G p.Thr1636Arg		AD_denovo	10.5	het	de novo	2	NDD	(+) Autistic behavior,(+) Delayed speech and language development,(+) Global developmental delay,(+) Premature birth
IPO13	ENST00000372339:c.174_177del p.Phe58Leufs*11		AD_unknown	7.6	het	unknown	2	NDD	Delayed speech and language development,(+) Obesity,(+) Moderate global developmental delay
GPM6A	ENST00000280187:c.387+2T>A None		AD_unknown	8.3	het	unknown	2	NDD	Delayed speech and language development,(+) Obesity,(+) Moderate global developmental delay
DQX1	ENST00000377526:c.769C>T p.(Arg257Trp)		AR_homo	4.6	homo	maternal&paternal	2	NDD	severe global developmental delay, microcephaly, joint contractures
UNC50	ENST00000328709:c.338C>G p.(Thr113Ser)		AR_homo	6.8	homo	maternal&paternal	2	NDD	severe global developmental delay, microcephaly, joint contractures
SPAG9	ENST00000262013:c.691C>T p.Gln231*	ENST00000262013:c.2146del p.Val716Leufs*19	AR_comphet	A	comphet	maternal&denovo	1	Auge	(+) Brachycephaly,(+) Microcephaly,(+) Hypertelorism,(+) Facial asymmetry,(+) Developmental cataract,(-) Motor delay
ARMC8	ENST00000358441:c.1024G>T p.Glu342*		AD_denovo	8.8	het	de novo	2	NDD	(+) Moderate global developmental delay
OMA1	ENST00000358603:c.185del p.Ser62Ilefs*48		AR_homo	9.3	homo	unknown	3	NDD + epilepsy	(-) Abnormality of the face,(+) Focal-onset seizure,(+) Moderate global developmental delay
AMER2	ENST00000357816:c.322G>T p.Gly108*		AR_homo	8.3	homo	unknown	3	NDD + epilepsy	(-) Abnormality of the face,(+) Focal-onset seizure,(+) Moderate global developmental delay
LAMP1	ENST00000332556:c.331dup p.Tyr111Leufs*11		AD_unknown	7.8	het	unknown	1	NDD + epilepsy	(+) Global developmental delay,(+) Tremor,(+) Generalized-onset seizure
YTHDC2	ENST00000161863:c.4172G>A p.Trp1391*		AD_unknown	7.1	het	unknown	1	NDD + epilepsy	(+) Epileptic encephalopathy
SHANK1	ENST00000293441:c.5387G>A p.Gly1796Glu	ENST00000293441:c.42C>T p.His14=	AR_comphet	6.8	comphet	maternal&paternal	1	NDD	Microcephaly,(+) Expressive language delay
TMEM194A	ENST00000300128:c.718C>T p.Gln240*		AD_unknown	6.2	het	unknown	1		(+) Global developmental delay,(+) Typical absence seizure
REV3L	ENST00000358835:c.8818C>T p.Arg2940Cys	ENST00000358835:c.5441C>T p.Ala1814Val	AR_comphet	6.0	comphet	maternal&paternal	2	NDD	Developmental delay, club foot, short stature, microcephaly, deafness
INSM2	ENST00000307169:c.282G>A p.Trp94*		AD_unknown	5.3	het	unknown	1	Epilepsy	Seizures

TRAP1	ENST00000246957:c.356A>G p.Asn119Ser	ENST00000246957:c.1804C>T p.Arg602*	AR_comphet	5.5	comphet	maternal&paternal	2	NDD	global developmental delay, expressive language delay, abnormality of metabolism/homeostasis, lower limb spasticity, tip-toe gait, hypotonia
NCAPH2	ENST00000299821:c.1628C>T p.Ala543Val	ENST00000299821:c.1769G>A p.Arg590Gln	AR_comphet	5.9	comphet	maternal&paternal	2	NDD	global developmental delay, expressive language delay, abnormality of metabolism/homeostasis, lower limb spasticity, tip-toe gait, hypotonia
ALG1L2	ENST00000425059:n.517G>A None		AD_denovo	3.3	het	de novo	1	NDD	(+) Microcephaly,(+) Intellectual disability,(+) Global developmental delay
GMDS	ENST00000380815:c.103-1G>A None		AD_unknown	7.4	het	unknown	1	NDD	(+) Microcephaly,(+) Multiple cafe-au-lait spots,(+) Mild global developmental delay
ERICH3	ENST00000326665:c.1732A>C p.Lys578Gln		AR_homo	5.1	homo	maternal&paternal	5	NDD	Delayed speech and language development,(+) Intellectual disability,(+) Motor delay,(+) Abnormal facial shape,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Cognitive impairment
ATAD2	ENST00000287394:c.3646G>A p.Gly1216Arg		AR_homo	5.3	homo	maternal&paternal	5	NDD	Delayed speech and language development,(+) Intellectual disability,(+) Motor delay,(+) Abnormal facial shape,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Cognitive impairment
NFRKB	ENST00000531755:c.1539C>T p.Leu513=		AR_homo	7.6	homo	maternal&paternal	5	NDD	Delayed speech and language development,(+) Intellectual disability,(+) Motor delay,(+) Abnormal facial shape,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Cognitive impairment
HIRIP3	ENST00000564026:c.420A>G p.Gln140=		AR_homo	8.9	homo	maternal&paternal	5	NDD	Delayed speech and language development,(+) Intellectual disability,(+) Motor delay,(+) Abnormal facial shape,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Cognitive impairment
MON2	ENST00000280379:c.1457C>T p.Ser486Phe		AD_unknown	4.8	het	unknown	1	NDD	(+) Short attention span,(+) Global developmental delay,(+) Intellectual disability, moderate,(+) Proportionate short stature,(+) Decreased body weight,(+) Abnormality of the Achilles tendon,(+) Tip-toe gait,(+) Decreased head circumference
ATP8B2	ENST00000368489:c.3617A>C p.Lys1206Thr		AD_denovo	5.4	het	de novo	1	Epilepsy	Focal-onset seizure
CACNG5	ENST00000169565:c.547G>A p.Ala183Thr		AD_denovo	6.1	het	de novo	1	NDD + epilepsy	severe global developmental delay, seizure, microcephaly, abnormal cerebral morphology
TMBIM6	ENST00000267115:c.385A>G p.Thr129Ala		AD_denovo	5.4	het	de novo	1	NDD + epilepsy	motor delay, hypotonia, seizure precipitated by febrile infection, abnormal tracheobronchial morphology, recurrent upper respiratory tract infections, obesity, epicanthus, hypertelorism, syndactyly, inverted nipples
RHOBTB2	ENST00000251822:c.2034C>G p.Tyr678*		AR_homo	10.7	homo	maternal&paternal	5	NDD	Delayed speech and language development,(+) Intellectual disability,(+) Motor delay,(+) Abnormal facial shape,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Cognitive impairment
ZNF787	ENST00000270459:c.350C>A p.Thr117Lys		AD_denovo	5.2	het	de novo	2	Epilepsy	Generalized non-motor (absence) seizure
THOC3	ENST00000265097:c.382G>C p.Asp128His		AD_denovo	5.0	het	de novo	2	Epilepsy	Generalized non-motor (absence) seizure
CLUH	ENST00000435359:c.1615C>T p.Arg539Cys		AR_homo	5.8	homo	maternal&paternal	2	NDD	global developmental delay, poor speech, behavioral abnormality with screaming fits, autistic behavior, sleep disturbance, facialis affection, polyphagia, macrosomia, elevated circulating creatine kinase concentration
SFXNG/PDZD7	ENST00000370215:c350C>G p.(Thr117Ser)		AR_homo	5.6	homo	maternal&paternal	2	NDD	global developmental delay, poor speech, behavioral abnormality with screaming fits, autistic behavior, sleep disturbance, facialis affection, polyphagia, macrosomia, elevated circulating creatine kinase concentration
NLRP14	ENST00000299481:c.1108dup p.Leu370Profs*19	ENST00000299481:c.2462+4_2462+7del None	AR_comphet	7.4	comphet	maternal&paternal	2	Epilepsy	Focal hemifacial clonic seizure
WDR7	ENST00000254442:c.2285A>T p.Glu762Val		AD_denovo	5.6	het	de novo	2	Epilepsy	Focal hemifacial clonic seizure
DIP2C	ENST00000280886:c.3430A>G p.Thr1144Ala		AD_unknown	5.9	het	unknown	1	NDD	intellectual disability, microcephaly, aplasia of the corpus callosum, hypotonia, increased body weight
ZBTB21	ENST00000310826:c.1731T>A p.Cys577*		AD_unknown	6.0	het	unknown	1	NDD	(+) Microcephaly,(+) Autism,(+) Moderate global developmental delay

FARP2	ENST00000264042:c.794T>C p.Phe265Ser		AR_homo	6.6	homo	maternal&paternal	3	NDD + epilepsy	global developmental delay, seizure, EEG abnormality, hypotonia alternating with opistho-tonus, abnormal heart morphology, ventricular septal defect, patent foramen ovale, mitral stenosis, recurrent infections, failure to thrive, poor suck, episodic vomiting, short stature, high palate, wide nasal bridge, facial hypertrichosis, brachytelomesophalangy
POLN	ENST00000382865:c.2308+1G>A None		AR_homo	8.9	homo	maternal&paternal	3	NDD + epilepsy	global developmental delay, seizure, EEG abnormality, hypotonia alternating with opistho-tonus, abnormal heart morphology, ventricular septal defect, patent foramen ovale, mitral stenosis, recurrent infections, failure to thrive, poor suck, episodic vomiting, short stature, high palate, wide nasal bridge, facial hypertrichosis, brachytelomesophalangy
ADAD2	ENST00000268624:c.731_732insA p.Leu245Alafs*37		AR_homo	8.6	homo	maternal&paternal	3	NDD + epilepsy	global developmental delay, seizure, EEG abnormality, hypotonia alternating with opistho-tonus, abnormal heart morphology, ventricular septal defect, patent foramen ovale, mitral stenosis, recurrent infections, failure to thrive, poor suck, episodic vomiting, short stature, high palate, wide nasal bridge, facial hypertrichosis, brachytelomesophalangy
SCAF1	ENST00000360565:c.111dup p.Ala38Serfs*17		AD_denovo	8.6	het	de novo	1	NDD	mild global developmental delay, speech development delay with blurred pronunciation, ne-glects motor skills in the right half of the body, hypotonia, poor suck, failure to thrive, facial dysmorphism including: narrow palate, narrow nasal bridge, long face, epicanthus, low anteri-or hairline, micrognathia
CELSR3	ENST00000164024:c.5386C>T p.Arg1796Trp		AD_unknown	6.5	het	unknown	2	NDD	(+) Autism,(+) Delayed speech and language development,(+) Global developmental delay
ESPL1	ENST00000257934:c.5364+6_5364+7dup None		AD_unknown	8.5	het	unknown	2	NDD	(+) Autism,(+) Delayed speech and language development,(+) Global developmental delay
NEURL3	ENST00000310865:n.824+1G>A None		AR_homo	7.5	homo	maternal&paternal	2	NDD + epilepsy	generalized tonic seizure, bilateral tonic-clonic seizure with generalized onset, nocturnal seizures, intellectual disability, delayed speech and language development, obesity
SCRN3	ENST00000272732:c.161G>A p.Cys54Tyr	ENST00000272732:c.1258_1261del p.Val420Lysfs*11	AR_comphet	5.5	comphet	maternal&paternal	2	NDD + epilepsy	generalized tonic seizure, bilateral tonic-clonic seizure with generalized onset, nocturnal seizures, intellectual disability, delayed speech and language development, obesity
NCOA4	ENST00000430396:c.36G>A p.Arg12=		AD_denovo	5.0	het	de novo	1	NDD	episodic ataxia, vertigo, falls, delayed gross motor development
C1orf109	ENST00000486637:c.218T>A p.Leu73Gln		AR_homo	5.0	homo	maternal&paternal	2	NDD	(+) Microcephaly,(+) Hypertonia,(+) Failure to thrive,(+) Increased serum lactate,(+) Opisthotonus,(+) Poor motor coordination,(+) Increased CSF lactate,(+) Hyperalaninemia,(+) Severe global developmental delay,(+) Abnormal visual fixation
USP21	ENST00000368002:c.709C>T p.Arg237Trp		AR_homo	4.9	homo	maternal&paternal	2	NDD	(+) Microcephaly,(+) Hypertonia,(+) Failure to thrive,(+) Increased serum lactate,(+) Opisthotonus,(+) Poor motor coordination,(+) Increased CSF lactate,(+) Hyperalaninemia,(+) Severe global developmental delay,(+) Abnormal visual fixation
ABHD6	ENST00000295962:c.349G>T p.Asp117Tyr		AD_denovo	6.0	het	de novo	2	NDD	global developmental delay, autistic behavior, hypotonia, tall stature
GDA	ENST00000238018:c.943G>A p.Val315Met		AD_denovo	6.5	het	de novo	1	NDD + epilepsy	myoclonic absence seizure, moderate global developmental delay, low levels of vitamin D
MTPN	ENST00000393085:c.248G>A p.Cys83Tyr		AD_denovo	5.6	het	de novo	1	epilepsy	bilateral tonic-clonic seizure, focal motor seizure
CEP89	ENST00000305768:c.457_458delinsGA p.Ser153Asp		AD_denovo	6.6	het	de novo	1	NDD	Moderate global developmental delay
GRIPAP1	ENST00000376423:c.1503C>A p.Asp501Glu		XL	5.8	hemi	maternal	5	NDD	(+) Microcephaly,(+) Abnormality of the face,(+) Hypertelorism,(+) Global developmental delay,(+) Severe expressive language delay
DENND4C	ENST00000434457:c.4285G>A p.Val1429Ile		AR_homo	5.2	homo	maternal&paternal	5	NDD	(+) Microcephaly,(+) Abnormality of the face,(+) Hypertelorism,(+) Global developmental delay,(+) Severe expressive language delay
SLC4A7	ENST00000295736:c.307G>A p.Asp103Asn		AR_homo	5.3	homo	maternal&paternal	5	NDD	(+) Microcephaly,(+) Abnormality of the face,(+) Hypertelorism,(+) Global developmental delay,(+) Severe expressive language delay
CCT3	ENST00000295688:c.791dup p.Thr265Hisfs*16		AD_unknown	7.9	het	unknown	1	NDD	(+) Microcephaly,(+) Intellectual disability,(+) Short stature,(+) Attention deficit hyperactivity disorder
KIF27	ENST00000297814:c.1016G>A p.Arg339Gln		AR_homo	6.0	homo	maternal&paternal	5	NDD	(+) Microcephaly,(+) Abnormality of the face,(+) Hypertelorism,(+) Global developmental delay,(+) Severe expressive language delay
UNC13C	ENST00000260323:c.422del p.Gln141Argfs*36		AR_homo	9.9	homo	maternal&paternal	5	NDD	(+) Microcephaly,(+) Abnormality of the face,(+) Hypertelorism,(+) Global developmental delay,(+) Severe expressive language delay
PDZRN3	ENST00000263666:c.506_536del p.His169Argfs*33		AD_unknown	B	het	unknown	1	Wachstum, Skelett	(+) Trigonocephaly,(+) Metopic synostosis

DIP2C	ENST00000280886:c.1303G>A p.Gly435Arg		AD_denovo	7.9	het	de novo	1	NDD + epilepsy	seizure, moderate global developmental delay, ataxia, spasticity, hypotonia, oculomotor apraxia, abnormality of brain morphology, small stature
ZFR	ENST00000265069:c.3G>A p.Met1?		AD_unknown	7.8	het	unknown	1	NDD + epilepsy	(+) Intellectual disability, mild,(+) Generalized-onset seizure
DNAH6	ENST00000237449:c.11225T>C p.Leu3742Pro		AD_denovo	7.0	het	de novo	5	NDD + epilepsy	focal autonomic seizure with epigastric sensation/nausea/vomiting/other gastrointestinal phenomena, focal tonic seizure, generalized tonic seizure, simple febrile seizure, mild glo-bal developmental delay, mild ataxia
SOX1	ENST00000330949:c.684_692del p.Ala229_Pro231del		AD_denovo	6.9	het	de novo	5	NDD + epilepsy	focal autonomic seizure with epigastric sensation/nausea/vomiting/other gastrointestinal phenomena, focal tonic seizure, generalized tonic seizure, simple febrile seizure, mild glo-bal developmental delay, mild ataxia
OTOP1	ENST00000296358:c.318G>A p.Trp106*		AR_homo	8.3	homo	maternal&paternal	5	NDD + epilepsy	focal autonomic seizure with epigastric sensation/nausea/vomiting/other gastrointestinal phenomena, focal tonic seizure, generalized tonic seizure, simple febrile seizure, mild glo-bal developmental delay, mild ataxia
FSTL4	ENST00000265342:c.784G>A p.Val262Met		AR_homo	5.0	homo	maternal&paternal	5	NDD + epilepsy	focal autonomic seizure with epigastric sensation/nausea/vomiting/other gastrointestinal phenomena, focal tonic seizure, generalized tonic seizure, simple febrile seizure, mild glo-bal developmental delay, mild ataxia
ZNF236	ENST00000253159:c.281C>T p.Thr94Ile		AR_homo	5.3	homo	maternal&paternal	5	NDD + epilepsy	focal autonomic seizure with epigastric sensation/nausea/vomiting/other gastrointestinal phenomena, focal tonic seizure, generalized tonic seizure, simple febrile seizure, mild glo-bal developmental delay, mild ataxia
BRWD1	ENST00000333229:c.1840T>C p.Ser614Pro		AD_denovo	6.9	het	de novo	1	epilepsy	Seizures with absences and myoclonias since 18 months of age
CNTN5	ENST00000524871:c.264_265insAACTGAGGAACC AGGCATTATTTTGTCTGATAGATCCAAAATTGACAAA GGTAGACAACATCTAGAAAATATTA p.Phe89Asnfs*2		AD_denovo	7.4	het	de novo	1	NDD	moderate global developmental delay, absent speech, autism, hypotonia, restlessness, joint hypermobility, clinodactyly of the 5th finger, hypertrichosis, facial dysmorphism (epi-canthus, depressed nasal ridge, upslanted palpebral fissure, synophrys)
PAXIP1	ENST00000397192:c.2177T>C p.Leu726Ser		AD_denovo	7.0	het	de novo	2	NDD	profound global developmental delay, developmental regression, hypotonia, hypoplasia of the corpus callosum, delayed CNS myelination, abnormal macular morphology, abnormal foot morphology, abnormality of the Achilles tendon, increased serum lactate, increased circulating lactate dehydrogenase concentration
PCM1	ENST00000325083:c.3391A>C p.Asn1131His	ENST00000325083:c.131C>T p.Ser44Leu	AR_comphet	6.2	comphet	maternal&paternal	2	NDD	profound global developmental delay, developmental regression, hypotonia, hypoplasia of the corpus callosum, delayed CNS myelination, abnormal macular morphology, abnormal foot morphology, abnormality of the Achilles tendon, increased serum lactate, increased circulating lactate dehydrogenase concentration
GRPEL1	ENST00000264954:c.238C>T p.Arg80*	ENST00000264954:c.613A>G p.Thr205Ala	AR_comphet	6.1	comphet	maternal&paternal	1	NDD + epilepsy	Pachygyria,(+) Generalized-onset seizure,(+) Focal-onset seizure
MARCH7	ENST00000259050:c.393del p.Gly133Aspfs*7		AD_inherited	7.0	het	unknown	1	NDD	Arachnodactyly,(+) Intellectual disability, moderate,(+) Long toe
SLIT2	ENST00000273739:c.2820del p.Pro941Glnfs*38		AD_unknown	8.7	het	unknown	1	NDD	(+) Hearing impairment,(+) Cholelithiasis,(+) Exocrine pancreatic insufficiency,(+) Spontaneous pneumothorax,(+) Mild global developmental delay
GRM5	ENST00000305432:c.2446T>C p.Cys816Arg		AD_unknown	10.5	het	unknown	1	NDD	moderate bis schwere Entwicklungsverzögerung, Sprachstörung, große Ohren
LRFN4	ENST00000309602:c.1160C>T p.Ser387Leu		AR_homo	6.0	homo	maternal&paternal	2	NDD + epilepsy	febrile status epilepticus, EEG abnormality, encephalopathy, reduced consciousness, global developmental delay, absent speech, spasticity, respiratory insufficiency, venous thrombosis internal jugular vein, microcephaly, poor eye contact, mask-like facies, elevated hepatic transaminase, increased body weight, increased blood pressure
PLXNA2	ENST00000367033:c.2594C>T p.Thr865Met		AR_homo	8.0	homo	maternal&paternal	1	NDD + epilepsy	(+) Microcephaly,(+) Global developmental delay,(+) Encephalopathy,(+) Increased body weight,(+) Febrile status epilepticus
ADGRF4/GPR115	ENST00000283303:c.1860del p.Phe620Leufs*3		AR_homo	8.5	homo	maternal&paternal	4	NDD + epilepsy	(+) Hypothyroidism,(+) Seizure,(+) Intellectual disability, mild,(+) Global developmental delay,(+) Gliosis
EPHB1	ENST00000398015:c.2204T>C p.Leu735Pro		AD_denovo	7.8	het	de novo	4	NDD + epilepsy	(+) Hypothyroidism,(+) Seizure,(+) Intellectual disability, mild,(+) Global developmental delay,(+) Gliosis
DOP1A/DOPEY1	ENST00000237163:c.5210A>T p.Glu1737Val		AR_homo	5.4	homo	maternal&paternal	4	NDD + epilepsy	(+) Hypothyroidism,(+) Seizure,(+) Intellectual disability, mild,(+) Global developmental delay,(+) Gliosis
PHTF2	ENST00000248550:c.2122C>T p.Leu708Phe		AD_denovo	5.4	het	de novo	4	NDD + epilepsy	(+) Hypothyroidism,(+) Seizure,(+) Intellectual disability, mild,(+) Global developmental delay,(+) Gliosis
BSN	ENST00000296452:c.8746_8749del p.Gln2916Cysfs*11		AD_unknown	9.5	het	unknown	1	NDD + epilepsy	(+) Intellectual disability,(+) Seizure,(+) Global developmental delay,(+) Postural instability,(+) Tetraparesis

MYCBP2	ENST00000357337:c.8674A>T p.Ser2892Cys		AD_unknown	6.3	het	unknown	1	Epilepsy	(+) Migraine,(+) EEG abnormality,(+) Focal-onset seizure,(+) Generalized tonic seizure,(-) Abnormality of brain morphology,(+) Seizure precipitated by febrile infection
TPR	ENST00000367478:c.2943+2T>C None		AD_unknown	9.0	het	unknown	1	NDD	(+) Autism,(+) Delayed speech and language development,(+) Global developmental delay,(+) EEG abnormality,(+) Tip-toe gait
PRMT9	ENST00000322396:c.1144C>A p.Gln382Lys		AR_homo	4.24	homo	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, dyslexia, Pes planus, Focal tonic seizure, Cognitive impairment
LAMA5	ENST00000252999:c.5408C>T p.Ser1803Phe		AR_homo	6.07	homo	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, dyslexia, Pes planus, Focal tonic seizure, Cognitive impairment
DBN1	ENST00000292385:c.1663_1664delinsCT p.Ser555Leu	ENST00000292385:c.1452C>G p.Asn484Lys	AR_comphet	5.18	comphet	maternal&paternal	3	NDD + Epilepsy	Intellectual disability, dyslexia, Pes planus, Focal tonic seizure, Cognitive impairment
ARSF	ENST00000359361:c.1156C>T p.Arg386*		XL	6.24	hemi	maternal	1	NDD	suspected neurodegenerative disease, leukoencephalopathy, horizontal nystagmus, slowed slurred speech, respiratory distress
PLCH1	ENST00000334686:c.2813del p.Asn938Thrfs*24		AD_unknown	5.55	het	unknown	1	NDD + Epilepsy	(+) Microcephaly,(+) Seizure,(+) Short stature,(+) Severe global developmental delay
DIP2B	ENST00000301180:c.3346C>T p.Arg1116*		AD	7.63	het	maternal	2	Epilepsy	(+) Generalized non-motor (absence) seizure
PLXNA1	ENST00000251772:c.4817C>T p.Thr1606Met		AD_denovo	6.43	het	de novo	1	NDD	leukoencephalopathy
SVEP1	ENST00000401783:c.2708T>A p.Leu903*	ENST00000401783:c.9653G>A p.Cys3218Tyr	AR_comphet	5.32	comphet	maternal&paternal	3	NDD + epilepsy	Structural epilepsy with epileptic spasms since the age of 10 months, bilateral extensive gyration disorder, severe developmental disorder, macrocephaly, former twin premature baby of 33+1 SSW, condition after hydrops fetals of unclear aetiology.
ATP13A4	ENST00000295548:c.826G>A p.Glu276Lys		AR_homo	6.18	homo	maternal&paternal	3	NDD + epilepsy	Structural epilepsy with epileptic spasms since the age of 10 months, bilateral extensive gyration disorder, severe developmental disorder, macrocephaly, former twin premature baby of 33+1 SSW, condition after hydrops fetals of unclear aetiology.
CFAP57/WDR65	ENST00000372492:c.176G>A p.Gly59Asp		AD_denovo	5.46	het	de novo	3	NDD + epilepsy	Structural epilepsy with epileptic spasms since the age of 10 months, bilateral extensive gyration disorder, severe developmental disorder, macrocephaly, former twin premature baby of 33+1 SSW, condition after hydrops fetals of unclear aetiology.
LPIN3	ENST00000373257:c.254A>G p.Glu85Gly		AD_denovo	5.9	het	de novo	1	Epilepsy	epileptic encephalopathy
MKRN1	ENST00000480552:c.262A>G p.Thr88Ala		AD_denovo	5.81	het	de novo	2	NDD	severe global developmental delay, autism, periventricular white matter hypodensities
HDAC9	ENST00000441542:c.800G>A p.Arg267His	ENST00000441542:c.2917G>C p.Val973Leu	AR_comphet	6.2	comphet	maternal&paternal	2	NDD	severe global developmental delay, autism, periventricular white matter hypodensities
EGFL6	ENST00000361306:c.954T>G p.Tyr318*		XL	5.54	hemi	maternal	2	NDD + epilepsy	epileptic encephalopathy, generalized non-motor (absence) seizure, mild intellectual disability, behavioral abnormality, myoclonus, excessive salivation
TAOK2	ENST00000279394:c.2529C>A p.Tyr843*		AD_unknown	7.51	het	unknown	1	NDD + epilepsy	(+) Global developmental delay,(+) Obesity,(+) Focal-onset seizure
KCNC2	ENST00000549446:c.1412T>C p.Val471Ala		AD_denovo	8.66	het	de novo	2	NDD + epilepsy	epileptic encephalopathy, generalized non-motor (absence) seizure, mild intellectual disability, behavioral abnormality, myoclonus, excessive salivation
HIP1	ENST00000336926:c.2377_2378del p.Ala793Tyrfs*2		AD_denovo	9.5	het	de novo	1	NDD + epilepsy	(+) Focal-onset seizure,(+) Mild global developmental delay
ARID3B	ENST00000346246:c.593G>A p.Arg198Gln		AD_denovo	5.08	het	de novo	1	NDD	severe global developmental delay, autism
ARHGAP21	ENST00000320481:c.806C>T p.Thr269Met		AD_denovo	6.64	het	de novo	1	NDD	developmental delay, macrocephalus, muscular hypotonia, strabismus convergens, obesity, tall stature
TCERG1	ENST00000296702:c.592C>T p.Gln198*		AD_unknown	7.43	het	unknown	1	NDD	(+) Behavioral abnormality,(+) Osteoporosis,(+) Intellectual disability,(+) Global developmental delay,(+) Spondylitis
HDAC2	ENST00000368632:c.88G>T p.Ala30Ser		AR_homo	7.72	homo	maternal&paternal	2	Epilepsy	ventriculomegaly, apneic episodes in infancy, cerebral hypomyelination, focal-onset seizure
ARHGAP21	ENST00000320481:c.2132C>T p.Ser711Phe		AD_denovo	5.77	het	de novo	2	Epilepsy	ventriculomegaly, apneic episodes in infancy, cerebral hypomyelination, focal-onset seizure
ELFN2	ENST00000402918:c.221C>A p.Ser74*		AD_unknown	B	het	unknown	1		Amyotrophic lateral sclerosis

EPHA1	ENST00000275815:c.1245C>A p.Tyr415*		AD_denovo	6.93	het	de novo	1	NDD + epilepsy	generalized-onset seizure, focal-onset seizure, profound global developmental delay, ab-normal cerebellum morphology, hypoplasia of the cerebellar vermis, spastic tetraparesis, flexion contractu
GAK	ENST00000314167:c.3742C>T p.Arg1248Cys	ENST00000314167:c.986_989del p.Thr329Serfs*90	AR_comphet	7.8	comphet	maternal& paternal	1	NDD	Intrauterine growth retardation, developmental delay emphasising speech, short stature, relative macrocephaly, benign enlargement of the external cerebrospinal fluid spaces, high hairline
INHBA	ENST00000242208:c.188T>C p.Leu63Ser		AD_denovo	5.67	het	de novo	3	NDD + epilepsy	Global developmental delay,(+) EEG with spike-wave complexes,(+) Nocturnal seizures
CKAP5	ENST00000312055:c.1157A>G p.Asp386Gly		AD_denovo	8.85	het	de novo	3	NDD + epilepsy	Global developmental delay,(+) EEG with spike-wave complexes,(+) Nocturnal seizures
CROCC	ENST00000375541:c.949del p.Thr317Leufs*26	ENST00000375541:c.3394G>A p.Ala1132Thr	AR_comphet	5.96	comphet	maternal& paternal	3	NDD + epilepsy	Global developmental delay,(+) EEG with spike-wave complexes,(+) Nocturnal seizures
AMOT	ENST00000304758:c.401A>G p.Lys134Arg		XL	5.47	hemi	maternal	3	NDD	Global developmental delay, autism
KLHL23	ENST00000392647:c.1573C>T p.Gln525*		AD_denovo	4.9	het	de novo	3	NDD	Global developmental delay, autism
SLC4A5	ENST00000377634:c.3122G>T p.*1041Leuext*5	ENST00000377634:c.1486G>A p.Gly496Ser	AR_comphet	5.28	comphet	maternal& paternal	3	NDD	Global developmental delay, autism
SNAP91	ENST00000195649:c.2516del p.Pro839Leufs*8		AD_unknown	10.0	het	unknown	1	Epilepsy	(+) Focal-onset seizure
ITPR2	ENST00000381340:c.6458G>A p.Arg2153Gln		AD_denovo	8.81	het	de novo	1	Epilepsy	Since 01/2021, daily recurrent seizures with nocturnal frequency (5-15 times per day). According to the EEG, multifocal onset with secondary generalisation. Occasional slurred speech since onset of seizures.
RNF2	ENST00000367509:c.442_443insC p.Met148Thrfs*5		AD_unknown	8.86	het	unknown	1	NDD	(+) Anal atresia,(+) Arnold-Chiari malformation,(+) Spina bifida,(+) Short stature,(+) Infantile muscular hypotonia,(+) Severe global developmental delay
ESPL1	ENST00000257934:c.32del p.Thr11Ilefs*3		AD_unknown	8.45	het	unknown	1	NDD + epilepsy	(+) Intellectual disability, mild,(+) Bilateral tonic-clonic seizure,(+) Generalized myoclonic-atonic seizure,(+) Nocturnal seizures
NOVA1	ENST00000267422:c.325C>T p.Arg109*		AR_homo	11.3	homo	unknown	1	NDD	(+) Retrognathia,(+) Hyperreflexia,(+) Flexion contracture,(+) Small for gestational age,(+) Dysphagia,(+) Respiratory insufficiency,(+) Muscle stiffness,(+) Severe global developmental delay
NRXN3	ENST00000281127:c.1030C>T p.Pro344Ser		AD_unknown	8.32	het	unknown	1	NDD + epilepsy	(+) Autism,(+) Global developmental delay,(+) Pachygyria,(+) Polymicrogyria,(+) Status epilepticus,(+) Intellectual disability, moderate,(+) Focal myoclonic seizure,(+) Focal tonic seizure
SNRNP70	ENST00000221448:c.1124G>A p.Gly375Asp		AD_unknown	5.06	het	unknown	1	NDD	(+) Macrocephaly,(+) Optic atrophy,(+) Global developmental delay,(+) Dandy-Walker malformation
TSKU	ENST00000333090:c.188_189dup p.Asp64Trpfs*11		AD_unknown	A	het	unknown	1	other	(+) Leukoencephalopathy,(+) Cerebral ischemia,(+) Recurrent subcortical infarcts,(+) Subcortical cerebral atrophy
MYO9B	ENST00000595618:c.5551G>A p.Ala1851Thr		AD_denovo	5.8	het	de novo	1	NDD	severe dystrophy (BMI 11), severe global developmental delay, dyskinetic-dystonic movement disorder, basal ganglia disease of unknown origin (not progressive, Segawa syndrome suspected), symmetric gliotic changes on both sides in globus pallidus, otherwise MRI without pathological findings, abnormal development since the 4th month of life until then normal development
ARHGAP21	ENST00000396432:c.4871G>A p.Ser1624Asn		AD_denovo	6.81	het	de novo	3	NDD	global developmental delay, hypotonia, pulmonic stenosis, bilateral single transverse palmar creases, microretrognathia, feeding difficulties
NOC4L	ENST00000330579:c.901G>T p.Gly301Trp		AD_denovo	5.58	het	de novo	3	NDD	global developmental delay, hypotonia, pulmonic stenosis, bilateral single transverse palmar creases, microretrognathia, feeding difficulties
CXorf36	ENST00000377934:c.233+4A>G None		XL	8.17	hemi	maternal	3	NDD	global developmental delay, hypotonia, pulmonic stenosis, bilateral single transverse palmar creases, microretrognathia, feeding difficulties
C9orf172	ENST00000436881:c.2006_2016del p.Arg669Hisfs*112		AD_unknown	6.38	het	unknown	1	NDD	(+) Moderate global developmental delay
TEKT4	ENST00000295201:c.1101_1104del p.Ser367Argfs*5		AR_homo	8.39	homo	unknown	1	NDD	Delayed speech and language development

ADRM1	ENST00000253003:c.1015-2A>G None		AD_unknown	7.05	het	unknown	1	NDD	(+) Microcephaly,(+) Autism,(+) Mild global developmental delay
MAP2K4	ENST00000353533:c.841C>T p.Arg281*		AD_denovo	9.05	het	de novo	1	NDD + epilepsy	Seizure. Intellectual disability, joint laxity
ATP8A2	ENST00000381655:c.560A>G p.Asp187Gly	ENST00000381655:c.368C>T p.Pro123Leu	AR_comphet	7.91	comphet	maternal&paternal	2	NDD + epilepsy	premature birth by sectio in breech presentation, severe mental retardation, no active speech, generalized ataxia, dystrophy (BMI 15.8), oculomotor dysfunction, dysphagia; significant developmental delay after first 6-vaccination, no head and trunk control, scoliosis, dystonia and spasticity, epilepsy with focal and generalized signs with tonic-clonic seizures. Human genetics (Spranger's practice, Bremen; 2018): Exclusion of SCN1A mutation and pathogenic CNV
SULT2A1	ENST00000222002:c.371G>C p.Arg124Thr		AD_denovo	5.28	het	de novo	2	NDD + epilepsy	premature birth by sectio in breech presentation, severe mental retardation, no active speech, generalized ataxia, dystrophy (BMI 15.8), oculomotor dysfunction, dysphagia; significant developmental delay after first 6-vaccination, no head and trunk control, scoliosis, dystonia and spasticity, epilepsy with focal and generalized signs with tonic-clonic seizures. Human genetics (Spranger's practice, Bremen; 2018): Exclusion of SCN1A mutation and pathogenic CNV
ABCA13	ENST00000435803:c.13921G>A p.Gly4641Ser	ENST00000435803:c.14182C>T p.Arg4728Ter	AR_comphet	5.04	comphet	maternal&paternal	2	NDD + epilepsy	mild developmental delay, focal seizure, ganglioglioma (with 5 years)
PDS5B	ENST00000315596:c.30del p.Asp10Gluufs*23		AD_denovo	10.7	het	de novo	2	NDD + epilepsy	mild developmental delay, focal seizure, ganglioglioma (with 5 years)
CLASP2	ENST00000313350:c.170C>T p.Pro57Leu		AD_unknown	6.28	het	unknown	1	NDD + epilepsy	(+) Autism,(+) Focal-onset seizure,(+) Moderate global developmental delay
UBE2H	ENST00000355621:c.449C>T p.Thr150Met		AD_denovo	7.89	het	de novo	1	NDD	Developmental delay with autistic features, muscular hypotonia, strabismus divergens, hyperopia, exotropia
GSE1	ENST00000253458:c.1921del p.Arg641Valfs*66		AD_unknown	7.04	het	unknown	1	NDD	(+) Autism,(+) Moderate global developmental delay
DHX15	ENST00000336812:c.955G>A p.Val319Ile		AD_denovo	7.28	het	de novo	1	NDD	moderate global developmental delay, absent speech, behavioral abnormality, autistic be-havior, growth delay
RNF44	ENST00000274811:c.802C>A p.Pro268Thr		AD_denovo	5.42	het	de novo	1	NDD	bilateral tonic-clonic seizure (first seizure 07/2017), autism, mixed hepatopathy of unclear etiology, abnormal circulating lipid concentration, left ventricular diastolic dysfunction
KCTD16	ENST00000507359:c.521G>A p.Cys174Tyr		AR_homo	5.18	homo	maternal&paternal	3	NDD + epilepsy	severe developmental delay, focal epilepsy
PDLIM5	ENST00000317968:c.737G>T p.Arg246Leu		AR_homo	5.64	homo	maternal&paternal	3	NDD + epilepsy	severe developmental delay, focal epilepsy
MTRF1L	ENST00000367230:c.641G>A p.Gly214Glu		AD_denovo	6.56	het	de novo	3	NDD + epilepsy	severe developmental delay, focal epilepsy
AGPAT3	ENST00000291572:c.250C>T p.Arg84Cys		AR_homo	4.54	homo	maternal&paternal	12	NDD + Epilepsy	Seizures, Global developmental delay, Microcephaly, Hearing impairment, Visual impairment, Intellectual disability,
AMY2B	ENST00000361355:c.944A>T p.Asp315Val		AR_homo	5.19	homo	maternal&paternal	12	NDD + Epilepsy	Seizures, Global developmental delay, Microcephaly, Hearing impairment, Visual impairment, Intellectual disability,
GIMAP2	ENST00000223293:c.783C>A p.Cys261*		AR_homo	6.97	homo	maternal&paternal	12	NDD + Epilepsy	Seizures, Global developmental delay, Microcephaly, Hearing impairment, Visual impairment, Intellectual disability,
ARID5B	ENST00000279873:c.1909G>T p.Asp637Tyr		AR_homo	6.21	homo	maternal&paternal	12	NDD + Epilepsy	Seizures, Global developmental delay, Microcephaly, Hearing impairment, Visual impairment, Intellectual disability,
YBX3	ENST00000228251:c.450G>T p.Lys150Asn		AR_homo	5.18	homo	maternal&paternal	12	NDD + Epilepsy	Seizures, Global developmental delay, Microcephaly, Hearing impairment, Visual impairment, Intellectual disability,
PTPRH	ENST00000376350:c.655C>T p.Gln219*		AR_homo	7.02	homo	maternal&paternal	12	NDD + Epilepsy	Seizures, Global developmental delay, Microcephaly, Hearing impairment, Visual impairment, Intellectual disability,
KCNJ15	ENST00000328656:c.83G>A p.Arg28His		AR_homo	5.74	homo	maternal&paternal	12	NDD + Epilepsy	Seizures, Global developmental delay, Microcephaly, Hearing impairment, Visual impairment, Intellectual disability,
AGPAT3	ENST00000291572:c.250C>T p.Arg84Cys		AR_homo	4.54	homo	maternal&paternal	12	NDD + Epilepsy	Seizures, Global developmental delay, Microcephaly, Hearing impairment, Visual impairment, Intellectual disability,
GRINA	ENST00000313269:c.967-6C>T None		AR_homo	4.08	homo	maternal&paternal	12	NDD + Epilepsy	Seizures, Global developmental delay, Microcephaly, Hearing impairment, Visual impairment, Intellectual disability,
MED16	ENST00000269814:c.1246G>T p.Glu416*		AD_unknown	6.54	het	unknown	1	NDD + Epilepsy	Intellectual disability, tonic seizure, myoclonic-atonic seizure, tonic-clonic seizure with generalized onset, myoclonic seizure, periventricular heterotopia
INPP5D	ENST00000359570:c.919G>A p.Asp307Asn		AR_homo	5.18	homo	unknown	1	NDD + Epilepsy	(+) Hypertension,(+) Global developmental delay,(+) Obesity,(+) Mitral regurgitation,(+) Bilateral tonic-clonic seizure,(+) Intellectual disability, moderate
WDFY4	ENST00000265453:c.3130G>A p.Val1044Met		AD_unknown	5.83	het	unknown	2	NDD+epilepsy	(+) Autism,(+) Gliosis,(+) Generalized-onset seizure,(+) Severe global developmental delay

