Comprehensive Epilepsy

Precision Panel

Overview

Epilepsy is a central nervous system disease characterized by recurrent unprovoked seizures, which are brief episodes of involuntary movement that may involve a part of the body (partial) or the entire body (generalized) and can be accompanied by loss of consciousness and loss of control of bowel or bladder function. Around 50 million people worldwide have epilepsy, making it one of the most common neurological diseases globally. Epilepsy entails an enduring predisposition to generate neurobiological, cognitive, psychological and social consequences. Multiple risk factors exist for epilepsy one of them being a strong genetic predisposition. The three major classes of epilepsy disorders are genetic generalized, focal and encephalopathic epilepsies, with several specific disorders within each class. Epilepsy genetics is shifting from an academic pursuit to a clinical discipline based on molecular diagnosis and stratified medicine. Mutations leading to epilepsy have been identified in genes encoding ion channels, neurotransmitter receptors, molecular cascade of cellular energy production and proteins involved in neuronal excitability. The mode of inheritance ranges from autosomal dominant, recessive all the way to mitochondrial.

The Igenomix Comprehensive Epilepsy Precision Panel can serve as an accurate and directed diagnostic tool as well as for a differential diagnosis of recurrent seizures ultimately leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes involved.

Indications

The Igenomix Comprehensive Epilepsy Precision Panel is indicated in patients with a clinical suspicion or diagnosis presenting with the following manifestations:

- Family history of epilepsy or treatment-resistant seizures
- Loss of consciousness or awareness
- Disturbances of movement
- Vision, hearing and taste disturbances
- Temporary confusion
- Uncontrollable jerking movements of the arms and legs
- Fear, anxiety or deja vu

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular diagnosis for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment with a multidisciplinary team in the form of medical with antiepileptic medication and monitoring of side effects, epilepsy surgery if indicated and dietary modifications.
- Establish recurrence risk depending on the type of epilepsy, genetic background and clinical presentation.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
AARS1	Charcot-Marie-Tooth Disease, Epileptic Encephalopathy	AD,AR	99.07	30 of 30
ABAT	Gaba-Transaminase Deficiency	AR	100	9 of 9
ABCA2	Intellectual Developmental Disorder, Seizures, Ataxia	AR	99.05	11 of 11
ABCC8	Diabetes Mellitus, Hyperinsulinemic Hypoglycemia, Dend Syndrome	AD,AR	99.98	710 of 712
ABCD1	Adrenoleukodystrophy	X,XR,G	100	-
ACTL6B	Epileptic Encephalopathy, Intellectual Developmental Disorder	AD,AR	100	21 of 21
ACY1	Aminoacylase 1 Deficiency	AR	100	15 of 15
ADAM22	Epileptic Encephalopathy	AR	99.98	4 of 4
ADAR	Aicardi-Goutieres Syndrome, Dyschromatosis Symmetrica, Bilateral Striatal Necrosis	AD,AR	99.93	252 of 252
ADGRG1	Polymicrogyria	AR	100	-
ADGRV1	Febrile Convulsions, Usher Syndrome, Epilepsy	AD,AR	97.53	-
ADPRS	Neurodegeneration, Ataxia	AR	99.86	11 of 11
ADRA2B	Epilepsy	-	100	5 of 5
ADSL	Adenylosuccinate Lyase Deficiency	AR	100	59 of 59
AFG3L2	Optic Atrophy, Ataxia, Epilepsy	AD,AR	99.74	42 of 42
AGA	Aspartylglucosaminuria	AR	100	35 of 35
AHI1	Joubert Syndrome, Retinitis Pigmentosa	AR	96.79	85 of 97
AIFM1	Oxidative Phosphorylation Deficiency, Cowchock Syndrome, Deafness, Spondyloepimetaphyseal Dysplasia, Leukoencephalopathy, Mitochondrial Encephalomyopathy, Charcot-Marie-Tooth Disease	X,XR,G	100	-
AIMP1	Leukodystrophy, Intellectual Disability	AR	100	10 of 10
AKT3	Megalencephaly-Polymicrogyria-Postaxial Polydactyly-Hydrocephalus Syndrome	AD	99.9	9 of 11
ALDH3A2	Sjogren-Larsson Syndrome	AR	96	119 of 119
ALDH4A1	Hyperprolinemia	AR	100	7 of 7
ALDH5A1	Succinic Semialdehyde Dehydrogenase Deficiency	AR	95.41	65 of 69
ALDH7A1	Epilepsy	AR	99.98	131 of 134
ALG1	Congenital Disorder Of Glycosylation	AR	100	46 of 46
ALG12	Congenital Disorder Of Glycosylation	AR	100	17 of 17
ALG13	Epileptic Encephalopathy, Intellectual Disability	X,XR,XD,G	99.62	-
ALG2	Congenital Disorder Of Glycosylation, Myasthenic Syndrome	AR	99.61	7 of 7
ALG3	Congenital Disorder Of Glycosylation	AR	99.2	25 of 25
ALG6	Congenital Disorder Of Glycosylation	AR	99.91	24 of 24
ALG8	Congenital Disorder Of Glycosylation, Polycystic Liver Disease	AD,AR	99.5	22 of 22
ALG9	Congenital Disorder Of Glycosylation, Polycystic Kidney Disease, Microbrachycephaly, Hypertelorism	AR	99.99	6 of 6

