Table e-1. Diagnostic yield of different genetic tests.

Author and year	Test	Diagnostic yield (when the study included patients without epilepsy we only considered patients with epilepsy)	Additional details
Chromosomal microarray			
Mefford et al, 2010 [1]	CMA	46/517 (0.089)	All 517 patients had idiopathic epilepsy, mostly without intellectual disability
Mefford et al, 2011 [2]	CMA	13/315 (0.041)	All 315 patients had epileptic encephalopathies
Bartnik et al, 2012 [3]	CMA	10/102 (0.098) -3/50 (0.06) in patients with isolated epilepsy -7/52 (0.135) in patients with epilepsy and other neurologic conditions	50 patients had isolated epilepsy 52 patients had epilepsy plus intellectual disability, dysmorphism, ASD, or other neurologic abnormalities
Michaud et al, 2014 [4]	CMA	6/44 (0.136)	44 patients with infantile spasms (40 of them with developmental delay)
Helbig et al, 2014 [5]	CMA	16/223 (0.072)	223 patients with childhood epilepsies and complex phenotypes including structural brain lesions
Olson et al, 2014 [6]	CMA	40/805 (0.05)	805 patients with epilepsy at a reference center
Hrabik et al, 2015 [7]	CMA	11/147 (0.075)	147 patients with epilepsy at a reference center
Berg et al, 2017 [8]	CMA	32/188 (0.1702)	188 patients with epilepsy onset before the third birthday in a multicenter study
Epilepsy gene panels			
Lemke et al, 2012 [9]	EP (265 genes)	16/33 (0.485)	33 patients with epilepsy in several reference centers
Wang et al, 2014 [10]	EP (53 genes or 38 genes)	6/28 (0.214)	28 patients with epilepsy in a reference center
Della Mina et al, 2015 [11]	EP (67 genes)	9/19 (0.474) -6/7 in patients with a clinical presentation suggestive of a specific syndrome -3/12 in patients with a phenotype not suggestive of any specific syndrome	19 patients with isolated or syndromic epilepsy
Mercimek-Mahmutoglu et al, 2015 [12]	EP (20 patients with 38 genes, 1 patient with 40 genes, 3 patients with 50 genes, 7 patients with 51 genes, 6 patients with 53 genes, 2 patients with 63 genes, 39 patients	12/93 (0.129)	All 93 children with intractable epilepsy, global developmental delay, and cognitive dysfunction and no recognizable syndromic clinical features, MRI, or MRS patterns, metabolic evaluation, and negative CMA

	with 70 genes, and 15 patients with 327 genes)		
Trump et al, 2016 [13]	EP (46 genes)	60/323 (0.1858)	323 patients with early-onset seizure disorders but without major structural brain malformations from tertiary centers
Segal et al, 2016 [14]	EP (87 genes or 455 genes)	7/49 (0.1429)	49 patients with refractory epilepsy and negative CMA results
Møller et al, 2016 [15]	EP (46 genes)	49/216 (0.2269)	216 patients with different types of epilepsy
Berg et al, 2017 [8]	EP (number of genes not specified)	31/114 (0.2719)	114 patients with epilepsy onset before the third birthday in a multicenter study
Butler et al, 2017 [16]	EP (110 genes)	62/339 (0.1829)	339 patients referred with epilepsy
Whole exome sequencing			
Veeramah et al, 2013 [17]	WES	7/10 (0.7)	10 trios of unaffected parents and a child with refractory epilepsy, normal or unspecific neuroimaging, and a variable combination of autistic features, cognitive impairment, and motor deficits
Michaud et al, 2014 [4]	WES	13/18 (0.722) families with a diagnosis	18 trios with the child having infantile spasms previously evaluated with a CMA and targeted sequencing of up to 2 genes associated with infantile spasms
Dyment et al, 2015 [18]	WES	7/9 (0.778) families with a diagnosis 8/11 (0.727) affected individuals	11 patients from 9 families with a child with seizures as the predominant clinical feature. All patients came from a network of rare diseases and had underwent prior CMA
Retterer et al, 2015 [19]	WES	232/830 (0.28)	830 patients in a single clinical laboratory
Helbig et al, 2016 [20]	WES	112/293 (0.3823)	293 patients in a single clinical laboratory
Berg et al, 2017 [8]	WES	11/33 (0.3333)	33 patients with epilepsy onset before the third birthday in a multicenter study

Legend: ASD: Autism spectrum disorder. **CMA:** Chromosomal microarray. **CNVs:** Copy number variations. **EP:** Epilepsy panel. **ID:** Intellectual disability. **MRI:** Magnetic resonance imaging. **MRS:** Magnetic resonance spectroscopy. **WES:** Whole-exome

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