PTPRCAP	ENST00000326294:c.280C>T p.Arg94*		AD_unknown	4.47	het	unknown	2	NDD+epilepsy	(+) Autism,(+) Gliosis,(+) Generalized-onset seizure,(+) Severe global developmental delay
CCDC121	ENST00000324364:c.816dup p.Ser273Ilefs*4		AD_denovo	7.02	het	de novo	1	NDD	early childhood autism with expressive language disorder, behavioural problems
HECW1	ENST00000395891:c.2588C>T p.Ala863Val		AD_unknown	5.16	het	unknown	1	NDD+epilepsy	(+) Generalized non-motor (absence) seizure,(+) EEG with focal spike waves,(+) Mild global developmental delay
IMPDH2	ENST00000326739:c.613A>G p.Lys205Glu		AD_denovo	7.55	het	de novo	1	NDD	combined developmental delay, crawling at 13 months, walking at 20 months, first words approx. 11 months, incorrect pronunciation, small hands, small feet, body measurements within normal range
APBB1	ENST00000299402:c.1781A>G p.Gln594Arg		AD_denovo	7.83	het	de novo	1	NDD+epilepsy	(+) Status epilepticus,(+) Focal-onset seizure
THOC2	ENST00000245838:c.2062G>A p.Gly688Arg		XL	8.77	het	de novo	1	NDD+epilepsy	Intellectual disability,(+) Brain very small,(+) Focal-onset seizure
FAT3	ENST00000298047:c.656T>G p.Leu219*		AD_denovo	8.99	het	de novo	1	NDD+epilepsy	(+) Global developmental delay,(+) Generalized-onset seizure,(+) Febrile seizure (within the age range of 3 months to 6 years)
FBXL19	ENST00000338343:c.26_32dup p.Ala12Glyfs*20		AD_inherited	8.85	het	paternal	1	NDD	(+) Microcephaly,(+) Delayed speech and language development,(+) Hirsutism,(+) Intellectual disability,(+) Intellectual disability, mild,(+) Global developmental delay,(+) Absent speech,(+) Intellectual disability, moderate,(+) Poor speech,(+) Short stature,(+) Frontal hirsutism,(+) Cognitive impairment
CAMSAP1	ENST00000312405:c.2749del p.Met917*		AD_unknown	6.92	het	unknown	1	Epilepsy	generalized onset seizure
NTRK2	ENST00000277120:c.2404A>T p.Met802Leu		AD_denovo	11.5	het	de novo	1	NDD	global developmental delay, short stature, recurrent hypoglycaemia under growth hormone therapy, postprandial hyperglycemia, obesity, anterior pituitary dysgenesis, ectopic posterior pituitary, posterior pituitary hypoplasia, pituitary growth hormone deficiency, aqueductal stenosis, condition after arachnoid cyst with hydrocephalus occlusus, space-occupying structure in epipharynx DD sphenoid cephalocele with hamartomatous lesion in epipharynx, aplasia/hypoplasia of the cervical spine, atlas dislocation, dens aplasia, hypoplasia of the spinous process C4 and C5, severe thinning and caudal displacement of the chiasm into the hypophyseal fossa and elongation of the optic tract, hiatus hernia, abnormal stomach morphology, secondary spleen, recurrent iron deficiency anemia
DLGAP2	ENST00000421627:c.1297C>A p.Gln433Lys		AD_inherited	6.21	het	paternal	2	NDD	(+) Behavioral abnormality,(+) Autistic behavior,(+) Sleep disturbance,(+) Developmental regression
DNAJC28	ENST00000314399:c.401G>A p.Arg134Gln		AR_homo	4.68	homo	maternal&paternal	2	NDD	(+) Behavioral abnormality,(+) Autistic behavior,(+) Sleep disturbance,(+) Developmental regression
AKAP7	ENST00000368123:c.611C>G p.Ser204*		AR_homo	8.23	homo	unknown	3	NDD+epilepsy	(+) Seizure,(+) Hypotonia,(+) Global developmental delay,(+) Intellectual disability, moderate,(+) Dystonic gait,(+) Periodic fever
UBE2O	ENST00000319380:c.3046C>T p.Pro1016Ser		AD_unknown	5.7	het	unknown	1	NDD	(+) Profound global developmental delay
RIMBP2	ENST00000261655:c.3121A>G p.Lys1041Glu	ENST00000261655:c.824C>T p.Ala275Val	AR_comphet	5.1	het	maternal&paternal	1	NDD	(+) Macrocephaly,(+) Abnormality of neuronal migration,(+) Cerebral white matter hypoplasia
CLDN5	ENST00000403084:c.358T>C p.Phe120Leu		AD_denovo	7.07	het	de novo	1	NDD+epilepsy	(+) Microcephaly,(+) Global developmental delay,(+) Bilateral tonic-clonic seizure,(+) Generalized-onset seizure,(+) Abnormality of neuronal migration,(+) Motor seizure,(+) Decreased head circumference
PTPRD	ENST00000355233:c.1318_1319insAGGA p.Glu440Glyfs*4		AD_unknown	9.64	het	unknown	2	epilepsy	(+) Seizure,(+) Mild global developmental delay
SIK2	ENST00000304987:c.1337dup p.Asn446Lysfs*8		AD_unknown	6.51	het	unknown	2	epilepsy	(+) Seizure,(+) Mild global developmental delay
KIAA1217	ENST00000376454:c.2698T>C p.Ser900Pro		AR_homo	5.23	homo	maternal&paternal	3	epilepsy	seizure, hypsarrhythmia, infantile spasms
RNF123	ENST00000327697:c.79A>C p.Thr27Pro		AD_denovo	6.33	het	de novo	3	epilepsy	seizure, hypsarrhythmia, infantile spasms
DOCK5	ENST00000276440:c.1595G>A p.Arg532Gln	ENST00000276440:c.3284-6_3284-2del None	AR_comphet	5.3	comphet	maternal&paternal	3	epilepsy	seizure, hypsarrhythmia, infantile spasms
PTBP3	ENST00000334318:c.1120_1121insACAC p.Thr374Asnfs*15		AD_unknown	5.72	het	unknown	2		(+) Hydrocephalus,(+) Premature birth,(+) Generalized-onset seizure,(+) Focal-onset seizure,(+) Intraventricular hemorrhage
HTR2C	ENST00000276198:c.896C>T p.Thr299Ile		XL	7.44	hemi	maternal	1	epilepsy	Therapy-resistant epilepsy (absence), intellectual disability

COPS2	ENST00000299259:c.784C>T p.His262Tyr		AD_denovo	8.22	het	de novo	2	NDD+epilepsy	(+) Microcephaly,(+) Abnormality of the face,(+) Strabismus,(+) 2-3 finger syndactyly,(+) Intellectual disability,(+) Severe global developmental delay
MBOAT2	ENST00000305997:c.1027A>G p.Ile343Val		AD_denovo	5.64	het	de novo	2	NDD+epilepsy	(+) Microcephaly,(+) Abnormality of the face,(+) Strabismus,(+) 2-3 finger syndactyly,(+) Intellectual disability,(+) Severe global developmental delay
DHX9	ENST00000367549:c.3787C>T p.Gln1263Ter		AD_unknown	7.04	het	unknown	2		(+) Hydrocephalus,(+) Premature birth,(+) Generalized-onset seizure,(+) Focal-onset seizure,(+) Intraventricular hemorrhage
KLHL20	ENST00000209884:c.1211C>G p.Thr404Arg		AD_unknown	4.77	het	unknown	1	NDD+epilepsy	(+) Intellectual disability, moderate,(+) Focal-onset seizure
PATL1	ENST00000300146:c.1031+1G>A None		AD_unknown	7.55	het	unknown	1	NDD+epilepsy	(+) Insulin resistance,(+) Obesity,(+) Generalized-onset seizure,(+) Severe global developmental delay
FAM222A	ENST00000358906:c.1231del p.Tyr411Ilefs*80		AD_unknown	6.31	het	unknown	2	NDD	(+) Hypotonia,(+) Growth delay,(+) Abnormality of acid-base homeostasis,(+) Relative macrocephaly,(+) Mild global developmental delay,(+) Feeding difficulties,(+) Vitamin B12 deficiency
HELZ	ENST00000358691:c.2117A>G p.Tyr706Cys		AD_unknown	5.29	het	unknown	2	NDD	(+) Hypotonia,(+) Growth delay,(+) Abnormality of acid-base homeostasis,(+) Relative macrocephaly,(+) Mild global developmental delay,(+) Feeding difficulties,(+) Vitamin B12 deficiency
HEATR1	ENST00000366581:c.360-1G>A None		AD_unknown	7.44	het	unknown	1	NDD + cardio	(+) Microcephaly,(+) Short attention span,(+) Global developmental delay,(+) Tetralogy of Fallot
DOCK11	ENST00000276202:c.1034A>G p.Gln345Arg		XL	5.03	hemi	maternal	1	NDD+epilepsy	(+) Focal-onset seizure,(+) Moderate global developmental delay,(+) Continuous spike and waves during slow sleep
ASCC3	ENST00000369162:c.4658C>T p.Ser1553Phe		AD_denovo	7.8	het	de novo	1	NDD	(+) Hearing impairment,(+) Poor eye contact,(+) Global developmental delay,(+) Expressive language delay,(+) No social interaction,(+) Receptive language delay
FHIP2A	ENST00000369248:c.1619T>C p.Leu540Ser		AD_denovo	6.71	het	de novo	2	NDD	Macrocephaly, Autism, Hypotonia, Delayed gross motor development, Incoordination
NOC4L	ENST00000330579:c.901+2T>A		AD_denovo	6.45	het	de novo	2	NDD+epilepsy	Global developmental delay,(+) Infantile spasms,(+) Epileptic encephalopathy
ZNF433	ENST00000344980:c.1809_1812delinsACAG p.Arg604Gln		AD_denovo	3.51	mosaik	de novo	2	NDD+epilepsy	Global developmental delay,(+) Infantile spasms,(+) Epileptic encephalopathy
DSCAM			AD_unknown		het	paternal	2	NDD	Macrocephaly, Autism, Hypotonia, Delayed gross motor development, Incoordination
MFSD14A	ENST00000370152:c.1042A>G p.Ser348Gly		AD_denovo	6.01	het	de novo	2	NDD+epilepsy	Tall stature, Global developmental delay, Abnormal cerebellum morphology, Status epilepticus, Focal aware seizure, Scoliosis, Grade IV vesicoureteral reflux
FAAH2	ENST00000374900: c.1362T>A p.His454Gln		XL	4.76	hemi	maternal	2	NDD+epilepsy	Tall stature, Global developmental delay, Abnormal cerebellum morphology, Status epilepticus, Focal aware seizure, Scoliosis, Grade IV vesicoureteral reflux
PCDHGB2	ENST00000522605:c.1841C>T p.Pro614Leu		AD_denovo	4.92	het	de novo	2	NDD+epilepsy	Gliososis, EEG abnormality, Dyscalculia, Focal-onset seizure, Dyslexia, Impaired visuospatial constructive cognition, Mild global developmental delay
PFAS	ENST00000314666:c.1520A>G p.Lys507Arg	ENST00000314666:c.2122G>A p.Val708Ile	AR_comphet	5.61	comphet	maternal&paternal	2	NDD+epilepsy	Gliososis, EEG abnormality, Dyscalculia, Focal-onset seizure, Dyslexia, Impaired visuospatial constructive cognition, Mild global developmental delay
DSCAML1	ENST00000321322:c.4921T>C p.Ser1641Pro		AD_denovo	7.64	het	de novo	1	NDD	Delayed speech and language development, Global developmental delay, Obesity, Simple febrile seizure
RHOBTB3	ENST00000379982: c.520G>A p.Glu174Lys		AD_denovo	6.46	het	de novo	1	epilepsy	(+) Pectus excavatum,(+) Striae distensae,(+) Joint laxity,(+) EEG abnormality,(+) Focal-onset seizure,(-) Abnormality of brain morphology
EXOC4	ENST00000253861:c.1766A>T p.Asp589Val		AD_denovo	8.22	het	de novo	2	Muskel	Hypotonia,(+) Flexion contracture,(+) Gowers sign,(+) Exercise-induced myalgia
PTPRD			AD_unknown		het	de novo	2	Muskel	Hypotonia,(+) Flexion contracture,(+) Gowers sign,(+) Exercise-induced myalgia
FRY	ENST00000380217:c.104A>T p.His35Leu		AD_denovo	B	het	de novo	1	Fehlbildungen	Hypertelorism, Low-set, posteriorly rotated ears, Broad neck, Downslanted palpebral fissures, Tetralogy of Fallot, Short stature
SYTL5	ENST00000297875:c.2063G>A p.Gly688Glu		XL	4.32	hemi	maternal	4	NDD	Renal insufficiency,(+) High palate,(+) Microretrognathia,(+) Hearing impairment,(+) Astigmatism,(+) Myopia,(+) Exotropia,(+) Delayed speech and language development,(+) Single transverse palmar crease,(+) Intellectual disability,(+) Hip dysplasia,(+) Pes planus,(+) Thrombocytopenia,(+) Anemia,(+) Fever,(+) Vomiting,(+) Respiratory insufficiency,(+) Delayed gross motor development,(+) Hypocalcemia,(+) Hypoalbuminemia,(+) 2-3 toe syndactyly,(+) Pes valgus,(+) Delayed fine motor development,(+) Abnormal circulating carnitine concentration,(+) Severe global developmental delay,(+) Tetralogy of Fallot with pulmonary stenosis,(+) Submucous cleft of soft and hard palate

FRA10AC1	ENST00000359204:c.481C>T p.Arg161*		AR_homo	7.78	homo	maternal&paternal	4	NDD	Renal insufficiency,(+) High palate,(+) Microretrognathia,(+) Hearing impairment,(+) Astigmatism,(+) Myopia,(+) Exotropia,(+) Delayed speech and language development,(+) Single transverse palmar crease,(+) Intellectual disability,(+) Hip dysplasia,(+) Pes planus,(+) Thrombocytopenia,(+) Anemia,(+) Fever,(+) Vomiting,(+) Respiratory insufficiency,(+) Delayed gross motor development,(+) Hypocalcemia,(+) Hypoalbuminemia,(+) 2-3 toe syndactyly,(+) Pes valgus,(+) Delayed fine motor development,(+) Abnormal circulating carnitine concentration,(+) Severe global developmental delay,(+) Tetralogy of Fallot with pulmonary stenosis,(+) Submucous cleft of soft and hard palate
DMXL1	ENST00000311085:c.7691G>C p.Gly2564Ala		AR_homo	4.96	homo	maternal&paternal	4	NDD	Renal insufficiency,(+) High palate,(+) Microretrognathia,(+) Hearing impairment,(+) Astigmatism,(+) Myopia,(+) Exotropia,(+) Delayed speech and language development,(+) Single transverse palmar crease,(+) Intellectual disability,(+) Hip dysplasia,(+) Pes planus,(+) Thrombocytopenia,(+) Anemia,(+) Fever,(+) Vomiting,(+) Respiratory insufficiency,(+) Delayed gross motor development,(+) Hypocalcemia,(+) Hypoalbuminemia,(+) 2-3 toe syndactyly,(+) Pes valgus,(+) Delayed fine motor development,(+) Abnormal circulating carnitine concentration,(+) Severe global developmental delay,(+) Tetralogy of Fallot with pulmonary stenosis,(+) Submucous cleft of soft and hard palate
FAM149B1	ENST00000242505:c.485C>G p.Pro162Arg		AR_homo	4.28	homo	maternal&paternal	4	NDD	Renal insufficiency,(+) High palate,(+) Microretrognathia,(+) Hearing impairment,(+) Astigmatism,(+) Myopia,(+) Exotropia,(+) Delayed speech and language development,(+) Single transverse palmar crease,(+) Intellectual disability,(+) Hip dysplasia,(+) Pes planus,(+) Thrombocytopenia,(+) Anemia,(+) Fever,(+) Vomiting,(+) Respiratory insufficiency,(+) Delayed gross motor development,(+) Hypocalcemia,(+) Hypoalbuminemia,(+) 2-3 toe syndactyly,(+) Pes valgus,(+) Delayed fine motor development,(+) Abnormal circulating carnitine concentration,(+) Severe global developmental delay,(+) Tetralogy of Fallot with pulmonary stenosis,(+) Submucous cleft of soft and hard palate
PLSCR1	ENST00000342435:c.881T>C p.Ile294Thr		AR_homo	4.22	homo	maternal&paternal	4	NDD	Sensorineural hearing impairment,(+) Hypotonia,(+) Moderate global developmental delay
PDLIM3	ENST00000284767:c.113C>T p.Ala38Val		AR_homo	4.46	homo	maternal&paternal	4	NDD	Sensorineural hearing impairment,(+) Hypotonia,(+) Moderate global developmental delay
NAA35	ENST00000361671:c.659A>G p.Asp220Gly		AR_homo	4.95	homo	maternal&paternal	4	NDD	Sensorineural hearing impairment,(+) Hypotonia,(+) Moderate global developmental delay
HABP4	ENST00000375249:c.401G>A p.Arg134His		AR_homo	4.58	homo	maternal&paternal	4	NDD	Sensorineural hearing impairment,(+) Hypotonia,(+) Moderate global developmental delay
KIF7 (TICRR)	ENST00000394412:None None		AD_denovo	8.87	het	de novo	2	NDD+epilepsy	Generalized non-motor (absence) seizure,(+) Memory impairment,(+) Mild global developmental delay
FAT3	ENST00000298047:c.11381G>A p.Gly3794Glu	ENST00000298047:c.9731C>T p.Thr3244Met	AR_comphet	4.64	comphet	maternal&paternal	2	NDD+epilepsy	Generalized non-motor (absence) seizure,(+) Memory impairment,(+) Mild global developmental delay
ATP2B4	ENST00000341360:c.2708G>A p.Arg903His		AD_denovo	6.63	het	de novo	2	NDD+epilepsy	Focal-onset seizure,(+) Cortical tubers,(+) Mild global developmental delay,(+) Simple renal cyst
BIRC6	ENST00000421745:c.13G>A p.Gly5Ser	ENST00000421745:c.10525G>A p.Val3509Ile	AR_comphet	4.51	comphet	maternal&paternal	2	NDD+epilepsy	Focal-onset seizure,(+) Cortical tubers,(+) Mild global developmental delay,(+) Simple renal cyst
PGK2	ENST00000304801:c.1121G>A p.Gly374Glu		AD_denovo	5.77	het	de novo	1	Neuro	Motor axonal neuropathy
NRXN2	ENST00000265459:c.1579A>G p.Asn527Asp		AD_unknown	8.35	het	unknown	1	NDD	(+) Delayed speech and language development,(+) Intellectual disability, mild
LRRC8B	ENST00000330947:c.1070del p.Ser357Metfs*40		AD_inherited	6.72	het	maternal	1	NDD	(+) Downslanted palpebral fissures,(+) Aggressive behavior,(+) Global developmental delay,(+) Abnormal nasal morphology,(+) Attention deficit hyperactivity disorder,(+) Skewfoot,(+) Finger clinodactyly
DDX55	ENST00000238146:c.112G>A p.Ala38Thr		AD_denovo	6.08	het	de novo	2	epilepsy	Anxiety,(+) Bilateral tonic-clonic seizure,(+) Generalized non-motor (absence) seizure
SEZ6L	ENST00000248933:c.2681G>A p.Gly894Glu		AD_denovo	7.09	het	de novo	2	epilepsy	Anxiety,(+) Bilateral tonic-clonic seizure,(+) Generalized non-motor (absence) seizure
CSMD1			AD_unknown		het	de novo	1	NDD+epilepsy	Focal-onset seizure,(+) Mild global developmental delay

KLHL12	ENST00000367258:c.335T>G p.Val112Gly		AD_unknown	5.16	het	unknown	1	NDD	(+) Macrocephaly,(+) Seizure,(+) Hypotonia,(+) Dandy-Walker malformation,(+) Muscle weakness,(+) Distal lower limb amyotrophy,(+) Mild global developmental delay
KCND1	NM_004979.5:c.343G>Ap.(Asp115Asn)		AD_denovo	5.5	hemi	de novo	1	NDD + Epilepsy	Epilepsy with absences and eyelid myoclonias, normal cMRI, EEG abnormalities, IQ 85 (low normal), speech delay, obstipation
HDAC3	ENST00000305264:c.277G>C p.Asp93His		AD_denovo	8.86	het	de novo	3	NDD	Macrotia,(+) Hypotonia,(+) Moderate global developmental delay
SP9	NM_001145250.1:c.1133A>Gp.(Glu378Gly)		AD_denovo	5.5	het	de novo	1	NDD + Epilepsy	picanthus, Seizures, Global developmental delay, Abnormal facial shape, Generalized-onset seizure, Severe muscular hypotonia, Muscular hypotonia of the trunk, Infantile muscular hypotonia
ADAMTS15	ENST00000299164:c.2820G>C p.Gln940His		AD_denovo	5.22	het	de novo	3	NDD	Macrotia,(+) Hypotonia,(+) Moderate global developmental delay
CNTNAP4	ENST00000307431:c.94G>C p.Asp32His		AD_denovo	7.83	het	de novo	3	NDD	Macrotia,(+) Hypotonia,(+) Moderate global developmental delay
CPZ	ENST00000360986:c.602G>A p.Ser201Asn	ENST00000360986:c.752C>T p.Ala251Val	AR_comphet	4.23	comphet	maternal& paternal	2	NDD	Delayed speech and language development,(+) Motor delay,(+) Aplasia/Hypoplasia of the cerebellar vermis,(+) Severe global developmental delay
CSPG4	ENST00000308508:c.1390C>T p.Arg464Cys		AD_denovo	7.33	het	de novo	2	NDD	Delayed speech and language development,(+) Motor delay,(+) Aplasia/Hypoplasia of the cerebellar vermis,(+) Severe global developmental delay
KCNAB2	ENST00000378083:c.989del p.Gly330Alafs*8		AD_denovo	12.0	het	de novo	2	Epilepsy	febrile seizure
NOC4L	ENST00000330579:c.884C>A p.Thr295Asn		AD_denovo	5.71	het	de novo	2	Epilepsy	febrile seizure
MTA1	ENST00000331320:c.1427C>T p.Thr476Met		AD_unknown	5.3	het	unknown	1	NDD+epilepsy	(+) Generalized-onset seizure,(+) Moderate global developmental delay
ASTN1	ENST00000361833:c.797del p.Ser266Thrfs*32		AD_unknown	B	het	unknown	1	Neuro	(+) Headache,(+) Abnormal cerebral white matter morphology,(+) Paresthesia,(+) Abnormal central sensory function,(+) Hypoesthesia
REV3L	ENST00000358835:c.5617C>T p.Arg1873*		AD_unknown	8.79	het	unknown	1	NDD	(+) Microcephaly,(+) Generalized-onset seizure,(+) Short stature,(+) Intellectual disability, severe
CLHC1	ENST00000401408:c.1441A>T p.Thr481Ser	ENST00000401408:c.499G>A p.Gly167Ser	AR_comphet	B	comphet	maternal& paternal	1	Stoffwechsel	Childhood-onset truncal obesity
C8orf76 (ZHX1-C8orf76)	ENST00000276704:c.357+1G>T None		AR_homo	8.1	homo	unknown	1	NDD	(+) Intellectual disability, mild,(+) EEG abnormality
C10orf10 (DEPP1)	ENST00000298295:c.121G>T p.Val41Leu		AD_denovo	5.66	het	de novo	1	Epilepsy	Atypical absence seizure,(+) Central nervous system cyst
ENTPD6	ENST00000354989:c.747+1G>T None		AD_denovo	5.46	het	de novo	2	NDD	Global developmental delay,(+) Abnormal facial shape,(+) Poor speech,(+) Intellectual disability, severe
MAP3K15	ENST00000338883:c.1621C>T p.Gln541*		AD_denovo	4.6	het	de novo	2	NDD	Global developmental delay,(+) Abnormal facial shape,(+) Poor speech,(+) Intellectual disability, severe
CSMD3	ENST00000297405:c.7385G>A p.Arg2462Gln	ENST00000297405:c.10088A>C p.Gln3363Pro	AR_comphet	4.57	comphet	maternal& paternal	1	NDD+epilepsy	Global developmental delay,(+) EEG abnormality,(+) Hypsarrhythmia,(+) Generalized tonic seizure,(+) Epileptic spasm
PPP6R2	ENST00000216061:c.1602+1G>T None		AD_unknown	6.22	het	unknown	1	NDD	(+) Macrocephaly,(+) Hypotonia,(+) Mild global developmental delay
ENPP2	ENST00000075322:c.1388del p.Lys463Argfs*27		AD_denovo	6.64	het	de novo	3	Neuro	(+) Tall stature,(+) Hearing impairment,(+) Precocious puberty,(+) Joint swelling,(+) Ankle swelling,(+) Areflexia of lower limbs,(+) Sensory axonal neuropathy,(+) Limb muscle weakness,(+) Lower limb muscle weakness,(+) Lower limb pain,(+) Abnormality of movement,(+) Hyperesthesia
GALNT9	ENST00000328957:c.1144A>T p.Arg382Trp		AR_homo	5.32	homo	maternal& paternal	3	Neuro	(+) Tall stature,(+) Hearing impairment,(+) Precocious puberty,(+) Joint swelling,(+) Ankle swelling,(+) Areflexia of lower limbs,(+) Sensory axonal neuropathy,(+) Limb muscle weakness,(+) Lower limb muscle weakness,(+) Lower limb pain,(+) Abnormality of movement,(+) Hyperesthesia
CA10	ENST00000285273:c.287G>A p.Gly96Glu		AD_denovo	7.07	mosaik	de novo	3	Neuro	(+) Tall stature,(+) Hearing impairment,(+) Precocious puberty,(+) Joint swelling,(+) Ankle swelling,(+) Areflexia of lower limbs,(+) Sensory axonal neuropathy,(+) Limb muscle weakness,(+) Lower limb muscle weakness,(+) Lower limb pain,(+) Abnormality of movement,(+) Hyperesthesia

FMN1	ENST00000559047:c.1878dup p.Glu627*		AD_unknown	7.16	het	unknown	1	NDD	(+) Conductive hearing impairment,(+) Delayed speech and language development,(+) Focal-onset seizure
GIGYF2	ENST00000373563:c.713-1G>C None		AD_unknown	10.5	het	maternal	1	NDD	(+) Hearing impairment,(+) Visual impairment,(+) Depression,(+) Intellectual disability,(+) Toe clinodactyly,(+) Scoliosis,(+) Thyroid hyperplasia,(+) Crohn's disease
IRAK1	ENST00000369974:c.698del p.Asn233Thrfs*6		AD_unknown	C	het	maternal	1	immunodeficiency	(+) Recurrent bacterial infections,(+) Neonatal sepsis
ZFHX3	ENST00000268489:c.10129C>T p.Gln3377*		AD_unknown	7.14	het	unknown	1	NDD+epilepsy	(+) Long face,(+) Macrotia,(+) Autistic behavior,(+) Osteoporosis,(+) Intellectual disability,(+) Seizure,(+) Tremor,(+) Dysphagia,(+) Kyphosis,(+) Self-injurious behavior
MYRIP	ENST00000302541:c.86G>A p.Arg29His	ENST00000302541:c.383G>A p.Arg128His	AR_comphet	4.36	comphet	maternal&paternal	2	NDD	(+) Behavioral abnormality,(+) Global developmental delay,(+) Sleep disturbance,(+) Flat face
SLITRK2	ENST00000370490:c.2485G>T p.Glu829*		XL	8.04	hemi	maternal	2	NDD	(+) Behavioral abnormality,(+) Global developmental delay,(+) Sleep disturbance,(+) Flat face
MAP4K4	ENST00000302217:c.1042-3A>G None		AD_unknown	B	het	unknown	1	Neuro	(+) Dysarthria,(+) Cerebellar atrophy,(+) Gait disturbance,(+) Adenomatous colonic polyposis,(+) Kinetic tremor
CRYBG3	ENST00000182096:c.2648G>A p.Arg883His		AR_homo	4.41	homo	maternal&paternal	5	NDD	(+) Deeply set eye,(+) Intellectual disability,(+) Failure to thrive
EPHA1	ENST00000275815:c.2884G>C p.Gly962Arg		AR_homo	7.15	homo	maternal&paternal	5	NDD	(+) Deeply set eye,(+) Intellectual disability,(+) Failure to thrive
PDP2	ENST00000311765:c.629G>A p.Arg210His		AR_homo	5.39	homo	maternal&paternal	5	NDD	(+) Deeply set eye,(+) Intellectual disability,(+) Failure to thrive
KCNG4	ENST00000308251:c.1022C>T p.Ala341Val		AR_homo	5.57	homo	maternal&paternal	5	NDD	(+) Deeply set eye,(+) Intellectual disability,(+) Failure to thrive
GSE1	ENST00000253458:c.2468G>A p.Arg823Gln		AR_homo	4.41	homo	maternal&paternal	5	NDD	(+) Deeply set eye,(+) Intellectual disability,(+) Failure to thrive
THOC1	ENST00000261600:c.189dup p.Ile64Tyrfs*7		AD_unknown	7.49	het	unknown	1	NDD+epilepsy	(+) Intellectual disability,(+) Global developmental delay,(+) Absent speech,(+) Generalized-onset seizure,(+) Mutism,(+) Atypical absence seizure,(+) EEG with spike-wave complexes
TUBA1B	ENST00000336023:c.686G>A p.Arg229His		AD_denovo	7.68	het	de novo	1		(+) Brachyturriccephaly,(+) Microcephaly,(+) Penoscrotal hypospadias,(+) Cutis marmorata,(+) Global developmental delay,(+) Craniosynostosis,(+) Abnormal facial shape,(+) Short stature,(+) Feeding difficulties in infancy,(+) Nasogastric tube feeding in infancy
PBX3	ENST00000373483:c.649T>C p.Tyr217His		AD_unknown	5.15	het	unknown	2	NDD+epilepsy	(+) Seizure,(+) Intellectual disability, moderate,(+) Myoclonic seizure
ATG13	ENST00000312040:c.898C>T p.Gln300*		AD_unknown	7.23	het	unknown	2	NDD+epilepsy	(+) Seizure,(+) Intellectual disability, moderate,(+) Myoclonic seizure
DAAM1	ENST00000351081:c.692G>T p.Cys231Phe		AD_denovo	7.22	het	de novo	1	NDD	(+) Open mouth,(+) Thin upper lip vermillion,(+) Webbed neck,(+) Motor delay,(+) Muscle weakness,(+) Ventricular septal defect,(+) Pes planus,(+) Recurrent fever,(+) Pes valgus,(+) Axial hypotonia,(+) Speech articulation difficulties,(+) Short finger,(+) Tip-toe gait,(+) Abnormal tendon morphology
PBX2	ENST00000375050:c.391G>A p.Glu131Lys		AD_denovo	6.04	het	de novo	1	NDD	(+) Global developmental delay,(+) Pes planus,(+) Abnormal form of the vertebral bodies
WNK2	ENST00000297954:c.1693del p.Arg565Glyfs*11		AD_unknown	6.69	het	unknown	1	Epilepsy	(+) Generalized non-motor (absence) seizure // normal development, cMRI unremarkable
CIZ1	ENST00000277465:c.2009G>A p.Arg670His		AD_denovo	8.43	het	de novo	2	NDD+epilepsy	(+) Focal-onset seizure,(+) EEG with focal epileptiform discharges,(+) EEG with generalized epileptiform discharges,(+) Mild global developmental delay
KAT2A	ENST00000225916:c.1111A>C p.Asn371His		AD_unknown	6.62	het	unknown	2	NDD+epilepsy	(+) Seizure,(+) Complex febrile seizure,(+) Mild global developmental delay
UBE3C	ENST00000348165:c.2600A>G p.Tyr867Cys		AD_unknown	5.61	het	unknown	2	NDD+epilepsy	(+) Seizure,(+) Complex febrile seizure,(+) Mild global developmental delay
SIDT2	ENST00000278951:c.2122-2A>C None		AD_denovo	5.77	het	de novo	2	NDD+epilepsy	(+) Global developmental delay,(+) Developmental regression,(+) Aphasia,(+) Focal-onset seizure
B9D1 (MAPK7)	ENST00000477478:None None		AR_homo	8.56	homo	maternal&paternal	2	NDD+epilepsy	(+) Global developmental delay,(+) Developmental regression,(+) Aphasia,(+) Focal-onset seizure
PODXL	ENST00000322985:c.1216-3C>A None		AD_unknown	7	het	unknown	1	NDD+epilepsy	(+) Absent speech,(+) Spastic tetraplegia,(+) Scoliosis,(+) Intellectual disability, severe,(+) Severe global developmental delay,(+) Multifocal seizures
KIF3B	ENST00000375712:c.1481del p.Gln494Argfs*58		AD_unknown	B	het	unknown	1	Auge	(+) Cone/cone-rod dystrophy
GPR107	ENST00000347136:c.1316_1320del p.Asn439Serfs*21		AD_denovo	5.54	het	de novo	1	NDD+epilepsy	(+) Autism,(+) Seizure,(+) Leukodystrophy,(+) Mild global developmental delay
TCERG1	ENST00000296702:c.2273C>G p.Ser758*		AD_unknown	7.58	het	unknown	1		(+) Microcephaly,(+) Ptosis,(+) Psoriasiform dermatitis,(+) Short stature,(+) Epileptic encephalopathy
NEURL4	ENST00000315614:c.4107C>A p.Cys1369*		AD_unknown	7.12	het	unknown	1	NDD+epilepsy	(+) Intellectual disability,(+) Focal-onset seizure,(+) Mild global developmental delay

JMJD1C	ENST00000399251:c.168C>A p.Ser56Arg		AD_unknown	5.42	het	unknown	2	NDD+epilepsy	(+) Autism,(+) Intellectual disability,(+) Focal impaired awareness motor seizure
AKAP8L	ENST00000397410:c.310C>T p.His104Tyr		AD_denovo	5.23	het	de novo	1	epilepsy	(+) Status epilepticus,(+) Generalized-onset seizure,(+) Focal-onset seizure
AFG3L2	ENST00000269143:c.851G>T p.Gly284Val		AR_comphet	9.8	het	maternal&denovo_on_paternal_allele	1	NDD	(+) Renal duplication,(+) Retrognathia,(+) Abnormal pinna morphology,(+) Global developmental delay,(+) Intrauterine growth retardation,(+) Cerebral atrophy,(+) Increased serum lactate,(+) Global brain atrophy,(+) EEG abnormality,(+) Neuronal loss in central nervous system,(+) Elevated hepatic transaminase,(+) Infantile muscular hypotonia,(+) Cerebral white matter atrophy,(+) Elevated gamma-glutamyltransferase level,(+) Brain imaging abnormality
TUBA1B	ENST00000336023:c.878A>G p.Asn293Ser		AD_denovo	7.92	het	de novo	1	NDD+epilepsy	(+) Hypospadias,(+) Delayed speech and language development,(+) Global developmental delay,(+) Short stature,(+) Delayed fine motor development,(+) Complex febrile seizure,(+) Feeding difficulties
MEX3C	ENST00000406189:c.1810C>T p.Arg604*		AD_denovo	7.27	het	de novo	3	NDD	Behavioral abnormality,(+) Delayed speech and language development,(+) Umbilical hernia
TENM1	ENST00000371130:c.1795C>T p.Pro599Ser		XL	5.49	hemi	maternal	3	NDD	Behavioral abnormality,(+) Delayed speech and language development,(+) Umbilical hernia
BRCC3	ENST00000330045:c.209G>C p.Arg70Thr		XL	4.3	hemi	maternal	3	NDD	Behavioral abnormality,(+) Delayed speech and language development,(+) Umbilical hernia
YWHAZ	ENST00000353245:c.168_169insGTCATCTTGAGGGTCG p.Ser57Valfs*40		AD_denovo	11.4	het	de novo	2	NDD+epilepsy	(+) Intellectual disability, mild,(+) Generalized non-motor (absence) seizure,(+) Generalized-onset seizure,(+) Dyscalculia
DGKI	ENST00000288490:c.111C>A p.Cys37*		AD_unknown	6.2	het	unknown	2	NDD+epilepsy	(+) Intellectual disability, mild,(+) Generalized non-motor (absence) seizure,(+) Generalized-onset seizure,(+) Dyscalculia
CTDSP1	ENST00000443891:c.67+1del None		AD_unknown	6.88	het	unknown	1	NDD+epilepsy	(+) Migraine,(+) Focal-onset seizure,(+) Moderate global developmental delay
FURIN	ENST00000268171:c.482C>T p.Pro161Leu		AD_denovo	7.38	het	de novo	1	NDD	(+) Microcephaly,(+) Choanal stenosis,(+) Hand polydactyly,(+) Global developmental delay,(+) Failure to thrive,(+) Ventricular septal defect,(+) Patent ductus arteriosus,(+) Short stature,(+) Persistent left superior vena cava,(+) Feeding difficulties,(+) Dermoid cyst
TAAR2	ENST00000275191:c.545G>A p.Gly182Glu		AD_denovo	4.45	mosaik	de novo	1	NDD+epilepsy	Autism,(+) Seizure,(+) Global developmental delay
IAH1	ENST00000497473:c.1A>G p.Met1?		AR_homo	6.7	homo	unknown	1	NDD+epilepsy	(+) Intellectual disability, mild,(+) Bilateral tonic-clonic seizure,(+) Periventricular heterotopia,(+) Generalized-onset motor seizure
LIMK1	ENST00000336180:c.1291C>T p.Gln431*		AD_unknown	9.9	het	maternal	2	NDD+epilepsy	(+) Autism,(+) Intellectual disability,(+) Focal impaired awareness motor seizure
UNC79	ENST00000256339:c.1466T>A p.Met489Lys		AD_unknown	6.32	het	unknown	1		(+) Autism,(+) Intellectual disability, borderline,(+) Mild global developmental delay
ARID4A	ENST00000348476:c.469G>T p.Glu157*		AD_unknown	7.24	het	unknown	1	NDD	(+) Delayed speech and language development,(+) Pectus excavatum,(+) Motor delay,(+) Basal cell carcinoma,(+) Odontoma,(+) Bone cyst
USP34	ENST00000398571:c.6682-5T>G None		AD_unknown	8.01	het	unknown	1	NDD	(+) Microcephaly,(+) Behavioral abnormality,(+) Autism,(+) Intellectual disability,(+) Absent speech
UBQLN1	ENST00000257468:c.140del p.Lys47Argfs*32		AD_unknown	8.06	het	unknown	1		(+) Generalized non-motor (absence) seizure,(+) Visually-induced seizure
ST6GAL1	ENST00000169298:c.1193T>C p.Leu398Pro		AD_denovo	5.82	het	de novo	2	NDD	(+) Dilated cardiomyopathy,(+) Neurodegeneration,(+) Muscle spasm,(+) Muscular dystrophy,(+) Short stature,(+) Infantile muscular hypotonia,(+) Severe global developmental delay
KIAA1024 (MINAR1)	ENST00000305428:c.2043G>T p.Trp681Cys		AD_denovo	4.44	het	de novo	2	NDD	(+) Dilated cardiomyopathy,(+) Neurodegeneration,(+) Muscle spasm,(+) Muscular dystrophy,(+) Short stature,(+) Infantile muscular hypotonia,(+) Severe global developmental delay
HECTD1	ENST00000399332:c.7789C>T p.Arg2597Cys		AD_denovo	8.04	het	de novo	1	NDD	(+) Generalized hypotonia,(+) Mild global developmental delay
SLC5A3	ENST00000381151:c.728del p.Pro243Leufs*6		AD_unknown	7.66	het	unknown	1	NDD	Intellectual disability, moderate,(+) Focal impaired awareness seizure,(+) Increased body weight,(+) Bilateral tonic-clonic seizure with focal onset,(+) Focal-onset seizure
CEP89	ENST00000305768:c.304C>T p.Arg102Trp		AR_homo	6.38	homo	maternal&paternal	2	NDD+epilepsy	(+) Intellectual disability,(+) Ataxia,(+) Gait disturbance,(+) Bilateral tonic-clonic seizure with focal onset,(+) Focal impaired awareness motor seizure
KCNAB1	ENST00000302490:c.1063C>A p.Leu355Ile		AD_denovo	7.14	het	de novo	1	Fehlbildung	(+) Oligohydramnios,(+) Abnormal heart morphology,(+) Morphological central nervous system abnormality,(+) Abnormality of bladder morphology
C20orf194 (DNAAF9)	ENST00000252032:c.1679-2A>C None	ENST00000252032:c.1960_1961del p.Ser654Argfs*64	AR_comphet	8.05	comphet	maternal&paternal	1	NDD+epilepsy	(+) Intellectual disability,(+) Global developmental delay,(+) Periventricular heterotopia,(+) Generalized myoclonic-atonic seizure,(+) Bilateral tonic-clonic seizure with generalized onset,(+) Tonic seizure,(+) Myoclonic seizure
ROCK1	ENST00000399799:c.2489+3A>G None		AD_unknown	8.04	het	unknown	2		(+) Insulin resistance,(+) Obesity,(+) Hypertriglyceridemia,(+) Hypercholesterolemia
UBR4	ENST00000375217:c.7720del p.Val2574*		AD_unknown	9.9	het	unknown	3	NDD	(+) Nevus flammeus,(+) Hypotonia,(+) Global developmental delay,(+) Abnormality of the voice,(+) Nasal speech,(+) Curly hair,(+) Depressed nasal bridge,(+) Prominent forehead