ALKBH8	Intellectual Developmental Disorder	AR	99.2	2 of 2
AMACR	Alpha-Methylacyl-Coa Racemase Deficiency	AR	100	8 of 8
AMT	Glycine Encephalopathy	AR	99.98	94 of 96
ANK3	Mental Retardation	AR	99.76	22 of 23
ANKRD11	Kbg Syndrome, 16q24.3 Microdeletion Syndrome	AD	99.6	119 of 124
AP2M1	Intellectual Developmental Disorder, Myoclonic-Astastic Epilepsy	AD	100	1 of 1
AP3B2	Epileptic Encephalopathy	AR	99.95	11 of 12
AP4B1	Spastic Paraplegia, Intellectual Disability	AR	99.64	22 of 22
AP4E1	Spastic Paraplegia, Stuttering, Severe Intellectual Disability	AD,AR	99.94	17 of 17
AP4M1	Spastic Paraplegia, Severe Intellectual Disability	AR	100	18 of 18
AP4S1	Spastic Paraplegia, Severe Intellectual Disability	AR	99.95	8 of 8
ARFGEF2	Microcephaly, Periventricular Nodular Heterotopia	AR	100	15 of 15
ARG1	Argininemia	AR	100	66 of 68
ARHGEF15	Angelman Syndrome, Epileptic Encephalopathy, Spastic Ataxia	-	99.89	3 of 3
ARHGEF9	Hyperekplexia, Epilepsy	X,XR,G	100	-
ARID1B	Coffin-Siris Syndrome, 6q25 Microdeletion Syndrome	AD	93.87	226 of 238
ARL13B	Joubert Syndrome	AR	99.77	10 of 10
ARSA	Metachromatic Leukodystrophy	AR	98	266 of 266
ARSB	Mucopolysaccharidosis	AR	99.83	217 of 220
ARV1	Epileptic Encephalopathy	AR	100	3 of 3
ARX	Corpus Callosum, Epileptic Encephalopathy, Lissencephaly, Mental Retardation, Partington Syndrome, West Syndrome	X,XR,G	81.92	-
ASAH1	Farber Lipogranulomatosis, Spinal Muscular Atrophy, Myoclonic Epilepsy	AR	99.98	69 of 70
ASNS	Asparagine Synthetase Deficiency	AR	99.98	37 of 37
ASPA	Canavan Disease	AR	99.56	93 of 94
ASPM	Microcephaly	AR	99.74	221 of 222
ASXL3	Bainbridge-Ropers Syndrome, Feeding Difficulties, Failure To Thrive, Microcephaly	AD	95.96	77 of 81
ATAD1	Hyperekplexia	AR	99.97	3 of 3
ATIC	Imp Cyclohydrolase, Charcot Marie Tooth Disease	AR	98.77	8 of 8
ATN1	Congenital Hypotonia, Chorea, Seizures, Dementia, Dentatorubral Pallidoluysian Atrophy	AD	99.86	11 of 11
ATP13A2	Kufor-Rakeb Syndrome, Spastic Paraplegia, Ceroid Lipofuscinosis	AR	99.97	53 of 53
ATP1A2	Alternating Hemiplegia Of Childhood, Migraine	AD	100	108 of 108
ATP1A3	Alternating Hemiplegia Of Childhood, Cerebellar Ataxia, Optic Atrophy, Sensorineuralhearing Loss, Dystonia, Areflexia, Pes Cavus, Parkinsonism	AD	99.94	138 of 138
ATP2A2	Acrokeratosis Verruciformis, Darier-White Disease	AD	100	298 of 301
ATP6AP2	Congenital Disorder Of Glycosylation, Mental Retardation, Epilepsy, Parkinsonism	X,XR,G	100	-
ATP6V0A2	Cutis Laxa, Wrinkly Skin Syndrome	AR	99.99	55 of 55
ATP6V1A	Cutis Laxa, Epileptic Encephalopathy	AD,AR	99.98	9 of 9
ATP7A	Cutis Laxa, Menkes Disease, Spinal Muscular Atrophy, Occipital Horn Syndrome	X,XR,G	99.83	-
ATPAF2	Atpase Deficiency	AR	100	2 of 2
ATRX	Alpha-Thalassemia Myelodysplasia Syndrome, Mental Retardation-Hypotonic Facies Syndrome, Carpenter-Waziri Syndrome, Chudley-Lowry-Hoar Syndrome, Holmes-Gang Syndrome, Juberg-Marsidi Syndrome, Smith-Fineman-Myers Syndrome	X,XR,XD,G	98.5	-
AUH	3-Methylglutaconic Aciduria	AR	99.99	11 of 11
B4GALT1	Congenital Disorder Of Glycosylation	AR	99.97	3 of 3
BCKDK	Branched-Chain Ketoacid Dehydrogenase Kinase Deficiency	-	99.91	6 of 6
BCS1L	Bjornstad Syndrome, Gracile Syndrome, Leigh Syndrome, Mitochondrial Complex III Deficiency	AR,MI	99.96	40 of 42
BOLA3	Mitochondrial Dysfunctions Syndrome	AR	100	8 of 8
BRAF	Cardiofaciocutaneous Syndrome, Leopard Syndrome, Lung Cancer, Craniopharyngioma, Noonan Syndrome	AD	100	80 of 80
BRAT1	Neurodevelopmental Disorder, Cerebellar Atrophy, Rigidity And Multifocal Seizure Syndrome	AR	99.95	29 of 29
BRD2	Photosensitive Epilepsy	-	92.11	1 of 1

