Data Dictionary Codebook

EURO-NMD Registry (PID: 747)

17.06.2024 09:59

Instruments	Events

#	Variable / Field Name	Field Label *Field Note*	Field Attributes (Field Type, Validation, Choices, Calculations, etc.)
Instrument:* Consent *(consent)			
1	[record_id]	Patient ID	text Field Annotation: @HIDDEN
2	[consent_note1]	The entry of data is only possible with the written informed consent of the patient.	descriptive
3	[consent_note2]	Patient ID [record-name] Please document the Patient ID within your local patient file, otherwise you may not be able to identify the patient at a later point. No personally identifying information about the patient is stored in this registry.	descriptive
4	[consent_h1]	CONSENT	descriptive
5	[date_of_consent]	Date of consent yyyy-mm-dd	text (date_ymd), Required
6	[has_consent_commercial]	"I CONSENT that my pseudonymized data [my child/the patient pseudonymized data] may also be used to support commercial projects aimed to improve healthcare."	radio, Required yes, Yes no, No unknown, Unknown
7	[has_consent_non_eu]	"I CONSENT that my pseudonymized data [my child/the patient pseudonymized data] may be transferred to non-EU countries, in compliance with GDPR, to support projects aimed to improve healthcare."	radio, Required yes, Yes no, No unknown, Unknown
8	[has_consent_linkage]	"I CONSENT that my pseudonymized data [my child/the patient pseudonymized data] may be linked to existing databases/registries to improve healthcare."	radio, Required yes, Yes no, No unknown, Unknown
9	[has_consent_be_contacted]	Agreement to be contacted for research purposes "I WOULD LIKE TO BE CONTACTED by my medical doctor [my child/the patient medical doctor] about any research project and/or clinical study related to my condition [my child/the patient condition]." *Patient's permission exists for being contacted for research purposes (Element 7.1. in the Set of common data elements for Rare Diseases Registration (link))*	radio, Required yes, Yes no, No unknown, Unknown
10	[has_consent_email] Show the field ONLY if: [has_consent_be_contacted] = 'yes'	"I CONSENT to provide my email address and be contacted to complete questionnaires about my health [the health of my child/the patient] and receive news and updates about the registry."	radio, Required yes, Yes no, No unknown, Unknown
11	[consent_complete]	Section Header: *Form Status* Complete?	dropdown 0, Incomplete 1, Unverified 2, Complete
Instrument:* Baseline *(baseline)			
12	[disease_group]	disease_group	dropdown enrolment_and_firs_arm_1, Neuropathies enrolment_and_firs_arm_2, Myopathies enrolment_and_firs_arm_3, Mitochondrial Diseases enrolment_and_firs_arm_4, Neuromuscular Juction Disorders enrolment_and_firs_arm_5, Motoneuron Diseases enrolment_and_firs_arm_6, Undiagnosed Cases

			Field Annotation: @READONLY @DEFAULT='[first-event-name]'
13	[baseline_h1]	BASELINE	descriptive
14	[internat_pat_id]	International patient identifier *Element 1.1. in the Set of common data elements for Rare Diseases Registration (link); more information available here*	text, Required Field Annotation: @HIDDEN @HIDDEN-PDF
15	[sex_at_birth]	Sex at birth *Patient's sex at birth (Element 2.2. in the Set of common data elements for Rare Diseases Registration (link))*	radio, Required female, Female male, Male undetermined, Undetermined foetus_unknown, Foetus (Unknown)
16	[date_of_birth]	Date of birth yyyy-mm-dd *Patient's date of birth (Element 2.1. in the Set of common data elements for Rare Diseases Registration (link))*	text (date_ymd), Required, Identifier
17	[date_of_visit_bl]	Date of baseline yyyy-mm-dd	text (date_ymd), Required
18	[age_at_baseline]	Age at baseline (years) *Age of patient (years)*	calc Calculation: rounddown(datediff([date_of_birth], [date_of_visit_bl], "y"))
19	[age_at_baseline_months]	Age at baseline (months) *Age of patient (months)*	calc Calculation: rounddown(datediff([date_of_birth], [date_of_visit_bl], "M"))
20	[date_of_first_contact_known]	Section Header: *DISEASE HISTORY* Date of first contact with specialised centre known *Element 4.1. in the Set of common data elements for Rare Diseases Registration (link)*	radio, Required yes, Yes no, No Field Annotation: @DEFAULT = 'yes'
21	[date_of_first_contact] Show the field ONLY if: [date_of_first_contact_known] = 'yes'	Date of first contact with specialised centre yyyy-mm-dd *Date of first contact with specialised centre (Element 4.1. in the Set of common data elements for Rare Diseases Registration (link)). If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.*	text (date_ymd), Required
22	[age_at_onset]	Age at onset of the neuromuscular disease *Age at which symptoms/signs first appeared (Element 5.1. in the Set of common data elements for Rare Diseases Registration (link)) Definitions: - Antenatal onset (accessed: 25.04.2022) - Congenital onset (accessed: 25.04.2022) - Neonatal onset (accessed: 25.04.2022) - Pediatric onset (accessed: 25.04.2022) - Adult onset (accessed: 25.04.2022)*	radio, Required date, Date antenatal, Antenatal onset congenital, Congenital onset neonatal, Neonatal onset pediatric, Pediatric onset adult, Adult onset undetermined, Undetermined Field Annotation: @DEFAULT = 'date'
23	[date_for_age_at_onset] Show the field ONLY if: [age_at_onset]='date'	Date of onset of the neuromuscular disease yyyy-mm-dd *If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year. (Element 5.1. in the Set of common data elements for Rare Diseases Registration (link))*	text (date_ymd), Required
24	[consanguinity]	Consanguinity	radio yes, Yes no, No unknown, Unknown
25	[affected_family]	Affected family members	radio yes, Yes no, No unknown, Unknown
26	[which_affected_family] Show the field ONLY if: [affected_family] = 'yes'	Affected family members (Click all that apply)	checkbox, Required mother, which_affected_family___mother, Mother father, which_affected_family___father, Father uncle, which_affected_family___uncle, Uncle aunt, which_affected_family___aunt, Aunt

			grandfather | which_affected_family___grandfather | Grandfather grandmother | which_affected_family___grandmother | Grandmother sister | which_affected_family___sister | Sister brother | which_affected_family___brother | Brother son | which_affected_family___son | Son daughter | which_affected_family___daughter | Daughter other | which_affected_family___other | Other
27	[had_reduced_foetal_mov] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6) AND [age_at_baseline] < 16	Reduced foetal movements	radio yes | Yes no | No unknown | Unknown
28	[had_polyhydramnios] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4)	Polyhydramnios	radio yes | Yes no | No unknown | Unknown
29	[s4_had_symp_fluc] Show the field ONLY if: ([arm-number]=4 OR [arm-number]=6)	Symptom fluctuation	radio yes | Yes no | No unknown | Unknown
30	[s4_had_exacerb_infection] Show the field ONLY if: [arm-number]=4	Exacerbation with infection	radio yes | Yes no | No unknown | Unknown
31	[s4_had_change_sev_puberty] Show the field ONLY if: [arm-number]=4 AND [age_at_baseline] >= 12	Change of severity in puberty	radio improvement | Improvement deterioration | Deterioration no_change | No change unknown | Unknown not_applicable | Not applicable
32	[s4_had_change_sev_menstr] Show the field ONLY if: [arm-number]=4 AND ([sex_at_birth] = 'female' AND [age_at_baseline] >= 12)	Change of severity with menstrual cycle	radio improvement | Improvement deterioration | Deterioration no_change | No change unknown | Unknown not_applicable | Not applicable
33	[s4_had_change_sev_horm] Show the field ONLY if: [arm-number]=4 AND ([sex_at_birth] = 'female' AND [age_at_baseline] >= 12)	Change of severity with hormonal therapy	radio improvement | Improvement deterioration | Deterioration no_change | No change unknown | Unknown not_applicable | Not applicable
34	[s4_had_change_sev_preg] Show the field ONLY if: [arm-number]=4 AND ([sex_at_birth] = 'female' AND [age_at_baseline] >= 12)	Change of severity with pregnancy or childbirth	radio improvement | Improvement deterioration | Deterioration no_change | No change unknown | Unknown not_applicable | Not applicable

35	[diagnosis_rd] Show the field ONLY if: [arm-number]=1 OR [arm-number]=2 OR [arm-number]=3 OR [arm-number]=4 OR [arm-number]=5	Section Header: *DIAGNOSIS* Diagnosis of the neuromuscular disease *Please answer "Neuromuscular disease diagnosed" if this patient has a known clinical diagnosis, even if the genetic cause has not been established. This is then the diagnosis retained by the specialised centre (Element 6.1. in the Set of common data elements for Rare Diseases Registration (link))*	radio, Required diagnosed: Neuromuscular disease diagnosed unknown: Unknown Field Annotation: @DEFAULT = 'diagnosed'
36	[diagnosis_rd_orpha] Show the field ONLY if: [diagnosis_rd] = 'diagnosed'	Diagnosis of the neuromuscular disease (ORPHAcode) *Diagnosis retained by the specialised centre (Element 6.1. in the Set of common data elements for Rare Diseases Registration (link))*	text, Required BIOPORTAL:ORDO: BIOPORTAL:ORDO
37	[diagnosis_rd_orpha_value] Show the field ONLY if: [diagnosis_rd] = 'diagnosed'	diagnosis_rd_orpha_value	text Field Annotation: @HIDDEN @HIDDEN-PDF @READONLY @CALCTEXT(if(!isblankormissingcode([diagnosis_rd_orpha]), mid([diagnosis_rd_orpha], 7, length([diagnosis_rd_orpha])), ''))
38	[diagnosis_rd_orpha_uri] Show the field ONLY if: [diagnosis_rd] = 'diagnosed'	diagnosis_rd_orpha_uri	text Field Annotation: @HIDDEN @HIDDEN-PDF @READONLY @CALCTEXT(if(!isblankormissingcode([diagnosis_rd_orpha]), concat('http://www.orpha.net/ORDO/Orphanet_', [diagnosis_rd_orpha_value]), ''))
39	[diagnosis_rd_orpha_disp] Show the field ONLY if: [diagnosis_rd] = 'diagnosed'	Diagnosis of the neuromuscular disease (ORDO-URI): [diagnosis_rd_orpha_uri]	descriptive
40	[pabpn1_v_report_def]	pabpn1_v_report_def	text Field Annotation: @READONLY @CALCTEXT(if([diagnosis_rd_orpha:value]='ORPHA:270', 'distinct_number', '')) @HIDDEN @HIDDEN-PDF
41	[dmpk_v_report_def]	dmpk_v_report_def	text Field Annotation: @READONLY @CALCTEXT(if([diagnosis_rd_orpha:value]='ORPHA:273', 'distinct_number', '')) @HIDDEN @HIDDEN-PDF
42	[cnbp_v_report_def]	cnbp_v_report_def	text Field Annotation: @READONLY @CALCTEXT(if([diagnosis_rd_orpha:value]='ORPHA:606', 'distinct_number', '')) @HIDDEN @HIDDEN-PDF
43	[ar_v1_report_def]	ar_v1_report_def	text Field Annotation: @READONLY @CALCTEXT(if([diagnosis_rd_orpha:value]='ORPHA:481', 'distinct_number', '')) @HIDDEN @HIDDEN-PDF
44	[age_at_diagnosis] Show the field ONLY if: [diagnosis_rd] = 'diagnosed'	Age at diagnosis of the neuromuscular disease *Age at which diagnosis was made (Element 5.2. in the Set of common data elements for Rare Diseases Registration (link))*	radio, Required antenatal: Antenatal (Prenatal) postnatal: Postnatal date: Date undetermined: Undetermined Field Annotation: @DEFAULT = 'date'
45	[date_for_age_at_diagnosis] Show the field ONLY if: [age_at_diagnosis]='date'	Date of diagnosis of the neuromuscular disease yyyy-mm-dd *If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year. (Element 5.2. in the Set of common data elements for Rare Diseases Registration (link))*	text (date_ymd), Required
46	[diagnosis_rd_method_known] Show the field ONLY if: [diagnosis_rd] = 'diagnosed'	Diagnostic method known	radio yes: Yes no: No
47	[diagnosis_rd_method] Show the field ONLY if: [diagnosis_rd] = 'diagnosed' AND [diagnosis_rd_method_known] = 'yes'	How was the diagnosis established? (select all that apply)	checkbox clinical_history: diagnosis_rd_method___clinical_history: Clinical History family_history: diagnosis_rd_method___family_history: Positive Family History clinical_exam: diagnosis_rd_method___clinical_exam: Clinical examination genetic_screening: diagnosis_rd_method___genetic_screening: Genetic screening other: diagnosis_rd_method___other: Other methodologies
48	[diagnosis_rd_2_exists] Show the field ONLY if: [diagnosis_rd] = 'diagnosed'	Section Header: *Second diagnosis* Does the patient have a second neuromuscular diagnosis?	radio yes: Yes no: No unknown: Unknown

49	[diagnosis_rd_2_orpha] Show the field ONLY if: [diagnosis_rd_2_exists] = 'yes'	Diagnosis of the second neuromuscular disease (ORPHAcode)	text BIOPORTAL:ORDO | BIOPORTAL:ORDO
50	[s3_main_mito_phenot_known] Show the field ONLY if: [arm-number] = 3	Section Header: *Disease specific* Main mitochondrial phenotype known	radio yes | Yes no | No Field Annotation: @DEFAULT = if([arm-number] = 3, 'yes', '')
51	[s3_main_mito_phenot] Show the field ONLY if: [arm-number]=3 AND [s3_main_mito_phenot_known]='yes'	Main mitochondrial phenotype	text, Required BIOPORTAL:HP | BIOPORTAL:HP
52	[s4_tensilon_test_resp] Show the field ONLY if: [arm-number]=4	Tensilon/prostigmin test response	radio improvement | Improvement deterioration | Deterioration no_change | No change not_done | Not done
53	[s4_chest_ct_findings] Show the field ONLY if: [arm-number]=4	Chest Computed Tomography (CT) findings	radio thymoma | Thymoma residual_thymus_tissue | Residual thymus tissue absent_thymus | Absent thymus not_done | Not done
54	[s5_sma_presymptomatic] Show the field ONLY if: [diagnosis_rd_orpha]='ORPHA:70'	Spinal muscular atrophy: Presymptomatic?	radio yes | Yes no | No unknown | Unknown
55	[s5_cgi_s_score] Show the field ONLY if: [arm-number]=5	Clinical Global Impression of Severity (CGI-S)	radio normal | Normal, not at all ill borderline_ill | Borderline ill mildly_ill | Mildly ill moderately_ill | Moderately ill markedly_ill | Markedly ill severly_ill | Severely ill most_extreme | Among the most extremely ill patients unknown | Unknown
56	[s5_esco_present_form] Show the field ONLY if: [diagnosis_rd_orpha]='ORPHA:803' OR [diagnosis_rd_orpha]='ORPHA:357043' OR [diagnosis_rd_orpha]='ORPHA:275872' OR [diagnosis_rd_orpha]='ORPHA:300605'	El Escorial presentation form	radio bulbar | Bulbar limb | Limb combined | Combined unknown | Unknown
57	[s5_onset_weight_loss] Show the field ONLY if: [arm-number]=5	>10% weight loss in 6 months prior to time of disease onset	radio yes | Yes no | No presymptomatic | Presymptomatic (no disease onset so far at the time of this Baseline-form) unknown | Unknown
58	[s5_onset_cognitive] Show the field ONLY if: [arm-number]=5	Cognitive symptoms or signs in 6 months prior to time of disease onset	radio yes | Yes no | No presymptomatic | Presymptomatic (no disease onset so far at the time of this Baseline-form)

			unknown: Unknown
59	[s5_onset_behavioural] Show the field ONLY if: [arm-number]=5	Behavioural symptoms or signs in 6 months prior to time of disease onset	radio yes: Yes no: No presymptomatic: Presymptomatic (no disease onset so far at the time of this Baseline-form) unknown: Unknown
60	[bio_sample_available]	Section Header: *BIOBANKING* Biological sample available for research *Patient's biological sample available for research (Element 7.3. in the Set of common data elements for Rare Diseases Registration (link))*	radio, Required yes: Yes no: No unknown: Unknown
61	[bio_sample_biobank] Show the field ONLY if: [bio_sample_available]='yes'	Biological sample stored in a biobank *Biological sample stored in a biobank (Element 7.4. in the Set of common data elements for Rare Diseases Registration (link))*	radio, Required yes: Yes no: No unknown: Unknown
62	[link_to_biobank] Show the field ONLY if: [bio_sample_biobank] = 'yes'	Link to a biobank *Link to a biobank (Element 7.4. in the Set of common data elements for Rare Diseases Registration (link))*	notes, Required
63	[has_bulbar_weakness] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Section Header: *MOBILITY AND GENERAL PHYSICAL FUNCTIONING* Bulbar weakness	radio yes: Yes no: No unknown: Unknown
64	[has_tongue_wasting] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Tongue wasting	radio yes: Yes no: No unknown: Unknown
65	[has_muscle_wasting] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Muscle wasting	radio yes: Yes no: No unknown: Unknown
66	[has_ptosis] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Section Header: *Muscular Functioning: Eye* Ptosis	radio yes: Yes no: No unknown: Unknown
67	[has_ophthalmoparesis] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Ophthalmoparesis	radio yes: Yes no: No unknown: Unknown
68	[has_diplopia] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Diplopia	radio yes: Yes no: No unknown: Unknown
69	[s4_has_episodic_apnoea] Show the field ONLY if: ([arm-number]=4 OR [arm-number]=6)	Section Header: *Respiratory / Ventilation* Episodic apnoea	radio yes: Yes no: No unknown: Unknown
70	[s4_has_stridor] Show the field ONLY if:	Stridor	radio yes: Yes

	([arm-number]=4 OR [arm-number]=6)		no, No unknown, Unknown
71	[s4_has_rec_resp_infection] Show the field ONLY if: ([arm-number]=4 OR [arm-number]=6)	Recurrent respiratory tract infection	radio yes, Yes no, No unknown, Unknown
72	[has_resp_weakness] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Respiratory weakness	radio yes, Yes no, No unknown, Unknown
73	[s5_has_resp_diff] Show the field ONLY if: [arm-number]=5	Respiratory difficulties	radio yes, Yes no, No unknown, Unknown
74	[has_high_arch_palate] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Section Header: *Skeletal* High arched palate	radio yes, Yes no, No unknown, Unknown
75	[has_rigid_spine] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Rigid spine	radio yes, Yes no, No unknown, Unknown
76	[has_scapular_winging] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Scapular winging	radio yes, Yes no, No unknown, Unknown
77	[has_joint_contr] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=5 OR [arm-number]=6)	Joint contractures	radio yes, Yes no, No unknown, Unknown
78	[which_joint_contr] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=5 OR [arm-number]=6) AND [has_joint_contr] = 'yes'	Joint contractures (select all that apply)	checkbox elbow, which_joint_contr___elbow, Elbow wrist, which_joint_contr___wrist, Wrist fingers, which_joint_contr___fingers, Fingers hip, which_joint_contr___hip, Hip knee, which_joint_contr___knee, Knee ankle, which_joint_contr___ankle, Ankle
79	[has_arthrogryposis] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6) AND [has_joint_contr] = 'yes'	Arthrogryposis	radio yes, Yes no, No unknown, Unknown
80	[had_delay_recov_anesth] Show the field ONLY if: ([arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Section Header: *TREATMENT* Delayed recovery from anesthetics	radio yes, Yes no, No unknown, Unknown
81	[s4_pyridost_treat_resp] Show the field ONLY if:	Pyridostigmine treatment response	radio improvement, Improvement

	[arm-number]=4		deterioration, Deterioration no_change, No change not_done, Not done unknown, Unknown
82	[baseline_desc2]	Section Header: *NOTE* To complete the baseline data collection, please remember to fill the following forms: - "Consent"- "Visit Data"- "Medication"- "Genetic Diagnosis" (or "Undiagnosed")	descriptive
83	[kpi_onset_contact_hcp_d]	Time from onset (first symptoms) to first HCP visit [days]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_for_age_at_onset]) AND !isblankormissingcode([date_of_first_contact]) AND [date_for_age_at_onset] <= [date_of_first_contact], datediff([date_for_age_at_onset], [date_of_first_contact], "d","ymd", true), ''))
84	[kpi_onset_contact_hcp_y]	Time from onset (first symptoms) to first HCP visit [years]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_for_age_at_onset]) AND !isblankormissingcode([date_of_first_contact]) AND [date_for_age_at_onset] <= [date_of_first_contact], datediff([date_for_age_at_onset], [date_of_first_contact], "y","ymd", true), ''))
85	[kpi_contact_hcp_onset_d]	Time from first HCP visit to onset (first symptoms) [days]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_of_first_contact]) AND !isblankormissingcode([date_for_age_at_onset]) AND [date_of_first_contact] < [date_for_age_at_onset], datediff([date_of_first_contact], [date_for_age_at_onset], "d","ymd", true), ''))
86	[kpi_contact_hcp_onset_y]	Time from first HCP visit to onset (first symptoms) [years]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_of_first_contact]) AND !isblankormissingcode([date_for_age_at_onset]) AND [date_of_first_contact] < [date_for_age_at_onset], datediff([date_of_first_contact], [date_for_age_at_onset], "y","ymd", true), ''))
87	[kpi_onset_diaclin_d]	Time from onset (first symptoms) to specific diagnosis [days]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_for_age_at_onset]) AND !isblankormissingcode([date_for_age_at_diagnosis]) AND [date_for_age_at_onset] <= [date_for_age_at_diagnosis], datediff([date_for_age_at_onset], [date_for_age_at_diagnosis], "d","ymd", true), ''))
88	[kpi_onset_diaclin_y]	Time from onset (first symptoms) to specific diagnosis [years]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_for_age_at_onset]) AND !isblankormissingcode([date_for_age_at_diagnosis]) AND [date_for_age_at_onset] <= [date_for_age_at_diagnosis], datediff([date_for_age_at_onset], [date_for_age_at_diagnosis], "y","ymd", true), ''))
89	[kpi_diaclin_onset_d]	Time from specific diagnosis to onset (first symptoms) [days]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_for_age_at_diagnosis]) AND !isblankormissingcode([date_for_age_at_onset]) AND [date_for_age_at_diagnosis] < [date_for_age_at_onset], datediff([date_for_age_at_diagnosis], [date_for_age_at_onset], "d","ymd", true), ''))
90	[kpi_diaclin_onset_y]	Time from specific diagnosis to onset (first symptoms) [years]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_for_age_at_diagnosis]) AND !isblankormissingcode([date_for_age_at_onset]) AND [date_for_age_at_diagnosis] < [date_for_age_at_onset], datediff([date_for_age_at_diagnosis], [date_for_age_at_onset], "y","ymd", true), ''))
91	[kpi_contact_hcp_diaclin_d]	Time from first HCP visit to specific diagnosis [days]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_of_first_contact]) AND !isblankormissingcode([date_for_age_at_diagnosis]) AND [date_of_first_contact] <= [date_for_age_at_diagnosis], datediff([date_of_first_contact], [date_for_age_at_diagnosis], "d","ymd", true), ''))
92	[kpi_contact_hcp_diaclin_y]	Time from first HCP visit to specific diagnosis [years]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_of_first_contact]) AND !isblankormissingcode([date_for_age_at_diagnosis]) AND [date_of_first_contact] <= [date_for_age_at_diagnosis], datediff([date_of_first_contact], [date_for_age_at_diagnosis], "y","ymd", true), ''))
93	[kpi_diaclin_contact_hcp_d]	Time from specific diagnosis to first HCP visit [days]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_for_age_at_diagnosis]) AND

			!isblankormissingcode([date_of_first_contact]) AND [date_for_age_at_diagnosis] < [date_of_first_contact], datediff([date_for_age_at_diagnosis], [date_of_first_contact], "d","ymd", true), ''))
94	[kpi_diaclin_contact_hcp_y]	Time from specific diagnosis to first HCP visit [years]	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([date_for_age_at_diagnosis]) AND !isblankormissingcode([date_of_first_contact]) AND [date_for_age_at_diagnosis] < [date_of_first_contact], datediff([date_for_age_at_diagnosis], [date_of_first_contact], "y","ymd", true), ''))
95	[baseline_complete]	Section Header: *Form Status* Complete?	dropdown 0, Incomplete 1, Unverified 2, Complete
Instrument:* Visit Data *(visit_data)			
96	[visit_data_h1]	VISIT DATA	descriptive
97	[date_of_visit_prev]	date_of_visit_prev: Value of date_of_visit in previous report. Is needed for piping for other fields (e.g. s5_hands_up_gain_d_known)	text Field Annotation: @CALCTEXT(if(starts_with([event-name], 'enrolment'), '', if(starts_with([event-name], 'visit') AND [current-instance]=1, [first-event-name][date_of_visit], if(starts_with([event-name], 'visit') AND [current-instance]>1, [event-name][date_of_visit][previous-instance], '')))) @READONLY @HIDDEN @HIDDEN-PDF
98	[fu_instance_id]	fu_instance_id	text Field Annotation: @CALCTEXT([current-instance]) @READONLY @HIDDEN @HIDDEN-PDF
99	[sex_at_birth_v]	sex_at_birth_v (needed for branching logic within this form)	text Field Annotation: @CALCTEXT(if(starts_with([event-name], 'visit'), [first-event-name][sex_at_birth], [event-name][sex_at_birth])) @READONLY @HIDDEN @HIDDEN-PDF
100	[diagnosis_rd_orpha_v]	diagnosis_rd_orpha_v (needed for branching logic within this form)	text Field Annotation: @CALCTEXT(if(starts_with([event-name], 'visit'), [first-event-name][diagnosis_rd_orpha], [event-name][diagnosis_rd_orpha])) @READONLY @HIDDEN @HIDDEN-PDF
101	[date_of_visit]	Section Header: *General* Date of visit yyyy-mm-dd	text (date_ymd), Required
102	[age_at_visit]	Age of patient (years)	calc Calculation: if(starts_with([event-name], 'visit'), rounddown(datediff([first-event-name][date_of_birth], [date_of_visit], "y")), rounddown(datediff([event-name][date_of_birth], [date_of_visit], "y")))
103	[age_at_visit_months]	Age of patient (months)	calc Calculation: if(starts_with([event-name], 'visit'), rounddown(datediff([first-event-name][date_of_birth], [date_of_visit], "M")), rounddown(datediff([event-name][date_of_birth], [date_of_visit], "M")))
104	[body_height_cm_known]	Body Height (cm) known	radio yes, Yes no, No Field Annotation: @DEFAULT = 'yes'
105	[body_height_cm] Show the field ONLY if: [body_height_cm_known] = 'yes'	Body Height (cm)	text (number), Required
106	[body_weight_kg_known]	Body Weight (kg) known	radio yes, Yes no, No
107	[body_weight_kg] Show the field ONLY if: [body_weight_kg_known] = 'yes'	Body Weight (kg)	text (number), Required
108	[affil_registries_desc1]	Section Header: *Research* Participation in affiliated registries* *registries affiliated to the EURO-NMD Registry Hub for Rare Neuromuscular Diseases	descriptive
109	[affil_registries_desc2] Show the field ONLY if: [pat_in_dmscope] = 'yes' OR [pat_in_cramp] = 'yes' OR [pat_in_ddp] = 'yes' OR [pat_in_smartcare] = 'yes' OR [pat_in_ot	NOTE: The ID of the patient in an other registry may only be entered here if the patient has consented to this procedure in writing.	descriptive

	her_reg] = 'yes'		
110	[pat_in_dmscope] Show the field ONLY if: [arm-number]=2	Patient registered in DM-Scope (French Observatory of Myotonic Dystrophies)	radio yes, Yes no, No unknown, Unknown Field Annotation: @DEFAULT='[event-name][pat_in_dmscope:value][previous-instance]'
111	[pat_id_dmscope] Show the field ONLY if: [arm-number]=2 AND [pat_in_dmscope]='yes'	Patient ID in DM-Scope *http://www.dmscope.fr/french-registry-of-myotonic-dystrophies/*	text Field Annotation: @DEFAULT='[event-name][pat_id_dmscope:value][previous-instance]'
112	[pat_in_cramp] Show the field ONLY if: ([arm-number]=1 OR [arm-number]=2)	Patient registered in CRAMP (Computer Registry of All Myopathies and Polyneuropathies)	radio yes, Yes no, No unknown, Unknown Field Annotation: @DEFAULT='[event-name][pat_in_cramp:value][previous-instance]'
113	[pat_id_cramp] Show the field ONLY if: ([arm-number]=1 OR [arm-number]=2) AND [pat_in_cramp] = 'yes'	Patient ID in CRAMP *CRAMP on Orphanet*	text Field Annotation: @DEFAULT='[event-name][pat_id_cramp:value][previous-instance]'
114	[pat_in_ddp] Show the field ONLY if: [arm-number]=2	Patient registered in DDP (Duchenne Data Platform)	radio yes, Yes no, No unknown, Unknown Field Annotation: @DEFAULT='[event-name][pat_in_ddp:value][previous-instance]'
115	[pat_id_ddp] Show the field ONLY if: [arm-number]=2 AND [pat_in_ddp] = 'yes'	Patient ID in DDP *https://duchenne.nl/duchenne-data-platform/*	text Field Annotation: @DEFAULT='[event-name][pat_id_ddp:value][previous-instance]'
116	[pat_in_smartcare] Show the field ONLY if: [arm-number]=5	Patient registered in SMArtCARE (Spinal Muscular Atrophy)	radio yes, Yes no, No unknown, Unknown Field Annotation: @DEFAULT='[event-name][pat_in_smartcare:value][previous-instance]'
117	[pat_id_smartcare] Show the field ONLY if: [arm-number]=5 AND [pat_in_smartcare] = 'yes'	Patient ID in SMArtCARE *https://www.smartcare.de/en/*	text Field Annotation: @DEFAULT='[event-name][pat_id_smartcare:value][previous-instance]'
118	[pat_in_other_reg]	Patient registered in a registry that is not listed above	radio yes, Yes no, No unknown, Unknown Field Annotation: @DEFAULT='[event-name][pat_in_other_reg:value][previous-instance]'
119	[link_to_patient_registry] Show the field ONLY if: [pat_in_other_reg] = 'yes'	Link to other patient registry (please provide a comma-separated list of URIs if this patient is registered in more than one other registry)	notes Field Annotation: @DEFAULT='[event-name][link_to_patient_registry:value][previous-instance]'
120	[pat_in_clinical_trial]	Patient in a clinical trial?	radio yes, Yes no, No unknown, Unknown Field Annotation: @DEFAULT='[event-name][pat_in_clinical_trial:value][previous-instance]'

121	[link_to_clinical_trial] Show the field ONLY if: [pat_in_clinical_trial] = 'yes'	Link to clinical trial (please provide a comma-separated list of URIs if this patient participates in more than one clinical trial)	notes Field Annotation: @DEFAULT='[event-name][link_to_clinical_trial:value][previous-instance]'
122	[number_of_preg_known] Show the field ONLY if: [sex_at_birth_v] = 'female' AND [age_at_visit] >= 16	Section Header: *Gestation* Number of pregnancies known	radio yes, Yes no, No
123	[number_of_preg] Show the field ONLY if: [sex_at_birth_v] = 'female' AND [number_of_preg_known] = 'yes'	Number of pregnancies	text (integer), Required
124	[number_of_child_known] Show the field ONLY if: [sex_at_birth_v] = 'female' AND [age_at_visit] >= 16	Number of children known	radio yes, Yes no, No
125	[number_of_child] Show the field ONLY if: [sex_at_birth_v] = 'female' AND [number_of_child_known] = 'yes'	Number of children	text (integer), Required
126	[had_genetic_couns] Show the field ONLY if: [arm-number]=1 OR [arm-number]=2 OR [arm-number]=3 OR [arm-number]=4 OR [arm-number]=5	Section Header: *Care pathway* Patient had genetic counselling *Has a genetic counselling taken place since the last registered contact?*	radio yes, Yes no, No unknown, Unknown
127	[genetic_couns_purp] Show the field ONLY if: [had_genetic_couns]= 'yes'	Genetic counselling (purpose)	checkbox post_test_report_re, genetic_couns_purp___post_test_report_re, Post-test report return counselling pre_natal, genetic_couns_purp___pre_natal, Pre-natal testing post_natal, genetic_couns_purp___post_natal, Post-natal testing pre_symp, genetic_couns_purp___pre_symp, Pre-symptomatic testing carrier, genetic_couns_purp___carrier, Carrier testing pre_conception, genetic_couns_purp___pre_conception, Pre-conception testing other, genetic_couns_purp___other, Other
128	[s2_had_malig_screening] Show the field ONLY if: [arm-number]=2	Patient had malignancy screening	radio yes, Yes no, No unknown, Unknown
129	[has_access_to_phys_therapy]	Does the patient have access to physical therapy?	radio yes, Yes no, No unknown, Unknown
130	[has_access_to_occ_therapy]	Does the patient have access to occupational therapy?	radio yes, Yes no, No unknown, Unknown
131	[s2_has_speech_therapy] Show the field ONLY if: [arm-number]=2	Does the patient have access to speech therapy?	radio yes, Yes no, No unknown, Unknown

132	[s2_has_dietitian_advice] Show the field ONLY if: [arm-number]=2	Does the patient have access to dietitian advice?	radio yes, Yes no, No unknown, Unknown
133	[delayed_motor_milestones] Show the field ONLY if: [age_at_visit] < 16	Section Header: *Developmental* Delayed motor milestones	radio yes, Yes no, No unknown, Unknown
134	[cgii] Show the field ONLY if: starts_with([event-name], 'visit')	Section Header: *Clinical Global Impression - Improvement scale (CGI-I)* Compared to the patient's condition at baseline, this patient's [average] condition has/is...?	radio 0, Not assessed 1, Very Much Improved 2, Much Improved 3, Minimally Improved 4, No Change 5, Minimally Worse 6, Much Worse 7, Very Much Worse
135	[s3_was_disc_in_boards] Show the field ONLY if: ([arm-number]=3 OR [arm-number]=6)	Section Header: *Events in the past 12 months* Patient discussed in MDT/CPMS/Board within the last 12 months? *Was the patient discussed in Multidisciplinary Team (MDT) / Clinical Patient Management System (CPMS) / Board within the last 12 months?*	radio yes, Yes no, No unknown, Unknown
136	[unpl_hosp_12m]	Have there been any unplanned hospitalizations in the last 12 months?	radio yes, Yes no, No unknown, Unknown
137	[unpl_hosp_12m_n] Show the field ONLY if: [unpl_hosp_12m] = 'yes'	Number of unplanned hospitalizations in the last 12 months	text (integer), Required
138	[unpl_hosp_12m_ndays_known] Show the field ONLY if: [unpl_hosp_12m] = 'yes'	Unplanned hospitalization in the last 12 months: Duration in days known	radio, Required yes, Yes no, No
139	[unpl_hosp_12m_ndays] Show the field ONLY if: [unpl_hosp_12m_ndays_known] = 'yes'	Unplanned hospitalization in the last 12 months: Duration in days	text (number), Required
140	[had_comp_remi] Show the field ONLY if: ([arm-number]=1 OR [arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Section Header: *Events since last visit* Complete remission *Has the patient had a complete remission since the last recorded contact in this registry?*	radio yes, Yes no, No unknown, Unknown not_applicable, Not applicable
141	[date_of_comp_remi] Show the field ONLY if: ([arm-number]=1 OR [arm-number]=2 OR [arm-number]=4 OR [arm-number]=6) AND [had_comp_remi]='yes'	Date of complete remission yyyy-mm-dd *If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.*	text (date_ymd)
142	[had_part_remi] Show the field ONLY if: ([arm-number]=1 OR [arm-number]=2 OR [arm-number]=4 OR [arm-number]=6)	Partial remission *Has the patient had a partial remission since the last recorded contact in this registry?*	radio yes, Yes no, No unknown, Unknown not_applicable, Not applicable

143	[date_of_part_remi] Show the field ONLY if: ([arm-number]=1 OR [arm-number]=2 OR [arm-number]=4 OR [arm-number]=6) AND [had_part_remi]='yes'	Date of partial remission yyyy-mm-dd *If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.*	text (date_ymd)
144	[s5_esco_diag_grade] Show the field ONLY if: [diagnosis_rd_orpha_v]='ORPHA:803' OR [diagnosis_rd_orpha_v]='ORPHA:357043' OR [diagnosis_rd_orpha_v]='ORPHA:275872' OR [diagnosis_rd_orpha_v]='ORPHA:300605'	Section Header: *Diagnosis* El Escorial diagnosis grade	radio suspected_als: Suspected ALS possible_als: Possible ALS probable_als_lab: Probable ALS lab supported probable_als: Probable ALS definitive_als: Definitive ALS unknown: Unknown
145	[s1_antibodies_present] Show the field ONLY if: [arm-number]=1	Section Header: *Inflammatory neuropathy auto-antibodies* Presence of inflammatory neuropathy auto-antibodies	radio yes: Yes no: No not_done: Not tested (not done) unknown: Unknown
146	[s1_aa_paraprot_result] Show the field ONLY if: [s1_antibodies_present] = 'yes'	Paraproteins test-result *e.g. in case of Chronic inflammatory demyelinating polyradiculoneuropathy (CIDP)*	radio positive: Positive negative: Negative not_done: Not done
147	[s1_aa_antimag_result] Show the field ONLY if: [s1_antibodies_present] = 'yes'	Anti-MAG antibodies test-result	radio positive: Positive negative: Negative not_done: Not done
148	[s1_aa_antigm1_result] Show the field ONLY if: [s1_antibodies_present] = 'yes'	Anti-GM1 antibodies test-result *e.g. in case of Multifocal motor neuropathy (MMN)*	radio positive: Positive negative: Negative not_done: Not done
149	[s1_aa_antigm1_igm_result] Show the field ONLY if: [s1_aa_antigm1_result] = 'positive'	IgM antibodies test-result	radio positive: Positive negative: Negative not_done: Not done
150	[s1_aa_antigm1_igm_od] Show the field ONLY if: [s1_aa_antigm1_igm_result] = 'positive'	IgM antibodies ELISA OD value	text (number)
151	[s1_aa_antigm1_igg_result] Show the field ONLY if: [s1_aa_antigm1_result] = 'positive'	IgG antibodies test-result	radio positive: Positive negative: Negative not_done: Not done
152	[s1_aa_antigm1_igg_od] Show the field ONLY if: [s1_aa_antigm1_igg_result] = 'positive'	IgG antibodies ELISA OD value	text (number)
153	[s1_aa_antigang_result] Show the field ONLY if: [s1_antibodies_present] = 'yes'	Other antiganglioside antibodies test-result	radio positive: Positive negative: Negative not_done: Not done

154	[s1_aa_antigang_which] Show the field ONLY if: [s1_aa_antigang_result] = 'positive'	Which other antiganglioside antibodies? Please enter the names of other antiganglioside antibodies tested positive as a comma-separated list.	notes
155	[s1_aa_nodal_result] Show the field ONLY if: [s1_antibodies_present] = 'yes'	Anti-nodal/paranodal protein antibodies test-result *e.g. in case of Chronic inflammatory demyelinating polyradiculoneuropathy (CIDP)*	radio positive, Positive negative, Negative not_done, Not done
156	[s1_aa_nodal_which] Show the field ONLY if: [s1_aa_nodal_result] = 'positive'	Which anti-nodal/paranodal protein antibodies? Please enter the names of anti-nodal/paranodal protein antibodies tested positive as a comma-separated list. *e.g.: anti-contactin, anti-CASPR, anti-neurofascin*	notes
157	[s2_antibodies_present] Show the field ONLY if: [arm-number]=2	Section Header: *Myositis auto-antibodies* Presence of myositis auto-antibodies	radio yes, Yes no, No not_done, Not tested (not done) unknown, Unknown
158	[s2_aa_mi_2alpha] Show the field ONLY if: [s2_antibodies_present] = 'yes'	Mi-2α test-result	radio positive, Positive negative, Negative not_done, Not done
159	[s2_aa_mi_2beta] Show the field ONLY if: [s2_antibodies_present] = 'yes'	Mi-2β test-result	radio positive, Positive negative, Negative not_done, Not done
160	[s2_aa_mup44] Show the field ONLY if: [s2_antibodies_present] = 'yes'	Mup44 test-result	radio positive, Positive negative, Negative not_done, Not done
161	[s2_aa_tif1gamma] Show the field ONLY if: [s2_antibodies_present] = 'yes'	TIF1γ test-result	radio positive, Positive negative, Negative not_done, Not done
162	[s2_aa_mda5] Show the field ONLY if: [s2_antibodies_present] = 'yes'	MDA5 test-result	radio positive, Positive negative, Negative not_done, Not done
163	[s2_aa_nxp2] Show the field ONLY if: [s2_antibodies_present] = 'yes'	NPX2 test-result	radio positive, Positive negative, Negative not_done, Not done
164	[s2_aa_sae1] Show the field ONLY if: [s2_antibodies_present] = 'yes'	SAE1 test-result	radio positive, Positive negative, Negative not_done, Not done
165	[s2_aa_ku] Show the field ONLY if: [s2_antibodies_present] = 'yes'	Ku test-result	radio positive, Positive negative, Negative not_done, Not done