SCAF4	ENST00000286835:c.1738G>C p.Ala580Pro		AD_unknown	4.51	het	unknown	3	NDD	(+) Nevus flammeus,(+) Hypotonia,(+) Global developmental delay,(+) Abnormality of the voice,(+) Nasal speech,(+) Curly hair,(+) Depressed nasal bridge,(+) Prominent forehead
HCN2	ENST00000251287:c.962G>A p.Arg321His		AD_unknown	7.12	het	unknown	3	NDD	(+) Nevus flammeus,(+) Hypotonia,(+) Global developmental delay,(+) Abnormality of the voice,(+) Nasal speech,(+) Curly hair,(+) Depressed nasal bridge,(+) Prominent forehead
RABEP1	ENST00000262477:c.2432T>G p.Leu811*		AD_unknown	6.6	het	maternal	1	Epilepsy	(+) Seizure,(+) Bilateral tonic-clonic seizure,(+) EEG with spike-wave complexes
ATP5B (ATP5F1B)	ENST00000262030:c.841C>T p.Arg281Trp		AD_unknown	6.35	het	unknown	1	NDD+epilepsy	(+) Cleft palate,(+) Hydrocephalus,(+) Macrocephaly,(+) Hypertelorism,(+) Broad forehead,(+) High forehead,(+) Wide nose,(+) Behavioral abnormality,(+) Abnormal corpus callosum morphology,(+) Pes planus,(+) Intellectual disability, moderate,(+) Focal-onset seizure,(+) Proximal placement of thumb,(+) Absent uvula,(+) Moderate global developmental delay,(+) Cleft lip
KDM5A	whole gene deletion as part of a bigger deletion seq[GRCh37]12p13.33p13.33(p*,146240_1026036x1) involved genes: IQSEC3, SLC6A12, SLC6A13, KDM5A, CCDC77, B4GALNT3, NINJ2, WNK1, RAD52		AD_unknown	C	het	unknown	2	metabolic disorder	(+) Insulin resistance,(+) extreme Obesity,(+) Hypertriglyceridemia,(+) Hypercholesterolemia
XPO1	ENST00000401558:c.2293C>T p.Arg765*		AD_denovo	8.48	het	de novo	1	NDD	(+) Microcephaly,(+) Attention deficit hyperactivity disorder,(+) Mild global developmental delay
AEBP2	ENST00000266508:c.537del p.Ser179Argfs*2		AD_unknown	7.23	het	unknown	1	NDD	(+) Intellectual disability,(+) Global developmental delay,(+) Poor speech
REXO1	ENST00000170168:c.2230+5G>A None		AD_unknown	7	het	unknown	1	NDD+epilepsy	(+) Intellectual disability, mild,(+) Progressive gait ataxia,(+) Multifocal seizures,(+) Focal cortical dysplasia
HIRA	ENST00000263208:c.1048G>A p.Gly350Ser		ad_inherited	8.03	het	maternal	1	NDD) Astigmatism,(+) Strabismus,(+) Visual impairment,(+) Aggressive behavior,(+) Global developmental delay
INO80	ENST00000361937:c.1015C>T p.Leu339Phe		AD_unknown	5.66	het	unknown	2	NDD	(+) Strabismus,(+) Behavioral abnormality,(+) Global developmental delay,(+) Polyhydramnios,(+) Nephroblastoma,(+) Intellectual disability, borderline,(+) Feeding difficulties
KLHL29	ENST00000486442:c.345G>A p.Trp115*		AD_unknown	5.8	het	unknown	2	NDD	(+) Strabismus,(+) Behavioral abnormality,(+) Global developmental delay,(+) Polyhydramnios,(+) Nephroblastoma,(+) Intellectual disability, borderline,(+) Feeding difficulties
NAV3	ENST00000228327:c.881-2A>T None		AD_unknown	7.27	het	unknown	1	NDD+epilepsy	(+) Global developmental delay,(+) Polymicrogyria,(+) Short stature,(+) Bilateral tonic-clonic seizure with focal onset,(+) Intellectual disability, severe,(+) Generalized myoclonic-atonic seizure,(+) Focal atonic seizure
FEM1B	ENST00000306917:c.1164del p.Phe388Leufs*8		AD_unknown	5.84	het	unknown	2	NDD	(+) Hearing impairment,(+) Hypermetropia,(+) Optic atrophy,(+) Delayed speech and language development,(+) Spastic tetraparesis,(+) Dandy-Walker malformation,(+) Progressive flexion contractures,(+) Severe global developmental delay
ICE1	ENST00000296564:c.6547G>T p.Glu2183*		AD_unknown	6.79	het	unknown	2	NDD	(+) Hearing impairment,(+) Hypermetropia,(+) Optic atrophy,(+) Delayed speech and language development,(+) Spastic tetraparesis,(+) Dandy-Walker malformation,(+) Progressive flexion contractures,(+) Severe global developmental delay
GIT1	ENST00000225394:c.1753-2A>G None		AD_unknown	10.4	het	unknown	1	NDD	(+) Microcephaly,(+) Autism,(+) Ataxia,(+) Severe global developmental delay,(+) Head-banging,(+) Neurodevelopmental delay
NRXN2	ENST00000265459:c.1654G>C p.Asp552His		AD_unknown	8.8	het	unknown	1		(+) Hearing abnormality,(+) Mild global developmental delay
UNC119B	ENST00000344651:c.463G>A p.Gly155Arg		AD_denovo	5.97	het	de novo	1	NDD	(+) Microcephaly,(+) Autism,(+) Delayed speech and language development
CDH10	ENST00000264463:c.2263A>G p.Thr755Ala		AD_unknown	6.53	het	unknown		NDD+epilepsy	(+) Autism,(+) Intellectual disability,(+) Hypotonia,(+) Gait ataxia,(+) Generalized non-motor (absence) seizure,(+) Bilateral tonic-clonic seizure with generalized onset
NF1 (EVI2A)	ENST00000356175:c.4773-7210A>G None		ad_inherited	7.96	het	paternal	2	NDD+epilepsy	(+) Focal-onset seizure,(+) Mild global developmental delay
PTPRD	ENST00000355233:c.3906C>G p.Tyr1302*		AD_unknown	9.19	het	unknown	1	NDD+epilepsy	(+) Obsessive-compulsive behavior,(+) Seizure,(+) Global developmental delay,(+) Generalized-onset seizure
PAPOLG	ENST00000238714:c.1680-1G>C None		AD_unknown	7.64	het	unknown	1	NDD+epilepsy	(+) Microcephaly,(+) Focal-onset seizure,(+) Severe global developmental delay
ACSS3	ENST00000261206:c.1124del p.Gly375Alafs*12		ad_inherited	5.62	het	maternal	3	Epilepsy	(+) Myoclonus,(+) Bilateral tonic-clonic seizure with generalized onset
MAPK8IP1	ENST00000241014:c.112G>A p.Asp38Asn		AD_denovo	7.17	het	de novo	3	Epilepsy	(+) Myoclonus,(+) Bilateral tonic-clonic seizure with generalized onset
AGAP3	ENST00000335367:c.528_536del p.Glu177_Pro179del		AD_denovo	5.1	het	de novo	3	Epilepsy	(+) Myoclonus,(+) Bilateral tonic-clonic seizure with generalized onset
YLPM1	ENST00000325680:c.5702A>G p.Tyr1901Cys		AD_denovo	6.51	het	de novo	1	Epilepsy	(+) Myoclonus,(+) Bilateral tonic-clonic seizure with generalized onset
SMARCC1	ENST00000254480:c.1297C>T p.Arg433*		AD_unknown		het	unknown	1	NDD	(+) Long eyelashes,(+) Synophrys,(+) Motor delay,(+) Coarctation of aorta,(+) Abnormal facial shape
MAP2K6	ENST00000589647:c.620C>T p.Pro207Leu		AD_denovo	6.91	het	de novo	1	NDD	(+) Macrocephaly,(+) Motor delay,(+) Moderate global developmental delay
GRIK4	ENST00000438375:c.-51+819C>A None		AD_denovo	6.79	het	de novo	1	NDD	(+) Microcephaly,(+) Intellectual disability,(+) Global developmental delay

ZFR	ENST00000265069:c.2740-3A>G None		AD_denovo	5.87	het	de novo	1	NDD	(+) Autism,(+) Motor stereotypy,(+) Intellectual disability,(+) Severe global developmental delay
DUOX1	ENST00000321429:c.2044C>T p.Gln682*		AD_unknown	5.04	het	unknown	1	NDD+epilepsy	(+) Autism,(+) Seizure,(+) Absent speech,(+) Neurodevelopmental delay
INPP4B	ENST00000262992:c.2603A>G p.His868Arg		de_novo	5.19	het	de novo	1	NDD	(+) Intellectual disability, mild,(+) Failure to thrive,(+) Growth delay,(+) Moderate global developmental delay
SNAP91	ENST00000195649:c.1391dup p.Thr465Asnfs*8		AD_unknown	10.0	het	unknown	1	NDD	(+) Tall stature,(+) Macrocephaly,(+) Restlessness,(+) Overgrowth,(+) Expressive language delay,(+) Neurodevelopmental delay
ACTR1B	ENST00000289228:c.942dup p.Leu315Thrfs*3		de_novo	6.34	het	de novo	1	NDD+epilepsy	Intellectual disability,(+) Seizure
GRIK5	ENST00000262895:c.1415dup p.Tyr472*		unknown	8.52	het	unknown	1		(+) Abnormal cerebral white matter morphology,(+) Abnormal myelination,(+) Tonic seizure,(+) Myoclonic seizure
MYCBP2	ENST00000357337:c.2858G>A p.Gly953Asp		unknown	6.21	het	unknown	1	NDD	(+) Autism,(+) Autistic behavior,(+) Global developmental delay,(+) Dissociative reaction
HK2	ENST00000290573:c.1202A>G p.Lys401Arg		homo	5.57	homo	maternal&paternal	2	NDD+epilepsy	(+) Intellectual disability,(+) Ataxia,(+) Gait disturbance,(+) Bilateral tonic-clonic seizure with focal onset,(+) Focal impaired awareness motor seizure
ARFGEF1	ENST00000262215:c.4036C>T p.Leu1346Phe		unknown	6.55	het	unknown		NDD+epilepsy	(+) Intellectual disability, mild,(+) Bilateral tonic-clonic seizure with focal onset,(+) Focal motor seizure
ABCB5	ENST00000404938:c.388A>G p.Ile130Val	ENST00000404938:c.1533-2A>G None	comphet	4.2	comphet	maternal&paternal		NDD+epilepsy	(+) Intellectual disability,(+) Generalized-onset seizure,(+) Focal-onset seizure
WIPI1	ENST00000262139:c.444_447del p.Ile150Thrfs*13		de_novo	6.07	het	de novo	3	NDD+epilepsy	(+) Global developmental delay,(+) Frontal bossing,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Chiari type I malformation,(+) Increased head circumference
VPS4B	ENST00000238497:c.572C>T p.Ala191Val		de_novo	5.97	het	de novo	3	NDD+epilepsy	(+) Global developmental delay,(+) Frontal bossing,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Chiari type I malformation,(+) Increased head circumference
HOOK3	ENST00000307602:c.1391+2T>A None		ad_inherited	7.77	het	maternal	3	NDD+epilepsy	(+) Global developmental delay,(+) Frontal bossing,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Chiari type I malformation,(+) Increased head circumference
DCLK2	ENST00000296550:c.1925C>T p.Ala642Val		unknown	6.53	het	unknown	2	NDD	(+) Personality changes,(+) Asthma,(+) Tetraparesis,(+) Sleep disturbance,(+) Falls,(+) Metachromatic leukodystrophy variant,(+) Poor fine motor coordination,(+) Chronic pain,(+) Short term memory impairment,(+) Erectile dysfunction
BAI2 (ADGRB2)	ENST00000257070:c.4470+1G>A None		unknown	7.65	het	unknown	2	NDD	(+) Personality changes,(+) Asthma,(+) Tetraparesis,(+) Sleep disturbance,(+) Falls,(+) Metachromatic leukodystrophy variant,(+) Poor fine motor coordination,(+) Chronic pain,(+) Short term memory impairment,(+) Erectile dysfunction
ICE1	ENST00000296564:c.310-2del None		unknown	7.1	het	unknown	1	epilepsy	(+) Generalized-onset seizure,(+) Focal-onset seizure,(+) Visually-induced seizure
TUBE1	ENST00000368662:c.290A>G p.Gln97Arg		homo	5.19	homo	maternal&paternal	3	NDD	(+) Microcephaly,(+) Aggressive behavior,(+) Spasticity,(+) Intellectual disability, severe,(+) Crohn's disease,(+) Self-injurious behavior
GNB1L	ENST00000329517:c.53G>A p.Gly18Asp		homo	5.37	homo	maternal&paternal	3	NDD	(+) Microcephaly,(+) Aggressive behavior,(+) Spasticity,(+) Intellectual disability, severe,(+) Crohn's disease,(+) Self-injurious behavior
LRP1B	ENST00000389484:c.3802C>T p.His1268Tyr		unknown	5.09	het	unknown	1	NDD	(+) Autistic behavior,(+) Intellectual disability,(+) Peripheral axonal neuropathy,(+) Sensorimotor neuropathy,(+) Focal-onset seizure,(+) Moderate global developmental delay
NKTR	ENST00000232978:c.3174_3175del p.Lys1060Ilefs*3		unknown	8.2	het	unknown	1	NDD+epilepsy	(+) Generalized-onset seizure,(+) Mild global developmental delay
MOV10	ENST00000357443:c.1787G>A p.Cys596Tyr		unknown	7.02	het	unknown	1	NDD+epilepsy	(+) Intellectual disability, mild,(+) Focal-onset seizure
SEL1L	ENST00000336735:c.108+1G>A None		unknown	7.16	het	unknown	1	NDD	(+) Hypotonia,(+) Delayed gross motor development,(+) Myopathy,(+) Peripheral neuropathy
ANKRD12	ENST00000262126:c.4760C>G p.Ser1587*		unknown	6.06	het	unknown	1	NDD	(+) Hypotonia,(+) Delayed gross motor development,(+) Myopathy,(+) Peripheral neuropathy
GRIK5	ENST00000262895:c.1171T>C p.Trp391Arg	ENST00000296452:c.9446C>T p.Ser3149Leu	unknown	B	het	unknown	1	NDD+epilepsy	(+) Delayed gross motor development,(+) Lower limb amyotrophy,(+) Infantile axial hypotonia

CSMD1	ENST00000537824:c.6598G>A p.Gly2200Ser	ENST00000537824:c.8697C>A p.Phe2899Leu	comphet	6.36	comphet	maternal&paternal	2	NDD+epilepsy	(+) Delayed speech and language development,(+) Generalized non-motor (absence) seizure
BSN	ENST00000296452:c.8648C>T p.Ala2883Val	ENST00000296452:c.9446C>T p.Ser3149Leu	comphet	6.73	comphet	maternal&paternal	2	NDD+epilepsy	(+) Delayed speech and language development,(+) Generalized non-motor (absence) seizure
PIGC	ENST00000258324:c.674C>G p.Pro225Arg		homo	8.19	homo	maternal&paternal	3	NDD	(+) Autism,(+) Attention deficit hyperactivity disorder,(+) severe global developmental delay
APC2	ENST00000233607:c.1519dup p.Gln507Profs*109		AD_unknown	10.3	het	unknown	1	NDD	(+) Autistic behavior,(+) Severe global developmental delay
CPSF2	ENST00000298875:c.460A>G p.Met154Val		unknown	5.06	het	unknown	1		(+) Autism,(+) Prominent fingertip pads,(+) Moderate global developmental delay
EIF4B	ENST00000262056:c.1306+1G>C None		unknown	7.7	het	unknown	1	NDD	(+) Abnormality of the face,(+) Global developmental delay
SEPT7	ENST00000350320:c.1271+1G>T None		de_novo	A	het	de novo	1	other	(+) Polycystic kidney dysplasia,(+) Hydronephrosis,(+) Agenesis of corpus callosum,(+) Ventriculomegaly,(+) Aplasia/Hypoplasia of the bladder
GMPPA (SPEGNB)	ENST00000313597:None None		homo	6.27	homo	maternal&paternal		NDD	(+) Hearing impairment,(+) Strabismus,(+) Hypermetropia,(+) Delayed speech and language development,(+) Intellectual disability,(+) Global developmental delay
HDAC7	ENST00000080059:c.184C>T p.Gln62*		unknown	6.74	het	unknown	1	NDD	(+) Hearing impairment,(+) Intellectual disability, moderate,(+) Moderate global developmental delay
MSI1	ENST00000257552:c.345dup p.Leu116Alafs*15		unknown	8.19	het	unknown	1	NDD	(+) Generalized-onset seizure,(+) Epileptic spasm,(+) Severe global developmental delay
SUSD4	ENST00000343846:c.662A>C p.Tyr221Ser		de_novo	4.93	het	de novo	1	NDD+epilepsy	(+) Hydrocephalus,(+) Strabismus,(+) Premature birth,(+) Double outlet right ventricle,(+) Esophageal atresia,(+) Generalized-onset seizure,(+) Meckel diverticulum,(+) Laryngeal cleft,(+) Prominent forehead,(+) Cerebral infarct
KIAA0100 (BLTP2)	ENST00000528896:c.5465G>A p.Arg1822Gln		de_novo	6.29	het	de novo	1	NDD+epilepsy	(+) Epicanthus,(+) Wide nasal bridge,(+) Delayed speech and language development,(+) Hypotonia,(+) Abnormal heart valve morphology,(+) Status epilepticus,(+) Leukodystrophy,(+) Focal-onset seizure,(+) Moderate global developmental delay,(+) Abnormal myelination,(+) Happy demeanor
EFNA5	ENST00000333274:c.125+56499T>C None		de_novo	5.4	het	de novo	1	NDD+epilepsy	(+) Generalized-onset seizure,(+) Focal-onset seizure,(+) Moderate global developmental delay
CHD9	ENST00000398510:c.4257T>G p.Asp1419Glu		unknown	5.4	het	unknown	1	NDD	(+) Myopia,(+) Behavioral abnormality,(+) Pectus excavatum,(+) Ataxia,(+) Intellectual disability, moderate
ABCE1	ENST00000296577:c.688G>C p.Val230Leu		unknown	5.47	het	unknown	2	Epilepsy+other	(+) Glaucoma,(+) Hypertension,(+) Rheumatoid arthritis,(+) Postural instability,(+) Abnormal rapid eye movement sleep,(+) Achalasia,(+) Scoliosis,(+) Hyposmia,(+) Epileptic spasm,(+) Chronic pain
MED1	ENST00000300651:c.2927A>T p.Asn976Ile		unknown	5.03	het	unknown	2	Epilepsy+other	(+) Glaucoma,(+) Hypertension,(+) Rheumatoid arthritis,(+) Postural instability,(+) Abnormal rapid eye movement sleep,(+) Achalasia,(+) Scoliosis,(+) Hyposmia,(+) Epileptic spasm,(+) Chronic pain
GTPBP1	ENST00000216044:c.1811del p.Glu604Glyfs*10		unknown	6.53	het	unknown	1	Epilepsy	(+) Generalized-onset seizure,(+) Focal-onset seizure
HMBX1	ENST00000287701:c.812G>A p.Arg271Gln		unknown	3.43	het	unknown		NDD+obesity	(+) Obesity,(+) Mild global developmental delay
TSPO (TTLL12)	ENST00000329563:None None		homo	5.49	homo	maternal&paternal	1	NDD+epilepsy	(+) Global developmental delay,(+) Complex febrile seizure
RAPGEF2	ENST00000264431:c.4475-8A>G None		de_novo	5.29	het	de novo	1	Ataxia+other	(+) Dysarthria,(+) Dysmetria,(+) Achilles tendon contracture,(+) Gait ataxia,(+) Intention tremor,(+) Poor fine motor coordination,(+) Abnormal pyramidal sign
ZNF219	ENST00000360947:c.745G>T p.Glu249*		unknown	5.58	het	unknown	1	NDD+epilepsy	(+) Dysphagia,(+) Generalized-onset seizure,(+) Developmental regression,(+) Severe global developmental delay
RABEP1	ENST00000262477:c.97C>T p.Gln33*		unknown	6.83	het	maternal	1	NDD	(+) Hypotonia,(+) Moderate global developmental delay
RFX2	ENST00000303657:c.446_447insGCTGG p.Ala150Leufs*66		unknown	7.56	het	unknown	1	NDD+epilepsy	(+) Intellectual disability,(+) Focal-onset seizure,(+) Moderate global developmental delay
PLXNA2	ENST00000367033:c.3548T>A p.Val1183Glu		unknown	7.33	het	unknown	1	NDD+epilepsy	(+) Seizure,(+) Spasticity,(+) Rigidity,(+) Intellectual disability, severe
SHANK1	ENST00000293441:c.6399_6403del p.Lys2134Glyfs*47		de_novo	12.9	het	de novo		NDD	Global developmental delay, severe expressive language delay, EEG and cMRT unremarkable
CCAR1	ENST00000265872:c.3187+2T>G None		unknown	7.56	het	unknown	1	NDD+epilepsy	(+) Intellectual disability, mild,(+) Focal-onset seizure,(+) Mild global developmental delay

THSD7A	ENST00000423059:c.1016del p.Asp339Valfs*2		unknown	6.59	het	unknown	1	NDD+epilepsy	(+) Behavioral abnormality,(+) Intellectual disability, mild,(+) Dysarthria,(+) Thick cerebral cortex,(+) Thick pachygyria,(+) Focal atonic seizure,(+) Bilateral perisylvian polymicrogyria
SF3A3	ENST00000373019:c.1005+1G>T None		de_novo	9.29	het	de novo	1	NDD	(+) Tall stature,(+) High palate,(+) Macrocephaly,(+) Abnormal earlobe morphology,(+) Autism,(+) Motor stereotypy,(+) Delayed speech and language development,(+) Large earlobe
NELFA	ENST00000382882:c.1335+3A>G None		unknown	8.82	het	unknown	1	NDD+epilepsy	(+) Urinary incontinence,(+) Microcephaly,(+) Behavioral abnormality,(+) Autism,(+) Autistic behavior,(+) Seizure,(+) Absent speech,(+) Hallux valgus,(+) Bilateral tonic-clonic seizure,(+) Intellectual disability, moderate,(+) Short stature,(+) Abnormality of the Achilles tendon,(+) Severe global developmental delay
DSCAML1	ENST00000321322:c.3914C>G p.Pro1305Arg		unknown	5.84	het	unknown	1	NDD	(+) Autism,(+) Moderate global developmental delay
NRXN3	ENST00000335750:c.904T>G p.Trp302Gly		unknown	7.56	het	unknown	1	NDD+epilepsy	(+) Seizure,(+) Hypotonia,(+) Generalized myoclonic seizure,(+) Focal hyperkinetic seizure,(+) Mild global developmental delay,(+) Moderate global developmental delay,(+) Startle-induced seizure,(+) Myoclonic seizure
CYP3A43	ENST00000222382:c.334G>A p.Gly112Arg		de_novo	5	het	de novo	1	NDD+epilepsy	(+) Seizure,(+) Global developmental delay,(+) Epileptic encephalopathy
PHLPP1	ENST00000262719:c.80_84del p.Ala27Glyfs*234		unknown	7.12	het	unknown	1	Epilepsy	(+) Seizure,(+) Continuous spike and waves during slow sleep
LMAN2L	ENST00000264963:c.425-331G>T None		homo	6	homo	maternal/paternal	1	other	(+) Cleft palate,(+) Pierre-Robin sequence,(+) Abnormality of the face,(+) Retrognathia,(+) Large hands,(+) Hypotonia,(+) Premature birth,(+) Long foot,(+) Neonatal asphyxia,(+) Abnormal ear morphology
DPYSL2	ENST00000311151:c.105A>G p.Ile35Met		unknown	7.07	het	unknown	1	NDD	(+) Short neck,(+) Strabismus,(+) Hypertrichosis,(+) Brachydactyly,(+) Motor delay,(+) Obesity,(+) Talipes equinovarus,(+) Supernumerary nipple,(+) Moderate global developmental delay,(+) Toe walking
BIRC6	ENST00000421745:c.4189G>T p.Gly1397*		unknown	7.13	het	unknown	1	Epilepsy	(+) Bilateral tonic-clonic seizure
KCNAB1	ENST00000302490:c.1067T>G p.Leu356Arg		unknown	5.72	het	unknown	1	NDD+epilepsy	(+) Seizure,(+) Intellectual disability, mild,(+) Spasticity
TOMM70A	ENST00000284320:c.324+2T>G None		unknown	7.59	het	unknown	1	Epilepsy	(+) Delayed speech and language development,(+) Seizure
PPP2R1A	ENST00000322088:c.1669C>T p.Gln557*		de_novo	11.3	het	de novo	1	NDD+epilepsy	Global developmental delay, Infantile spasms, Vertical nystagmus
BAZ1B	ENST00000339594:c.728G>A p.Arg243His		unknown	8.28	het	unknown		Epilepsy	(+) Seizure,(+) Focal-onset seizure
NLE1	ENST00000360831:c.899del p.Glu300Glyfs*27		de_novo	5.98	het	de novo	1	other	(+) Microcephaly,(+) Bilateral talipes equinovarus,(+) Peripheral neuropathy
EP400	ENST00000330386:c.3126+1G>C None		unknown	8.34	het	maternal	2		(+) Intellectual disability, mild,(+) Generalized-onset seizure
NRXN2	ENST00000265459:c.1201A>T p.Thr401Ser		unknown	8.62	het	unknown		NDD/ID	(+) Thin upper lip vermillion,(+) Posteriorly rotated ears,(+) Behavioral abnormality,(+) Delayed speech and language development,(+) Intellectual disability,(+) Moderate global developmental delay,(+) Encopresis
PTPRD	ENST00000355233:c.4259_4262dup p.His1422Serfs*3		unknown	9.64	het	unknown		Epilepsy	(+) Delayed speech and language development,(+) Seizure,(+) Generalized-onset seizure,(+) EEG abnormality,(+) Febrile seizure (within the age range of 3 months to 6 years)
PCSK5	ENST00000376752:c.2240G>A p.Cys747Tyr		unknown	5.65	het	unknown	1	NDD	(+) Motor delay,(+) Periventricular leukomalacia,(+) Motor regression
BIRC6	ENST00000421745:c.9344C>A p.Thr3115Lys		unknown	5.29	het	unknown	3		(+) Hydrocephalus,(+) Nystagmus,(+) Autism,(+) Aggressive behavior,(+) Seizure,(+) Ventriculomegaly,(+) Intracranial hemorrhage,(+) Scoliosis,(+) Coxa valga,(+) Intellectual disability, severe,(+) Abnormality of brain morphology
GRM2	ENST00000395052:c.941C>T p.Ala314Val		unknown	6.06	het	unknown	3		(+) Hydrocephalus,(+) Nystagmus,(+) Autism,(+) Aggressive behavior,(+) Seizure,(+) Ventriculomegaly,(+) Intracranial hemorrhage,(+) Scoliosis,(+) Coxa valga,(+) Intellectual disability, severe,(+) Abnormality of brain morphology
OLFM1	ENST00000252854:c.191A>C p.Gln64Pro		unknown	5.35	het	unknown	3		(+) Hydrocephalus,(+) Nystagmus,(+) Autism,(+) Aggressive behavior,(+) Seizure,(+) Ventriculomegaly,(+) Intracranial hemorrhage,(+) Scoliosis,(+) Coxa valga,(+) Intellectual disability, severe,(+) Abnormality of brain morphology
NCOR1	ENST00000580554:c.458+4A>T None		unknown	8.83	het	maternal	2		(+) Intellectual disability, mild,(+) Generalized-onset seizure
TOPBP1	ENST00000260810:c.3870A>G p.Leu1290=		unknown	7.98	het	unknown		NDD+epilepsy	(+) Autism,(+) Seizure,(+) Bilateral tonic-clonic seizure,(+) EEG abnormality,(+) Focal impaired awareness seizure,(+) Focal-onset seizure,(+) Moderate global developmental delay,(+) Autonomic epileptic aura
ATP13A1	ENST00000291503:c.1872G>T p.Arg624=		unknown	8.13	het	unknown		NDD+Autism	(+) Macrocephaly,(+) Autistic behavior,(+) Delayed speech and language development,(+) Obesity,(+) Expressive language delay,(+) Abnormal social behavior,(+) Abnormal eating behavior
SHB	ENST00000377707:c.903C>A p.Tyr301*		unknown	6.44	het	unknown		NDD+Autism	(+) Macrocephaly,(+) Autistic behavior,(+) Delayed speech and language development,(+) Obesity,(+) Expressive language delay,(+) Abnormal social behavior,(+) Abnormal eating behavior
NOP2	ENST00000322166:c.104-4A>G None		unknown	7.99	het	unknown	1	NDD	(+) Tall stature,(+) Macrocephaly,(+) Atypical behavior,(+) Autistic behavior,(+) Delayed speech and language development,(+) Obesity
SPATA21	ENST00000335496:c.974C>G p.Pro325Arg		de_novo	4.25	het	de novo	1	Neurofibromatosis	(+) Cafe-au-lait spot,(+) Neurofibromas,(+) Allergy

ARHGAP30	ENST00000368013:c.2276_2277del p.Glu759Glyfs*21		unknown	6.62	het	unknown		NDD+epilepsy	(+) Microcephaly,(+) Hypertelorism,(+) Upslanted palpebral fissure,(+) Ataxia,(+) Hypotonia,(+) Failure to thrive,(+) Ventricular septal defect,(+) Atrial septal defect,(+) Aortic valve stenosis,(+) Double outlet right ventricle,(+) Generalized-onset seizure,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Short stature,(+) Mild global developmental delay,(+) Midface retrusion,(+) Hypoplastic aortic arch,(+) Wide nasal base
LIF	ENST00000249075:c.320G>T p.Arg107Leu		de_novo	6.67	het	de novo	1	NDD+Epilepsy	Intellectual disability, seizure, epileptic encephalopathy
PIP5K1B	ENST00000265382:c.125G>A p.Gly42Asp		de_novo	5.74	het	de novo		NDD	increased nuchal translucency
ALG1	ENST00000588623:c.-125-12666C>T None		unknown	5.44	het	unknown		other	(+) Cryptorchidism,(+) Hearing impairment,(+) Small for gestational age,(+) Abnormal lung morphology,(+) Tachypnea,(+) Stridor,(+) Dilatation of the renal pelvis
MTMR12	ENST00000264934:c.358+1G>A None		unknown	6.5	het	unknown		NDD + Autism	(+) Autistic behavior,(+) Delayed speech and language development,(+) Mild global developmental delay
MYO15B	ENST00000582561:None None		comphet	2.13	het	maternal/paternal	1	NDD	Hypertension HP:0000822 Global developmental delay HP:0001263 Hyperammonemia HP:0001987 Abnormality of mitochondrial metabolism HP:0003287 Infantile muscular hypotonia HP:0008947 Spastic hemiparesis
PRPF31	ENST00000321030:c.822C>G p.Ile274Met		unknown	6.39	het	unknown	1	NDD	Moderate global developmental delay
LMO4	ENST00000370542:c.377A>C p.Asp126Ala		unknown	5.48	het	unknown		NDD	(+) Delayed speech and language development,(+) Atopic dermatitis,(+) Polysplenia,(+) Abnormal facial shape,(+) Biliary atresia,(+) Moderate global developmental delay,(+) Hodgkin lymphoma,(+) Abnormal inferior vena cava morphology,(+) Reduced serum alpha-1-antitrypsin
TTBK1	ENST00000259750:c.471+1G>C None		unknown	8.49	het	unknown		NDD	(+) Delayed speech and language development,(+) Atopic dermatitis,(+) Polysplenia,(+) Abnormal facial shape,(+) Biliary atresia,(+) Moderate global developmental delay,(+) Hodgkin lymphoma,(+) Abnormal inferior vena cava morphology,(+) Reduced serum alpha-1-antitrypsin
DGKI	ENST00000288490:c.1670G>A p.Arg557His		unknown	4.56	het	unknown		Epilepsy	(+) Focal-onset seizure
LRRC41	ENST00000343304:c.521del p.Gln174Argfs*76		unknown	7.53	het	unknown	3	NDD+ Epilepsy	(+) Intellectual disability,(+) Seizure,(+) Generalized-onset seizure,(+) Difficulty walking,(+) Developmental regression,(+) Moderate global developmental delay
DENND2A	ENST00000275884:c.1889+43C>T None		homo	3.15	homo	maternal/paternal		NDD	Micropenis, Intellectual disability, Intellectual disability, mild, Global developmental delay, Motor delay, Intellectual disability, profound, Delayed gross motor development, Intellectual disability, moderate, Expressive language delay, Delayed fine motor development, Intellectual disability, severe
IQSEC3	ENST00000326261:c.2531A>G p.Asp844Gly		de_novo	7.95	het	de novo	2	NDD+ Epilepsy	Bilateral tonic-clonic seizure with generalized onset, Spastic ataxia, Spastic gait, Gait ataxia, Absent speech, Autistic behavior, Intellectual disability, Self-injurious behavior
TIMM17A	ENST00000367287:c.244C>G p.Gln82Glu		de_novo	5.89	het	de novo	2	NDD+ Epilepsy	Bilateral tonic-clonic seizure with generalized onset, Spastic ataxia, Spastic gait, Gait ataxia, Absent speech, Autistic behavior, Intellectual disability, Self-injurious behavior
NPL	ENST00000258317:c.416C>T p.Pro139Leu		de_novo	6.4	het	de novo		NDD	Optic atrophy, Strabismus, Hypermetropia, Mild global developmental delay, Hydrocephalus, Intraventricular hemorrhage
KIAA0368 (ECPAS)	ENST00000259335:c.4323del p.Val1442Phefs*10		unknown	7.24	het	unknown			(+) Microcephaly,(+) Hypotonia,(+) Spasticity,(+) Abnormal cerebellum morphology,(+) Generalized-onset seizure,(+) Severe global developmental delay,(+) Brain imaging abnormality
MAPK9	ENST00000343111:c.214C>T p.Arg72Cys		unknown	5.68	het	unknown			(+) Microcephaly,(+) Hypotonia,(+) Spasticity,(+) Abnormal cerebellum morphology,(+) Generalized-onset seizure,(+) Severe global developmental delay,(+) Brain imaging abnormality
ARPC2	ENST00000295685:c.3G>T p.Met1?		unknown	7.07	het	unknown	3	NDD	(+) Delayed speech and language development,(+) Intellectual disability, mild,(+) Limb tremor
TBL3	ENST00000332704:c.323C>G p.Ser108*		unknown	6.24	het	unknown	3	NDD	(+) Delayed speech and language development,(+) Intellectual disability, mild,(+) Limb tremor
PLEKH3	ENST00000293349:c.1632_1633insGCAT p.Arg545Alafs*98		unknown	5.59	het	unknown	3	NDD	(+) Delayed speech and language development,(+) Intellectual disability, mild,(+) Limb tremor
TAAR2	ENST00000367931:c.737T>G p.Leu246Trp		AR_homo	4.6	homo	maternal/paternal	6	NDD	moderate global developmental delay, restlessness, spenomegaly
ABHD14B	ENST00000315877:c.193G>A p.Val65Met		AR_homo	5.19	homo	maternal/paternal	6	NDD	moderate global developmental delay, restlessness, spenomegaly
TIGD5	ENST00000504548:c.1319G>T p.Cys440Phe		AR_homo	4.39	homo	maternal/paternal	6	NDD	moderate global developmental delay, restlessness, spenomegaly
NXF1	ENST00000294172:c.1208G>A p.Arg403Gln		de_novo	8.65	het	de novo	6	NDD	moderate global developmental delay, restlessness, spenomegaly, ADHD
CMPK2 NRIR			AR_homo	6.31	homo	maternal/paternal	6	NDD	moderate global developmental delay, restlessness, spenomegaly