BTD	Biotinidase Deficiencymultiple Carboxylase Deficiency	AR	100	261 of 262
BUB1B	Colorectal Cancer, Mosaic Variegated Aneuploidy Syndrome	AD,AR	99.84	30 of 31
C12ORF57	Craniofacial Dysmorphism, Ocular Coloboma, Temtamy Syndrome	AR	-	-
CACNA1A	Epileptic Encephalopathy, Ataxia, Migraine, Benign Paroxysmal Torticollis Of Infancy	AD	96.13	249 of 266
CACNA1B	Neurodevelopmental Disorder, Seizures, Hyperkinetic Movements, Epileptic Encephalopathy	AR	95.83	7 of 7
CACNA1D	Primary Aldosteronism, Seizures, Neurologic Abnormalities, Sinoatrial Node Dysfunction, Deafness	AD,AR	100	18 of 18
CACNA1E	Epileptic Encephalopathy	AD	99.94	25 of 25
CACNA1H	Hyperaldosteronism, Epilepsy	AD	98.05	71 of 71
CACNA2D2	Cerebellar Atrophy, Seizures, Developmental Delay	AR	94	10 of 10
CACNB4	Epilepsy, Ataxia	AD	99.87	5 of 5
CAD	Epileptic Encephalopathy	AR	100	12 of 12
CARS2	Oxidative Phosphorylation Deficiency	AR	99.14	6 of 6
CASK	Anemia, Fg Syndrome, Mental Retardation, Microcephaly, Pontine And Cerebellar Hypoplasia, Epileptic Encephalopathy	X,XR,XD,G	99.98	-
CASR	Hyperparathyroidism, Hypocalcemia, Pancreatitis	AD,AR	100	445 of 446
CBL	Myelomonocytic Leukemia, Noonan Syndrome, Mastocytosis, Noonan Syndrome	AD	100	46 of 47
CC2D1A	Mental Retardation	AR	100	7 of 7
CC2D2A	Coach Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.43	98 of 100
CCDC88C	Hydrocephalus, Spinocerebellar Ataxia	AD,AR	99.44	13 of 14
CCL2	Neural Tube Defects	AD	100	-
CDK9	Inmune Deficiency Disease, Myeloma	-	82.69	2 of 2
CDKL5	Epileptic Encephalopathy, Atypical Rett Syndrome, West Syndrome	X,XD,G	99.92	-
CENPJ	Microcephaly, Seckel Syndrome	AR	99.97	13 of 13
CEP290	Bardet-Biedl Syndrome, Joubert Syndrome, Leber Congenital Amaurosis, Meckel Syndrome, Senior-Loken Syndrome	AR	96.47	293 of 327
CERS1	Epilepsy	AR	72.1	2 of 2
CERT1	Mental Retardation	AD	99.98	8 of 8
CHD2	Epileptic Encephalopathy, Lennox-Gastaut Syndrome	AD	98.91	103 of 103
CHRNA2	Epilepsy	AD	99.91	8 of 8
CHRNA4	Epilepsy	AD	99.8	24 of 24
CHRNB2	Epilepsy	AD	100	13 of 13
CILK1	Endocrine-Cerebroosteodysplasia, Epilepsy	AD,AR	100	-
CLCN2	Epilepsy, Hyperaldosteronism, Leukoencephalopathy	AD,AR	100	39 of 39
CLCN4	Mental Retardation	X,XR,XD,G	99.69	-
CLN3	Ceroid Lipofuscinosis	AR	99.93	73 of 75
CLN5	Ceroid Lipofuscinosis	AR	99.56	52 of 55
CLN6	Ceroid Lipofuscinosis	AR	99.94	98 of 99
CLN8	Ceroid Lipofuscinosis, Intellectual Disability	AR	100	44 of 45
CLTC	Mental Retardation, Epileptic Encephalopathy	AD	98.81	14 of 14
CNKSR2	Mental Retardation, Epileptic Encephalopathy	X,G	99.11	-
CNPY3	Epileptic Encephalopathy, West Syndrome	AR	100	5 of 5
CNTN2	Epilepsy	AR	99.98	6 of 6
CNTNAP2	Pitt-Hopkins-Like Syndrome	AR	99.91	39 of 41
COA7	Spinocerebellar Ataxia	AR	99.99	6 of 6
COA8	Mitochondrial Complex Iv Deficiency, Leukoencephalopathy	AR,MI	86.26	4 of 5
COG7	Congenital Disorder Of Glycosylation	AR	99.94	6 of 6
COG8	Congenital Disorder Of Glycosylation	AR	100	8 of 8
COL18A1	Glaucoma, Knobloch Syndrome	AD,AR	99.76	-
COL4A1	Angiopathy, Microangiopathy, Leukoencephalopathy, Porencephaly, Retinal Arteries, Hanac Syndrome, Walker-Warburg Syndrome	AD	99.99	173 of 173
COQ2	Coenzyme Q10 Deficiency, Multiple System Atrophy, Leigh Syndrome, Nephrotic Syndrome	AD,AR	99.61	37 of 38
COQ4	Coenzyme Q10 Deficiency	AR	91.05	21 of 21
COQ8A	Coenzyme Q10 Deficiency, Ataxia	AR	100	-