166	[s2_aa_pm_scl100] Show the field ONLY if: [s2_antibodies_present] = 'yes'	PM-Scl100 test-result	radio positive, Positive negative, Negative not_done, Not done
167	[s2_aa_pm_scl75] Show the field ONLY if: [s2_antibodies_present] = 'yes'	PM-Scl75 test-result	radio positive, Positive negative, Negative not_done, Not done
168	[s2_aa_jo_1] Show the field ONLY if: [s2_antibodies_present] = 'yes'	Jo-1 test-result	radio positive, Positive negative, Negative not_done, Not done
169	[s2_aa_srp] Show the field ONLY if: [s2_antibodies_present] = 'yes'	SRP test-result	radio positive, Positive negative, Negative not_done, Not done
170	[s2_aa_pl_7] Show the field ONLY if: [s2_antibodies_present] = 'yes'	PL-7 test-result	radio positive, Positive negative, Negative not_done, Not done
171	[s2_aa_pl_12] Show the field ONLY if: [s2_antibodies_present] = 'yes'	PL-12 test-result	radio positive, Positive negative, Negative not_done, Not done
172	[s2_aa_ej] Show the field ONLY if: [s2_antibodies_present] = 'yes'	EJ test-result	radio positive, Positive negative, Negative not_done, Not done
173	[s2_aa_oj] Show the field ONLY if: [s2_antibodies_present] = 'yes'	OJ test-result	radio positive, Positive negative, Negative not_done, Not done
174	[s2_aa_ro_52] Show the field ONLY if: [s2_antibodies_present] = 'yes'	Ro-52 test-result	radio positive, Positive negative, Negative not_done, Not done
175	[s2_aa_anti_srp] Show the field ONLY if: [s2_antibodies_present] = 'yes'	anti-SRP test-result	radio positive, Positive negative, Negative not_done, Not done
176	[s2_aa_anti_hmgcr] Show the field ONLY if: [s2_antibodies_present] = 'yes'	anti-HMGCR test-result	radio positive, Positive negative, Negative not_done, Not done
177	[s4_antibodies_present] Show the field ONLY if: [arm-number]=4	Section Header: *Myasthenia auto-antibodies* Presence of myasthenia auto-antibodies	radio yes, Yes no, No

			not_done | Not tested (not done) unknown | Unknown
178	[s4_aa_achr_ria_result] Show the field ONLY if: [s4_antibodies_present] = 'yes'	AChR (standard, RIA) test-result	radio positive | Positive negative | Negative not_done | Not done
179	[s4_aa_achr_cba_result] Show the field ONLY if: [s4_antibodies_present] = 'yes'	AChR (low-affinity, CBA) test-result	radio positive | Positive negative | Negative not_done | Not done
180	[s4_aa_musk_ria_result] Show the field ONLY if: [s4_antibodies_present] = 'yes'	Musk (standard,RIA) test-result	radio positive | Positive negative | Negative not_done | Not done
181	[s4_aa_musk_cba_result] Show the field ONLY if: [s4_antibodies_present] = 'yes'	MuSK (low-affinity, CBA) test-result	radio positive | Positive negative | Negative not_done | Not done
182	[s4_aa_titin_result] Show the field ONLY if: [s4_antibodies_present] = 'yes'	Titin test-result	radio positive | Positive negative | Negative not_done | Not done
183	[s4_aa_ryr1_result] Show the field ONLY if: [s4_antibodies_present] = 'yes'	Ryanodine receptor (RYR1) test-result	radio positive | Positive negative | Negative not_done | Not done
184	[s4_aa_vgcc_result] Show the field ONLY if: [s4_antibodies_present] = 'yes'	Voltage-gated calcium channel antibodies (VGCC) test-result	radio positive | Positive negative | Negative not_done | Not done
185	[s2_ck_level] Show the field ONLY if: [arm-number]=2	Section Header: *Creatine Kinase* Creatine kinase level	radio normal | Normal elevated_up_to_10_times | Elevated - less than 10x normal or 10x normal elevated_10_times_plus | Elevated - more than 10x normal not_done | Not done
186	[s2_ck_exact_val_known] Show the field ONLY if: [arm-number]=2 AND ([s2_ck_level]='elevated_up_to_10_times' OR [s2_ck_level]='elevated_10_times_plus')	Exact creatine kinase value known	radio yes | Yes no | No
187	[s2_ck_exact_val] Show the field ONLY if: [arm-number]=2 AND ([s2_ck_level]='elevated_up_to_10_times' OR [s2_ck_level]='elevated_10_times_plus') AND [s2_ck_exact_val_known] = 'yes'	Exact creatine kinase value [U/l])	text (number)
188	[s2_ck_value_ref_report] Show the field ONLY if: [arm-number]=2 AND ([s2_ck_level]='elevated_up_to_10_times' OR [s2_ck_level]='elevated_10_times_plus')	Referenced normal value for creatine kinase level known	radio yes | Yes no | No

189	[s2_ck_value_ref] Show the field ONLY if: [arm-number]=2 AND [s2_ck_value_ref_report] = 'yes'	Upper level of normal (ULN) for reference creatine kinase level (in enzyme units per litre [U/l]):	text (number)
190	[s2_ck_value_report] Show the field ONLY if: [arm-number]=2 AND ([s2_ck_level]='elevated_up_to_10_times' OR [s2_ck_level]='elevated_10_times_plus')	Report value of creatine kinase	radio distinct_number: Report a distinct number range: Report a range unknown: Unknown
191	[s2_ck_value] Show the field ONLY if: [s2_ck_value_report] = 'distinct_number'	Creatine Kinase value (in enzyme units per litre [U/l]):	text (number)
192	[s2_ck_value_r1] Show the field ONLY if: [s2_ck_value_report] = 'range'	Start of range for Creatine Kinase value (in enzyme units per litre [U/l]):	text (number)
193	[s2_ck_value_r2] Show the field ONLY if: [s2_ck_value_report] = 'range'	End of range for Creatine Kinase value (in enzyme units per litre [U/l]):	text (number)
194	[s2_musc_biopsy_result] Show the field ONLY if: [arm-number]=2	Section Header: *Muscular* Muscle biopsy result	radio normal: Normal abnormal: Abnormal not_done: Not done
195	[s2_musc_biopsy_abnorm_type] Show the field ONLY if: [arm-number]=2 AND [s2_musc_biopsy_result]='abnormal'	Muscle biopsy abnormal type	radio neurogenic: Neurogenic myopathic: Myopathic dystrophic: Dystrophic unknown: Unknown
196	[s2_musc_mri] Show the field ONLY if: [arm-number]=2	Muscle MRI	radio normal: Normal abnormal: Abnormal not_done: Not done
197	[emg_result_type]	Section Header: *Neuromuscular* Electromyography (EMG): result type	radio normal: Normal abnormal: Abnormal equivocal: Equivocal not_done: Not done
198	[emg_abnormality_type] Show the field ONLY if: [emg_result_type]='abnormal'	Electromyography (EMG): abnormality type	checkbox myopathic: emg_abnormality_type___myopathic: Myopathic neurogenic: emg_abnormality_type___neurogenic: Neurogenic
199	[s4_sfemg_result_type] Show the field ONLY if: [arm-number]=4	Single Fiber Electromyography (SFEMG): result type	radio normal: Normal abnormal: Abnormal equivocal: Equivocal not_done: Not done
200	[s4_rep_nerve_stimu] Show the field ONLY if: ([arm-number]=4 OR [arm-number]=6)	Repetitive nerve stimulation	radio normal: Normal abnormal: Abnormal not_done: Not done
201	[s4_rep_nerve_stimu_decr] Show the field ONLY if:	Repetitive nerve stimulation: Decrement percentage *expected: number, minimum: 0, maximum: 100*	text (number, Min: 0, Max: 100)

	([arm-number]=4 OR [arm-number]=6) AND [s4_rep_nerve_stimu]='abnormal'		
202	[s4_rep_nerve_stimu_incr] Show the field ONLY if: ([arm-number]=4 OR [arm-number]=6) AND [s4_rep_nerve_stimu]='abnormal'	Repetitive nerve stimulation: Increment percentage *expected: number, minimum: 0*	text (number, Min: 0)
203	[ambulatory_status]	Section Header: *Mobility and General Physical Functioning* Current ambulatory status	radio walk_unsupported, Walk without support walk_supported, Walk with support wheelchair_pt, Wheelchair parttime wheelchair_ft, Wheelchair fulltime unknown, Unknown
204	[ambulatory_status_prev]	ambulatory_status_prev: Value of ambulatory_status in previous report. Is needed for piping and branching for other fields.	text Field Annotation: @CALCTEXT(if(starts_with([event-name], 'enrolment'), '', if(starts_with([event-name], 'visit') AND [current-instance]=1, [first-event-name][ambulatory_status], if(starts_with([event-name], 'visit') AND [current-instance]>1, [event-name][ambulatory_status][previous-instance], '')))) @READONLY @HIDDEN @HIDDEN-PDF
205	[ambulation_change_d_known] Show the field ONLY if: [ambulatory_status_prev] != 'unknown' AND [ambulatory_status] != 'unknown' AND !isblankormissingcode([ambulatory_status_prev]) AND !isblankormissingcode([ambulatory_status]) AND [ambulatory_status_prev] != [ambulatory_status]	When comparing this report to the last report, there was a change in in terms of the patients ambulatory status. Last time (date: [date_of_visit_prev]) you reported: [ambulatory_status_prev] Here (date: [date_of_visit]) you reported: [ambulatory_status:value] Do you know when this ability changed?	radio yes, Yes no, No
206	[date_of_change_of_ambu] Show the field ONLY if: [ambulation_change_d_known] = 'yes'	Date of change of ambulation yyyy-mm-dd *If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.*	text (date_ymd)
207	[s2_max_walking_m] Show the field ONLY if: [arm-number]=2	Maximum walking distance estimate (m)	text (number)
208	[has_limb_weakness]	Limb weakness	radio yes, Yes no, No unknown, Unknown
209	[has_neck_weakness]	Neck muscle weakness	radio yes, Yes no, No unknown, Unknown
210	[has_facial_weakness]	Weakness of facial musculature	radio yes, Yes no, No unknown, Unknown
211	[s5_hands_up_curr] Show the field ONLY if: [arm-number]=5	Can the patient currently raise hands to mouth in a sitting position?	radio yes, Yes no, No unknown, Unknown
212	[s5_hands_up_curr_prev]	s5_hands_up_curr_prev: Value of s5_hands_up_curr in previous report. Is needed for piping and branching for other fields.	text Field Annotation: @CALCTEXT(if(starts_with([event-name], 'enrolment'), '', if(starts_with([event-name], 'visit') AND [current-instance]=1, [first-event-name][s5_hands_up_curr], if(starts_with([event-name], 'visit') AND [current-instance]>1, [event-name][s5_hands_up_curr][previous-instance], '')))) @READONLY @HIDDEN @HIDDEN-PDF
213	[s5_hands_up_d_known] Show the field ONLY if:	When comparing this report to the last report, there was a change in in terms of the patients ability to raise hands to mouth in a sitting position. Last time (date: [date_of_visit_prev]) you reported: Is patient able? [s5_hands_up_curr_prev]	radio yes, Yes

	[arm-number]=5 AND ([s5_hands_up_curr]='yes' AND [s5_hands_up_curr_prev]='no' OR [s5_hands_up_curr]='no' AND [s5_hands_up_curr_prev]='yes')	Here (date: [date_of_visit]) you reported: Is patient able? [s5_hands_up_curr:value] Do you know when this ability changed?	no, No
214	[s5_date_of_hands_up_gain] Show the field ONLY if: [s5_hands_up_curr]='yes' AND [s5_hands_up_curr_prev]='no' AND [s5_hands_up_d_known]='yes'	Date of gain of raising hands ability yyyy-mm-dd *If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.*	text (date_ymd)
215	[s5_date_of_hands_up_loss] Show the field ONLY if: [s5_hands_up_curr]='no' AND [s5_hands_up_curr_prev]='yes' AND [s5_hands_up_d_known]='yes'	Date of loss of raising hands ability yyyy-mm-dd *If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.*	text (date_ymd)
216	[s5_hold_head_curr] Show the field ONLY if: [arm-number]=5 AND [age_at_visit] < 16	Can the patient currently hold their head up without support?	radio yes, Yes no, No unknown, Unknown
217	[s5_hold_head_curr_prev]	s5_hold_head_curr_prev: Value of s5_hold_head_curr in previous report. Is needed for piping and branching for other fields.	text Field Annotation: @CALCTEXT(if(starts_with([event-name], 'enrolment'), '', if(starts_with([event-name], 'visit') AND [current-instance]=1, [first-event-name][s5_hold_head_curr], if(starts_with([event-name], 'visit') AND [current-instance]>1, [event-name][s5_hold_head_curr][previous-instance], ''))))) @READONLY @HIDDEN @HIDDEN-PDF
218	[s5_hold_head_d_known] Show the field ONLY if: [arm-number]=5 AND ([s5_hold_head_curr]='yes' AND [s5_hold_head_curr_prev]='no' OR [s5_hold_head_curr]='no' AND [s5_hold_head_curr_prev]='yes')	When comparing this report to the last report, there was a change in in terms of the patients ability to hold their head up without support. Last time (date: [date_of_visit_prev]) you reported: Is patient able? [s5_hold_head_curr_prev] Here (date: [date_of_visit]) you reported: Is patient able? [s5_hold_head_curr:value] Do you know when this ability changed?	radio yes, Yes no, No
219	[s5_date_of_hold_head_gain] Show the field ONLY if: [s5_hold_head_curr]='yes' AND [s5_hold_head_curr_prev]='no' AND [s5_hold_head_d_known]='yes'	Date of gain of holding head up yyyy-mm-dd *If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.*	text (date_ymd)
220	[s5_date_of_hold_head_loss] Show the field ONLY if: [s5_hold_head_curr]='no' AND [s5_hold_head_curr_prev]='yes' AND [s5_hold_head_d_known]='yes'	Date of loss of holding head up yyyy-mm-dd *If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.*	text (date_ymd)
221	[s5_sitting_curr] Show the field ONLY if: [arm-number]=5 AND [age_at_visit] < 16	Can the patient currently sit without support?	radio yes, Yes no, No unknown, Unknown
222	[s5_sitting_curr_prev]	s5_sitting_curr_prev: Value of s5_sitting_curr in previous report. Is needed for piping and branching for other fields.	text Field Annotation: @CALCTEXT(if(starts_with([event-name], 'enrolment'), '', if(starts_with([event-name], 'visit') AND [current-instance]=1, [first-event-name][s5_sitting_curr], if(starts_with([event-name], 'visit') AND [current-instance]>1, [event-name][s5_sitting_curr][previous-instance], '')))) @READONLY @HIDDEN @HIDDEN-PDF
223	[s5_sitting_d_known] Show the field ONLY if: [arm-number]=5 AND ([s5_sitting_curr]='yes' AND [s5_sitting_curr_prev]='no' OR [s5_sitting_curr]='no' AND [s5_sitting_curr_prev]='yes')	When comparing this report to the last report, there was a change in in terms of the patients ability to sit without support. Last time (date: [date_of_visit_prev]) you reported: Is patient able? [s5_sitting_curr_prev] Here (date: [date_of_visit]) you reported: Is patient able? [s5_sitting_curr:value] Do you know when this ability changed?	radio yes, Yes no, No
224	[s5_date_of_sitting_gain] Show the field ONLY if: [s5_sitting_curr]='yes' AND [s5_sitting_curr_prev]='no' AND [s5_sitting_d_known]='yes'	Date of gain of sitting ability yyyy-mm-dd *If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.*	text (date_ymd)
225	[s5_date_of_sitting_loss] Show the field ONLY if: [s5_sitting_curr]='no' AND [s5_sitting_curr_prev]='yes' AND [s5_sitting_d_known]='yes'	Date of loss of sitting ability yyyy-mm-dd *If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.*	text (date_ymd)

226	[has_cognitive_impairment]	Section Header: *Cognitive Impairment* Cognitive impairment	radio yes, Yes no, No unknown, Unknown
227	[cognitive_impairment_type] Show the field ONLY if: [has_cognitive_impairment] = 'yes'	Type of cognitive impairment	radio developmental, Developmental aqcuired, Acquired unknown, Unknown
228	[has_breathing_difficulties]	Section Header: *Respiratory / Ventilation* Breathing difficulties	radio yes, Yes no, No unknown, Unknown
229	[has_noct_hypovent]	Symptoms of nocturnal hypoventilation	radio yes, Yes no, No unknown, Unknown
230	[fvc_seated_known] Show the field ONLY if: [has_breathing_difficulties] = 'yes'	Forced Vital Capacity (FVC) seated known	radio yes, Yes no, No
231	[fvc_seated] Show the field ONLY if: [fvc_seated_known] = 'yes'	Forced Vital Capacity (FVC) seated % of estimated	text (number)
232	[fvc_lying_known] Show the field ONLY if: [has_breathing_difficulties] = 'yes'	Forced Vital Capacity (FVC) lying known	radio yes, Yes no, No
233	[fvc_lying] Show the field ONLY if: [fvc_lying_known] = 'yes'	Forced Vital Capacity (FVC) lying % of estimated	text (number)
234	[ventilation_required] Show the field ONLY if: [has_breathing_difficulties] = 'yes'	Assisted ventilation required	radio yes, Yes no, No unknown, Unknown
235	[ventilation_times_noninv] Show the field ONLY if: [ventilation_required] = 'yes'	Noninvasive ventilation	radio night, Night day_night, Day & Night during_exacerb, During exacerbations unknown, Unknown not_applicable, Not applicable
236	[ventilation_times_inv] Show the field ONLY if: [ventilation_required] = 'yes'	Invasive Ventilation	radio night, Night day_night, Day & Night during_exacerb, During exacerbations unknown, Unknown not_applicable, Not applicable
237	[s4_vent_days_py_known] Show the field ONLY if: [arm-number]=4	Number of ventilation days in the last 12 months known	radio yes, Yes no, No

238	[s4_vent_days_py] Show the field ONLY if: [arm-number]=4	Number of ventilation days in the last 12 months	text (integer), Required
239	[has_feed_swal_difficulties]	Section Header: *Feeding* Feeding or swallowing difficulties	radio yes, Yes no, No unknown, Unknown
240	[has_feeding_difficulties] Show the field ONLY if: [has_feed_swal_difficulties]='yes'	Feeding difficulties	radio yes, Yes no, No unknown, Unknown
241	[has_swallow_difficulties] Show the field ONLY if: [has_feed_swal_difficulties]='yes'	Swallowing difficulties	radio yes, Yes no, No unknown, Unknown
242	[independent_feeding] Show the field ONLY if: isblankormissingcode([age_at_visit]) OR [age_at_visit] >= 3	Independent feeding (hand to mouth)	radio yes, Yes no, No unknown, Unknown
243	[has_tube_feeding_gastro] Show the field ONLY if: [has_feed_swal_difficulties]='yes'	Gastrostomy tube feeding	radio yes, Yes no, No unknown, Unknown
244	[has_tube_feeding_nasog] Show the field ONLY if: [has_feed_swal_difficulties]='yes'	Nasogastric tube feeding	radio yes, Yes no, No unknown, Unknown
245	[s3_receives_mitococktail] Show the field ONLY if: [arm-number]=3	"Mitococktail" or food supplements? *Does the patient receive "Mitococktail" or food supplements?*	radio yes, Yes no, No unknown, Unknown
246	[has_cardiac_involvement]	Section Header: *Cardiac* Cardiac involvement	radio yes, Yes no, No unknown, Unknown
247	[which_cardiac_syndrome] Show the field ONLY if: [has_cardiac_involvement] = 'yes'	Cardiac syndrome (ORPHAcode)	text BIOPORTAL:ORDO, BIOPORTAL:ORDO
248	[has_impl_cardio_defib] Show the field ONLY if: [has_cardiac_involvement]='yes'	Does the patient have a cardioverter defibrillator?	radio yes, Yes no, No unknown, Unknown
249	[had_heart_transplantation] Show the field ONLY if: [has_cardiac_involvement]='yes'	Has the patient had a heart transplantation?	radio yes, Yes no, No unknown, Unknown
250	[has_pacemaker] Show the field ONLY if:	Does the patient have a pacemaker?	radio yes, Yes

	[has_cardiac_involvement]='yes'		no, No unknown, Unknown
251	[had_cardiac_resync_therapy] Show the field ONLY if: [has_cardiac_involvement]='yes'	Has the patient had Cardiac Resynchronization Therapy (CRT)?	radio yes, Yes no, No unknown, Unknown
252	[has_scoliosis]	Section Header: *Scoliosis* Scoliosis	radio yes, Yes no, No unknown, Unknown
253	[had_scoliosis_surgery] Show the field ONLY if: [has_scoliosis] = 'yes'	Has the patient had a scoliosis surgery?	radio yes, Yes no, No unknown, Unknown
254	[scoliosis_cobb_angle_known] Show the field ONLY if: [has_scoliosis] = 'yes'	Scoliosis Cobb Angle known	radio yes, Yes no, No
255	[scoliosis_cobb_angle] Show the field ONLY if: [has_scoliosis] = 'yes' AND [scoliosis_cobb_angle_known] = 'yes'	Scoliosis Cobb Angle	text (number), Required
256	[s3_has_mito_asso_epilepsy] Show the field ONLY if: [arm-number]=3	Section Header: *Epilepsy* Does the patient have Mito-associated epilepsy?	radio yes, Yes no, No unknown, Unknown
257	[phenotypes_description1]	Section Header: *PHENOTYPES* Please enter the most important symptom(s) for the neuromuscular clinic appointment here. Some of the following fields may be prepopulated with data from a previous visit. Please make sure that all values are correct.Phenotypes are to be coded with the Human Phenotype Ontology (HPO). Element 6.3. in the Set of common data elements for Rare Diseases Registration (link)	descriptive
258	[phenotype_matrix]	No Provide phenotype Phenotype (HPO) 1 {pheno_yes_1} {pheno_hpo_1} 2 {pheno_yes_2} {pheno_hpo_2} 3 {pheno_yes_3} {pheno_hpo_3} 4 {pheno_yes_4} {pheno_hpo_4} 5 {pheno_yes_5} {pheno_hpo_5} 6 {pheno_yes_6} {pheno_hpo_6} 7 {pheno_yes_7} {pheno_hpo_7} 8 {pheno_yes_8} {pheno_hpo_8} 9 {pheno_yes_9} {pheno_hpo_9} 10 {pheno_yes_10} {pheno_hpo_10}	descriptive Field Annotation: @HIDDEN-PDF
259	[pheno_yes_1]	Phenotype 1 yes/no	yesno, Required 1, Yes 0, No Custom alignment: LV Field Annotation: @DEFAULT='1' @READONLY
260	[pheno_hpo_1] Show the field ONLY if: [pheno_yes_1]='1'	Phenotype: HPO-code (1)	text, Required BIOPORTAL:HP, BIOPORTAL:HP Field Annotation: @DEFAULT='[event-name][pheno_hpo_1:value][previous-instance]'
261	[pheno_yes_2]	Phenotype 2 yes/no	yesno 1, Yes 0, No Custom alignment: LV Field Annotation: @DEFAULT='[event-name][pheno_yes_2:value][previous-instance]'

262	[pheno_hpo_2] Show the field ONLY if: [pheno_yes_2]='1'	Phenotype: HPO-code (2)	text, Required BIOPORTAL:HP | BIOPORTAL:HP Field Annotation: @DEFAULT='[event-name][pheno_hpo_2:value][previous-instance]'
263	[pheno_yes_3]	Phenotype 3 yes/no	yesno 1 Yes 0 No Custom alignment: LV Field Annotation: @DEFAULT='[event-name][pheno_yes_3:value][previous-instance]'
264	[pheno_hpo_3] Show the field ONLY if: [pheno_yes_3]='1'	Phenotype: HPO-code (3)	text, Required BIOPORTAL:HP | BIOPORTAL:HP Field Annotation: @DEFAULT='[event-name][pheno_hpo_3:value][previous-instance]'
265	[pheno_yes_4]	Phenotype 4 yes/no	yesno 1 Yes 0 No Custom alignment: LV Field Annotation: @DEFAULT='[event-name][pheno_yes_4:value][previous-instance]'
266	[pheno_hpo_4] Show the field ONLY if: [pheno_yes_4]='1'	Phenotype: HPO-code (4)	text, Required BIOPORTAL:HP | BIOPORTAL:HP Field Annotation: @DEFAULT='[event-name][pheno_hpo_4:value][previous-instance]'
267	[pheno_yes_5]	Phenotype 5 yes/no	yesno 1 Yes 0 No Custom alignment: LV Field Annotation: @DEFAULT='[event-name][pheno_yes_5:value][previous-instance]'
268	[pheno_hpo_5] Show the field ONLY if: [pheno_yes_5]='1'	Phenotype: HPO-code (5)	text, Required BIOPORTAL:HP | BIOPORTAL:HP Field Annotation: @DEFAULT='[event-name][pheno_hpo_5:value][previous-instance]'
269	[pheno_yes_6]	Phenotype 6 yes/no	yesno 1 Yes 0 No Custom alignment: LV Field Annotation: @DEFAULT='[event-name][pheno_yes_6:value][previous-instance]'
270	[pheno_hpo_6] Show the field ONLY if: [pheno_yes_6]='1'	Phenotype: HPO-code (6)	text, Required BIOPORTAL:HP | BIOPORTAL:HP Field Annotation: @DEFAULT='[event-name][pheno_hpo_6:value][previous-instance]'
271	[pheno_yes_7]	Phenotype 7 yes/no	yesno 1 Yes 0 No Custom alignment: LV Field Annotation: @DEFAULT='[event-name][pheno_yes_7:value][previous-instance]'
272	[pheno_hpo_7] Show the field ONLY if: [pheno_yes_7]='1'	Phenotype: HPO-code (7)	text, Required BIOPORTAL:HP | BIOPORTAL:HP Field Annotation: @DEFAULT='[event-name][pheno_hpo_7:value][previous-instance]'
273	[pheno_yes_8]	Phenotype 8 yes/no	yesno 1 Yes

			0 | No Custom alignment: LV Field Annotation: @DEFAULT='[event-name][pheno_yes_8:value][previous-instance]'
274	[pheno_hpo_8] Show the field ONLY if: [pheno_yes_8]='1'	Phenotype: HPO-code (8)	text, Required BIOPORTAL:HP | BIOPORTAL:HP Field Annotation: @DEFAULT='[event-name][pheno_hpo_8:value][previous-instance]'
275	[pheno_yes_9]	Phenotype 9 yes/no	yesno 1 | Yes 0 | No Custom alignment: LV Field Annotation: @DEFAULT='[event-name][pheno_yes_9:value][previous-instance]'
276	[pheno_hpo_9] Show the field ONLY if: [pheno_yes_9]='1'	Phenotype: HPO-code (9)	text, Required BIOPORTAL:HP | BIOPORTAL:HP Field Annotation: @DEFAULT='[event-name][pheno_hpo_9:value][previous-instance]'
277	[pheno_yes_10]	Phenotype 10 yes/no	yesno 1 | Yes 0 | No Custom alignment: LV Field Annotation: @DEFAULT='[event-name][pheno_yes_10:value][previous-instance]'
278	[pheno_hpo_10] Show the field ONLY if: [pheno_yes_10]='1'	Phenotype: HPO-code (10)	text, Required BIOPORTAL:HP | BIOPORTAL:HP Field Annotation: @DEFAULT='[event-name][pheno_hpo_10:value][previous-instance]'
279	[review_medi_desc1]	Section Header: *Medication Review* This section is intended to review the recorded information on this patient's medications. An overview of all recorded medications is also visible on the patient's Record Home Page.	descriptive
280	[review_medi_all]	Show all recorded medications for this patient	sql
281	[review_medi_ongoing]	Show all recorded ongoing medications for this patient	sql Field Annotation: @HIDDEN @HIDDEN-PDF
282	[medi_change_yes] Show the field ONLY if: !starts_with([event-name], 'enrolment')	Are there any changes in the patient's medications compared to the available data listed in the dropdowns above?	radio yes | Yes no | No unknown | Unknown
283	[review_medi_desc2] Show the field ONLY if: [medi_change_yes]=1	Please complete the medication questionnaire(s) with the updated medication details (accessible from the patient's Record Home Page).	descriptive
284	[visit_data_complete]	Section Header: *Form Status* Complete?	dropdown 0 | Incomplete 1 | Unverified 2 | Complete
Instrument:* NJD: Thymus *(njd_thymus)			
285	[s4_thymus_h1]	NJD: THYMUS	descriptive
286	[date_of_assess_thym]	Date of assessment yyyy-mm-dd	text (date_ymd), Required
287	[s4_thymectomy_type] Show the field ONLY if: [arm-number]=4	Thymectomy	radio open_surgery | Open surgery endocopic | Endocopic not_done | Not done

			unknown: Unknown
288	[s4_date_of_thymectomy] Show the field ONLY if: [arm-number]=4 AND ([s4_thymectomy_type] = 'open_surgery' OR [s4_thymectomy_type] = 'endocopic')	Date of thymectomy yyyy-mm-dd *If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.*	text (date_ymd)
289	[s4_thym_histology] Show the field ONLY if: [arm-number] = 4 AND ([s4_thymectomy_type] = 'open_surgery' OR [s4_thymectomy_type] = 'endocopic')	Thymus histology	radio thymoma: Thymoma other: Other unknown: Unknown
290	[s4_thym_mk_stage] Show the field ONLY if: [arm-number]=4 AND [s4_thym_histology] = 'thymoma'	Masaoka-Koga stage	radio I: I IIa: IIa IIb: IIb III: III IVa: IVa IVb: IVb unknown: Unknown
291	[s4_thym_who_class] Show the field ONLY if: [arm-number]=4 AND [s4_thym_histology] = 'thymoma'	WHO classification	radio A: A AB: AB B1: B1 B2: B2 B3: B3 C: C unknown: Unknown
292	[s4_thym_ther_type_known] Show the field ONLY if: [arm-number]=4	Thymoma therapy known	radio yes: Yes no: No no_thymoma: No Thymoma Field Annotation: @DEFAULT = 'yes'
293	[s4_thym_ther_type] Show the field ONLY if: [arm-number]=4 AND [s4_thym_ther_type_known] = 'yes'	Thymoma therapy	checkbox, Required surgery: s4_thym_ther_type___surgery: Surgery chemotherapy: s4_thym_ther_type___chemotherapy: Chemotherapy radiotherapy: s4_thym_ther_type___radiotherapy: Radiotherapy
294	[s4_had_thym_relapse] Show the field ONLY if: [arm-number]=4	Thymoma relapse	radio yes: Yes no: No unknown: Unknown
295	[s4_date_of_thym_relapse] Show the field ONLY if: [arm-number]=4 AND [s4_had_thym_relapse]='yes'	Date of thymoma relapse yyyy-mm-dd *If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.*	text (date_ymd)
296	[njd_thymus_complete]	Section Header: *Form Status* Complete?	dropdown 0: Incomplete 1: Unverified 2: Complete
Instrument: **NJD: Quantitative Myasthenia Gravis Test (QMG)** **(*njd_quantitative_myasthenia_gravis_test_qmg*)**			
297	[cms_qmg_h1]	Quantitative Myasthenia Gravis Test (QMG)	descriptive

298	[date_of_assess_qmg]	Date of assessment yyyy-mm-dd	text (date_ymd), Required
299	[cms_qmg_subscores_known]	Score of the individual data items is known	radio yes, Yes no, No
300	[cms_qmg_score_manual] Show the field ONLY if: [cms_qmg_subscores_known] = 'no'	Total QMG Score (manual) *Please enter a number in the range [0, 39]*	text (number, Min: 0, Max: 39)
301	[cms_qmg1] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	1. Double vision on lateral gaze right or left (circle one), seconds	radio 0, 61 1, 11-60 2, 1-10 3, Spontaneous
302	[cms_qmg1_score] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Item 1 Score	calc Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg1] = '0', 0, 0) or if([cms_qmg1] = '1', 1, 0) or if([cms_qmg1] = '2', 2, 0) or if([cms_qmg1] = '3', 3, 0), '') Field Annotation: @HIDDEN @HIDDEN-PDF
303	[cms_qmg2] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	2. Ptosis (upward gaze), seconds	radio 0, 61 1, 11-60 2, 1-10 3, Spontaneous
304	[cms_qmg2_score] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Item 2 Score	calc Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg2] = '0', 0, 0) or if([cms_qmg2] = '1', 1, 0) or if([cms_qmg2] = '2', 2, 0) or if([cms_qmg2] = '3', 3, 0), '') Field Annotation: @HIDDEN @HIDDEN-PDF
305	[cms_qmg3] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	3. Facial Muscles	radio 0, Normal lid closure 1, Complete weak, some resistance 2, Complete, without resistance 3, Incomplete
306	[cms_qmg3_score] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Item 3 Score	calc Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg3] = '0', 0, 0) or if([cms_qmg3] = '1', 1, 0) or if([cms_qmg3] = '2', 2, 0) or if([cms_qmg3] = '3', 3, 0), '') Field Annotation: @HIDDEN @HIDDEN-PDF
307	[cms_qmg4] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	4. Swallowing 4 oz. / 120 mL water	radio 0, Normal 1, Minimal coughing or throat clearing 2, Severe coughing/choking or nasal regurgitation 3, Cannot swallow (test not attempted)
308	[cms_qmg4_score] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Item 4 Score	calc Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg4] = '0', 0, 0) or if([cms_qmg4] = '1', 1, 0) or if([cms_qmg4] = '2', 2, 0) or if([cms_qmg4] = '3', 3, 0), '') Field Annotation: @HIDDEN @HIDDEN-PDF
309	[cms_qmg5] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	5. Speech following counting aloud from 1 to 50 (onset of dysarthria)	radio 0, None at #50 1, Dysarthria at #30-49 2, Dysarthria at #10-29 3, Dysarthria at #9

310	[cms_qmg5_score] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Item 5 Score	calc Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg5] = '0', 0, 0) or if([cms_qmg5] = '1', 1, 0) or if([cms_qmg5] = '2', 2, 0) or if([cms_qmg5] = '3', 3, 0), '') Field Annotation: @HIDDEN @HIDDEN-PDF
311	[cms_qmg6] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	6. Right arm outstretched (90 degrees sitting), seconds	radio 0, 240 1, 90-239 2, 10-89 3, 0-9
312	[cms_qmg6_score] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Item 6 Score	calc Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg6] = '0', 0, 0) or if([cms_qmg6] = '1', 1, 0) or if([cms_qmg6] = '2', 2, 0) or if([cms_qmg6] = '3', 3, 0), '') Field Annotation: @HIDDEN @HIDDEN-PDF
313	[cms_qmg7] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	7. Left arm outstretched (90 degrees sitting), seconds	radio 0, 240 1, 90-239 2, 10-89 3, 0-9
314	[cms_qmg7_score] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Item 7 Score	calc Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg7] = '0', 0, 0) or if([cms_qmg7] = '1', 1, 0) or if([cms_qmg7] = '2', 2, 0) or if([cms_qmg7] = '3', 3, 0), '') Field Annotation: @HIDDEN @HIDDEN-PDF
315	[cms_qmg8] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	8. Vital capacity (% predicted) mouthpiece or facemask (circle 1, best of 3)	radio 0, greater than or equal to 80% 1, 65-79% 2, 50-64% 3, < 50%
316	[cms_qmg8_score] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Item 8 Score	calc Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg8] = '0', 0, 0) or if([cms_qmg8] = '1', 1, 0) or if([cms_qmg8] = '2', 2, 0) or if([cms_qmg8] = '3', 3, 0), '') Field Annotation: @HIDDEN @HIDDEN-PDF
317	[cms_qmg9] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	9. Right hand grip: (best of 2), kgW	radio 0, Male 45+, Female 30+ 1, Male 15-44, Female 10-29 2, Male 5-14, Female 5-9 3, 0-4
318	[cms_qmg9_score] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Item 9 Score	calc Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg9] = '0', 0, 0) or if([cms_qmg9] = '1', 1, 0) or if([cms_qmg9] = '2', 2, 0) or if([cms_qmg9] = '3', 3, 0), '') Field Annotation: @HIDDEN @HIDDEN-PDF
319	[cms_qmg10] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	10. Left hand grip: (best of 2), kgW	radio 0, Male 45+, Female 30+ 1, Male 15-44, Female 10-29 2, Male 5-14, Female 5-9 3, 0-4
320	[cms_qmg10_score] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Item 10 Score	calc Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg10] = '0', 0, 0) or if([cms_qmg10] = '1', 1, 0) or if([cms_qmg10] = '2', 2, 0) or if([cms_qmg10] = '3', 3, 0), '') Field Annotation: @HIDDEN @HIDDEN-PDF
321	[cms_qmg11] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	11. Head, lifted (45 degrees supine), seconds	radio 0, 120 1, 30-119 2, 1-29

			3 | 0
322	[cms_qmg11_score] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Item 11 Score	calc Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg11] = '0', 0, 0) or if([cms_qmg11] = '1', 1, 0) or if([cms_qmg11] = '2', 2, 0) or if([cms_qmg11] = '3', 3, 0), '') Field Annotation: @HIDDEN @HIDDEN-PDF
323	[cms_qmg12] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	12. Right leg outstretched (45 degrees supine), seconds	radio 0 | 100 1 | 31-99 2 | 1-30 3 | 0
324	[cms_qmg12_score] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Item 12 Score	calc Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg12] = '0', 0, 0) or if([cms_qmg12] = '1', 1, 0) or if([cms_qmg12] = '2', 2, 0) or if([cms_qmg12] = '3', 3, 0), '') Field Annotation: @HIDDEN @HIDDEN-PDF
325	[cms_qmg13] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	13. Left leg outstretched (45 degrees supine), seconds	radio 0 | 100 1 | 31-99 2 | 1-30 3 | 0
326	[cms_qmg13_score] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Item 13 Score	calc Calculation: if([cms_qmg_subscores_known] = 'yes', if([cms_qmg13] = '0', 0, 0) or if([cms_qmg13] = '1', 1, 0) or if([cms_qmg13] = '2', 2, 0) or if([cms_qmg13] = '3', 3, 0), '') Field Annotation: @HIDDEN @HIDDEN-PDF
327	[cms_qmg_score_calc] Show the field ONLY if: [cms_qmg_subscores_known] = 'yes'	Total QMG Score (calculated)	calc Calculation: if([cms_qmg_subscores_known] = 'yes', sum([cms_qmg1_score], [cms_qmg2_score], [cms_qmg3_score], [cms_qmg4_score], [cms_qmg5_score], [cms_qmg6_score], [cms_qmg7_score], [cms_qmg8_score], [cms_qmg9_score], [cms_qmg10_score], [cms_qmg11_score], [cms_qmg12_score], [cms_qmg13_score]), '') Field Annotation: @READONLY
328	[njd_quantitative_myasthenia_gravis_test_qmg_complete]	Section Header: *Form Status* Complete?	dropdown 0 | Incomplete 1 | Unverified 2 | Complete
Instrument:* Motorneuron FRS-R (Age 16+ Only) *(motorneuron_frsr_age_16_only)			
329	[s5_als_frs_h1]	MOTORNEURON: FRS-R (AGE >= 16 ONLY)	descriptive
330	[s5_als_frs_description1]	Section Header: *Notes on editing* ALS Functional Rating Scale - Revised (ALS-FRS-R) must be completed for adults only (age at vist >= 16).	descriptive
331	[date_of_assess_frsr]	Date of assessment yyyy-mm-dd	text (date_ymd), Required
332	[s5_als_frs_r_speech]	Section Header: *ALS Functional Rating Scale - Revised (ALS-FRS-R)* 1. Speech	radio 0 | Loss of useful speech 1 | Speech combined with nonvocal communication 2 | Intelligible with repeating 3 | Detectable speech disturbance 4 | Normal speech process
333	[s5_als_frs_r_saliv]	2. Salivation	radio 0 | Marked drooling; requires constant tissue or handkerchief 1 | Marked escessive saliva wth some drooling 2 | Moderately excessive saliva; may have minimal drooling 3 | Slight but definitive excessive saliva in mouth; may have nighttime drooling 4 | Normal