CACNA2D1	ENST00000356253:c.716G>A p.Arg239His		de_novo	8.8	het	de novo	1	NDD	Moderate global developmental delay, hypospadias, single transverse palmar crease
PRR14L	ENST00000327423:c.6131T>G p.Leu2044*		unknown	6.24	het	de novo		Epilepsy	(+) Seizure,(+) Generalized-onset seizure
PKHD1L1	ENST00000378402:c.6351-1G>C None	ENST00000378402:c.11408A>G p.His3803Arg	AR_comphet	5.54	het	maternal/ paternal	6	NDD	moderate global developmental delay, restlessness, spenomegaly
CACNB3	ENST00000301050:c.604G>A p.Asp202Asn		unknown	5.73	het	unknown	1	NDD	(+) Fused labia minora,(+) Epicanthus,(+) Wide nose,(+) Myopia,(+) Delayed speech and language development,(+) Intrauterine growth retardation,(+) Premature birth,(+) Sandal gap,(+) Recurrent infections,(+) Depressed nasal bridge,(+) Patent ductus arteriosus after premature birth,(+) Slanting of the palpebral fissure,(+) Retinopathy of prematurity
LONRF3	ENST00000371628:c.1940A>C p.Asn647Thr		XL	4.81	hemi	maternal	1	Epilepsy	Early childhood absence epilepsy (onset at 1-2 years of age), exclusion of a glucose transporter defect 12/2020, unremarkable cranial MRI, age-appropriate development, short attention span
RIPK2	ENST00000220751:c.1165C>T p.Pro389Ser		de_novo	4.46	het	de novo	1	NDD	(+) Narrow mouth,(+) Abnormal pinna morphology,(+) Iris coloboma,(+) Glandular hypospadias,(+) Polymicrogyria,(+) Cerebral hypoplasia,(+) Aplasia/Hypoplasia of the corpus callosum,(+) Bilateral microphthalmos,(+) Severe global developmental delay
TCEB3	ENST00000418390:c.1869+2T>C None		unknown	7.6	het	unknown	1	Epilepsy	(+) Focal-onset seizure,(+) Sleep apnea,(+) Focal sensory seizure with auditory features,(+) Bilateral tonic-clonic seizure with generalized onset,(+) Psychic epileptic aura
TAAR9	ENST00000434551:c.611T>C p.Leu204Pro		AR_homo	4.64	homo	maternal/ paternal	4	NDD+Epilepsy	seizure, hypotonia, global developmental delay, moderate intellectual disability, dystonic gait, periodic fever
MSL2	ENST00000309993:c.1231_1232del p.His412Ter		unknown	6.3	het	unknown	1	Epilepsy, Schizophrenia	(+) Generalized myoclonic seizure,(+) Bilateral tonic-clonic seizure with generalized onset,(+) Schizophrenia
CORO2A	ENST00000343933:c.1010T>C p.Ile337Thr		de_novo	4.91	het	de novo	4	NDD+Epilepsy	seizure, hypotonia, global developmental delay, moderate intellectual disability, dystonic gait, periodic fever
OSTF1	ENST00000346234:c.251G>A p.Gly84Asp		AR_homo	5.19	homo	maternal/ paternal	4	NDD+Epilepsy	seizure, hypotonia, global developmental delay, moderate intellectual disability, dystonic gait, periodic fever
VPS8	ENST00000287546:c.4189C>T		AR_homo	3.76	homo	maternal/ paternal	6	NDD+epilepsy	autism, aggressive behavior, delayed speech and language development, seizure, severe global developmental delay
GNL1	ENST00000376621:c.562T>A		AR_homo	5.35	homo	maternal/ paternal	6	NDD+epilepsy	autism, aggressive behavior, delayed speech and language development, seizure, severe global developmental delay
OARD1	ENST00000244558:c.6C>T		AR_homo	6.44	homo	maternal/ paternal	6	NDD+epilepsy	autism, aggressive behavior, delayed speech and language development, seizure, severe global developmental delay
XPO5	ENST00000265351:c.625A>T		AR_homo	4.92	homo	maternal/ paternal	6	NDD+epilepsy	autism, aggressive behavior, delayed speech and language development, seizure, severe global developmental delay
KIF16B	ENST00000354981:c.2564A>G		AR_homo	5.34	homo	maternal/ paternal	6	NDD+epilepsy	autism, aggressive behavior, delayed speech and language development, seizure, severe global developmental delay
EFCC1	ENST00000480450:c.988G>A		de_novo	4.58	het	de novo	6	NDD+epilepsy	autism, aggressive behavior, delayed speech and language development, seizure, severe global developmental delay
PRPSAP2	ENST00000268835:c.122G>A p.Arg41Gln		de_novo	6.03	het	de novo	1	NDD	Suspected mitochondriopathy (MELAS), with stroke-like episodes, primarily neurodegen-erative disease, impaired diffusion in cMRI, ataxia, dystrophy, right abducens palsy, left temporal ischaemia, pronounced inner and outer cerebrospinal fluid spaces and periventricular white matter abnormalities.
LRP1B	ENST00000389484:c.1014-3C>G None		de_novo	7.14	het	de novo	1	NDD+epilepsy	severe developmental delay, focal epilepsy, epileptic encephalopathy
NUMB	ENST00000554546:c.1592A>G p.Asn531Ser		AR_homo	7.23	homo	maternal/ paternal	2		connatal hyperinsulinism (ABCC8 variant), ASD, moderate developmental delay, generalized onset epilepsy, muscular hypotonia, microcephaly
MAP3K9	ENST00000381250:c.1723_1728del p.Arg575_Ser576del		AR_homo	4.1	homo	maternal/ paternal	2		connatal hyperinsulinism (ABCC8 variant), ASD, moderate developmental delay, generalized onset epilepsy, muscular hypotonia, microcephaly
APBA1	ENST00000265381:c.1482G>A p.Lys494=		de_novo	10.3	het	de novo	1		autism, moderate global developmental delay
POLQ	ENST00000264233:c.7537_7538inv p.Gln2513*		unknown	6.62	het	unknown		Epilpesy	Myoclonus,(+) Focal clonic seizure,(+) Focal tonic seizure,(+) Focal impaired awareness motor seizure,(+) Focal impaired awareness non-motor seizure
PPP2R2C	ENST00000335585:c.754A>G p.Met252Val		unknown	5.04	het	unknown	2	NDD	(+) Tall stature,(+) Macrocephaly,(+) Hypertelorism,(+) Hyperhidrosis,(+) Obesity,(+) Frontal bossing,(+) Attention deficit hyperactivity disorder,(+) Mild global developmental delay,(+) Abnormal social behavior
SPIN1	ENST00000375859:c.473G>A p.Arg158His		unknown	5.55	het	unknown	2	NDD	(+) Tall stature,(+) Macrocephaly,(+) Hypertelorism,(+) Hyperhidrosis,(+) Obesity,(+) Frontal bossing,(+) Attention deficit hyperactivity disorder,(+) Mild global developmental delay,(+) Abnormal social behavior

HMG20B	ENST00000262949:c.562G>T p.Gly188*		de_novo	7.63	het	de novo	1	NDD+other	Moderate global developmental delay, Protruding ear, Abnormal morphology of the nasal alae, Behavioral abnormality, Delayed speech and language development, Eczema, Prominent fingertip pads, Generalized hypotonia, Aortic valve stenosis, Inverted nipples, Pulmonary artery stenosis, Sparse lateral eyebrow
BPIFC	ENST00000300399:c.6T>A p.Cys2*		unknown	4.28	het	unknown	1	NDD	(+) Abnormality of the face,(+) Intellectual disability,(+) Short stature
TLX2	ENST00000233638:c.629C>T p.Thr210Ile		de_novo	5.17	het	de novo	1		focal epilepsy, ADHD
GRIN3A	ENST00000361820:c.1635C>A p.Asp545Glu		de_novo	6.56	het	de novo	2	NDD+epilepsy	seizure, global developmental delay, motor tics, epistaxis, abnormality of von Willebrandt factor
TTYH2	ENST00000269346:c.728C>T p.Ala243Val		de_novo	4.88	het	de novo	2	NDD+epilepsy	seizure, global developmental delay, motor tics, epistaxis, abnormality of von Willebrandt factor
SLC4A8	ENST00000358657:c.1745A>G p.Tyr582Cys		unknown	4.45	het	unknown		Epilepsy, speech delay	(+) Delayed speech and language development,(+) Generalized non-motor (absence) seizure,(+) Generalized-onset seizure
SBNO1	ENST00000267176:c.1084T>A p.Leu362Ile		unknown	4.86	het	unknown		Epilepsy, speech delay	(+) Delayed speech and language development,(+) Generalized non-motor (absence) seizure,(+) Generalized-onset seizure
KCNJ3	ENST00000295101:c.863T>C p.Met288Thr		ad_inherited	6.63	het	unknown	2	NDD	(+) Intellectual disability,(+) Severe global developmental delay
HECTD2	ENST00000298068:c.1523T>G p.Leu508Arg		ad_inherited	4.65	het	unknown	2	NDD	(+) Intellectual disability,(+) Severe global developmental delay
HECW1	ENST00000395891:c.4172T>C p.Leu1391Ser		ad_inherited	5.59	het	unknown	1	NDD	(+) Delayed speech and language development,(+) Intellectual disability, mild,(+) Attention deficit hyperactivity disorder
USP31	ENST00000219689:c.1240C>G p.His414Asp		de_novo	4.71	het	de novo	2	NDD+epilepsy	ataxia, progressive muscle weakness, slowed slurred speech, focal-onset seizure, mild global developmental delay, hemihypertrophy of lower limb
ESYT1	ENST00000394048:c.2986C>T p.Arg996Trp		de_novo	5.67	het	de novo	1	Epilepsy	tonic clonic seizures, eyelid myoclonia, absence seizures
RECQL5	ENST00000317905:c.1235G>A p.Arg412His		de_novo	6.2	het	de novo	2	NDD+epilepsy	ataxia, progressive muscle weakness, slowed slurred speech, focal-onset seizure, mild global developmental delay, hemihypertrophy of lower limb
PROKR1	ENST00000303786:c.731C>T p.Pro244Leu		de_novo	4.43	het	de novo	1	NDD+epilepsy	microcephaly, hypothyroidism, generalized-onset seizure, moderate global developmental delay
TMF1	ENST00000398559:c.716G>A	ENST00000398559:c.2716A>G	comphet	4.4	comphet	maternal/paternal	1	NDD+epilepsy	Macrocephaly, seizure, hypotonia, Dandy-Walker malformation, muscle weakness, distal lower limb amyotrophy, mild global developmental delay
CUL5	ENST00000393094:c.194T>C p.Leu65Ser		unknown	5.75	het	unknown	1	NDD/spasticity	Global developmental delay,(+) Spastic tetraparesis
DRC3	ENST00000399182:c.205A>G p.Lys69Glu	ENST00000399182:c.605A>G p.Glu202Gly	comphet	4.88	comphet	maternal/paternal	2	NDD+epilepsy	moderate global developmental delay, focal epilepsy, intellectual impairment, autistic features, bilateral spastic cerebral palsy, visual impairment with bilateral optic atrophy, cerebral malformation
PLXNB2	ENST000003593377:c.4580C>T p.Ser1527Leu	ENST000003593377:c.2875A>G p.Met959Val	comphet	5.67	comphet	maternal/paternal	2	NDD+epilepsy	moderate global developmental delay, focal epilepsy, intellectual impairment, autistic features, bilateral spastic cerebral palsy, visual impairment with bilateral optic atrophy, cerebral malformation
HP1BP3	ENST00000312239:c.717_718del p.Lys240Ilefs*7		unknown	7.14	het	unknown	1	NDD	(+) Hypotonia,(+) Mild global developmental delay,(+) Abnormal myelination
JRKL	ENST00000332349:c.788G>A p.Arg263Gln		de_novo	4.23	het	de novo	1	NDD	tall stature, macrocephaly, global developmental delay, sleep disturbance, obesity, autistic behavior
FAM222A	ENST00000538780:c.1231del p.Tyr411Ilefs*80		de_novo	7.81	het	de novo	3	NDD	hypotonia, abnormality of acid-base homeostasis, mild global developmental delay, feeding difficulties, vitamin B12 deficiency
PSMD10	ENST00000217958:c.623C>A p.Pro208His		de_novo	6.51	het	de novo	3	NDD	hypotonia, abnormality of acid-base homeostasis, mild global developmental delay, feeding difficulties, vitamin B12 deficiency
PFAS	ENST00000314666:c.1981G>A p.Val661Met, c.3072G>T p.Glu1024Asp		AR_comphet	5.6	comphet	maternal/paternal	3	NDD	hypotonia, abnormality of acid-base homeostasis, mild global developmental delay, feeding difficulties, vitamin B12 deficiency
EPHA8	ENST00000166244:c.2388G>A p.Thr796=		homo	9.13	homo	maternal&paternal	3	NDD	(+) Microcephaly,(+) Aggressive behavior,(+) Spasticity,(+) Intellectual disability, severe,(+) Crohn's disease,(+) Self-injurious behavior
ADGRB2	NM_001703.2:c.4572+1G>A p.(?)			B	het	unknown		NDD	(+) Personality changes,(+) Asthma,(+) Tetraparesis,(+) Sleep disturbance,(+) Falls,(+) Metachromatic leukodystrophy variant,(+) Poor fine motor coordination,(+) Chronic pain,(+) Short term memory impairment,(+) Erectile dysfunction

DIP2A	ENST00000417564:c.1612C>T p.Arg538Trp	ENST00000417564:c.1894A>G p.Met632Val	comphet	5.74	comphet	maternal/ paternal	2	NDD+epilepsy	bilateral tonic-clonic seizures, focal epilepsy, severe GDD, severe ID, bilateral spastic cerebral palsy, muscular hypotonia, microcephaly, short stature, dystrophia, coloboma, multiple malformations, anophthalmia, microphthalmia, Aicardi syndrome
FRYL	ENST00000358350:c.2021C>T p.Pro674Leu		de_novo	6.74	het	de novo	3	NDD	(+) Autism,(+) Attention deficit hyperactivity disorder,(+) severe global developmental delay
GCN1	ENST00000300648:c.689A>T p.Asn230Ile	ENST00000300648:c.2044G>A p.Val682Met	comphet	4.22	comphet	maternal/ paternal	2	NDD	moderate global developmental delay, intellectual disability, obesity
AKAP13	ENST00000361243:c.3689T>C p.Leu1230Pro	ENST00000361243:c.8030A>G p.Gln2677Arg	comphet	4.36	comphet	maternal/ paternal	2	NDD	moderate global developmental delay, intellectual disability, obesity
FIZ1	ENST00000221665:c.665A>C p.His222Pro		de_novo	B	het	de novo	2	other	recurrent neuroinflammation, partial albinism, recurrent petechiae, optic atrophy, mild hypogammaglobulinemia
PRR14L	ENST00000327423:c.4908A>C p.Arg1636Ser		de_novo	B	het	de novo	2	other	recurrent neuroinflammation, partial albinism, recurrent petechiae, optic atrophy, mild hypogammaglobulinemia
CHST2	ENST00000309575:c.1124A>G p.Lys375Arg		de_novo	5.11	het	de novo	2	NDD	mild intellectual disability, global developmental delay
PLCB1	ENST00000338037:c.3298C>T p.Arg1100Trp		de_novo	10.0	het	de novo	2	NDD	mild intellectual disability, global developmental delay
RGPD2	ENST00000327544:c.2066_2070del p.Lys689Argfs*8		unknown	5.43	het	unknown	1	NDD+epilepsy	Epilepsy with spasms, tonic seizure, complex seizure, GTKA, intellectual disability, ataxia
KIF13A	ENST00000259711:c.46A>C p.Asn16His		de_novo	6.54	het	de novo	2	NDD+other	Microcephaly, Global developmental delay, Failure to thrive, constipation, infantile muscular hypotonia
HEATR6	ENST00000184956:c.1311del p.Val438Phefs*24, c.3320T>C p.Leu1107Pro		comphet	5.03	comphet	maternal/ paternal	2	NDD+other	Microcephaly, Global developmental delay, Failure to thrive, constipation, infantile muscular hypotonia
ATRNL1	ENST00000355044:c.402del p.Arg134Serfs*26		unknown	7	het	unknown	2	NDD+myopathy+Autistic behaviour	(+) Atypical behavior,(+) Delayed speech and language development,(+) Enuresis,(+) Intellectual disability,(+) Hypotonia,(+) Motor delay,(+) Myopathy,(+) Peripheral neuropathy,(+) Severe global developmental delay
PRMT9	ENST00000322396:c.734_735del p.Ile245Thrfs*41, c.792A>C p.Glu264Asp		comphet	5.55	comphet	paternal/ maternal	1	NDD	Global developmental delay, hypotonia, plagiocephaly, hearing impairment, cow milk allergy
INSYN1	ENST00000559817:c.182T>C		de_novo	4.37	het	de novo	1	NDD+other	Leukoencephalopathy, developmental regression, moderate global developmental delay
CASKIN2	ENST00000321617:c.3061_3068dup p.Ser1024Hisfs*99		unknown	7.03	het	unknown	2	NDD	(+) Obesity,(+) Intellectual disability, moderate,(+) Focal-onset seizure
PLXNA4	ENST00000321063:c.5057G>A p.Gly1686Asp		unknown	5.83	het	unknown	2	NDD	(+) Obesity,(+) Intellectual disability, moderate,(+) Focal-onset seizure
LRFN1	ENST00000248668:c.1520C>A p.Thr507Lys		unknown	4.74	het	unknown	1	Epilepsy	(+) Bilateral tonic-clonic seizure with focal onset,(+) Focal-onset seizure
PLXNA1	ENST00000393409:c.2225A>G p.Tyr742Cys		unknown	5.27	het	unknown	1	NDD+other	Crohn's disease, Developmental delay, Intellectual disability, borderline, Feeding difficulties, Behavioral abnormality, Abnormal fear/anxiety-related behavior, Depression
TMEM104	ENST00000335464:c.983G>T		de_novo	4.54	het	de novo	1	Epilepsy	Generalized myoclonic-atonic seizure
SPACA9	ENST00000350499:c.495+1G>A		AR_homo	8.14	homo	maternal/ paternal	3	NDD	autism, moderate global developmental delay
BCO2	ENST00000357685:c.709A>C p.Asn237His,		comphet	3.69	comphet	maternal/ paternal	3	NDD	autism, moderate global developmental delay
TSPYL2	ENST00000375442:c.815A>G p.Asn272Ser		XL	4.91	hemi	maternal	3	NDD	autism, moderate global developmental delay
DPYSL3	ENST00000343218:c.477del p.Ile159Metfs*5		unknown	8.45	het	unknown	2	NDD	(+) Microcephaly,(+) Short stature,(+) Mild global developmental delay
KCNH4	ENST00000264661:c.908A>T p.His303Leu		unknown	4.18	het	unknown	2	NDD	(+) Microcephaly,(+) Short stature,(+) Mild global developmental delay
SYVN1	ENST00000294256:c.883C>T p.Arg295*		unknown	6.74	het	unknown		Epilepsy	(+) Generalized-onset seizure,(+) Continuous spike and waves during slow sleep
CSTF3	ENST00000323959:c.203G>T p.Trp68Leu		unknown	5	het	unknown		Epilepsy	(+) Generalized-onset seizure,(+) Continuous spike and waves during slow sleep
GRN	ENST00000053867:c.-9_-8+16dup None		de_novo	8.03	het	de novo	2	NDD+epilepsy	bilateral tonic-clonic seizures, focal epilepsy, severe global developmental delay, severe intellectual disability, bilateral spastic cerebral palsy, muscular hypotonia, microcephaly, short stature, dystrophia, iris and retinal coloboma, multiple malformations, dysphagia, anophthalmia, microphthalmia, Aicardi syndrome

JADE2	ENST00000282605:c.83C>G p.Ser28*		de_novo	8.22	het	de novo	1	NDD	autism, severe global developmental delay
RPS5	ENST00000196551:c.380G>A p.Arg127His		de_novo	7.4	het	de novo	1	NDD+epilepsy	Lennox-Gastaut-syndrome, moderate intellectual disability, autistic features, possible microcephaly, cerebellar atrophy, reflux oesophagitis, fT3/ fT4/ TSH in reference range, severe sensorimotor axonal and demyelinating polyneuropathy
TRPC3	ENST00000264811:c.1694C>T p.Pro565Leu		de_novo	8.43	het	de novo	3	NDD + epilepsy	(+) Strabismus,(+) Delayed speech and language development,(+) Global developmental delay,(+) Motor delay,(+) Gait ataxia,(+) Generalized-onset seizure,(+) Recurrent respiratory infections,(+) Secondary microcephaly,(+) Intracranial cystic lesion,(+) Complex febrile seizure,(+) Neonatal seizure,(+) Abnormality of movement
ESRRG	ENST00000408911:.550C>T p.Arg184Cys		de_novo	7.08	het	de novo	1	NDD+other	Prolonged neonatal jaundice, Moderate global developmental delay, Upgaze palsy, Joint hyperflexibility, Growth delay, Ataxia, Hypotonia, Dysarthria, Atypical behavior
RBM26	ENST00000267229:c.899dup p.Cys303Leufs*10		unknown	7.16	het	paternal	3	NDD	(+) High forehead,(+) Autism,(+) Delayed speech and language development,(+) Hyperactivity,(+) Pectus excavatum,(+) Global developmental delay,(+) Abnormal renal morphology,(+) Supravalvar pulmonary stenosis,(+) Self-injurious behavior
PUM2	ENST00000319801:c.1534C>T p.Gln512*		unknown	8.53	het	maternal	3	NDD	(+) High forehead,(+) Autism,(+) Delayed speech and language development,(+) Hyperactivity,(+) Pectus excavatum,(+) Global developmental delay,(+) Abnormal renal morphology,(+) Supravalvar pulmonary stenosis,(+) Self-injurious behavior
LRRC1	ENST00000370882:c.206A>G p.Asn69Ser		de_novo	5.56	het	de novo	2	Epilepsy	(+) Anxiety,(+) Seizure,(+) Attention deficit hyperactivity disorder,(+) Focal-onset seizure,(+) Panic attack,(+) Abnormal fear/anxiety-related behavior
ADAMTS14	ENST00000373208:c.870+1G>A	ENST00000373208:c.2106G>A	comphet	4.39	homo	maternal/paternal	1	NDD+other	microcephaly, hip dysplasia, short stature, attention deficit hyperactivity disorder, receptive language delay, growth delay, mild global developmental delay.
CPO	ENST00000272852:c.551G>A p.Arg184Gln	ENST00000272852:c.484-365C>G	comphet	3.28	comphet	paternal/maternal	3	NDD+Epilepsy	Hearing impairment, Obesity, Polymicrogyria, Spastic paraparesis, Intellectual disability (borderline), Moderate global developmental delay, Bilateral tonic-clonic seizure with generalized onset
UBE2D2	ENST00000253815:c.284C>T p.Ala95Val		unknown	5.53	het	unknown	2	Epilepsy	(+) Seizure,(+) Paroxysmal dyskinesia,(+) Focal-onset seizure
IRF2BP1	ENST00000302165:c.1726A>G		homo	3.96	homo	maternal/paternal	2	NDD + other	Thin vermilion border, microcephaly, high forehead, plagiocephaly, failure to thrive, small for gestational age, expressive language delay, delayed fine motor development, midface retrusion
DGCR8	ENST00000351989:c.805G>A		homo	8.45	homo	maternal/paternal	2	NDD + other	Thin vermilion border, microcephaly, high forehead, plagiocephaly, failure to thrive, small for gestational age, expressive language delay, delayed fine motor development, midface retrusion
RASSF6	ENST00000307439:c.683C>T,p.Pro228Leu		homo	4.58	homo	maternal/paternal	3	NDD+Epilepsy	Hearing impairment, Obesity, Polymicrogyria, Spastic paraparesis, Intellectual disability (borderline), Moderate global developmental delay, Bilateral tonic-clonic seizure with generalized onset
TRAK1	ENST00000327628:c.2498A>C p.Gln833Pro		de_novo	9.18	het	de novo	3	NDD+Epilepsy	Hearing impairment, Obesity, Polymicrogyria, Spastic paraparesis, Intellectual disability (borderline), Moderate global developmental delay, Bilateral tonic-clonic seizure with generalized onset
STAM	ENST00000377524:c.1073C>G p.Ser358*		unknown	8.34	het	unknown	1	NDD	(+) Narrow nose,(+) Synophrys,(+) Autism,(+) Impaired social interactions,(+) Pes planus,(+) Unsteady gait,(+) Supernumerary nipple
NOC4L	ENST00000330579:c.910del p.Leu304Serfs*6	ENST00000330579:c.1418T>C p.Leu473Pro	comphet	6.08	comphet	paternal/maternal	2	NDD+epilepsy	neurodevelopmental delay, seizure
ARSF	ENST00000359361:c.784C>T p.Arg262*		XL	5.97	hemi	maternal	2	NDD+epilepsy	neurodevelopmental delay, seizure
GRM5	ENST00000418177:c.-201+1G>C		comphet	9.04	het	paternal/maternal	1	NDD + other	hypotonia, bulbar palsy, respiratory insufficiency, myopathy, elevated circulating creatine kinase concentration, myalgia, fatigable weakness, mild global developmental delay.
THBS2	ENST00000366787:c.2095G>A p.Gly699Ser		de_novo	5.34	het	paternal/maternal	1	NDD + epilepsy + other	seizure, global developmental delay, dilated cardiomyopathy, gastrointestinal hemorrhage, colitis, bilateral renal dysplasia, systemic autoinflammation, middle cerebral artery stroke
DOPEY1/DOP1A	ENST00000237163:c.2897T>C p.Leu966Pro		unknown	5.3	het	unknown		NDD	(+) Delayed speech and language development,(+) Moderate global developmental delay
CSTF1	ENST00000217109:c.686G>A p.Gly229Glu		unknown	4.4	het	unknown		Epilepsy	(+) Generalized-onset seizure,(+) Focal-onset seizure,(+) Visually-induced seizure
DSCAML1	ENST00000321322:c.2373C>A p.Asn791Lys		unknown	5.37	het	unknown	1	NDD+Epilepsy	(+) Seizure,(+) Abnormality of neuronal migration,(+) Intellectual disability, moderate,(+) Abnormal periventricular white matter morphology,(+) Typical absence seizure

LPHN3	ENST00000502815:c.772G>A p.Val258Met		unknown	6.71	het	unknown	1	NDD+Epilepsy	(+) Generalized-onset seizure,(+) Severe global developmental delay,(+) Infantile spasms
CHD9	ENST00000398510:c.6361C>A p.Pro2121Thr		de_novo	7.32	het	de novo	1	NDD + other	mild global developmental delay, short stature, growth hormone deficiency, glucose intolerance, hypoglycaemia, increased intracranial pressure, headache, atypical behaviour, patent ductus arteriosus after birth at term, hyperlordosis, cMRI: heterotopia
TRIM71	ENST00000383763:c.2229_2230del p.Trp744Glufs*4		unknown	8.14	het	unknown	1	Short stature	(+) Hypospadias,(+) Multiple lentiginos,(+) Preaxial hand polydactyly,(+) Disproportionate short stature
GCN1L1	ENST00000300648:c.6478A>C p.Thr2160Pro		unknown	5	het	unknown	2	NDD	(+) Microcephaly,(+) Atypical behavior,(+) Intellectual disability,(+) Spasticity,(+) Global developmental delay,(+) Progressive neurologic deterioration,(+) Periventricular leukomalacia
CELSR3	ENST00000164024:c.5791C>G p.Leu1931Val		AD_unknown	6.28	het	unknown	2	NDD	(+) Microcephaly,(+) Atypical behavior,(+) Intellectual disability,(+) Spasticity,(+) Global developmental delay,(+) Progressive neurologic deterioration,(+) Periventricular leukomalacia
MBP	ENST00000354542:c.177+6378A>G None		de_novo	B	het	de novo	1	other	congenital heart defect, functionally univentricular heart, dilated, non-contractile and non-perfused left ventricle, dysplastic aortic and mitral valves, multiple muscular ventricular septal defects, hypoplastic aortic arch; no other malformations known
ANKRD23	ENST00000318357:c.748G>A p.Ala250Thr		de_novo	5.34	het	de novo	2	NDD + epilepsy + other	premature birth at 25+2 weeks of gestational age, microcephaly, bilateral renal dysplasia, retinopathy, multiple hernias, 2-3 toe syndactyly, seizures, motor delay, delayed speech and language developmental, choroid plexus cysts, bronchodysplasia, hypospadias, big ears
GABRE	ENST00000370328.3:c.1148A>G p.Asn383Ser		AD_denovo	4.9	het	de novo	2	epilepsy	Seizure, abnormality of metabolism, epileptic encephalopathy
BCL2L11	ENST00000393256:c.268T>C p.Ser90Pro		de_novo	6.11	het	de novo	2	NDD + epilepsy + other	premature birth at 25+2 weeks of gestational age, microcephaly, bilateral renal dysplasia, retinopathy, multiple hernias, 2-3 toe syndactyly, seizures, motor delay, delayed speech and language developmental, choroid plexus cysts, bronchodysplasia, hypospadias, big ears
CRMP1	ENST00000324989:c.1234G>T p.Ala412Ser		unknown	6.09	het	unknown	1	NDD	(+) Intellectual disability, moderate,(+) Short stature
BIRC6	ENST00000421745:c.2213C>A p.Pro738His		unknown	5.28	het	unknown	2	NDD	(+) Delayed speech and language development,(+) Mild global developmental delay
TLN1	ENST00000314888:c.5672G>C p.Ser1891Thr		unknown	5.06	het	unknown	2	NDD	(+) Delayed speech and language development,(+) Mild global developmental delay
PNMA6F	ENST00000436629		XL	3.34	het	maternal	1	NDD	Microcephaly, Syndactyly, Intellectual disability, Dandy-Walker malformation, Hip dysplasia, Polymicrogyria, Partial duplication of thumb phalanx, Severe intellectual disability, Epileptic spasm, Abnormal brain morphology
UBE4B	ENST00000253251:c.1885T>C p.Phe629Leu		AD_unknown	5.69	het	unknown	2	NDD + epilepsy + other	(+) Autistic behavior,(+) Seizure,(+) Global developmental delay,(+) Hypoglycemia,(+) Hyponatremia,(+) Inappropriate antidiuretic hormone secretion
ATRNL1	ENST00000355044:c.402del p.Arg134Serfs*26		unknown	7	het	unknown	1	NDD	(+) Atypical behavior,(+) Developmental regression,(+) Aplasia/Hypoplasia of the corpus callosum,(+) Severe global developmental delay
BAIAP2	ENST00000321280:c.1019C>T p.Thr340Ile		de_novo	8.23	het	de novo	1	NDD+ Epilepsy	mild global developmental delay, generalized clonic seizure, EEG with spike-wave complexes
DHX15	ENST00000336812:c.1277C>T p.Thr426Met		unknown	5.68	het	unknown	2		(+) Macrocephaly,(+) Global developmental delay,(+) Short stature
USP3	ENST00000268049:c.615_616del p.Ala206Phefs*10		unknown	6.63	het	unknown	2		(+) Macrocephaly,(+) Global developmental delay,(+) Short stature
GALNT8	ENST00000252318:c.1431T>G p.Phe477Leu		de_novo	4.49	het	de novo	2	NDD + epilepsy	Seizure, moderate global developmental delay, ataxia, dystonia, moderate intellectual disability, atypical behavior
ADCY8	ENST00000286355:c.3523C>T p.Gln1175*		de_novo	8.03	het	de novo	2	NDD + epilepsy	Seizure, moderate global developmental delay, ataxia, dystonia, moderate intellectual disability, atypical behavior
FAM65A	ENST00000042381:c.1855dup p.Ser619Phefs*84		unknown	6.97	het	unknown	1	NDD + epilepsy	(+) Atypical behavior,(+) Global developmental delay,(+) Expressive language delay,(+) Focal-onset seizure,(+) Abnormal eye contact
PARP6	ENST00000260376:c.631dup p.Arg211Profs*74		unknown	6.54	het	unknown	1	epilepsy	(+) Bilateral tonic-clonic seizure with focal onset,(+) Focal-onset seizure,(+) Motor seizure
JADE1	ENST00000226319:c.1546A>T p.Lys516*		unknown	6.5	het	unknown	1	NDD	(+) Orofacial cleft,(+) Global developmental delay,(+) Metopic synostosis
RANBP2	ENST00000283195:c.1684_1685del p.Leu562Lysfs*28		unknown	9.9	het	unknown	1	Optic atrophy	optic atrophy, myopia, ADHD, fine motor delay
MAOB	ENST00000378069:c.857T>G p.Ile286Ser		homo	8.81	homo	paternal/maternal	3	NDD + epilepsy	(-) Abnormality of the face,(+) Focal-onset seizure,(+) Moderate global developmental delay
TRPC5	ENST00000262839:c.778C>G p.Arg260Gly		x_linked	7.09	hemi	maternal	1	NDD + other	microcephaly, ataxia, cerebellar vermis hypoplasia, short stature, unilateral renal atrophy, mild global developmental delay

ASCC3	ENST00000369162:c.1597-2A>G None	ENST00000369162:c.5996T>C p.Leu1999Pro	comphet	7.22	comphet	unknown	1	NDD	(+) High palate,(+) Retrognathia,(+) Low-set ears,(+) Nystagmus,(+) Hypotonia,(+) Muscle weakness,(+) Talipes equinovarus,(+) Arthrogryposis-like hand anomaly,(+) Moderate global developmental delay
SYMPK	ENST00000245934:c.3027C>A p.Tyr1009*		unknown	6.94		unknown	1	NDD	(+) Glandular hypospadias,(+) Global developmental delay,(+) Umbilical hernia
CEP170	ENST00000336415:c.4603G>T p.Glu1535*		unknown	7.85		unknown	1	NDD	(+) Glandular hypospadias,(+) Global developmental delay,(+) Umbilical hernia
SCRIB	ENST00000356994:c.1866C>T	ENST00000356994:c.787+6T>C	AR_comphet	7.51	comphet	maternal&paternal	1	NDD+epilepsy	Microcephaly, focal-onset seizure, moderate global developmental delay
GABRD	ENST00000378585:c.1336T>C p.Tyr446His		unknown	8.4	het	unknown	1	NDD	(+) High palate,(+) Brachycephaly,(+) Microcephaly,(+) Pointed chin,(+) Triangular face,(+) Low-set ears,(+) Hypotonia,(+) Failure to thrive,(+) Patent foramen ovale,(+) Iron deficiency anemia,(+) Moderate global developmental delay,(+) Feeding difficulties,(+) Tube feeding
KALRN	ENST00000291478:c.910C>T p.Gln304*		unknown	9.49	het	unknown	4	NDD	(+) Intellectual disability,(+) Hip dysplasia,(+) Bilateral talipes equinovarus,(+) Mild global developmental delay,(+) Overweight,(+) Psychogenic non-epileptic seizure,(+) Reduced impulse control
MAPK3	ENST00000263025:c.776-1G>A None		unknown	10.0	het	unknown	1	epilepsy	(+) Focal-onset seizure,(+) Focal-onset epileptic spasm
WIZ	ENST00000389282:c.247dup p.Gln83Profs*10		unknown	7.05	het	unknown	1	NDD +Autism	(+) Autistic behavior,(+) Impaired social interactions,(+) Delayed speech and language development,(+) Intellectual disability, mild
IKZF4	ENST00000262032:c.1487_1488del p.Lys496Argfs*29		unknown	6.04	het	unknown	1	NDD + spastic tetraparesis	(+) Seizure,(+) Spastic tetraparesis,(+) Severe global developmental delay,(+) Abnormal lateral ventricle morphology
POLR2B	ENST00000314595:c.1404+250A>G None		unknown	3.06	het	unknown	1	NDD + other	mild global developmental delay, agenesis of corpus callosum, longitudinal callosal fasci-cles
NXPH1	ENST00000405863:c.54+38925G>A None		de_novo	5.5	het	de novo	2	NDD + other	small fiber neuropathy, ID, IQ 55, cerebral palsy, small intestinal perforation
H4C6	ENST00000244537:c.3G>A p.Met1?	ENST00000244537:c.195dup p.Val66CysfsTer15	AR_comphet	7.74	comphet	maternal/paternal	2	NDD + other	small fiber neuropathy, ID, IQ 55, cerebral palsy, small intestinal perforation
CRISPLD2	ENST00000262424:c.289T>C p.Cys97Arg		de_novo	4.77	het	de novo	2	NDD+epilepsy	Seizure, Global developmental delay, Atonic seizure, Generalized myoclonic-atonic seizure, Epileptic encephalopathy
XRCC5	ENST00000392132:c.1113+141A>G		de_novo	5.45	het	de novo	2	NDD+epilepsy	Seizure, Global developmental delay, Atonic seizure, Generalized myoclonic-atonic seizure, Epileptic encephalopathy
SIPA1L1	ENST00000358550:c.2428C>T p.Gln810Ter		unknown	7.66	het	unkown	1	NDD+epilepsy	(+) Microcephaly,(+) Hypotonia,(+) Spasticity,(+) Focal-onset seizure,(+) Severe global developmental delay,(+) Perisylvian polymicrogyria
UBE4B	ENST00000343090:c.979C>T p.Leu327Phe		comphet	6.44	comphet	paternal/de novo	1	NDD	(+) Cyanosis,(+) Seizure,(+) Global developmental delay,(+) Generalized hypotonia
ADCY2	ENST00000338316:c.1026C>A p.Tyr342Ter		unknown	7.97	het	unkown	1	Epilepsy	(+) Focal-onset seizure,(+) Focal sensory seizure
SUGP1	ENST00000247001:c.1911+5G>T	ENST00000247001:c.698A>G p.Tyr233Cys	AR_comphet	7.57	comphet	paternal/maternal	1	NDD+epilepsy	High palate, Thin upper lip vermillion, Retrognathia, Broad philtrum, Posteriorly rotated ears, Delayed speech and language development, Intellectual disability, Febrile seizure (within the age range of 3 months to 6 years), Moderate global developmental delay, Finger clinodactyly
CADPS	ENST00000283269:c.969+4836A>G		de_novo	6.43	het	de novo	1	NDD+epilepsy	Intellectual disability, seizure, aphasia, restless legs
WDR7	ENST00000254442:c.3451C>T p.Arg1151Ter		unknown	6.41	het	unknown	2	NDD+epilepsy	(+) Seizure,(+) Mild global developmental delay
TERF2	ENST00000254942:c.1341-1G>C None		unknown	8.21	het	unknown	2	NDD+epilepsy	(+) Seizure,(+) Mild global developmental delay
CSMD2	ENST00000373388:c.1111G>A p.Glu371Lys	ENST00000373388:c.10310G>C p.Arg3437Thr	AR_comphet	5.34	comphet	paternal/maternal	2	NDD+epilepsy	autism, global developmental delay, generalized-onset seizure, focal-onset seizure
WDR13	ENST00000218056:c.1017G>C p.Lys339Asn		XL	4.7	hemi	maternal	2	NDD+epilepsy	autism, global developmental delay, generalized-onset seizure, focal-onset seizure