COQ9	Coenzyme Q10 Deficiency	AR	99.87	6 of 6
COX10	Leigh Syndrome, Mitochondrial Complex Iv Deficiency	AR,MI	100	13 of 13
COX15	Cardioencephalomyopathy, Leigh Syndrome, Leukodystrophy	AR,MI	100	5 of 5
COX6B1	Mitochondrial Complex Iv Deficiency	AR,MI	100	3 of 3
CPA6	Epilepsy, Febrile Seizures	AD,AR	99.97	9 of 9
CPLX1	Epileptic Encephalopathy, Wolf-Hirschhorn Syndrome	AD,AR	99.81	3 of 3
CPT2	Carnitine Palmitoyltransferase Ii Deficiency, Encephalopathy	AD,AR	99.99	116 of 116
CRH	Epilepsy, Conn Syndrome, Depression	-	99.84	1 of 2
CSF1R	Brain Abnormalities, Gliosis	AD,AR	100	122 of 124
CSNK2B	Poirier-Bienvenu Neurodevelopmental Syndrome	AD	99.98	14 of 17
CSTB	Epilepsy, Hypohidrotic Ectodermal Dysplasia, Unverricht-Lundborg Disease	AR	100	14 of 14
CTC1	Cerebroretinal Microangiopathy, Dyskeratosis Congenita	AR	99.73	43 of 44
CTNND2	Benign Adult Familial Myoclonic Epilepsy	-	94.3	10 of 12
CTSA	Neuraminidase Deficiency, Galactosialidosis	AR	100	40 of 40
CTSD	Ceroid Lipofuscinosis	AR	100	18 of 18
CTSF	Ceroid Lipofuscinosis	AR	92.18	12 of 12
CUL4B	Mental Retardation, Short Stature, Musclewasting	X,XR,G	99.77	-
CUX2	Epileptic Encephalopathy, Lennox-Gastaut Syndrome	AD	99.72	2 of 2
CYFIP2	Epileptic Encephalopathy	AD	100	8 of 8
CYP27A1	Cerebrotendinous Xanthomatosis	AR	100	118 of 118
D2HGDH	D-2-Hydroxyglutaric Aciduria	AR	100	42 of 42
DARS1	Hypomyelination	AR	99.99	18 of 18
DARS2	Leukoencephalopathy	AR	100	65 of 65
DCX	Lissencephaly	X,G	100	-
DDC	Amino Acid Decarboxylase Deficiency	AR	100	59 of 59
DDX3X	Intellectual Developmental Disorder	X,XR,XD,G	99.03	-
DEAF1	Dyskinesia, Seizures, Intellectual Developmental Disorder, Smith-Magenis Syndrome	AD,AR	93.55	42 of 42
DEGS1	Leukodystrophy	AR	86.16	12 of 14
DENND5A	Epileptic Encephalopathy	AR	100	9 of 9
DEPDC5	Epilepsy	AD	100	127 of 127
DHCR7	Smith-Lemli-Opitz Syndrome	AR	100	217 of 217
DHDDS	Developmental Delay, Seizures, Retinitis Pigmentosa, Epileptic Encephalopathy	AD,AR	96.32	8 of 8
DHFR	Megaloblastic Anemia, Dihydrofolate Reductase Deficiency	AR	99.7	4 of 4
DHPS	Neurodevelopmental Disorder, Seizures	AR	99.85	4 of 4
DIAPH1	Deafness, Seizures	AD,AR	99.94	15 of 15
DLD	Pyruvate Dehydrogenase Deficiency	AR	100	26 of 26
DNAJC5	Ceroid Lipofuscinosis	AD	100	2 of 2
DNM1	Epileptic Encephalopathy, Lennox-Gastaut Syndrome	AD	94.8	30 of 30
DNM1L	Encephalopathy, Optic Atrophy	AD,AR	100	29 of 29
DOCK7	Epileptic Encephalopathy, Cortical Blindness	AR	99.95	11 of 11
DOLK	Congenital Disorder Of Glycosylation, Dilated Cardiomyopathy	AR	99.98	13 of 13
DPAGT1	Congenital Disorder Of Glycosylation, Myasthenic Syndrome	AR	100	41 of 41
DPM1	Congenital Disorder Of Glycosylation	AR	97.25	9 of 9
DPM2	Congenital Disorder Of Glycosylation, Muscular Dystrophy, Intellectual Disability, Epilepsy	AR	99.87	2 of 2
DPYD	Dihydropyrimidine Dehydrogenase Deficiency, 1p21.3 Microdeletion Syndrome	AR	100	74 of 75
DPYS	Dihydropyrimidinuria	AR	100	31 of 31
DYNC1H1	Charcot-Marie-Tooth Disease, Mental Retardation, Spinal Muscular Atrophy	AD	100	104 of 104
DYRK1A	Mental Retardation	AD	99.85	78 of 81
EARS2	Oxidative Phosphorylation Deficiency	AR	98.8	31 of 31
ECHS1	Mitochondrial Short-Chain Enoyl-Coa Hydratase 1 Deficiency, Leigh Syndrome, Leukodystrophy	AR	100	39 of 39
ECM1	Lipoid Proteinosis	AR	99.99	64 of 64
EEF1A2	Epileptic Encephalopathy, Mental Retardation	AD	100	14 of 14
EFHC1	Epilepsy	AD	100	38 of 39

