

Genetic Testing for Epilepsy

AVAILABLE TESTS

Comprehensive Epilepsy Panel (144 Genes)

Infantile Epilepsy Panel (126 Genes)

Childhood Epilepsy Panel (87 Genes)

CLINICAL FEATURES

Epilepsy is defined by the occurrence of at least two unprovoked seizures occurring more than 24 hours apart. It is a common neurological disorder that affects at least 0.8% of the population. The International League against Epilepsy (ILAE) classifies seizures into two main categories.¹ **Generalized epileptic seizures** originate in and rapidly engage both cerebral hemispheres. Tonic-clonic, absence, myoclonic, clonic, tonic, and atonic seizures are common types of generalized seizures. **Focal seizures** originate from neuronal networks within a single hemisphere. Traditionally, focal seizures have been classified as “simple partial seizures,” which do not result in an alteration of consciousness, and “complex partial seizures,” which cause a change in behavior or consciousness. Some types of seizures, such as infantile spasms, do not fit into either category and remain unclassified.

Seizures can be self-limiting or controlled by standard therapeutic treatments in some cases; however, individuals with epileptic encephalopathy have severe seizures that are refractory to treatment, leading to cognitive and behavioral impairment secondary to the epileptic activity. Epilepsy may be an isolated neurological symptom, or it may occur in association with other neurological symptoms or medical problems.² Some individuals with epilepsy are diagnosed with an electroclinical syndrome such as West or Ohtahara syndrome based on the presence of characteristic EEG findings and the clinical and family history.¹

GENETICS

Epilepsy can be caused by genetic disorders, metabolic diseases, trauma, infection, and structural brain abnormalities, although the cause is not known in many cases. A genetic etiology underlies epilepsy in approximately 40% of individuals.³ Genes have been identified that cause both generalized seizures and focal seizures, as well as unclassified epilepsy types such as infantile spasms. The genetic etiology of idiopathic generalized epilepsy (IGE) is frequently complex because it is due to a combination of multiple genetic factors that each confer a small risk for epilepsy and may be modified by environmental influences.³ Currently, approximately 2% of patients with IGE harbor an identifiable variant in a single gene associated with Mendelian inheritance of epilepsy.⁴ However, the percentage of patients with Mendelian epilepsy is higher for specific epilepsy types such as infantile spasms, benign familial neonatal and neonatal-infantile seizures (BFNS and BFNIS), and others.^{3,5,6,7} The inheritance pattern can be autosomal dominant, autosomal recessive, or X-linked. Pathogenic variants in a single gene may be associated with different types of seizures (clinical heterogeneity), and conversely, pathogenic variants in different genes can cause the same epilepsy phenotype (genetic heterogeneity).

The Epilepsy Panels at GeneDx includes sequencing and deletion/duplication analysis of genes causing Mendelian forms of epilepsy. Many of these genes encode subunits of ion channels involved in stabilizing or propagating neuronal activity, including components of the voltage-gated sodium, potassium, and calcium channels and the ligand-gated gamma-aminobutyric (GABA) channel.^{5,7,8,9} The panels also includes non-ion channel genes associated with a variety of neurotransmitter disorders, storage and other neurometabolic disorders, as well as genes causing syndromic forms of epilepsy, many of which are involved in transcriptional activation or repression.^{5,6,8,10,11,12}

Several different Epilepsy Panels are available at GeneDx, based on seizure age-of-onset and/or clinical presentation. The complete list of genes included on each panel is included in the table in the appendix.

In addition to the epilepsy panels mentioned above, GeneDx also offers panels or single gene testing for many other disorders causing epilepsy, such as tuberous sclerosis complex, mitochondrial disorders, and metabolic disorders.

TEST METHODS

Using genomic DNA from the submitted specimen, the complete coding regions and splice site junctions of the genes on this panel are enriched using a proprietary targeted capture system developed by GeneDx for next-generation sequencing with CNV calling (NGS-CNV). The enriched targets are simultaneously sequenced with paired-end reads on an Illumina platform. Bi-directional sequence reads are assembled and aligned to reference sequences based on NCBI RefSeq transcripts and human genome build GRCh37/UCSC hg19. After gene specific filtering, data are analyzed to identify sequence variants and most deletions and duplications involving coding exons. Alternative sequencing or copy number detection methods are used to analyze or confirm regions with inadequate sequence or copy number data by NGS. Reportable variants include pathogenic variants, likely pathogenic variants and variants of uncertain significance. Likely benign and benign variants, if present, are not routinely reported but are available upon request.