IGSF9B	ENST00000321016:c.2549T>C p.Ile850Thr		unknown	5.1	het	unknown	1	Epilepsy	(+) Seizure,(+) Specific learning disability,(+) Focal-onset seizure,(+) Mild global developmental delay
CSNK1D	ENST00000314028:c.581A>G p.Asp194Gly		de_novo	8.71	het	de novo	1	NDD + Epilepsy + other	generalized-onset seizure, febrile seizure, frontotemporal cerebral atrophy, subdural hemorrhage, muscular hypotonia, mild global developmental delay, plagiocephaly
SAP130	ENST00000259234:c.2134C>T p.Gln712Ter		unknown	6.93	het	unknown	2	NDD	(+) Pancreatitis,(+) Eosinophilia,(+) Short stature,(+) Mild global developmental delay,(+) Esophagitis,(+) Food allergy
SMAP1	ENST00000316999:c.52C>T p.Gln18Ter		unknown	5.87	het	unknown	2	NDD	(+) Pancreatitis,(+) Eosinophilia,(+) Short stature,(+) Mild global developmental delay,(+) Esophagitis,(+) Food allergy
YME1L1	ENST00000326799:c.955C>T p.Arg319Trp		unknown	6.77	het	unknown		NDD	(+) Hemangioma,(+) Abnormal cerebral cortex morphology,(+) Bilateral tonic-clonic seizure with focal onset,(+) Focal-onset seizure,(+) Mild global developmental delay,(+) Abnormality of brain morphology,(+) Perisylvian polymicrogyria,(+) Nevus sebaceus
FBXO41	ENST00000295133:c.670G>T p.Glu224Ter		unknown	8.41	het	unkown			Focal-onset seizure
SLC4A8	ENST00000358657:c.1157del p.Gly386ValfsTer70		unknown	6.99	het	unkown	1	NDD+ other	(+) Hearing impairment,(+) Delayed speech and language development,(+) Motor delay,(+) Severe global developmental delay
NAV3	ENST00000397909:c.6782G>A p.Trp2261Ter		unknown	6.82	het	unkown	3	NDD	(+) Microcephaly,(+) Delayed speech and language development,(+) Moderate global developmental delay
NCOR1	ENST00000268712:c.1279C>G p.Pro427Ala		de_novo	8.27	het	de novo	1	NDD + other	myopia, behavioural abnormality, compulsive behaviour, pectus excavatum, ataxia, moderate intellectual disability, patellar dislocation
FBH1	ENST00000362091:c.2474T>C p.Val825Ala		de_novo	B	het	de novo	1	other	Fallot-Tetralogy, Ptosis, Anosmia
CSMD1	ENST00000335551:c.2084T>C p.Leu695Pro		de_novo	7.47	het	de novo	3	NDD+epile psy	Intellectual disability, Seizure, Global developmental delay, Generalized hypotonia, Bilateral tonic-clonic seizure, Neurodevelopmental delay, Epileptic encephalopathy, Intellectual disability, severe, Myoclonic absence seizure, Interictal epileptiform activity, EMG: myotonic discharges
DPY19L4	ENST00000414645:c.1870C>T p.Arg624Ter	ENST00000414645:c.1256C>T p.Ser419Phe	AR_comphet	4.17	comphet	paternal/maternal	3	NDD+epile psy	Intellectual disability, Seizure, Global developmental delay, Generalized hypotonia, Bilateral tonic-clonic seizure, Neurodevelopmental delay, Epileptic encephalopathy, Intellectual disability, severe, Myoclonic absence seizure, Interictal epileptiform activity, EMG: myotonic discharges
MFAP1	ENST00000267812:c.88T>C p.Ser30Pro		de_novo	6.45	het	de novo	3	NDD+epile psy	Intellectual disability, Seizure, Global developmental delay, Generalized hypotonia, Bilateral tonic-clonic seizure, Neurodevelopmental delay, Epileptic encephalopathy, Intellectual disability, severe, Myoclonic absence seizure, Interictal epileptiform activity, EMG: myotonic discharges
ARHGAP23	ENST00000616767:c.1384C>T		de_novo	5.35	het	de novo	2	Epilepsy	Generalized non-motor (absence) seizure
GRM4	ENST00000374177:c.1302C>G p.Tyr434Ter		unknown	8.34	het	unkown	1	Epilepsy	(+) Focal-onset seizure,(+) Focal cortical dysplasia type I
ERG	ENST00000288319:c.1338del p.Phe446LeufsTer59		de_novo	9.9	het	de novo	1	NDD	hypotonia, muscle weakness, progressive spastic paraplegia, pes cavus
ARHGEF7	ENST00000218789:c.973C>T p.Leu325Phe		unknown	6.58	het	unknown		NDD+Epile psy	(+) Psychotic episodes,(+) Intellectual disability,(+) Focal-onset seizure,(+) Focal polymicrogyria,(+) Schizophrenia
HDLBP	ENST00000310931:c.1981C>A p.Pro661Thr		unknown	6.23	het	unknown		NDD+ Adipositas	(+) Abnormality of the face,(+) Abnormal repetitive mannerisms,(+) Intellectual disability,(+) Global developmental delay,(+) Overweight
SUMO1	ENST00000392244:c.90+1G>A None		unknown	8.07	het	unknown		NDD	(+) Global developmental delay,(+) Neurodevelopmental delay,(+) Cognitive impairment
PCSK5	ENST00000424854:c.3819C>A p.Cys1273Ter		homo	9.06	homo	unknown	1	NDD	(+) Abnormality of the face,(+) Autism,(+) Sleep disturbance,(+) Supraventricular tachycardia,(+) Severe global developmental delay
PUM2	ENST00000338086:c.3067C>G p.Arg1023Gly		unknown	6.37	het	unknown		DLD	(+) Tall stature,(+) Abnormal repetitive mannerisms,(+) Delayed speech and language development,(+) Overgrowth,(+) Moderate global developmental delay
EP400	ENST00000389561:c.5512del p.Gln1838SerfsTer17		unknown	8.34	het	maternal	1	NDD+Epile psy	(+) Microcephaly,(+) Generalized non-motor (absence) seizure,(+) Intellectual disability, borderline,(+) Mild global developmental delay
RNF157	ENST00000269391:c.721-1G>A		homo	8.42	homo	paternal/maternal	1	NDD	Macrocephaly, autistic behavior, absent speech, moderate global developmental delay
PLXNA3	ENST00000369682:c.4372G>A p.Glu1458Lys		XL	6.37	hemi	maternal	1	NDD + epilepsy	Microcephaly, absent speech, EEG abnormality, intellectual disability severe, severe global developmental delay, bilateral tonic-clonic seizure with generalized onset, arm dystonia
PHACTR1	ENST00000332995:c.497-1G>C None		unknown	8.23	het	unknown	1	Epilepsy	(+) Hypotonia,(+) Abnormal cerebral morphology,(+) Focal-onset seizure
EPHB1	ENST00000398015:c.2570T>C p.Met857Thr		de_novo	7.77	het	de novo	2	NDD + other	drug-resistant generalized-onset seizure, mild global developmental delay
CELF2	ENST00000354897:c.604_624dup p.Ala202_Leu208dup		ad_inherited	4.93	het	maternal	1	NDD + epilepsy + other	moderate global developmental delay, intellectual disability, autistic behaviour, peripheral axonal neuropathy, focal-onset seizure since age of 6

PIM2			de_novo	7.05	het	de novo	1	NDD + epilepsy + other	severe intellectual disability, behavioural abnormalities, autistic features, pes planus, difference in leg length left < right, infantile spasm, short stature (150 cm; 3 cm below 3rd percentile), facial dysmorphism: high forehead, low-set and posteriorly rotated ears, strabismus, 2-3 toe syndactyly; cMRI: delayed myelination
TRAF3	ENST00000351691:c.1078_1094del p.Leu360AspfsTer63		de_novo	9.09	het	de novo	2	NDD + other	mild developmental delay/ delayed gross motor development, short stature, global muscu-lar hypotonia, FLG-associated atopic dermatitis, hip dysplasia, talipes calcaneovalgus, nasal poly-posis, food allergy
INTS8	ENST00000523731:c.2295+3A>G		de_novo	4.92	het	de novo	2	NDD + other	Seizure, global developmental delay, bilateral tonic-clonic seizure, myoclonic seizure
IL13RA1	ENST00000371642:c.553T>A		x_linked	4.22	hemi	maternal	2	NDD + other	Seizure, global developmental delay, bilateral tonic-clonic seizure, myoclonic seizure
HERC1	ENST00000443617:c.5122_5132del p.Asp1708SerfsTer3		de_novo	12.2	het	de novo	1	NDD	Delayed speech and language development, hearing impairment, aggressive behavior, self-injurious behavior
ILDR2	ENST00000271417:c.242G>T p.Ser81Ile		de_novo	5.41	het	de novo	2	NDD	autistic behavior, global developmental delay, delayed speech and language development
ZFHX4	ENST00000518282:c.5208dup p.Gly1737TrpfsTer14		unknown	6.67	het	not paternal	1	NDD	(+) Moderate global developmental delay
ITIH2	ENST00000358415:c.1721A>G p.His574Arg		homo	4.47	homo	maternal/ paternal	2	NDD	autistic behavior, global developmental delay, delayed speech and language development
WDR33	ENST00000322313:c.3691C>T p.Arg1231Ter		de_novo	9.16	het	de novo	3	NDD + epilepsy	Microcephaly, Nystagmus, Hypotonia, Status epilepticus, EEG abnormality, Severe global developmental delay
SPATA13	ENST00000343003:c.1724C>T p.Ala575Val		homo	6.52	homo	maternal/ paternal	3	NDD + epilepsy	Microcephaly, Nystagmus, Hypotonia, Status epilepticus, EEG abnormality, Severe global developmental delay
NOP56	ENST00000329276:c.862A>G p.Lys288Glu		homo	7.68	homo	maternal/ paternal	3	NDD + epilepsy	Microcephaly, Nystagmus, Hypotonia, Status epilepticus, EEG abnormality, Severe global developmental delay
FAM9C	ENST00000333995:c.62-6A>G		XL	6	hemi	maternal	1	NDD + epilepsy	Seizure, Bilateral tonic-clonic seizure, Generalized myoclonic seizure, Atonic seizure, Epileptic spasm, Epileptic encephalopathy, Autism, Moderate global developmental delay
HECTD1	ENST00000399332:c.1238A>C p.Glu413Ala		unknown	5.89	het	unkown		NDD + epilepsy	(+) Autism,(+) Intellectual disability,(+) Focal-onset seizure,(+) Spastic hemiparesis,(+) Severe global developmental delay
SRCIN1	ENST00000617146:c.665T>A		de_novo	7.29	het	de novo	1	NDD + epilepsy	Hypotonia, generalized-onset seizure, focal-onset seizure, severe global developmental delay, hippocampal sclerosis
UBC	ENST00000339647:c.224G>A p.Gly75Asp		de_novo	6.26	het	de novo	1	NDD+other	Focal-onset seizure, moderate global developmental delay, grade IV vesicoureteral reflux, generalized hypotonia, atrial septal defect, micropenis
TTC30B	ENST00000408939:c.408dup p.Glu137ArgfsTer6		unknown	6.23	het	unknown	1	NDD+epilepsy+ataxia	(+) Abnormal saccadic eye movements,(+) Nystagmus,(+) Ataxia,(+) Encephalopathy,(+) Focal-onset seizure,(+) Neurodevelopmental delay,(+) Cognitive impairment
C2CD2L	ENST00000336702:c.1949G>T p.Arg650Leu		de_novo	5.93	het	de novo	1	NDD + other	global developmental delay, delayed speech and language development, atypical behaviour, gait disturbance, cerebellar gliosis
CCNI	ENST00000237654:c.1080_1081insT p.Gln361SerfsTer31		unknown	6.98	het	unknown	1	Epilepsy	(+) Absence seizure with eyelid myoclonia
MYOF	ENST00000358334:c.5321+1G>A	ENST00000358334:c.4673A>G p.Asn1558Ser	AR_comphet	5.24	comphet	maternal/ paternal	1		Seizure, Hypsarrhythmia, Mild global developmental delay, Infantile spasms
RGS6	ENST00000343854:c.1207-1G>A None		ad_inherited	5.97	het	paternal	2	other	mixed demyelinating and axonal polyneuropathy, tremor, migraine with aura, pes cavus, ulnar claw
DNAJC1	ENST00000376980:c.356_357del p.Asp119GlyfsTer7		ad_inherited	5.07	het	paternal	2	other	mixed demyelinating and axonal polyneuropathy, tremor, migraine with aura, pes cavus, ulnar claw
KIAA1958	ENST00000337530:c.1879C>T p.Gln627Ter		unknown	4.66	het	unknown	3	other	(+) Polyneuropathy,(+) Gait disturbance,(+) Lower limb hyperreflexia,(+) Muscle spasm,(+) Progressive spastic paraplegia,(+) Stress urinary incontinence,(+) Fatigue,(+) Restless legs,(+) Spondylitis,(+) Hypoesthesia
PSD	ENST00000020673:c.1537del p.Ala513GlnfsTer10		unknown	7.97	het	unknown	2	Epilepsy	(+) Migraine with aura,(+) Generalized-onset seizure,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Bilateral tonic-clonic seizure with generalized onset,(+) Seizure precipitated by febrile infection
FIGN	ENST00000333129:c.1624C>T p.Gln542Ter		de_novo	9.09	het	de novo	4	NDD	global developmental delay, intellectual disability, visual impairment, enuresis
PKN1	ENST00000242783:c.2766_2767insT p.Pro923SerfsTer?		unknown	6.74	het	unknown		Epilepsy	(+) Bilateral tonic-clonic seizure,(+) Generalized non-motor (absence) seizure,(+) Episodic ataxia,(+) Generalized-onset seizure,(+) Paroxysmal vertigo

C4orf50	ENST00000531445:c.496A>T p.Lys166Ter		AR_homo	7.5	homo	maternal/ paternal	4	NDD	global developmental delay, intellectual disability, visual impairment, enuresis
TCP1	ENST00000321394:c.1112C>T p.Thr371Met		AR_homo	6.4	homo	maternal/ paternal	4	NDD	global developmental delay, intellectual disability, visual impairment, enuresis
ADHFE1	ENST00000396623:c.1097C>T p.Pro366Leu		AR_homo	6.78	homo	maternal/ paternal	4	NDD	global developmental delay, intellectual disability, visual impairment, enuresis
HSPA12B	ENST00000254963:c.1714G>C p.Asp572His		de_novo	5.88	het	de novo		other	(+) Ventricular septal defect,(+) Atrioventricular canal defect,(+) Abnormal fetal cardiovascular morphology,(+) Anomalous origin of the right subclavian artery from the descending aorta
PAPSS1	ENST00000265174:c.1051C>T p.Arg351Cys		homo	5.66	homo	mater/pat ernal	1	NDD + other	muscular hypotonia, motor delay, nystagmus, facial dysmorphism (high palate, low-set ears, tented philtrum), turricephaly, lacticaciduria, methylmalonic aciduria, increased urine succinate level, cafe au lait spot (3 cm, on the lower back), genital hypoplasia
FBN3	ENST00000270509:c.3887C>G p.Ser1296Cys	ENST00000270509:c.697C>G p.Arg233Gly	AR_comphet	3.7	comphet	maternal/ paternal	1	NDD	(+) High palate,(+) Brachycephaly,(+) Microcephaly,(+) Pointed chin,(+) Triangular face,(+) Low-set ears,(+) Hypotonia,(+) Failure to thrive,(+) Patent foramen ovale,(+) Iron deficiency anemia,(+) Moderate global developmental delay,(+) Feeding difficulties,(+) Tube feeding
GOT1	ENST00000370508:c.761C>T p.Ala254Val	ENST00000370508:c.239T>C p.Phe80Ser	comphet	5.48	comphet	maternal/ paternal	1	NDD + other	mild global developmental delay (since age of 5 years), microcephaly, hypertelorism, sen-sorineural hearing impairment, myopia (-4.5 dpt), decreased body weight, depressed nasal bridge, swan neck-like deformities of the fingers, recurrent pneumonia, short 5th finger, abnormal protein n-linked glycosylation
DOCK9	ENST00000427887:c.734G>A		homo	5.27	homo	maternal/ paternal	2	NDD + epilepsy	Moderate global developmental delay, complex febrile seizure, seizure, intellectual disability, bilateral tonic-clonic seizure
MFSD5	ENST00000329548:c.1310G>A		homo	4.2	homo	maternal/ paternal	2	NDD + epilepsy	Moderate global developmental delay, complex febrile seizure, seizure, intellectual disability, bilateral tonic-clonic seizure
PCSK5	ENST00000376752:c.536C>G p.Pro179Arg		unknown	5.67	het	unknown	2	NDD + epilepsy + other	intellectual disability, focal-onset seizure, hemimegalencephaly, focal cortical dysplasia, autistic features, muscular hypotonia, hemiparesis on the right side, unilateral visual im-pairment, nystagmus, divergent strabismus, astigmatism, scoliosis, pes valgus, small hand, small thin upper and lower lip vermilion, abnormality of the dentition
CEP350	ENST00000367607:c.5363C>G p.Thr1788Ser		unknown	4.99	het	unknown	2	NDD + epilepsy + other	intellectual disability, focal-onset seizure, hemimegalencephaly, focal cortical dysplasia, autistic features, muscular hypotonia, hemiparesis on the right side, unilateral visual im-pairment, nystagmus, divergent strabismus, astigmatism, scoliosis, pes valgus, small hand, small thin upper and lower lip vermilion, abnormality of the dentition
CAD	ENST00000264705:c.6212A>G p.Asn2071Ser		de_novo	7.5	het	de novo		NDD	(+) Short stature,(+) Global developmental delay,(+) Intellectual disability,(+) Microcephaly
ADAMDEC1	ENST00000256412:c.321C>G p.Tyr107Ter		unknown	B	het	unknown	3	other	immunodeficiency, recurrent respiratory infections, recurrent pneumonia, recurrent fever, increased circulating IgG level, pilocytic astrocytoma at the age of 5 1/2 months, hydrocephalus occlusus
PROSER2	ENST00000277570:c.621dup p.Asn208GlufsTer120		unknown	B	het	unknown	3	other	immunodeficiency, recurrent respiratory infections, recurrent pneumonia, recurrent fever, increased circulating IgG level, pilocytic astrocytoma at the age of 5 1/2 months, hydrocephalus occlusus
A1BG	ENST00000263100:c.1227del p.Trp409Ter		unknown	B	het	unknown	3	other	immunodeficiency, recurrent respiratory infections, recurrent pneumonia, recurrent fever, increased circulating IgG level, pilocytic astrocytoma at the age of 5 1/2 months, hydrocephalus occlusus
TENM4	ENST00000278550:c.6892C>T p.Arg2298Trp		unknown	6.3	het	unknown	1	Epilepsy	(+) Generalized-onset seizure,(+) Focal-onset seizure,(+) Myoclonic seizure
HNRNPL	ENST00000221419:c.1696C>T p.Gln566Ter		unknown	7.83	het	unknown	1	ID	(+) Intellectual disability,(+) Global developmental delay,(+) Asthma,(+) Scoliosis
GRIN2C	ENST00000293190:c.1326-9_1326-2del None		homo	12.1	homo	unknown	1	Epilepsie	(+) Status epilepticus,(+) EEG with focal epileptiform discharges,(+) Focal motor seizure with paresis/paralysis,(+) Cerebral cavernous malformation
KAZN	ENST00000376030:c.1358C>A p.Ala453Asp	ENST00000376030:c.1257C>A p.Ser419Arg	AR_comphet	3.7	comphet	paternal/ maternal	3	NDD	Atypical behavior, Autism, Aggressive behavior, Gait disturbance, Absent speech, Short stature, Severe global developmental delay, Abnormal skeletal muscle

TSPAN1	ENST00000372003:c.220G>A p.Gly74Ser		AR_homo	9.36	homo	maternal/ paternal	3	NDD	Atypical behavior, Autism, Aggressive behavior, Gait disturbance, Absent speech, Short stature, Severe global developmental delay, Abnormal skeletal muscle
KPNA2	ENST00000330459:c.1347+1G>T		AR_homo	9.19	homo	maternal/ paternal	3	NDD	Atypical behavior, Autism, Aggressive behavior, Gait disturbance, Absent speech, Short stature, Severe global developmental delay, Abnormal skeletal muscle
PSD3	ENST00000286485:c.1099G>A p.Ala367Thr		de_novo	6.33	het	de novo	2	NDD	moderate global developmental delay
NFASC	ENST00000404076:c.530T>A		AD_denovo	10.1	het	de novo	1	NDD+epilepsy	Microcephaly, seizure, intellectual disability, global developmental delay, absent speech, tetraparesis and short stature.
ATP6V1B2	ENST00000276390:c.1316_1318del p.Val439del		de_novo	8.49	het	de novo	1	NDD+Epilepsy	(+) Microcephaly,(+) Nystagmus,(+) Delayed eruption of permanent teeth,(+) Hypotonia,(+) Absent speech,(+) Hypoplasia of the corpus callosum,(+) Ventriculomegaly,(+) Hyperhomocystinemia,(+) Generalized-onset seizure,(+) Inability to walk,(+) Hypercholesterolemia,(+) Enamel hypoplasia,(+) Severe global developmental delay,(+) Feeding difficulties
TENM2	ENST00000518659:c.7883A>G p.Asp2628Gly		de_novo	6.68	het	de novo	1	NDD	moderate global developmental delay, autism
KCNA3	NM_002232.4:c.1430C>A, p.(Pro477His)		AD_denovo	4.64	het	de novo	1	NDD + epilepsy	(-) Infantile onset (+) Aggressive behavior (+) Atonic seizure (+) Delayed speech and language development (+) EEG abnormality (+) Focal seizure with eyelid myoclonia (+) Gait ataxia (+) Generalized non-motor (absence) seizure (+) Generalized-onset seizure (+) Global developmental delay (+) Poor speech (+) Seizure (+) Sleep disturbance (+) Spastic ataxia
PTPRD	ENST00000356435:c.2345_2349+1del None		unknown	9.64	het	unknown	1	epilepsy	focal onset seizures, onset with 6 years, male
OSBPL1A	ENST00000319481:c.1433A>T p.Asp478Val		de_novo	6.37	het	de novo	1	NDD + epilepsy	generalised-onset seizure, mild global developmental delay
SLC1A6	ENST00000221742:c.1475T>C p.Leu492Pro		unknown	5.52	het	unknown	1	NDD	(+) Macrocephaly,(+) Delayed speech and language development,(+) Global developmental delay
ATP6V1B2	ENST00000276390:c.488G>A p.Arg163Gln		unknown	7.75	het	unkown	1		Dysarthria,(+) Episodic ataxia,(+) Focal-onset seizure
NSF	ENST00000398238:c.1913G>C p.Arg638Pro		unknown	7.69	het	unkown	2	Epilepsy	(+) Seizure,(+) Bilateral tonic-clonic seizure,(+) Febrile seizure (within the age range of 3 months to 6 years)
TMEM131	ENST00000186436:c.2024C>G		homo	5.17	homo	maternal/ paternal	1	NDD + other	Motor and speech developmental delay, microcephaly, short philtrum, large ears, syndactyly of 2/3 toes bilateral
PARG	ENST00000402038:c.217+1G>A None		unknown	6.17	het	unkown	1		(+) Abnormality of skin pigmentation,(+) Nevus flammeus,(+) Wrist swelling,(+) Arthralgia/arthritis,(+) Mild global developmental delay
NRXN3	ENST00000335750:c.1814A>G p.Tyr605Cys		unknown	8.04	het	unknown	1	NDD, Autism	(+) Autism,(+) Delayed speech and language development,(+) Global developmental delay,(+) Oromotor apraxia,(+) Severe global developmental delay
APBA3	ENST00000316757:c.616G>A p.Val206Met	ENST00000316757:c.574C>T p.Gln192Ter	AR_comphet	5.23	comphet	maternal/ paternal	2	NDD + other	mild developmental delay/ delayed gross motor development, short stature, global muscular hypotonia, FLG -associated atopic dermatitis, hip dysplasia, talipes calcaneovalgus, nasal polyposis, food allergy
LENG8	ENST00000326764:c.721C>T p.Arg241Ter		unknown	6.32	het	unknown	1	NDD + epilepsy	(+) Generalized myoclonic-atonic seizure,(+) Mild global developmental delay,(+) Myoclonic seizure,(+) Epileptic encephalopathy
ADGRB1	ENST00000517894:c.206_215del p.Ser69TyrfsTer13		unknown	7.06	het	unknown	1	NDD + autism	(+) Autism,(+) Intellectual disability, mild,(+) Intellectual disability, moderate,(+) Mild global developmental delay
ADCY2	ENST00000338316:c.3059T>C p.Ile1020Thr		unknown	6.34	het	unknown	2		(+) Abnormality of eye movement,(+) Bilateral ptosis,(+) Moderate global developmental delay,(+) Abnormal optic disc morphology
EP400	ENST00000389561:c.5585-2A>G None		unknown	8.34	het	paternal	2		(+) Abnormality of eye movement,(+) Bilateral ptosis,(+) Moderate global developmental delay,(+) Abnormal optic disc morphology
TRAPPC8	ENST00000283351:c.3189-2A>G None		unknown	8.03	het	unknown	1	severe NDD+Epilepsy	(+) Microcephaly,(+) Macrotia,(+) Seizure,(+) EEG abnormality,(+) Severe global developmental delay,(+) Epileptic encephalopathy
ATP6V0B	ENST00000236067:None None		homo	6.21	het	unknown	1		(+) Microcephaly,(+) Intellectual disability,(+) Seizure,(+) Short stature,(+) Moderate global developmental delay
RXRB	ENST00000374680:c.766C>T p.Arg256Cys		unknown	4.77	het	unknown	1		(+) Microcephaly,(+) Global developmental delay
KCNH4	ENST00000264661:c.1118C>T p.Ala373Val		unknown	3.88	het	unknown	2	Epilepsy	(+) Seizure,(+) Generalized non-motor (absence) seizure
APLP1	ENST00000221891:c.981+1G>A None		unknown	7.67	het	unkown	1	NDD	(+) Hypotonia,(+) Specific learning disability,(+) Mild global developmental delay
CPEB4	ENST00000265085:c.1427T>G p.Val476Gly		unknown	4.79	het	unkown	1	other	(+) Hydrocephalus,(+) Global developmental delay,(+) Lower limb spasticity,(+) Ventriculomegaly,(+) Progressive muscle weakness,(+) Short stature,(+) Periventricular leukomalacia
KDM5A	ENST00000399788:c.3617A>C p.Lys1206Thr		unknown	6.09	het	unkown	1	other	(+) Obesity,(+) Polyphagia
TTC7B	ENST00000328459:c.2281C>T p.Pro761Ser		unknown	3.39	het	unkown	1	NDD + epilepsy	(+) Global developmental delay,(+) Focal impaired awareness seizure,(+) Myoclonic spasms,(+) Short stature,(+) Focal-onset seizure
ACAP2	ENST00000326793:c.669+2T>G None		unknown	6.78	het	unkown	1	NDD+Autism	(+) Autism,(+) Developmental regression,(+) Expressive language delay,(+) Receptive language delay,(+) Severe global developmental delay

PIP5K1C	ENST00000335312:c.1640_1641insCTCC p.Arg548SerfsTer63		homo	10.7	homo	unkonwn	2	NDD + epilepsy	(+) Hypotonia,(+) Focal-onset seizure,(+) Moderate global developmental delay
NR1D2	ENST00000312521:c.1626A>C p.Lys542Asn		unknown	3.15	homo	unknown	1	hypogonadism	(+) Female hypogonadism,(+) Hypogonadism,(+) Intellectual disability, mild,(+) EEG abnormality
MON2	ENST00000393629:c.2768G>C p.Arg923Pro		unknown	4.86	het	unkonwn	1	NDD + other	(+) Progressive microcephaly,(+) Retinal detachment,(+) Microphthalmia,(+) Blindness,(+) Optic atrophy,(+) Hypotonia,(+) Failure to thrive,(+) Kyphosis,(+) Short stature,(+) Simplified gyral pattern,(+) Severe global developmental delay,(+) Primary microcephaly,(+) Abnormality of brain morphology,(+) Brain imaging abnormality,(+) Abnormality in emotional social interactions
CHD6	ENST00000373233:c.7336C>G p.Pro2446Ala		unknown	5.87	het	unkonwn	1	NDD + other	(+) Progressive microcephaly,(+) Retinal detachment,(+) Microphthalmia,(+) Blindness,(+) Optic atrophy,(+) Hypotonia,(+) Failure to thrive,(+) Kyphosis,(+) Short stature,(+) Simplified gyral pattern,(+) Severe global developmental delay,(+) Primary microcephaly,(+) Abnormality of brain morphology,(+) Brain imaging abnormality,(+) Abnormality in emotional social interactions
RBM15B	ENST00000563281:c.157del p.Ala53ProfsTer94		unknown	5.78	het	unkonwn	1	other	(+) Patent ductus arteriosus,(+) Dyspnea,(+) Respiratory distress,(+) Laryngeal edema,(+) Bilateral vocal cord paresis,(+) Abnormal larynx morphology,(+) Extremely preterm birth,(+) Abnormal gastrointestinal motility
OSBP	ENST00000263847:c.1856del p.Phe619SerfsTer5		unknown	6.91	het	unkonwn	1	NDD+ Epilepsy	(+) Intellectual disability,(+) Focal-onset seizure,(+) Moderate global developmental delay,(+) Focal cortical dysplasia type IIb
SUPT6H	ENST00000314616:c.4507A>C		de_novo	7.13	het	de novo	1	other	Cutis laxa, abnormal facial shape, frontal bossing
SRPK2	ENST00000357311:c.1029_1030delinsACCATGA p.Glu344ProfsTer2		unknown	7.37	het	not maternal	1	NDD+Epilepsy	autistic behavior, seizure, intellectual disability, mild, global developmental delay, bilat-eral tonic-clonic seizure, epileptic spasm
DNAH1	ENST00000420323:c.3583C>T p.Gln1195Ter		unknown	7.06	het	unknown	1	NDD	(+) Low-set, posteriorly rotated ears,(+) Strabismus,(+) Autistic behavior,(+) Hypotonia,(+) Global developmental delay,(+) Gait disturbance,(+) Growth delay,(+) Abnormal foot morphology,(+) Scapular winging,(+) Abnormal aggressive, impulsive or violent behavior,(+) Tip-toe gait
LRRC41	ENST00000343304:c.2393C>G p.Ser798Ter		unknown	6.87	het	unknown	1	Epilepsy	(+) Status epilepticus,(+) EEG abnormality,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Focal-onset seizure
ATP6V0B	ENST00000236067:c.444dup p.Leu149SerfsTer10		unknown	7.8	het	unknown	1	Epilepsy	(+) Focal-onset seizure,(+) Marcus Gunn jaw winking synkinesis
CLVS1	ENST00000325897:c.978-2A>G None		unknown	5.81	het	unknown	1		(+) Focal-onset seizure,(+) Marcus Gunn jaw winking synkinesis
RBPJ	ENST00000342295:c.60-23747del None		de_novo	5.96	het	de novo	1	NDD+ Epilepsy	(+) Intellectual disability, mild,(+) Generalized-onset seizure
SACS	ENST00000382292:c.2775A>G p.Ala925=		de_novo	10.1	het	de novo		NDD	Combined developmental delay, dyscalculia, arterial hypertension, thyroid hypoplasia, con-genital bilateral ptosis, migraine, patent ductus ateriosus, VSD, downslanted palpebral fissures, strabismus divergens intermittens, hypermetropia, tip-toe gait, atopic eczema, asthma, preterm birth (36+0)
MAPK14	ENST00000229794:c.571C>G p.Pro191Ala		unknown	6.79	het	unknown	1		(+) Ataxia,(+) Abnormal corpus callosum morphology,(+) Abnormal heart morphology,(+) Focal myoclonic seizure,(+) Mild global developmental delay,(+) Delayed myelination
VSIG10	ENST00000359236:c.877dup p.Ser293LysfsTer24		homo	8.02	homo	mat and pat	1	NDD	(+) Microcephaly,(+) Abnormality of the face,(+) Pectus excavatum,(+) Hypotonia,(+) Atrial septal defect,(+) Dysphagia,(+) Short stature,(+) Moderate global developmental delay
PYGB	ENST00000216962:c.1451C>A p.Thr484Asn		homo	6.44	homo	mat and pat	1	NDD	(+) Microcephaly,(+) Abnormality of the face,(+) Pectus excavatum,(+) Hypotonia,(+) Atrial septal defect,(+) Dysphagia,(+) Short stature,(+) Moderate global developmental delay
ARID5B	ENST00000279873:c.192del p.Thr65ProfsTer38		unknown	7.63	het	unknown	1	other	(+) Joint laxity,(+) Abnormal foot morphology,(+) Skeletal muscle atrophy,(+) Exercise intolerance,(+) Abnormality of connective tissue,(+) Proximal muscle weakness
PLXNB3	ENST00000361971:c.5093C>T p.Ser1698Phe		de_novo	7.5	het	de novo	1	NDD	Strabismus, Thick eyebrow, Optic atrophy, Autism, Motor delay, Sandal gap, entriculomegaly
TAOK2	ENST00000279394:c.671C>T p.Pro224Leu		unknown	5.95	het	unknown	1	epilepsy	(+) Seizure,(+) Intellectual disability, mild,(+) Focal impaired awareness seizure,(+) Atypical absence seizure,(+) Epileptic spasm,(+) Tonic seizure
UBR4	ENST00000375254:c.11893-1_11900del None		unknown	9.9	het	unknown	1	epilepsy	(+) Seizure,(+) Intellectual disability, mild,(+) Focal impaired awareness seizure,(+) Atypical absence seizure,(+) Epileptic spasm,(+) Tonic seizure
CADPS	ENST00000283269:c.888+17291A>G None		unknown	C	het	unknown	1	Ataxia+ other	,Ataxia, Cerebellar atrophy, Cerebellar hypoplasia, Leukoencephalopathy, Leukodystrophy, Seizure
CAMKMT	ENST00000378494:c.376+116522G>T None		de_novo	6.49	het	de novo	1	NDD	inguinal hernia, cryptorchidism, hypospadias, renal cyst, hemangioma, bicuspid aortic valve, coarctation of aorta, anal atresia, choroid plexus cyst, abnormal vertebral mor-phology, multiple renal cysts, global developmental delay, short stature, failure to thrive, hearing impairment

TKT	ENST00000296289:c.1555+1G>A None		de_novo	7.91	het	de novo		NDD	moderate/severe global developmental delay, macrocephaly, ataxia, muscular hypotonia, agenesis of corpus callosum, periventricular heterotopia, generalized-/focal-onset seizure, hip dysplasia
CLEC6A	ENST00000382073:c.223G>C p.Ala75Pro		homo	3.47	hom	maternal&paternal		NDD	moderate/severe global developmental delay, macrocephaly, ataxia, muscular hypotonia, agenesis of corpus callosum, periventricular heterotopia, generalized-/focal-onset seizure, hip dysplasia
LINC02641	ENST00000448347:n.746+8175G>A None		de_novo	3	het	de novo	1	NDD	severe global developmental delay, tall stature, hypotonia, obesity
DDX5	ENST00000450599:c.1217-440G>A None		de_novo	5.16	het	de novo	1	NDD	severe global developmental delay, tall stature, hypotonia, obesity
CALU	ENST00000249364:c.558del p.Tyr187ThrfsTer2		unknown	6.5	het	unknown	1	NDD	(+) Autism,(+) Brachydactyly,(+) Global developmental delay,(+) Pectoralis hypoplasia,(+) Upper limb undergrowth
TTC28	ENST00000397906:c.4238G>T p.Gly1413Val		unknown	4.03	het	unknown	1	NDD	allgemeine, moderate Entwicklungsverzögerung, V.a. Autismus, Poland-Syndrom mit verkürzten Arm, Brachydaktylie und Pectoralishypoplasie, Chromosomen und Array unauffällig
GRM5	ENST00000305432:c.3448T>C p.Ser1150Pro		unknown	7.59	het	unknown	1	NDD+hearing loss	(+) Strabismus,(+) Frontal bossing,(+) Abnormal nasal morphology,(+) Mild global developmental delay,(+) Childhood onset sensorineural hearing impairment
DGKH	ENST00000261491:c.2305T>C p.Cys769Arg		unknown	5.53	het	unknown	1	NDD	(+) Mutism,(+) Severe expressive language delay,(+) Clinodactyly
DPYSL2	ENST00000311151:c.163G>A p.Val55Met		unknown	7.31	het	unknown	1	Epilepsy	(+) Hypertension,(+) Seizure,(+) Intellectual disability, mild,(+) Bilateral tonic-clonic seizure,(+) Status epilepticus,(+) Focal impaired awareness seizure,(+) Tension-type headache,(+) Seizure precipitated by febrile infection,(+) Hippocampal sclerosis
GCN1	ENST00000300648:c.4397T>C p.Leu1466Pro		unknown	4	het	unknown	1	NDD	schwere Entwicklungsverzögerung, muskuläre Hypotonie, West-Syndrom bei Astrozytom
RASSF9	ENST00000361228:c.423G>A p.Trp141Ter		homo	7.56	hom	maternal&paternal	2	NDD	(+) Microcephaly,(+) Short attention span,(+) Delayed speech and language development,(+) Failure to thrive,(+) Expressive language delay,(+) Receptive language delay,(+) Moderate global developmental delay,(+) Diminished ability to concentrate,(+) Cognitive impairment,(+) Abnormal social emotional interactions
DMXL1	ENST00000311085:c.4594+1G>T, p.?		unknown	7.1	het	unknown	1	NDD+epilepsy	(+) Microcephaly,(+) Short stature,(+) Focal-onset seizure,(+) Moderate global developmental delay,(+) Abnormal growth hormone level
DOCK7	ENST00000251157:c.1872G>A p.Arg624=		unknown	8.35	het	unknown	1	NDD+epilepsy	(+) Seizure,(+) Hypsarrhythmia,(+) Focal-onset seizure,(+) Moderate global developmental delay,(+) Epileptic encephalopathy
BAIAP2	ENST00000321280:c.843del p.Glu282SerfsTer18		unknown	8.54	het	unknown	1	NDD	(+) Intellectual disability,(+) Obesity,(+) Increased body mass index
PDS5A	ENST00000303538:c.259C>T p.Arg87Cys		unknown	6.66	het	unknown	1	NDD	severe intellectual disability, short stature
PTPRM	ENST00000332175:c.3191A>G p.His1064Arg		unknown	4.97	het	unknown	1	NDD+epilepsy	(+) Optic atrophy,(+) Single transverse palmar crease,(+) Absent septum pellucidum,(+) Generalized-onset seizure,(+) Short stature,(+) Abnormality of the palpebral fissures,(+) Mild global developmental delay,(+) Septo-optic dysplasia
ARHGEF7	ENST00000218789:c.356A>G p.Tyr119Cys		ad_inherited	6.61	het	maternal	1	NDD	Intelligenzminderung, Lipödem Stadium 3-4
SLIT1	ENST00000266058:c.4418G>A p.Gly1473Asp		unknown	6.17	het	unknown	1	Fehlbildungen	Hypospadias, Unilateral renal agenesis, Hemangioma, Syringomyelia, Lipomyelomeningocele, Skin ulcer
TRAPPC8	ENST00000283351:c.1708C>T p.Arg570Ter		unknown	7.53	het	unknown	1	NDD+epilepsy	(+) Seizure,(+) Global developmental delay
SUCO	ENST00000263688:c.1222_1223del p.Ser408PhefsTer5		unknown	7.38	het	unknown	1	NDD+epilepsy	(+) Retrognathia,(+) Abnormal sternum morphology,(+) Hypotonia,(+) Generalized myoclonic seizure,(+) Generalized-onset seizure,(+) Developmental regression,(+) Short stature,(+) Atypical absence seizure,(+) Severe global developmental delay,(+) Bilateral tonic-clonic seizure with generalized onset,(+) Epileptic encephalopathy
SEC24B	ENST00000265175:c.1924_1925delinsG p.Asn642AlafsTer43		unknown	8.64	het	unknown	1	epilepsy	(+) Seizure,(+) Status epilepticus,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Febrile status epilepticus
LRRC7	ENST00000310961:c.3167dup p.Gln1058SerfsTer20		unknown	8.58	het	unknown		NDD	(+) Microcephaly,(+) Epicanthus,(+) Preauricular skin tag,(+) Upslanted palpebral fissure,(+) Delayed speech and language development,(+) Hyperactivity,(+) Gait disturbance,(+) Moderate global developmental delay,(+) Overfriendliness
HIF1A	ENST00000323441:c.2180C>G p.Ser727Ter		unknown	9.07	het	unknown	1	epilepsy	(+) Bilateral tonic-clonic seizure,(+) EEG abnormality,(+) Febrile seizure outside the age of 3 months to 6 years
ATG9A	ENST00000361242:c.2229del p.Asp744MetfsTer100		unknown	7.2	het	unknown	1	ataxia	(+) Gaze-evoked nystagmus,(+) Ataxia,(+) Gait disturbance,(+) Episodic ataxia,(+) Headache,(+) Paroxysmal vertigo
UNC5D	ENST00000287272:c.1560-2A>C None		unknown	8.69	het	unknown	1	Neuropathy	(+) Postural instability,(+) Paraparesis,(+) Spondylolisthesis,(+) Low back pain,(+) Sensorimotor neuropathy,(+) Peripheral demyelination,(+) Pain,(+) Neuralgia,(+) Hypoesthesia
TTL	ENST00000233336:c.740_741del p.Lys247ArgfsTer10		unknown	7.7	het	unknown	1	NDD	(+) Delayed speech and language development,(+) Hypotonia,(+) Global developmental delay,(+) Motor delay,(+) Muscle weakness,(+) Hepatomegaly,(+) Elevated circulating hepatic transaminase concentration,(+) Elevated circulating creatine kinase concentration
XPR1	ENST00000367589:c.385A>T p.Lys129Ter		unknown	6.82	het	unknown	1	epilepsy	(+) Generalized-onset seizure