EHMT1	Kleefstra Syndrome	AD	98.58	58 of 75
EIF2B1	Leukoencephalopathy, Vanishing White Matter	AR	100	9 of 9
EIF2B2	Leukoencephalopathy, Vanishing White Matter	AR	100	30 of 30
EIF2B3	Leukoencephalopathy, Vanishing White Matter	AR	97.55	26 of 26
EIF2B4	Leukoencephalopathy, Vanishing White Matter	AR	100	31 of 31
EIF2B5	Leukoencephalopathy, Vanishing White Matter	AR	100	99 of 99
EIF3F	Intellectual Developmental Disorder	AR	99.99	1 of 1
EMX2	Schizencephaly	-	100	5 of 5
EPM2A	Lafora Disease	AR	89.2	63 of 70
EPRS1	Leukodystrophy	AR	99.62	6 of 6
ETFA	Acyl-Coa Dehydrogenase Deficiency	AR	92.33	32 of 32
ETFB	Acyl-Coa Dehydrogenase Deficiency	AR	100	21 of 21
ETFDH	Acyl-Coa Dehydrogenase Deficiency	AR	100	221 of 222
ETHE1	Encephalopathy	AR	100	32 of 33
FA2H	Spastic Paraplegia, Fatty Acid Hydroxylase-Associated Neurodegeneration	AR	88.77	60 of 62
FAM126A	Hypomyelination, Congenital Cataract	AR	100	11 of 12
FAR1	Fatty Acyl-Coa Reductase 1 Deficiency	AR	98.77	4 of 4
FARS2	Oxidative Phosphorylation Deficiency, Spastic Paraplegia	AR	99.98	23 of 23
FASN	Fatty Liver Disease	-	100	6 of 6
FCSK	Congenital Disorder Of Glycosylation	AR	97.99	-
FDFT1	Squalene Synthase Deficiency	AR	99.77	3 of 4
FDX2	Mitochondrial Myopathy, Leukoencephalopathy	AR,MI	100	-
FGD1	Aarskog-Scott Syndrome	X,XR,G	98.95	-
FGF12	Epileptic Encephalopathy	AD	99.98	4 of 6
FGFR3	Achondroplasia, Hypochondroplasia, Camptodactyly, Crouzon Syndrome, Epidermal Nevus, Lacrimoauriculodentodigital Syndrome, Muenke Syndrome, Thanatophoric Dysplasia, Isolated Brachycephaly, Isolated Plagiocephaly, Saethre-Chotzen Syndrome	AD,AR	99.89	77 of 78
FH	Fumarase Deficiency, Leiomyomatosis	AD,AR	100	229 of 232
FKRP	Dystroglycanopathy, Limb-Girdle Muscular Dystrophy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	99.9	157 of 157
FKTN	Cardiomyopathy, Dystroglycanopathy, Limb-Girdle Muscular Dystrophy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	98	54 of 56
FLNA	Cardiac Valvular Dysplasia, Fg Syndrome, Frontometaphyseal Dysplasia, Heterotopia, Intestinal Pseudoobstruction, Melnick-Needles Syndrome, Otopalatodigital Syndrome, Terminal Osseous Dysplasia, Short Bowel Syndrome, Ehlers-Danlos Syndrome	X,XR,XD,G	100	-
FOLR1	Neurodegeneration	AR	100	19 of 23
FOXG1	Rett Syndrome, 14q12 Microdeletion Syndrome	AD	88.71	93 of 109
FOXRED1	Mitochondrial Complex I Deficiency, Leigh Syndrome, Leukodystrophy	AR	100	13 of 13
FRRS1L	Epileptic Encephalopathy, Intellectual Disability	AR	85.58	7 of 7
FUCA1	Fucosidosis	AR	100	31 of 32
FUT8	Congenital Disorder Of Glycosylation, Fucosylation	AR	99.73	4 of 4
GABBR2	Epileptic Encephalopathy, Neurodevelopmental Disorder, Rett Syndrome	AD	95.98	7 of 7
GABRA1	Epileptic Encephalopathy, Dravet Syndrome	AD	100	45 of 46
GABRA2	Alcohol Dependence, Epileptic Encephalopathy	AD,MU	99.08	3 of 3
GABRA3	Thyrotoxic Periodic Paralysis	-	99.91	-
GABRA5	Epileptic Encephalopathy	AD	99.94	9 of 9
GABRB1	Epileptic Encephalopathy	AD	99.98	9 of 9
GABRB2	Epileptic Encephalopathy	AD	99.19	16 of 19
GABRB3	Epileptic Encephalopathy, Lennox-Gastaut Syndrome	AD	100	54 of 62
GABRD	Epilepsy, 1p36 Deletion Syndrome	AD	95.23	3 of 3
GABRG2	Epileptic Encephalopathy, Dravet Syndrome	AD	99.67	53 of 53
GAL	Epilepsy	AD	100	1 of 1
GALC	Krabbe Disease	AR	99.38	252 of 254