334	[s5_als_frs_r_swallow]	3. Swallowing	radio 0 Nothing by mouth; exclusively parenteral or enteral feeding 1 Needs supplemental tube feeding 2 Dietary consistency changes 3 Early eating problems; occasional chocking 4 Normal eating habits
335	[s5_als_frs_r_handwr]	4. Handwriting	radio 0 Unable to grip pen 1 Able to grip pen but unable to write 2 Not all words are legible 3 Slow or sloppy; all words are legible 4 Normal
336	[s5_als_frs_r_gastron]	ALS-FRS-R Patient with gastrostomy and >50% daily nutrition intake via G-tube	yesno 1 Yes 0 No Field Annotation: @DEFAULT = 0
337	[s5_als_frs_r_utensils_a] Show the field ONLY if: [s5_als_frs_r_gastron] = 0	5. Cutting food and handling utensils (patients without gastrostomy)	radio 0 Needs to be fed 1 Food must be cut by someone but can still feed slowly 2 Can cut most foods although clumsy and slow; some help needed 3 Somewhat slow and clumsy but no help neded 4 Normal
338	[s5_als_frs_r_utensils_b] Show the field ONLY if: [s5_als_frs_r_gastron] = 1	5. Cutting food and handling utensils (alternate scale for patients with gastrostomy)	radio 0 Unable to perform any aspect of task 1 Provides minimal assistance to caregiver 2 Some help needed with closures and fasteners 3 Clumsy but able to perform all manipulations independently 4 Normal
339	[s5_als_frs_r_dressing]	6. Dressing and hygiene	radio 0 Total dependence 1 Needs attendant for self-care 2 Intermittent assistance or substitute methods 3 Independent and complete self-care with effort or decreased efficieny 4 Normal function
340	[s5_als_frs_r_bed]	7. Turning in bed and adjusting bed clothes	radio 0 Helpless 1 Can initiate but not turn or ajust sheets alone 2 Can turn alone or adjust sheets but with great difficulty 3 Somewhat slow and clumsy but no help needed 4 Normal
341	[s5_als_frs_r_walking]	8. Walking	radio 0 No porposeful leg movement 1 Nonambulatory functional movement 2 Walks with assistance 3 Early ambulation difficulties 4 Normal

342	[s5_als_frs_r_stairs]	9. Climbing stairs	radio 0, Cannot do 1, Needs assitance 2, Mild unsteadiness or fatigue 3, Slow 4, Normal
343	[s5_als_frs_r_dyspnea]	10. Dyspnea	radio 0, Significant difficulty; consdering using mechanical respiratory support 1, Occurs at rest; dificulty breathing when either sitting or lying 2, Occurs with one or more of the following: eating, bathing, dressing (ADL) 3, Occurs when walking 4, None
344	[s5_als_frs_r_orthopnea]	11. Orthopnea	radio 0, Unable to sleep 1, Can only sleep sittng up 2, Needs extra pillows in order to sleep (more than two) 3, Some difficulty sleeping at night due to sortness of breath; does not routinely use more than two pillows 4, None
345	[s5_als_frs_r_resp_insuf]	12. Respiratory insufficiency	radio 0, Invasive mechanical ventilation by intubation or trachostomy 1, Continuous use of BiPAP during night and day 2, Continuous use of BiPAP during the night 3, Intermittent use of BiPAP 4, None
346	[s5_als_frs_description2]	Section Header: *ALS-FRS-R Total Score* Please note: ALS-FRS-R Total score will only be calculated if all 12 questions have been answered.	descriptive
347	[s5_als_frs_r_total_score]	ALS-FRS-R total score	calc Calculation: if([s5_als_frs_r_gastron]=0, ([s5_als_frs_r_speech]+ [s5_als_frs_r_saliv]+ [s5_als_frs_r_swallow]+ [s5_als_frs_r_handwr]+ [s5_als_frs_r_utensils_a]+ [s5_als_frs_r_dressing]+ [s5_als_frs_r_bed]+ [s5_als_frs_r_walking]+ [s5_als_frs_r_stairs]+ [s5_als_frs_r_dyspnea]+ [s5_als_frs_r_orthopnea]+ [s5_als_frs_r_resp_insuf]), ([s5_als_frs_r_speech]+ [s5_als_frs_r_saliv]+ [s5_als_frs_r_swallow]+ [s5_als_frs_r_handwr]+ [s5_als_frs_r_utensils_b]+ [s5_als_frs_r_dressing]+ [s5_als_frs_r_bed]+ [s5_als_frs_r_walking]+ [s5_als_frs_r_stairs]+ [s5_als_frs_r_dyspnea]+ [s5_als_frs_r_orthopnea]+ [s5_als_frs_r_resp_insuf]))
348	[motorneuron_frsr_age_16_only_complete]	Section Header: *Form Status* Complete?	dropdown 0, Incomplete 1, Unverified 2, Complete
Instrument:* Motor Function Measures *(motor_function_measures)			
349	[motor_measures_h1]	Motor Function Measures	descriptive
350	[date_of_assess_mm]	Date of assessment yyyy-mm-dd	text (date_ymd), Required
351	[age_at_visit_mm] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2)	Age of patient (years)	calc Calculation: if([arm-number]=5, rounddown(datediff([enrolment_and_firs_arm_5][date_of_birth], [date_of_assess_mm], "y")), if([arm-number]=2, rounddown(datediff([enrolment_and_firs_arm_2][date_of_birth], [date_of_assess_mm], "y")), -999))
352	[show_mm_child] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [age_at_visit_mm] < 16	Section Header: *Children only motor measures (age < 16)* Motor Measure Performed? Value / Result Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP-INTEND) {s5_chop_intend_taken} {s5_chop_intend_score} Hammersmith Functional Motor Scale (HFMS) {hfms_taken} {hfms_score} Hammersmith Functional Motor Scale Expanded (HFMS-E) {hfmse_taken} {hfmse_score} Hammersmith Infant Neurological Examination (HINE) Section 2 {hine_taken} {hine_score}	descriptive

353	[s5_chop_intend_taken] Show the field ONLY if: [arm-number]=5 AND [age_at_visit_mm] < 16	s5_chop_intend_taken	yesno 1 Yes 0 No
354	[s5_chop_intend_score] Show the field ONLY if: [arm-number]=5 AND [age_at_visit_mm] < 16 AND [s5_chop_intend_taken] = 1	CHOP-INTEND-Score *Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders*	text (number)
355	[hfms_taken] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [age_at_visit_mm] < 16	hfms_taken	yesno 1 Yes 0 No
356	[hfms_score] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [age_at_visit_mm] < 16 AND [hfms_taken] = 1	HFMS-Score *Hammersmith Functional Motor Scale*	text (number)
357	[hfmse_taken] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [age_at_visit_mm] < 16	hfmse_taken	yesno 1 Yes 0 No
358	[hfmse_score] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [age_at_visit_mm] < 16 AND [hfmse_taken] = 1	HFMS-E-Score *Hammersmith Functional Motor Scale Expanded*	text (number)
359	[hine_taken] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [age_at_visit_mm] < 16	hine_taken	yesno 1 Yes 0 No
360	[hine_score] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [age_at_visit_mm] < 16 AND [hine_taken] = 1	HINE-Section2-Score *Hammersmith Infant Neurological Examination (HINE) Section 2*	text (number)
361	[s5_show_mm_adult] Show the field ONLY if: [arm-number]=5 AND [age_at_visit_mm] >= 16	Section Header: *Adult only motor mesures (age >= 16)* Motor Measure Performed? Value / Result Adult Test of Neuromuscular Disorders (CHOP-ATEND) {s5_chop_atend_taken} {s5_chop_atend_score}	descriptive
362	[s5_chop_atend_taken] Show the field ONLY if: [arm-number]=5 AND [age_at_visit_mm] >= 16	s5_chop_atend_taken	yesno 1 Yes 0 No
363	[s5_chop_atend_score] Show the field ONLY if: [arm-number]=5 AND [age_at_visit_mm] >= 16 AND [s5_chop_atend_taken] = 1	CHOP-ATEND-Score *Adult Test of Neuromuscular Disorders (CHOP-ATEND)*	text (number)
364	[show_mm_general] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2)	Section Header: *Age independent motor measures* Motor Measure Performed? Value / Result Revised Upper Limb Modul (RULM) {rulm_taken} {rulm_score} Motor Function Measurement (MFM) {mfm_taken} {mfm_result} 6 Minute Walk Test: Result in metres (m) {six_mwt_taken} {six_mwt_result} 10 Minute Walk Test: Result in metres (m) {ten_mwt_taken} {ten_mwt_result} Timed "Up and Go" (TUG) {tug_taken} {tug_result} Other motor measure {other_mm_taken} {which_other_mm}	descriptive
365	[rulm_taken] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2)	rulm_taken	yesno 1 Yes 0 No
366	[rulm_score] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [rulm_taken] = 1	RULM-Score *Revised Upper Limb Measure (RULM)*	text (number)

367	[mfm_taken] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2)	mfm_taken	yesno 1 Yes 0 No
368	[mfm_result] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [mfm_taken] = 1	MFM-Result *Motor Function Measurement (MFM)*	text (number)
369	[six_mwt_taken] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2)	six_mwt_taken	yesno 1 Yes 0 No
370	[six_mwt_result] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [six_mwt_taken] = 1	6 Minute Walk Test (6MWT)	text (number)
371	[ten_mwt_taken] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2)	ten_mwt_taken	yesno 1 Yes 0 No
372	[ten_mwt_result] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [ten_mwt_taken] = 1	10 Minute Walk Test (10MWT)	text (number)
373	[tug_taken] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2)	tug_taken	yesno 1 Yes 0 No
374	[tug_result] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [tug_taken] = 1	TUG-Result *Timed "Up & Go" (TUG)*	text (number)
375	[other_mm_taken] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2)	s5_other_mm_taken	yesno 1 Yes 0 No
376	[which_other_mm] Show the field ONLY if: ([arm-number]=5 OR [arm-number]=2) AND [other_mm_taken] = 1	Other motor measure *Which other motor measure has been performed?*	text
377	[motor_function_measures_complete]	Section Header: *Form Status* Complete?	dropdown 0 Incomplete 1 Unverified 2 Complete
Instrument: **Genetic Diagnosis: Changes in nuclear DNA** ***(genetic_diagnosis_changes_in_nuclear_dna)***			
378	[gendia_h1]	Genetic Diagnosis: Changes in nuclear DNA	descriptive
379	[date_of_gendia]	Date of genetic diagnosis [or if no genetic diagnosis: date of report] yyyy-mm-dd	text (date_ymd), Required
380	[gendia_known]	Do you have a genetic diagnosis for this patient?	radio, Required yes Yes no No
381	[gendia_label] Show the field ONLY if: [gendia_known] = 'yes'	Genetic diagnosis name *This is a label to identify this diagnostic record to the registry user. It will be displayed on the record home page together with the date of diagnosis. You can name it anything you find helpful - consider something like the gene name and inheritance pattern or similar.*	text, Required

382	[gendia_label_disp]	gendia_label_disp	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_known]='yes',concat([gendia_label], ' (', [date_of_gendia], ')'),'[date_of_gendia]'))
383	[gendia_desc1] Show the field ONLY if: [gendia_known] = 'yes'	The disease-group of this patient is: [arm-label]The clinical diagnosis of the neuromuscular disease for this patient is (from Baseline-form): [first-event-name][diagnosis_rd_orpha] ([first-event-name][diagnosis_rd_orpha:value])	descriptive
384	[gendia_listpref] Show the field ONLY if: [gendia_known] = 'yes'	Which gene is affected? *Genetic diagnosis retained by the specialised centre (Element 6.2. in the Set of common data elements for Rare Diseases Registration (link))*	radio, Required all_nmd | Search in full neuromuscular disease gene list ontology | Gene is not in neuromuscular disease gene list Field Annotation: @DEFAULT=if([show_snv_fields]=1,'disease_group','')
385	[gendia_omim_list] Show the field ONLY if: [gendia_listpref]='all_nmd'	Gene Symbol (HGNC) for all neuromuscular diseases *Genetic diagnosis retained by the specialised centre (Element 6.2. in the Set of common data elements for Rare Diseases Registration (link))*	dropdown (autocomplete), Required 601065 | AARS1 / HGNC:20 / OMIM:601065 (alanyl-tRNA synthetase 1) 612035 | AARS2 / HGNC:21022 / OMIM:612035 (alanyl-tRNA synthetase 2, mitochondrial) 601439 | ABCC9 / HGNC:60 / OMIM:601439 (ATP binding cassette subfamily C member 9) 604780 | ABHD5 / HGNC:21396 / OMIM:604780 (abhydrolase domain containing 5, lysophosphatidic acid acyltransferase) 611103 | ACAD9 / HGNC:21497 / OMIM:611103 (acyl-CoA dehydrogenase family member 9) 609575 | ACADVL / HGNC:92 / OMIM:609575 (acyl-CoA dehydrogenase very long chain) 617036 | ACER3 / HGNC:16066 / OMIM:617036 (alkaline ceramidase 3) 102610 | ACTA1 / HGNC:129 / OMIM:102610 (actin alpha 1, skeletal muscle) 102540 | ACTC1 / HGNC:143 / OMIM:102540 (actin alpha cardiac muscle 1) 102573 | ACTN2 / HGNC:164 / OMIM:102573 (actinin alpha 2) 102576 | ACVR1 / HGNC:171 / OMIM:102576 (activin A receptor type 1) 606980 | COQ8A / HGNC:16812 / OMIM:606980 (coenzyme Q8A) 600294 | ADCY6 / HGNC:237 / OMIM:600294 (adenylate cyclase 6) 612243 | ADGRG6 / HGNC:13841 / OMIM:612243 (adhesion G protein-coupled receptor G6) 612498 | ADSS1 / HGNC:20093 / OMIM:612498 (adenylosuccinate synthase 1) 604581 | AFG3L2 / HGNC:315 / OMIM:604581 (AFG3 like matrix AAA peptidase subunit 2) 610860 | AGL / HGNC:321 / OMIM:610860 (amylo-alpha-1, 6-glucosidase, 4-alpha-glucanotransferase) 103320 | AGRN / HGNC:329 / OMIM:103320 (agrin) 606830 | AGTPBP1 / HGNC:17258 / OMIM:606830 (ATP/GTP binding carboxypeptidase 1) 608570 | AHNAK2 / HGNC:20125 / OMIM:608570 (AHNAK nucleoprotein 2) 300169 | AIFM1 / HGNC:8768 / OMIM:300169 (apoptosis inducing factor mitochondria associated 1) 604001 | AKAP9 / HGNC:379 / OMIM:604001 (A-kinase anchoring protein 9) 138250 | ALDH18A1 / HGNC:9722 / OMIM:138250 (aldehyde dehydrogenase 18 family member A1) 609523 | ALDH3A2 / HGNC:403 / OMIM:609523 (aldehyde dehydrogenase 3 family member A2) 300776 | ALG13 / HGNC:30881 / OMIM:300776 (ALG13 UDP-N-acetylglucosaminyltransferase subunit) 612866 | ALG14 / HGNC:28287 / OMIM:612866 (ALG14 UDP-N-acetylglucosaminyltransferase subunit) 607905 | ALG2 / HGNC:23159 / OMIM:607905 (ALG2 alpha-1,3/1,6-mannosyltransferase) 617608 | ALPK3 / HGNC:17574 / OMIM:617608 (alpha kinase 3) 606352 | ALS2 / HGNC:443 / OMIM:606352 (alsin Rho guanine nucleotide exchange factor ALS2) 102771 | AMPD2 / HGNC:469 / OMIM:102771 (adenosine monophosphate deaminase 2) 105850 | ANG / HGNC:483 / OMIM:105850 (angiogenin) 106410 | ANK2 / HGNC:493 / OMIM:106410 (ankyrin 2) 609599 | ANKRD1 / HGNC:15819 / OMIM:609599 (ankyrin repeat domain 1) 613726 | ANO10 / HGNC:25519 / OMIM:613726 (anoctamin 10) 608662 | ANO5 / HGNC:27337 / OMIM:608662 (anoctamin 5) 602572 | ANXA11 / HGNC:535 / OMIM:602572 (annexin A11)

607245	AP4B1 / HGNC:572 / OMIM:607245 (adaptor related protein complex 4 subunit beta 1)
607244	AP4E1 / HGNC:573 / OMIM:607244 (adaptor related protein complex 4 subunit epsilon 1)
602296	AP4M1 / HGNC:574 / OMIM:602296 (adaptor related protein complex 4 subunit mu 1)
607243	AP4S1 / HGNC:575 / OMIM:607243 (adaptor related protein complex 4 subunit sigma 1)
613653	AP5Z1 / HGNC:22197 / OMIM:613653 (adaptor related protein complex 5 subunit zeta 1)
606350	APTX / HGNC:15984 / OMIM:606350 (aprataxin)
313700	AR / HGNC:644 / OMIM:313700 (androgen receptor)
608136	ARHGEF10 / HGNC:14103 / OMIM:608136 (Rho guanine nucleotide exchange factor 10)
607669	ARL6IP1 / HGNC:697 / OMIM:607669 (ADP ribosylation factor like GTPase 6 interacting protein 1)
613468	ASAH1 / HGNC:735 / OMIM:613468 (N-acylsphingosine amidohydrolase 1)
614215	ASCC1 / HGNC:24268 / OMIM:614215 (activating signal cointegrator 1 complex subunit 1)
604261	ATG5 / HGNC:589 / OMIM:604261 (autophagy related 5)
606439	ATL1 / HGNC:11231 / OMIM:606439 (atlastin GTPase 1)
609369	ATL3 / HGNC:24526 / OMIM:609369 (atlastin GTPase 3)
607585	ATM / HGNC:795 / OMIM:607585 (ATM serine/threonine kinase)
610513	ATP13A2 / HGNC:30213 / OMIM:610513 (ATPase cation transporting 13A2)
182310	ATP1A1 / HGNC:799 / OMIM:182310 (ATPase Na+/K+ transporting subunit alpha 1)
182340	ATP1A2 / HGNC:800 / OMIM:182340 (ATPase Na+/K+ transporting subunit alpha 2)
108730	ATP2A1 / HGNC:811 / OMIM:108730 (ATPase sarcoplasmic/endoplasmic reticulum Ca2+ transporting 1)
300011	ATP7A / HGNC:869 / OMIM:300011 (ATPase copper transporting alpha)
601556	ATXN1 / HGNC:10548 / OMIM:601556 (ataxin 1)
611150	ATXN10 / HGNC:10549 / OMIM:611150 (ataxin 10)
601517	ATXN2 / HGNC:10555 / OMIM:601517 (ataxin 2)
607047	ATXN3 / HGNC:7106 / OMIM:607047 (ataxin 3)
607640	ATXN7 / HGNC:10560 / OMIM:607640 (ataxin 7)
603680	ATXN8OS / HGNC:10561 / OMIM:603680 (ATXN8 opposite strand lncRNA)
610194	B3GALNT2 / HGNC:28596 / OMIM:610194 (beta-1,3-N-acetylgalactosaminyltransferase 2)
605517	B4GAT1 / HGNC:15685 / OMIM:605517 (beta-1,4-glucuronyltransferase 1)
601873	B4GALNT1 / HGNC:4117 / OMIM:601873 (beta-1,4-N-acetyl-galactosaminyltransferase 1)
603883	BAG3 / HGNC:939 / OMIM:603883 (BAG cochaperone 3)
612051	BEAN1 / HGNC:24160 / OMIM:612051 (brain expressed associated with NEDD4 1)
609797	BICD2 / HGNC:17208 / OMIM:609797 (BICD cargo adaptor 2)
601248	BIN1 / HGNC:1052 / OMIM:601248 (bridging integrator 1)
606158	BSCL2 / HGNC:15832 / OMIM:606158 (BSCL2 lipid droplet biogenesis associated, seipin)
604577	BVES / HGNC:1152 / OMIM:604577 (blood vessel epicardial substance)
613541	MTRFR / HGNC:26784 / OMIM:613541 (mitochondrial translation release factor in rescue)
614297	C19orf12 / HGNC:25443 / OMIM:614297 (chromosome 19 open reading frame 12)
618682	C1orf194 / HGNC:32331 / OMIM:618682 (chromosome 1 open reading frame 194)
601269	C1QBP / HGNC:1243 / OMIM:601269 (complement C1q binding protein)
614260	C9orf72 / HGNC:28337 / OMIM:614260 (C9orf72-SMCR8 complex subunit)
601011	CACNA1A / HGNC:1388 / OMIM:601011 (calcium voltage-gated channel subunit alpha1 A)
114205	CACNA1C / HGNC:1390 / OMIM:114205 (calcium voltage-gated channel subunit alpha1 C)
604065	CACNA1G / HGNC:1394 / OMIM:604065 (calcium voltage-gated channel subunit alpha1 G)
607904	CACNA1H / HGNC:1395 / OMIM:607904 (calcium voltage-gated channel subunit alpha1
114208	CACNA1S / HGNC:1397 / OMIM:114208 (calcium voltage-gated channel subunit alpha

600003	CACNB2 / HGNC:1402 / OMIM:600003 (calcium voltage-gated channel auxiliary subunit beta 2)
601949	CACNB4 / HGNC:1404 / OMIM:601949 (calcium voltage-gated channel auxiliary subunit beta 4)
114180	CALM1 / HGNC:1442 / OMIM:114180 (calmodulin 1)
114182	CALM2 / HGNC:1445 / OMIM:114182 (calmodulin 2)
611414	CALR3 / HGNC:20407 / OMIM:611414 (calreticulin 3)
114220	CAPN1 / HGNC:1476 / OMIM:114220 (calpain 1)
114240	CAPN3 / HGNC:1480 / OMIM:114240 (calpain 3)
114250	CASQ1 / HGNC:1512 / OMIM:114250 (calsequestrin 1)
114251	CASQ2 / HGNC:1513 / OMIM:114251 (calsequestrin 2)
601253	CAV3 / HGNC:1529 / OMIM:601253 (caveolin 3)
603198	CAVIN1 / HGNC:9688 / OMIM:603198 (caveolae associated protein 1)
617714	CAVIN4 / HGNC:33742 / OMIM:617714 (caveolae associated protein 4)
614666	CCDC78 / HGNC:14153 / OMIM:614666 (coiled-coil domain containing 78)
611204	CCDC88C / HGNC:19967 / OMIM:611204 (coiled-coil domain containing 88C)
610150	CCT5 / HGNC:1618 / OMIM:610150 (chaperonin containing TCP1 subunit 5)
601443	CFL2 / HGNC:1875 / OMIM:601443 (cofilin 2)
118490	CHAT / HGNC:1912 / OMIM:118490 (choline O-acetyltransferase)
615903	CHCHD10 / HGNC:15559 / OMIM:615903 (coiled-coil-helix-coiled-coil-helix domain containing 10)
612395	CHKB / HGNC:1938 / OMIM:612395 (choline kinase beta)
609512	CHMP2B / HGNC:24537 / OMIM:609512 (charged multivesicular body protein 2B)
606988	CHP1 / HGNC:17433 / OMIM:606988 (calcineurin like EF-hand protein 1)
100690	CHRNA1 / HGNC:1955 / OMIM:100690 (cholinergic receptor nicotinic alpha 1 subunit)
100710	CHRNB1 / HGNC:1961 / OMIM:100710 (cholinergic receptor nicotinic beta 1 subunit)
100720	CHRND / HGNC:1965 / OMIM:100720 (cholinergic receptor nicotinic delta subunit)
100725	CHRNE / HGNC:1966 / OMIM:100725 (cholinergic receptor nicotinic epsilon subunit)
100730	CHRNG / HGNC:1967 / OMIM:100730 (cholinergic receptor nicotinic gamma subunit)
118425	CLCN1 / HGNC:2019 / OMIM:118425 (chloride voltage-gated channel 1)
607042	CLN3 / HGNC:2074 / OMIM:607042 (CLN3 lysosomal/endosomal transmembrane protein, battenin)
601273	CLTCL1 / HGNC:2093 / OMIM:601273 (clathrin heavy chain like 1)
116955	CNBP / HGNC:13164 / OMIM:116955 (CCHC-type zinc finger nucleic acid binding protein)
600016	CNTN1 / HGNC:2171 / OMIM:600016 (contactin 1)
602346	CNTNAP1 / HGNC:8011 / OMIM:602346 (contactin associated protein 1)
615623	COA7 / HGNC:25716 / OMIM:615623 (cytochrome c oxidase assembly factor 7)
120320	COL12A1 / HGNC:2188 / OMIM:120320 (collagen type XII alpha 1 chain)
120350	COL13A1 / HGNC:2190 / OMIM:120350 (collagen type XIII alpha 1 chain)
610004	COL25A1 / HGNC:18603 / OMIM:610004 (collagen type XXV alpha 1 chain)
120215	COL5A1 / HGNC:2209 / OMIM:120215 (collagen type V alpha 1 chain)
120220	COL6A1 / HGNC:2211 / OMIM:120220 (collagen type VI alpha 1 chain)
120240	COL6A2 / HGNC:2212 / OMIM:120240 (collagen type VI alpha 2 chain)
120250	COL6A3 / HGNC:2213 / OMIM:120250 (collagen type VI alpha 3 chain)
603033	COLQ / HGNC:2226 / OMIM:603033 (collagen like tail subunit of asymmetric acetylcholinesterase)
609825	COQ2 / HGNC:25223 / OMIM:609825 (coenzyme Q2, polyprenyltransferase)
612898	COQ4 / HGNC:19693 / OMIM:612898 (coenzyme Q4)
614647	COQ6 / HGNC:20233 / OMIM:614647 (coenzyme Q6, monooxygenase)

601683	COQ7 / HGNC:2244 / OMIM:601683 (coenzyme Q7, hydroxylase)
612837	COQ9 / HGNC:25302 / OMIM:612837 (coenzyme Q9)
603646	COX15 / HGNC:2263 / OMIM:603646 (cytochrome c oxidase assembly homolog COX15)
602072	COX6A1 / HGNC:2277 / OMIM:602072 (cytochrome c oxidase subunit 6A1)
602009	COX6A2 / HGNC:2279 / OMIM:602009 (cytochrome c oxidase subunit 6A2)
608846	CPT1C / HGNC:18540 / OMIM:608846 (carnitine palmitoyltransferase 1C)
600650	CPT2 / HGNC:2330 / OMIM:600650 (carnitine palmitoyltransferase 2)
614631	CRPPA / HGNC:37276 / OMIM:614631 (CDP-L-ribitol pyrophosphorylase A)
123590	CRYAB / HGNC:2389 / OMIM:123590 (crystallin alpha B)
600824	CSRP3 / HGNC:2472 / OMIM:600824 (cysteine and glycine rich protein 3)
604927	CTDP1 / HGNC:2498 / OMIM:604927 (CTD phosphatase subunit 1)
607667	CTNNA3 / HGNC:2511 / OMIM:607667 (catenin alpha 3)
616120	CWF19L1 / HGNC:25613 / OMIM:616120 (CWF19 like cell cycle control factor 1)
610670	CYP2U1 / HGNC:20582 / OMIM:610670 (cytochrome P450 family 2 subfamily U member 1)
603711	CYP7B1 / HGNC:2652 / OMIM:603711 (cytochrome P450 family 7 subfamily B member 1)
128239	DAG1 / HGNC:2666 / OMIM:128239 (dystroglycan 1)
615820	DCAF8 / HGNC:24891 / OMIM:615820 (DDB1 and CUL4 associated factor 8)
601143	DCTN1 / HGNC:2711 / OMIM:601143 (dynactin subunit 1)
614603	DDHD1 / HGNC:19714 / OMIM:614603 (DDHD domain containing 1)
615003	DDHD2 / HGNC:29106 / OMIM:615003 (DDHD domain containing 2)
125660	DES / HGNC:2770 / OMIM:125660 (desmin)
606983	DGAT2 / HGNC:16940 / OMIM:606983 (diacylglycerol O-acyltransferase 2)
601465	DGUOK / HGNC:2858 / OMIM:601465 (deoxyguanosine kinase)
614984	DHTKD1 / HGNC:23537 / OMIM:614984 (dehydrogenase E1 and transketolase domain containing 1)
300377	DMD / HGNC:2928 / OMIM:300377 (dystrophin)
605377	DMPK / HGNC:2933 / OMIM:605377 (DM1 protein kinase)
601810	DNA2 / HGNC:2939 / OMIM:601810 (DNA replication helicase/nuclease 2)
604139	DNAJB2 / HGNC:5228 / OMIM:604139 (DnaJ heat shock protein family (Hsp40) member B2)
611332	DNAJB6 / HGNC:14888 / OMIM:611332 (DnaJ heat shock protein family (Hsp40) member B6)
602378	DNM2 / HGNC:2974 / OMIM:602378 (dynamin 2)
126375	DNMT1 / HGNC:2976 / OMIM:126375 (DNA methyltransferase 1)
610285	DOK7 / HGNC:26594 / OMIM:610285 (docking protein 7)
610746	DOLK / HGNC:23406 / OMIM:610746 (dolichol kinase)
191350	DPAGT1 / HGNC:2995 / OMIM:191350 (dolichyl-phosphate N-acetylglucosaminephosphotransferase 1)
603503	DPM1 / HGNC:3005 / OMIM:603503 (dolichyl-phosphate mannosyltransferase subunit 1, catalytic)
603564	DPM2 / HGNC:3006 / OMIM:603564 (dolichyl-phosphate mannosyltransferase subunit 2, regulatory)
605951	DPM3 / HGNC:3007 / OMIM:605951 (dolichyl-phosphate mannosyltransferase subunit 3, regulatory)
125645	DSC2 / HGNC:3036 / OMIM:125645 (desmocollin 2)
125671	DSG2 / HGNC:3049 / OMIM:125671 (desmoglein 2)
125647	DSP / HGNC:3052 / OMIM:125647 (desmoplakin)
113810	DST / HGNC:1090 / OMIM:113810 (dystonin)
601239	DTNA / HGNC:3057 / OMIM:601239 (dystrobrevin alpha)
606009	DUX4L1 / HGNC:3082 / OMIM:606009 (double homeobox 4 like 1 (pseudogene))
600112	DYNC1H1 / HGNC:2961 / OMIM:600112 (dynein cytoplasmic 1 heavy chain 1)

603009	DYSF / HGNC:3097 / OMIM:603009 (dysferlin)
130610	EEF2 / HGNC:3214 / OMIM:130610 (eukaryotic translation elongation factor 2)
129010	EGR2 / HGNC:3239 / OMIM:129010 (early growth response 2)
605512	ELOVL4 / HGNC:14415 / OMIM:605512 (ELOVL fatty acid elongase 4)
611805	ELOVL5 / HGNC:21308 / OMIM:611805 (ELOVL fatty acid elongase 5)
603722	ELP1 / HGNC:5959 / OMIM:603722 (elongator acetyltransferase complex subunit 1)
300384	EMD / HGNC:3331 / OMIM:300384 (emerin)
131370	ENO3 / HGNC:3354 / OMIM:131370 (enolase 3)
601752	ENTPD1 / HGNC:3363 / OMIM:601752 (ectonucleoside triphosphate diphosphohydrolase 1)
190151	ERBB3 / HGNC:3431 / OMIM:190151 (erb-b2 receptor tyrosine kinase 3)
600543	ERBB4 / HGNC:3432 / OMIM:600543 (erb-b2 receptor tyrosine kinase 4)
611604	ERLIN1 / HGNC:16947 / OMIM:611604 (ER lipid raft associated 1)
611605	ERLIN2 / HGNC:1356 / OMIM:611605 (ER lipid raft associated 2)
608053	ETFA / HGNC:3481 / OMIM:608053 (electron transfer flavoprotein subunit alpha)
231675	ETFDH / HGNC:3483 / OMIM:231675 (electron transfer flavoprotein dehydrogenase)
606489	EXOSC3 / HGNC:17944 / OMIM:606489 (exosome component 3)
606019	EXOSC8 / HGNC:17035 / OMIM:606019 (exosome component 8)
603550	EYA4 / HGNC:3522 / OMIM:603550 (EYA transcriptional coactivator and phosphatase 4)
611026	FA2H / HGNC:21197 / OMIM:611026 (fatty acid 2-hydroxylase)
615584	FAM111B / HGNC:24200 / OMIM:615584 (FAM111 trypsin like peptidase B)
611592	FARS2 / HGNC:21062 / OMIM:611592 (phenylalanyl-tRNA synthetase 2, mitochondrial)
612322	FASTKD2 / HGNC:29160 / OMIM:612322 (FAST kinase domains 2)
604269	FAT2 / HGNC:3596 / OMIM:604269 (FAT atypical cadherin 2)
604580	FBLN5 / HGNC:3602 / OMIM:604580 (fibulin 5)
605654	FBXL4 / HGNC:13601 / OMIM:605654 (F-box and leucine rich repeat protein 4)
608533	FBXO38 / HGNC:28844 / OMIM:608533 (F-box protein 38)
614585	FDX2 / HGNC:30546 / OMIM:614585 (ferredoxin 2)
611104	FGD4 / HGNC:19125 / OMIM:611104 (FYVE, RhoGEF and PH domain containing 4)
601515	FGF14 / HGNC:3671 / OMIM:601515 (fibroblast growth factor 14)
300163	FHL1 / HGNC:3702 / OMIM:300163 (four and a half LIM domains 1)
609390	FIG4 / HGNC:16873 / OMIM:609390 (FIG4 phosphoinositide 5-phosphatase)
606596	FKRP / HGNC:17997 / OMIM:606596 (fukutin related protein)
607440	FKTN / HGNC:3622 / OMIM:607440 (fukutin)
610595	FLAD1 / HGNC:24671 / OMIM:610595 (flavin adenine dinucleotide synthetase 1)
300017	FLNA / HGNC:3754 / OMIM:300017 (filamin A)
102565	FLNC / HGNC:3756 / OMIM:102565 (filamin C)
609144	FLVCR1 / HGNC:24682 / OMIM:609144 (FLVCR heme transporter 1)
601278	FRG1 / HGNC:3954 / OMIM:601278 (FSHD region gene 1)
137070	FUS / HGNC:4010 / OMIM:137070 (FUS RNA binding protein)
606829	FXN / HGNC:3951 / OMIM:606829 (frataxin)
600819	FXR1 / HGNC:4023 / OMIM:600819 (FMR1 autosomal homolog 1)
606800	GAA / HGNC:4065 / OMIM:606800 (alpha glucosidase)
605379	GAN / HGNC:4137 / OMIM:605379 (gigaxonin)
600287	GARS1 / HGNC:4162 / OMIM:600287 (glycyl-tRNA synthetase 1)
614518	GATAD1 / HGNC:29941 / OMIM:614518 (GATA zinc finger domain containing 1)
606463	GBA / HGNC:4177 / OMIM:606463 (glucosylceramidase beta)
609471	GBA2 / HGNC:18986 / OMIM:609471 (glucosylceramidase beta 2)

607839	GBE1 / HGNC:4180 / OMIM:607839 (1,4-alpha-glucan branching enzyme 1)
603698	GBF1 / HGNC:4181 / OMIM:603698 (golgi brefeldin A resistant guanine nucleotide exchange factor 1)
606598	GDAP1 / HGNC:15968 / OMIM:606598 (ganglioside induced differentiation associated protein 1)
618128	GDAP2 / HGNC:18010 / OMIM:618128 (ganglioside induced differentiation associated protein 2)
138292	GFPT1 / HGNC:4241 / OMIM:138292 (glutamine--fructose-6-phosphate transaminase 1)
605072	GIPC1 / HGNC:1226 / OMIM:605072 (GIPC PDZ domain containing family member 1)
121013	GJA5 / HGNC:4279 / OMIM:121013 (gap junction protein alpha 5)
304040	GJB1 / HGNC:4283 / OMIM:304040 (gap junction protein beta 1)
603324	GJB3 / HGNC:4285 / OMIM:603324 (gap junction protein beta 3)
608803	GJC2 / HGNC:17494 / OMIM:608803 (gap junction protein gamma 2)
608603	GLDN / HGNC:29514 / OMIM:608603 (gliomedin)
603371	GLE1 / HGNC:4315 / OMIM:603371 (GLE1 RNA export mediator)
615320	GMPPB / HGNC:22932 / OMIM:615320 (GDP-mannose pyrophosphorylase B)
610863	GNB4 / HGNC:20731 / OMIM:610863 (G protein subunit beta 4)
603824	GNE / HGNC:23657 / OMIM:603824 (glucosamine (UDP-N-acetyl)-2-epimerase/N-acetylmannosamine kinase)
602580	GOLGA2 / HGNC:4425 / OMIM:602580 (golgin A2)
604027	GOSR2 / HGNC:4431 / OMIM:604027 (golgi SNAP receptor complex member 2)
611778	GPD1L / HGNC:28956 / OMIM:611778 (glycerol-3-phosphate dehydrogenase 1 like)
602368	GRID2 / HGNC:4576 / OMIM:602368 (glutamate ionotropic receptor delta type subunit 2)
604473	GRM1 / HGNC:4593 / OMIM:604473 (glutamate metabotropic receptor 1)
603942	GYG1 / HGNC:4699 / OMIM:603942 (glycogenin 1)
138570	GYS1 / HGNC:4706 / OMIM:138570 (glycogen synthase 1)
610467	HACD1 / HGNC:9639 / OMIM:610467 (3-hydroxyacyl-CoA dehydratase 1)
610876	HACE1 / HGNC:21033 / OMIM:610876 (HECT domain and ankyrin repeat containing E3 ubiquitin protein ligase 1)
142810	HARS1 / HGNC:4816 / OMIM:142810 (histidyl-tRNA synthetase 1)
605206	HCN4 / HGNC:16882 / OMIM:605206 (hyperpolarization activated cyclic nucleotide gated potassium channel 4)
606873	HEXB / HGNC:4879 / OMIM:606873 (hexosaminidase subunit beta)
601314	HINT1 / HGNC:4912 / OMIM:601314 (histidine triad nucleotide binding protein 1)
142600	HK1 / HGNC:4922 / OMIM:142600 (hexokinase 1)
164017	HNRNPA1 / HGNC:5031 / OMIM:164017 (heterogeneous nuclear ribonucleoprotein A1)
600124	HNRNPA2B1 / HGNC:5033 / OMIM:600124 (heterogeneous nuclear ribonucleoprotein A2/B1)
607137	HNRNPDL / HGNC:5037 / OMIM:607137 (heterogeneous nuclear ribonucleoprotein D like)
142984	HOXD10 / HGNC:5133 / OMIM:142984 (homeobox D10)
190020	HRAS / HGNC:5173 / OMIM:190020 (HRas proto-oncogene, GTPase)
602195	HSPB1 / HGNC:5246 / OMIM:602195 (heat shock protein family B (small) member 1)
604624	HSPB3 / HGNC:5248 / OMIM:604624 (heat shock protein family B (small) member 3)
608014	HSPB8 / HGNC:30171 / OMIM:608014 (heat shock protein family B (small) member 8)
118190	HSPD1 / HGNC:5261 / OMIM:118190 (heat shock protein family D (Hsp60) member 1)
142461	HSPG2 / HGNC:5273 / OMIM:142461 (heparan sulfate proteoglycan 2)
615316	IBA57 / HGNC:27302 / OMIM:615316 (iron-sulfur cluster assembly factor IBA57)
603502	IFRD1 / HGNC:5456 / OMIM:603502 (interferon related developmental regulator 1)
600502	IGHMBP2 / HGNC:5542 / OMIM:600502 (immunoglobulin mu DNA binding protein 2)
602366	ILK / HGNC:6040 / OMIM:602366 (integrin linked kinase)