The technical sensitivity of sequencing is estimated to be > 99% at detecting single nucleotide events. It will not reliably detect deletions greater than 20 base pairs, insertions or rearrangements greater than 10 base pairs, or low-level mosaicism. The copy number assessment methods used with this test cannot reliably detect copy number variants of less than 500 base pairs or mosaicism and cannot identify balanced chromosome aberrations. Assessment of exon-level copy number events is dependent on the inherent sequence properties of the targeted regions, including shared homology and exon size. For the *CHRNA7* and *MAGI2* gene(s), deletion/duplication analysis but not sequencing is performed. For the *ARG1*, *C12orf57*, *FGF12*, *FOXP1*, *PHGDH*, *PIGG*, *PIGT*, *SLC35A2*, *SLC6A8*, and *SNAP25* gene(s), sequencing but not deletion/duplication analysis is performed. For the *ARX*, *ASNS*, *CHRNA7*, *FOLR1*, *GAMT*, *KCNT1*, *NR2F1*, *PCDH19*, *SCN1B*, *SHANK3*, and *TUBB2A* genes, only whole gene deletions or duplications may be detected, and deletions/duplications involving the 3' end of the *TSC2* gene (exons 36-42) may not be identified. If indicated based on the patient's specific clinical features, multiplex ligation-dependent probe amplification (MLPA) of the *FOXP1* or *SLC6A8* genes is available as a separate test (test code 906).

CLINICAL SENSITIVITY

The clinical sensitivity of sequencing and deletion/duplication analysis of the genes included in the Comprehensive Epilepsy Panel depends in part on the patient's clinical phenotype. In a prior study, 31% of individuals with infantile spasms who were tested using an epilepsy gene panel were found to harbor definitive pathogenic variant(s) to explain the phenotype (Wirrell et al., 2015). Overall, 17-20% of epileptic encephalopathies have an identifiable genetic etiology (EpiPM Consortium, 2015). Specific information about the diagnostic yield for each gene in selected populations is summarized in the table below.

DIAGNOSTIC YIELD OF EPILEPSY PANEL GENES IN SELECTED POPULATIONS

Epilepsy Type	Gene	Protein	Inh	Diagnostic Yield in Selected Population(s)
Benign familial neonatal seizures (BFNS)	<i>KCNQ2</i>	Potassium voltage-gated channel subfamily KQT member 2	AD	>50% BFNS ⁶
	<i>KCNQ3</i>	Potassium voltage-gated channel subfamily KQT member 3	AD	~7% BFNS ⁶