TRANK1	ENST00000429976:c.4411C>T p.His1471Tyr		unknown	4.54	het	unknown	1	NDD+obesity	(+) Atypical behavior,(+) Aggressive behavior,(+) Gynecomastia,(+) Atopic dermatitis,(+) Global developmental delay,(+) Overweight,(+) Abnormal eating behavior
GRM4	ENST00000374177:c.419T>C p.Met140Thr		maternal	6.45	het	maternal	1	NDD	(+) Synophrys,(+) Intellectual disability, mild,(+) Global developmental delay,(+) Pes cavus,(+) Decreased body weight,(+) Attention deficit hyperactivity disorder
UHRF2	ENST00000276893:c.1240A>G p.Lys414Glu		unknown	4.05	het	unknown	2	Polyneuropathy	(+) Tall stature,(+) Polyneuropathy
PLXNA4	ENST00000321063:c.577_582dup p.Pro193_Glu194dup		de_novo	6.8	het	unknown	1	other	(+) Patent ductus arteriosus,(+) Dyspnea,(+) Respiratory distress,(+) Laryngeal edema,(+) Bilateral vocal cord paresis,(+) Abnormal larynx morphology,(+) Extremely preterm birth,(+) Abnormal gastrointestinal motility
PRKAR1B	ENST00000360274:c.494T>C p.Ile165Thr		unknown	5.38	het	unknown	2	obesity	obesity
BBX	ENST00000325805:c.78_79insAA p.Gln27AsnfsTer7		unknown	6.58	het	unknown	1	NDD+epilepsy	(+) Autism,(+) Generalized-onset seizure,(+) Focal-onset seizure,(+) Moderate global developmental delay
MPRIIP	ENST00000313485:c.1045C>T p.Gln349Ter		unknown	7.77	het	unknown	1	epilepsy	(+) Atypical behavior,(+) Focal-onset seizure,(+) Focal tonic seizure,(+) Nocturnal seizures,(+) Selective mutism
MINK1	ENST00000347992:c.3552_3571dup p.Asn1191MetfsTer54		AD_unknown	7.94	het	unknown	1	NDD+epilepsy	(+) Generalized-onset seizure,(+) EEG abnormality,(+) Mild global developmental delay
CAMK1D	ENST00000378845:c.1035del p.Asp346ThrfsTer5		unknown	7.54	het	unknown	1	other	(+) Recurrent bacterial infections,(+) Recurrent infections,(+) Immunodeficiency,(+) Decreased circulating antibody level
ATP11B	ENST00000323116:c.2830C>G p.Pro944Ala		unknown	3.82	het	unknown	1	NDD	(+) Thick upper lip vermillion,(+) Microcephaly,(+) Oval face,(+) Short philtrum,(+) Micrognathia,(+) Upslanted palpebral fissure,(+) Atypical behavior,(+) Delayed speech and language development,(+) Hypertrichosis,(+) Growth delay,(+) Mild global developmental delay
KIF1A	ENST00000320389:c.3080A>G p.His1027Arg		unknown	9.35	het	unknown	2	migraine	(+) Migraine,(+) Episodic hemiplegia
FRMD4A	ENST00000357447:c.2324C>A p.Ser775Ter		unknown	7.71	het	unknown	1	NDD	(+) Tall stature,(+) Autism,(+) Intellectual disability,(+) Moderate global developmental delay
TNK2	ENST00000333602:c.3022del p.Glu1008SerfsTer128		unknown	7.01	het	unknown	1	NDD+epilepsy	(+) Autism,(+) Generalized-onset seizure,(+) Focal-onset seizure,(+) Moderate global developmental delay
HNRNPA0	ENST00000314940:c.689G>A p.Gly230Asp		unknown	3.9	het	unknown	1	other	(+) Functional abnormality of the bladder,(+) Vesicoureteral reflux,(+) Unilateral renal hypoplasia,(+) Primary obstructive megaureter
MAPK10	ENST00000359221:c.839G>A p.Gly280Glu		unknown	6.22	het	unknown	1	NDD+epilepsy	(+) High palate,(+) Atypical behavior,(+) Global developmental delay,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Scoliosis,(+) Pes valgus
PCDH9	ENST00000328454:c.1906G>C p.Glu636Gln		unknown	6.9	het	unknown	1	NDD	(+) Autistic behavior,(+) Delayed speech and language development,(+) Mild global developmental delay
GIT1	ENST00000225394:c.174del p.Thr59ArgfsTer129		unknown	10.4	het	unknown	1		(+) Microcephaly,(+) Strabismus,(+) Motor delay,(+) Failure to thrive,(+) Short stature
BIRC6	ENST00000421745:c.9226C>T p.Gln3076Ter		unknown	7.39	het	unknown	1		(+) Generalized-onset seizure,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Attention deficit hyperactivity disorder
RBM6	ENST00000266022:c.412G>A p.Gly138Arg		unknown	3.49	homo	unknown	1	NDD	(+) Hypotonia,(+) Absent speech,(+) Supernumerary nipple,(+) Profound global developmental delay
SEC24B	ENST00000265175:c.2126+2_2126+5del None		unknown	8.64	het	unknown	1	NDD+epilepsy	(+) Generalized non-motor (absence) seizure,(+) Intellectual disability, borderline,(+) Moderate global developmental delay
KLHL22	ENST00000328879:c.595del p.Leu199SerfsTer50		unknown	6.55	het	unknown	2	NDD+epilepsy	(+) Generalized non-motor (absence) seizure,(+) Intellectual disability, borderline,(+) Moderate global developmental delay
PIAS2	ENST00000324794:c.1347_1353dup p.Lys452ProfsTer3		unknown	6.55	het	unknown	1	NDD	(+) External ophthalmoplegia,(+) Ataxia,(+) Dysphagia,(+) Tetraparesis,(+) Developmental regression,(+) Cachexia
BTAF1	ENST00000265990:c.1264-2A>G None		unknown	8.57	het	unknown	1	NDD	(+) Microcephaly,(+) Epicanthus,(+) Strabismus,(+) Hypermetropia,(+) Delayed speech and language development,(+) Sacral dimple,(+) Intellectual disability, mild,(+) Global developmental delay,(+) Short stature,(+) Infantile muscular hypotonia,(+) Dimple chin,(+) Clinodactyly
LINC01398	ENST00000416581:?		de_novo	3	het	de novo			
NDUFA13	ENST00000512771:c.524del p.Gly175GluTer6		unknown	8.82	het	unknown	1	NDD	(+) Autism,(+) Hypertrichosis,(+) Feeding difficulties in infancy,(+) Mild global developmental delay
ATG4B	ENST00000396411:c.77_80del p.Gln26ArgfsTer28		unknown	6.83	het	unknown	1	NDD+epilepsy	(+) Bilateral tonic-clonic seizure,(+) Generalized non-motor (absence) seizure,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Mild global developmental delay

NC119	ENST00000301032:c.*236C>T None		homo	6.81	het	maternal&paternal		NDD+Autism	(+) Abnormal skeletal muscle morphology (+) Absent speech (+) Aggressive behavior (+) Autism (+) Behavioral abnormality (+) Gait disturbance (+) Severe global developmental delay (+) Short stature
NCBP1	ENST00000375147:c.1408C>T p.Pro470Ser		unknown	5.86	het	unknown	1	Immune	(+) Asthma,(+) Immunodeficiency,(+) Recurrent bronchitis,(+) Recurrent viral infections
CTPS2	ENST00000359276:c.245T>G p.Leu82Arg		unknown	4.78	het	unknown	1	Epilepsy	(+) Seizure,(+) Generalized non-motor (absence) seizure,(+) Reduced circulating vitamin B6 circulating
ARHGEF11	ENST00000361409:c.3337G>T p.Glu1113Ter		unknown	7.07	het	unknown	1	NDD+Autism	(+) Atypical behavior,(+) Autism,(+) Hypertrichosis,(+) Syndactyly,(+) Absent speech,(+) Phimosis,(+) Sandal gap,(+) Moderate global developmental delay
ZMYM4	ENST00000314607:c.3834C>A p.Cys1278Ter		unknown	6.11	het	unknown	1	NDD	(+) Macroglossia,(+) Thin upper lip vermilion,(+) Global developmental delay,(+) Obesity,(+) Sandal gap,(+) Deviation of the thumb,(+) Disturbed perception with a stimulus
TMEM132B	ENST00000299308:c.2026C>T p.Arg676Ter		unknown	5.59	het	unknown	1	Epilepsy	(+) Generalized non-motor (absence) seizure,(+) EEG abnormality,(+) Atypical absence seizure
XPO7	ENST00000252512:c.1932G>C p.Thr644=		unknown	6.81	het	unknown	1	other	(+) Precocious puberty,(+) Acne,(+) Growth abnormality,(+) Clitoral hypertrophy,(+) Premature pubarche,(+) Increased serum testosterone level,(+) Ovarian neoplasm
MFN1	ENST00000263969:c.109dup p.Glu37GlyfsTer5		unknown	9.14	het	unknown	1	NDD+epilepsy	(+) Obesity,(+) Focal impaired awareness seizure,(+) Obstructive sleep apnea,(+) Focal-onset seizure,(+) Mild global developmental delay
BCLAF1	ENST00000353331:c.257_258del p.Tyr86SerfsTer6		unknown	6.59	het	unknown	1	NDD	(+) Delayed speech and language development,(+) Global developmental delay,(+) Absent speech,(+) No social interaction,(+) Abnormal social behavior
SEC62	ENST00000337002:c.420_421del p.Lys143ArgfsTer3		unknown	7.24	het	unknown	1	NDD + Immune	(+) Abnormality of the face,(+) Hypotonia,(+) Gait disturbance,(+) Anemia,(+) Unsteady gait,(+) Increased circulating antibody level,(+) Mild global developmental delay,(+) Abnormal brain morphology,(+) Abscess,(+) Systemic autoinflammation
SMG7	ENST00000347615:c.2082_2089del p.Phe695AlafsTer135		unknown	8.04	het	unknown	1	NDD + Immune	(+) Abnormality of the face,(+) Hypotonia,(+) Gait disturbance,(+) Anemia,(+) Unsteady gait,(+) Increased circulating antibody level,(+) Mild global developmental delay,(+) Abnormal brain morphology,(+) Abscess,(+) Systemic autoinflammation
TRIM33	ENST00000358465:c.1594dup p.Gln532ProfsTer24		unknown	7.84	het	unknown	1	NDD	(+) Upslanted palpebral fissure,(+) Intellectual disability,(+) Depressed nasal bridge,(+) Moderate global developmental delay
MYO18A	ENST00000527372:c.10dup p.Leu4ProfsTer52		unknown	7.65	het	unknown	1	NDD + epilepsy	(+) Seizure,(+) Intellectual disability, mild,(+) Generalized non-motor (absence) seizure,(+) Bilateral tonic-clonic seizure with generalized onset
ADGRB2	ENST00000373655:c.2463C>A p.Tyr821Ter		unknown	7.05	het	unknown	1	syndromal disease, ndd	(+) Narrow mouth,(+) Thin upper lip vermilion,(+) Retrognathia,(+) Pointed chin,(+) Triangular face,(+) Long philtrum,(+) Low-set, posteriorly rotated ears,(+) Abnormality iris morphology,(+) Upslanted palpebral fissure,(+) Single transverse palmar crease,(+) Hypotonia,(+) Global developmental delay,(+) Failure to thrive,(+) Skin tags
NFE2L1	ENST00000357480:c.2101C>T p.Arg701Ter		unknown	6.51	het	unknown	1	syndromal disease, ndd	(+) Narrow mouth,(+) Thin upper lip vermilion,(+) Retrognathia,(+) Pointed chin,(+) Triangular face,(+) Long philtrum,(+) Low-set, posteriorly rotated ears,(+) Abnormality iris morphology,(+) Upslanted palpebral fissure,(+) Single transverse palmar crease,(+) Hypotonia,(+) Global developmental delay,(+) Failure to thrive,(+) Skin tags
AMPD3	ENST00000396553:c.1499T>C p.Leu500Pro		unknown	3.9	het	unknown	1	Immune	(+) Recurrent fever,(+) Vertigo,(+) Immunodeficiency,(+) Autoimmunity,(+) Fatigue,(+) Acroparesthesia,(+) Body ache,(+) Systemic autoinflammation
AMPD3	ENST00000396553:c.2023C>A p.Leu675Ile		unknown	4.2	het	unknown	1	Immune	(+) Recurrent fever,(+) Vertigo,(+) Immunodeficiency,(+) Autoimmunity,(+) Fatigue,(+) Acroparesthesia,(+) Body ache,(+) Systemic autoinflammation
SEC24B	ENST00000265175:c.3247A>G p.Lys1083Glu		unknown	5.76	het	unknown	1	Epilepsy	(+) Seizure,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Complex febrile seizure
STIM2	ENST00000237364:c.653C>G p.Ser218Ter		unknown	7.28	het	unknown	1	NDD + epilepsy	(+) Generalized-onset seizure,(+) Moderate global developmental delay
ZNF638	ENST00000264447:c.3079_3080del p.Val1027TrpfsTer15		unknown	6.94	het	unknown	1	malformation	(+) Microcephaly,(+) Retrognathia,(+) Hearing impairment,(+) Hypotonia,(+) Umbilical hernia,(+) Diastasis recti,(+) Patent urachus
PDZD2	ENST00000438447:c.5100del p.Ser1701AlafsTer14		unknown	7.24	het	unknown	1	NDD + epilepsy	(+) Autism,(+) Seizure,(+) Global developmental delay,(+) Status epilepticus,(+) Severe global developmental delay
PKN1	ENST00000242783:c.2262del p.Lys755ArgfsTer33		homo	9.74	homo	unknown	1	NDD	(+) Long face,(+) Oval face,(+) Long philtrum,(+) Hypotelorism,(+) Nystagmus,(+) Autism,(+) Aggressive behavior,(+) Delayed speech and language development,(+) Prominent fingertip pads,(+) Hypotonia,(+) Absent speech,(+) Ventricular septal defect,(+) Short stature,(+) Secondary microcephaly,(+) Hepatic arteriovenous malformation,(+) Repetitive compulsive behavior,(+) Severe global developmental delay,(+) Reduced circulating alpha-1-antitrypsin concentration

ZNF638	ENST00000264447:c.3079_3080del p.Val1027TrpfsTer15		unknown	6.94	het	unknown	1		(+) Microcephaly,(+) Retrognathia,(+) Hearing impairment,(+) Hypotonia,(+) Umbilical hernia,(+) Diastasis recti,(+) Patent urachus
EPHB1	ENST00000398015:c.1993T>C p.Phe665Leu		unknown	5.5	het	unknown	1	Epilepsy	(+) Motor delay,(+) Generalized-onset seizure
GTF2I	ENST00000573035:c.1304+3A>G None		unknown	8.69	het	unknown	1	NDD	(+) Atypical behavior,(+) Autism,(+) Delayed speech and language development,(+) Sacral dimple,(+) Hypopigmented skin patches,(+) Global developmental delay,(+) Celiac disease
ZNF827	ENST00000379448:c.2416G>T p.Gly806Ter		unknown	6.51	het	unknown	1	NDD	(+) Microcephaly,(+) Global developmental delay,(+) Abnormal relationship
AZIN1	ENST00000337198:c.646C>T p.Arg216Ter		unknown	6.36	het	unknown	1	Epilepsy	(+) Myoclonus,(+) Obesity,(+) Bilateral tonic-clonic seizure,(+) Migraine,(+) Generalized-onset seizure,(+) Focal-onset seizure
VEZF1	ENST00000581208:c.1190dup p.Thr398HisfsTer38		de_novo	9.9	het	de novo		Myopathy	(+) Ptosis,(+) Muscle weakness,(+) Abnormality of the voice,(+) Weakness of facial musculature
OSBP	ENST00000263847:c.822+1G>T None		unknown	6.91	het	unknown		NDD	(+) Round face,(+) Upslanted palpebral fissure,(+) Low frustration tolerance,(+) Delayed speech and language development,(+) Prominent fingertip pads,(+) Sleep abnormality,(+) Secondary microcephaly,(+) Attention deficit hyperactivity disorder,(+) Mild global developmental delay,(+) Narrow palpebral fissure
PRKCE	ENST00000306156:c.1475G>A p.Gly492Glu		de_novo	7.5	het	de novo	1	NDD	Tall stature, Abnormality of the face, Secundum atrial septal defect, Nail dystrophy, Mild global developmental delay, Unilateral renal hypoplasia
SLC2A3	ENST00000075120:c.1283G>C	p.Gly428Ala	de_novo	7.43	het	de novo	1	NDD + autism	Behavioral abnormality, Delayed speech and language development, Autism
GRIK3	ENST00000373091:c.1142A>G	p.Asn381Ser	de_novo	8.16	het	de novo		NDD	Abnormality of the face, Delayed speech and language development, Global developmental delay, Abnormal cerebrospinal fluid morphology, Pendular nystagmus
SON	ENST00000300278:c.1889T>C p.Leu630Pro		de_novo	7.61	het	de novo	1	NDD + epilepsy	Seizure, Febrile seizure (within the age range of 3 months to 6 years), Mild global developmental delay, Periorbital edema
FBXO33	ENST00000298097:c.523_524del p.Ala176SerfsTer30		homo	9.72	homo	maternal& paternal	4	NDD + epilepsy	Focal-onset seizure, Moderate global developmental delay
TLL4	ENST00000392102:c.-99+737G>A		homo	3.18	homo	maternal& paternal	4	NDD + epilepsy	Focal-onset seizure, Moderate global developmental delay
ADGRB1	ENST00000517894:c.2228G>A p.Arg743Gln		homo	4.9	homo	maternal& paternal	4	NDD + epilepsy	Focal-onset seizure, Moderate global developmental delay
CDR2	ENST00000268383:c.514G>T p.Val172Leu		de_novo	5.81	het	de novo	4	NDD + epilepsy	Focal-onset seizure, Moderate global developmental delay
LRRC41	ENST00000343304:c.199+4A>G None		unknown	6.53	het	unknown	3	other	(+) Muscle weakness,(+) Rheumatoid arthritis,(+) Immune dysregulation,(+) Elevated circulating creatine kinase concentration,(+) Myalgia,(+) Muscle stiffness,(+) Sclerosis of hand bone,(+) Chilblains,(+) Allergy,(+) Raynaud phenomenon,(+) Sclerosis of foot bone
UBR2	ENST00000372899:c.3743G>C p.Arg1248Thr		homo	6.54	homo	maternal& paternal	4	NDD + autism	Autism, Delayed speech and language development, Hypotonia, Sleep abnormality, Hyperactivity
PIP4K2B	ENST00000617499:c.-191+573G>A		homo	5.49	homo	maternal& paternal	4	NDD + autism	Autism, Delayed speech and language development, Hypotonia, Sleep abnormality, Hyperactivity
OLFML2A	ENST00000288815:c.1084G>A p.Gly362Arg		homo	5.45	homo	maternal& paternal	4	NDD + autism	Autism, Delayed speech and language development, Hypotonia, Sleep abnormality, Hyperactivity
MTMR1	ENST00000370390:c.2T>C p.Met1?		unknown	6.56	hemi	unknown	1	NDD	(+) Macroglossia,(+) Neonatal hypotonia,(+) Severe global developmental delay
GRAMD1B	ENST00000450171:c.218A>G p.Asn73Ser		de_novo	6.32	het	de novo	2	NDD + autism	Microcephaly, Atypical behavior, Autism, Delayed speech and language development, Short stature, Severe global developmental delay, Abnormal eating behavior
NAV2	ENST00000349880:c.6017A>G p.Tyr2006Cys	ENST00000349880:c.3466T>A p.Ser1156Thr	AR_comphet	6.18	comphet	maternal/ paternal	2	NDD + autism	Microcephaly, Atypical behavior, Autism, Delayed speech and language development, Short stature, Severe global developmental delay, Abnormal eating behavior
PSKH1	ENST00000291041:c.509T>C p.Met170Thr		de_novo	5.57	het	de novo	2	NDD + Epilepsie	Microcephaly, Macrotia, Seizure, EEG abnormality, Severe global developmental delay, Epileptic encephalopathy
WWP2	ENST00000356003:c.986C>G p.Ala329Gly		de_novo	5.86	het	de novo	2	NDD + Epilepsie	Microcephaly, Macrotia, Seizure, EEG abnormality, Severe global developmental delay, Epileptic encephalopathy
MINK1	ENST00000347992:c.72C>G p.Ile24Met		de_novo	7.09	het	de novo	1	NDD	Secondary microcephaly, Polycystic kidney dysplasia, Hypertelorism, Blue sclerae, Low-set ears, Small earlobe , Patent foramen ovale, Hip dysplasia, Laryngomalacia, Thin vermilion border, Omphalocele, Global developmental delay
RAPH1	ENST00000319170:c.1922dup p.Pro642SerfsTer44		unknown	7.15	het	unkown	1	other	(+) Microcephaly,(+) Failure to thrive,(+) Atrial septal defect,(+) Pulmonic stenosis,(+) Short stature,(+) Tricuspid atresia,(+) Abnormal emotion
AHCTF1	ENST00000326225:c.1759dup p.Thr587AsnfsTer2		unknown	8.15	het	unknown	1	other	(+) Cardiomyopathy,(+) Hypertrophic cardiomyopathy

PTPRS	ENST00000262963:c.2179G>T p.Val727Leu		de_novo	7.68	het	de novo	2	Cortical tubers	Cortical tubers, Seizure, Cerebral hamartoma, Polycystic kidney dysplasia, Localized skin lesion
NCKAP1	ENST00000360982:c.126+56C>A		de_novo	6.65	het	de novo	2	Cortical tubers	Cortical tubers, Seizure, Cerebral hamartoma, Polycystic kidney dysplasia, Localized skin lesion
DGKG	ENST00000265022.8:c.1798G>C p.Glu600Gln	ENST00000265022.8:c.1096del p.Cys366AlafsTer6	AR_comphet	5.03	comphet	maternal/paternal	1		Short philtrum, Low-set ears, Long eyelashes, Long palpebral fissure, Global developmental delay, Bilateral tonic-clonic seizure with focal onset, EEG abnormality, Broad-based gait, Delayed speech and language development
SSR2	ENST00000295702:c.355C>A p.Pro119Thr		homo	4.98	homo	maternal and paternal	2	NDD + autism	Autistic behavior, Moderate global developmental delay
IFT80	ENST00000475677:c.665A>G p.His222Arg		homo	8.18	homo	maternal and paternal	2	NDD + autism	Autistic behavior, Moderate global developmental delay
SEPTIN11	ENST00000264893:c.773G>T p.Gly258Val		de_novo	6.85	het	de novo	1	NDD	Triangular face, Aggressive behavior, Intellectual disability, mild, Global developmental delay
FEN1	ENST00000535307:None None		de_novo	5.25	het	de novo	1	NDD	Expressive language delay
BIRC6	ENST00000421745:c.7438G>T p.Asp2480Tyr		homo	6.01	homo	maternal/paternal	3	multiple malformations; DD VACTERL-association	Ectopic kidney, Renal dysplasia, Patent ductus arteriosus, Patent foramen ovale, Esophageal atresia, Intracranial hemorrhage, Duodenal atresia, Tracheoesophageal fistula
RFX1	ENST00000254325:c.2693A>G p.Lys898Arg		homo	5.7	homo	maternal/paternal	3	multiple malformations; DD VACTERL-association	Ectopic kidney, Renal dysplasia, Patent ductus arteriosus, Patent foramen ovale, Esophageal atresia, Intracranial hemorrhage, Duodenal atresia, Tracheoesophageal fistula
ZNF468	ENST00000595646:c.968G>C p.Gly323Ala		homo	3.56	homo	maternal/paternal	3	multiple malformations; DD VACTERL-association	Ectopic kidney, Renal dysplasia, Patent ductus arteriosus, Patent foramen ovale, Esophageal atresia, Intracranial hemorrhage, Duodenal atresia, Tracheoesophageal fistula
CLU	ENST00000316403:c.1225C>T p.Leu409Phe		de_novo	8.6	het	de novo	2	NDD +hypotonia	Microcephaly, Strabismus, Delayed speech and language development, Hypotonia, Plagiocephaly, Skull asymmetry, Mild global developmental delay
CRACR2A	ENST00000440314:c.2008C>T p.Arg670Trp	ENST00000440314:c.1109A>G p.Asp370Gly	comphet	3.54	comphet	maternal/paternal	2	NDD +hypotonia	Microcephaly, Strabismus, Delayed speech and language development, Hypotonia, Plagiocephaly, Skull asymmetry, Mild global developmental delay
GLI4	ENST00000340042:c.97C>T p.Gln33Ter		homo	7.84	het	unknown	2	NDD	Mild global developmental delay
TNN	ENST00000239462:c.2297del p.Gly766AlafsTer2		homo	8.76	het	unknown	2	NDD	Mild global developmental delay
ASTN1	ENST00000361833:c.1936del p.Arg646Alafs*75		unknown	10.4	het	unknown	1	progressive Macroceph	(+) Macrocephaly
SHPRH	ENST00000275233:c.1180G>C p.Asp394His		de_novo	5.8	het	de novo	2	NDD + learning	Hypotonia, Specific learning disability, Mild global developmental delay
APLP1	ENST00000221891:c.981+1G>A None		de_novo	7.67	het	de novo	2	NDD + learning	Hypotonia, Specific learning disability, Mild global developmental delay
GRIPAP1	ENST00000376423:c.2073C>A p.Ser691Arg		x_linked	6.01	hemi	maternal	1	NDD + autism	Autism, Global developmental delay
ZFHX4	ENST00000050961:c.6719A>G p.Tyr2240Cys		unknown	3.45	het	unknown	1	NDD	(+) Microcephaly,(+) Aggressive behavior,(+) Global developmental delay,(+) EEG abnormality,(+) Short stature,(+) Cognitive impairment,(+) Reduced attention regulation
POC1B	ENST00000313546:c.100+2115G>T None		de_novo	6.25	het	de novo	2		Seizure, Hypsarrhythmia, Focal-onset seizure, Moderate global developmental delay, Epileptic encephalopathy
BTBD1	ENST00000261721:c.1120A>G p.Thr374Ala		de_novo	6.36	het	de novo	1		Focal-onset seizure, Gray matter heterotopia
AGMO	ENST00000342526:c.958-3T>G None		homo	10.2	homo	unkown	1	NDD + Epilepsie	(+) Progressive microcephaly,(+) Retrognathia,(+) Synophrys,(+) Delayed speech and language development,(+) Hypotonia,(+) Joint hypermobility,(+) Failure to thrive,(+) Bilateral tonic-clonic seizure,(+) Generalized-onset seizure,(+) EEG abnormality,(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Clinodactyly of the 5th finger,(+) Short stature,(+) Focal-onset seizure,(+) Mild global developmental delay,(+) Primary microcephaly,(+) Clinodactyly of the 4th toe

ACSL4	ENST00000340800:c.1124del p.Pro375ArgfsTer12		x_linked	11.1	hemi	maternal	2		Seizure, Hypsarrhythmia, Focal-onset seizure, Moderate global developmental delay, Epileptic encephalopathy
KDM1A	ENST00000693156:c.2070G>A p.Thr690=		de_novo	11.7	het	de novo	1	NDD + autism	(+) Long face,(+) Oval face,(+) Long philtrum,(+) Hypotelorism,(+) Nystagmus,(+) Autism,(+) Aggressive behavior,(+) Delayed speech and language development,(+) Prominent fingertip pads,(+) Hypotonia,(+) Absent speech,(+) Ventricular septal defect,(+) Short stature,(+) Secondary microcephaly,(+) Hepatic arteriovenous malformation,(+) Repetitive compulsive behavior,(+) Severe global developmental delay,(+) Reduced circulating alpha-1-antitrypsin concentration
RNPS1	ENST00000301730:c.448G>T p.Glu150Ter		de_novo	10.2	het	de novo	1	NDD	Tall stature, Pectus excavatum, Hypotonia, Global developmental delay, Alopecia, Recurrent fever, Precocious puberty in males
SRXN1	ENST00000381962:c.238G>C p.Asp80His		de_novo	5.12	het	de novo	1	NDD + Epilepsy	Seizure, Generalized myoclonic-atonic seizure, Global developmental delay, Expressive language delay
CTNND2	ENST00000304623:c.1517T>C p.Leu506Pro		unknown	8.56	het	unknown	2	NDD + seizure	Synophrys,(+) Delayed speech and language development,(+) Intellectual disability, moderate,(+) Focal-onset seizure,(+) Everted upper lip vermilion,(+) Moderate global developmental delay
BSN	ENST00000296452:c.3335del p.Ala1112GlyfsTer80		AD_unknown	9.48	het	unknown	1	Seizure	(+) Generalized-onset seizure,(+) Febrile seizure (within the age range of 3 months to 6 years)
PPP1R9B	ENST00000316878:c.1214_1215del p.Gly405GlufsTer75		unknown	9.21	het	unknown	2	NDD + seizure	Synophrys,(+) Delayed speech and language development,(+) Intellectual disability, moderate,(+) Focal-onset seizure,(+) Everted upper lip vermilion,(+) Moderate global developmental delay
JPH3	ENST00000284262:c.2101dup p.Gln701ProfsTer17		unknown	9.66	het	unknown	1	NDD + ataxia	(+) Microcephaly,(+) Delayed speech and language development,(+) Ataxia,(+) Myoclonic seizure
STAU1	ENST00000340954:c.137A>C p.Tyr46Ser		unknown	5.69	het	unknown	1	NDD + autism	(+) Epicanthus,(+) High forehead,(+) Atypical behavior,(+) Autism,(+) Frontal bossing,(+) Clinodactyly of the 5th finger,(+) Severe global developmental delay,(+) Narrow palpebral fissure,(+) Cognitive impairment
NTM	ENST00000374784:c.503C>T p.Thr168Ile		de_novo	6.76	het	de novo	1	NDD + seizure	Autism, Atonic seizure, Epileptic spasm, Severe global developmental delay, Tonic seizure, Generalized-onset seizure
ATP8A1	ENST00000264449:c.2744C>A p.Ser915Tyr		unknown	5.05	het	unknown	1	NDD	(+) Atypical behavior,(+) Autism,(+) Delayed speech and language development,(+) Intellectual disability, mild,(+) Severe global developmental delay
ZBTB41	ENST00000367405:c.434C>T p.Thr145Ile		unknown	3.91	het	unknown	1	NDD + seizure	(+) Generalized-onset seizure,(+) Cognitive impairment
PSME4	ENST00000404125:c.421C>T p.Pro141Ser		de_novo	6.66	het	de novo	2	NDD + seizure	Macroglossia, Epicanthus, Long philtrum, Upslanted palpebral fissure, Autistic behavior, Hypotonia, Depressed nasal bridge, Focal-onset seizure, Mild global developmental delay, Tip-toe gait, Macroglossia, Epicanthus, Long philtrum, Upslanted palpebral fissure, Autistic behavior, Hypotonia, Depressed nasal bridge, Focal-onset seizure, Mild global developmental delay, Tip-toe gait
ADAMTS6	ENST00000381055:c.843+1G>T None		de_novo	9.11	het	de novo	2	NDD + seizure	Macroglossia, Epicanthus, Long philtrum, Upslanted palpebral fissure, Autistic behavior, Hypotonia, Depressed nasal bridge, Focal-onset seizure, Mild global developmental delay, Tip-toe gait, Macroglossia, Epicanthus, Long philtrum, Upslanted palpebral fissure, Autistic behavior, Hypotonia, Depressed nasal bridge, Focal-onset seizure, Mild global developmental delay, Tip-toe gait
MYOZ1	ENST00000359322:c.481G>T p.Gly161Trp		de_novo	5.31	het	de novo	1	Myopathy	Tremor, Talipes equinovarus, Motor axonal neuropathy, Demyelinating motor neuropathy, Increased CSF albumin concentration
GARIN1A	ENST00000682356:c.428A>C p.Tyr143Ser		homo	5.02	homo	maternal& paternal	1	NDD + seizure	Focal clonic seizure, Moderate global developmental delay
WDR47	ENST00000361054:c.2407G>A p.Gly803Arg		unknown	4.33	het	unkown	2	Seizure	(+) Seizure,(+) Focal-onset seizure
OTUB1	ENST00000428192:c.22C>T p.Gln8Ter		unknown	6.39	het	unkown	1	Seizure	(+) Focal-onset seizure,(+) Focal cortical dysplasia
TMEM132D	ENST00000389441:c.1718dup p.Thr574AsnfsTer34		unknown	6.84	het	unkown	1	Autism	(+) Atypical behavior,(+) Autistic behavior,(+) Abnormality of speech or vocalization,(+) Mild global developmental delay
DPYSL5	ENST00000288699:c.1366_1370dup p.Thr458ArgfsTer9		unknown	9.83	het	unkown	1	Seizure	(+) Microcephaly,(+) Hypotonia,(+) EEG abnormality,(+) Feeding difficulties in infancy,(+) EEG with burst suppression,(+) Mild global developmental delay,(+) Abnormal brain morphology,(+) Neonatal seizure,(+) Brain imaging abnormality
SH2D3C	ENST00000314830:c.806dup p.Val270GlyfsTer17		unknown	5.88	het	unkown	1	NDD	(+) Premature birth,(+) Secondary microcephaly,(+) Mild global developmental delay,(+) Epidural hemorrhage
PTK7	ENST00000230419:c.1388del p.Asn463MetfsTer20		unknown	8.6	het	unknown	1	Neuro	(+) Polyneuropathy,(+) Gait disturbance,(+) Poor motor coordination,(+) Distal sensory impairment,(+) Poor fine motor coordination,(+) Reduced visual acuity
EIF3A	ENST00000369144:c.568C>T p.Arg190Cys		unknown	5.56	het	unknown	1	NDD+ Seizure	(+) Ataxia,(+) Hypotonia,(+) Polymicrogyria,(+) Generalized-onset seizure,(+) Abnormal cortical gyration,(+) Focal-onset seizure,(+) Severe global developmental delay,(+) Abnormal brain morphology
RPRD1A	ENST00000357384:c.506C>T p.Ala169Val		de_novo	5.71	het	de novo	2	NDD	Global developmental delay, Intellectual disability, moderate, Mild microcephaly, Developmental regression

SLC35F3	ENST00000366617:c.667_668dup p.Ala224ThrfsTer91		de_novo	5.09	het	de novo	2	Immunodeficiency	Diarrhea, T lymphocytopenia, Abnormality of immune system physiology, Abnormal T cell count, Bloody diarrhea, Unusual infection, Sepsis, Abnormal natural killer subset distribution
UNC5B	ENST00000335350:c.1148C>A p.Ala383Asp	ENST00000335350:c.1579G>C p.Ala527Pro	comphet	4.56	comphet	maternal/paternal	2	Immunodeficiency	Diarrhea, T lymphocytopenia, Abnormality of immune system physiology, Abnormal T cell count, Bloody diarrhea, Unusual infection, Sepsis, Abnormal natural killer subset distribution
BRPF3	ENST00000357641:c.2320G>A p.Glu774Lys		unknown	5	het	unknown	1	other	(+) Renal cyst,(+) Optic atrophy,(+) Atypical behavior,(+) Pancreatic cysts,(+) Global brain atrophy,(+) Unsteady gait,(+) Type II diabetes mellitus,(+) Abnormality of von Willebrand factor,(+) Cognitive impairment
TTC28	ENST00000397906:c.5065G>T p.Ala1689Ser		unknown	4.2	het	unknown	1	other	(+) Wide nose,(+) Downslanted palpebral fissures,(+) Depression,(+) Arthrogryposis multiplex congenita,(+) Arthralgia,(+) Abnormal phalangeal joint morphology of the hand,(+) Camptodactyly
ELFN1	ENST00000424383:c.1842del p.Tyr616ThrfsTer68		unknown	7.74	het	unknown	1	lokale selbstlimitierende Epilepsie, milde globale Entwicklungsverzögerung mit schwerer	Delayed speech and language development, Global developmental delay, Focal-onset seizure
MRPL42	ENST00000393128:c.143A>G p.Glu48Gly		de_novo	4.9	het	de novo	1	NDD + brain atrophy	Atypical behavior, Dementia, Intellectual disability, mild, Motor delay, Abnormality of speech or vocalization, Global brain atrophy, Sleep abnormality, Infectious encephalitis, Pica
ALDH1L1	ENST00000273450:c.435C>A p.Phe145Leu		de_novo	7.41	het	de novo	3	intracranial hemorrhage, incoordination	Intracranial hemorrhage, Spontaneous hematomas, Incoordination
TBKBP1	ENST00000361722:c.316C>G p.Leu106Val	ENST00000361722:c.317T>G p.Leu106Arg	comphet	3.76	comphet	de novo	3	Intracranial hemorrhage, incoordination	Intracranial hemorrhage, Spontaneous hematomas, Incoordination
CXXC4	ENST00000394767:c.233C>T p.Ala78Val		de_novo	4.56	het	de novo	1		Intellectual disability, Seizure, Myoclonus, Developmental regression, Atypical absence seizure, Epileptic spasm, Multifocal seizures, Tonic seizure
NAV3	ENST00000397909:c.4683+5G>A None		unknown	7.27	het	unkown		NDD+ seizure	(+) Generalized-onset seizure,(+) Absence seizure with eyelid myoclonia,(+) Moderate global developmental delay,(+) Bilateral tonic-clonic seizure with generalized onset,(+) Eyelid myoclonia seizure,(+) Cognitive impairment
NINL	ENST00000278886:c.578C>T p.Ser193Phe		de_novo	5.9	het	de novo	1	neonatal hypotonia, Polyhydramnios, Muscular ventricular septal defect, Aortic	Macrotia, Neonatal hypotonia, Polyhydramnios, Frontal bossing, Hypocalcemia, Depressed nasal bridge, Prominent forehead, Muscular ventricular septal defect, Aortic isthmus hypoplasia, Fetal abdominal cyst, Polycystic kidney dysplasia, Leukodystrophy, Syringomyelia, Hydromyelia
TSPAN2	ENST00000369516:c.517-2846G>A p.Gly193Asp	ENST00000369516:c.353G>A p.Arg118Gln	comphet	4.3	comphet	maternal/paternal	2	NDD+ autism	Autism, Delayed speech and language development, Global developmental delay, Hyperactivity
TRIB2	ENST00000155926:c.208C>G p.Leu70Val		de_novo	5.62	het	de novo	2	NDD+ autism	Autism, Delayed speech and language development, Global developmental delay, Hyperactivity
DSCAML1	ENST00000321322:c.2408G>A p.Arg803His		unknown	5.59	het		3	Seizure + Obesity	(+) Obesity,(+) Focal-onset seizure