610982	INF2 / HGNC:23791 / OMIM:610982 (inverted formin 2)
607875	INPP5K / HGNC:33882 / OMIM:607875 (inositol polyphosphate-5-phosphatase K)
611911	ISCU / HGNC:29882 / OMIM:611911 (iron-sulfur cluster assembly enzyme)
600536	ITGA7 / HGNC:6143 / OMIM:600536 (integrin subunit alpha 7)
147265	ITPR1 / HGNC:6180 / OMIM:147265 (inositol 1,4,5-trisphosphate receptor type 1)
147267	ITPR3 / HGNC:6182 / OMIM:147267 (inositol 1,4,5-trisphosphate receptor type 3)
601920	JAG1 / HGNC:6188 / OMIM:601920 (jagged canonical Notch ligand 1)
605267	JPH2 / HGNC:14202 / OMIM:605267 (junctophilin 2)
173325	JUP / HGNC:6207 / OMIM:173325 (junction plakoglobin)
601421	KARS1 / HGNC:6215 / OMIM:601421 (lysyl-tRNA synthetase 1)
613727	KBTBD13 / HGNC:37227 / OMIM:613727 (kelch repeat and BTB domain containing 13)
176260	KCNA1 / HGNC:6218 / OMIM:176260 (potassium voltage-gated channel subfamily A member 1)
176267	KCNA5 / HGNC:6224 / OMIM:176267 (potassium voltage-gated channel subfamily A member 5)
176264	KCNC3 / HGNC:6235 / OMIM:176264 (potassium voltage-gated channel subfamily C member 3)
605411	KCND3 / HGNC:6239 / OMIM:605411 (potassium voltage-gated channel subfamily D member 3)
176261	KCNE1 / HGNC:6240 / OMIM:176261 (potassium voltage-gated channel subfamily E regulatory subunit 1)
603796	KCNE2 / HGNC:6242 / OMIM:603796 (potassium voltage-gated channel subfamily E regulatory subunit 2)
604433	KCNE3 / HGNC:6243 / OMIM:604433 (potassium voltage-gated channel subfamily E regulatory subunit 3)
152427	KCNH2 / HGNC:6251 / OMIM:152427 (potassium voltage-gated channel subfamily H member 2)
602323	KCNJ12 / HGNC:6258 / OMIM:602323 (potassium inwardly rectifying channel subfamily J member 12)
600681	KCNJ2 / HGNC:6263 / OMIM:600681 (potassium inwardly rectifying channel subfamily J member 2)
600734	KCNJ5 / HGNC:6266 / OMIM:600734 (potassium inwardly rectifying channel subfamily J member 5)
607542	KCNQ1 / HGNC:6294 / OMIM:607542 (potassium voltage-gated channel subfamily Q member 1)
610657	WASHC5 / HGNC:28984 / OMIM:610657 (WASH complex subunit 5)
615759	KIDINS220 / HGNC:29508 / OMIM:615759 (kinase D interacting substrate 220)
601255	KIF1A / HGNC:888 / OMIM:601255 (kinesin family member 1A)
605995	KIF1B / HGNC:16636 / OMIM:605995 (kinesin family member 1B)
603060	KIF1C / HGNC:6317 / OMIM:603060 (kinesin family member 1C)
608283	KIF21A / HGNC:19349 / OMIM:608283 (kinesin family member 21A)
614026	KIF26B / HGNC:25484 / OMIM:614026 (kinesin family member 26B)
602821	KIF5A / HGNC:6323 / OMIM:602821 (kinesin family member 5A)
611729	KLC2 / HGNC:20716 / OMIM:611729 (kinesin light chain 2)
615340	KLHL40 / HGNC:30372 / OMIM:615340 (kelch like family member 40)
607701	KLHL41 / HGNC:16905 / OMIM:607701 (kelch like family member 41)
611201	KLHL9 / HGNC:18732 / OMIM:611201 (kelch like family member 9)
605739	KY / HGNC:26576 / OMIM:605739 (kyphoscoliosis peptidase)
308840	L1CAM / HGNC:6470 / OMIM:308840 (L1 cell adhesion molecule)
156225	LAMA2 / HGNC:6482 / OMIM:156225 (laminin subunit alpha 2)
600133	LAMA4 / HGNC:6484 / OMIM:600133 (laminin subunit alpha 4)

601033	LAMA5 / HGNC:6485 / OMIM:601033 (laminin subunit alpha 5)
150325	LAMB2 / HGNC:6487 / OMIM:150325 (laminin subunit beta 2)
309060	LAMP2 / HGNC:6501 / OMIM:309060 (lysosomal associated membrane protein 2)
603590	LARGE1 / HGNC:6511 / OMIM:603590 (LARGE xylosyl- and glucuronyltransferase 1)
605906	LDB3 / HGNC:15710 / OMIM:605906 (LIM domain binding 3)
150000	LDHA / HGNC:6535 / OMIM:150000 (lactate dehydrogenase A)
607908	LIMS2 / HGNC:16084 / OMIM:607908 (LIM zinc finger domain containing 2)
603795	LITAF / HGNC:16841 / OMIM:603795 (lipopolysaccharide induced TNF factor)
150330	LMNA / HGNC:6636 / OMIM:150330 (lamin A/C)
616112	LMOD3 / HGNC:6649 / OMIM:616112 (leiomodin 3)
605518	LPIN1 / HGNC:13345 / OMIM:605518 (lipin 1)
618299	LRP12 / HGNC:31708 / OMIM:618299 (LDL receptor related protein 12)
604270	LRP4 / HGNC:6696 / OMIM:604270 (LDL receptor related protein 4)
610933	LRSAM1 / HGNC:25135 / OMIM:610933 (leucine rich repeat and sterile alpha motif containing 1)
159460	MAG / HGNC:6783 / OMIM:159460 (myelin associated glycoprotein)
609479	MAP3K20 / HGNC:17797 / OMIM:609479 (mitogen-activated protein kinase kinase kinase 20)
157140	MAPT / HGNC:6893 / OMIM:157140 (microtubule associated protein tau)
156560	MARS1 / HGNC:6898 / OMIM:156560 (methionyl-tRNA synthetase 1)
609728	MARS2 / HGNC:25133 / OMIM:609728 (methionyl-tRNA synthetase 2, mitochondrial)
164015	MATR3 / HGNC:6912 / OMIM:164015 (matrin 3)
160000	MB / HGNC:6915 / OMIM:160000 (myoglobin)
603294	MCM3AP / HGNC:6946 / OMIM:603294 (minichromosome maintenance complex component 3 associated protein)
612453	MEGF10 / HGNC:29634 / OMIM:612453 (multiple EGF like domains 10)
608507	MFN2 / HGNC:16877 / OMIM:608507 (mitofusin 2)
615076	MGME1 / HGNC:16205 / OMIM:615076 (mitochondrial genome maintenance exonuclease 1)
608677	MIB1 / HGNC:21086 / OMIM:608677 (MIB E3 ubiquitin protein ligase 1)
120520	MME / HGNC:7154 / OMIM:120520 (membrane metalloendopeptidase)
616661	MORC2 / HGNC:23573 / OMIM:616661 (MORC family CW-type zinc finger 2)
604041	MPDU1 / HGNC:7207 / OMIM:604041 (mannose-P-dolichol utilization defect 1)
137960	MPV17 / HGNC:7224 / OMIM:137960 (mitochondrial inner membrane protein MPV17)
159440	MPZ / HGNC:7225 / OMIM:159440 (myelin protein zero)
600814	MRE11 / HGNC:7230 / OMIM:600814 (MRE11 homolog, double strand break repair nuclease)
607118	MRPL3 / HGNC:10379 / OMIM:607118 (mitochondrial ribosomal protein L3)
611849	MRPL44 / HGNC:16650 / OMIM:611849 (mitochondrial ribosomal protein L44)
611987	MRPS25 / HGNC:14511 / OMIM:611987 (mitochondrial ribosomal protein S25)
601788	MSTN / HGNC:4223 / OMIM:601788 (myostatin)
617619	MSTO1 / HGNC:29678 / OMIM:617619 (misato mitochondrial distribution and morphology regulator 1)
590065	MT-TM / HGNC:7492 / OMIM:590065 (mitochondrially encoded tRNA-Met (AUA/G))
590090	MT-TT / HGNC:7499 / OMIM:590090 (mitochondrially encoded tRNA-Thr (ACN))
300415	MTM1 / HGNC:7448 / OMIM:300415 (myotubularin 1)
603557	MTMR2 / HGNC:7450 / OMIM:603557 (myotubularin related protein 2)
614667	MTO1 / HGNC:19261 / OMIM:614667 (mitochondrial tRNA translation optimization 1)
613669	MTPAP / HGNC:25532 / OMIM:613669 (mitochondrial poly(A) polymerase)
601296	MUSK / HGNC:7525 / OMIM:601296 (muscle associated receptor tyrosine kinase)
160794	MYBPC1 / HGNC:7549 / OMIM:160794 (myosin binding protein C1)

600958	MYBPC3 / HGNC:7551 / OMIM:600958 (myosin binding protein C3)
608568	MYH14 / HGNC:23212 / OMIM:608568 (myosin heavy chain 14)
160740	MYH2 / HGNC:7572 / OMIM:160740 (myosin heavy chain 2)
160720	MYH3 / HGNC:7573 / OMIM:160720 (myosin heavy chain 3)
160710	MYH6 / HGNC:7576 / OMIM:160710 (myosin heavy chain 6)
160760	MYH7 / HGNC:7577 / OMIM:160760 (myosin heavy chain 7)
160741	MYH8 / HGNC:7578 / OMIM:160741 (myosin heavy chain 8)
160780	MYL1 / HGNC:7582 / OMIM:160780 (myosin light chain 1)
160781	MYL2 / HGNC:7583 / OMIM:160781 (myosin light chain 2)
160790	MYL3 / HGNC:7584 / OMIM:160790 (myosin light chain 3)
160770	MYL4 / HGNC:7585 / OMIM:160770 (myosin light chain 4)
606566	MYLK2 / HGNC:16243 / OMIM:606566 (myosin light chain kinase 2)
615345	MYMK / HGNC:33778 / OMIM:615345 (myomaker, myoblast fusion factor)
607295	MYO18B / HGNC:18150 / OMIM:607295 (myosin XVIIIB)
604875	MYO9A / HGNC:7608 / OMIM:604875 (myosin IXA)
604103	MYOT / HGNC:12399 / OMIM:604103 (myotilin)
605602	MYOZ2 / HGNC:1330 / OMIM:605602 (myozenin 2)
608517	MYPN / HGNC:23246 / OMIM:608517 (myopalladin)
609701	NAGLU / HGNC:7632 / OMIM:609701 (N-acetyl-alpha-glucosaminidase)
612803	NARS2 / HGNC:26274 / OMIM:612803 (asparaginyl-tRNA synthetase 2, mitochondrial)
605262	NDRG1 / HGNC:7679 / OMIM:605262 (N-myc downstream regulated 1)
606934	NDUFAF1 / HGNC:18828 / OMIM:606934 (NADH:ubiquinone oxidoreductase complex assembly factor 1)
161650	NEB / HGNC:7720 / OMIM:161650 (nebulin)
162230	NEFH / HGNC:7737 / OMIM:162230 (neurofilament heavy chain)
162280	NEFL / HGNC:7739 / OMIM:162280 (neurofilament light chain)
604588	NEK1 / HGNC:7744 / OMIM:604588 (NIMA related kinase 1)
609798	NEK9 / HGNC:18591 / OMIM:609798 (NIMA related kinase 9)
613121	NEXN / HGNC:29557 / OMIM:613121 (nexilin F-actin binding protein)
162030	NGF / HGNC:7808 / OMIM:162030 (nerve growth factor)
608145	NIPA1 / HGNC:17043 / OMIM:608145 (NIPA magnesium transporter 1)
605955	NKX6-2 / HGNC:19321 / OMIM:605955 (NK6 homeobox 2)
608701	NMNAT2 / HGNC:16789 / OMIM:608701 (nicotinamide nucleotide adenylyltransferase 2)
614154	NOP56 / HGNC:15911 / OMIM:614154 (NOP56 ribonucleoprotein)
108780	NPPA / HGNC:7939 / OMIM:108780 (natriuretic peptide A)
600417	NT5C2 / HGNC:8022 / OMIM:600417 (5'-nucleotidase, cytosolic II)
191315	NTRK1 / HGNC:8031 / OMIM:191315 (neurotrophic receptor tyrosine kinase 1)
606694	NUP155 / HGNC:8063 / OMIM:606694 (nucleoporin 155)
602552	NUP88 / HGNC:8067 / OMIM:602552 (nucleoporin 88)
605290	OPA1 / HGNC:8140 / OMIM:605290 (OPA1 mitochondrial dynamin like GTPase)
602432	OPTN / HGNC:17142 / OMIM:602432 (optineurin)
610277	ORAI1 / HGNC:25896 / OMIM:610277 (ORAI calcium release-activated calcium modulator 1)
602279	PABPN1 / HGNC:8565 / OMIM:602279 (poly(A) binding protein nuclear 1)
167410	PAX7 / HGNC:8621 / OMIM:167410 (paired box 7)
176740	PCNA / HGNC:8729 / OMIM:176740 (proliferating cell nuclear antigen)
300906	PDK3 / HGNC:8811 / OMIM:300906 (pyruvate dehydrogenase kinase 3)
131340	PDYN / HGNC:8820 / OMIM:131340 (prodynorphin)

601757	PEX7 / HGNC:8860 / OMIM:601757 (peroxisomal biogenesis factor 7)
610681	PFKM / HGNC:8877 / OMIM:610681 (phosphofructokinase, muscle)
176610	PFN1 / HGNC:8881 / OMIM:176610 (profilin 1)
612931	PGAM2 / HGNC:8889 / OMIM:612931 (phosphoglycerate mutase 2)
311800	PGK1 / HGNC:8896 / OMIM:311800 (phosphoglycerate kinase 1)
171900	PGM1 / HGNC:8905 / OMIM:171900 (phosphoglucomutase 1)
311870	PHKA1 / HGNC:8925 / OMIM:311870 (phosphorylase kinase regulatory subunit alpha 1)
602753	PHOX2A / HGNC:691 / OMIM:602753 (paired like homeobox 2A)
602026	PHYH / HGNC:8940 / OMIM:602026 (phytanoyl-CoA 2-hydroxylase)
613629	PIEZO2 / HGNC:26270 / OMIM:613629 (piezo type mechanosensitive ion channel component 2)
606102	PIP5K1C / HGNC:8996 / OMIM:606102 (phosphatidylinositol-4-phosphate 5-kinase type 1 gamma)
602861	PKP2 / HGNC:9024 / OMIM:602861 (plakophilin 2)
615698	PLD3 / HGNC:17158 / OMIM:615698 (phospholipase D family member 3)
601282	PLEC / HGNC:9069 / OMIM:601282 (plectin)
611101	PLEKHG5 / HGNC:29105 / OMIM:611101 (pleckstrin homology and RhoGEF domain containing G5)
172405	PLN / HGNC:9080 / OMIM:172405 (phospholamban)
300401	PLP1 / HGNC:9086 / OMIM:300401 (proteolipid protein 1)
170715	PMP2 / HGNC:9117 / OMIM:170715 (peripheral myelin protein 2)
601097	PMP22 / HGNC:9118 / OMIM:601097 (peripheral myelin protein 22)
605610	PNKP / HGNC:9154 / OMIM:605610 (polynucleotide kinase 3'-phosphatase)
609059	PNPLA2 / HGNC:30802 / OMIM:609059 (patatin like phospholipase domain containing 2)
603197	PNPLA6 / HGNC:16268 / OMIM:603197 (patatin like phospholipase domain containing 6)
612123	PNPLA8 / HGNC:28900 / OMIM:612123 (patatin like phospholipase domain containing 8)
615618	POGLUT1 / HGNC:22954 / OMIM:615618 (protein O-glucosyltransferase 1)
174763	POLG / HGNC:9179 / OMIM:174763 (DNA polymerase gamma, catalytic subunit)
604983	POLG2 / HGNC:9180 / OMIM:604983 (DNA polymerase gamma 2, accessory subunit)
606822	POMGNT1 / HGNC:19139 / OMIM:606822 (protein O-linked mannose N-acetylglucosaminyltransferase 1 (beta 1,2-))
614828	POMGNT2 / HGNC:25902 / OMIM:614828 (protein O-linked mannose N-acetylglucosaminyltransferase 2 (beta 1,4-))
615247	POMK / HGNC:26267 / OMIM:615247 (protein O-mannose kinase)
607423	POMT1 / HGNC:9202 / OMIM:607423 (protein O-mannosyltransferase 1)
607439	POMT2 / HGNC:19743 / OMIM:607439 (protein O-mannosyltransferase 2)
605824	POPDC3 / HGNC:17649 / OMIM:605824 (popeye domain containing 3)
604325	PPP2R2B / HGNC:9305 / OMIM:604325 (protein phosphatase 2 regulatory subunit Bbeta)
616458	PRDM12 / HGNC:13997 / OMIM:616458 (PR/SET domain 12)
605557	PRDM16 / HGNC:14000 / OMIM:605557 (PR/SET domain 16)
609557	PREPL / HGNC:30228 / OMIM:609557 (prolyl endopeptidase like)
602743	PRKAG2 / HGNC:9386 / OMIM:602743 (protein kinase AMP-activated non-catalytic subunit gamma 2)
176980	PRKCG / HGNC:9402 / OMIM:176980 (protein kinase C gamma)
170710	PRPH / HGNC:9461 / OMIM:170710 (peripherin)
311850	PRPS1 / HGNC:9462 / OMIM:311850 (phosphoribosyl pyrophosphate synthetase 1)
617413	PRUNE1 / HGNC:13420 / OMIM:617413 (prune exopolyphosphatase 1)
605725	PRX / HGNC:13797 / OMIM:605725 (periaxin)
104311	PSEN1 / HGNC:9508 / OMIM:104311 (presenilin 1)

600759	PSEN2 / HGNC:9509 / OMIM:600759 (presenilin 2)
608625	PTRH2 / HGNC:24265 / OMIM:608625 (peptidyl-tRNA hydrolase 2)
607204	PUM1 / HGNC:14957 / OMIM:607204 (pumilio RNA binding family member 1)
600473	PURA / HGNC:9701 / OMIM:600473 (purine rich element binding protein A)
608109	PUS1 / HGNC:15508 / OMIM:608109 (pseudouridine synthase 1)
608455	PYGM / HGNC:9726 / OMIM:608455 (glycogen phosphorylase, muscle associated)
617220	PYROXD1 / HGNC:26162 / OMIM:617220 (pyridine nucleotide-disulphide oxidoreductase domain 1)
602298	RAB7A / HGNC:9788 / OMIM:602298 (RAB7A, member RAS oncogene family)
164760	RAF1 / HGNC:9829 / OMIM:164760 (Raf-1 proto-oncogene, serine/threonine kinase)
601592	RAPSN / HGNC:9863 / OMIM:601592 (receptor associated protein of the synapse)
610924	RBCK1 / HGNC:15864 / OMIM:610924 (RANBP2-type and C3HC4-type zinc finger containing 1)
613171	RBM20 / HGNC:27424 / OMIM:613171 (RNA binding motif protein 20)
612413	RBM7 / HGNC:9904 / OMIM:612413 (RNA binding motif protein 7)
609139	REEP1 / HGNC:25786 / OMIM:609139 (receptor accessory protein 1)
609347	REEP2 / HGNC:17975 / OMIM:609347 (receptor accessory protein 2)
613114	RETREG1 / HGNC:25964 / OMIM:613114 (reticulophagy regulator 1)
102579	RFC1 / HGNC:9969 / OMIM:102579 (replication factor C subunit 1)
604123	RNASEH1 / HGNC:18466 / OMIM:604123 (ribonuclease H1)
609948	RNF216 / HGNC:21698 / OMIM:609948 (ring finger protein 216)
612159	RPH3A / HGNC:17056 / OMIM:612159 (rabphilin 3A)
604712	RRM2B / HGNC:17296 / OMIM:604712 (ribonucleotide reductase regulatory TP53 inducible subunit M2B)
603183	RTN2 / HGNC:10468 / OMIM:603183 (reticulon 2)
613516	RUBCN / HGNC:28991 / OMIM:613516 (rubicon autophagy regulator)
605862	RXYLT1 / HGNC:13530 / OMIM:605862 (ribitol xylosyltransferase 1)
180901	RYR1 / HGNC:10483 / OMIM:180901 (ryanodine receptor 1)
180902	RYR2 / HGNC:10484 / OMIM:180902 (ryanodine receptor 2)
180903	RYR3 / HGNC:10485 / OMIM:180903 (ryanodine receptor 3)
604490	SACS / HGNC:10519 / OMIM:604490 (sacsin molecular chaperone)
603560	SBF1 / HGNC:10542 / OMIM:603560 (SET binding factor 1)
607697	SBF2 / HGNC:2135 / OMIM:607697 (SET binding factor 2)
604385	SCN11A / HGNC:10583 / OMIM:604385 (sodium voltage-gated channel alpha subunit 11)
600235	SCN1B / HGNC:10586 / OMIM:600235 (sodium voltage-gated channel beta subunit 1)
601327	SCN2B / HGNC:10589 / OMIM:601327 (sodium voltage-gated channel beta subunit 2)
608214	SCN3B / HGNC:20665 / OMIM:608214 (sodium voltage-gated channel beta subunit 3)
603967	SCN4A / HGNC:10591 / OMIM:603967 (sodium voltage-gated channel alpha subunit 4)
608256	SCN4B / HGNC:10592 / OMIM:608256 (sodium voltage-gated channel beta subunit 4)
600163	SCN5A / HGNC:10593 / OMIM:600163 (sodium voltage-gated channel alpha subunit 5)
603415	SCN9A / HGNC:10597 / OMIM:603415 (sodium voltage-gated channel alpha subunit 9)
604272	SCO2 / HGNC:10604 / OMIM:604272 (synthesis of cytochrome C oxidase 2)
607982	SCYL1 / HGNC:14372 / OMIM:607982 (SCY1 like pseudokinase 1)
600857	SDHA / HGNC:10680 / OMIM:600857 (succinate dehydrogenase complex flavoprotein subunit A)
606210	SELENON / HGNC:15999 / OMIM:606210 (selenoprotein N)
604061	SEPTIN9 / HGNC:7323 / OMIM:604061 (septin 9)
608465	SETX / HGNC:445 / OMIM:608465 (senataxin)
600119	SGCA / HGNC:10805 / OMIM:600119 (sarcoglycan alpha)

600900	SGCB / HGNC:10806 / OMIM:600900 (sarcoglycan beta)
601411	SGCD / HGNC:10807 / OMIM:601411 (sarcoglycan delta)
608896	SGCG / HGNC:10809 / OMIM:608896 (sarcoglycan gamma)
603729	SGPL1 / HGNC:10817 / OMIM:603729 (sphingosine-1-phosphate lyase 1)
608206	SH3TC2 / HGNC:29427 / OMIM:608206 (SH3 domain and tetratricopeptide repeats 2)
601978	SIGMAR1 / HGNC:8157 / OMIM:601978 (sigma non-opioid intracellular receptor 1)
608005	SIL1 / HGNC:24624 / OMIM:608005 (SIL1 nucleotide exchange factor)
604878	SLC12A6 / HGNC:10914 / OMIM:604878 (solute carrier family 12 member 6)
600682	SLC16A1 / HGNC:10922 / OMIM:600682 (solute carrier family 16 member 1)
600336	SLC18A3 / HGNC:10936 / OMIM:600336 (solute carrier family 18 member A3)
600111	SLC1A3 / HGNC:10941 / OMIM:600111 (solute carrier family 1 member 3)
603377	SLC22A5 / HGNC:10969 / OMIM:603377 (solute carrier family 22 member 5)
190315	SLC25A1 / HGNC:10979 / OMIM:190315 (solute carrier family 25 member 1)
606521	SLC25A19 / HGNC:14409 / OMIM:606521 (solute carrier family 25 member 19)
613698	SLC25A20 / HGNC:1421 / OMIM:613698 (solute carrier family 25 member 20)
103220	SLC25A4 / HGNC:10990 / OMIM:103220 (solute carrier family 25 member 4)
610823	SLC25A42 / HGNC:28380 / OMIM:610823 (solute carrier family 25 member 42)
610826	SLC25A46 / HGNC:25198 / OMIM:610826 (solute carrier family 25 member 46)
603690	SLC33A1 / HGNC:95 / OMIM:603690 (solute carrier family 33 member 1)
607882	SLC52A2 / HGNC:30224 / OMIM:607882 (solute carrier family 52 member 2)
613350	SLC52A3 / HGNC:16187 / OMIM:613350 (solute carrier family 52 member 3)
608761	SLC5A7 / HGNC:14025 / OMIM:608761 (solute carrier family 5 member 7)
107310	SLC9A1 / HGNC:11071 / OMIM:107310 (solute carrier family 9 member A1)
604990	SLC9A3R1 / HGNC:11075 / OMIM:604990 (SLC9A3 regulator 1)
614982	SMCHD1 / HGNC:29090 / OMIM:614982 (structural maintenance of chromosomes flexible hinge domain containing 1)
600354	SMN1 / HGNC:11117 / OMIM:600354 (survival of motor neuron 1, telomeric)
601627	SMN2 / HGNC:11118 / OMIM:601627 (survival of motor neuron 2, centromeric)
610457	SMPD4 / HGNC:32949 / OMIM:610457 (sphingomyelin phosphodiesterase 4)
600322	SNAP25 / HGNC:11132 / OMIM:600322 (synaptosome associated protein 25)
601017	SNTA1 / HGNC:11167 / OMIM:601017 (syntrophin alpha 1)
616105	SNX14 / HGNC:14977 / OMIM:616105 (sorting nexin 14)
147450	SOD1 / HGNC:11179 / OMIM:147450 (superoxide dismutase 1)
182500	SORD / HGNC:11184 / OMIM:182500 (sorbitol dehydrogenase)
607111	SPART / HGNC:18514 / OMIM:607111 (spartin)
604277	SPAST / HGNC:11233 / OMIM:604277 (spastin)
615950	SPEG / HGNC:16901 / OMIM:615950 (striated muscle enriched protein kinase)
610844	SPG11 / HGNC:11226 / OMIM:610844 (SPG11 vesicle trafficking associated, spatacsin)
608181	SPG21 / HGNC:20373 / OMIM:608181 (SPG21 abhydrolase domain containing, maspardin)
602783	SPG7 / HGNC:11237 / OMIM:602783 (SPG7 matrix AAA peptidase subunit, paraplegin)
182810	SPTAN1 / HGNC:11273 / OMIM:182810 (spectrin alpha, non-erythrocytic 1)
604985	SPTBN2 / HGNC:11276 / OMIM:604985 (spectrin beta, non-erythrocytic 2)
606214	SPTBN4 / HGNC:14896 / OMIM:606214 (spectrin beta, non-erythrocytic 4)
605712	SPTLC1 / HGNC:11277 / OMIM:605712 (serine palmitoyltransferase long chain base subunit 1)
605713	SPTLC2 / HGNC:11278 / OMIM:605713 (serine palmitoyltransferase long chain base sub 2)
601530	SQSTM1 / HGNC:11280 / OMIM:601530 (sequestosome 1)

615521	STAC3 / HGNC:28423 / OMIM:615521 (SH3 and cysteine rich domain 3)
605921	STIM1 / HGNC:11386 / OMIM:605921 (stromal interaction molecule 1)
607207	STUB1 / HGNC:11427 / OMIM:607207 (STIP1 homology and U-box containing protein 1)
603921	SUCLA2 / HGNC:11448 / OMIM:603921 (succinate-CoA ligase ADP-forming subunit beta)
611224	SUCLG1 / HGNC:11449 / OMIM:611224 (succinate-CoA ligase GDP/ADP-forming subunit alpha)
185620	SURF1 / HGNC:11474 / OMIM:185620 (SURF1 cytochrome c oxidase assembly factor)
604126	SVIL / HGNC:11480 / OMIM:604126 (supervillin)
608441	SYNE1 / HGNC:17089 / OMIM:608441 (spectrin repeat containing nuclear envelope protein 1)
608442	SYNE2 / HGNC:17084 / OMIM:608442 (spectrin repeat containing nuclear envelope protein 2)
610949	SYT14 / HGNC:23143 / OMIM:610949 (synaptotagmin 14)
600104	SYT2 / HGNC:11510 / OMIM:600104 (synaptotagmin 2)
605078	TARDBP / HGNC:11571 / OMIM:605078 (TAR DNA binding protein)
300394	TAFAZZIN / HGNC:11577 / OMIM:300394 (tafazzin, phospholipid-lysophospholipid transacylase)
604649	TBCD / HGNC:11581 / OMIM:604649 (tubulin folding cofactor D)
604834	TBK1 / HGNC:11584 / OMIM:604834 (TANK binding kinase 1)
600075	TBP / HGNC:11588 / OMIM:600075 (TATA-box binding protein)
604488	TCAP / HGNC:11610 / OMIM:604488 (titin-cap)
607198	TDP1 / HGNC:18884 / OMIM:607198 (tyrosyl-DNA phosphodiesterase 1)
605764	TDP2 / HGNC:17768 / OMIM:605764 (tyrosyl-DNA phosphodiesterase 2)
615000	TECPR2 / HGNC:19957 / OMIM:615000 (tectonin beta-propeller repeat containing 2)
617242	TECRL / HGNC:27365 / OMIM:617242 (trans-2,3-enoyl-CoA reductase like)
602498	TFG / HGNC:11758 / OMIM:602498 (trafficking from ER to golgi regulator)
190230	TGFB3 / HGNC:11769 / OMIM:190230 (transforming growth factor beta 3)
613900	TGM6 / HGNC:16255 / OMIM:613900 (transglutaminase 6)
603518	TIA1 / HGNC:11802 / OMIM:603518 (TIA1 cytotoxic granule associated RNA binding protein)
607251	TIMM22 / HGNC:17317 / OMIM:607251 (translocase of inner mitochondrial membrane 22)
188250	TK2 / HGNC:11831 / OMIM:188250 (thymidine kinase 2)
616101	TMEM240 / HGNC:25186 / OMIM:616101 (transmembrane protein 240)
612048	TMEM43 / HGNC:28472 / OMIM:612048 (transmembrane protein 43)
616609	TMEM65 / HGNC:25203 / OMIM:616609 (transmembrane protein 65)
188380	TMPO / HGNC:11875 / OMIM:188380 (thymopoietin)
191040	TNNC1 / HGNC:11943 / OMIM:191040 (troponin C1, slow skeletal and cardiac type)
191043	TNNI2 / HGNC:11946 / OMIM:191043 (troponin I2, fast skeletal type)
191044	TNNI3 / HGNC:11947 / OMIM:191044 (troponin I3, cardiac type)
191041	TNNT1 / HGNC:11948 / OMIM:191041 (troponin T1, slow skeletal type)
191045	TNNT2 / HGNC:11949 / OMIM:191045 (troponin T2, cardiac type)
600692	TNNT3 / HGNC:11950 / OMIM:600692 (troponin T3, fast skeletal type)
610032	TNPO3 / HGNC:17103 / OMIM:610032 (transportin 3)
601243	TOP3A / HGNC:11992 / OMIM:601243 (DNA topoisomerase III alpha)
605204	TOR1A / HGNC:3098 / OMIM:605204 (torsin family 1 member A)
614512	TOR1AIP1 / HGNC:29456 / OMIM:614512 (torsin 1A interacting protein 1)
191010	TPM1 / HGNC:12010 / OMIM:191010 (tropomyosin 1)
190990	TPM2 / HGNC:12011 / OMIM:190990 (tropomyosin 2)
191030	TPM3 / HGNC:12012 / OMIM:191030 (tropomyosin 3)
614138	TRAPPC11 / HGNC:25751 / OMIM:614138 (trafficking protein particle complex subuni
603283	TRDN / HGNC:12261 / OMIM:603283 (triadin)

			614141 | TRIM2 / HGNC:15974 / OMIM:614141 (tripartite motif containing 2) 602290 | TRIM32 / HGNC:16380 / OMIM:602290 (tripartite motif containing 32) 606474 | TRIM54 / HGNC:16008 / OMIM:606474 (tripartite motif containing 54) 606131 | TRIM63 / HGNC:16007 / OMIM:606131 (tripartite motif containing 63) 604501 | TRIP4 / HGNC:12310 / OMIM:604501 (thyroid hormone receptor interactor 4) 602345 | TRPC3 / HGNC:12335 / OMIM:602345 (transient receptor potential cation channel subfamily C member 3) 605427 | TRPV4 / HGNC:18083 / OMIM:605427 (transient receptor potential cation channel subfamily V member 4) 604723 | TSFM / HGNC:12367 / OMIM:604723 (Ts translation elongation factor, mitochondrial) 611695 | TTBK2 / HGNC:19141 / OMIM:611695 (tau tubulin kinase 2) 188840 | TTN / HGNC:12403 / OMIM:188840 (titin) 600415 | TTPA / HGNC:12404 / OMIM:600415 (alpha tocopherol transfer protein) 176300 | TTR / HGNC:12405 / OMIM:176300 (transthyretin) 191110 | TUBA4A / HGNC:12407 / OMIM:191110 (tubulin alpha 4a) 602661 | TUBB3 / HGNC:20772 / OMIM:602661 (tubulin beta 3 class III) 606075 | TWNK / HGNC:1160 / OMIM:606075 (twinkle mtDNA helicase) 131222 | TYMP / HGNC:3148 / OMIM:131222 (thymidine phosphorylase) 314370 | UBA1 / HGNC:12469 / OMIM:314370 (ubiquitin like modifier activating enzyme 1) 609787 | UBAP1 / HGNC:12461 / OMIM:609787 (ubiquitin associated protein 1) 300264 | UBQLN2 / HGNC:12509 / OMIM:300264 (ubiquilin 2) 191342 | UCHL1 / HGNC:12513 / OMIM:191342 (ubiquitin C-terminal hydrolase L1) 605836 | UNC13B / HGNC:12566 / OMIM:605836 (unc-13 homolog B) 611220 | UNC45B / HGNC:14304 / OMIM:611220 (unc-45 myosin chaperone B) 185880 | VAMP1 / HGNC:12642 / OMIM:185880 (vesicle associated membrane protein 1) 605704 | VAPB / HGNC:12649 / OMIM:605704 (VAMP associated protein B and C) 193065 | VCL / HGNC:12665 / OMIM:193065 (vinculin) 601023 | VCP / HGNC:12666 / OMIM:601023 (valosin containing protein) 300913 | VMA21 / HGNC:22082 / OMIM:300913 (vacuolar ATPase assembly factor VMA21) 608877 | VPS13D / HGNC:23595 / OMIM:608877 (vacuolar protein sorting 13 homolog D) 609927 | VPS37A / HGNC:24928 / OMIM:609927 (VPS37A subunit of ESCRT-I) 602168 | VRK1 / HGNC:12718 / OMIM:602168 (VRK serine/threonine kinase 1) 614884 | VWA3B / HGNC:28385 / OMIM:614884 (von Willebrand factor A domain containing 3B) 191050 | WARS1 / HGNC:12729 / OMIM:191050 (tryptophanyl-tRNA synthetase 1) 605232 | WNK1 / HGNC:14540 / OMIM:605232 (WNK lysine deficient protein kinase 1) 605131 | WWOX / HGNC:12799 / OMIM:605131 (WW domain containing oxidoreductase) 194360 | XRCC1 / HGNC:12828 / OMIM:194360 (X-ray repair cross complementing 1) 603623 | YARS1 / HGNC:12840 / OMIM:603623 (tyrosyl-tRNA synthetase 1) 610957 | YARS2 / HGNC:24249 / OMIM:610957 (tyrosyl-tRNA synthetase 2) 613915 | ZBTB42 / HGNC:32550 / OMIM:613915 (zinc finger and BTB domain containing 42) 617828 | ZFHX2 / HGNC:20152 / OMIM:617828 (zinc finger homeobox 2) 612012 | ZFYVE26 / HGNC:20761 / OMIM:612012 (zinc finger FYVE-type containing 26) 610243 | ZFYVE27 / HGNC:26559 / OMIM:610243 (zinc finger FYVE-type containing 27)
386	[gendia_hgnc_ont] Show the field ONLY if: [gendia_listpref]='ontology'	Gene HGNC-code (browse ontology)If the affected gene is not present in the neuromuscular gene list above (e.g. a new gene not previously associated with neuromuscular disease) please search for the gene at https://www.genenames.org/tools/search/ and enter the numerical HGNC ID here (e.g. for DMD HGNC:2928 you would enter HGNC_2928) *Genetic diagnosis retained by the specialised centre (Element 6.2. in the Set of common data elements for Rare Diseases Registration (link))*	text, Required BIOPORTAL:HGNC | BIOPORTAL:HGNC
387	[gendia_omim_ont] Show the field ONLY if:	Gene OMIM-code (browse ontology)If the affected gene is not present in the neuromuscular gene list above (e.g. a new gene not previously associated with neuromuscular disease) please search for the gene at https://www.omim.org/ and enter the numerical OMIM ID here (e.g. for DMD you would enter 300377; you also find the code for this example if you	text, Required BIOPORTAL:OMIM | BIOPORTAL:OMIM

	[gendia_listpref]='ontology'	type 'Dystrophin' to the box on the right) *Genetic diagnosis retained by the specialised centre (Element 6.2. in the Set of common data elements for Rare Diseases Registration (link))*	
388	[gendia_omim]	Gene OMIM code calculated field	calc Calculation: if([gendia_listpref]='ontology', [gendia_omim_ont], if([gendia_listpref]='all_nmd', [gendia_omim_list], '')) Field Annotation: @HIDDEN @HIDDEN-PDF @READONLY
389	[gendia_inher] Show the field ONLY if: [gendia_known] = 'yes'	Inheritance	dropdown autosomal_dominant: Autosomal dominant autosomal_dominant_dn: Autosomal dominant - de novo sporadic: Sporadic autosomal_recessive: Autosomal recessive x_linked: X-linked x_linked_dom: X-linked dominant x_linked_rec: X-linked recessive y_linked: Y-linked inheritance digentic: Digentic inheritance mitochindrial: Mitochondrial inheritance oligogenic: Oligogenic inheritance autosomal_dominant_pat: Autosomal dominant with paternal imprinting autosomal_dominant_mat: Autosomal dominant with maternal imprinting unknown: Unknown
390	[gendia_square_bracket]	Section Header: *DISEASE-SPECIFIC GENETIC DETAILS* gendia_square_bracket	text Field Annotation: @DEFAULT='[' @HIDDEN @HIDDEN-PDF
391	[gendia_desc4] Show the field ONLY if: [gendia_known] = 'yes'	The genetic reporting methods in this section are displayed based on the disease group ([arm-label]) or the gene selected above (OMIM:[gendia_omim]) or the clinical diagnosis indicated in the "Baseline"-form ([first-event-name][diagnosis_rd_orpha] / [first-event-name][diagnosis_rd_orpha:value]).	descriptive
392	[helper_var1_v2]	#SECTION Oculopharyngeal Muscular Dystrophy (OPMD, ORPHA:270)	descriptive Field Annotation: @HIDDEN @HIDDEN-PDF
393	[gendia_pabpn1_report_v1] Show the field ONLY if: [gendia_omim] = 602279	Section Header: *PABPN1 (OMIM:602279; HGNC:8565; e.g. for diagnosis ORPHA:270)* Report number of GCG-repeats in PABPN1-gene (OMIM:602279; HGNC:8565) at NM_004643.3:c.4_6GCG	radio, Required distinct_number: Report a distinct number unknown: Unknown not_applicable: Not applicable not_present: Not present Field Annotation: @DEFAULT='[pabpn1_v_report_def]'
394	[gendia_pabpn1_nrep_v1] Show the field ONLY if: [gendia_omim] = 602279 AND [gendia_pabpn1_report_v1] = 'distinct_number'	Number of GCG repeats in PABPN1-gene (OMIM:602279; HGNC:8565) at NM_004643.3:c.4_6GCG *expected: integer between 8 and 13*	text (integer, Min: 8, Max: 13), Required
395	[gendia_pabpn1_hgvs_v1] Show the field ONLY if: [gendia_omim] = 602279 AND [gendia_pabpn1_report_v1] = 'distinct_number'	gendia_pabpn1_hgvs_v1	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_pabpn1_nrep_v1]!='',concat('NM_004643.3:c.4_6GCN', [gendia_square_bracket], [gendia_pabpn1_nrep_v1], ']'), ''))
396	[gendia_pabpn1_hgvs_disp] Show the field ONLY if: [gendia_omim] = 602279 AND [gendia_pabpn1_report_v1] = 'distinct_number'	Resulting HGVS variant notation:[gendia_pabpn1_hgvs_v1]	descriptive
397	[gendia_pabpn1_gen_build_v1] Show the field ONLY if: [gendia_omim] = 602279 AND [gendia_pabpn1_report_v1] = 'distinct_number'	Genomic build	dropdown GRCh38: GRCh38 (hg38) GRCh37: GRCh37 (hg19)
398	[gendia_pabpn1_gen_app_v1] Show the field ONLY if:	Genetic approach	dropdown single_gene_screening: Single Gene Screening