Benign familial neonatal-infantile seizures (BFNIS)	<i>SCN2A</i>	Sodium channel protein type 2 subunit alpha	AD	Unknown ⁶
Benign familial infantile seizures (BFIS)	<i>PRRT2</i>	Proline-rich transmembrane protein 2	AD	83% familial and 30% sporadic BFIS ^{14,15,16} ; 62-96% of familial PKD/IC; 36% sporadic PKD/IC ^{14,17,18}
Familial infantile myoclonic epilepsy (FIME)	<i>TBC1D24</i>	TBC1 domain family member 24	AR	Unknown ^{19,20}
Developmental and epileptic encephalopathy (includes West, Ohtahara, and Lennox Gastaut and other electroclinical syndromes)	<i>ALDH7A1</i>	Alpha-aminoacidic semialdehyde dehydrogenase (antiqutin)	AR	>90% pyridoxine-responsive epilepsy ³²
	<i>ALG13</i>	ALG13, UDP-N-acetylglucosaminyltransferase subunit	XL	Rare ⁹²
	<i>ANKRD11</i>	Ankyrin repeat domain-containing protein	AD	Rare ¹⁵⁴
	<i>ARHGEF9</i>	Cdc42 guanine nucleotide exchange factor (GEF) 9	XL	Rare ⁹⁴
	<i>ARX</i>	Aristaless related homeobox	XL	5% males with infantile spasms ⁶
	<i>ATP1A3</i>	Sodium/potassium-transporting ATPase subunit alpha-3	AD	Rare in early life epilepsy ¹³¹ , 78% alternating hemiplegia of childhood ¹³²
	<i>ATP6AP2</i>	Renin receptor	XL	Rare ⁴⁵
	<i>BRAT1</i>	BRCA1 associated ATM activator 1	AR	Rare ¹²³
	<i>CACNA1A</i>	Voltage-dependent P/Q-type calcium channel subunit alpha-1A	AD	Rare ⁹⁵
	<i>CACNA1E</i>	Voltage-dependent R-type calcium channel subunit alpha-1E	AD	Rare ¹⁵⁸
	<i>CACNA1G</i>	Voltage-dependent T-type calcium channel subunit alpha-1G	AD	Rare ¹⁵⁹
	<i>CASK</i>	Peripheral plasma membrane protein CASK	XL	Rare ¹⁵¹
	<i>CDKL5</i>	Cyclin-dependent kinase-like 5	XL	10-17% infantile spasms ⁶
	<i>CHD2</i>	Chromodomain helicase DNA binding protein 2	AD	1% of epileptic encephalopathy ⁹¹
	<i>CLCN4</i>	Chloride voltage-gated channel 4	XL	Rare ¹³³
	<i>DNM1**</i>	Dynamins 1	AD	1% of epileptic encephalopathy ⁹²
	<i>DOCK7</i>	Dedicator of cytokinesis protein 7	AR	Rare ¹⁶⁴
	<i>EEF1A2</i>	Eukaryotic translation elongation factor 1 alpha 2	AD	Rare ^{96, 97}
	<i>FGF12</i>	Fibroblast growth factor 12	AD	Rare ¹⁶⁶
	<i>FRRS1L</i>	DOMON domain-containing protein FRRS1L	AR	Rare ¹³⁵
	<i>GABBR2</i>	Gamma-aminobutyric acid type B receptor subunit 2	AD	Rare ¹¹⁸
	<i>GABRA1</i>	Gamma-aminobutyric acid receptor subunit alpha-1	AD	Rare ⁸⁸
	<i>GABRB2</i>	Gamma-aminobutyric acid (GABA) A receptor, beta 2	AD	Rare ¹¹²
	<i>GABRB3</i>	Gamma-aminobutyric acid (GABA) A receptor, beta 3	AD	Rare ⁹⁵
	<i>GABRG2</i>	Gamma-aminobutyric acid receptor subunit gamma-2	AD	Rare ⁶
	<i>GNAO1</i>	Guanine nucleotide binding protein (G protein), alpha activating activity polypeptide O	AD	Rare in epileptic encephalopathy and early-onset hyperkinetic movement disorders ¹³⁶
	<i>GRIN1</i>	Glutamate receptor, ionotropic, NMDA 1	AD	Rare ⁹⁴
<i>GRIN2A</i>	Glutamate receptor ionotropic, NMDA 2A	AD	Rare ⁹³	
<i>GRIN2B</i>	Glutamate receptor ionotropic, NMDA 2B	AD	Rare ⁹⁰	
<i>HCN1</i>	Potassium/sodium hyperpolarization-activated cyclic nucleotide-gated channel 1	AD	Rare ¹⁶⁷	
<i>IQSEC2</i>	IQ motif and sec7 domain 7	XL	Rare ⁹⁵	
<i>KCNA2</i>	Potassium voltage-gated channel subfamily A member 2	AD	Rare ¹³⁸	