NTN4	ENST00000343702:c.55+1G>T None		unknown	6.93	het	unknown	3	Aneurysm	Abdominal aortic aneurysm, Thoracic aortic aneurysm
MYO15B	ENST00000583140:c.254+1G>T None		unknown	5.13	het	unknown	3	Aneurysm	Abdominal aortic aneurysm, Thoracic aortic aneurysm
HSP90AB1	ENST00000353801:c.2020C>T p.Gln674Ter		unknown	7.11	het	unknown	1	NDD+ Seizure	(+) Microcephaly,(+) Acidosis,(+) Generalized-onset seizure,(+) Muscle spasm,(+) Severe global developmental delay,(+) Abnormal brain morphology
NCL	ENST00000322723:c.459_462del p.Ser153ArgfsTer22		de_novo	11.3	het	de novo	1		(+) Microcephaly,(+) Hypotonia,(+) Global developmental delay,(+) Intellectual disability, moderate,(+) Feeding difficulties in infancy
TRPV2	ENST00000338560:c.1654G>A p.Ala552Thr		unknown	3.11	het	unknown	3	Aneurysm	Abdominal aortic aneurysm, Thoracic aortic aneurysm
FAM89A	ENST00000366654:c.133G>C p.Gly45Arg		de_novo	4.74	het	de novo	1	NDD + Hypercholesterolemia	Atypical behavior, Hypothyroidism, Intellectual disability, Global developmental delay, Obesity, Polyphagia, Hypercholesterolemia, Increased LDL cholesterol concentration,
RASL10B	ENST00000603017:c.594C>A p.Asn198Lys		de_novo	5.22	het	de novo	1	NDD + Epilepsy	Seizure, Global developmental delay, Partial agenesis of the corpus callosum, Interictal epileptiform activity, Arachnoid cyst, Optic atrophy, Talipes equinovarus, Cortical dysplasia, Inability to walk, Secondary microcephaly, Focal-onset seizure, EEG with focal sharp slow waves, Joint contracture
MED15	ENST00000263205:c.1697T>C p.Met566Thr		de_novo	8.26	het	de novo	1	NDD + seizure	Hypotonia, Global developmental delay, Generalized non-motor (absence) seizure, Intellectual disability, moderate
SLIT2	ENST00000273739:c.2729_2736dup p.Gly913ProfsTer11		unknown	8.66	het	unknown	1	NDD + seizure	Hypotonia,(+) Spasticity,(+) Generalized-onset seizure,(+) Intellectual disability, severe,(+) Severe global developmental delay
P2RX3	ENST00000263314:c.1072T>G p.Phe358Val		de_novo	6.11	het	de novo	1	Short stature + Facial dysmorphism	Microcephaly, Hypertelorism, Depressed nasal ridge, Downslanted palpebral fissures, Failure to thrive, Short stature, Abnormal eating behavior, Triangular-shaped open mouth
RASGEF1C	ENST00000361132:c.458A>T p.His153Leu		de_novo	5.23	het	de novo	1	NDD	Abnormal thumb morphology, Broad toe, Gait ataxia, Elevated circulating creatine kinase concentration, Severe global developmental delay, Short palpebral fissure, Thin eyebrow, Long fingers
KCNK2	ENST00000391894:c.97G>A p.Val33Met		de_novo	6.66	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizure, Generalized-onset seizure, Focal-onset seizure, Moderate global developmental delay
HSD17B8	ENST00000374662:c.52G>A p.Gly18Ser		de_novo	5.64	het	de novo	2	NDD + Epilepsy	Intellectual disability, Seizure, Generalized-onset seizure, Focal-onset seizure, Moderate global developmental delay
CACNG3	ENST00000005284:c.437-1G>A None		ad_inherited	9.47	het	paternal	2	NDD	Global developmental delay, Intellectual disability, moderate, Mild microcephaly, Developmental regression
PAK6	ENST00000260404:c.1028C>T p.Pro343Leu		de_novo	6.25	het	de novo	2	NDD + Epilepsy	Bilateral tonic-clonic seizure, Febrile seizure (within the age range of 3 months to 6 years), Focal-onset seizure, Moderate global developmental delay, Diminished ability to concentrate, Seizure precipitated by febrile infection
MAP4K1	ENST00000396857:c.1447-154G>T None		de_novo	3.39	het	de novo	2	NDD + Epilepsy	Bilateral tonic-clonic seizure, Febrile seizure (within the age range of 3 months to 6 years), Focal-onset seizure, Moderate global developmental delay, Diminished ability to concentrate, Seizure precipitated by febrile infection
KAT2A	ENST00000225916:c.527T>G p.Val176Gly		unknown	7.17	het	unknown	1	Myopathy	Seizure, Malignant hyperthermia, Myopathy, Elevated circulating creatine kinase concentration, Intermittent painful muscle spasms
FAM171A1	ENST00000378116:c.566G>T p.Gly189Val		de_novo	5.32	het	de novo	2	NDD	Microcephaly, Autism, Delayed speech and language development, Global developmental delay, Absent speech, Receptive language delay
TARBP1	ENST00000040877:c.3737C>A p.Thr1246Lys	ENST00000040877:c.4201C>T p.Leu1401Phe	comphet	4.41	comphet	maternal&paternal	2	NDD	Microcephaly, Autism, Delayed speech and language development, Global developmental delay, Absent speech, Receptive language delay
BSN	ENST00000296452:c.5886del p.Pro1963LeufsTer57		AD_unknown	9.48	het	unknown	3	Epilepsy	(+) Focal-onset seizure
ZNF337	ENST00000252979:c.147_154+1del None		de_novo	5.15	het	de novo	1	macular degeneration	Retinal degeneration Macular degeneration
HDAC5	ENST00000225983:c.2194C>T p.Arg732Ter		unknown	8.18	het	unkown	2	Epilepsy	(+) Myoclonus,(+) Generalized-onset seizure,(+) Bilateral tonic-clonic seizure with generalized onset
SEMA4B	ENST00000332496:c.158-2518C>G None		de_novo	4.41	het	de novo	1	NDD	Macrocephaly, Atypical behavior, Global developmental delay, Obesity, Developmental regression, Few cafe-au-lait spots, Intellectual disability
SRCIN1	ENST00000612431:c.686C>G p.Ser229Cys		de_novo	7.45	het	de novo	1	NDD	Pierre-Robin sequence, Strabismus, Myopia, Agenesis of corpus callosum, Talipes equinovarus, Intellectual disability, moderate, Focal-onset seizure, Anisocoria, Anisometropia
SCPPPQ1	ENST00000715253:c.168+1G>A None		unknown	5	het	unkown	1	Basal cell carcinoma	Basal cell carcinoma Arachnoid cyst Abnormal coagulation factor V activity

CC2D2A	ENST00000389652:None None		unknown	5.76	het	unkown	1	NDD + Epilepsy	(+) Autism,(+) Neurodegeneration,(+) Generalized-onset seizure,(+) Global brain atrophy,(+) Severe global developmental delay,(+) Epileptic encephalopathy
NFKBID	ENST00000585544:c.-200-2A>C None		homo	8.02	homo	maternal& paternal	8	NDD	Microcephaly, Hypotonia, Developmental regression, Severe global developmental delay, CNS hypomyelination
GABPA	ENST00000354828:c.23A>G p.Glu8Gly		homo	6.55	homo	maternal& paternal	8	NDD	Microcephaly, Hypotonia, Developmental regression, Severe global developmental delay, CNS hypomyelination
SLC25A14	ENST00000218197:c.880G>T p.Ala294Ser		x_linked	5.17	hemi	maternal	8	NDD	Microcephaly, Hypotonia, Developmental regression, Severe global developmental delay, CNS hypomyelination
NCOA2	ENST00000452400:c.3022A>C p.Asn1008His		homo	5.92	homo	maternal& paternal	8	NDD	Microcephaly, Hypotonia, Developmental regression, Severe global developmental delay, CNS hypomyelination
PKN1	ENST00000242783:None None		homo	5.31	homo	maternal& paternal	8	NDD	Microcephaly, Hypotonia, Developmental regression, Severe global developmental delay, CNS hypomyelination
MAP3K10	ENST00000253055:None None		homo	5.26	homo	maternal& paternal	8	NDD	Microcephaly, Hypotonia, Developmental regression, Severe global developmental delay, CNS hypomyelination
ZSWIM8	ENST00000398706:c.313C>T p.Gln105Ter		unknown	6.65	het	unknown	1	NDD	(+) Nystagmus,(+) Atypical behavior,(+) Hypotonia,(+) Moderate global developmental delay
FZD7	ENST00000286201:c.1059G>A p.Trp353Ter		de_novo	6.25	het	de novo	1	NDD	High forehead, Autism, Delayed speech and language development, Hyperactivity, Pectus excavatum, Global developmental delay, Abnormal renal morphology, Supravalvar pulmonary stenosis, Self-injurious behavior
PPP1R13B	ENST00000202556:c.26del p.Phe9SerfsTer2		unknown	6.71	het	unknown	1	other	(+) Renal dysplasia,(+) Astigmatism,(+) Bronchiectasis,(+) Immunodeficiency,(+) Chronic bronchitis,(+) Recurrent pneumonia,(+) Abnormal pulmonary thoracic imaging finding,(+) Atelectasis
CHD6	ENST00000309279:c.1709T>G p.Phe570Cys		unknown	5.8	het	unknown	1	NDD + Epilepsy	(+) Polymicrogyria,(+) Focal-onset seizure,(+) Severe global developmental delay,(+) Focal cortical dysplasia
MAP1A	ENST00000300231:c.5534G>A p.Trp1845Ter		unknown	9.16	het	unknown	1	NDD + Epilepsy	+) Autism,(+) Intellectual disability,(+) Status epilepticus,(+) Attention deficit hyperactivity disorder
PRPF39	ENST00000355765:c.1757+2_1757+5del None		ad_inherited	6.9	het	maternal or paternal	1	NDD	High palate, Aggressive behavior, Autistic behavior, Intellectual disability, Global developmental delay, Hepatosplenomegaly, Protuberant abdomen, Facial shape deformation, Axial hypotonia, Infantile muscular hypotonia, Decreased circulating iron concentration, Decreased circulating vitamin D concentration, Self-injurious behavior, Intellectual disability, moderate
RNASE2	ENST00000304625:c.127A>G p.Ile43Val		de_novo	5.42	het	de novo	2	NDD	Microcephaly, Spasticity, Acidosis, Cerebral ischemia, Mild global developmental delay, Intraventricular hemorrhage, Neonatal seizure
SNTG2	ENST00000308624:c.217A>G p.Thr73Ala	ENST00000308624:c.289_290del p.Pro97CysfsTer13	comphet	6.76	comphet	maternalp aternal	2	NDD	Microcephaly, Spasticity, Acidosis, Cerebral ischemia, Mild global developmental delay, Intraventricular hemorrhage, Neonatal seizure
KRT19	ENST00000361566:c.1136C>G p.Thr379Ser		de_novo	5.3	het	de novo	1	NDD	Aggressive behavior, Gynecomastia, Atopic dermatitis, Global developmental delay, Overweight, Abnormal eating behavior, Atypical behavior
LOC400499	ENST00000598234:c.836-2A>G None		homo	8	homo	maternal& paternal	1	NDD + epilepsy	Coarse facial features, Autism, Intellectual disability, Seizure, EEG abnormality, Moderate global developmental delay, Infantile spasms, Overweight, Global developmental delay
PCSK6	ENST00000331826:c.1532+1836C>T None		homo	4.2	homo	maternal& paternal	1	Motor DD + Myopathy	Hypospadias, Hearing impairment, Global developmental delay, Infantile muscular hypotonia, Camptodactyly, Long philtrum, Recurrent pneumonia, Inappropriate crying, Wide nasal base, Low-set ears, Retrognathia, Pectus excavatum, Abnormal testis morphology, Prominent calcaneus, Motor delay
TAGLN3	ENST00000273368:c.406G>T p.Ala136Ser		de_novo	6.92	het	de novo	2	NDD	Pachygyria, Polymicrogyria, Arthrogryposis multiplex congenita, Severe global developmental delay, Wrist drop, Bilateral talipes equinovarus, Hemangioma
PPM1M	ENST00000323588:c.28T>G p.Phe10Val		de_novo	4.14	het	de novo	2	NDD	Pachygyria, Polymicrogyria, Arthrogryposis multiplex congenita, Severe global developmental delay, Wrist drop, Bilateral talipes equinovarus, Hemangioma
SEPTIN6	ENST00000343984:c.410G>A p.Arg137Gln		x_linked	4.5	hemi	maternal	1	NDD	Tall stature, Hearing impairment, Nystagmus, Hypotonia, Global developmental delay, Abnormal facial shape, Generalized hypotonia
RIMS4	ENST00000372851:c.763del p.Gln255SerfsTer89		unknown	B	het	unknown	1	Movement disorder	Spasticity, Gait disturbance, Chorea, Hyperkinetic movements, Impaired pain sensation, Weakness due to upper motor neuron dysfunction, Impaired temperature sensation, Impaired continence
PPFIA4	ENST00000295706:None None		homo	4.75	homo	maternal& paternal	8	NDD	Microcephaly, Hypotonia, Developmental regression, Severe global developmental delay, CNS hypomyelination

ARHGEF28	ENST00000296794:c.1679C>T p.Ser560Leu		homo	5.98	homo	maternal&paternal	8	NDD	Microcephaly, Hypotonia, Developmental regression, Severe global developmental delay, CNS hypomyelination
XRN2	ENST00000377191:c.2256-2A>G None		unknown	7.6	het	unknown	1	NDD + epilepsy	(+) Unilateral renal agenesis,(+) Stage 5 chronic kidney disease,(+) Aortic aneurysm,(+) Bilateral tonic-clonic seizure with focal onset,(+) Mild global developmental delay,(+) Unilateral renal hypoplasia
MINK1	ENST00000347992:c.1189C>G p.Arg397Gly		de_novo	7.3	het	de novo	1	other	Omphalocele, Large for gestational age, Recurrent hypoglycemia, Enlarged kidney
PSMC4	ENST00000157812:c.651G>T p.Lys217Asn		ad_inherited	5.7	het	paternal	1	NDD	(+) Astigmatism,(+) Short stature,(+) Decreased body weight,(+) Moderate global developmental delay
DTD2	ENST00000310850:c.240_241del p.Leu81GlyfsTer28		homo	8.1	homo	unknown	1		(+) Autistic behavior,(+) Moderate global developmental delay
CADM2	ENST00000383699:c.308del p.Val103GlyfsTer48		unknown	7.8	het	unknown	1	Seizure	(+) Depression,(+) Autism,(+) Generalized non-motor (absence) seizure,(+) Bilateral tonic-clonic seizure with generalized onset
NAA40	ENST00000377793:c.373C>T p.Arg125Trp		de_novo	6.2	het	de novo	1	NDD + epilepsy	Cataract, Autism, Seizure, Severe global developmental delay, Global developmental delay
VASH1	ENST00000167106:c.530T>A p.Ile177Asn		de_novo	5.5	het	de novo	1	NDD	Intellectual disability, Global developmental delay
TRIM73	ENST00000323819:c.233dup p.Arg79GlufsTer64		unknown	5.6	het	unknown	2	NDD	(+) Autistic behavior,(+) Delayed speech and language development,(+) Cafe-au-lait spot,(+) Obesity
RBM5	ENST00000347869:c.1810C>T p.Arg604Ter		unknown	6.6	het	unknown	2	NDD	(+) Autistic behavior,(+) Delayed speech and language development,(+) Cafe-au-lait spot,(+) Obesity
CGN	ENST00000271636:c.1406_1416del p.Leu469ProfsTer40		unknown	6.3	het	unknown	1	NF	Axillary freckling, Atypical neurofibromatosis, Multiple cafe-au-lait spots, Inguinal freckling, Cafe-au-lait spot
CASZ1	ENST00000344008:c.2440_2443dup p.Val815AlafsTer22		de_novo	9.1	het	de novo	1	Kardiomyopathie	Dilated cardiomyopathy, Congestive Heart Failure, Feeding Difficulties
RUSC1	ENST00000368352:c.693del p.Asn232ThrfsTer52		unknown	7.0	het	unknown	1	NDD - epilepsy	(+) Autism,(+) Intellectual disability,(+) Bilateral tonic-clonic seizure,(+) Generalized non-motor (absence) seizure,(+) Focal-onset seizure,(+) Focal cortical dysplasia
DHX57	ENST00000457308:c.3816+4A>G None		ad_inherited	5.8	het	maternal	1	Epilepsy	Nausea, Migraine with aura, Dyscalculia, Episodic vomiting, Dyslexia, Paroxysmal lethargy, Bilateral tonic-clonic seizure, Chronic fatigue, Iron deficiency anemia
RPS6KB1	ENST00000225577:c.26G>A p.Gly9Asp		de_novo	7.9	het	de novo	1	NDD + Epilepsy	Generalized non-motor (absence) seizure, Thyroid hypoplasia, focal-onset seizure, Severe global developmental delay, myoclonic seizure, Epileptic encephalopathy, Seizure
HERC3	ENST00000402738:c.506_536dup p.Ser180TrpfsTer49		unknown	6.1	het	unknown	1	Other	(+) Granulomatosis,(+) Abnormal lymphocyte morphology,(+) Pulmonary granulomatosis,(+) Raynaud phenomenon,(+) Increased circulating interleukin 2 concentration,(+) Cold intolerance
NOP9	ENST00000267425:c.1175T>G p.Val392Gly		de_novo	5.5	het	de novo	1	NDD	Autism, Delayed speech and language development, Intellectual disability, Global developmental delay, Autistic behavior
ZSWIM8	ENST00000398706:c.2013dup p.Gly672TrpfsTer9		unknown	A	het	unknown	1	Movement disorder	(+) Visual impairment,(+) Hashimoto thyroiditis,(+) Seizure,(+) Gait disturbance,(+) Episodic ataxia,(+) Headache,(+) Frequent falls,(+) Paroxysmal vertigo
ADCY2	ENST00000338316:c.1714A>G p.Lys572Glu		unknown	6.1	het	unknown	1	NDD + Immune	(+) Unilateral renal agenesis,(+) Microcephaly,(+) Cervical ribs,(+) Abnormal finger morphology,(+) Global developmental delay,(+) Meningitis,(+) Abnormal foot morphology,(+) Coxa valga,(+) Immunodeficiency,(+) Syringomyelia,(+) Dysplastic aortic valve,(+) Abnormality of complement system,(+) Aplasia/Hypoplasia of the sacrum,(+) Cochlear malformation,(+) Paraplegia,(+) Childhood onset sensorineural hearing impairment,(+) Decreased circulating level of specific antibody,(+) Decreased specific antibody response to vaccination,(+) Obstipation,(+) Hydromyelia,(+) Sepsis
PHF12	ENST00000268756:c.248+4A>G None		unknown	6.7	het	unknown	1	Immunodeficiency	(+) Immunodeficiency,(+) Recurrent lower respiratory tract infections,(+) Decreased circulating total IgM,(+) Decreased circulating antibody concentration,(+) Decreased circulating IgG concentration,(+) Defective B cell differentiation,(+) Atelectasis
CDYL	ENST00000328908:c.1545_1546dup p.Gly516ValfsTer19		unknown	8.0	het	unknown	1	NDD + Immune	(+) Abnormal eyebrow morphology,(+) Delayed speech and language development,(+) Alopecia areata,(+) Immunodeficiency,(+) Autoimmunity,(+) Decreased circulating vitamin D concentration,(+) Recurrent oral herpes
PIK3C2B	ENST00000367187:c.4622A>G p.Tyr1541Cys		unknown	5.1	het	unknown	1	NDD + epilepsy	(+) Atypical behavior,(+) Seizure,(+) Global developmental delay
HDAC4	ENST00000345617:c.1792G>C p.Glu598Gln		de_novo	10.0	het	de novo	1	Intellectual disability,(+) Obesity,(+) Increased	

MYO1D	ENST00000318217:c.1539-1G>A None		homo	9.6	homo	unknown	1	(+) Microcephaly,(+) Hearing impairment,(+) Motor delay,(+) Infantile muscular hypotonia	
STK40	ENST00000359297:c.631dup p.Arg211ProfsTer46		unknown	6.8			1	NDD	(+) Respiratory insufficiency,(+) Developmental regression,(+) Muscle spasm,(+) Infantile muscular hypotonia,(+) Moderate global developmental delay
ZNF319	ENST00000299237:c.1375C>T p.Arg459Cys		de_novo	5.0	het	de novo	1	NDD + epilepsy	Cerebellar atrophy, Cerebral atrophy, Focal-onset seizure, Mild global developmental delay, Multifocal seizures
CDADC1	ENST00000251108:c.1219A>G p.Arg407Gly		de_novo	5.4	het	de novo	1	NDD + epilepsy	Seizure, Hypotonia, Global developmental delay, Polymicrogyria, Gray matter heterotopia, Abnormal cortical gyration, Focal-onset seizure, Aplasia/Hypoplasia of the corpus callosum, Severe global developmental delay, Brain atrophy, Abnormal myelination, Cerebellar atrophy, Cerebral atrophy
CEP350	ENST00000367607:c.7712A>G p.Asp2571Gly		de_novo	6.6	het	de novo	1	NDD	Motor stereotypy, Delayed speech and language development, Global developmental delay, Abnormal social behavior
INTS3	ENST00000318967:c.1008C>G p.Tyr336Ter		unknown	6.4	het	unknown	1	NDD	(+) Hydrocephalus,(+) Microcephaly,(+) Hypotonia,(+) Global developmental delay,(+) Failure to thrive,(+) Ventricular septal defect,(+) Spina bifida,(+) Hip contracture,(+) Short stature,(+) Autonomic bladder dysfunction,(+) Chiari type II malformation,(+) Myeloschisis,(+) Joint contracture,(+) Obstipation
MAP3K12	ENST00000267079:c.901T>A p.Trp301Arg		unknown	6.2	het	unknown	1	Epilepsy	(+) Bilateral tonic-clonic seizure,(+) Generalized non-motor (absence) seizure,(+) Generalized-onset seizure
KIF3C	ENST00000264712:c.1230_1243del p.Arg411ValfsTer8		unknown	9.2	het	unknown	1	NDD	(+) Global developmental delay,(+) Obesity
UBR4	ENST00000375254:c.7391C>G p.Ser2464Ter		de_novo	11.7	het	de novo	1	NDD + epilepsy	Polyhydramnios, Hypotonia, Seizure, Global developmental delay, Developmental regression, Receptive language delay, Abnormal myelination
UCHL5	ENST00000367448:c.271C>T p.Gln91Ter		unknown	6.6	het	unknown	1	NDD + obesity	Autism, obesity, infantile muscular hypotonia, moderate global developmental delay
MAML3	ENST00000327122:c.1472_1479del p.Gln491ProfsTer?		unknown	7.7	het	unknown	1	Hypermobility	(+) Trismus,(+) Tooth malposition,(+) Depression,(+) Striae distensae,(+) Joint dislocation,(+) Joint hypermobility,(+) Hip dysplasia,(+) Pes planus,(+) Back pain,(+) Chronic pain,(+) Piezogenic pedal papules,(+) Bowel irritability
GLI1	ENST00000228682:None None		homo	8.1	homo	maternal&paternal	1	NDD	Autism Intellectual disability, severe Severe global developmental delay Type I diabetes mellitus
SMTN	ENST00000333137:c.1222C>T p.Arg408Ter		homo	7.3	homo	maternal&paternal	2	NDD+ seizure	Focal-onset seizure Severe global developmental delay
TSSK4	ENST00000287913:c.865A>T p.Lys289Ter		homo	8.0	homo	maternal&paternal	2	NDD+ seizure	Focal-onset seizure Severe global developmental delay
IGF2BP1, UBE2Z			het		het	unknown	1	Failure to thrive	(+) Failure to thrive,(+) Feeding difficulties in infancy,(+) Moderate global developmental delay
CYTH3	ENST00000350796:c.1074C>G p.Tyr358Ter		unknown	5.1	het	unknown	1	Failure to thrive	(+) Failure to thrive,(+) Feeding difficulties in infancy,(+) Moderate global developmental delay
GALNT17	ENST00000333538:c.413G>T p.Arg138Leu		ad_inherited	5.4	het	maternal	2	other	Osteoarthritis, Premature osteoarthritis, Hip osteoarthritis, Knee osteoarthritis, Osteoarthritis of the elbow, Sacroiliac arthritis, Hip dysplasia, Arthralgia of the hip, Shoulder pain, Elbow pain, Hip pain, Knee pain, Ankle pain, Joint swelling, Joint stiffness, Skeletal dysplasia, Multiple epiphyseal dysplasia, Hypertension, Decreased circulating vitamin D concentration, Abnormal circulating lipid concentration, Hypertriglyceridemia
SLC8A3	ENST00000381269:c.2534T>G p.Ile845Ser		de_novo	8.6	het	de novo	2	other	Osteoarthritis, Premature osteoarthritis, Hip osteoarthritis, Knee osteoarthritis, Osteoarthritis of the elbow, Sacroiliac arthritis, Hip dysplasia, Arthralgia of the hip, Shoulder pain, Elbow pain, Hip pain, Knee pain, Ankle pain, Joint swelling, Joint stiffness, Skeletal dysplasia, Multiple epiphyseal dysplasia, Hypertension, Decreased circulating vitamin D concentration, Abnormal circulating lipid concentration, Hypertriglyceridemia
PPARGC1B	ENST00000309241:c.1807G>T p.Gly603Ter		unknown	6.2	het	unknown	1	NDD	(+) Atypical behavior,(+) Autism,(+) Severe global developmental delay,(+) Auditory sensitivity,(+) Recurrent hand flapping

NOP2	ENST00000322166:c.958C>T p.Arg320Ter		unknown	7.4	het	unknown	4	other	(+) Abnormal vagina morphology,(+) Primary amenorrhea,(+) Thoracolumbar scoliosis,(+) Cervical C2/C3 vertebral fusion,(+) Vertebral wedging,(+) Aplasia/hypoplasia of the uterus,(+) Aplasia/Hypoplasia involving the pelvis,(+) Unilateral renal hypoplasia
GCN1	ENST00000300648:c.3312+5G>A None		unknown	7.5	het	unknown	1	Autism	(+) Atypical behavior,(+) Autistic behavior
PCNX3	ENST00000355703:c.5315_5317del p.Ser1772del		de_novo	5.0	het	de novo	1	NDD + epilepsy	Pointed chin Smooth philtrum Protruding ear Downslanted palpebral fissures Autism Hypotonia Global developmental delay Generalized-onset seizure Delayed myelination Infantile spasms
ACO1	ENST00000309951:c.770C>G p.Ser257Cys		unknown	5.6	het	unknown	1	NDD + Epilepsy	(+) Microcephaly,(+) Autism,(+) Intellectual disability,(+) Global developmental delay,(+) Bilateral tonic-clonic seizure,(+) Myoclonic seizure
AGFG2	ENST00000300176:c.221+1G>A None		unknown	5.7	het	unknown	1	other	Proportionate short stature,(+) Relative macrocephaly
NTNG1	ENST00000370065:c.281C>G p.Ala94Gly		unknown	6.8	het	unknown	2	NDD + Epilepsy	(+) Hypotonia,(+) Generalized-onset seizure,(+) Tetraparesis,(+) Scoliosis,(+) Intellectual disability, severe,(+) Severe global developmental delay
BIRC6	ENST00000421745:c.6983G>T p.Cys2328Phe		unknown	5.3	het	unknown	2	NDD + Epilepsy	(+) Hypotonia,(+) Generalized-onset seizure,(+) Tetraparesis,(+) Scoliosis,(+) Intellectual disability, severe,(+) Severe global developmental delay
DIP2C	ENST00000280886:c.3554G>T p.Gly1185Val		unknown	6.1	het	unknown	1	NDD + Epilepsy	(+) Depression,(+) Intellectual disability, borderline,(+) Focal hyperkinetic seizure
MBD1	ENST00000269468:c.990dup p.Asn331GlufsTer64		unknown	8.1	het	unknown	1	short stature	(+) Generalized-onset seizure,(+) Short stature
PRDM2	ENST00000235372:c.5037-2A>C None		unknown	7.3	het	unknown	1	NDD+ hypotonia	(+) Poor head control,(+) Infantile muscular hypotonia,(+) Infantile axial hypotonia,(+) Mild global developmental delay
CDC42BPA	ENST00000334218:c.3977dup p.Leu1327SerfsTer16		unknown	7.4	het	unknown	1	NDD+ hypotonia	(+) Poor head control,(+) Infantile muscular hypotonia,(+) Infantile axial hypotonia,(+) Mild global developmental delay
VIRMA	ENST00000297591:c.1282del p.Gly429AlafsTer4		unknown	7.9	het	unknown	1	NDD+ hypotonia	(+) Poor head control,(+) Infantile muscular hypotonia,(+) Infantile axial hypotonia,(+) Mild global developmental delay
RPS12	ENST00000230050:c.223A>C p.Asn75His		de_novo	7.2	het	de novo	1		Restlessness, Hypotonia, Paresthesia, Enuresis nocturna
DDX19A	ENST00000302243:c.433_434del p.Ala145CysfsTer27		homo	8.9	het	unknown	1	NDD + Epilepsy	(+) Seizure,(+) Ataxia,(+) Spastic tetraparesis,(+) Dysphagia,(+) Moderate global developmental delay,(+) Joint contracture
ASAP1	ENST00000357668:c.2044-2A>G None		unknown	7.1	het	unknown	2	NDD	(+) Autistic behavior,(+) Hypotonia,(+) Attention deficit hyperactivity disorder,(+) Dyslexia,(+) Mild global developmental delay
SCAI	ENST00000336505:c.639_642del p.His214AspfsTer17		unknown	6.5	het	unknown	2	NDD	(+) Autistic behavior,(+) Hypotonia,(+) Attention deficit hyperactivity disorder,(+) Dyslexia,(+) Mild global developmental delay
PYGO1	ENST00000302000:c.832C>T p.Arg278Ter		unknown	5.6	het	unknown	1	NDD + Epilepsy	(+) Focal-onset seizure,(+) Moderate global developmental delay
ILF3	ENST00000588657:c.2252_2253insCTA p.Ser751_Gly752insTer		unknown	4.4	het	unknown	1	NDD + Epilepsy	(+) Focal-onset seizure,(+) Moderate global developmental delay
DIE	ENST00000265986.11:c.1327-16T>G, p.?		de_novo	6.8	het	de novo	2	NDD	(+) Delayed speech and language development,(+) Global developmental delay,(+) Hip dysplasia,(+) Ventricular septal defect,(+) Atrial septal defect,(+) Patent foramen ovale,(+) Gliosis,(+) Scoliosis,(+) EEG with focal epileptiform discharges,(+) Hyperintensity of cerebral white matter on MRI,(+) Diminished ability to concentrate
MAOB	ENST00000378069:c.280-13317C>T None		x_linked	6.2	hemi	maternal	2	NDD	(+) Delayed speech and language development,(+) Global developmental delay,(+) Hip dysplasia,(+) Ventricular septal defect,(+) Atrial septal defect,(+) Patent foramen ovale,(+) Gliosis,(+) Scoliosis,(+) EEG with focal epileptiform discharges,(+) Hyperintensity of cerebral white matter on MRI,(+) Diminished ability to concentrate
					het	unknown	1	NDD + Epilepsy	(+) Depression,(+) Aggressive behavior,(+) Intellectual disability, mild,(+) Obesity,(+) Ventriculomegaly,(+) Abnormal aggressive, impulsive or violent behavior,(+) Bilateral tonic-clonic seizure with focal onset,(+) Cerebral white matter atrophy
USP12	ENST00000282344:c.1106G>A p.Arg369Gln		homo	4.7	homo	maternal& paternal	1	NDD	Hypotonia, Feeding difficulties in infancy, Infantile muscular hypotonia, Severe global developmental delay
NTRK3	ENST00000317501:c.678_680delinsTGT p.Ala227Val		unknown	5.3	het	unknown	1	NDD	(+) Global developmental delay,(+) Developmental regression,(+) Cognitive impairment
RAB11FIP2	ENST00000355624:c.22C>T p.Gln8Ter		unknown	5.9	het	unknown	1	NDD + Epilepsy	(+) Autism,(+) Seizure,(+) Intellectual disability, mild,(+) Mild global developmental delay

PTPRM	ENST00000580170:c.4073_4076dup p.Phe1359LeufsTer17		unknown	7.3	het	unknown	1	NDD	(+) Autistic behavior,(+) Delayed speech and language development,(+) Pallor,(+) Intellectual disability,(+) Fatigue
ZBTB4	ENST00000311403:c.2035_2039dup p.Gly681LeufsTer60		unknown	B	het	unknown	1	Leukenzep halopathie	(+) Neurogenic bladder,(+) Gait disturbance,(+) Leukoencephalopathy,(+) Cognitive impairment
ACOT7	ENST00000361521:c.958T>C p.Phe320Leu		homo	6.6	homo	maternal& paternal	2	NDD + Epilepsie	Delayed speech and language development, Seizure, Cortical dysplasia
IQGAP1	ENST00000268182:c.2363G>C p.Gly788Ala		homo	6.3	homo	maternal& paternal	2	NDD + Epilepsie	Delayed speech and language development, Seizure, Cortical dysplasia
TULP4	ENST00000367094:c.1744C>T p.Arg582Trp		homo	5.9	homo	unknown	1	NDD + Epilepsy	Generalized hypotonia, Nystagmus, Astigmatism, Hypermetropia, Epicanthus, Toe clinodactyly, Lumbar hyperlordosis, Bilateral tonic-clonic seizure with focal onset
NGEF	ENST00000264051:c.990-3_990-1del None		unknown	8.1	het	unknown	1	other	(+) Paroxysmal dystonia
ADGRB2	ENST00000373655:c.2873G>A p.Trp958Ter		unknown	7.2	het	unknown	1	NDD	(+) Delayed speech and language development,(+) Intellectual disability, mild,(+) Moderate global developmental delay
XRCC6	ENST00000359308:c.244C>G p.Leu82Val		unknown	5.8	het	unknown	1	NDD + Epilepsie	(+) Spasticity,(+) Bilateral tonic-clonic seizure with focal onset,(+) Focal tonic seizure,(+) Mild global developmental delay,(+) Bilateral polymicrogyria
RAB11FIP4	ENST00000394744:c.355G>T p.Glu119Ter		unknown	6.9	het	unknown	1	NDD+ Epilepsie	(+) Hypotonia,(+) Early onset absence seizures,(+) Mild global developmental delay,(+) Focal atonic seizure
NRK	ENST00000243300:c.215del p.Val72GlyfsTer26		unknown	6.1	het	unknown	1	other	(+) Tall stature,(+) Unilateral renal agenesis,(+) Delayed puberty,(+) Septate vagina,(+) Uterus didelphys,(+) Decreased body mass index
QKI					het	unknown	1	NDD+ Epilepsie	(+) Delayed speech and language development,(+) Intellectual disability,(+) Hypotonia,(+) Obesity,(+) Focal-onset seizure,(+) Mild global developmental delay
JMJD6	ENST00000397625:c.856_862del p.Gln286LeufsTer15		unknown	8.0	het	unknown	1	Epilepsy	(+) Focal-onset seizure,(+) Focal hyperkinetic seizure,(+) Nocturnal seizures
RAB5A	ENST00000273047:c.278C>G p.Ala93Gly		unknown	5.5	het	unknown	1	Epilepsy	(+) Focal-onset seizure, (+) Aggressive behavior,(+) Attention deficit hyperactivity disorder
ARHGAP5	ENST00000345122:c.3563_3566del p.Arg1188LysfsTer34		unknown	7.9	het	unknown	1	NDD	(+) Myopia,(+) Delayed speech and language development,(+) Global developmental delay,(+) Delayed gross motor development,(+) Poor fine motor coordination
TRAF2	ENST00000247668:c.604-2A>G None		unknown	7.6	het	paternal	1	NDD+Epile psy	(+) Autism,(+) Hypotonia,(+) Generalized non-motor (absence) seizure,(+) Developmental regression,(+) EEG with abnormally slow frequencies,(+) Severe global developmental delay,(+) Generalized-onset motor seizure,(+) Abnormality of movement
EIF3L	ENST00000624234:c.853G>A p.Gly285Arg		unknown	4.9	het	unknown	1	NDD + Epilepsy	(+) Atypical behavior,(+) Delayed speech and language development,(+) Ataxia,(+) Bilateral tonic-clonic seizure,(+) Severe global developmental delay
EHMT2	ENST00000375537:c.3218C>G p.Ser1073Cys		unknown	7.2	het	unknown	1	other	(+) Functional abnormality of the bladder,(+) Urinary retention,(+) Spastic paraplegia,(+) Spastic tetraparesis,(+) Gait disturbance,(+) Asthma,(+) Spastic tetraplegia
PIK3C3	ENST00000262039:c.2432G>T p.Arg811Met		unknown	5.7	het	maternal	1	NDD	(+) Delayed speech and language development,(+) Global developmental delay,(+) Motor delay,(+) Short stature,(+) Cerebral hypomyelination
FAM171A1	ENST00000378116:c.1306del p.Glu436LysfsTer31		unknown	6.5	het	unknown	2	Epilepsy	(+) Vesicoureteral reflux,(+) Bilateral tonic-clonic seizure,(+) Generalized-onset seizure,(+) Attention deficit hyperactivity disorder,(+) Unilateral renal hypoplasia
NCAM1	ENST00000316851:c.875A>C p.Lys292Thr		unknown	7.0	het	unknown	2	Epilepsy	(+) Vesicoureteral reflux,(+) Bilateral tonic-clonic seizure,(+) Generalized-onset seizure,(+) Attention deficit hyperactivity disorder,(+) Unilateral renal hypoplasia
XRCC5	ENST00000392132:c.1373_1376del p.Ile458ThrfsTer3		unknown	8.5	het	unknown	1	NDD	(+) Irritability,(+) Premature birth,(+) Progressive language deterioration,(+) Severe global developmental delay
KDM4C	ENST00000381309:c.629+6T>G None		de_novo	6.4	het	de novo	1	Muscle	Generalized hypotonia, Motor axonal neuropathy, Progressive distal muscle weakness, Motor delay, Hyporeflexia
PHC2	ENST00000257118:c.1505dup p.Gly503TrpfsTer9		unknown	7.9	het	unknown	1	NDD	(+) Lymphedema,(+) Hypotonia,(+) Global developmental delay
RBM5	ENST00000347869:c.959_965del p.Leu320SerfsTer12		unknown	7.3	het	unknown	1	NDD	(+) Delayed speech and language development,(+) Diabetes mellitus,(+) Neonatal insulin-dependent diabetes mellitus,(+) Gait disturbance,(+) Hip dysplasia,(+) Short stature,(+) Moderate global developmental delay
LRP8	ENST00000306052:c.1948G>A p.Ala650Thr		unknown	7.0	het	unknown	1	NDD	(+) Intellectual disability, mild,(+) Global developmental delay,(+) Motor delay,(+) Monocular strabismus
GSE1	ENST00000253458:c.248C>A p.Ser83Ter		unknown	6.4	het	unknown	2	NDD	(+) Autistic behavior,(+) Delayed speech and language development,(+) Global developmental delay,(+) Recurrent respiratory infections,(+) Poor motor coordination
UBE4B	ENST00000253251:c.3314-2A>G None		unknown	8.4	het	unknown	2	NDD	(+) Autistic behavior,(+) Delayed speech and language development,(+) Global developmental delay,(+) Recurrent respiratory infections,(+) Poor motor coordination
HRH2	ENST00000377291:c.452_455del p.Phe151CysfsTer45		unknown	5.6	het	unknown	1	NDD	(+) Microcephaly,(+) Myopia,(+) Failure to thrive,(+) Hiatus hernia,(+) Attention deficit hyperactivity disorder,(+) Neurodevelopmental abnormality,(+) Abnormal social development, Joint hypermobility
TMEM131	ENST00000186436:c.5109-2A>G None		unknown	6.6	het	unknown	1	NDD	(+) Motor delay,(+) Failure to thrive