	[gendia_omim] = 602279 AND [gendia_pabpn1_report_v1] = 'distinct_number'		acgh, aCGH snp_array, SNP array mlpa, MLPA karyotype, Karyotype ngs_panel, NGS panel wes, WES wgs, WGS
399	[gendia_pabpn1_chrom_v1] Show the field ONLY if: [gendia_omim] = 602279 AND [gendia_pabpn1_report_v1] = 'distinct_number'	Chromosome	dropdown 1, 1 2, 2 3, 3 4, 4 5, 5 6, 6 7, 7 8, 8 9, 9 10, 10 11, 11 12, 12 13, 13 14, 14 15, 15 16, 16 17, 17 18, 18 19, 19 20, 20 21, 21 22, 22 X, X Y, Y MT, MT
400	[gendia_pabpn1_zygos_v1] Show the field ONLY if: [gendia_omim] = 602279 AND [gendia_pabpn1_report_v1] = 'distinct_number'	Zygosity	dropdown heterozygous, Heterozygous compound_heterozygous, Compound heterozygous homozygous, Homozygous hemizygous, Hemizygous mosaic, Mosaic unknown, Unknown
401	[helper_var2_v2]	#SECTION Myotonic Dystrophy type 1 (DM1, ORPHA:273)	descriptive Field Annotation: @HIDDEN @HIDDEN-PDF
402	[gendia_dmpk_report_v1] Show the field ONLY if: [gendia_omim] = 605377	Section Header: *DMPK (OMIM:605377; HGNC:2933; e.g. for diagnosis ORPHA:273)* Report number of CTG-repeats in DMPK-gene (OMIM:605377; HGNC:2933) at NM_001081563.1:c.*224_226CTG	radio, Required distinct_number, Report a distinct number range, [for higher repeats]: Report a range unknown, Unknown not_applicable, Not applicable

			not_present: Not present Field Annotation: @DEFAULT='[dmpk_v_report_def]'
403	[gendia_dmpk_nrep_v1] Show the field ONLY if: [gendia_omim] = 605377 AND [gendia_dmpk_report_v1] = 'distinct_number'	Number of CTG-repeatsin DMPK-gene (OMIM:605377; HGNC:2933)at NM_001081563.1:c.*224_226CTG *expected: integer, minimum: 50*	text (integer, Min: 50), Required
404	[gendia_dmpk_nrep_r1_v1] Show the field ONLY if: [gendia_omim] = 605377 AND [gendia_dmpk_report_v1] = 'range'	Inclusive lower bound of range for number of CTG-repeats in DMPK-gene (OMIM:605377; HGNC:2933)at NM_001081563.1:c.*224_226CTG *expected: integer, minimum: 50*	text (integer, Min: 50), Required
405	[gendia_dmpk_nrep_r2_v1] Show the field ONLY if: [gendia_omim] = 605377 AND [gendia_dmpk_report_v1] = 'range'	Inclusive upper bound of range for number of CTG-repeatsin DMPK-gene (OMIM:605377; HGNC:2933)at NM_001081563.1:c.*224_226CTG *expected: integer*	text (integer, Min: 51), Required
406	[gendia_dmpk_hgvs_v1] Show the field ONLY if: [gendia_omim] = 605377 AND ([gendia_dmpk_report_v1] = 'distinct_number' OR [gendia_dmpk_report_v1] = 'range')	gendia_dmpk_hgvs_v1	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_dmpk_report_v1] = 'distinct_number', concat('NM_001081563.1:c.*224_226CTG', [gendia_square_bracket], [gendia_dmpk_nrep_v1], ']'), if([gendia_dmpk_report_v1]='range', concat('NM_001081563.1:c.*224_226CTG', [gendia_square_bracket], [gendia_dmpk_nrep_r1_v1],'_',[gendia_dmpk_nrep_r2_v1],']'), ''))
407	[gendia_dmpk_hgvs_disp_v1] Show the field ONLY if: [gendia_omim] = 605377 AND ([gendia_dmpk_report_v1] = 'distinct_number' OR [gendia_dmpk_report_v1] = 'range')	Resulting variant notation:[gendia_dmpk_hgvs_v1]	descriptive
408	[gendia_dmpk_gen_build_v1] Show the field ONLY if: [gendia_omim] = 605377 AND ([gendia_dmpk_report_v1] = 'distinct_number' OR [gendia_dmpk_report_v1] = 'range')	Genomic build	dropdown GRCh38: GRCh38 (hg38) GRCh37: GRCh37 (hg19)
409	[gendia_dmpk_gen_app_v1] Show the field ONLY if: [gendia_omim] = 605377 AND ([gendia_dmpk_report_v1] = 'distinct_number' OR [gendia_dmpk_report_v1] = 'range')	Genetic approach	dropdown single_gene_screening: Single Gene Screening acgh: aCGH snp_array: SNP array mlpa: MLPA karyotype: Karyotype ngs_panel: NGS panel wes: WES wgs: WGS
410	[gendia_dmpk_chrom_v1] Show the field ONLY if: [gendia_omim] = 605377 AND ([gendia_dmpk_report_v1] = 'distinct_number' OR [gendia_dmpk_report_v1] = 'range')	Chromosome	dropdown 1: 1 2: 2 3: 3 4: 4 5: 5 6: 6 7: 7 8: 8 9: 9 10: 10 11: 11 12: 12 13: 13 14: 14

			15 15 16 16 17 17 18 18 19 19 20 20 21 21 22 22 X X Y Y MT MT
411	[gendia_dmpk_zygos_v1] Show the field ONLY if: [gendia_omim] = 605377 AND ([gendia_dmpk_report_v1] = 'distinct_number' OR [gendia_dmpk_report_v1] = 'range')	Zygosity	dropdown heterozygous | Heterozygous compound_heterozygous | Compound heterozygous homozygous | Homozygous hemizygous | Hemizygous mosaic | Mosaic unknown | Unknown
412	[helper_var3_v2]	#SECTION Myotonic Dystrophy type 2 (DM2, ORPHA:606)	descriptive Field Annotation: @HIDDEN @HIDDEN-PDF
413	[gendia_cnbp_report_v1] Show the field ONLY if: [gendia_omim] = 116955	Section Header: *CNBP (OMIM:116955; HGNC:13164; e.g. for diagnosis ORPHA:606)* Report number of CCTG-repeats in CNBP-gene (OMIM:116955; HGNC:13164) at NM_003418.5(CNBP):c.-14-833_-14-806CCTG	radio, Required distinct_number | Report a distinct number range | [for higher repeats]: Report a range unknown | Unknown not_applicable | Not applicable not_present | Not present Field Annotation: @DEFAULT='[cnbp_v_report_def]'
414	[gendia_cnbp_nrep_v1] Show the field ONLY if: [gendia_omim] = 116955 AND [gendia_cnbp_report_v1] = 'distinct_number'	Number of CCTG-repeatsin CNBP-gene (OMIM:116955; HGNC:13164)at NM_003418.5(CNBP):c.-14-833_-14-806CCTG *expected: integer, minimum: 75*	text (integer, Min: 75), Required
415	[gendia_cnbp_nrep_r1_v1] Show the field ONLY if: [gendia_omim] = 116955 AND [gendia_cnbp_report_v1] = 'range'	Inclusive lower bound of range for number of CCTG-repeatsin CNBP-gene (OMIM:116955; HGNC:13164)at NM_003418.5(CNBP):c.-14-833_-14-806CCTG *expected: integer, minimum: 75*	text (integer, Min: 75), Required
416	[gendia_cnbp_nrep_r2_v1] Show the field ONLY if: [gendia_omim] = 116955 AND [gendia_cnbp_report_v1] = 'range'	Inclusive upper bound of range for number of CCTG-repeatsin CNBP-gene (OMIM:116955; HGNC:13164)at NM_003418.5(CNBP):c.-14-833_-14-806CCTG *expected: integer*	text (integer, Min: 76), Required
417	[gendia_cnbp_hgvs_v1] Show the field ONLY if: [gendia_omim] = 116955 AND ([gendia_cnbp_report_v1] = 'distinct_number' OR [gendia_cnbp_report_v1]='range')	cnbp_v1_hgvs	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_cnbp_report_v1]='distinct_number', concat('NM_003418.5(CNBP):c.-14-833_-14-806CCTG', [gendia_square_bracket], [gendia_cnbp_nrep_v1],']'), if([gendia_cnbp_report_v1]='range', concat('NM_003418.5(CNBP):c.-14-833_-14-806CCTG', [gendia_square_bracket],[gendia_cnbp_nrep_r1_v1],'_',[gendia_cnbp_nrep_r2_v1],']'), ''))
418	[gendia_cnbp_hgvs_disp_v1] Show the field ONLY if: [gendia_omim] = 116955 AND ([gendia_cnbp_report_v1] = 'distinct_number' OR [gendia_cnbp_report_v1]='range')	Resulting variant notation:[gendia_cnbp_hgvs_v1]	descriptive
419	[gendia_cnbp_gen_build_v1] Show the field ONLY if: [gendia_omim] = 116955 AND ([gendia_cnbp_report_v1] = '	Genomic build	dropdown GRCh38 | GRCh38 (hg38) GRCh37 | GRCh37 (hg19)

	distinct_number' OR [gendia_cnbp_report_v1]='range')		
420	[gendia_cnbp_gen_app_v1] Show the field ONLY if: [gendia_omim] = 116955 AND ([gendia_cnbp_report_v1] = ' distinct_number' OR [gendia_cnbp_report_v1]='range')	Genetic approach	dropdown single_gene_screening: Single Gene Screening acgh: aCGH snp_array: SNP array mlpa: MLPA karyotype: Karyotype ngs_panel: NGS panel wes: WES wgs: WGS
421	[gendia_cnbp_chrom_v1] Show the field ONLY if: [gendia_omim] = 116955 AND ([gendia_cnbp_report_v1] = ' distinct_number' OR [gendia_cnbp_report_v1]='range')	Chromosome	dropdown 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13, 14, 15, 16, 17, 18, 19, 20, 21, 22, X, Y, MT
422	[gendia_cnbp_zygos_v1] Show the field ONLY if: [gendia_omim] = 116955 AND ([gendia_cnbp_report_v1] = ' distinct_number' OR [gendia_cnbp_report_v1]='range')	Zygosity	dropdown heterozygous: Heterozygous compound_heterozygous: Compound heterozygous homozygous: Homozygous hemizygous: Hemizygous mosaic: Mosaic unknown: Unknown
423	[helper_var8_v2]	#SECTION Facioscapulohumeral Muscular Dystrophy type 1 (FSHD, OMIM:158900, ORPHA:269 - orphacode not for type1 only)	descriptive Field Annotation: @HIDDEN @HIDDEN-PDF
424	[gendia_frag_4q35_report] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_omim] = 601278	Section Header: *FRG1 (OMIM:601278; HGNC:3954; e.g. for diagnosis ORPHA:269): 4q35 fragment* 4q35 fragment: Report size of remaining EcoRI/BlnI fragment in kB	radio distinct_number: Report a distinct number unknown: Unknown

			not_applicable | Not applicable not_present | Not present Field Annotation: @DEFAULT='[fragment_4q35_report_def]'
425	[gendia_frag_4q35_size] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_omim] = 601278 AND [gendia_frag_4q35_report]='distinct_number'	4q35 fragment: Size of remaining EcoRI/BlnI fragment in kB *expected: integer between 0 and 40*	text (integer, Min: 0, Max: 40), Required
426	[gendia_4q35_gen_build] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_omim] = 601278 AND [gendia_frag_4q35_report]='distinct_number'	Genomic build	dropdown GRCh38 | GRCh38 (hg38) GRCh37 | GRCh37 (hg19)
427	[gendia_4q35_gen_app] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_omim] = 601278 AND [gendia_frag_4q35_report]='distinct_number'	Genetic approach	dropdown single_gene_screening | Single Gene Screening acgh | aCGH snp_array | SNP array mlpa | MLPA karyotype | Karyotype ngs_panel | NGS panel wes | WES wgs | WGS
428	[gendia_4q35_chrom] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_omim] = 601278 AND [gendia_frag_4q35_report]='distinct_number'	Chromosome	dropdown 1 | 1 2 | 2 3 | 3 4 | 4 5 | 5 6 | 6 7 | 7 8 | 8 9 | 9 10 | 10 11 | 11 12 | 12 13 | 13 14 | 14 15 | 15 16 | 16 17 | 17 18 | 18 19 | 19 20 | 20 21 | 21 22 | 22 X | X Y | Y MT | MT
429	[gendia_4q35_zygos] Show the field ONLY if:	Zygosity	dropdown heterozygous | Heterozygous

	[gendia_known] = 'yes' AND [gendia_omim] = 601278 AND [gendia_frag_4q35_report]='distinct_number'		<table><tr><td>compound_heterozygous</td><td>Compound heterozygous</td></tr><tr><td>homozygous</td><td>Homozygous</td></tr><tr><td>hemizygous</td><td>Hemizygous</td></tr><tr><td>mosaic</td><td>Mosaic</td></tr><tr><td>unknown</td><td>Unknown</td></tr></table>
430	[helper_var4_v2]	#SECTION Charcot-Marie-Tooth 1A (CMT1A, ORPHA:101081) #SECTION Hereditary neuropathy with liability to pressure palsies (ORPHA:640)	descriptive Field Annotation: @HIDDEN @HIDDEN-PDF
431	[gd_pmp22_dup_del] Show the field ONLY if: [gendia_omim] = 601097	Section Header: *Duplication of PMP22 (OMIM:601097; HGNC:9118; e.g. for diagnosis ORPHA:101081) OR Deletion of PMP22 (OMIM:601097; HGNC:9118; e.g. for diagnosis ORPHA:640)* Which change applies to PMP22-gene (OMIM:601097; HGNC:9118)?	radio <table><tr><td>duplication</td><td>Duplication of the gene</td></tr><tr><td>deletion</td><td>Deletion of the gene</td></tr><tr><td>not_present</td><td>Not present (none of the above)</td></tr><tr><td>unknown</td><td>Unknown</td></tr></table>
432	[gd_pmp22_dup_hgvs_v1] Show the field ONLY if: [gendia_omim] = 601097 AND [gd_pmp22_dup_del] = 'duplication'	gd_pmp22_dup_hgvs_v1	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_omim] = 601097 AND [gd_pmp22_dup_del] = 'duplication', 'NM_000304.3:c.-238_*1140{2}', ''))
433	[gd_pmp22_dup_hgvs_v2] Show the field ONLY if: [gendia_omim] = 601097 AND [gd_pmp22_dup_del] = 'duplication'	gd_pmp22_dup_hgvs_v2	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_omim] = 601097 AND [gd_pmp22_dup_del] = 'duplication', 'NC_000017.10:g.(?_15133094)_(15168674_?)dup', ''))
434	[gd_pmp22_del_hgvs_v1] Show the field ONLY if: [gendia_omim] = 601097 AND [gd_pmp22_dup_del] = 'deletion'	gd_pmp22_del_hgvs_v1	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_omim] = 601097 AND [gd_pmp22_dup_del] = 'deletion', 'NM_000304.3:c.-238_*1140{0}', ''))
435	[gd_pmp22_del_hgvs_v2] Show the field ONLY if: [gendia_omim] = 601097 AND [gd_pmp22_dup_del] = 'deletion'	gd_pmp22_del_hgvs_v2	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_omim] = 601097 AND [gd_pmp22_dup_del] = 'deletion', 'NC_000017.10:g.(?_15133094)_(15168674_?)del', ''))
436	[gd_pmp22_hgvs_dup_disp_v1] Show the field ONLY if: [gendia_omim] = 601097 AND [gd_pmp22_dup_del] = 'duplication'	Resulting variant notation:[gd_pmp22_dup_hgvs_v1]	descriptive
437	[gd_pmp22_hgvs_dup_disp_v2] Show the field ONLY if: [gendia_omim] = 601097 AND [gd_pmp22_dup_del] = 'duplication'	Resulting variant notation:[gd_pmp22_dup_hgvs_v2]	descriptive
438	[gd_pmp22_hgvs_del_disp_v1] Show the field ONLY if: [gendia_omim] = 601097 AND [gd_pmp22_dup_del] = 'deletion'	Resulting variant notation:[gd_pmp22_del_hgvs_v1]	descriptive
439	[gd_pmp22_hgvs_del_disp_v2] Show the field ONLY if: [gendia_omim] = 601097 AND [gd_pmp22_dup_del] = 'deletion'	Resulting variant notation:[gd_pmp22_del_hgvs_v2]	descriptive
440	[gd_pmp22_gen_build_v1] Show the field ONLY if: [gendia_omim] = 601097 AND ([gd_pmp22_dup_del] = 'duplication' OR [gd_pmp22_dup_del] = 'deletion')	Genomic build	dropdown <table><tr><td>GRCh38</td><td>GRCh38 (hg38)</td></tr><tr><td>GRCh37</td><td>GRCh37 (hg19)</td></tr></table>
441	[gd_pmp22_gen_app_v1] Show the field ONLY if: [gendia_omim] = 601097 AND ([gd_pmp22_dup_del] = 'duplication' OR [gd_pmp22_dup_del] = 'deletion')	Genetic approach	dropdown <table><tr><td>single_gene_screening</td><td>Single Gene Screening</td></tr><tr><td>acgh</td><td>aCGH</td></tr><tr><td>snp_array</td><td>SNP array</td></tr><tr><td>mlpa</td><td>MLPA</td></tr></table>

			karyotype: Karyotype ngs_panel: NGS panel wes: WES wgs: WGS
442	[gd_pmp22_chrom_v1] Show the field ONLY if: [gendia_omim] = 601097 AND ([gd_pmp22_dup_del] = 'duplication' OR [gd_pmp22_dup_del] = 'deletion')	Chromosome	dropdown 1: 1 2: 2 3: 3 4: 4 5: 5 6: 6 7: 7 8: 8 9: 9 10: 10 11: 11 12: 12 13: 13 14: 14 15: 15 16: 16 17: 17 18: 18 19: 19 20: 20 21: 21 22: 22 X: X Y: Y MT: MT
443	[gd_pmp22_zygos_v1] Show the field ONLY if: [gendia_omim] = 601097 AND ([gd_pmp22_dup_del] = 'duplication' OR [gd_pmp22_dup_del] = 'deletion')	Zygosity	dropdown heterozygous: Heterozygous compound_heterozygous: Compound heterozygous homozygous: Homozygous hemizygous: Hemizygous mosaic: Mosaic unknown: Unknown
444	[helper_var6_v2]	#SECTION Spinobulbar Muscular Atrophy (SBMA, ORPHA:481)	descriptive Field Annotation: @HIDDEN @HIDDEN-PDF
445	[gendia_ar_report_v1] Show the field ONLY if: [gendia_omim] = 313700	Section Header: *AR (OMIM:313700; HGNC:644; e.g. for diagnosis ORPHA:481)* Report number of CAG-repeats in AR-gene (OMIM:313700; HGNC:644) at NM_000044.3:c.172_174CAG	radio, Required distinct_number: Report a distinct number unknown: Unknown not_applicable: Not applicable not_present: Not present Field Annotation: @DEFAULT='[ar_v1_report_def]'
446	[gendia_ar_nrep_v1] Show the field ONLY if:	Number of CAG-repeatsin AR-gene (OMIM:313700; HGNC:644)at NM_000044.3:c.172_174CAG *expected: integer between 38 and 62*	text (integer, Min: 38, Max: 62)

	[gendia_omim] = 313700 AND [gendia_ar_report_v1] = 'distinct_number'		
447	[gendia_ar_hgvs_v1] Show the field ONLY if: [gendia_omim] = 313700 AND [gendia_ar_report_v1] = 'distinct_number'	gendia_ar_hgvs_v1	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_ar_nrep_v1]!='',concat('NM_000044.3:c.172_174CAG', [gendia_square_bracket], [gendia_ar_nrep_v1], ']'), ''))
448	[gendia_ar_hgvs_disp_v1] Show the field ONLY if: [gendia_omim] = 313700 AND [gendia_ar_report_v1] = 'distinct_number'	Resulting variant notation:[gendia_ar_hgvs_v1]	descriptive
449	[gendia_ar_gen_build_v1] Show the field ONLY if: [gendia_omim] = 313700 AND [gendia_ar_report_v1] = 'distinct_number'	Genomic build	dropdown GRCh38, GRCh38 (hg38) GRCh37, GRCh37 (hg19)
450	[gendia_ar_gen_app_v1] Show the field ONLY if: [gendia_omim] = 313700 AND [gendia_ar_report_v1] = 'distinct_number'	Genetic approach	dropdown single_gene_screening, Single Gene Screening acgh, aCGH snp_array, SNP array mlpa, MLPA karyotype, Karyotype ngs_panel, NGS panel wes, WES wgs, WGS
451	[gendia_ar_chrom_v1] Show the field ONLY if: [gendia_omim] = 313700 AND [gendia_ar_report_v1] = 'distinct_number'	Chromosome	dropdown 1, 1 2, 2 3, 3 4, 4 5, 5 6, 6 7, 7 8, 8 9, 9 10, 10 11, 11 12, 12 13, 13 14, 14 15, 15 16, 16 17, 17 18, 18 19, 19 20, 20 21, 21 22, 22 X, X Y, Y MT, MT

452	[gendia_ar_zygos_v1] Show the field ONLY if: [gendia_omim] = 313700 AND [gendia_ar_report_v1] = 'distinct_number'	Zygosity	dropdown heterozygous: Heterozygous compound_heterozygous: Compound heterozygous homozygous: Homozygous hemizygous: Hemizygous mosaic: Mosaic unknown: Unknown
453	[helper_var9_v2]	#SECTION Spinal Muscular Atrophy (SMA types 1, 2, 3)	descriptive Field Annotation: @HIDDEN @HIDDEN-PDF
454	[gendia_smn2_copy_n_known] Show the field ONLY if: [gendia_known] = 'yes' AND ([arm-number] = 5 OR [gendia_omim] = 601627)	Section Header: *SMN2 (OMIM:601627; HGNC:11118) copy number* SMN2 (OMIM:601627; HGNC:11118) copy number known	radio yes: Yes no: No
455	[gendia_smn2_copy_n] Show the field ONLY if: [gendia_known] = 'yes' AND ([arm-number] = 5 OR [gendia_omim] = 601627) AND [gendia_smn2_copy_n_known]='yes'	SMN2 copy number *expected: integer between 1 and 6*	text (number, Min: 1, Max: 6), Required
456	[gendia_smn2_gen_build_v1] Show the field ONLY if: [gendia_smn2_copy_n] >= 1	Genomic build	dropdown GRCh38: GRCh38 (hg38) GRCh37: GRCh37 (hg19)
457	[gendia_smn2_gen_app_v1] Show the field ONLY if: [gendia_smn2_copy_n] >= 1	Genetic approach	dropdown single_gene_screening: Single Gene Screening acgh: aCGH snp_array: SNP array mlpa: MLPA karyotype: Karyotype ngs_panel: NGS panel wes: WES wgs: WGS
458	[gendia_smn2_chrom_v1] Show the field ONLY if: [gendia_smn2_copy_n] >= 1	Chromosome	dropdown 1: 1 2: 2 3: 3 4: 4 5: 5 6: 6 7: 7 8: 8 9: 9 10: 10 11: 11 12: 12 13: 13 14: 14 15: 15 16: 16 17: 17 18: 18

			19 | 19 20 | 20 21 | 21 22 | 22 X | X Y | Y MT | MT
459	[gendia_smn2_zygos_v1] Show the field ONLY if: [gendia_smn2_copy_n] >= 1	Zygosity	dropdown heterozygous | Heterozygous compound_heterozygous | Compound heterozygous homozygous | Homozygous hemizygous | Hemizygous mosaic | Mosaic unknown | Unknown
460	[gendia_smn1_variant_v1] Show the field ONLY if: [gendia_known] = 'yes' AND ([arm-number]=5 OR [gendia_omim]=600354)	Section Header: *SMN1 (OMIM:600354; HGNC:11117)* SMN1 (OMIM:600354; HGNC:11117) variant	radio homoz_del_ex7 | Homozygous deletion of exon 7 of SMN1 comp_heteroz_del_ex7 | Compound heterozygous for a deletion of exon 7 and a point mutation in SMN1 comp_heteroz_2sub | Compound heterozygous for two substitutions (point mutations) in SMN1 homoz_1sub | Homozygous for a single substitution (point mutation) in SMN1 (rare, may occur in consanguinous families) none_of_above | None of the above unknown | Unknown
461	[gendia_smn1_variant_desc2] Show the field ONLY if: [gendia_smn1_variant_v1] = 'comp_heteroz_del_ex7'	Please fill in the point mutation in variant 1 of "REPORTING CAUSATIVE VARIANT(S)" below.	descriptive
462	[gendia_smn1_variant_desc3] Show the field ONLY if: [gendia_smn1_variant_v1] = 'comp_heteroz_2sub'	Please fill in both variants in "REPORTING CAUSATIVE VARIANT(S)" below.	descriptive
463	[gendia_smn1_variant_desc4] Show the field ONLY if: [gendia_smn1_variant_v1] = 'homoz_1sub'	Please fill in same variant in both variant 1 and variant 2 in "REPORTING CAUSATIVE VARIANT(S)" below.	descriptive
464	[gendia_smn1_gen_build_v1] Show the field ONLY if: [gendia_known] = 'yes' AND ([arm-number]=5 OR [gendia_omim]=600354) AND ([gendia_smn1_variant_v1] = 'homoz_del_ex7' OR [gendia_smn1_variant_v1] = 'comp_heteroz_del_ex7' OR [gendia_smn1_variant_v1] = 'comp_heteroz_2sub' OR [gendia_smn1_variant_v1] = 'homoz_1sub')	Genomic build	dropdown GRCh38 | GRCh38 (hg38) GRCh37 | GRCh37 (hg19)
465	[gendia_smn1_gen_app_v1] Show the field ONLY if: [gendia_known] = 'yes' AND ([arm-number]=5 OR [gendia_omim]=600354) AND ([gendia_smn1_variant_v1] = 'homoz_del_ex7' OR [gendia_smn1_variant_v1] = 'comp_heteroz_del_ex7' OR [gendia_smn1_variant_v1] = 'comp_heteroz_2sub' OR [gendia_smn1_variant_v1] = 'homoz_1sub')	Genetic approach	dropdown single_gene_screening | Single Gene Screening acgh | aCGH snp_array | SNP array mlpa | MLPA karyotype | Karyotype ngs_panel | NGS panel wes | WES wgs | WGS
466	[gendia_smn1_chrom_v1] Show the field ONLY if:	Chromosome	dropdown 1 | 1

	[gendia_known] = 'yes' AND ([arm-number]=5 OR [gendia_omim]=600354) AND ([gendia_smn1_variant_v1] = 'homoz_del_ex7' OR [gendia_smn1_variant_v1] = 'comp_heteroz_del_ex7' OR [gendia_smn1_variant_v1] = 'comp_heteroz_2sub' OR [gendia_smn1_variant_v1] = 'homoz_1sub')		2 2 3 3 4 4 5 5 6 6 7 7 8 8 9 9 10 10 11 11 12 12 13 13 14 14 15 15 16 16 17 17 18 18 19 19 20 20 21 21 22 22 X X Y Y MT MT
467	[helper_var7_v2]	#SECTION DMD/BMD	descriptive Field Annotation: @HIDDEN @HIDDEN-PDF
468	[gendia_dmd_desc1] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_omim]='300377'	Section Header: *DMD (OMIM:300377; HGNC:2928; e.g. for diagnoses ORPHA:98895, ORPHA:98896, ORPHA:206546)* If... the change is described in the NCBI ClinVar database OR you can describe the change in HGVS-simple notation OR there is a point mutation in the DMD-gene as well ...please fill the section "REPORTING CAUSATIVE VARIANT(S)" below. The LOVD-tool (accessed: 01.03.2022) may be helpful to convert exon deletions in the DMD gene to HGVS nomenclature.	descriptive
469	[gendia_dmd_mutation_type] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_omim]='300377'	Mutation type of DMD-gene (OMIM:300377; HGNC:2928)	radio, Required deletion: Deletion of one or several exons duplication: Duplication of one or several exons triplication: Triplication of one or several exons not_present: Not present (none of the above) unknown: Unknown
470	[gendia_dmd_exon_del_first] Show the field ONLY if: [gendia_dmd_mutation_type] = 'deletion'	Section Header: *Deletion* First deleted exon	text (integer), Required
471	[gendia_dmd_exon_del_last] Show the field ONLY if: [gendia_dmd_mutation_type] = 'deletion'	Last deleted exon	text (integer), Required
472	[gendia_dmd_exon_del] Show the field ONLY if: [gendia_dmd_mutation_type] = 'deletion'	gendia_dmd_exon_del	text Field Annotation: @CALCTEXT(if([gendia_dmd_mutation_type] = 'deletion', if([gendia_dmd_exon_del_first]=[gendia_dmd_exon_del_last], concat('del ex', [gendia_dmd_exon_del_first]), concat('del ex', [gendia_dmd_exon_del_first], '-', [gendia_dmd_exon_del_last])), '')) @READONLY @HIDDEN @HIDDEN-PDF
473	[exon_v_del_disp_v2] Show the field ONLY if: [gendia_dmd_mutation_type] = 'deletion'	Resulting exon-based description of the variant: [gendia_dmd_exon_del] (see guidelines for curators on mutation entries in DMD registries, accessed 18.11.2021)	descriptive

474	[gendia_dmd_exon_dup_first] Show the field ONLY if: [gendia_dmd_mutation_type] = 'duplication'	Section Header: *Duplication* First duplicated exon	text (integer), Required
475	[gendia_dmd_exon_dup_last] Show the field ONLY if: [gendia_dmd_mutation_type] = 'duplication'	Last duplicated exon	text (integer), Required
476	[gendia_dmd_exon_dup] Show the field ONLY if: [gendia_dmd_mutation_type] = 'duplication'	gendia_dmd_exon_dup	text Field Annotation: @CALCTEXT(if([gendia_dmd_mutation_type] = 'duplication', if([gendia_dmd_exon_dup_first]=[gendia_dmd_exon_dup_last], concat('dup ex', [gendia_dmd_exon_dup_first]), concat('dup ex', [gendia_dmd_exon_dup_first], '-', [gendia_dmd_exon_dup_last])), '')) @READONLY @HIDDEN @HIDDEN-PDF
477	[gendia_dmd_exon_dup_disp] Show the field ONLY if: [gendia_dmd_mutation_type] = 'duplication'	Resulting exon-based description of the variant: [gendia_dmd_exon_dup] (see guidelines for curators on mutation entries in DMD registries, accessed 18.11.2021)	descriptive
478	[gendia_dmd_exon_tri_first] Show the field ONLY if: [gendia_dmd_mutation_type] = 'triplication'	Section Header: *Triplication* First triplicated exon	text (integer), Required
479	[gendia_dmd_exon_tri_last] Show the field ONLY if: [gendia_dmd_mutation_type] = 'triplication'	Last triplicated exon	text (integer), Required
480	[gendia_dmd_exon_tri] Show the field ONLY if: [gendia_dmd_mutation_type] = 'triplication'	gendia_dmd_exon_tri	text Field Annotation: @CALCTEXT(if([gendia_dmd_mutation_type] = 'triplication', if([gendia_dmd_exon_tri_first]=[gendia_dmd_exon_tri_last], concat('tri ex', [gendia_dmd_exon_tri_first]), concat('tri ex', [gendia_dmd_exon_tri_first], '-', [gendia_dmd_exon_tri_last])), '')) @READONLY @HIDDEN @HIDDEN-PDF
481	[gendia_dmd_exon_tri_disp] Show the field ONLY if: [gendia_dmd_mutation_type] = 'triplication'	Resulting exon-based description of the variant: [gendia_dmd_exon_tri] (see guidelines for curators on mutation entries in DMD registries, accessed 18.11.2021)	descriptive
482	[gendia_dmd_gen_build_v1] Show the field ONLY if: [gendia_dmd_mutation_type] = 'deletion' OR [gendia_dmd_mutation_type] = 'duplication' OR [gendia_dmd_mutation_type] = 'triplication'	Section Header: *Additional Information* Genomic build	dropdown GRCh38, GRCh38 (hg38) GRCh37, GRCh37 (hg19)
483	[gendia_dmd_gen_app_v1] Show the field ONLY if: [gendia_dmd_mutation_type] = 'deletion' OR [gendia_dmd_mutation_type] = 'duplication' OR [gendia_dmd_mutation_type] = 'triplication'	Genetic approach	dropdown single_gene_screening, Single Gene Screening acgh, aCGH snp_array, SNP array mlpa, MLPA karyotype, Karyotype ngs_panel, NGS panel wes, WES wgs, WGS
484	[gendia_dmd_chrom_v1] Show the field ONLY if: [gendia_dmd_mutation_type] = 'deletion' OR [gendia_dmd_mutation_type] = 'duplication' OR [gendia_dmd_mutation_type] = 'triplication'	Chromosome	dropdown 1, 1 2, 2 3, 3 4, 4 5, 5 6, 6 7, 7 8, 8 9, 9

			10 | 10 11 | 11 12 | 12 13 | 13 14 | 14 15 | 15 16 | 16 17 | 17 18 | 18 19 | 19 20 | 20 21 | 21 22 | 22 X | X Y | Y MT | MT
485	[gendia_dmd_zygos_v1] Show the field ONLY if: [gendia_dmd_mutation_type] = 'deletion' OR [gendia_dmd_mutation_type] = 'duplication' OR [gendia_dmd_mutation_type] = 'triplication'	Zygosity	dropdown heterozygous | Heterozygous compound_heterozygous | Compound heterozygous homozygous | Homozygous hemizygous | Hemizygous mosaic | Mosaic unknown | Unknown
486	[gendia_desc2] Show the field ONLY if: [gendia_known] = 'yes'	Section Header: *REPORTING CAUSATIVE VARIANT(S)* Please note: You only need to fill in this section ifa) if no data entry fields are shown in the above section "DISEASE-SPECIFIC GENETIC DETAILS". In this case, none of the more frequent variants for which we have already stored a correspondingly simplified data entry applies to this patient.b) if you want to report variant(s) in the selected gene (OMIM: [gendia_omim]) that are not covered by the variant(s) described under "DISEASE-SPECIFIC GENETIC DETAILS" above Please report only an additional variant for the gene selected above (OMIM:[gendia_omim]). If you wish to report a variant for another gene, please report that in a separate 'Genetic Diagnosis'-form for that gene. This section is intended to record the change(s) in DNA, RNA or protein sequences according to the standards of the Sequence Variant Nomenclature "HGVS simple" (accessed: 18.13.22). HGVS-codes can be checked with Mutalyzer (accessed: 18.03.22). Please enter all the details that you have available from the report. For autosomal recessive cases, you must enter both variant 1 and variant 2. If the variant is homozygous, enter the identical details twice. For autosomal dominant and X-linked, only one is required.	descriptive
487	[gendia_report_causa_variant] Show the field ONLY if: [gendia_known] = 'yes'	Report (additional) variant(s) / variant(s) not covered by the predefined variant(s) under "DISEASE-SPECIFIC GENETIC DETAILS" above	yesno, Required 1 | Yes 0 | No
488	[gendia_v1_report] Show the field ONLY if: [gendia_report_causa_variant] = 1	Section Header: *Variant 1* How would you like to describe variant 1?Note: We prefer NCBI ClinVar but if the change in question is not yet described there, you can give a dbSNP ID if available, or you can type in the protein or cDNA change given in the genetic report.	radio, Required clinvar | NCBI ClinVar dbsnp | dbSNP custom_hgvs | Custom HGVS-simple notation
489	[gendia_v1_clinvar_uri] Show the field ONLY if: [gendia_v1_report] = 'clinvar'	Variant 1: NCBI ClinVar URI(please provide website-link) *If the change is described in the NCBI ClinVar database, please provide website-link (e.g. https://www.ncbi.nlm.nih.gov/clinvar/variation/4886/)*	text, Required
490	[gendia_v1_dbsnp_id] Show the field ONLY if: [gendia_v1_report] = 'dbsnp'	Variant 1: dbSNP RS ID *If the change has a dbSNP ID, enter it here, e.g. rs606231128*	text, Required
491	[gendia_v1_hgvs_opts] Show the field ONLY if: [gendia_v1_report] = 'custom_hgvs'	Variant 1: What kind of change(s) would you like to report in "HGVS simple"-notation? Please enter all the details that you have available from the report - e.g. if the report lists both the cDNA and protein changes, please provide both.	checkbox, Required cdna | gendia_v1_hgvs_opts___cdna | Report cDNA change (c.) nucleotide | gendia_v1_hgvs_opts___nucleotide | Report nucleotide (genomic) change (g.) protein | gendia_v1_hgvs_opts___protein | Report protein change (p.)