	<i>KCNB1</i>	Potassium channel, voltage gated shab related subfamily B, member 1	AD	Rare ⁹⁸
	<i>KCNQ2</i>	Potassium voltage-gated channel subfamily KQT member 2	AD	10% neonatal epileptic encephalopathy ³¹
	<i>KCNT1</i>	Potassium channel, sodium activated subfamily T, member 1	AD	35% MMPSI ⁹⁹⁻¹⁰¹ ; Rare in other epileptic encephalopathies ⁹⁹
	<i>MAGI2 (dels only)</i>	Membrane-associated guanylate kinase, WW and PDZ domain-containing protein 2	AD	Rare ⁴³
	<i>MEF2C</i>	Myocyte-specific enhancer factor 2C	AD	2% epileptic encephalopathy ³⁶
	<i>NR2F1</i>	Nuclear receptor subfamily 2, group F, member 1	AD	Rare ¹⁰³
	<i>PCDH19</i>	Protocadherin-19	XL	2-14% females with infantile/childhood epilepsy ^{26,27,28,29,30}
	<i>PIGA</i>	Phosphatidylinositol glycan anchor biosynthesis, class A	XL	Rare ¹⁰⁹
	<i>PIGG</i>	GPI ethanolamine phosphate transferase 2	AR	Rare ¹⁶⁹
	<i>PIGN</i>	Phosphatidylinositol glycan anchor biosynthesis, class N	AR	Rare ¹⁴⁵
	<i>PIGO</i>	Phosphatidylinositol glycan anchor biosynthesis, class O	AR	Rare ¹¹⁰
	<i>PIGT</i>	GPI transamidase component PIG-T	AR	Rare ¹⁷⁰
	<i>PIGV</i>	Phosphatidylinositol glycan anchor biosynthesis, class V	AR	Rare ¹¹¹
	<i>PLCB1</i>	1-phosphatidylinositol 4,5-bisphosphate phosphodiesterase beta-1	AR	Rare ¹⁵²
	<i>PNPO</i>	Pyridoxine-5'-phosphate oxidase	AR	Rare ⁴⁰
	<i>POLG</i>	DNA polymerase subunit gamma-1	AR	63-87% Alpers syndrome ^{33,34,35} ; 4-5% infantile/childhood epileptic encephalopathy ³⁴
	<i>QARS</i>	Glutamyl-tRNA synthetase	AR	Rare ¹⁰⁴
	<i>SCN1A</i>	Sodium channel protein type 1 alpha	AD	70-80% Dravet syndrome ⁶ ; 20-24% early-onset cryptic epilepsy ^{24,25}
	<i>SCN2A</i>	Sodium channel protein type 2 alpha	AD	1-2% early-onset epileptic encephalopathy ^{37,38}
	<i>SCN8A</i>	Sodium channel protein type 8 subunit alpha	AD	Unknown ³⁹
	<i>SLC13A5</i>	Solute carrier family 13 (sodium-dependent citrate transporter), member 5	AR	Rare ¹⁰⁸
	<i>SLC25A22</i>	Mitochondrial glutamate carrier 1	AR	Rare ⁴⁴
	<i>SLC2A1</i>	Solute carrier family 2, facilitated glucose transporter member 1	AD	91% GLUT1 deficiency ³ ; ~10% early-onset absence epilepsy ⁶
	<i>SLC35A2</i>	UDP-galactose translocator	XL	Rare ¹⁷²
	<i>SLC6A1</i>	Solute carrier family 6 member 1	AD	4% myoclonic-astatic epilepsy (MAE) ¹⁴⁹
	<i>SMC1A</i>	Structural maintenance of chromosomes protein 1A	AD	Rare ^{151,152}
	<i>SNAP25</i>	Synaptosomal-associated protein 25	AD	Rare ¹⁷³
	<i>SPATA5</i>	ATPase family protein 2 homolog	AR	Rare ¹⁷⁴
	<i>SPTAN1</i>	Alpha-II spectrin	AD	Rare ⁴¹
	<i>STX1B</i>	Syntaxin 1B	AD	Rare in MAE and fever-associated epilepsy syndromes ¹⁴⁹
	<i>STXBP1</i>	Syntaxin binding protein 1	AD	35% Ohtahara syndrome ⁶ ; Unknown in Dravet syndrome ⁸⁸
	<i>SYNGAP1</i>	Ras/Rap GTPase-activating protein SynGAP	AD	Rare ¹⁵¹
	<i>SZT2</i>	KICSTOR complex protein SZT2	AR	Rare ¹⁴⁹
	<i>TBL1XR1</i>	F-box-like/WD repeat-containing protein TBL1XR1	AD	Rare ¹⁵⁰
	<i>TSC1</i>	Hamartin	AD	2-4% infantile spasms ^{21,22,23}
	<i>TSC2</i>	Tuberin	AD	10-16% infantile spasms ^{21,22,23}
	<i>WWOX</i>	WW domain-containing oxidoreductase	AR	Rare
Autosomal Dominant Focal Epilepsies	<i>CHRNA4</i>	Neuronal acetylcholine receptor alpha-4	AD	~10% autosomal dominant nocturnal frontal lobe epilepsy (ADNFLE) ⁶
	<i>CHRN2</i>	Neuronal acetylcholine receptor beta-2	AD	< 5% ADNFLE ⁶
	<i>CHRNA2</i>	Neuronal acetylcholine receptor alpha-2	AD	Rare in ADNFLE ⁶