GAMT	Cerebral Creatine Deficiency Syndrome, Guanidinoacetate Methyltransferase Deficiency	AR	99.92	60 of 60
GATM	Cerebral Creatine Deficiency Syndrome, Fanconi Renotubular Syndrome	AD,AR	99.98	21 of 21
GCDH	Glutaric Acidemia, Glutaryl-Coa Dehydrogenase Deficiency	AR	88.74	254 of 254
GCH1	Dystonia, Gtp Cyclohydrolase I Deficiency	AD,AR	99.41	225 of 244
GCSH	Glycine Encephalopathy	AR	93.52	1 of 1
GFAP	Alexander Disease	AD	99.98	143 of 143
GFM1	Combined Oxidative Phosphorylation Deficiency	AR	100	27 of 27
GFM2	Combined Oxidative Phosphorylation Deficiency	AR	99.35	5 of 7
GJC2	Leukodystrophy, Lymphedema, Spastic Paraplegia, Milroy Disease	AD,AR	95.37	52 of 63
GLB1	Gangliosidosis, Morquio Syndrome	AR	100	242 of 243
GLDC	Glycine Encephalopathy	AR	98.69	359 of 367
GLI2	Holoprosencephaly, Pallister-Hall Syndrome, Pituitary Hormone Deficiencies	AD	98.38	83 of 88
GLI3	Greig Cephalopolysyndactyly Syndrome, Hypothalamic Hamartomascongenital Hypothalamic Hamartoma Syndrome, Pallister-Hall Syndrome, Acrocallosal Syndrome, Tibial Hemimelia	AD,AR	100	231 of 231
GLRA1	Hyperekplexia	AD,AR	99.6	71 of 72
GLRB	Hyperekplexia	AR	99.3	16 of 18
GLS	Epileptic Encephalopathy, Global Developmental Delay, Infantile Cataract	AD,AR	97.77	8 of 9
GLUD1	Hyperinsulinemic Hypoglycemia, Hyperinsulinism-Hyperammonemia Syndrome	AD	99.98	39 of 39
GNAO1	Epileptic Encephalopathy, Neurodevelopmental Disorder	AD	100	47 of 47
GNB1	Leukemia, Mental Retardation, Global Developmental Delay	AD,MU,P	100	31 of 31
GNE	Nonaka Myopathy, Sialuria	AD,AR	99.97	248 of 253
GNS	Mucopolysaccharidosis	AR	99.92	22 of 22
GOLGA2	Vohwinkel Syndrome, Smith-Mccort Dysplasia	-	99.89	3 of 3
GOSR2	Epilepsy	AR	88.39	6 of 6
GPAA1	Glycosylphosphatidylinositol Biosynthesis Defect, Neurodevelopmental Delay	AR	99.98	11 of 11
GPC3	Simpson-Golabi-Behmel Syndrome, Wilms Tumor, Nephroblastoma	AD,X,XR,G	99.84	-
GPHN	Hyperekplexia, Molybdenum Cofactor Deficiency	AD,AR	99.2	6 of 6
GRIA3	Mental Retardation	X,XR,G	98.39	-
GRIA4	Neurodevelopmental Disorder	AD	99.94	5 of 5
GRIK2	Mental Retardation	AR	96.98	5 of 6
GRIN1	Neurodevelopmental Disorder	AD,AR	100	43 of 43
GRIN2A	Epileptic Encephalopathy	AD	100	143 of 143
GRIN2B	Epileptic Encephalopathy, Mental Retardation, West Syndrome	AD	99.99	108 of 108
GRIN2D	Epileptic Encephalopathy	AD	79.74	17 of 18
GRN	Ceroid Lipofuscinosis, Frontotemporal Lobar Degeneration, Semantic Dementia	AD,AR	100	220 of 229
GTPBP3	Oxidative Phosphorylation Deficiency	AR	99.94	17 of 17
GUF1	Epileptic Encephalopathy, West Syndrome	AR	99.88	4 of 4
HACE1	Neuroblastoma, Spastic Paraplegia, Developmental Delay, Epilepsy	AR	100	15 of 15
HCN1	Epileptic Encephalopathy	AD	98.43	42 of 43
HCN2	Epilepsy, Retinitis Pigmentosa	-	70.45	6 of 9
HCN4	Brugada Syndrome, Sick Sinus Syndrome	AD	98.01	40 of 41
HDAC4	2q37 Microdeletion Syndrome	-	100	10 of 10
HECW2	Neurodevelopmental Disorder, Hypotonia, Seizures	AD	99.85	13 of 13
HEPACAM	Megalencephalic Leukoencephalopathy	AD,AR	97.87	30 of 30
HEXA	Tay-Sachs Disease	AR	100	205 of 206
HEXB	Sandhoff Disease	AR	99.92	109 of 115
HGSNAT	Mucopolysaccharidosis, Retinitis Pigmentosa	AR	87.91	69 of 73
HIBCH	3-Hydroxyisobutyryl-Coa Hydrolase Deficiency, Neurodegeneration	AR	96.47	27 of 27
HNRNPU	Epileptic Encephalopathy, 1q44 Microdeletion Syndrome	AD	99.8	36 of 36
HPD	Hawkinsinuria , Tyrosinemia	AD,AR	100	10 of 10