			other	gendia_v1_hgvs_opts___other	Report other type of change, e.g. RNA (r.) or non-coding (n.)
492	[gendia_v1_refseq_c] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v1_hgvs_opts(cdna)] = 1	Section Header: *Variant 1: cDNA change* Reference sequence for cDNA change *Enter the RefSeq transcript ID with the prefix NM_ or NR_ etc., e.g. 'NM_173660.5'. More information and examples see here and here*			text, Required
493	[gendia_v1_desc_c] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v1_hgvs_opts(cdna)] = 1	Description of cDNA change *Enter the cDNA change in HGVS nomenclature, e.g. c.1124_1127dupTGCC or c.346G>A More information and examples see here*			text, Required
494	[gendia_v1_hgvs_c] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v1_hgvs_opts(cdna)] = 1	gendia_v1_hgvs_c			text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_v1_refseq_c]!='' and [gendia_v1_desc_c]!='', concat([gendia_v1_refseq_c], ':', [gendia_v1_desc_c]), ''))
495	[gendia_v1_hgvs_c_disp] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v1_hgvs_opts(cdna)] = 1	Resulting HGVS notation for variant 1 cDNA change:[gendia_v1_hgvs_c]			descriptive
496	[gendia_v1_refseq_g] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v1_hgvs_opts(nucleotide)] = 1	Section Header: *Variant 1: Nucleotide (genomic) change* Reference sequence for nucleotide (genomic) change *Enter the RefSeq transcript ID with the prefix NC_ or NG_ etc., e.g. 'NC_000023.10'. More information and examples see here and here*			text, Required
497	[gendia_v1_desc_g] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v1_hgvs_opts(nucleotide)] = 1	Description of nucleotide (genomic) change *Enter the nucleotide change in HGVS nomenclature, e.g. g.34805_34808dup More information and examples see here*			text, Required
498	[gendia_v1_hgvs_g] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v1_hgvs_opts(nucleotide)] = 1	gendia_v1_hgvs_g			text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_v1_refseq_g]!='' and [gendia_v1_desc_g]!='', concat([gendia_v1_refseq_g], ':', [gendia_v1_desc_g]), ''))
499	[gendia_v1_hgvs_g_disp] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v1_hgvs_opts(nucleotide)] = 1	Resulting HGVS notation for variant 1 nucleotide (genomic) change:[gendia_v1_hgvs_g]			descriptive
500	[gendia_v1_refseq_p] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v1_hgvs_opts(protein)] = 1	Section Header: *Variant 1: Protein change* Reference sequence for protein change *Enter the RefSeq transcript ID with the prefix NP_, e.g. NP_003997.1 More information and examples see here and here*			text, Required
501	[gendia_v1_desc_p] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v1_hgvs_opts(protein)] = 1	Description of protein change *Enter the protein change in HGVS nomenclature, e.g. p.Ala378fs More information and examples see here*			text, Required
502	[gendia_v1_hgvs_p] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v1_hgvs_opts(protein)] = 1	gendia_v1_hgvs_p			text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_v1_refseq_p]!='' and [gendia_v1_desc_p]!='', concat([gendia_v1_refseq_p], ':', [gendia_v1_desc_p]), ''))
503	[gendia_v1_hgvs_p_disp] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v1_hgvs_opts(protein)] = 1	Resulting HGVS notation for variant 1 protein change:[gendia_v1_hgvs_p]			descriptive
504	[gendia_v1_refseq_o] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v1_hgvs_opts(other)] = 1	Section Header: *Variant 1: Other change* Reference sequence for other change *Examples for RNA reference sequences: NC_000023.10(NM_004006.2) or LRG_199t1. More information and examples see here and here*			text, Required
505	[gendia_v1_desc_o]	Description of other change *Examples for descriptions of RNA changes: r.357_358ins357+1_357+12 or r.11u>g More information and examples see here*			text, Required

	Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v1_hgvs_opts(other)] = 1		
506	[gendia_v1_hgvs_o] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v1_hgvs_opts(other)] = 1	gendia_v1_hgvs_o	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_v1_refseq_o]!='' and [gendia_v1_desc_o]!='', concat([gendia_v1_refseq_o], ':', [gendia_v1_desc_o]), ''))
507	[gendia_v1_hgvs_o_disp] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v1_hgvs_opts(other)] = 1	Resulting HGVS notation for variant 1 other change:[gendia_v1_hgvs_o]	descriptive
508	[gendia_v1_zygos] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_report_causa_variant] = 1	Section Header: *Variant 1: Additional Information* Zygosity	dropdown heterozygous, Heterozygous compound_heterozygous, Compound heterozygous homozygous, Homozygous hemizygous, Hemizygous mosaic, Mosaic unknown, Unknown
509	[gendia_v1_gen_build] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_report_causa_variant] = 1 AND [gendia_v1_report] = 'custom_hgvs'	Genomic build	dropdown GRCh38, GRCh38 (hg38) GRCh37, GRCh37 (hg19)
510	[gendia_v1_gen_app] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_report_causa_variant] = 1 AND [gendia_v1_report] = 'custom_hgvs'	Genetic approach	dropdown single_gene_screening, Single Gene Screening acgh, aCGH snp_array, SNP array mlpa, MLPA karyotype, Karyotype ngs_panel, NGS panel wes, WES wgs, WGS
511	[gendia_v1_chrom] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_report_causa_variant] = 1 AND [gendia_v1_report] = 'custom_hgvs'	Chromosome	dropdown 1, 1 2, 2 3, 3 4, 4 5, 5 6, 6 7, 7 8, 8 9, 9 10, 10 11, 11 12, 12 13, 13 14, 14 15, 15 16, 16 17, 17

			18 | 18 19 | 19 20 | 20 21 | 21 22 | 22 X | X Y | Y MT | MT
512	[gendia_desc3] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_report_causa_variant] = 1	Section Header: *Variant 2 (if applicable)* Please report only an additional variant for the gene selected above (OMIM:[gendia_omim]). If you wish to report a variant for another gene, please report that in a separate 'Genetic Diagnosis'-form for that gene. Please enter all the details that you have available from the report. For autosomal recessive cases, you must enter both variant 1 and variant 2. If the variant is homozygous, enter the identical details twice. For autosomal dominant and X-linked, only one is required.	descriptive
513	[gendia_v2_yes] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_report_causa_variant] = 1	Report variant 2	yesno 1 | Yes 0 | No
514	[gendia_v2_report] Show the field ONLY if: [gendia_v2_yes] = 1	How would you like to describe variant 2?Note: We prefer NCBI ClinVar but if the change in question is not yet described there, you can give a dbSNP ID if available, or you can type in the protein or cDNA change given in the genetic report.	radio, Required clinvar | NCBI ClinVar dbsnp | dbSNP custom_hgvs | Custom HGVS-simple notation
515	[gendia_v2_clinvar_uri] Show the field ONLY if: [gendia_v2_report] = 'clinvar'	Variant 2: NCBI ClinVar URI(please provide website-link) *If the change is described in the NCBI ClinVar database, please provide website-link (e.g. https://www.ncbi.nlm.nih.gov/clinvar/variation/4886/)*	text, Required
516	[gendia_v2_dbsnp_id] Show the field ONLY if: [gendia_v2_report] = 'dbsnp'	Variant 2: dbSNP RS ID *If the change has a dbSNP ID, enter it here, e.g. rs606231128*	text, Required
517	[gendia_v2_hgvs_opts] Show the field ONLY if: [gendia_v2_report] = 'custom_hgvs'	Variant 2: What kind of change(s) would you like to report in "HGVS simple"-notation? Please enter all the details that you have available from the report - e.g. if the report lists both the cDNA and protein changes, please provide both.	checkbox, Required cdna | gendia_v2_hgvs_opts___cdna | Report cDNA change (c.) nucleotide | gendia_v2_hgvs_opts___nucleotide | Report nucleotide (genomic) change (g.) protein | gendia_v2_hgvs_opts___protein | Report protein change (p.) other | gendia_v2_hgvs_opts___other | Report other type of change, e.g. RNA (r.) or non-coding (n.)
518	[gendia_v2_refseq_c] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(cdna)] = 1	Section Header: *Variant 2: cDNA change* Reference sequence for cDNA change *Enter the RefSeq transcript ID with the prefix NM_ or NR_ etc., e.g. 'NM_173660.5'. More information and examples see here and here*	text, Required
519	[gendia_v2_desc_c] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(cdna)] = 1	Description of cDNA change *Enter the cDNA change in HGVS nomenclature, e.g. c.1124_1127dupTGCC or c.346G>A More information and examples see here*	text, Required
520	[gendia_v2_hgvs_c] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(cdna)] = 1	gendia_v1_hgvs_c	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_v2_refseq_c]!='' and [gendia_v2_desc_c]!='', concat([gendia_v2_refseq_c], ':', [gendia_v2_desc_c]), ''))
521	[gendia_v2_hgvs_c_disp] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(cdna)] = 1	Resulting HGVS notation for variant 2 cDNA change:[gendia_v2_hgvs_c]	descriptive
522	[gendia_v2_refseq_g] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(nucleotide)] = 1	Section Header: *Variant 2: Nucleotide (genomic) change* Reference sequence for nucleotide (genomic) change *Enter the RefSeq transcript ID with the prefix NC_ or NG_ etc., e.g. 'NC_000023.10'. More information and examples see here and here*	text, Required

523	[gendia_v2_desc_g] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(nucleotide)] = 1	Description of nucleotide (genomic) change *Enter the nucleotide change in HGVS nomenclature, e.g. g.34805_34808dup More information and examples see here*	text, Required
524	[gendia_v2_hgvs_g] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(nucleotide)] = 1	gendia_v2_hgvs_g	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_v2_refseq_g]!='' and [gendia_v2_desc_g]!='', concat([gendia_v2_refseq_g], ':', [gendia_v2_desc_g]), ''))
525	[gendia_v2_hgvs_g_disp] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(nucleotide)] = 1	Resulting HGVS notation for variant 2 nucleotide (genomic) change:[gendia_v2_hgvs_g]	descriptive
526	[gendia_v2_refseq_p] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(protein)] = 1	Section Header: *Variant 2: Protein change* Reference sequence for protein change *Enter the RefSeq transcript ID with the prefix NP_, e.g. NP_003997.1 More information and examples see here and here*	text, Required
527	[gendia_v2_desc_p] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(protein)] = 1	Description of protein change *Enter the protein change in HGVS nomenclature, e.g. p.Ala378fs More information and examples see here*	text, Required
528	[gendia_v2_hgvs_p] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(protein)] = 1	gendia_v2_hgvs_p	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_v2_refseq_p]!='' and [gendia_v2_desc_p]!='', concat([gendia_v2_refseq_p], ':', [gendia_v2_desc_p]), ''))
529	[gendia_v2_hgvs_p_disp] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(protein)] = 1	Resulting HGVS notation for variant 2 protein change:[gendia_v2_hgvs_p]	descriptive
530	[gendia_v2_refseq_o] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(other)] = 1	Section Header: *Variant 2: Other change* Reference sequence for other change *Examples for RNA reference sequences: NC_000023.10(NM_004006.2) or LRG_199t1. More information and examples see here and here*	text, Required
531	[gendia_v2_desc_o] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(other)] = 1	Description of other change *Examples for descriptions of RNA changes: r.357_358ins357+1_357+12 or r.11u>g More information and examples see here*	text, Required
532	[gendia_v2_hgvs_o] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(other)] = 1	gendia_v2_hgvs_o	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([gendia_v2_refseq_o]!='' and [gendia_v2_desc_o]!='', concat([gendia_v2_refseq_o], ':', [gendia_v2_desc_o]), ''))
533	[gendia_v2_hgvs_o_disp] Show the field ONLY if: [gendia_report_causa_variant]=1 AND [gendia_v2_hgvs_opts(other)] = 1	Resulting HGVS notation for variant 1 other change:[gendia_v2_hgvs_o]	descriptive
534	[gendia_v2_zygos] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_report_causa_variant] = 1 AND [gendia_v2_yes] = 1	Section Header: *Variant 2: Additional Information* Zygosity	dropdown heterozygous, Heterozygous compound_heterozygous, Compound heterozygous homozygous, Homozygous hemizygous, Hemizygous mosaic, Mosaic unknown, Unknown
535	[gendia_v2_gen_build] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_report_causa_variant]	Genomic build	dropdown GRCh38, GRCh38 (hg38) GRCh37, GRCh37 (hg19)

	= 1 AND [gendia_v2_report] = 'custom_hgvs'		
536	[gendia_v2_gen_app] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_report_causa_variant] = 1 AND [gendia_v2_report] = 'custom_hgvs'	Genetic approach	dropdown single_gene_screening, Single Gene Screening acgh, aCGH snp_array, SNP array mlpa, MLPA karyotype, Karyotype ngs_panel, NGS panel wes, WES wgs, WGS
537	[gendia_v2_chrom] Show the field ONLY if: [gendia_known] = 'yes' AND [gendia_report_causa_variant] = 1 AND [gendia_v2_report] = 'custom_hgvs'	Chromosome	dropdown 1, 1 2, 2 3, 3 4, 4 5, 5 6, 6 7, 7 8, 8 9, 9 10, 10 11, 11 12, 12 13, 13 14, 14 15, 15 16, 16 17, 17 18, 18 19, 19 20, 20 21, 21 22, 22 X, X Y, Y MT, MT
538	[genetic_diagnosis_changes_in_nuclear_dna_complete]	Section Header: *Form Status* Complete?	dropdown 0, Incomplete 1, Unverified 2, Complete
Instrument: **Genetic Diagnosis Changes in mitochondrial DNA** **(*genetic_diagnosis_changes_in_mitochondrial_dna*)**			
539	[mtdna_h1]	Genetic Diagnosis: Changes in mitochondrial DNA	descriptive
540	[date_of_mtdna]	Date of assessment yyyy-mm-dd	text (date_ymd), Required
541	[mtdna_desc1]	Please note: this form is only for recording changes in mitochondrial DNA (mtDNA). If you wish to report nuclear DNA variant(s), please use the "Genetic Diagnosis" form.	descriptive
542	[mtdna_label]	Genetic diagnosis name *This is a label to identify this record to the registry user. It will be displayed on the patient's record home page together with the date of assessment. You can name it anything you find helpful.*	text, Required

543	[mtdna_label_disp]	mtdna_label_disp	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(concat([mtdna_label], ' (', [date_of_mtdna], ')'))
544	[mtdna_change_type_known]	Type of mtDNA change known	radio yes, Yes no, No not_present, Not present Field Annotation: @DEFAULT = 'yes'
545	[mtdna_change_type] Show the field ONLY if: [mtdna_change_type_known] = 'yes'	Type of mtDNA change	checkbox, Required common_del, mtdna_change_type___common_del, Common deletion multiple_del, mtdna_change_type___multiple_del, Multiple deletions depletion, mtdna_change_type___depletion, Depletion
546	[mtdna_cnv]	Section Header: *mtDNA copy number variation* Copy number variation in mtDNA	radio yes, Yes not_present, Not present unknown, Unknown
547	[mtdna_cnv_where] Show the field ONLY if: [mtdna_cnv] = 'yes'	Copy number variation in mtDNA detected	checkbox muscle, mtdna_cnv_where___muscle, in muscle blood, mtdna_cnv_where___blood, in blood other, mtdna_cnv_where___other, other tissue
548	[mtdna_cnv_plasmic] Show the field ONLY if: [mtdna_cnv] = 'yes'	State of copy number variation in mtDNA	radio homoplasmic, Homoplasmic heteroplasmic, Heteroplasmic unknown, Unknown
549	[mtdna_snp]	Section Header: *mtDNA SNP variant* mtDNA SNP variant	radio yes, Yes not_present, Not present unknown, Unknown
550	[mtdna_snp_where] Show the field ONLY if: [mtdna_snp] = 'yes'	mtDNA SNP variant detected	checkbox muscle, mtdna_snp_where___muscle, in muscle blood, mtdna_snp_where___blood, in blood other, mtdna_snp_where___other, other tissue
551	[mtdna_snp_plasmic] Show the field ONLY if: [mtdna_snp] = 'yes'	State of mtDNA SNP variant	radio homoplasmic, Homoplasmic heteroplasmic, Heteroplasmic unknown, Unknown
552	[mtdna_snp_report] Show the field ONLY if: [mtdna_snp] = 'yes'	How would you like to describe this mtDNA SNP variant?Note: We prefer NCBI ClinVar but if the change in question is not yet described there, you can type in the mtDNA change given in the genetic report.	radio, Required clinvar, NCBI ClinVar custom_hgvs, Custom HGVS-simple notation
553	[mtdna_clinvar_uri] Show the field ONLY if: [mtdna_snp_report] = 'clinvar'	mtDNA variant: NCBI ClinVar URI(please provide website-link) *If the change is described in the NCBI ClinVar database, please provide website-link (e.g. https://www.ncbi.nlm.nih.gov/clinvar/variation/689805/)*	text, Required
554	[mtdna_variant] Show the field ONLY if: [mtdna_snp_report] = 'custom_hgvs'	Description of mtDNA change based on the Revised Cambridge Reference Sequence (rCRS) of the Human Mitochondrial DNA: NC_012920.1(MT-CYB) *Enter the mtDNA change in HGVS nomenclature, e.g. m.5781>C More information and examples see here*	text, Required
555	[mtdna_hgvs] Show the field ONLY if: [mtdna_snp_report] = 'custom_hgvs'	mtdna_hgvs	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([mtdna_variant]!='', concat('NC_012920 [mtdna_variant]), ''))

556	[mtdna_hgvs_disp] Show the field ONLY if: [mtdna_snp_report] = 'custom_hgvs'	Resulting HGVS notation for mtDNA change:[mtdna_hgvs]	descriptive
557	[genetic_diagnosis_changes_in_mitochondrial_dna_complete]	Section Header: *Form Status* Complete?	dropdown 0, Incomplete 1, Unverified 2, Complete
Instrument:* Negative Genetic Tests (only when no genetic diagnosis) *(negative_genetic_tests_only_when_no_genetic_diagno)			
558	[neggentests_h1]	Negative Genetic Tests (only when no genetic diagnosis)	descriptive
559	[gps_done]	Section Header: *Gene Panel Sequencing (GPS)* Gene Panel Sequencing (GPS) done	radio yes, Yes no, No unknown, Unknown
560	[date_of_gps] Show the field ONLY if: [gps_done] = 'yes'	Date of Gene Panel Sequencing (GPS) yyyy-mm-dd *If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.*	text (date_ymd)
561	[gps_panel_name] Show the field ONLY if: [gps_done] = 'yes'	Name of Gene Panel	text
562	[gps_n_excluded] Show the field ONLY if: [gps_done] = 'yes'	Number of Genes excluded by GPS	text (integer)
563	[wes_done]	Section Header: *Whole Exome Sequencing (WES)* Whole Exome Sequencing (WES) done	radio yes, Yes no, No unknown, Unknown
564	[date_of_wes] Show the field ONLY if: [wes_done] = 'yes'	Date of Whole Exome Sequencing (WES) yyyy-mm-dd *If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.*	text (date_ymd)
565	[wgs_done]	Section Header: *Whole-Genome Sequencing (WGS)* Whole-Genome Sequencing (WGS) done	radio yes, Yes no, No unknown, Unknown
566	[date_of_wgs] Show the field ONLY if: [wgs_done] = 'yes'	Date of Whole-Genome Sequencing (WGS) yyyy-mm-dd *If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.*	text (date_ymd)
567	[neggentests_comment]	Section Header: *Comments* Comments about negative genetic tests	notes
568	[gps_label_disp]	gps_label_disp	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([gps_done]), concat('GPS: ', [gps_done], if(!isblankormissingcode([date_of_gps]), concat(' (', [date_of_gps], ')'), ''), ''), ''))
569	[wes_label_disp]	wes_label_disp	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([wes_done]), concat('WES: ', [wes_done], if(!isblankormissingcode([date_of_wes]), concat(' (', [date_of_wes], ')'), ''), ''), ''))
570	[wgs_label_disp]	wgs_label_disp	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if(!isblankormissingcode([wgs_done]), concat('WGS: ', [wgs_done], if(!isblankormissingcode([date_of_wgs]), concat(' (', [date_of_wgs], ')'), ''), ''), ''))
571	[ngt_label_disp]	ngt_label_disp	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(concat(if(!isblankormissingcode([gps_label_disp]), [gps_label_disp], 'GPS: missing'), if(!isblankormissingcode([wes_label_disp]), [wes_label_disp], 'WES: missing'), '; ',

			if(!isblankormissingcode([wgs_label_disp]), [wgs_label_disp], 'WGS: missing')))
572	[negative_genetic_tests_only_when_no_genetic_diagno_complete]	Section Header: *Form Status* Complete?	dropdown 0, Incomplete 1, Unverified 2, Complete
Instrument:* Undiagnosed *(undiagnosed)			
573	[undiag_h1]	UNDIAGNOSED	descriptive
574	[undiag_desc1]	For undiagnosed patients: Please enter a combination of a phenotype and a genetic variant that cannot be definitively diagnosed.	descriptive
575	[date_of_undiag]	Date of assessment yyyy-mm-dd	text (date_ymd), Required
576	[date_of_undiag_2]	helper-field for generation of triples. contains same value as date_of_undiag but will only be filled if a second variant is reported.	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v2_yes]='yes', [date_of_undiag], ''))
577	[undiag_label]	Label for findings *This is a label to identify this record to the registry user. It will be displayed on the patient's record home page together with the date of assessment. You can name it anything you find helpful.*	text, Required
578	[undiag_label_disp]	undiag_label_disp	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(concat([undiag_label], ' (', [date_of_undiag], ')'))
579	[undiag_pheno_hpo]	Section Header: *PHENOTYPE* Phenotype: HPO-code *Element 6.3. in the Set of common data elements for Rare Diseases Registration (link)*	text, Required BIOPORTAL:HP, BIOPORTAL:HP
580	[undiag_pheno_hpo_2]	helper-field for generation of triples. contains same value as undiag_pheno_hpo but will only be filled if a second variant is reported.	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v2_yes]='yes', [undiag_pheno_hpo], ''))
581	[undiag_desc2]	Section Header: *GENOTYPE* This section is intended to record the change(s) in DNA, RNA or protein sequences according to the standards of the Sequence Variant Nomenclature "HGVS simple" (accessed: 18.13.22). HGVS-codes can be checked with Mutalyzer (accessed: 18.03.22). Please enter all the details that you have available from the report. For autosomal recessice cases, you must enter both variant 1 and variant 2. If the variant is homocygous, enter the identical details twice. For autosomal dominant and X-linked, only one is required.	descriptive
582	[undiag_report_variant]	Report genetic variant(s)	radio yes, Yes unknown, Unknown not_applicable, Not applicable not_present, Not present
583	[undiag_listpref] Show the field ONLY if: [undiag_report_variant] = 'yes'	Gene list preference *Element 6.3. in the Set of common data elements for Rare Diseases Registration (link)*	radio all_nmd, Search gene in full list for neuromuscular diseases ontology, If gene not in provided lists: Search gene in biomedical ontologies
584	[undiag_omim_armx] Show the field ONLY if: [undiag_report_variant] = 'yes' AND [undiag_listpref]='all_nmd'	Gene Symbol (HGNC) for all neuromuscular diseases *Element 6.3. in the Set of common data elements for Rare Diseases Registration (link)*	dropdown (autocomplete) 601065, AARS1 / HGNC:20 / OMIM:601065 (alanyl-tRNA synthetase 1) 612035, AARS2 / HGNC:21022 / OMIM:612035 (alanyl-tRNA synthetase 2, mitochondrial) 601439, ABCC9 / HGNC:60 / OMIM:601439 (ATP binding cassette subfamily C member 9) 604780, ABHD5 / HGNC:21396 / OMIM:604780 (abhydrolase domain containing 5, lysophosphatidic acid acyltransferase) 611103, ACAD9 / HGNC:21497 / OMIM:611103 (acyl-CoA dehydrogenase family member 9) 609575, ACADVL / HGNC:92 / OMIM:609575 (acyl-CoA dehydrogenase very long chain) 617036, ACER3 / HGNC:16066 / OMIM:617036 (alkaline ceramidase 3) 102610, ACTA1 / HGNC:129 / OMIM:102610 (actin alpha 1, skeletal muscle) 102540, ACTC1 / HGNC:143 / OMIM:102540 (actin alpha cardiac muscle 1) 102573, ACTN2 / HGNC:164 / OMIM:102573 (actinin alpha 2) 102576, ACVR1 / HGNC:171 / OMIM:102576 (activin A receptor type 1) 606980, COQ8A / HGNC:16812 / OMIM:606980 (coenzyme Q8A)

600294	ADCY6 / HGNC:237 / OMIM:600294 (adenylate cyclase 6)
612243	ADGRG6 / HGNC:13841 / OMIM:612243 (adhesion G protein-coupled receptor G6)
612498	ADSS1 / HGNC:20093 / OMIM:612498 (adenylosuccinate synthase 1)
604581	AFG3L2 / HGNC:315 / OMIM:604581 (AFG3 like matrix AAA peptidase subunit 2)
610860	AGL / HGNC:321 / OMIM:610860 (amylo-alpha-1, 6-glucosidase, 4-alpha-glucanotransferase)
103320	AGRN / HGNC:329 / OMIM:103320 (agrin)
606830	AGTPBP1 / HGNC:17258 / OMIM:606830 (ATP/GTP binding carboxypeptidase 1)
608570	AHNAK2 / HGNC:20125 / OMIM:608570 (AHNAK nucleoprotein 2)
300169	AIFM1 / HGNC:8768 / OMIM:300169 (apoptosis inducing factor mitochondria associated 1)
604001	AKAP9 / HGNC:379 / OMIM:604001 (A-kinase anchoring protein 9)
138250	ALDH18A1 / HGNC:9722 / OMIM:138250 (aldehyde dehydrogenase 18 family member A1)
609523	ALDH3A2 / HGNC:403 / OMIM:609523 (aldehyde dehydrogenase 3 family member A2)
300776	ALG13 / HGNC:30881 / OMIM:300776 (ALG13 UDP-N-acetylglucosaminyltransferase subunit)
612866	ALG14 / HGNC:28287 / OMIM:612866 (ALG14 UDP-N-acetylglucosaminyltransferase subunit)
607905	ALG2 / HGNC:23159 / OMIM:607905 (ALG2 alpha-1,3/1,6-mannosyltransferase)
617608	ALPK3 / HGNC:17574 / OMIM:617608 (alpha kinase 3)
606352	ALS2 / HGNC:443 / OMIM:606352 (alsin Rho guanine nucleotide exchange factor ALS2)
102771	AMPD2 / HGNC:469 / OMIM:102771 (adenosine monophosphate deaminase 2)
105850	ANG / HGNC:483 / OMIM:105850 (angiogenin)
106410	ANK2 / HGNC:493 / OMIM:106410 (ankyrin 2)
609599	ANKRD1 / HGNC:15819 / OMIM:609599 (ankyrin repeat domain 1)
613726	ANO10 / HGNC:25519 / OMIM:613726 (anoctamin 10)
608662	ANO5 / HGNC:27337 / OMIM:608662 (anoctamin 5)
602572	ANXA11 / HGNC:535 / OMIM:602572 (annexin A11)
607245	AP4B1 / HGNC:572 / OMIM:607245 (adaptor related protein complex 4 subunit beta 1)
607244	AP4E1 / HGNC:573 / OMIM:607244 (adaptor related protein complex 4 subunit epsilon 1)
602296	AP4M1 / HGNC:574 / OMIM:602296 (adaptor related protein complex 4 subunit mu 1)
607243	AP4S1 / HGNC:575 / OMIM:607243 (adaptor related protein complex 4 subunit sigma 1)
613653	AP5Z1 / HGNC:22197 / OMIM:613653 (adaptor related protein complex 5 subunit zeta 1)
606350	APTX / HGNC:15984 / OMIM:606350 (aprataxin)
313700	AR / HGNC:644 / OMIM:313700 (androgen receptor)
608136	ARHGEF10 / HGNC:14103 / OMIM:608136 (Rho guanine nucleotide exchange factor 10)
607669	ARL6IP1 / HGNC:697 / OMIM:607669 (ADP ribosylation factor like GTPase 6 interacting protein 1)
613468	ASAH1 / HGNC:735 / OMIM:613468 (N-acylsphingosine amidohydrolase 1)
614215	ASCC1 / HGNC:24268 / OMIM:614215 (activating signal cointegrator 1 complex subunit 1)
604261	ATG5 / HGNC:589 / OMIM:604261 (autophagy related 5)
606439	ATL1 / HGNC:11231 / OMIM:606439 (atlastin GTPase 1)
609369	ATL3 / HGNC:24526 / OMIM:609369 (atlastin GTPase 3)
607585	ATM / HGNC:795 / OMIM:607585 (ATM serine/threonine kinase)
610513	ATP13A2 / HGNC:30213 / OMIM:610513 (ATPase cation transporting 13A2)
182310	ATP1A1 / HGNC:799 / OMIM:182310 (ATPase Na+/K+ transporting subunit alpha 1)
182340	ATP1A2 / HGNC:800 / OMIM:182340 (ATPase Na+/K+ transporting subunit alpha 2)
108730	ATP2A1 / HGNC:811 / OMIM:108730 (ATPase sarcoplasmic/endoplasmic reticulum Ca2+ transporting 1)
300011	ATP7A / HGNC:869 / OMIM:300011 (ATPase copper transporting alpha)
601556	ATXN1 / HGNC:10548 / OMIM:601556 (ataxin 1)
611150	ATXN10 / HGNC:10549 / OMIM:611150 (ataxin 10)

601517	ATXN2 / HGNC:10555 / OMIM:601517 (ataxin 2)
607047	ATXN3 / HGNC:7106 / OMIM:607047 (ataxin 3)
607640	ATXN7 / HGNC:10560 / OMIM:607640 (ataxin 7)
603680	ATXN8OS / HGNC:10561 / OMIM:603680 (ATXN8 opposite strand lncRNA)
610194	B3GALNT2 / HGNC:28596 / OMIM:610194 (beta-1,3-N-acetylgalactosaminyltransferase 2)
605517	B4GAT1 / HGNC:15685 / OMIM:605517 (beta-1,4-glucuronyltransferase 1)
601873	B4GALNT1 / HGNC:4117 / OMIM:601873 (beta-1,4-N-acetyl-galactosaminyltransferase 1)
603883	BAG3 / HGNC:939 / OMIM:603883 (BAG cochaperone 3)
612051	BEAN1 / HGNC:24160 / OMIM:612051 (brain expressed associated with NEDD4 1)
609797	BICD2 / HGNC:17208 / OMIM:609797 (BICD cargo adaptor 2)
601248	BIN1 / HGNC:1052 / OMIM:601248 (bridging integrator 1)
606158	BSCL2 / HGNC:15832 / OMIM:606158 (BSCL2 lipid droplet biogenesis associated, seipin)
604577	BVES / HGNC:1152 / OMIM:604577 (blood vessel epicardial substance)
613541	MTRFR / HGNC:26784 / OMIM:613541 (mitochondrial translation release factor in rescue)
614297	C19orf12 / HGNC:25443 / OMIM:614297 (chromosome 19 open reading frame 12)
618682	C1orf194 / HGNC:32331 / OMIM:618682 (chromosome 1 open reading frame 194)
601269	C1QBP / HGNC:1243 / OMIM:601269 (complement C1q binding protein)
614260	C9orf72 / HGNC:28337 / OMIM:614260 (C9orf72-SMCR8 complex subunit)
601011	CACNA1A / HGNC:1388 / OMIM:601011 (calcium voltage-gated channel subunit alpha1 A)
114205	CACNA1C / HGNC:1390 / OMIM:114205 (calcium voltage-gated channel subunit alpha1 C)
604065	CACNA1G / HGNC:1394 / OMIM:604065 (calcium voltage-gated channel subunit alpha1 G)
607904	CACNA1H / HGNC:1395 / OMIM:607904 (calcium voltage-gated channel subunit alpha1 H)
114208	CACNA1S / HGNC:1397 / OMIM:114208 (calcium voltage-gated channel subunit alpha1 S)
600003	CACNB2 / HGNC:1402 / OMIM:600003 (calcium voltage-gated channel auxiliary subunit beta 2)
601949	CACNB4 / HGNC:1404 / OMIM:601949 (calcium voltage-gated channel auxiliary subunit beta 4)
114180	CALM1 / HGNC:1442 / OMIM:114180 (calmodulin 1)
114182	CALM2 / HGNC:1445 / OMIM:114182 (calmodulin 2)
611414	CALR3 / HGNC:20407 / OMIM:611414 (calreticulin 3)
114220	CAPN1 / HGNC:1476 / OMIM:114220 (calpain 1)
114240	CAPN3 / HGNC:1480 / OMIM:114240 (calpain 3)
114250	CASQ1 / HGNC:1512 / OMIM:114250 (calsequestrin 1)
114251	CASQ2 / HGNC:1513 / OMIM:114251 (calsequestrin 2)
601253	CAV3 / HGNC:1529 / OMIM:601253 (caveolin 3)
603198	CAVIN1 / HGNC:9688 / OMIM:603198 (caveolae associated protein 1)
617714	CAVIN4 / HGNC:33742 / OMIM:617714 (caveolae associated protein 4)
614666	CCDC78 / HGNC:14153 / OMIM:614666 (coiled-coil domain containing 78)
611204	CCDC88C / HGNC:19967 / OMIM:611204 (coiled-coil domain containing 88C)
610150	CCT5 / HGNC:1618 / OMIM:610150 (chaperonin containing TCP1 subunit 5)
601443	CFL2 / HGNC:1875 / OMIM:601443 (cofilin 2)
118490	CHAT / HGNC:1912 / OMIM:118490 (choline O-acetyltransferase)
615903	CHCHD10 / HGNC:15559 / OMIM:615903 (coiled-coil-helix-coiled-coil-helix domain containing 10)
612395	CHKB / HGNC:1938 / OMIM:612395 (choline kinase beta)
609512	CHMP2B / HGNC:24537 / OMIM:609512 (charged multivesicular body protein 2B)
606988	CHP1 / HGNC:17433 / OMIM:606988 (calcineurin like EF-hand protein 1)
100690	CHRNA1 / HGNC:1955 / OMIM:100690 (cholinergic receptor nicotinic alpha 1 subunit)

100710	CHRNB1 / HGNC:1961 / OMIM:100710 (cholinergic receptor nicotinic beta 1 subunit)
100720	CHRND / HGNC:1965 / OMIM:100720 (cholinergic receptor nicotinic delta subunit)
100725	CHRNE / HGNC:1966 / OMIM:100725 (cholinergic receptor nicotinic epsilon subunit)
100730	CHRNG / HGNC:1967 / OMIM:100730 (cholinergic receptor nicotinic gamma subunit)
118425	CLCN1 / HGNC:2019 / OMIM:118425 (chloride voltage-gated channel 1)
607042	CLN3 / HGNC:2074 / OMIM:607042 (CLN3 lysosomal/endosomal transmembrane protein, battenin)
601273	CLTCL1 / HGNC:2093 / OMIM:601273 (clathrin heavy chain like 1)
116955	CNBP / HGNC:13164 / OMIM:116955 (CCHC-type zinc finger nucleic acid binding protein)
600016	CNTN1 / HGNC:2171 / OMIM:600016 (contactin 1)
602346	CNTNAP1 / HGNC:8011 / OMIM:602346 (contactin associated protein 1)
615623	COA7 / HGNC:25716 / OMIM:615623 (cytochrome c oxidase assembly factor 7)
120320	COL12A1 / HGNC:2188 / OMIM:120320 (collagen type XII alpha 1 chain)
120350	COL13A1 / HGNC:2190 / OMIM:120350 (collagen type XIII alpha 1 chain)
610004	COL25A1 / HGNC:18603 / OMIM:610004 (collagen type XXV alpha 1 chain)
120215	COL5A1 / HGNC:2209 / OMIM:120215 (collagen type V alpha 1 chain)
120220	COL6A1 / HGNC:2211 / OMIM:120220 (collagen type VI alpha 1 chain)
120240	COL6A2 / HGNC:2212 / OMIM:120240 (collagen type VI alpha 2 chain)
120250	COL6A3 / HGNC:2213 / OMIM:120250 (collagen type VI alpha 3 chain)
603033	COLQ / HGNC:2226 / OMIM:603033 (collagen like tail subunit of asymmetric acetylcholinesterase)
609825	COQ2 / HGNC:25223 / OMIM:609825 (coenzyme Q2, polyprenyltransferase)
612898	COQ4 / HGNC:19693 / OMIM:612898 (coenzyme Q4)
614647	COQ6 / HGNC:20233 / OMIM:614647 (coenzyme Q6, monooxygenase)
601683	COQ7 / HGNC:2244 / OMIM:601683 (coenzyme Q7, hydroxylase)
612837	COQ9 / HGNC:25302 / OMIM:612837 (coenzyme Q9)
603646	COX15 / HGNC:2263 / OMIM:603646 (cytochrome c oxidase assembly homolog COX15)
602072	COX6A1 / HGNC:2277 / OMIM:602072 (cytochrome c oxidase subunit 6A1)
602009	COX6A2 / HGNC:2279 / OMIM:602009 (cytochrome c oxidase subunit 6A2)
608846	CPT1C / HGNC:18540 / OMIM:608846 (carnitine palmitoyltransferase 1C)
600650	CPT2 / HGNC:2330 / OMIM:600650 (carnitine palmitoyltransferase 2)
614631	CRPPA / HGNC:37276 / OMIM:614631 (CDP-L-ribitol pyrophosphorylase A)
123590	CRYAB / HGNC:2389 / OMIM:123590 (crystallin alpha B)
600824	CSRP3 / HGNC:2472 / OMIM:600824 (cysteine and glycine rich protein 3)
604927	CTDP1 / HGNC:2498 / OMIM:604927 (CTD phosphatase subunit 1)
607667	CTNNA3 / HGNC:2511 / OMIM:607667 (catenin alpha 3)
616120	CWF19L1 / HGNC:25613 / OMIM:616120 (CWF19 like cell cycle control factor 1)
610670	CYP2U1 / HGNC:20582 / OMIM:610670 (cytochrome P450 family 2 subfamily U member 1)
603711	CYP7B1 / HGNC:2652 / OMIM:603711 (cytochrome P450 family 7 subfamily B member 1)
128239	DAG1 / HGNC:2666 / OMIM:128239 (dystroglycan 1)
615820	DCAF8 / HGNC:24891 / OMIM:615820 (DDB1 and CUL4 associated factor 8)
601143	DCTN1 / HGNC:2711 / OMIM:601143 (dynactin subunit 1)
614603	DDHD1 / HGNC:19714 / OMIM:614603 (DDHD domain containing 1)
615003	DDHD2 / HGNC:29106 / OMIM:615003 (DDHD domain containing 2)
125660	DES / HGNC:2770 / OMIM:125660 (desmin)
606983	DGAT2 / HGNC:16940 / OMIM:606983 (diacylglycerol O-acyltransferase 2)
601465	DGUOK / HGNC:2858 / OMIM:601465 (deoxyguanosine kinase)

614984	DHTKD1 / HGNC:23537 / OMIM:614984 (dehydrogenase E1 and transketolase domain containing 1)
300377	DMD / HGNC:2928 / OMIM:300377 (dystrophin)
605377	DMPK / HGNC:2933 / OMIM:605377 (DM1 protein kinase)
601810	DNA2 / HGNC:2939 / OMIM:601810 (DNA replication helicase/nuclease 2)
604139	DNAJB2 / HGNC:5228 / OMIM:604139 (DnaJ heat shock protein family (Hsp40) member B2)
611332	DNAJB6 / HGNC:14888 / OMIM:611332 (DnaJ heat shock protein family (Hsp40) member B6)
602378	DNM2 / HGNC:2974 / OMIM:602378 (dynamin 2)
126375	DNMT1 / HGNC:2976 / OMIM:126375 (DNA methyltransferase 1)
610285	DOK7 / HGNC:26594 / OMIM:610285 (docking protein 7)
610746	DOLK / HGNC:23406 / OMIM:610746 (dolichol kinase)
191350	DPAGT1 / HGNC:2995 / OMIM:191350 (dolichyl-phosphate N-acetylglucosaminephosphotransferase 1)
603503	DPM1 / HGNC:3005 / OMIM:603503 (dolichyl-phosphate mannosyltransferase subunit 1, catalytic)
603564	DPM2 / HGNC:3006 / OMIM:603564 (dolichyl-phosphate mannosyltransferase subunit 2, regulatory)
605951	DPM3 / HGNC:3007 / OMIM:605951 (dolichyl-phosphate mannosyltransferase subunit 3, regulatory)
125645	DSC2 / HGNC:3036 / OMIM:125645 (desmocollin 2)
125671	DSG2 / HGNC:3049 / OMIM:125671 (desmoglein 2)
125647	DSP / HGNC:3052 / OMIM:125647 (desmoplakin)
113810	DST / HGNC:1090 / OMIM:113810 (dystonin)
601239	DTNA / HGNC:3057 / OMIM:601239 (dystrobrevin alpha)
606009	DUX4L1 / HGNC:3082 / OMIM:606009 (double homeobox 4 like 1 (pseudogene))
600112	DYNC1H1 / HGNC:2961 / OMIM:600112 (dynein cytoplasmic 1 heavy chain 1)
603009	DYSF / HGNC:3097 / OMIM:603009 (dysferlin)
130610	EEF2 / HGNC:3214 / OMIM:130610 (eukaryotic translation elongation factor 2)
129010	EGR2 / HGNC:3239 / OMIM:129010 (early growth response 2)
605512	ELOVL4 / HGNC:14415 / OMIM:605512 (ELOVL fatty acid elongase 4)
611805	ELOVL5 / HGNC:21308 / OMIM:611805 (ELOVL fatty acid elongase 5)
603722	ELP1 / HGNC:5959 / OMIM:603722 (elongator acetyltransferase complex subunit 1)
300384	EMD / HGNC:3331 / OMIM:300384 (emerin)
131370	ENO3 / HGNC:3354 / OMIM:131370 (enolase 3)
601752	ENTPD1 / HGNC:3363 / OMIM:601752 (ectonucleoside triphosphate diphosphohydrolase 1)
190151	ERBB3 / HGNC:3431 / OMIM:190151 (erb-b2 receptor tyrosine kinase 3)
600543	ERBB4 / HGNC:3432 / OMIM:600543 (erb-b2 receptor tyrosine kinase 4)
611604	ERLIN1 / HGNC:16947 / OMIM:611604 (ER lipid raft associated 1)
611605	ERLIN2 / HGNC:1356 / OMIM:611605 (ER lipid raft associated 2)
608053	ETFA / HGNC:3481 / OMIM:608053 (electron transfer flavoprotein subunit alpha)
231675	ETFDH / HGNC:3483 / OMIM:231675 (electron transfer flavoprotein dehydrogenase)
606489	EXOSC3 / HGNC:17944 / OMIM:606489 (exosome component 3)
606019	EXOSC8 / HGNC:17035 / OMIM:606019 (exosome component 8)
603550	EYA4 / HGNC:3522 / OMIM:603550 (EYA transcriptional coactivator and phosphatase 4)
611026	FA2H / HGNC:21197 / OMIM:611026 (fatty acid 2-hydroxylase)
615584	FAM111B / HGNC:24200 / OMIM:615584 (FAM111 trypsin like peptidase B)
611592	FARS2 / HGNC:21062 / OMIM:611592 (phenylalanyl-tRNA synthetase 2, mitochondrial)
612322	FASTKD2 / HGNC:29160 / OMIM:612322 (FAST kinase domains 2)
604269	FAT2 / HGNC:3596 / OMIM:604269 (FAT atypical cadherin 2)