	<i>DEPDC5</i>	DEP domain containing 5	AD	5-37% autosomal dominant focal epilepsies, with or without cortical dysplasia ¹²⁶
	<i>KCNT1</i>	Potassium channel, sodium-activated subfamily T, member 1	AD	< 5% ADNPLE ^{99,102}
	<i>LG11</i>	Leucine-rich glioma-inactivated protein 1	AD	~50% autosomal dominant partial epilepsy with auditory features (ADTLE) ⁶
	<i>NPRL3</i>	GATOR complex protein NPRL3	AD	~2% autosomal dominant focal epilepsies, with or without cortical dysplasia ¹²⁷
Generalized Epilepsy with Febrile Seizures Plus (GEFS+)	<i>SCN1A</i>	Sodium channel protein type 1 alpha	AD	5-10% GEFS+ ⁶
	<i>SCN1B</i>	Sodium channel subunit beta-1	AD	<5% GEFS+ ⁶
	<i>GABRG2</i>	Gamma-aminobutyric acid receptor subunit gamma-2	AD	<1% GEFS+ ⁶
	<i>SCN2A</i>	Sodium channel protein type 2 alpha	AD	Rare ⁵
Epilepsy with paroxysmal dyskinesia	<i>PRRT2</i>	Proline-rich transmembrane protein 2	AD	62-96% of familial PKD/infantile convulsions (PKD/IC); 36% sporadic PKD/IC ^{14,17,18}
	<i>KCNMA1</i>	Potassium calcium-activated channel subfamily M alpha 1	AD/AR	Rare ¹⁴⁰
Progressive Myoclonic Epilepsy	<i>EPM2A</i>	Laforin	AR	53% Lafora disease ⁴⁸
	<i>NHLRC1 (EPM2B)</i>	NHL repeat-containing protein 1 (malin)	AR	40% Lafora disease ⁴⁸
	<i>CSTB* (EPM1)</i>	Cystatin-B	AR	Rare sequencing variants in Unverricht-Lundborg disease ⁴⁹
	<i>KCTD7 (EPM3)</i>	BTB/POZ domain-containing protein KCTD7	AR	Rare ⁵⁰
	<i>SCARB2 (EPM4)</i>	Lysosome membrane protein 2	AR	~7% progressive myoclonic epilepsy ⁵¹ ; Unknown in action myoclonus-renal failure syndrome ⁵²
	<i>GOSR2</i>	Golgi SNAP receptor complex member 2	AR	Rare ^{53,54}
	<i>KCNC1</i>	Potassium voltage-gated channel subfamily C member 1	AD	Rare KCNC1-associated myoclonus epilepsy and ataxia (MEAK) ¹¹⁵
	<i>FOLR1</i>	Folate receptor alpha	AR	Rare ⁵⁵
Rett/atypical Rett syndromes	<i>MECP2</i>	Methyl CpG binding protein 2	XL	88% females with Rett syndrome ⁵⁷
	<i>CDKL5</i>	Cyclin-dependent kinase-like 5	XL	2-8% females with atypical Rett syndrome ^{58,59}
	<i>CTNNA1</i>	Catenin beta 1	AD	Rare in Rett-like syndromes ¹¹⁶
	<i>DDX3X</i>	DEAD-box helicase 3, X-linked	XL	Rare in Rett-like syndromes ¹¹⁷
	<i>FOXP1**</i>	Forkhead box protein G1	AD	~1% Rett syndrome overall ⁶⁰ ; 25% congenital variant of Rett ⁶¹
	<i>GABBR2</i>	Gamma-aminobutyric acid type B receptor subunit 2	AD	Rare in epileptic encephalopathy and intellectual disability with Rett-like features ¹¹⁸
	<i>KCNA2</i>	Potassium voltage-gated channel subfamily A member 2	AD	Rare in Rett-like syndromes ¹¹⁹
	<i>MBD5</i>	Methyl-CpG-binding domain protein 5	AD	Rare in Rett-like syndromes ⁶²
	<i>MEF2C</i>	Myocyte-specific enhancer factor 2C	AD	Rare in Rett-like syndromes ⁶³
	<i>SATB2</i>	SATB homeobox 2	AD	Rare in Rett-like syndromes ¹²⁰
	<i>STXBP1</i>	Syntaxin binding protein 1	AD	Rare in Rett-like syndromes ¹²¹
	<i>TBL1XR1</i>	Transducin (beta)-like 1 X-linked receptor 1	AD	Rare in Rett-like syndromes ¹²²
	<i>WDR45</i>	WD repeat domain 45	XL	Rare in Rett-like syndromes ^{106, 107}
	Angelman/ Angelman-like/ Pitt-Hopkins syndromes	<i>UBE3A</i>	Ubiquitin protein ligase E3A	AD
<i>SLC9A6</i>		Sodium/hydrogen exchanger 6	XL	6% Angelman-like syndrome ⁶⁵
<i>MBD5</i>		Methyl-CpG-binding domain protein 5	AD	Rare in Angelman-like syndrome ⁶⁶
<i>TCF4</i>		Transcription factor 4	AD	36% Pitt-Hopkins syndrome (PHS) ⁶⁷ ; 2% Angelman syndrome ⁶⁷
<i>ATRX</i>		Transcriptional regulator ATRX	XL	95% males with alpha-thalassemia X-linked intellectual disability syndrome ¹⁵³
<i>NRXN1</i>		Neurexin-1	AR	Rare in PHS ⁶⁸
Creatine deficiency syndromes	<i>CNTNAP2</i>	Contactin-associated protein-like 2	AR	Rare in PHS ⁶⁸
	<i>SLC6A8**</i>	Solute carrier family 6 (neurotransmitter transporter), member 8	XL	2% males with epilepsy and intellectual disability ⁴⁶ ; 65% males with biochemical creatine deficiency ⁴⁷
	<i>GAMT**</i>	Guanidinoacetate N-methyltransferase	AR	Rare ⁷¹