HRAS	Bladder Cancer, Costello Syndrome, Epidermal Nevus, Giant Pigmented Hairy Nevus, Schimmelpenning-Feuerstein-Mims Syndrome, Linear Nevus Sebaceus Syndrome	AD	100	34 of 34
HSD17B10	Hydroxyacyl-Coa Dehydrogenase Ii Deficiency	X,XD,G	100	-
HSD17B4	D-Bifunctional Protein Deficiency, Perrault Syndrome	AR	99.52	85 of 85
HSPD1	Leukodystrophy, Spastic Paraplegia	AD,AR	100	7 of 7
HTRA1	Cerebral Arteriopathy, Leukoencephalopathy	AD,AR	87.47	55 of 57
HTT	Huntington Disease, Lopes-Maciel-Rodan Syndrome	AD,AR	99	-
IBA57	Multiple Mitochondrial Dysfunctions Syndrome, Spastic Paraplegia	AR	93.35	25 of 27
IDH2	D-2-Hydroxyglutaric Aciduria, Maffucci Syndrome, Ollier Disease	AD	99.99	4 of 4
IDS	Mucopolysaccharidosis	X,XR,G	99.86	-
IER3IP1	Microcephaly, Epilepsy	AR	99.97	5 of 5
IQSEC2	Mental Retardation, Microduplication Xp11.22p11.23 Syndrome, Smith-Magenis Syndrome	X,XR,XD,G	99.73	-
IRF2BPL	Neurodevelopmental Disorder, Seizures	AD	95.01	24 of 25
ITPA	Epileptic Encephalopathy	AR	100	5 of 6
JMJD1C	22q11.2 Deletion Syndrome	-	99.09	27 of 27
JRK	Epilepsy	-	-	-
KANSL1	Koolen-De Vries Syndrome	AD	96.03	22 of 27
KCNA1	Ataxia, Epileptic Encephalopathy, Continuous Muscle Fiber Activity, Paroxysmal Kinesigenic Dyskinesia	AD	100	49 of 49
KCNA2	Epileptic Encephalopathy	AD	99.86	23 of 23
KCNAB2	1p36 Deletion Syndrome	-	79	3 of 3
KCNB1	Epileptic Encephalopathy	AD	99.95	55 of 55
KCNC1	Epilepsy	AD	99.87	10 of 10
KCND2	Autism, Epileptic Encephalopathy	-	100	4 of 4
KCNH1	Temple-Baraitser Syndrome, Zimmermann-Laband Syndrome	AD	99.69	15 of 15
KCNH2	Long Qt Syndrome, Short Qt Syndrome, Romano-Ward Syndrome	AD	98.69	908 of 930
KCNH5	Epileptic Encephalopathy, Neuropathy	-	98.72	1 of 1
KCNJ10	Enlarged Vestibular Aqueduct, Pendred Syndrome, Seizures, East Syndrome	AR	93.53	27 of 32
KCNJ11	Diabetes Mellitus, Hyperinsulinemic Hypoglycemia, Maturity-Onset Diabetes Of The Young, Hyperinsulinism, Dend Syndrome	AD,AR	100	190 of 191
KCNK4	Facial Dysmorphism, Epilepsy, Gingival Overgrowth	AD	94.93	2 of 2
KCNMA1	Cerebellar Atrophy, Epilepsy, Paroxysmal Dyskinesia, Liang-Wang Syndrome	AD,AR	99.98	24 of 26
KCNQ2	Epileptic Encephalopathy	AD	99.94	333 of 334
KCNQ3	Epilepsy	AD	97.94	40 of 40
KCNQ5	Mental Retardation	AD	95.08	8 of 8
KCNT1	Epileptic Encephalopathy	AD	95.98	64 of 64
KCNT2	Epileptic Encephalopathy	AD	98.26	4 of 4
KCNV2	Retinal Cone Dystrophy	AR	99.98	86 of 88
KCTD3	Variegate Porphyria, Niemann-Pick Disease	-	96.19	2 of 2
KCTD7	Epilepsy	AR	99.99	40 of 40
KDM5C	Mental Retardation	X,XR,G	100	-
KDM6A	Kabuki Syndrome	AD,X,XD,G	99.98	-
KIF1A	Mental Retardation, Neuropathy, Spastic Paraplegia, Peho Syndrome	AD,AR	100	76 of 76
KIFBP	Goldberg-Shprintzen Syndrome	AR	99.27	-
KMT2D	Kabuki Syndrome	AD	99.71	839 of 847
KMT2E	O'donnell-Luria-Rodan Syndrome, Intellectual Disability	AD	99.83	34 of 34
KPNA7	Cerebellar Malformation, Cerebellar Vermis Hypoplasia	-	100	3 of 3
KRAS	Aplasia Cutis Congenita, Cardiofaciocutaneous Syndrome, Leukemia, Noonan Syndrome, Autoimmune Lymphoproliferative Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome, Encephalocraniocutaneous Lipomatosis, Linear Nevus Sebaceus Syndrome, Lynch Syndrome, Toriello-Lacassie-Droste Syndrome	AD	100	38 of 38
L2HGDH	L-2-Hydroxyglutaric Aciduria	AR	100	72 of 73
LAMA2	Limb-Girdle Muscular Dystrophy	AR	100	363 of 377