604580	FBLN5 / HGNC:3602 / OMIM:604580 (fibulin 5)
605654	FBXL4 / HGNC:13601 / OMIM:605654 (F-box and leucine rich repeat protein 4)
608533	FBXO38 / HGNC:28844 / OMIM:608533 (F-box protein 38)
614585	FDX2 / HGNC:30546 / OMIM:614585 (ferredoxin 2)
611104	FGD4 / HGNC:19125 / OMIM:611104 (FYVE, RhoGEF and PH domain containing 4)
601515	FGF14 / HGNC:3671 / OMIM:601515 (fibroblast growth factor 14)
300163	FHL1 / HGNC:3702 / OMIM:300163 (four and a half LIM domains 1)
609390	FIG4 / HGNC:16873 / OMIM:609390 (FIG4 phosphoinositide 5-phosphatase)
606596	FKRP / HGNC:17997 / OMIM:606596 (fukutin related protein)
607440	FKTN / HGNC:3622 / OMIM:607440 (fukutin)
610595	FLAD1 / HGNC:24671 / OMIM:610595 (flavin adenine dinucleotide synthetase 1)
300017	FLNA / HGNC:3754 / OMIM:300017 (filamin A)
102565	FLNC / HGNC:3756 / OMIM:102565 (filamin C)
609144	FLVCR1 / HGNC:24682 / OMIM:609144 (FLVCR heme transporter 1)
601278	FRG1 / HGNC:3954 / OMIM:601278 (FSHD region gene 1)
137070	FUS / HGNC:4010 / OMIM:137070 (FUS RNA binding protein)
606829	FXN / HGNC:3951 / OMIM:606829 (frataxin)
600819	FXR1 / HGNC:4023 / OMIM:600819 (FMR1 autosomal homolog 1)
606800	GAA / HGNC:4065 / OMIM:606800 (alpha glucosidase)
605379	GAN / HGNC:4137 / OMIM:605379 (gigaxonin)
600287	GARS1 / HGNC:4162 / OMIM:600287 (glycyl-tRNA synthetase 1)
614518	GATAD1 / HGNC:29941 / OMIM:614518 (GATA zinc finger domain containing 1)
606463	GBA / HGNC:4177 / OMIM:606463 (glucosylceramidase beta)
609471	GBA2 / HGNC:18986 / OMIM:609471 (glucosylceramidase beta 2)
607839	GBE1 / HGNC:4180 / OMIM:607839 (1,4-alpha-glucan branching enzyme 1)
603698	GBF1 / HGNC:4181 / OMIM:603698 (golgi brefeldin A resistant guanine nucleotide exchange factor 1)
606598	GDAP1 / HGNC:15968 / OMIM:606598 (ganglioside induced differentiation associated protein 1)
618128	GDAP2 / HGNC:18010 / OMIM:618128 (ganglioside induced differentiation associated protein 2)
138292	GFPT1 / HGNC:4241 / OMIM:138292 (glutamine--fructose-6-phosphate transaminase 1)
605072	GIPC1 / HGNC:1226 / OMIM:605072 (GIPC PDZ domain containing family member 1)
121013	GJA5 / HGNC:4279 / OMIM:121013 (gap junction protein alpha 5)
304040	GJB1 / HGNC:4283 / OMIM:304040 (gap junction protein beta 1)
603324	GJB3 / HGNC:4285 / OMIM:603324 (gap junction protein beta 3)
608803	GJC2 / HGNC:17494 / OMIM:608803 (gap junction protein gamma 2)
608603	GLDN / HGNC:29514 / OMIM:608603 (gliomedin)
603371	GLE1 / HGNC:4315 / OMIM:603371 (GLE1 RNA export mediator)
615320	GMPPB / HGNC:22932 / OMIM:615320 (GDP-mannose pyrophosphorylase B)
610863	GNB4 / HGNC:20731 / OMIM:610863 (G protein subunit beta 4)
603824	GNE / HGNC:23657 / OMIM:603824 (glucosamine (UDP-N-acetyl)-2-epimerase/N-acetylmannosamine kinase)
602580	GOLGA2 / HGNC:4425 / OMIM:602580 (golgin A2)
604027	GOSR2 / HGNC:4431 / OMIM:604027 (golgi SNAP receptor complex member 2)
611778	GPD1L / HGNC:28956 / OMIM:611778 (glycerol-3-phosphate dehydrogenase 1 like)
602368	GRID2 / HGNC:4576 / OMIM:602368 (glutamate ionotropic receptor delta type subuni
604473	GRM1 / HGNC:4593 / OMIM:604473 (glutamate metabotropic receptor 1)

603942	GYG1 / HGNC:4699 / OMIM:603942 (glycogenin 1)
138570	GYS1 / HGNC:4706 / OMIM:138570 (glycogen synthase 1)
610467	HACD1 / HGNC:9639 / OMIM:610467 (3-hydroxyacyl-CoA dehydratase 1)
610876	HACE1 / HGNC:21033 / OMIM:610876 (HECT domain and ankyrin repeat containing E3 ubiquitin protein ligase 1)
142810	HARS1 / HGNC:4816 / OMIM:142810 (histidyl-tRNA synthetase 1)
605206	HCN4 / HGNC:16882 / OMIM:605206 (hyperpolarization activated cyclic nucleotide gated potassium channel 4)
606873	HEXB / HGNC:4879 / OMIM:606873 (hexosaminidase subunit beta)
601314	HINT1 / HGNC:4912 / OMIM:601314 (histidine triad nucleotide binding protein 1)
142600	HK1 / HGNC:4922 / OMIM:142600 (hexokinase 1)
164017	HNRNPA1 / HGNC:5031 / OMIM:164017 (heterogeneous nuclear ribonucleoprotein A1)
600124	HNRNPA2B1 / HGNC:5033 / OMIM:600124 (heterogeneous nuclear ribonucleoprotein A2/B1)
607137	HNRNPDL / HGNC:5037 / OMIM:607137 (heterogeneous nuclear ribonucleoprotein D like)
142984	HOXD10 / HGNC:5133 / OMIM:142984 (homeobox D10)
190020	HRAS / HGNC:5173 / OMIM:190020 (HRas proto-oncogene, GTPase)
602195	HSPB1 / HGNC:5246 / OMIM:602195 (heat shock protein family B (small) member 1)
604624	HSPB3 / HGNC:5248 / OMIM:604624 (heat shock protein family B (small) member 3)
608014	HSPB8 / HGNC:30171 / OMIM:608014 (heat shock protein family B (small) member 8)
118190	HSPD1 / HGNC:5261 / OMIM:118190 (heat shock protein family D (Hsp60) member 1)
142461	HSPG2 / HGNC:5273 / OMIM:142461 (heparan sulfate proteoglycan 2)
615316	IBA57 / HGNC:27302 / OMIM:615316 (iron-sulfur cluster assembly factor IBA57)
603502	IFRD1 / HGNC:5456 / OMIM:603502 (interferon related developmental regulator 1)
600502	IGHMBP2 / HGNC:5542 / OMIM:600502 (immunoglobulin mu DNA binding protein 2)
602366	ILK / HGNC:6040 / OMIM:602366 (integrin linked kinase)
610982	INF2 / HGNC:23791 / OMIM:610982 (inverted formin 2)
607875	INPP5K / HGNC:33882 / OMIM:607875 (inositol polyphosphate-5-phosphatase K)
611911	ISCU / HGNC:29882 / OMIM:611911 (iron-sulfur cluster assembly enzyme)
600536	ITGA7 / HGNC:6143 / OMIM:600536 (integrin subunit alpha 7)
147265	ITPR1 / HGNC:6180 / OMIM:147265 (inositol 1,4,5-trisphosphate receptor type 1)
147267	ITPR3 / HGNC:6182 / OMIM:147267 (inositol 1,4,5-trisphosphate receptor type 3)
601920	JAG1 / HGNC:6188 / OMIM:601920 (jagged canonical Notch ligand 1)
605267	JPH2 / HGNC:14202 / OMIM:605267 (junctophilin 2)
173325	JUP / HGNC:6207 / OMIM:173325 (junction plakoglobin)
601421	KARS1 / HGNC:6215 / OMIM:601421 (lysyl-tRNA synthetase 1)
613727	KBTBD13 / HGNC:37227 / OMIM:613727 (kelch repeat and BTB domain containing 13)
176260	KCNA1 / HGNC:6218 / OMIM:176260 (potassium voltage-gated channel subfamily A member 1)
176267	KCNA5 / HGNC:6224 / OMIM:176267 (potassium voltage-gated channel subfamily A member 5)
176264	KCNC3 / HGNC:6235 / OMIM:176264 (potassium voltage-gated channel subfamily C member 3)
605411	KCND3 / HGNC:6239 / OMIM:605411 (potassium voltage-gated channel subfamily D member 3)
176261	KCNE1 / HGNC:6240 / OMIM:176261 (potassium voltage-gated channel subfamily E regulatory subunit 1)
603796	KCNE2 / HGNC:6242 / OMIM:603796 (potassium voltage-gated channel subfamily E regulatory subunit 2)
604433	KCNE3 / HGNC:6243 / OMIM:604433 (potassium voltage-gated channel subfamily E regulatory subunit 3)

152427	KCNH2 / HGNC:6251 / OMIM:152427 (potassium voltage-gated channel subfamily H member 2)
602323	KCNJ12 / HGNC:6258 / OMIM:602323 (potassium inwardly rectifying channel subfamily J member 12)
600681	KCNJ2 / HGNC:6263 / OMIM:600681 (potassium inwardly rectifying channel subfamily J member 2)
600734	KCNJ5 / HGNC:6266 / OMIM:600734 (potassium inwardly rectifying channel subfamily J member 5)
607542	KCNQ1 / HGNC:6294 / OMIM:607542 (potassium voltage-gated channel subfamily Q member 1)
610657	WASHC5 / HGNC:28984 / OMIM:610657 (WASH complex subunit 5)
615759	KIDINS220 / HGNC:29508 / OMIM:615759 (kinase D interacting substrate 220)
601255	KIF1A / HGNC:888 / OMIM:601255 (kinesin family member 1A)
605995	KIF1B / HGNC:16636 / OMIM:605995 (kinesin family member 1B)
603060	KIF1C / HGNC:6317 / OMIM:603060 (kinesin family member 1C)
608283	KIF21A / HGNC:19349 / OMIM:608283 (kinesin family member 21A)
614026	KIF26B / HGNC:25484 / OMIM:614026 (kinesin family member 26B)
602821	KIF5A / HGNC:6323 / OMIM:602821 (kinesin family member 5A)
611729	KLC2 / HGNC:20716 / OMIM:611729 (kinesin light chain 2)
615340	KLHL40 / HGNC:30372 / OMIM:615340 (kelch like family member 40)
607701	KLHL41 / HGNC:16905 / OMIM:607701 (kelch like family member 41)
611201	KLHL9 / HGNC:18732 / OMIM:611201 (kelch like family member 9)
605739	KY / HGNC:26576 / OMIM:605739 (kyphoscoliosis peptidase)
308840	L1CAM / HGNC:6470 / OMIM:308840 (L1 cell adhesion molecule)
156225	LAMA2 / HGNC:6482 / OMIM:156225 (laminin subunit alpha 2)
600133	LAMA4 / HGNC:6484 / OMIM:600133 (laminin subunit alpha 4)
601033	LAMA5 / HGNC:6485 / OMIM:601033 (laminin subunit alpha 5)
150325	LAMB2 / HGNC:6487 / OMIM:150325 (laminin subunit beta 2)
309060	LAMP2 / HGNC:6501 / OMIM:309060 (lysosomal associated membrane protein 2)
603590	LARGE1 / HGNC:6511 / OMIM:603590 (LARGE xylosyl- and glucuronyltransferase 1)
605906	LDB3 / HGNC:15710 / OMIM:605906 (LIM domain binding 3)
150000	LDHA / HGNC:6535 / OMIM:150000 (lactate dehydrogenase A)
607908	LIMS2 / HGNC:16084 / OMIM:607908 (LIM zinc finger domain containing 2)
603795	LITAF / HGNC:16841 / OMIM:603795 (lipopolysaccharide induced TNF factor)
150330	LMNA / HGNC:6636 / OMIM:150330 (lamin A/C)
616112	LMOD3 / HGNC:6649 / OMIM:616112 (leiomodin 3)
605518	LPIN1 / HGNC:13345 / OMIM:605518 (lipin 1)
618299	LRP12 / HGNC:31708 / OMIM:618299 (LDL receptor related protein 12)
604270	LRP4 / HGNC:6696 / OMIM:604270 (LDL receptor related protein 4)
610933	LRSAM1 / HGNC:25135 / OMIM:610933 (leucine rich repeat and sterile alpha motif containing 1)
159460	MAG / HGNC:6783 / OMIM:159460 (myelin associated glycoprotein)
609479	MAP3K20 / HGNC:17797 / OMIM:609479 (mitogen-activated protein kinase kinase kinase 20)
157140	MAPT / HGNC:6893 / OMIM:157140 (microtubule associated protein tau)
156560	MARS1 / HGNC:6898 / OMIM:156560 (methionyl-tRNA synthetase 1)
609728	MARS2 / HGNC:25133 / OMIM:609728 (methionyl-tRNA synthetase 2, mitochondrial)
164015	MATR3 / HGNC:6912 / OMIM:164015 (matrin 3)
160000	MB / HGNC:6915 / OMIM:160000 (myoglobin)

603294	MCM3AP / HGNC:6946 / OMIM:603294 (minichromosome maintenance complex component 3 associated protein)
612453	MEGF10 / HGNC:29634 / OMIM:612453 (multiple EGF like domains 10)
608507	MFN2 / HGNC:16877 / OMIM:608507 (mitofusin 2)
615076	MGME1 / HGNC:16205 / OMIM:615076 (mitochondrial genome maintenance exonuclease 1)
608677	MIB1 / HGNC:21086 / OMIM:608677 (MIB E3 ubiquitin protein ligase 1)
120520	MME / HGNC:7154 / OMIM:120520 (membrane metalloendopeptidase)
616661	MORC2 / HGNC:23573 / OMIM:616661 (MORC family CW-type zinc finger 2)
604041	MPDU1 / HGNC:7207 / OMIM:604041 (mannose-P-dolichol utilization defect 1)
137960	MPV17 / HGNC:7224 / OMIM:137960 (mitochondrial inner membrane protein MPV17)
159440	MPZ / HGNC:7225 / OMIM:159440 (myelin protein zero)
600814	MRE11 / HGNC:7230 / OMIM:600814 (MRE11 homolog, double strand break repair nuclease)
607118	MRPL3 / HGNC:10379 / OMIM:607118 (mitochondrial ribosomal protein L3)
611849	MRPL44 / HGNC:16650 / OMIM:611849 (mitochondrial ribosomal protein L44)
611987	MRPS25 / HGNC:14511 / OMIM:611987 (mitochondrial ribosomal protein S25)
601788	MSTN / HGNC:4223 / OMIM:601788 (myostatin)
617619	MSTO1 / HGNC:29678 / OMIM:617619 (misato mitochondrial distribution and morphology regulator 1)
590065	MT-TM / HGNC:7492 / OMIM:590065 (mitochondrially encoded tRNA-Met (AUA/G))
590090	MT-TT / HGNC:7499 / OMIM:590090 (mitochondrially encoded tRNA-Thr (ACN))
300415	MTM1 / HGNC:7448 / OMIM:300415 (myotubularin 1)
603557	MTMR2 / HGNC:7450 / OMIM:603557 (myotubularin related protein 2)
614667	MTO1 / HGNC:19261 / OMIM:614667 (mitochondrial tRNA translation optimization 1)
613669	MTPAP / HGNC:25532 / OMIM:613669 (mitochondrial poly(A) polymerase)
601296	MUSK / HGNC:7525 / OMIM:601296 (muscle associated receptor tyrosine kinase)
160794	MYBPC1 / HGNC:7549 / OMIM:160794 (myosin binding protein C1)
600958	MYBPC3 / HGNC:7551 / OMIM:600958 (myosin binding protein C3)
608568	MYH14 / HGNC:23212 / OMIM:608568 (myosin heavy chain 14)
160740	MYH2 / HGNC:7572 / OMIM:160740 (myosin heavy chain 2)
160720	MYH3 / HGNC:7573 / OMIM:160720 (myosin heavy chain 3)
160710	MYH6 / HGNC:7576 / OMIM:160710 (myosin heavy chain 6)
160760	MYH7 / HGNC:7577 / OMIM:160760 (myosin heavy chain 7)
160741	MYH8 / HGNC:7578 / OMIM:160741 (myosin heavy chain 8)
160780	MYL1 / HGNC:7582 / OMIM:160780 (myosin light chain 1)
160781	MYL2 / HGNC:7583 / OMIM:160781 (myosin light chain 2)
160790	MYL3 / HGNC:7584 / OMIM:160790 (myosin light chain 3)
160770	MYL4 / HGNC:7585 / OMIM:160770 (myosin light chain 4)
606566	MYLK2 / HGNC:16243 / OMIM:606566 (myosin light chain kinase 2)
615345	MYMK / HGNC:33778 / OMIM:615345 (myomaker, myoblast fusion factor)
607295	MYO18B / HGNC:18150 / OMIM:607295 (myosin XVIIIB)
604875	MYO9A / HGNC:7608 / OMIM:604875 (myosin IXA)
604103	MYOT / HGNC:12399 / OMIM:604103 (myotilin)
605602	MYOZ2 / HGNC:1330 / OMIM:605602 (myozenin 2)
608517	MYPN / HGNC:23246 / OMIM:608517 (myopalladin)
609701	NAGLU / HGNC:7632 / OMIM:609701 (N-acetyl-alpha-glucosaminidase)
612803	NARS2 / HGNC:26274 / OMIM:612803 (asparaginyl-tRNA synthetase 2, mitochondrial)
605262	NDRG1 / HGNC:7679 / OMIM:605262 (N-myc downstream regulated 1)

606934	NDUFAF1 / HGNC:18828 / OMIM:606934 (NADH:ubiquinone oxidoreductase complex assembly factor 1)
161650	NEB / HGNC:7720 / OMIM:161650 (nebulin)
162230	NEFH / HGNC:7737 / OMIM:162230 (neurofilament heavy chain)
162280	NEFL / HGNC:7739 / OMIM:162280 (neurofilament light chain)
604588	NEK1 / HGNC:7744 / OMIM:604588 (NIMA related kinase 1)
609798	NEK9 / HGNC:18591 / OMIM:609798 (NIMA related kinase 9)
613121	NEXN / HGNC:29557 / OMIM:613121 (nexilin F-actin binding protein)
162030	NGF / HGNC:7808 / OMIM:162030 (nerve growth factor)
608145	NIPA1 / HGNC:17043 / OMIM:608145 (NIPA magnesium transporter 1)
605955	NKX6-2 / HGNC:19321 / OMIM:605955 (NK6 homeobox 2)
608701	NMNAT2 / HGNC:16789 / OMIM:608701 (nicotinamide nucleotide adenylyltransferase 2)
614154	NOP56 / HGNC:15911 / OMIM:614154 (NOP56 ribonucleoprotein)
108780	NPPA / HGNC:7939 / OMIM:108780 (natriuretic peptide A)
600417	NT5C2 / HGNC:8022 / OMIM:600417 (5'-nucleotidase, cytosolic II)
191315	NTRK1 / HGNC:8031 / OMIM:191315 (neurotrophic receptor tyrosine kinase 1)
606694	NUP155 / HGNC:8063 / OMIM:606694 (nucleoporin 155)
602552	NUP88 / HGNC:8067 / OMIM:602552 (nucleoporin 88)
605290	OPA1 / HGNC:8140 / OMIM:605290 (OPA1 mitochondrial dynamin like GTPase)
602432	OPTN / HGNC:17142 / OMIM:602432 (optineurin)
610277	ORAI1 / HGNC:25896 / OMIM:610277 (ORAI calcium release-activated calcium modulator 1)
602279	PABPN1 / HGNC:8565 / OMIM:602279 (poly(A) binding protein nuclear 1)
167410	PAX7 / HGNC:8621 / OMIM:167410 (paired box 7)
176740	PCNA / HGNC:8729 / OMIM:176740 (proliferating cell nuclear antigen)
300906	PDK3 / HGNC:8811 / OMIM:300906 (pyruvate dehydrogenase kinase 3)
131340	PDYN / HGNC:8820 / OMIM:131340 (prodynorphin)
601757	PEX7 / HGNC:8860 / OMIM:601757 (peroxisomal biogenesis factor 7)
610681	PFKM / HGNC:8877 / OMIM:610681 (phosphofructokinase, muscle)
176610	PFN1 / HGNC:8881 / OMIM:176610 (profilin 1)
612931	PGAM2 / HGNC:8889 / OMIM:612931 (phosphoglycerate mutase 2)
311800	PGK1 / HGNC:8896 / OMIM:311800 (phosphoglycerate kinase 1)
171900	PGM1 / HGNC:8905 / OMIM:171900 (phosphoglucomutase 1)
311870	PHKA1 / HGNC:8925 / OMIM:311870 (phosphorylase kinase regulatory subunit alpha 1)
602753	PHOX2A / HGNC:691 / OMIM:602753 (paired like homeobox 2A)
602026	PHYH / HGNC:8940 / OMIM:602026 (phytanoyl-CoA 2-hydroxylase)
613629	PIEZO2 / HGNC:26270 / OMIM:613629 (piezo type mechanosensitive ion channel component 2)
606102	PIP5K1C / HGNC:8996 / OMIM:606102 (phosphatidylinositol-4-phosphate 5-kinase type 1 gamma)
602861	PKP2 / HGNC:9024 / OMIM:602861 (plakophilin 2)
615698	PLD3 / HGNC:17158 / OMIM:615698 (phospholipase D family member 3)
601282	PLEC / HGNC:9069 / OMIM:601282 (plectin)
611101	PLEKHG5 / HGNC:29105 / OMIM:611101 (pleckstrin homology and RhoGEF domain containing G5)
172405	PLN / HGNC:9080 / OMIM:172405 (phospholamban)
300401	PLP1 / HGNC:9086 / OMIM:300401 (proteolipid protein 1)
170715	PMP2 / HGNC:9117 / OMIM:170715 (peripheral myelin protein 2)
601097	PMP22 / HGNC:9118 / OMIM:601097 (peripheral myelin protein 22)

605610	PNKP / HGNC:9154 / OMIM:605610 (polynucleotide kinase 3'-phosphatase)
609059	PNPLA2 / HGNC:30802 / OMIM:609059 (patatin like phospholipase domain containing 2)
603197	PNPLA6 / HGNC:16268 / OMIM:603197 (patatin like phospholipase domain containing 6)
612123	PNPLA8 / HGNC:28900 / OMIM:612123 (patatin like phospholipase domain containing 8)
615618	POGLUT1 / HGNC:22954 / OMIM:615618 (protein O-glucosyltransferase 1)
174763	POLG / HGNC:9179 / OMIM:174763 (DNA polymerase gamma, catalytic subunit)
604983	POLG2 / HGNC:9180 / OMIM:604983 (DNA polymerase gamma 2, accessory subunit)
606822	POMGNT1 / HGNC:19139 / OMIM:606822 (protein O-linked mannose N-acetylglucosaminyltransferase 1 (beta 1,2-))
614828	POMGNT2 / HGNC:25902 / OMIM:614828 (protein O-linked mannose N-acetylglucosaminyltransferase 2 (beta 1,4-))
615247	POMK / HGNC:26267 / OMIM:615247 (protein O-mannose kinase)
607423	POMT1 / HGNC:9202 / OMIM:607423 (protein O-mannosyltransferase 1)
607439	POMT2 / HGNC:19743 / OMIM:607439 (protein O-mannosyltransferase 2)
605824	POPDC3 / HGNC:17649 / OMIM:605824 (popeye domain containing 3)
604325	PPP2R2B / HGNC:9305 / OMIM:604325 (protein phosphatase 2 regulatory subunit Bbeta)
616458	PRDM12 / HGNC:13997 / OMIM:616458 (PR/SET domain 12)
605557	PRDM16 / HGNC:14000 / OMIM:605557 (PR/SET domain 16)
609557	PREPL / HGNC:30228 / OMIM:609557 (prolyl endopeptidase like)
602743	PRKAG2 / HGNC:9386 / OMIM:602743 (protein kinase AMP-activated non-catalytic subunit gamma 2)
176980	PRKCG / HGNC:9402 / OMIM:176980 (protein kinase C gamma)
170710	PRPH / HGNC:9461 / OMIM:170710 (peripherin)
311850	PRPS1 / HGNC:9462 / OMIM:311850 (phosphoribosyl pyrophosphate synthetase 1)
617413	PRUNE1 / HGNC:13420 / OMIM:617413 (prune exopolyphosphatase 1)
605725	PRX / HGNC:13797 / OMIM:605725 (periaxin)
104311	PSEN1 / HGNC:9508 / OMIM:104311 (presenilin 1)
600759	PSEN2 / HGNC:9509 / OMIM:600759 (presenilin 2)
608625	PTRH2 / HGNC:24265 / OMIM:608625 (peptidyl-tRNA hydrolase 2)
607204	PUM1 / HGNC:14957 / OMIM:607204 (pumilio RNA binding family member 1)
600473	PURA / HGNC:9701 / OMIM:600473 (purine rich element binding protein A)
608109	PUS1 / HGNC:15508 / OMIM:608109 (pseudouridine synthase 1)
608455	PYGM / HGNC:9726 / OMIM:608455 (glycogen phosphorylase, muscle associated)
617220	PYROXD1 / HGNC:26162 / OMIM:617220 (pyridine nucleotide-disulphide oxidoreductase domain 1)
602298	RAB7A / HGNC:9788 / OMIM:602298 (RAB7A, member RAS oncogene family)
164760	RAF1 / HGNC:9829 / OMIM:164760 (Raf-1 proto-oncogene, serine/threonine kinase)
601592	RAPSN / HGNC:9863 / OMIM:601592 (receptor associated protein of the synapse)
610924	RBCK1 / HGNC:15864 / OMIM:610924 (RANBP2-type and C3HC4-type zinc finger containing 1)
613171	RBM20 / HGNC:27424 / OMIM:613171 (RNA binding motif protein 20)
612413	RBM7 / HGNC:9904 / OMIM:612413 (RNA binding motif protein 7)
609139	REEP1 / HGNC:25786 / OMIM:609139 (receptor accessory protein 1)
609347	REEP2 / HGNC:17975 / OMIM:609347 (receptor accessory protein 2)
613114	RETREG1 / HGNC:25964 / OMIM:613114 (reticulophagy regulator 1)
102579	RFC1 / HGNC:9969 / OMIM:102579 (replication factor C subunit 1)
604123	RNASEH1 / HGNC:18466 / OMIM:604123 (ribonuclease H1)
609948	RNF216 / HGNC:21698 / OMIM:609948 (ring finger protein 216)
612159	RPH3A / HGNC:17056 / OMIM:612159 (rabphilin 3A)

604712	RRM2B / HGNC:17296 / OMIM:604712 (ribonucleotide reductase regulatory TP53 inducible subunit M2B)
603183	RTN2 / HGNC:10468 / OMIM:603183 (reticulon 2)
613516	RUBCN / HGNC:28991 / OMIM:613516 (rubicon autophagy regulator)
605862	RXYLT1 / HGNC:13530 / OMIM:605862 (ribitol xylosyltransferase 1)
180901	RYR1 / HGNC:10483 / OMIM:180901 (ryanodine receptor 1)
180902	RYR2 / HGNC:10484 / OMIM:180902 (ryanodine receptor 2)
180903	RYR3 / HGNC:10485 / OMIM:180903 (ryanodine receptor 3)
604490	SACS / HGNC:10519 / OMIM:604490 (sacsin molecular chaperone)
603560	SBF1 / HGNC:10542 / OMIM:603560 (SET binding factor 1)
607697	SBF2 / HGNC:2135 / OMIM:607697 (SET binding factor 2)
604385	SCN11A / HGNC:10583 / OMIM:604385 (sodium voltage-gated channel alpha subunit 11)
600235	SCN1B / HGNC:10586 / OMIM:600235 (sodium voltage-gated channel beta subunit 1)
601327	SCN2B / HGNC:10589 / OMIM:601327 (sodium voltage-gated channel beta subunit 2)
608214	SCN3B / HGNC:20665 / OMIM:608214 (sodium voltage-gated channel beta subunit 3)
603967	SCN4A / HGNC:10591 / OMIM:603967 (sodium voltage-gated channel alpha subunit 4)
608256	SCN4B / HGNC:10592 / OMIM:608256 (sodium voltage-gated channel beta subunit 4)
600163	SCN5A / HGNC:10593 / OMIM:600163 (sodium voltage-gated channel alpha subunit 5)
603415	SCN9A / HGNC:10597 / OMIM:603415 (sodium voltage-gated channel alpha subunit 9)
604272	SCO2 / HGNC:10604 / OMIM:604272 (synthesis of cytochrome C oxidase 2)
607982	SCYL1 / HGNC:14372 / OMIM:607982 (SCY1 like pseudokinase 1)
600857	SDHA / HGNC:10680 / OMIM:600857 (succinate dehydrogenase complex flavoprotein subunit A)
606210	SELENON / HGNC:15999 / OMIM:606210 (selenoprotein N)
604061	SEPTIN9 / HGNC:7323 / OMIM:604061 (septin 9)
608465	SETX / HGNC:445 / OMIM:608465 (senataxin)
600119	SGCA / HGNC:10805 / OMIM:600119 (sarcoglycan alpha)
600900	SGCB / HGNC:10806 / OMIM:600900 (sarcoglycan beta)
601411	SGCD / HGNC:10807 / OMIM:601411 (sarcoglycan delta)
608896	SGCG / HGNC:10809 / OMIM:608896 (sarcoglycan gamma)
603729	SGPL1 / HGNC:10817 / OMIM:603729 (sphingosine-1-phosphate lyase 1)
608206	SH3TC2 / HGNC:29427 / OMIM:608206 (SH3 domain and tetratricopeptide repeats 2)
601978	SIGMAR1 / HGNC:8157 / OMIM:601978 (sigma non-opioid intracellular receptor 1)
608005	SIL1 / HGNC:24624 / OMIM:608005 (SIL1 nucleotide exchange factor)
604878	SLC12A6 / HGNC:10914 / OMIM:604878 (solute carrier family 12 member 6)
600682	SLC16A1 / HGNC:10922 / OMIM:600682 (solute carrier family 16 member 1)
600336	SLC18A3 / HGNC:10936 / OMIM:600336 (solute carrier family 18 member A3)
600111	SLC1A3 / HGNC:10941 / OMIM:600111 (solute carrier family 1 member 3)
603377	SLC22A5 / HGNC:10969 / OMIM:603377 (solute carrier family 22 member 5)
190315	SLC25A1 / HGNC:10979 / OMIM:190315 (solute carrier family 25 member 1)
606521	SLC25A19 / HGNC:14409 / OMIM:606521 (solute carrier family 25 member 19)
613698	SLC25A20 / HGNC:1421 / OMIM:613698 (solute carrier family 25 member 20)
103220	SLC25A4 / HGNC:10990 / OMIM:103220 (solute carrier family 25 member 4)
610823	SLC25A42 / HGNC:28380 / OMIM:610823 (solute carrier family 25 member 42)
610826	SLC25A46 / HGNC:25198 / OMIM:610826 (solute carrier family 25 member 46)
603690	SLC33A1 / HGNC:95 / OMIM:603690 (solute carrier family 33 member 1)
607882	SLC52A2 / HGNC:30224 / OMIM:607882 (solute carrier family 52 member 2)
613350	SLC52A3 / HGNC:16187 / OMIM:613350 (solute carrier family 52 member 3)

608761	SLC5A7 / HGNC:14025 / OMIM:608761 (solute carrier family 5 member 7)
107310	SLC9A1 / HGNC:11071 / OMIM:107310 (solute carrier family 9 member A1)
604990	SLC9A3R1 / HGNC:11075 / OMIM:604990 (SLC9A3 regulator 1)
614982	SMCHD1 / HGNC:29090 / OMIM:614982 (structural maintenance of chromosomes flexible hinge domain containing 1)
600354	SMN1 / HGNC:11117 / OMIM:600354 (survival of motor neuron 1, telomeric)
601627	SMN2 / HGNC:11118 / OMIM:601627 (survival of motor neuron 2, centromeric)
610457	SMPD4 / HGNC:32949 / OMIM:610457 (sphingomyelin phosphodiesterase 4)
600322	SNAP25 / HGNC:11132 / OMIM:600322 (synaptosome associated protein 25)
601017	SNTA1 / HGNC:11167 / OMIM:601017 (syntrophin alpha 1)
616105	SNX14 / HGNC:14977 / OMIM:616105 (sorting nexin 14)
147450	SOD1 / HGNC:11179 / OMIM:147450 (superoxide dismutase 1)
182500	SORD / HGNC:11184 / OMIM:182500 (sorbitol dehydrogenase)
607111	SPART / HGNC:18514 / OMIM:607111 (spartin)
604277	SPAST / HGNC:11233 / OMIM:604277 (spastin)
615950	SPEG / HGNC:16901 / OMIM:615950 (striated muscle enriched protein kinase)
610844	SPG11 / HGNC:11226 / OMIM:610844 (SPG11 vesicle trafficking associated, spatacsin)
608181	SPG21 / HGNC:20373 / OMIM:608181 (SPG21 abhydrolase domain containing, maspardin)
602783	SPG7 / HGNC:11237 / OMIM:602783 (SPG7 matrix AAA peptidase subunit, paraplegin)
182810	SPTAN1 / HGNC:11273 / OMIM:182810 (spectrin alpha, non-erythrocytic 1)
604985	SPTBN2 / HGNC:11276 / OMIM:604985 (spectrin beta, non-erythrocytic 2)
606214	SPTBN4 / HGNC:14896 / OMIM:606214 (spectrin beta, non-erythrocytic 4)
605712	SPTLC1 / HGNC:11277 / OMIM:605712 (serine palmitoyltransferase long chain base subunit 1)
605713	SPTLC2 / HGNC:11278 / OMIM:605713 (serine palmitoyltransferase long chain base subunit 2)
601530	SQSTM1 / HGNC:11280 / OMIM:601530 (sequestosome 1)
615521	STAC3 / HGNC:28423 / OMIM:615521 (SH3 and cysteine rich domain 3)
605921	STIM1 / HGNC:11386 / OMIM:605921 (stromal interaction molecule 1)
607207	STUB1 / HGNC:11427 / OMIM:607207 (STIP1 homology and U-box containing protein 1)
603921	SUCLA2 / HGNC:11448 / OMIM:603921 (succinate-CoA ligase ADP-forming subunit beta)
611224	SUCLG1 / HGNC:11449 / OMIM:611224 (succinate-CoA ligase GDP/ADP-forming subunit alpha)
185620	SURF1 / HGNC:11474 / OMIM:185620 (SURF1 cytochrome c oxidase assembly factor)
604126	SVIL / HGNC:11480 / OMIM:604126 (supervillin)
608441	SYNE1 / HGNC:17089 / OMIM:608441 (spectrin repeat containing nuclear envelope protein 1)
608442	SYNE2 / HGNC:17084 / OMIM:608442 (spectrin repeat containing nuclear envelope protein 2)
610949	SYT14 / HGNC:23143 / OMIM:610949 (synaptotagmin 14)
600104	SYT2 / HGNC:11510 / OMIM:600104 (synaptotagmin 2)
605078	TARDBP / HGNC:11571 / OMIM:605078 (TAR DNA binding protein)
300394	TAFAZZIN / HGNC:11577 / OMIM:300394 (tafazzin, phospholipid-lysophospholipid transacylase)
604649	TBCD / HGNC:11581 / OMIM:604649 (tubulin folding cofactor D)
604834	TBK1 / HGNC:11584 / OMIM:604834 (TANK binding kinase 1)
600075	TBP / HGNC:11588 / OMIM:600075 (TATA-box binding protein)
604488	TCAP / HGNC:11610 / OMIM:604488 (titin-cap)
607198	TDP1 / HGNC:18884 / OMIM:607198 (tyrosyl-DNA phosphodiesterase 1)
605764	TDP2 / HGNC:17768 / OMIM:605764 (tyrosyl-DNA phosphodiesterase 2)
615000	TECPR2 / HGNC:19957 / OMIM:615000 (tectonin beta-propeller repeat containing 2)