Neuronal Ceroid Lipofuscinoses (NCL)	<i>GATM</i>	Glycine amidinotransferase, mitochondrial	AR	Rare ⁷¹
	<i>PPT1 (CLN1)</i>	Palmitoyl-protein thioesterase 1	AR	98% PPT1 deficiency ⁷²
	<i>TPP1 (CLN2)</i>	Tripeptidyl-peptidase 1	AR	95% TPP1 deficiency ⁷³
	<i>CLN3</i>	Battenin	AR	92% Juvenile NCL ⁷⁴
	<i>DNAJC5 (CLN4B)</i>	DnaJ homolog subfamily C member 5	AD	25% Kufs disease ^{75,76}
	<i>CLN5</i>	Ceroid-lipofuscinosis neuronal protein 5	AR	94% Finnish late-infantile NCL ⁷⁷ ; Otherwise rare
	<i>CLN6</i>	Ceroid-lipofuscinosis neuronal protein 6	AR	Rare ⁷⁷
	<i>MFSD8 (CLN7)</i>	Major facilitator superfamily domain-containing protein 8	AR	Rare ⁷⁷
	<i>CLN8</i>	Ceroid-lipofuscinosis neuronal protein 8	AR	100% Finnish Northern epilepsy ⁷⁷ ; Otherwise rare
	<i>CTSD (CLN10)</i>	Cathepsin D	AR	Rare ⁷⁷
	<i>CTSF (CLN13)</i>	Cathepsin F	AR	Rare ⁷⁷
<i>KCTD7 (CLN14)</i>	BTB/POZ domain-containing protein KCTD7	AR	Rare ⁷⁸	
Metabolic disorders	<i>ADSL</i>	Adenylosuccinate lyase	AR	Rare (adenosuccinate lyase deficiency) ⁷⁹
	<i>ALDH5A1</i>	Aldehyde dehydrogenase 5 family member A1	AR	96% (SSADH deficiency) ¹²⁹
	<i>ARG1</i>	Arginase 1	AR	Rare (argininemia) ¹⁵⁵
	<i>ASNS</i>	Asparagine synthetase (glutamine-hydrolyzing)	AR	Rare (asparagine synthetase deficiency) ¹³⁰
	<i>FOLR1</i>	Folate receptor alpha	AR	Rare (cerebral folate deficiency) ⁵⁶
	<i>GLDC</i>	Glycine decarboxylase	AR	70-75% glycine encephalopathy (nonketotic hyperglycinemia) ¹²⁴
	<i>NGLY1</i>	N-glycanase 1	AR	Rare (congenital disorder of deglycosylation) ¹⁴³
	<i>PHGDH</i>	D-3-phosphoglycerate dehydrogenase	AR	Rare (phosphoglycerate dehydrogenase deficiency / Neu- Laxova syndrome) ¹⁶⁸
<i>SLC19A3</i>	Solute carrier family 19 member 3	AR	Rare (thiamine metabolism dysfunction disorders) ¹²⁵	
Brain malformations	<i>ARX</i>	Aristaless related homeobox	XL	70-95% of X-linked lissencephaly with ambiguous genitalia (XLAG) ¹⁵⁶
	<i>CASK</i>	Peripheral plasma membrane protein CASK	XL	~96% females with microcephaly with pontine and cerebellar hypoplasia (MICPCH) ¹⁵¹
	<i>CUL4B</i>	Cullin 4B	XL	Rare in cerebral malformations ¹⁶⁰
	<i>DCX</i>	Doublecortin	XL	Up to 100% XL lissencephaly; 10% of classic lissencephaly; 85% females and ~30% males with subcortical band heterotopia (SBH) ¹⁶¹⁻¹⁶³
	<i>FLNA</i>	Filamin A	XL	93% classic bilateral periventricular nodular heterotopia ¹³⁴
	<i>PAFAH1B1 (LIS1)</i>	Platelet-activating factor acetylhydrolase IB subunit alpha	AD	~40-65% of classic lissencephaly ¹⁶²
	<i>TUBB2A</i>	Tubulin, Beta-2A	AD	Rare ⁷⁰
EAST/SeSAME syndrome	<i>KCNJ10</i>	ATP-sensitive inward rectifier potassium channel 10	AR	Rare ^{81,82}
Epilepsy and hemiplegic migraine	<i>ATP1A2</i>	Sodium/potassium-transporting ATPase subunit alpha-2	AD	Unknown ⁶
Idiopathic generalized epilepsy	<i>CHRNA7 (dels only)</i>	Neuronal acetylcholine receptor subunit alpha-7	AD	~1% patients with IGE ⁵⁶
Epilepsy associated with other neuro-developmental disorders	<i>ANKRD11</i>	Ankyrin repeat domain-containing protein	AD	83% patients with KBG syndrome ¹⁵⁴
	<i>C12orf57</i>	Protein C10	AR	Rare (Temtamy syndrome or TEMTYS) ¹⁵⁷
	<i>DYRK1A</i>	Dual-specificity tyrosine-(Y)-phosphorylation regulated kinase 1A	AD	Rare ⁹³
	<i>EEF1A2</i>	Eukaryotic translation elongation factor 1 alpha 2	AD	Rare ⁹⁶
	<i>EHMT1</i>	Histone-lysine N-methyltransferase EHMT1	AD	~75% of patients with Kleefstra syndrome, including microdeletion of 9q34.3 ¹⁶⁵