TENM4	ENST00000278550:c.1847G>A p.Ser616Asn		homo	7.25	homo	maternal&paternal	3	NDD	mild ID, facial dysmorphisms, Missing anterior teeth in the lower jaw, simian crease, dry skin, narrow mouth, behavioral abnormalities
LHX1	ENST00000614239:c.194dup p.Cys66LeufsTer8		unknown	7.9	het	unknown	1	NDD	(+) Microcephaly,(+) Strabismus,(+) Intellectual disability,(+) Coxa valga,(+) Syringomyelia,(+) Increased overbite,(+) Mild global developmental delay,(+) Chronic constipation,(+) Impulsivity
RALGPS1	ENST00000259351:c.1582C>T p.Arg528Ter		unknown	A	het	unknown	1	Stonweckers el+Fehlbild	Elevated circulating glutarylcarnitine concentration, close Dextrotransposition of the great arteries
ZFPM2	ENST00000407775:c.250_253del p.Ser84ArgfsTer51		de_novo	10.6	het	de novo		NDD	(+) High forehead,(+) Autism,(+) Delayed speech and language development,(+) Hyperactivity,(+) Pectus excavatum,(+) Global developmental delay,(+) Abnormal renal morphology,(+) Supravalvar pulmonary stenosis,(+) Self-injurious behavior
FBXO40			AR_comphet	A	comphet	maternal&paternal	1	Stonweckers el+Fehlbild	Hyperinsulinemic hypoglycemia,Hypotonia,Increased serum lactate,Hypoglycemia,Thrombocytopenia
GRM5	ENST00000305432:c.2943dup p.Ala982ArgfsTer185		unknown	10.76	het	unknown	2	NDD+epilepsy	(+) Abnormality of the face,(+) Focal-onset seizure,(+) Moderate global developmental delay
DLGAP1	ENST00000315677:c.2835dup p.Met946AspfsTer70		unknown	9.7	het	unknown	2	NDD+epilepsy	(+) Abnormality of the face,(+) Focal-onset seizure,(+) Moderate global developmental delay
SYMPK	ENST00000245934:c.3355-7T>G None		unknown	6.8	het	unknown	3	NDD	(+) Atypical behavior,(+) Autism,(+) Delayed speech and language development,(+) Hypotonia,(+) Global developmental delay,(+) Sleep abnormality,(+) Axial hypotonia
BCL2L11	ENST00000393256:c.79del p.Gln27SerfsTer57		de_novo	8.5	het	de novo	2	NDD	moderate global developmental delay
NRP1	ENST00000265371:c.2248C>T p.Gln750Ter		unknown	8.1	het	unknown	1	NDD	(+) Retrognathia,(+) Hypotonia,(+) Laryngomalacia,(+) Obstructive sleep apnea,(+) Mild global developmental delay
CHAF1A					het	de novo	1	NDD	(+) Hearing impairment,(+) Preauricular skin tag,(+) Hypotonia,(+) Failure to thrive,(+) Mild global developmental delay
KLHL2	ENST00000226725:c.1692C>A p.Asn564Lys		unknown	4.9	het	unknown	1	Epilepsie	(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Focal-onset seizure,(+) Focal seizures, afebril
SLC25A28	ENST00000370495:c.1089dup p.Lys364GlnfsTer4		homo	8.73	homo	maternal + paternal	1	NDD+Epilepsy	(+) Autism,(+) Hypotonia,(+) Generalized non-motor (absence) seizure,(+) Developmental regression,(+) EEG with abnormally slow frequencies,(+) Severe global developmental delay,(+) Generalized-onset motor seizure,(+) Abnormality of movement
SLC5A3	ENST00000381151:c.1531T>C p.Tyr511His		unknown	5.4	het	unknown	1	other	(+) Recurrent infections,(+) Arthralgia,(+) Calcium oxalate nephrolithiasis,(+) Raynaud phenomenon,(+) Hypoesthesia,(+) Tendon rupture
ARNT	ENST00000354396:c.179T>C p.Leu60Ser		de_novo	5.2	het	de novo	1	NDD + epilepsy	Microcephaly, Generalized non-motor (absence) seizure, Generalized myoclonic seizure, Mild global developmental delay
RSF1	ENST00000308488:c.1094dup p.Lys366GlufsTer4		unknown	7.5	het	unknown	1	NDD + epilepsy	Abnormal facial shape, Wide nose, Wide mouth, Primary microcephaly, Bilateral tonic-clonic seizure, Eyelid myoclonia seizure, Generalized myoclonic seizure, Atonic seizure, Posteriorly rotated ears
TMEM132B	ENST00000299308:c.3103G>T p.Glu1035Ter		unknown	6.4	het	materanl	3	other	Focal myoclonic seizure, EEG abnormality, Global developmental delay, Pes planus, Elbow hypertrichosis, Abnormal facial shape, Hypertelorism, Wide nasal bridge, Downslanted palpebral fissures, High, narrow palate, Abnormality of movement, Hyperreflexia, Hypotonia, Lower limb spasticity, Hand tremor, Gait disturbance, Poor fine motor coordination, Attention deficit hyperactivity disorder, Decreased body weight
ADCY1			unknown	7.7	het	unknown	1		(+) Hypsarrhythmia,(+) Epileptic spasm
LAMC1	ENST00000258341:c.2980del p.Gln994SerfsTer45		unknown	8.4	het	unknown	1		(+) Epileptic encephalopathy, (+) Focal-onset seizure, (+) Intellectual disability, (+) Atypical behavior
THRAP3	ENST00000354618:c.404C>T p.Pro135Leu		de_novo	7.0	het	de novo	1	NDD	Intellectual disability, mild, Global developmental delay, Delayed speech and language development, Generalized hypertrichosis
MAP3K12	ENST00000547488:c.980+1G>C None		unknown	8.8	het	unknown	1	NDD	(+) Microcephaly,(+) Delayed speech and language development,(+) Intellectual disability,(+) Hypotonia
MORC4	ENST00000355610:c.1700del p.Ile567ThrfsTer2		x_linked	7.0	het	unknown	1	other	Abnormality of the hand, Flexion contracture of finger, Upper limb undergrowth
HECTD1	ENST00000399332:c.3109A>T p.Lys1037Ter		unknown	7.8	het	unknown	2	NDD+epilepsy	(+) Polymicrogyria,(+) Abnormal cerebral cortex morphology,(+) Unilateral polymicrogyria,(+) Focal-onset seizure,(+) Mild global developmental delay
CHD9	ENST00000398510:c.5270A>T p.Asp1757Val		unknown	5.7	het	unknown	2	NDD+epilepsy	(+) Polymicrogyria,(+) Abnormal cerebral cortex morphology,(+) Unilateral polymicrogyria,(+) Focal-onset seizure,(+) Mild global developmental delay
ANKHD1	ENST00000360839:c.1126C>A p.Leu376Ile		unknown	5.6	het	unknown	1	Autism	(+) Autism,(+) Absent speech,(+) Receptive language delay,(+) Abnormal interpretation of external stimuli
ASIC1	ENST00000228468:c.474C>G p.Tyr158Ter		unknown	6.7	het	unknown	1	epilepsy	(+) Generalized non-motor (absence) seizure,(+) Early onset absence seizures
DPP9						de novo	1	NDD	(+) Hearing impairment,(+) Preauricular skin tag,(+) Hypotonia,(+) Failure to thrive,(+) Mild global developmental delay

RANBP2	ENST00000283195:c.3591C>G p.Asn1197Lys		de_novo	7.5	het	de novo	1	NDD	Anal stenosis, Scoliosis, Grade IV vesicoureteral reflux, Unilateral ptosis, Hypotonia, Mild global developmental delay
ATAD2B	ENST00000238789:c.3257G>T p.Gly1086Val		unknown	5.6	het	unknown	1	epilepsy	(+) Febrile seizure (within the age range of 3 months to 6 years),(+) Bilateral tonic-clonic seizure with generalized onset,(+) Generalized-onset motor seizure
RXRB	ENST00000374680:c.1493C>G p.Ser498Cys		unknown	5.0	het	unknown	1	NDD	(+) Atypical behavior,(+) Intellectual disability, mild
ZNF76	ENST00000339411:c.432+5G>A None		homo	8.7	homo	maternal&paternal	1	NDD	Global developmental delay, Delayed speech and language development, Intellectual disability, Tip-toe gait, Infantile muscular hypotonia, Genu valgum, Hydronephrosis, Autism, Restlessness
KCNJ4	ENST00000303592:c.406G>A p.Gly136Ser		unknown	5.7	het	unknown	1	NDD+epilepsy	(+) Thick upper lip vermillion,(+) Microcephaly,(+) Smooth philtrum,(+) Low-set ears,(+) Intellectual disability,(+) Focal-onset seizure,(+) Severe global developmental delay,(+) Brain atrophy
PPP1R13B	ENST00000202556:c.457-2A>T None		unknown	A	het	unknown	1	Leukencephalopathie	Leukoencephalopathy, memory impairment, dminished ability to concentrate, depression, antinuclear antibody positivty
RPS2	ENST00000343262:c.752T>C p.Leu251Pro		unknown	5.7	het	unknown	1	NDD+epilepsy	(+) Seizure,(+) Ataxia,(+) Spasticity,(+) Global developmental delay,(+) Short stature,(+) Intellectual disability, severe
NSD3	ENST00000317025:c.3410A>G p.Gln1137Arg		unknown	5.8	het	unknown	1	NDD+epilepsy	(+) Polymicrogyria,(+) Focal-onset seizure,(+) Mild global developmental delay
NSD3	ENST00000317025:c.4278G>A p.Trp1426Ter		unknown	7.7	het	unknown	3	NDD	(+) Microcephaly,(+) Atypical behavior,(+) Short stature,(+) Moderate global developmental delay
LRRC37B	ENST00000341671:c.639del p.Lys214AsnfsTer14		unknown	6.8	het	unknown	3	NDD	(+) Microcephaly,(+) Atypical behavior,(+) Short stature,(+) Moderate global developmental delay
PRKCA	ENST00000413366:c.355G>A p.Gly119Arg		unknown	6.5	het	unknown	3	NDD	(+) Microcephaly,(+) Atypical behavior,(+) Short stature,(+) Moderate global developmental delay
CLIP2	ENST00000223398:c.803+1G>C None		unknown	10.7	het	unknown	1	NDD+epilepsy	(+) Intellectual disability, mild,(+) Myoclonic seizure
FMNL2	ENST00000288670:c.1037A>C p.Lys346Thr		unknown	4.7	het	unknown	2	NDD+epilepsy	(+) Intellectual disability, mild,(+) Generalized non-motor (absence) seizure,(+) Focal clonic seizure,(+) Bilateral tonic-clonic seizure with focal onset,(+) Focal tonic seizure
XPO5	ENST00000265351:c.218A>G p.His73Arg		unknown	5.3	het	unknown	2	NDD+epilepsy	(+) Intellectual disability, mild,(+) Generalized non-motor (absence) seizure,(+) Focal clonic seizure,(+) Bilateral tonic-clonic seizure with focal onset,(+) Focal tonic seizure
CMTR1	ENST00000373451:c.704+5G>A None		ad_inherited	6.7	het	unknown	1	NDD+epilepsy	(+) Wide mouth,(+) Coarse facial features,(+) Abnormal facial shape,(+) Generalized myoclonic-atonic seizure,(+) Focal hyperkinetic seizure,(+) Severe global developmental delay
IFT20	ENST00000357896:c.346del p.Glu116AsnfsTer57		unknown	7.5	het	unknown	1	NDD	(+) Restlessness,(+) Aggressive behavior,(+) Autistic behavior,(+) Motor stereotypy,(+) Delayed speech and language development,(+) Plagiocephaly,(+) Moderate global developmental delay
FMNL3	ENST00000335154:c.2228T>A p.Leu743His		unknown	3.8	het	unknown	1	NDD+Epilepsie	(+) Atypical behavior,(+) Abnormal skull morphology,(+) Ataxia,(+) Focal-onset seizure,(+) Severe global developmental delay
TNIP1	ENST00000315050:c.955C>T p.Gln319Ter		unknown	4.9	het	unknown	1	other	(+) Failure to thrive,(+) Colitis,(+) Celiac disease,(+) Autoimmunity,(+) Myopathy,(+) Short stature,(+) Dust mite allergy,(+) Myositis-specific autoantibody positivity
ANKFY1	ENST00000341657:c.1331T>C p.Ile444Thr		unknown	5.4	het	unknown	1	other	(+) Depression,(+) Parkinsonism,(+) Parkinsonism with favorable response to dopaminergic medication
KMT5C	ENST00000255613:c.125del p.Leu42ArgfsTer15		de_novo	7.9	het	de novo	1	NDD + epilepsy	Intellectual disability, mild, Bilateral tonic-clonic seizure with focal onset, Focal motor seizure
ZNF385B	ENST00000336917:c.299-72908A>G None		homo	4.6	homo	maternal&paternal	1	NDD	Microcephaly, Cerebellar atrophy, Syncope, Neurodegeneration, EEG abnormality, Moderate global developmental delay
FMO4	ENST00000367749:c.1180+3A>G None		homo	8.4	homo	maternal&paternal	4	NDD + epilepsy	Hypotonia, Focal-onset seizure, Severe global developmental delay
TMEM221	ENST00000341130:c.320+2T>A None		homo	8.4	homo	maternal&paternal	4	NDD + epilepsy	Hypotonia, Focal-onset seizure, Severe global developmental delay
ENY2	ENST00000521662:c.-10+3A>G None		homo	8.2	homo	maternal&paternal	4	NDD + epilepsy	Hypotonia, Focal-onset seizure, Severe global developmental delay
NUP210	ENST00000254508:c.3608A>G p.Asn1203Ser		homo	5.9	homo	maternal&paternal	4	NDD + epilepsy	Hypotonia, Focal-onset seizure, Severe global developmental delay
UBQLN4	ENST00000368309:c.1688T>C p.Leu563Pro		de_novo	7.4	het	de novo	1	NDD	Microcephaly, Autism, Delayed speech and language development, Moderate global developmental delay
ARHGEF11	ENST00000361409:c.2095G>T p.Asp699Tyr		de_novo	6.7	het	de novo	1	other	Hemiparesis, Stroke, Ischemic stroke, Aphasia, Arterial stenosis
PGD	ENST00000270776:c.354del p.Lys119ArgfsTer65		unknown	7.8	het	unknown	1	other	EBV encephalitis, Immunodeficiency, Recurrent infections
NUDT13	ENST00000349051:c.359C>T p.Ala120Val		de_novo	4.8	het	de novo	1	malformation	(+) Astigmatism,(+) Myopia,(+) Atrial septal defect,(+) Radioulnar synostosis,(+) Dislocated radial head,(+) Abnormality of the radial head,(+) Pseudobulbar paralysis,(+) Sacralization of the fifth lumbar vertebra,(+) Abnormality of the humeroradial joint

PIK3CB	ENST00000289153:c.2636A>T p.Asp879Val		unknown	6,38	het	mother?	1	perisylvian Polymikrogyrie, fokale Epilepsie	(+) Focal-onset seizure,(+) Perisylvian polymicrogyria
PARP8	ENST00000281631:c.2384C>T p.Thr795Ile		homo	5.1	homo	maternal&paternal	2	NDD	Severe global developmental delay, Short stature, Hypotonia
MGAM	ENST00000475668:c.2185C>T, p.Arg729Cys	7:141736731:C:T	AR_comphet	4.5	het	maternal&paternal	2	NDD	Severe global developmental delay, Short stature, Hypotonia
PCDH1	ENST00000287008:c.2237_2238del p.Ser746TrpfsTer5		de_novo	8.8	het	de novo	1	NDD + epilepsy	(+) Atypical behavior,(+) Delayed speech and language development,(+) Generalized non-motor (absence) seizure,(+) Intellectual disability, borderline
FBP2	ENST00000375337:c.710G>A p.Gly237Asp		AR_comphet	5.6	het	maternal&paternal	1	NDD	Microcephaly, Seizure, Absent speech, Developmental regression, Inability to walk, Severe global developmental delay, Dyskinesia
REV1	ENST00000258428:c.54+1G>A None		unknown	7.8	het	unknown	1	other + epilepsy	(+) Cerebral hemorrhage,(+) Recurrent cerebral hemorrhage,(+) Focal-onset seizure
SMU1	ENST00000397149:c.478C>T p.Arg160Cys		de_novo	6.8	het	de novo	1	NDD + other	Intrauterine growth retardation, Short stature, Microcephaly, Abnormality of the face, High palate, Tooth malposition, Global developmental delay, Generalized non-motor (absence) seizure
VPS18	ENST00000220509:c.724C>T p.Gln242Ter		unknown	8.0	het	unknown	1	other	(+) Rickets,(+) Hyperphosphatemia,(+) Genu varum,(+) Hypercalcemia,(+) Childhood-onset truncal obesity,(+) Abnormality of vitamin D metabolism
ZNF106	ENST00000263805:c.4677+5G>A None		unknown	6.9	het	unknown	1	NDD + seizure	(+) Abnormal periventricular white matter morphology,(+) Neurodevelopmental delay,(+) Focal-onset epileptic spasm
SCHIP1	ENST00000412423:c.978T>G p.Tyr326Ter		ad_inherited	5.5	het	paternal inherited	1	NDD + seizure	Seizure, Nocturnal seizures, Exercise-induced muscle cramps, Focal dystonia, Eyelid myoclonia seizure, Focal impaired awareness seizure, EEG abnormality, Hyperparathyroidism
EPHA4	ENST00000281821:c.2030T>G p.Phe677Cys		unknown	7.2	het	unknown		ID+NDD, ADHD	(+) Intellectual disability,(+) Obesity,(+) Attention deficit hyperactivity disorder,(+) Severe global developmental delay
H2AX	ENST00000530167:c.300_301del p.Val101AspfsTer129		unknown	7.9	het	unknown		NDD+ID, language delay	(+) Cleft palate,(+) Ptosis,(+) Carious teeth,(+) Hypotonia,(+) Global developmental delay,(+) Unsteady gait,(+) Expressive language delay,(+) Hyperlordosis,(+) Intellectual disability, borderline,(+) Skewfoot
NSF	ENST00000398238:c.1660G>A p.Ala554Thr		unknown	7.5	het	unknown		NDD+ID, language delay	(+) Cleft palate,(+) Ptosis,(+) Carious teeth,(+) Hypotonia,(+) Global developmental delay,(+) Unsteady gait,(+) Expressive language delay,(+) Hyperlordosis,(+) Intellectual disability, borderline,(+) Skewfoot
KDM4A	ENST00000372396:c.146C>G p.Pro49Arg		unknown	5.1	het	unknown		hemimegalencephalie, NDD, Epilepsy	(+) Spasticity,(+) Talipes equinovarus,(+) Hemimegalencephaly,(+) Focal-onset seizure,(+) Spastic hemiparesis,(+) Moderate global developmental delay,(+) Exodeviation,(+) Focal-onset epileptic spasm
TNK2	ENST00000333602:c.2692_2695del p.Ser898ProfsTer72		unknown	7.0	het	unknown		NDD, Autism, Epilepsy	(+) Severe global developmental delay,(+) Self-injurious behavior
AREL1	ENST00000356357:c.736C>T p.Arg246Ter		unknown	5.5	het	unknown		NDD, Autism	(+) Severe global developmental delay,(+) Self-injurious behavior
ADRM1	ENST00000253003:c.447del p.Leu150Ter		unknown	7.1	het	unknown		NDD+ID	(+) Atypical behavior,(+) Intellectual disability,(+) Neurodevelopmental delay
AKAP13	ENST00000361243:c.7524+2del None		unknown	7.8	het	unknown	2	NDD	(+) Atopic dermatitis,(+) Failure to thrive,(+) Ventriculomegaly,(+) Increased circulating IgE concentration,(+) Cessation of head growth,(+) Severe global developmental delay,(+) Cutaneous abscess,(+) Food allergy
WWP1	ENST00000265428:c.1045G>T p.Glu349Ter		unknown	6.94	het	unknown	2	NDD	(+) Atopic dermatitis,(+) Failure to thrive,(+) Ventriculomegaly,(+) Increased circulating IgE concentration,(+) Cessation of head growth,(+) Severe global developmental delay,(+) Cutaneous abscess,(+) Food allergy
IWS1	ENST00000295321:c.1565+1_1565+2insA None		unknown	7.1	het	unknown	1	Epilepsy	(+) Absence seizure with eyelid myoclonia,(+) Bilateral tonic-clonic seizure with generalized onset
PRP4K	ENST00000337659:c.1861+1G>A None		unknown	6.9	het	unknown		Epilepsy	Focal motor seizure,(+) Focal motor status epilepticus
DDX42	ENST00000359353:c.2170G>T p.Gly724Ter		unknown	7.1	het	unknown		Epilepsy	Focal motor seizure,(+) Focal motor status epilepticus
DGKD	ENST00000264057:c.3164T>C p.Leu1055Pro		unknown	4.3	het	unknown		NDD + epilepsy	(+) Tall stature,(+) Intellectual disability,(+) Spasticity,(+) Focal-onset seizure,(+) Severe global developmental delay
ECPAS	ENST00000259335:c.4711+2T>G None		unknown	7.2	het	unknown		NDD	(+) Autism,(+) Global developmental delay
ACTR2	ENST00000260641:c.695A>T p.Lys232Ile		unknown	6.4	het	unknown		NDD	(+) Obesity,(+) Intellectual disability, severe,(+) Severe global developmental delay
DNAJC11	ENST00000294401:c.817_818insT p.Arg273LeufsTer11		unknown	7,27	het	unknown	2	NDD	(+) Dysphagia,(+) Progressive spasticity,(+) Intellectual disability, moderate,(+) Generalized dystonia,(+) Rigors
H2AC21	ENST00000331128:c.88dup p.Arg30ProfsTer62		unknown	6.9	het	unknown	2	NDD	(+) Dysphagia,(+) Progressive spasticity,(+) Intellectual disability, moderate,(+) Generalized dystonia,(+) Rigors
ATXN7L1	ENST00000318724:c.72dup p.Glu25ArgfsTer57		unknown	6.0	het	unknown	2	NDD + epilepsy	(+) Hypotonia,(+) Global developmental delay,(+) Hip dysplasia,(+) Intellectual disability, moderate,(+) Focal-onset seizure

SETDB2	ENST00000317257:c.728T>C p.Ile243Thr		unknown	3.9	het	unknown	2	NDD + epilepsy	(+) Hypotonia,(+) Global developmental delay,(+) Hip dysplasia,(+) Intellectual disability, moderate,(+) Focal-onset seizure
ZNF107	ENST00000620827:c.1162A>T p.Lys388Ter		homo	6.9	homo	unknown	1	NDD	(+) Hypotonia,(+) Mild global developmental delay
CAND1	ENST00000545606:c.176A>G p.Asp59Gly		unknown	5.0	het	unknown	1	NDD	(+) Microcephaly,(+) Mild global developmental delay
SND1	ENST00000354725:c.391G>A p.Gly131Ser		unknown	6.52	het	unknown		NDD, Microcephaly	(+) Microcephaly,(+) Premature birth,(+) EEG abnormality,(+) Short stature,(+) Attention deficit hyperactivity disorder,(+) Neurodevelopmental delay
BAZ2A	ENST00000379441:c.1789_1790del p.Thr597AlafsTer32		unknown	7.8	het	unknown	1	NDD, Microcephaly	(+) Microcephaly,(+) Premature birth,(+) Mild global developmental delay,(+) Intraventricular hemorrhage
PICK1	ENST00000356976:c.674T>C p.Leu225Pro		unknown	5.9	het	unknown	1	other	(+) Motor delay,(+) Elevated circulating creatine kinase concentration,(+) Muscular dystrophy
TNS3	ENST00000311160:c.4156C>T p.Gln1386Ter		unknown	6.9	het	unknown		NDD, brain malformation	(+) Neurogenic bladder,(+) Hypotonia,(+) Cerebellar hypoplasia,(+) Bradycardia,(+) Unexplained fevers,(+) Syringomyelia,(+) Severe hydrocephalus,(+) Paraplegia,(+) Neurodevelopmental delay,(+) Chiari type II malformation,(+) Myeloschisis,(+) Small posterior fossa
DROSHA	ENST00000344624:c.3218A>T p.Asp1073Val		ad_inherited	5.4	het	maternal	1	other	Splenomegaly, Decreased total lymphocyte count, Lymphadenopathy, Recurrent infections, Decreased circulating IgA concentration, Immunodeficiency, Autoimmunity, Decreased circulating total IgA, Increased circulating IgM level, Decreased circulating IgG concentration, Decreased total T cell count, Decreased total B cell count, Thrombocytopenia
OSBP	ENST00000263847:c.2054C>G p.Pro685Arg		unknown	4.6	het	unknown	1	NDD	(+) Autism,(+) Intellectual disability,(+) Moderate global developmental delay
FAM160B1	ENST00000369248:c.1772A>G p.Tyr591Cys		unknown	5.0	het	unkown	1	NDD	(+) Tall stature,(+) Atypical behavior,(+) Intellectual disability, mild,(+) Obesity,(+) Mild global developmental delay
PPP1R26	ENST00000356818:c.3027del p.Ser1010AlafsTer144		homo	9.4	homo	unknown	1	NDD	(+) Autism,(+) Short stature,(+) Moderate global developmental delay
PIGX			comphet	6.1	comphet	maternal&paternal	1	NDD	Seizure, Hypotonia, Rotary nystagmus, Horizontal jerk nystagmus, Atonic seizure, Delayed myelination, Myoclonic seizure, Global developmental delay
CSE1L	ENST00000262982:c.660T>A p.Tyr220Ter		ad_inherited	7.5	het	unknown	1	NND + epilepsy	(+) Focal-onset seizure,(+) Reduced visual acuity,(+) Moderate global developmental delay,(+) Abnormal hippocampus morphology
TMEM131	ENST00000186436:c.2701C>T p.Gln901Ter		unknown	6.1	het	unknown	1	NDD	(+) Autistic behavior,(+) Moderate global developmental delay,(+) Abnormal social development
TNKS	ENST00000310430:c.2740G>A p.Gly914Arg		ad_inherited	A	het	unknown	1	obesity	(+) Anxiety,(+) Enuresis,(+) Global developmental delay,(+) Obesity
PLK2	ENST00000274289:c.379-1G>C None		unknown	7.5	het	unknown	1	other	(+) Depression,(+) Vertigo,(+) Fatigue,(+) Focal hyperintensity of cerebral white matter on MRI
NOL4	ENST00000261592:c.2T>G p.Met1?		unknown	6.0	het	unknown	2	NDD + epilepsy	(+) Specific learning disability,(+) Typical absence seizure,(+) Refractory drug response
FBXO32	ENST00000517956:c.804del p.Lys268AsnfsTer16		unknown	7.1	het	unknown	2	NDD + epilepsy	(+) Specific learning disability,(+) Typical absence seizure,(+) Refractory drug response
SERBP1					het	unknown	1	other	(+) Immunodeficiency,(+) Decreased circulating antibody concentration,(+) Agammaglobulinemia,(+) B lymphocytopenia
TRPM7	ENST00000560955:c.4-2097G>A None		homo	4.8	het	unknown	1	other	(+) Seizure,(+) Muscle weakness,(+) Failure to thrive in infancy,(+) Hypokinesia,(+) Respiratory failure requiring assisted ventilation,(+) Infantile muscular hypotonia,(+) Elevated urinary 4-hydroxybutyric acid,(+) Increased CSF glutamine concentration,(+) Elevated urine 4,5-dihydroxyhexanoic acid level
TM9SF2	ENST00000376387:c.1475G>A p.Gly492Asp		unknown	4.9	het	unknown	1	other	(+) Urticaria,(+) Leukopenia,(+) Immunodeficiency,(+) T lymphocytopenia,(+) Systemic autoinflammation
UPF1	ENST00000262803:c.1627C>T p.Arg543Cys		unknown	6.5	het	unknown	1	NDD + epilepsy	(+) Atypical behavior,(+) Intellectual disability, mild,(+) Pes planus,(+) Bilateral tonic-clonic seizure,(+) Genu valgum,(+) Focal motor seizure,(+) Moderate global developmental delay,(+) Cutaneous syndactyly
NAV2					het	unknown	1	Epilepsy	(+) Generalized-onset seizure,(+) Tonic seizure
CDH8	ENST00000299345:c.1414+2T>C None		unknown	8.8	het	maternal	1	NDD	Global developmental delay, Delayed speech and language development, Hyperactivity, Autistic behavior, Hypermetropia
ZBTB4	ENST00000311403:c.1131C>A p.Tyr377Ter		unknown	6.1	het	unknown	1	NDD	Global developmental delay Intellectual disability check Absent speech close Relative macrocephaly High, narrow palate check Wide mouth Low-set ears
BRD1	ENST00000404760:c.2737dup p.Thr913AsnfsTer88		unknown	A	het	unknown	1	other	(+) Headache,(+) Exercise-induced muscle cramps,(+) Exercise-induced leg cramps
RPS6KB1	ENST00000225577:c.904del p.Ile302LeufsTer27		unknown	8.8	het	maternal	1	NDD	Autistic behavior, Global developmental delay, Absent speech, Short palpebral fissure, Hypotelorism, Midface retrusion, Abnormal facial shape, Aggressive behavior
MDGA2	ENST00000439988:c.2592A>T p.Glu864Asp		unknown	2.7	het	unknown	1	other	(+) Hydrocephalus,(+) Strabismus,(+) Spasticity
EPHA4	ENST00000281821:c.2846A>G p.Glu949Gly		unknown	7.2	het	unknown	1	other	Abnormal morphology of female internal genitalia, Sensorineural hearing impairment, Microscopic hematuria, Female infertility, Feeding difficulties in infancy, Albuminuria, Osteoporosis

DENR	ENST00000280557:c.126+2del None		unknown	6.9	het	unknown	1	Epilepsy	(+) Intellectual disability, mild,(+) Focal-onset seizure,(+) Nocturnal seizures,(+) Focal impaired awareness motor seizure
CKAP5	ENST00000529230:c.1875+5_1875+6del None		unknown	4.2	het	unknown	1	NDD + epilepsy	(+) Generalized-onset seizure,(+) Mild global developmental delay
CIAO1	ENST00000488633:c.563C>T p.Ala188Val		homo	5.0	homo		1	Epilepsy	family is consanguineous and as we got the probe, it was a one-month-old infant with epileptic encephalopathy, seizures, EEG with burst suppression, abnormal brain stem MRI signal intensity, and abnormal basal ganglia MRI signal intensity
NOL4L	ENST00000359676:c.388dup p.Ser130PhefsTer21		unknown	6.2	het	unknown	1	NDD + epilepsy	(+) Delayed speech and language development,(+) Generalized-onset seizure
AQR	ENST00000156471:c.1960A>T p.Asn654Tyr		unknown	5.2		unknown	1	epilepsy	(+) Intellectual disability, mild,(+) Focal-onset seizure,(+) Mild global developmental delay,(+) Febrile status epilepticus,(+) Febrile seizure outside the age of 3 months to 6 years
PPARD	ENST00000311565:c.-101-1G>A None		unknown	7.9	het	unknown	1	other	(+) Macrocephaly,(+) Strabismus,(+) Hypermetropia,(+) Nystagmus,(+) Atypical behavior,(+) Ocular albinism,(+) Sensory hypersensitivity
DLG2	ENST00000280241:c.2446+1G>A None		unknown	9.1	het	unknown	1	NDD + epilepsy	(+) Global developmental delay,(+) Generalized-onset seizure,(+) Focal-onset seizure
ADD2	ENST00000264436:c.555+5G>A None		unknown	7.3	het	unknown	1	Epilepsy	(+) Generalized-onset seizure,(+) Bilateral tonic-clonic seizure with generalized onset
DCP1A	ENST00000294241:c.250_262del p.Lys84PhefsTer31		unknown	6.6	het	unknown	1	NDD	(+) Carious teeth,(+) Dyscalculia
DPYSL2	ENST00000311151:c.1160A>T p.Asn387Ile		unknown	7.1	het	unknown	1	NDD	(+) Atypical behavior,(+) Autistic behavior,(+) Delayed speech and language development,(+) Moderate global developmental delay
PTPRS	ENST00000262963:c.4733A>T p.Asn1578Ile		unknown	5.5	het	unknown	1	other	Hemangioma Bilateral talipes equinovarus Iron deficiency anemia Epicanthus Feeding difficulties Neonatal hypoglycemia Abnormal immune system morphology Floppy infant
MAP3K2	ENST00000682094:c.745+1G>A None		unknown	6.9	het	unknown	1	NDD + epilepsy	(+) Unilateral polymicrogyria,(+) Focal-onset seizure,(+) Speech apraxia,(+) Mild global developmental delay
ZNF644	ENST00000337393:c.3856C>T p.Arg1286Ter		unknown	7.5	het	unknown	1	NDD	rinary incontinence, Intellectual disability, Obesity, Mild hearing impairment, Myopia, Global developmental delay, Small for gestational age, Enuresis diurna, Depression
RPTOR	ENST00000306801:c.3693-3C>A None		unknown	9.2	het	unknown	1	NDD+Epi	(+) Autism,(+) Short stature,(+) Focal-onset seizure,(+) Intellectual disability, severe,(+) Severe global developmental delay
PSPC1	ENST00000338910:c.1213del p.(Val405Leufs*9)		unknown	7.2	het	unknown	1	NDD+Epi	(+) Cleft palate,(+) Atresia of the external auditory canal,(+) Hypotonia,(+) Polymicrogyria,(+) Focal-onset seizure,(+) Intellectual disability, severe,(+) Rhombencephalosynapsis,(+) Cleft lip,(+) Goldenhar syndrome
DPF3	ENST00000556509:c.282_285del p.Leu95TrpfsTer3		unknown	6.7	het	unknown	2	NDD + other	(+) Global developmental delay,(+) Ventricular septal defect,(+) Esophageal atresia,(+) Hiatus hernia,(+) Short stature
EIF4A1	ENST00000293831:c.1075A>G p.Arg359Gly		unknown	6.1	het	unknown	2	NDD + other	(+) Global developmental delay,(+) Ventricular septal defect,(+) Esophageal atresia,(+) Hiatus hernia,(+) Short stature
BRD8	ENST00000230901:c.1048C>T p.Gln350Ter		de_novo	6.3	het	de novo	1	other	Hypospadias, Ambiguous genitalia, Hypertyrosinemia, Extremely preterm birth, Periauricular skin pits, Micropenis, Intrauterine growth retardation, Decreased total neutrophil count, Wide anterior fontanel, Inguinal hernia, Increased circulating androstenedione concentration, Elevated circulating 17-hydroxyprogesterone concentration, Abnormal circulating dehydroepiandrosterone concentration, Abnormal circulating luteinizing hormone concentration
PEX5L	ENST00000467460:c.1411_1412del p.Met471AspfsTer20		unknown	7.8	het	unknown	1	NDD	(+) Autistic behavior,(+) Delayed speech and language development,(+) Global developmental delay
SIPA1L1	ENST00000381232:c.172dup p.Arg58ProfsTer3		unknown	7.9	het	unknown	1	NDD + epilepsy	Myopia, Focal aware autonomic seizure, Atonic seizure, Focal aware sensory seizure, Atypical behavior, Allergy, Focal impaired awareness autonomic seizure, Self-injurious behavior, Depression, Febrile seizure (within the age range of 3 months to 6 years), EEG abnormality, Short attention span
OSBP	ENST00000263847:c.1311G>A p.Pro437=		unknown	6.9	het	unknown	2	NDD	Motor delay, Generalized hypotonia, Severe global developmental delay, Plagiocephaly, Hypermetropia, Bicuspid aortic valve, Atopic dermatitis, Motor stereotypy
TUT7	ENST00000277141:c.1909C>T p.Arg637Ter		unknown	5.9	het	unknown	2	NDD	Motor delay, Generalized hypotonia, Severe global developmental delay, Plagiocephaly, Hypermetropia, Bicuspid aortic valve, Atopic dermatitis, Motor stereotypy
NPAS2	ENST00000335681:c.40del p.Arg14GlufsTer75		unknown	8.65	het	unknown	1	other	Tall stature Megalencephaly
PTK2	ENST00000340930:c.895G>C p.Val299Leu		AD_inherited	6.0	het	paternal	1	NDD	Cryptorchidism, Microcephaly, Expressive language delay, Short stature, Decreased body weight, Decreased circulating total IgG concentration, Failure to thrive, Immunodeficiency, Recurrent fever, Autistic behavior
CKAP5	ENST00000312055:c.4417del p.His1473IlefsTer16		unknown	9.2	het	unkown	1	NDD	(+) Hearing impairment,(+) Visual impairment,(+) Intellectual disability,(+) Severe global developmental delay

ASCL1	ENST00000266744:c.64C>T p.Gln22Ter		unknown	8.9	het	unkown	1	NDD + Epilepsie	(+) Dysarthria,(+) Focal-onset seizure,(+) Mild global developmental delay
DNMT1	ENST00000340748:c.2159del p.Gln720ArgfsTer57		unknown	10.2	het	unkown		other	(+) Vesicoureteral reflux,(+) Wide mouth,(+) Hypertelorism,(+) Delayed speech and language development,(+) Tremor,(+) Failure to thrive,(+) Frontal bossing,(+) Elevated circulating hepatic transaminase concentration,(+) Elevated circulating creatine kinase concentration
HNRNPUL1	ENST00000378215:c.2T>C p.Met1?		unknown	6.3	het	unknown	1	Epilepsy	(+) Bilateral tonic-clonic seizure with generalized onset
VEZF1	ENST00000581208.2:c.550C>T p.(Arg184*)		unknown	7.3	het	unknown	1	NDD	(+) Delayed speech and language development,(+) Hyperactivity,(+) Intellectual disability, mild,(+) Mild global developmental delay
SSBP3	ENST00000357475:c.164C>G p.Pro55Arg		unknown	5.6	het	unknown	1	other	Edema, Increased circulating lactate concentration, Paraparesis, Arrhythmia, Limb tremor, Chilblains, Raynaud phenomenon, Myopathy, Exercise-induced myalgia, Exercise-induced muscle cramps, Abnormality of pain sensation, Chronic pain, Chronic fatigue, Arthralgia, Urticaria, Limb muscle weakness
TTC17	ENST00000039989:c.2153T>G p.(Leu718*)		unknown	6.5	het	unknown	1	NDD	(+) Tall stature,(+) Autism,(+) Delayed speech and language development,(+) Mild global developmental delay,(+) Cognitive impairment
NAA25	ENST00000261745.9:c.475C>G p.(Gln159Glu)		unknown	4.5	het	unknown	1	NADD	(+) Pierre-Robin sequence,(+) Microcephaly,(+) Conductive hearing impairment,(+) Glaucoma,(+) Developmental cataract,(+) Atypical behavior,(+) Intellectual disability,(+) Expressive language delay,(+) Focal-onset seizure,(+) Anisocoria,(+) Moderate global developmental delay,(+) Abnormal social behavior,(+) Diminished ability to concentrate
USP46	ENST00000441222:c.193del p.Ala65HisfsTer13		unknown	6.6	het	unknown	1	NDD	(+) Epicanthus,(+) Hypotonia,(+) Motor delay,(+) Delayed gross motor development,(+) Depressed nasal bridge
SLC5A3	ENST00000381151:c.168_171del p.Phe56LeufsTer75		ad_inherited	7.7	het	unknown	1	NDD	(+) Renal insufficiency,(+) Obesity,(+) Intellectual disability, moderate