617242	TECRL / HGNC:27365 / OMIM:617242 (trans-2,3-enoyl-CoA reductase like)
602498	TFG / HGNC:11758 / OMIM:602498 (trafficking from ER to golgi regulator)
190230	TGFB3 / HGNC:11769 / OMIM:190230 (transforming growth factor beta 3)
613900	TGM6 / HGNC:16255 / OMIM:613900 (transglutaminase 6)
603518	TIA1 / HGNC:11802 / OMIM:603518 (TIA1 cytotoxic granule associated RNA binding protein)
607251	TIMM22 / HGNC:17317 / OMIM:607251 (translocase of inner mitochondrial membrane 22)
188250	TK2 / HGNC:11831 / OMIM:188250 (thymidine kinase 2)
616101	TMEM240 / HGNC:25186 / OMIM:616101 (transmembrane protein 240)
612048	TMEM43 / HGNC:28472 / OMIM:612048 (transmembrane protein 43)
616609	TMEM65 / HGNC:25203 / OMIM:616609 (transmembrane protein 65)
188380	TMPO / HGNC:11875 / OMIM:188380 (thymopoietin)
191040	TNNC1 / HGNC:11943 / OMIM:191040 (troponin C1, slow skeletal and cardiac type)
191043	TNNI2 / HGNC:11946 / OMIM:191043 (troponin I2, fast skeletal type)
191044	TNNI3 / HGNC:11947 / OMIM:191044 (troponin I3, cardiac type)
191041	TNNT1 / HGNC:11948 / OMIM:191041 (troponin T1, slow skeletal type)
191045	TNNT2 / HGNC:11949 / OMIM:191045 (troponin T2, cardiac type)
600692	TNNT3 / HGNC:11950 / OMIM:600692 (troponin T3, fast skeletal type)
610032	TNPO3 / HGNC:17103 / OMIM:610032 (transportin 3)
601243	TOP3A / HGNC:11992 / OMIM:601243 (DNA topoisomerase III alpha)
605204	TOR1A / HGNC:3098 / OMIM:605204 (torsin family 1 member A)
614512	TOR1AIP1 / HGNC:29456 / OMIM:614512 (torsin 1A interacting protein 1)
191010	TPM1 / HGNC:12010 / OMIM:191010 (tropomyosin 1)
190990	TPM2 / HGNC:12011 / OMIM:190990 (tropomyosin 2)
191030	TPM3 / HGNC:12012 / OMIM:191030 (tropomyosin 3)
614138	TRAPPC11 / HGNC:25751 / OMIM:614138 (trafficking protein particle complex subunit 11)
603283	TRDN / HGNC:12261 / OMIM:603283 (triadin)
614141	TRIM2 / HGNC:15974 / OMIM:614141 (tripartite motif containing 2)
602290	TRIM32 / HGNC:16380 / OMIM:602290 (tripartite motif containing 32)
606474	TRIM54 / HGNC:16008 / OMIM:606474 (tripartite motif containing 54)
606131	TRIM63 / HGNC:16007 / OMIM:606131 (tripartite motif containing 63)
604501	TRIP4 / HGNC:12310 / OMIM:604501 (thyroid hormone receptor interactor 4)
602345	TRPC3 / HGNC:12335 / OMIM:602345 (transient receptor potential cation channel subfamily C member 3)
605427	TRPV4 / HGNC:18083 / OMIM:605427 (transient receptor potential cation channel subfamily V member 4)
604723	TSFM / HGNC:12367 / OMIM:604723 (Ts translation elongation factor, mitochondrial)
611695	TTBK2 / HGNC:19141 / OMIM:611695 (tau tubulin kinase 2)
188840	TTN / HGNC:12403 / OMIM:188840 (titin)
600415	TTPA / HGNC:12404 / OMIM:600415 (alpha tocopherol transfer protein)
176300	TTR / HGNC:12405 / OMIM:176300 (transthyretin)
191110	TUBA4A / HGNC:12407 / OMIM:191110 (tubulin alpha 4a)
602661	TUBB3 / HGNC:20772 / OMIM:602661 (tubulin beta 3 class III)
606075	TWNK / HGNC:1160 / OMIM:606075 (twinkle mtDNA helicase)
131222	TYMP / HGNC:3148 / OMIM:131222 (thymidine phosphorylase)
314370	UBA1 / HGNC:12469 / OMIM:314370 (ubiquitin like modifier activating enzyme 1)
609787	UBAP1 / HGNC:12461 / OMIM:609787 (ubiquitin associated protein 1)
300264	UBQLN2 / HGNC:12509 / OMIM:300264 (ubiquilin 2)
191342	UCHL1 / HGNC:12513 / OMIM:191342 (ubiquitin C-terminal hydrolase L1)

			605836 | UNC13B / HGNC:12566 / OMIM:605836 (unc-13 homolog B) 611220 | UNC45B / HGNC:14304 / OMIM:611220 (unc-45 myosin chaperone B) 185880 | VAMP1 / HGNC:12642 / OMIM:185880 (vesicle associated membrane protein 1) 605704 | VAPB / HGNC:12649 / OMIM:605704 (VAMP associated protein B and C) 193065 | VCL / HGNC:12665 / OMIM:193065 (vinculin) 601023 | VCP / HGNC:12666 / OMIM:601023 (valosin containing protein) 300913 | VMA21 / HGNC:22082 / OMIM:300913 (vacuolar ATPase assembly factor VMA21) 608877 | VPS13D / HGNC:23595 / OMIM:608877 (vacuolar protein sorting 13 homolog D) 609927 | VPS37A / HGNC:24928 / OMIM:609927 (VPS37A subunit of ESCRT-I) 602168 | VRK1 / HGNC:12718 / OMIM:602168 (VRK serine/threonine kinase 1) 614884 | VWA3B / HGNC:28385 / OMIM:614884 (von Willebrand factor A domain containing 3B) 191050 | WARS1 / HGNC:12729 / OMIM:191050 (tryptophanyl-tRNA synthetase 1) 605232 | WNK1 / HGNC:14540 / OMIM:605232 (WNK lysine deficient protein kinase 1) 605131 | WWOX / HGNC:12799 / OMIM:605131 (WW domain containing oxidoreductase) 194360 | XRCC1 / HGNC:12828 / OMIM:194360 (X-ray repair cross complementing 1) 603623 | YARS1 / HGNC:12840 / OMIM:603623 (tyrosyl-tRNA synthetase 1) 610957 | YARS2 / HGNC:24249 / OMIM:610957 (tyrosyl-tRNA synthetase 2) 613915 | ZBTB42 / HGNC:32550 / OMIM:613915 (zinc finger and BTB domain containing 42) 617828 | ZFHX2 / HGNC:20152 / OMIM:617828 (zinc finger homeobox 2) 612012 | ZFYVE26 / HGNC:20761 / OMIM:612012 (zinc finger FYVE-type containing 26) 610243 | ZFYVE27 / HGNC:26559 / OMIM:610243 (zinc finger FYVE-type containing 27)
585	[undiag_hgnc_ont] Show the field ONLY if: [undiag_report_variant] = 'yes' AND [undiag_listpref]='ontology'	Gene HGNC-code (browse ontology)If the affected gene is not present in the neuromuscular gene list above (e.g. a new gene not previously associated with neuromuscular disease) please search for the gene at https://www.genenames.org/tools/search/ and enter the numerical HGNC ID here (e.g. for DMD HGNC:2928 you would enter HGNC_2928) *Element 6.3. in the Set of common data elements for Rare Diseases Registration (link)*	text, Required BIOPORTAL:HGNC | BIOPORTAL:HGNC
586	[undiag_omim_ont] Show the field ONLY if: [undiag_report_variant] = 'yes' AND [undiag_listpref]='ontology'	Gene OMIM-code (browse ontology)If the affected gene is not present in the neuromuscular gene list above (e.g. a new gene not previously associated with neuromuscular disease) please search for the gene at https://www.omim.org/ and enter the numerical OMIM ID here (e.g. for DMD you would enter 300377; you also find the code for this example if you type 'Dystrophin' to the box on the right) *Element 6.3. in the Set of common data elements for Rare Diseases Registration (link)*	text, Required BIOPORTAL:OMIM | BIOPORTAL:OMIM
587	[undiag_omim] Show the field ONLY if: [undiag_report_variant] = 'yes'	Gene OMIM code calculated field	calc Calculation: if([undiag_report_variant] = 'yes', if([undiag_listpref]='ontology', [undiag_omim_ont], if([undiag_listpref]='all_nmd', [undiag_omim_armx], '')), '') Field Annotation: @READONLY @HIDDEN @HIDDEN-PDF
588	[undiag_v1_report] Show the field ONLY if: [undiag_report_variant] = 'yes'	Section Header: *Variant 1* How would you like to describe variant 1?Note: We prefer NCBI ClinVar but if the change in question is not yet described there, you can give a dbSNP ID if available, or you can type in the protein or cDNA change given in the genetic report.	radio, Required clinvar | NCBI ClinVar dbsnp | dbSNP custom_hgvs | Custom HGVS-simple notation
589	[undiag_v1_clinvar_uri] Show the field ONLY if: [undiag_v1_report] = 'clinvar'	NCBI ClinVar URI (please provide website-link) *If the change is described in the NCBI ClinVar database, please provide website-link (e.g. https://www.ncbi.nlm.nih.gov/clinvar/variation/4886/)*	text
590	[undiag_v1_snp_id] Show the field ONLY if: [undiag_v1_report] = 'dbsnp'	dbSNP RS ID *If the change has a dbSNP ID, enter it here, e.g. 'rs606231128'*	text
591	[undiag_v1_hgvs_opts] Show the field ONLY if: [undiag_v1_report] = 'custom_hgvs'	Variant 1: What kind of change(s) would you like to report in "HGVS simple"-notation? Please enter all the details that you have available from the report - e.g. if the report lists both the cDNA and protein changes, please provide both.	checkbox, Required cdna | undiag_v1_hgvs_opts___cdna | Report cDNA change (c.) nucleotide | undiag_v1_hgvs_opts___nucleotide | Report nucleotide (genomic) change (g.) protein | undiag_v1_hgvs_opts___protein | Report protein change (p.) other | undiag_v1_hgvs_opts___other | Report other type of change, e.g. RNA (r.) or non-coding (n.)

592	[undiag_v1_refseq_c] Show the field ONLY if: [undiag_v1_hgvs_opts(cdna)] = 1	Section Header: *Variant 1: cDNA change* Reference sequence for cDNA change *Enter the RefSeq transcript ID with the prefix NM_ or NR_ etc., e.g. 'NM_173660.5'. More information and examples see here and here*	text, Required
593	[undiag_v1_desc_c] Show the field ONLY if: [undiag_v1_hgvs_opts(cdna)] = 1	Description of cDNA change *Enter the cDNA change in HGVS nomenclature, e.g. c.1124_1127dupTGCC or c.346G>A More information and examples see here*	text, Required
594	[undiag_v1_hgvs_c] Show the field ONLY if: [undiag_v1_hgvs_opts(cdna)] = 1	undiag_v1_hgvs_c	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v1_refseq_c]!='' and [undiag_v1_desc_c]!='', concat([undiag_v1_refseq_c], ':', [undiag_v1_desc_c]), ''))
595	[undiag_v1_hgvs_c_disp] Show the field ONLY if: [undiag_v1_hgvs_opts(cdna)] = 1	Resulting HGVS notation for variant 1 cDNA change:[undiag_v1_hgvs_c]	descriptive
596	[undiag_v1_refseq_g] Show the field ONLY if: [undiag_v1_hgvs_opts(nucleotide)] = 1	Section Header: *Variant 1: Nucleotide (genomic) change* Reference sequence for nucleotide (genomic) change *Enter the RefSeq transcript ID with the prefix NC_ or NG_ etc., e.g. 'NC_000023.10'. More information and examples see here and here*	text, Required
597	[undiag_v1_desc_g] Show the field ONLY if: [undiag_v1_hgvs_opts(nucleotide)] = 1	Description of nucleotide (genomic) change *Enter the nucleotide change in HGVS nomenclature, e.g. g.34805_34808dup More information and examples see here*	text, Required
598	[undiag_v1_hgvs_g] Show the field ONLY if: [undiag_v1_hgvs_opts(nucleotide)] = 1	undiag_v1_hgvs_g	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v1_refseq_g]!='' and [undiag_v1_desc_g]!='', concat([undiag_v1_refseq_g], ':', [undiag_v1_desc_g]), ''))
599	[undiag_v1_hgvs_g_disp] Show the field ONLY if: [undiag_v1_hgvs_opts(nucleotide)] = 1	Resulting HGVS notation for variant 1 nucleotide (genomic) change:[undiag_v1_hgvs_g]	descriptive
600	[undiag_v1_refseq_p] Show the field ONLY if: [undiag_v1_hgvs_opts(protein)] = 1	Section Header: *Variant 1: Protein change* Reference sequence for protein change *Enter the RefSeq transcript ID with the prefix NP_, e.g. NP_003997.1 More information and examples see here and here*	text, Required
601	[undiag_v1_desc_p] Show the field ONLY if: [undiag_v1_hgvs_opts(protein)] = 1	Description of protein change *Enter the protein change in HGVS nomenclature, e.g. p.Ala378fs More information and examples see here*	text, Required
602	[undiag_v1_hgvs_p] Show the field ONLY if: [undiag_v1_hgvs_opts(protein)] = 1	undiag_v1_hgvs_p	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v1_refseq_p]!='' and [undiag_v1_desc_p]!='', concat([undiag_v1_refseq_p], ':', [undiag_v1_desc_p]), ''))
603	[undiag_v1_hgvs_p_disp] Show the field ONLY if: [undiag_v1_hgvs_opts(protein)] = 1	Resulting HGVS notation for variant 1 protein change:[undiag_v1_hgvs_p]	descriptive
604	[undiag_v1_refseq_o] Show the field ONLY if: [undiag_v1_hgvs_opts(other)] = 1	Section Header: *Variant 1: Other change* Reference sequence for other change *More information and examples see here and here*	text, Required
605	[undiag_v1_desc_o] Show the field ONLY if: [undiag_v1_hgvs_opts(other)] = 1	Description of other change *Examples for descriptions of RNA changes: r.357_358ins357+1_357+12 or r.11u>g More information and examples see here*	text, Required
606	[undiag_v1_hgvs_o] Show the field ONLY if: [undiag_v1_hgvs_opts(other)] = 1	undiag_v1_hgvs_o	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v1_refseq_o]!='' and [undiag_v1_desc_o]!='', concat([undiag_v1_refseq_o], ':', [undiag_v1_desc_o]), ''))
607	[undiag_v1_hgvs_o_disp] Show the field ONLY if: [undiag_v1_hgvs_opts(other)] = 1	Resulting HGVS notation for variant 1 other change:[undiag_v1_hgvs_o]	descriptive
608	[undiag_v1_zygos] Show the field ONLY if: [undiag_report_variant] = 'yes'	Section Header: *Variant 1: Additional Information* Zygosity	dropdown heterozygous, Heterozygous compound_heterozygous, Compound heterozygous homozygous, Homozygous

			hemizygous | Hemizygous mosaic | Mosaic unknown | Unknown
609	[undiag_v1_gen_build] Show the field ONLY if: [undiag_report_variant] = 'yes' AND [undiag_v1_report] = 'custom_hgvs'	Genomic build	dropdown GRCh38 | GRCh38 (hg38) GRCh37 | GRCh37 (hg19)
610	[undiag_v1_gen_app] Show the field ONLY if: [undiag_report_variant] = 'yes' AND [undiag_v1_report] = 'custom_hgvs'	Genetic approach	dropdown single_gene_screening | Single Gene Screening acgh | aCGH snp_array | SNP array mlpa | MLPA karyotype | Karyotype ngs_panel | NGS panel wes | WES wgs | WGS
611	[undiag_v1_chrom] Show the field ONLY if: [undiag_report_variant] = 'yes' AND [undiag_v1_report] = 'custom_hgvs'	Chromosome	dropdown 1 | 1 2 | 2 3 | 3 4 | 4 5 | 5 6 | 6 7 | 7 8 | 8 9 | 9 10 | 10 11 | 11 12 | 12 13 | 13 14 | 14 15 | 15 16 | 16 17 | 17 18 | 18 19 | 19 20 | 20 21 | 21 22 | 22 X | X Y | Y MT | MT
612	[undiag_desc3] Show the field ONLY if: [undiag_report_variant] = 'yes'	Section Header: *Variant 2 (if applicable)* Please report only an additional variant for the gene selected above (selected gene-OMIM: [undiag_omim]). If you wish to report a variant for another gene, please report that in a separate 'Undiagnosed'-form for that gene.	descriptive
613	[undiag_v2_yes] Show the field ONLY if: [undiag_report_variant] = 'yes'	Report a second variant for gene-OMIM: [undiag_omim]	radio yes | Yes not_present | Not present

			unknown: Unknown
614	[undiag_v2_report] Show the field ONLY if: [undiag_v2_yes] = 'yes'	How would you like to describe variant 2?Note: We prefer NCBI ClinVar but if the change in question is not yet described there, you can give a dbSNP ID if available, or you can type in the protein or cDNA change given in the genetic report.	radio, Required clinvar: NCBI ClinVar dbsnp: dbSNP custom_hgvs: Custom HGVS-simple notation
615	[undiag_v2_clinvar_uri] Show the field ONLY if: [undiag_v2_report] = 'clinvar'	NCBI ClinVar URI (please provide website-link) *If the change is described in the NCBI ClinVar database, please provide website-link (e.g. https://www.ncbi.nlm.nih.gov/clinvar/variation/4886/)*	text
616	[undiag_v2_snp_id] Show the field ONLY if: [undiag_v2_report] = 'dbsnp'	dbSNP RS ID *If the change has a dbSNP ID, enter it here, e.g. 'rs606231128'*	text
617	[undiag_v2_hgvs_opts] Show the field ONLY if: [undiag_v2_report] = 'custom_hgvs'	Variant 2: What kind of change(s) would you like to report in "HGVS simple"-notation? Please enter all the details that you have available from the report - e.g. if the report lists both the cDNA and protein changes, please provide both.	checkbox, Required cdna: undiag_v2_hgvs_opts___cdna: Report cDNA change (c.) nucleotide: undiag_v2_hgvs_opts___nucleotide: Report nucleotide (genomic) change (g.) protein: undiag_v2_hgvs_opts___protein: Report protein change (p.) other: undiag_v2_hgvs_opts___other: Report other type of change, e.g. RNA (r.) or non-coding (n.)
618	[undiag_v2_refseq_c] Show the field ONLY if: [undiag_v2_hgvs_opts(cdna)] = 1	Section Header: *Variant 2: cDNA change* Reference sequence for cDNA change *Enter the RefSeq transcript ID with the prefix NM_ or NR_ etc., e.g. 'NM_173660.5'. More information and examples see here and here*	text, Required
619	[undiag_v2_desc_c] Show the field ONLY if: [undiag_v2_hgvs_opts(cdna)] = 1	Description of cDNA change *Enter the cDNA change in HGVS nomenclature, e.g. c.1124_1127dupTGCC or c.346G>A More information and examples see here*	text, Required
620	[undiag_v2_hgvs_c] Show the field ONLY if: [undiag_v2_hgvs_opts(cdna)] = 1	undiag_v2_hgvs_c	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v2_refseq_c]!='' and [undiag_v2_desc_c]!='', concat([undiag_v2_refseq_c], ':', [undiag_v2_desc_c]), ''))
621	[undiag_v2_hgvs_c_disp] Show the field ONLY if: [undiag_v2_hgvs_opts(cdna)] = 1	Resulting HGVS notation for variant 1 cDNA change:[undiag_v2_hgvs_c]	descriptive
622	[undiag_v2_refseq_g] Show the field ONLY if: [undiag_v2_hgvs_opts(nucleotide)] = 1	Section Header: *Variant 2: Nucleotide (genomic) change* Reference sequence for nucleotide (genomic) change *Enter the RefSeq transcript ID with the prefix NC_ or NG_ etc., e.g. 'NC_000023.10'. More information and examples see here and here*	text, Required
623	[undiag_v2_desc_g] Show the field ONLY if: [undiag_v2_hgvs_opts(nucleotide)] = 1	Description of nucleotide (genomic) change *Enter the nucleotide change in HGVS nomenclature, e.g. g.34805_34808dup More information and examples see here*	text, Required
624	[undiag_v2_hgvs_g] Show the field ONLY if: [undiag_v2_hgvs_opts(nucleotide)] = 1	undiag_v2_hgvs_g	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v2_refseq_g]!='' and [undiag_v2_desc_g]!='', concat([undiag_v2_refseq_g], ':', [undiag_v2_desc_g]), ''))
625	[undiag_v2_hgvs_g_disp] Show the field ONLY if: [undiag_v2_hgvs_opts(nucleotide)] = 1	Resulting HGVS notation for variant 1 nucleotide (genomic) change:[undiag_v2_hgvs_g]	descriptive
626	[undiag_v2_refseq_p] Show the field ONLY if: [undiag_v2_hgvs_opts(protein)] = 1	Section Header: *Variant 2: Protein change* Reference sequence for protein change *Enter the RefSeq transcript ID with the prefix NP_, e.g. NP_003997.1 More information and examples see here and here*	text, Required
627	[undiag_v2_desc_p] Show the field ONLY if: [undiag_v2_hgvs_opts(protein)] = 1	Description of protein change *Enter the protein change in HGVS nomenclature, e.g. p.Ala378fs More information and examples see here*	text, Required
628	[undiag_v2_hgvs_p] Show the field ONLY if: [undiag_v2_hgvs_opts(protein)] = 1	undiag_v2_hgvs_p	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v2_refseq_p]!='' and [undiag_v2_desc_p]!='', concat([undiag_v2_refseq_p], ':', [undiag_v2_desc_p]), ''))

629	[undiag_v2_hgvs_p_disp] Show the field ONLY if: [undiag_v2_hgvs_opts(protein)] = 1	Resulting HGVS notation for variant 1 protein change:[undiag_v2_hgvs_p]	descriptive
630	[undiag_v2_refseq_o] Show the field ONLY if: [undiag_v2_hgvs_opts(other)] = 1	Section Header: *Variant 2: Other change* Reference sequence for other change *Examples for RNA reference sequences: NC_000023.10(NM_004006.2) or LRG_199t1. More information and examples see here and here*	text, Required
631	[undiag_v2_desc_o] Show the field ONLY if: [undiag_v2_hgvs_opts(other)] = 1	Description of other change *Examples for descriptions of RNA changes: r.357_358ins357+1_357+12 or r.11u>g More information and examples see here*	text, Required
632	[undiag_v2_hgvs_o] Show the field ONLY if: [undiag_v2_hgvs_opts(other)] = 1	undiag_v2_hgvs_o	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT(if([undiag_v2_refseq_o]!='' and [undiag_v2_desc_o]!='', concat([undiag_v2_refseq_o], ':', [undiag_v2_desc_o]), ''))
633	[undiag_v2_hgvs_o_disp] Show the field ONLY if: [undiag_v2_hgvs_opts(other)] = 1	Resulting HGVS notation for variant 1 other change:[undiag_v2_hgvs_o]	descriptive
634	[undiag_v2_zygos] Show the field ONLY if: [undiag_report_variant] = 'yes' AND [undiag_v2_yes] = 'yes'	Section Header: *Variant 2: Additional Information* Zygosity	dropdown heterozygous, Heterozygous compound_heterozygous, Compound heterozygous homozygous, Homozygous hemizygous, Hemizygous mosaic, Mosaic unknown, Unknown
635	[undiag_v2_gen_build] Show the field ONLY if: [undiag_report_variant] = 'yes' AND [undiag_v2_report] = 'custom_hgvs'	Genomic build	dropdown GRCh38, GRCh38 (hg38) GRCh37, GRCh37 (hg19)
636	[undiag_v2_gen_app] Show the field ONLY if: [undiag_report_variant] = 'yes' AND [undiag_v2_report] = 'custom_hgvs'	Genetic approach	dropdown single_gene_screening, Single Gene Screening acgh, aCGH snp_array, SNP array mlpa, MLPA karyotype, Karyotype ngs_panel, NGS panel wes, WES wgs, WGS
637	[undiag_v2_chrom] Show the field ONLY if: [undiag_report_variant] = 'yes' AND [undiag_v2_report] = 'custom_hgvs'	Chromosome	dropdown 1, 1 2, 2 3, 3 4, 4 5, 5 6, 6 7, 7 8, 8 9, 9 10, 10 11, 11 12, 12 13, 13

			14 | 14 15 | 15 16 | 16 17 | 17 18 | 18 19 | 19 20 | 20 21 | 21 22 | 22 X | X Y | Y MT | MT
638	[undiagnosed_complete]	Section Header: *Form Status* Complete?	dropdown 0 | Incomplete 1 | Unverified 2 | Complete
Instrument:* Medication *(medication)			
639	[medication_h1]	MEDICATION	descriptive
640	[medi_desc1]	NOTE: Please fill in this form more than once if the patient has interrupted taking the same medication in the meantime and do not replace start date in the existing form.	descriptive
641	[medi_desc2] Show the field ONLY if: [medi_status] = 'discontinued'	WARNING: You reported that this medication has already been discontinued. The current status of this form is: [medication_complete]. Are you sure you want to edit this form instead of filling an additional medication form?	descriptive
642	[date_of_assess_medi]	Date of assessment yyyy-mm-dd *Date of visit when the medication was documented. If this date is not known and cannot be estimated, please use the current date (date you enter this information here).*	text (date_ymd), Required
643	[medication_label]	Medication label *This is a label to identify this record to the registry user. It will be displayed on the patient's record home page together with the dosage and timeframe. You can name it anything you find helpful - consider something like the medication name.*	text
644	[medication_label_display]	medication_label_display	text Field Annotation: @CALCTEXT(concat([medication_label], ' (', [medi_dosage], ' ', [medi_dosage_unit], '; ', [medi_freq], ' ', if([medi_freq_timeunit] = 'other', [medi_freq_timeunit_custom], [medi_freq_timeunit]), '): ', concat(right([date_of_medi_start],2), '.', mid([date_of_medi_start],6,2), '.', left([date_of_medi_start],4)), ' - ', if([medi_status]="ongoing", "ongoing", concat(right([date_of_medi_stop],2), '.', mid([date_of_medi_stop],6,2), '.', left([date_of_medi_stop],4)))))) @HIDDEN @HIDDEN-PDF
645	[medication_matrix]	Codesystem Code registered? Medication name Chemical Entities of Biological Interest (CheBI) {medi_has_chebi_code} {medi_chebi_code} Medical Subject Headings (MeSH) {medi_has_mesh_code} {medi_mesh_code} ?	descriptive
646	[medi_has_chebi_code]	CheBI code registered for this medication	yesno, Required 1 | Yes 0 | No
647	[medi_chebi_code] Show the field ONLY if: [medi_has_chebi_code]=1	CheBI-code	text, Required BIOPORTAL:CHEBI | BIOPORTAL:CHEBI
648	[medi_has_mesh_code]	MeSH code registered for this medication	yesno, Required 1 | Yes 0 | No
649	[medi_mesh_code] Show the field ONLY if: [medi_has_mesh_code]=1	MeSH-code	text, Required BIOPORTAL:MESH | BIOPORTAL:MESH
650	[is_specific_treatment_nmd]	Is this medication a specific treatment for the neuromuscular disease? *If this medication is used to treat other symptoms of the patient, please select "No".*	yesno, Required 1 | Yes

			0 No
651	[treatment_effect] Show the field ONLY if: [is_specific_treatment_nmd]=1	Treatment effect *Effect of the specific treatment for neuromuscular disease*	radio positive Positive negative Negative none None unknown Unknown
652	[is_cardiac_medication]	Cardiac medication *Is this medication a cardiac medication?*	yesno, Required 1 Yes 0 No
653	[date_of_medi_start_i]	Section Header: *Timeframe* Medication start date imputation	radio, Required no_imputation Exact date of medication start known day Day of medication start date unknown day_month Day and month of medication start date unknown Field Annotation: @DEFAULT = 'no_imputation'
654	[date_of_medi_start]	Medication start date yyyy-mm-dd *On what date did the patient start taking this medication? If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.*	text (date_ymd), Required
655	[medi_status]	Medication status	radio, Required ongoing Medication ongoing discontinued Medication discontinued Field Annotation: @DEFAULT = 'ongoing'
656	[date_of_medi_stop_i] Show the field ONLY if: [medi_status] = 'discontinued'	Medication end date imputation	radio, Required no_imputation Exact date of medication discontinuation known day Day of medication end date unknown day_month Day and month of medication end date unknown
657	[date_of_medi_stop] Show the field ONLY if: [medi_status] = 'discontinued'	Medication end date yyyy-mm-dd *On what date did the patient stop taking this medication? If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.*	text (date_ymd), Required
658	[medi_dosage_known]	Section Header: *Dosage* Dosage known	radio, Required yes Yes no No not_applicable Not applicable Field Annotation: @DEFAULT = 'yes'
659	[medi_dosage] Show the field ONLY if: [medi_dosage_known] = 'yes'	Dosage *If the same treatment is administered several times in different doses on the same day, please indicate the cumulative dose.*	text (number), Required
660	[medi_dosage_unit] Show the field ONLY if: [medi_dosage_known] = 'yes'	Dosage unit	dropdown, Required g g (gram) mg mg (milligram) microg µg (microgramm) mmol mmol (millimol) micromol µmol (micromol) ml ml (millilitre) microl µl (microlitre) sessions session(s)
661	[medi_admin_freq_known]	Frequency of administration known	radio, Required yes Yes

			no: No not_applicable: Not applicable Field Annotation: @DEFAULT = 'yes'
662	[medi_freq] Show the field ONLY if: [medi_admin_freq_known] = 'yes'	Frequency of administration	text (number), Required
663	[medi_freq_timeunit] Show the field ONLY if: [medi_admin_freq_known] = 'yes'	Frequency of administration: time unit	dropdown (autocomplete), Required once: once per_hour: per hour per_day: per day per_week: per week t2weeks: every 2 weeks t3weeks: every 3 weeks t4weeks: every 4 weeks t6weeks: every 6 weeks t8weeks: every 8 weeks t3months: every 3 months t4months: every 4 months t6months: every 6 months other: other
664	[medi_freq_timeunit_custom] Show the field ONLY if: [medi_admin_freq_known] = 'yes' AND [medi_freq_timeunit] = 'other'	Frequency of administration: custom time unit	text, Required
665	[medication_complete]	Section Header: *Form Status* Complete?	dropdown 0: Incomplete 1: Unverified 2: Complete
Instrument:* WHODAS 2.0 12-item *(whodas_20_12item)			
666	[whodas_title]	WHODAS 2.0 12-item self-administered	descriptive
667	[whodas_desc1]	This questionnaire asks about difficulties due to health conditions. Health conditions include diseases orillnesses, other health problems that may be short or long lasting, injuries, mental or emotional problems,and problems with alcohol or drugs.Think back over the past 30 days and answer these questions, thinking about how much difficulty youhad doing the following activities.	descriptive
668	[date_of_whodas]	Current Date yyyy-mm-dd	text (date_ymd), Required
669	[whodas_answered_by]	Who answers this survey?	radio patient: Patient caregiver: Caregiver (on behalf of the patient)
670	[whodas_subscores_known]	Result of the individual data items is known	radio, Required yes: Yes no: No Field Annotation: @HIDDEN-SURVEY @DEFAULT = 'yes'
671	[whodas_score_manual] Show the field ONLY if: [whodas_subscores_known] = 'no'	Manual Overall WHODAS-Score [%] *Please enter a number in the range [0, 100]. Element 8.1. in the Set of common data elements for Rare Diseases Registration (link)*	text (number, Min: 0, Max: 100), Required Field Annotation: @HIDDEN-SURVEY
672	[whodas_desc2] Show the field ONLY if: [whodas_subscores_known] = 'yes'	In the past 30 days, how much difficulty did you have in:	descriptive

673	[whodas_s1] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Standing for long periods such as 30 minutes?	radio, Required 0 None 1 Mild 2 Moderate 3 Severe 4 Extreme or cannot do Custom alignment: LH
674	[whodas_s2] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Taking care of your household responsibilities?	radio, Required 0 None 1 Mild 2 Moderate 3 Severe 4 Extreme or cannot do Custom alignment: LH
675	[whodas_s3] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Learning a new task, for example, learning how to get to a new place?	radio, Required 0 None 1 Mild 2 Moderate 3 Severe 4 Extreme or cannot do Custom alignment: LH
676	[whodas_s4] Show the field ONLY if: [whodas_subscores_known] = 'yes'	How much of a problem did you have joining in community activities (for example, festivities, religious or other activities) in the same way as anyone else can?	radio, Required 0 None 1 Mild 2 Moderate 3 Severe 4 Extreme or cannot do Custom alignment: LH
677	[whodas_s5] Show the field ONLY if: [whodas_subscores_known] = 'yes'	How much have you been emotionally affected by your health problems?	radio, Required 0 None 1 Mild 2 Moderate 3 Severe 4 Extreme or cannot do Custom alignment: LH
678	[whodas_s6] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Concentrating on doing something for ten minutes?	radio, Required 0 None 1 Mild 2 Moderate 3 Severe 4 Extreme or cannot do Custom alignment: LH
679	[whodas_s7] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Walking a long distance such as a kilometre [or equivalent]?	radio, Required 0 None 1 Mild

			2 Moderate 3 Severe 4 Extreme or cannot do Custom alignment: LH
680	[whodas_s8] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Washing your whole body?	radio, Required 0 None 1 Mild 2 Moderate 3 Severe 4 Extreme or cannot do Custom alignment: LH
681	[whodas_s9] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Getting dressed?	radio, Required 0 None 1 Mild 2 Moderate 3 Severe 4 Extreme or cannot do Custom alignment: LH
682	[whodas_s10] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Dealing with people you do not know?	radio, Required 0 None 1 Mild 2 Moderate 3 Severe 4 Extreme or cannot do Custom alignment: LH
683	[whodas_s11] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Maintaining a friendship?	radio, Required 0 None 1 Mild 2 Moderate 3 Severe 4 Extreme or cannot do Custom alignment: LH
684	[whodas_s12] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Your day-to-day work?	radio, Required 0 None 1 Mild 2 Moderate 3 Severe 4 Extreme or cannot do Custom alignment: LH
685	[whodas_h1] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Overall, in the past 30 days, how many days were these difficulties present? *Record number of days*	text (number, Min: 0, Max: 30) Custom alignment: LH
686	[whodas_h2] Show the field ONLY if: [whodas_subscores_known] = 'yes'	In the past 30 days, for how many days were you totally unable to carry out your usual activities or work because of any health condition? *Record number of days*	text (number, Min: 0, Max: 30) Custom alignment: LH

687	[whodas_h3] Show the field ONLY if: [whodas_subscores_known] = 'yes'	In the past 30 days, not counting the days that you were totally unable, for how many days did you cut back or reduce your usual activities or work because of any health condition? *Record number of days*	text (number, Min: 0, Max: 30) Custom alignment: LH
688	[whodas_sc_simple_calc] Show the field ONLY if: [whodas_subscores_known] = 'yes'	Calculated Overall WHODAS-Score [%] *Will only contain values if none of whodas_s1 - whodas_s12 is missing. Element 8.1. in the Set of common data elements for Rare Diseases Registration (link)*	calc Calculation: if((!isblankormissingcode([whodas_s1]) AND !isblankormissingcode([whodas_s2]) AND !isblankormissingcode([whodas_s3]) AND !isblankormissingcode([whodas_s4]) AND !isblankormissingcode([whodas_s5]) AND !isblankormissingcode([whodas_s6]) AND !isblankormissingcode([whodas_s7]) AND !isblankormissingcode([whodas_s8]) AND !isblankormissingcode([whodas_s9]) AND !isblankormissingcode([whodas_s10]) AND !isblankormissingcode([whodas_s11]) AND !isblankormissingcode([whodas_s12])), (round(sum([whodas_s1], [whodas_s2], [whodas_s3], [whodas_s4], [whodas_s5], [whodas_s6], [whodas_s7], [whodas_s8], [whodas_s9], [whodas_s10], [whodas_s11], [whodas_s12]) / 48 * 100, 2)), '') Field Annotation: @HIDDEN-SURVEY
689	[whodas_pdf]	Auto-uploaded survey PDF	file Field Annotation: @HIDDEN-SURVEY @HIDDEN-PDF
690	[whodas_20_12item_complete]	Section Header: *Form Status* Complete?	dropdown 0, Incomplete 1, Unverified 2, Complete
Instrument:* PGI-C (only available if date of birth filled in Baseline) *(pgic_only_available_if_date_of_birth_filled_in_bas)			
691	[pgic_h1]	Patient Global Impression scale: Change (PGI-C)	descriptive
692	[date_of_pgic]	Date of assessment yyyy-mm-dd	text (date_ymd), Required
693	[age_at_pgic]	Age of patient (years)	calc Calculation: rounddown(datediff([first-event-name][date_of_birth], [date_of_pgic], "y")) Field Annotation: @READONLY
694	[pgic_answered_by]	Who answers this survey?	radio patient, Patient caregiver, Caregiver (on behalf of the patient)
695	[pgic_1a] Show the field ONLY if: starts_with([event-name], 'enrolment') AND [age_at_pgic] < 18	How has your neuromuscular condition changed over the last 6 months?	radio 1, Very Much Improved 2, Much Improved 3, Minimally Improved 4, No Change 5, Minimally Worse 6, Much Worse 7, Very Much Worse
696	[pgic_1b] Show the field ONLY if: starts_with([event-name], 'enrolment') AND [age_at_pgic] >= 18	How has your neuromuscular condition changed over the last year?	radio 1, Very Much Improved 2, Much Improved 3, Minimally Improved 4, No Change 5, Minimally Worse 6, Much Worse 7, Very Much Worse
697	[pgic_2] Show the field ONLY if: starts_with([event-name], 'visit')	How has your neuromuscular condition changed since you were first enrolled in this patient registry on [first-event-name][date_of_consent]	radio 1, Very Much Improved 2, Much Improved 3, Minimally Improved 4, No Change 5, Minimally Worse 6, Much Worse

			7 | Very Much Worse
698	[pgic_3a] Show the field ONLY if: starts_with([event-name], 'visit')	Have you started a new treatment or medicine based on advice from your specialist doctor at this site since you last answered this questionnaire?	radio yes | Yes no | No
699	[pgic_3b] Show the field ONLY if: [pgic_3a] = 'yes'	What new treatment or medicine are you thinking about?	text
700	[pgic_3c] Show the field ONLY if: [pgic_3a] = 'yes'	How has your neuromuscular condition changed since you started your new treatment plan?	radio 1 | Very Much Improved 2 | Much Improved 3 | Minimally Improved 4 | No Change 5 | Minimally Worse 6 | Much Worse 7 | Very Much Worse
701	[pgic_3b_prev]	pgic_3b_prev	text Field Annotation: @HIDDEN @HIDDEN-PDF @CALCTEXT([event-name][pgic_3b][previous-instance])
702	[pgic_4a] Show the field ONLY if: starts_with([event-name], 'visit') AND [pgic_3b_prev] != ""	Last time you answered this questionnaire you said you had started the following new treatment: [event-name][pgic_3b][previous-instance] Are you still on this treatment?	radio yes | Yes no | No
703	[pgic_4b] Show the field ONLY if: [pgic_4a] = 'yes'	How has your neuromuscular condition changed since you started this treatment plan?	radio 1 | Very Much Improved 2 | Much Improved 3 | Minimally Improved 4 | No Change 5 | Minimally Worse 6 | Much Worse 7 | Very Much Worse
704	[pgic_only_available_if_date_of_birth_filled_in_bas_complete]	Section Header: *Form Status* Complete?	dropdown 0 | Incomplete 1 | Unverified 2 | Complete
Instrument:* End of Data Collection *(end_of_data_collection)			
705	[eodc_h1]	END OF DATA COLLECTION	descriptive
706	[eodc_current_date]	Current date	text (date_ymd), Required
707	[eodc_reason]	Reason for end of data collection *Element 3.1. in the Set of common data elements for Rare Diseases Registration (link))*	radio, Required death | Death of patient lost_fu | Patient lost in follow-up opted_out | Patient opted-out
708	[eodc_date_of_death_known] Show the field ONLY if: [eodc_reason] = 'death'	Date of death known *Element 3.2. in the Set of common data elements for Rare Diseases Registration (link)).*	radio, Required yes | Yes no | No
709	[eodc_date_of_death] Show the field ONLY if: [eodc_date_of_death_known] = 'yes'	Date of deathyyyy-mm-dd *Element 3.2. in the Set of common data elements for Rare Diseases Registration (link)). If the exact day is not known, please enter 15th. If the month is not known, please enter 1st of July of that year.*	text (date_ymd), Required
710	[eodc_date_of_loss_fu_known] Show the field ONLY if: [eodc_reason] = 'lost_fu'	Date of loss to follow-up known	radio, Required yes | Yes no | No

711	[eodc_date_of_loss_fu] Show the field ONLY if: [eodc_date_of_loss_fu_known] = 'yes'	Date of loss to follow-up yyyy-mm-dd	text (date_ymd), Required
712	[eodc_date_of_opt_out_known] Show the field ONLY if: [eodc_reason] = 'opted_out'	Date of opt-out known	radio, Required yes, Yes no, No
713	[eodc_date_of_opt_out] Show the field ONLY if: [eodc_date_of_opt_out_known] = 'yes'	Date of opt-out yyyy-mm-dd	text (date_ymd), Required
714	[eodc_date]	eodc_date: Hidden calculated field that will always contain the date of end of data collection regardless of the reason for end of data collection. We use this to simplify writing of logic e.g. for Data Quality rules.	text Field Annotation: @CALCTEXT(if([eodc_date_of_death_known] = 'yes', [eodc_date_of_death], if([eodc_date_of_loss_fu_known] = 'yes', [eodc_date_of_loss_fu], if([eodc_date_of_opt_out_known] = 'yes', [eodc_date_of_opt_out], '')))) @READONLY @HIDDEN @HIDDEN-PDF
715	[end_of_data_collection_complete]	Section Header: *Form Status* Complete?	dropdown 0, Incomplete 1, Unverified 2, Complete