<i>GRIN2A</i>	Glutamate receptor ionotropic, NMDA 2A	AD	9-20% atypical Rolandic epilepsy and epilepsy-aphasia ⁸⁹ ; ~2% patients with intellectual disability and epilepsy ^{83,84}
<i>GRIN2B</i>	Glutamate receptor ionotropic, NMDA 2B	AD	Unknown ⁸³
<i>KANSL1</i>	KAT8 regulatory NSL complex subunit 1	AD	Rare (Koolen-de Vries syndrome, including microdeletion of 17q21.31) ^{85,86}
<i>HNRNPU</i>	HNRNPU antisense RNA 1	AD	Rare ¹³⁷
<i>KCNH1</i>	Potassium voltage-gated channel subfamily H member 1	AD	Rare (Temple-Baraitser and Zimmermann-Laband syndromes) ¹³⁹
<i>KDM6A</i>	Lysine demethylase 6A	XL	Rare (Kabuki syndrome) ¹²⁸
<i>NEXMIF</i> (<i>KIAA2022</i>)	Neurite extension and migration factor	XL	Rare ¹⁴¹
<i>NALCN</i>	Sodium leak channel, non-selective	AD/ AR	Rare (CLIFAHDD and infantile hypotonia with psychomotor retardation and characteristic facies (IHPRF)) ¹⁴²
<i>PACS1</i>	Phosphofurin acidic cluster sorting protein 1	AD	Rare ¹⁴⁴
<i>PNKP</i>	Bifunctional polynucleotide phosphatase/kinase	AR	Rare ⁸⁶
<i>PPP2R5D</i>	protein phosphatase 2 regulatory subunit B', delta	AD	Rare ¹⁴⁶
<i>PURA</i>	Transcriptional activator protein Pur-alpha	AD	Rare (Including microdeletion of 5q31.3) ¹⁴⁷
<i>SHANK3</i>	SH3 and multiple ankyrin repeat domains protein 3	AD	Rare (Phelan-McDermid syndrome, including microdeletion of 22q13.3) ¹⁷¹
<i>SMARCA2</i>	Probable global transcription activator SNF2L2	AD	Rare (Nicolaidis-Baraitser and Coffin Siris syndromes) ¹⁷²

* The dodecamer repeat expansion that accounts for ~90% of all CSTB mutations is not be detectable by this test

** Does not include deletion/duplication testing of DNM1, FOXG1, and SLC6A8; duplications of GATM may not be detectable

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Appendix: Genes Included on Comprehensive Epilepsy Panel and Subpanels

Gene	Comprehensive	Infantile	Childhood	STAT	Progressive	Rett/Angelman
ADSL	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
ALDH5A1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
ALDH7A1	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>		
ALG13	<input type="checkbox"/>	<input type="checkbox"/>				
ANKRD11	<input type="checkbox"/>					
ARG1	<input type="checkbox"/>		<input type="checkbox"/>			
ARHGEF9	<input type="checkbox"/>	<input type="checkbox"/>				
ARX	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>		
ASNS	<input type="checkbox"/>	<input type="checkbox"/>				
ATP1A2	<input type="checkbox"/>					
ATP1A3	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
ATP6AP2	<input type="checkbox"/>	<input type="checkbox"/>				
ATRX	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			<input type="checkbox"/>
BRAT1	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>		
C12orf57	<input type="checkbox"/>	<input type="checkbox"/>				
CACNA1A	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
CACNA1E	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>		
CACNA1G	<input type="checkbox"/>	<input type="checkbox"/>				
CASK	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
CDKL5	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>
CHD2	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
CHRNA2	<input type="checkbox"/>		<input type="checkbox"/>			
CHRNA4	<input type="checkbox"/>		<input type="checkbox"/>			
CHRNA7**	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
CHRN2	<input type="checkbox"/>		<input type="checkbox"/>			
CLCN4	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
CLN3	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>	
CLN5	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>	
CLN6	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>	
CLN8	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>	
CNTNAP2	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			<input type="checkbox"/>
CSTB***	<input type="checkbox"/>		<input type="checkbox"/>		<input type="checkbox"/>	
CTNNB1	<input type="checkbox"/>					<input type="checkbox"/>
CTSD (CLN10)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>	
CTSF	<input type="checkbox"/>				<input type="checkbox"/>	
CUL4B	<input type="checkbox"/>	<input type="checkbox"/>				
DCX	<input type="checkbox"/>	<input type="checkbox"/>				
DDX3X	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			<input type="checkbox"/>
DEPDC5	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
DNAJC5 (CLN4B)	<input type="checkbox"/>				<input type="checkbox"/>	
DNM1*	<input type="checkbox"/>	<input type="checkbox"/>				
DOCK7	<input type="checkbox"/>	<input type="checkbox"/>				
DYRK1A	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			<input type="checkbox"/>
EEF1A2	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
EHMT1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			<input type="checkbox"/>
EPM2A	<input type="checkbox"/>		<input type="checkbox"/>		<input type="checkbox"/>	
FGF12	<input type="checkbox"/>	<input type="checkbox"/>				
FLNA	<input type="checkbox"/>					
FOLR1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
FOXP1*	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			<input type="checkbox"/>
FRRS1L	<input type="checkbox"/>	<input type="checkbox"/>				
GABBR2	<input type="checkbox"/>	<input type="checkbox"/>				<input type="checkbox"/>

GABRA1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
GABRB2	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
GABRB3	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
GABRG2	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
GAMT*	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
GATM	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
GLDC	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>		
GNAO1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
GOSR2	<input type="checkbox"/>		<input type="checkbox"/>			
GRIN1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>	
GRIN2A	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
GRIN2B	<input type="checkbox"/>	<input type="checkbox"/>				
HCN1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
HNRNPU	<input type="checkbox"/>	<input type="checkbox"/>				
IQSEC2	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			<input type="checkbox"/>
KANSL1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
KCNA2	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			<input type="checkbox"/>
KCNB1	<input type="checkbox"/>	<input type="checkbox"/>				
KCNC1	<input type="checkbox"/>		<input type="checkbox"/>		<input type="checkbox"/>	
KCNH1	<input type="checkbox"/>	<input type="checkbox"/>				
KCNJ10	<input type="checkbox"/>	<input type="checkbox"/>				
KCNMA1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
KCNQ2	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>		
KCNQ3	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>		
KCNT1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>		
KCTD7 (CLN14)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>	
KDM6A	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
LGI1	<input type="checkbox"/>		<input type="checkbox"/>			
MAGI2**	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
MBD5	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			<input type="checkbox"/>
MECP2	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>
MEF2C	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>
MFSD8 (CLN7)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>	
NALCN	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
NEXMIF (KIAA2022)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
NGLY1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
NHLRC1 (EPM2B)	<input type="checkbox"/>		<input type="checkbox"/>		<input type="checkbox"/>	
NPRL3	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
NR2F1	<input type="checkbox"/>	<input type="checkbox"/>				
NRXN1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			<input type="checkbox"/>
PACS1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
PAFAH1B1 (LIS1)	<input type="checkbox"/>	<input type="checkbox"/>				
PCDH19	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>
PHGDH	<input type="checkbox"/>		<input type="checkbox"/>			
PIGA	<input type="checkbox"/>	<input type="checkbox"/>				
PIGG	<input type="checkbox"/>	<input type="checkbox"/>				
PIGN	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
PIGO	<input type="checkbox"/>	<input type="checkbox"/>				
PIGT	<input type="checkbox"/>	<input type="checkbox"/>				
PIGV	<input type="checkbox"/>	<input type="checkbox"/>				
PLCB1	<input type="checkbox"/>	<input type="checkbox"/>				
PNKP	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			
PNPO	<input type="checkbox"/>	<input type="checkbox"/>		<input type="checkbox"/>		
POLG	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>		
PPP2R5D	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>			

PPT1 (CLN1)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
PRRT2	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
PURA	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
QARS	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SATB2	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SCARB2	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SCN1A	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SCN1B	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SCN2A	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SCN8A	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SHANK3	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SLC13A5	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SLC19A3	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SLC25A22	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SLC2A1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SLC35A2	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SLC6A1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SLC6A8*	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SLC9A6	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SMARCA2	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SMC1A	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SNAP25	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SPATA5	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SPTAN1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
STX1B	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
STXBP1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SYNGAP1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
SZT2	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
TBC1D24	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
TBL1XR1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
TCF4	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
TPP1 (CLN2)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
TSC1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
TSC2	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
TUBB2A	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
UBE3A	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
WDR45	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
WWOX	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
ZEB2	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

* Sequencing only (no deletion/duplication analysis) for DNMI1, FOXG1, or SLC6A8. This test can detect only complete gene deletions/duplications of GAMT. No sequence coverage of exon 22 of DNMI1 (NM_004408.3).

** Deletion/duplication testing only (no sequence analysis) for CHRNA7 and MAGI2.

*** The dodecamer repeat expansion that accounts for ~90% of all CSTB mutations is not be detectable by this test.