LBR	Hydrops-Ectopic Calcification-Moth-Eaten Skeletal Dysplasia, Pelger-Huet Anomaly, Reynolds Syndrome, Greenberg Dysplasia, Reynolds Syndrome	AD,AR	99.98	34 of 34
LGI1	Epilepsy	AD	99.94	54 of 54
LIAS	Pyruvate Dehydrogenase Lipoic Acid Synthetase Deficiency	AR	99.82	8 of 8
LMNB1	Leukodystrophy	AD	99.66	4 of 4
LMNB2	Barraquer-Simons Syndrome, Epilepsy, Acquired Partial Lipodystrophy	AD,AR	95.03	5 of 5
LNPK	Neurodevelopmental Disorder With Epilepsy And Hypoplasia Of The Corpus Callosum	AR	99.26	-
LRPPRC	Leigh Syndrome	AR	98.94	18 of 18
LYRM7	Mitochondrial Complex Iii Deficiency	AR	99.86	9 of 9
MACF1	Lissencephaly	AD	99.94	18 of 18
MAGI2	Nephrotic Syndrome	AR	93.82	7 of 9
MAP2K1	Cardiofaciocutaneous Syndrome, Melorheostosis, Noonan Syndrome	AD	100	31 of 31
MAP2K2	Cardiofaciocutaneous Syndrome, Neurofibromatosis-Noonan Syndrome	AD	100	37 of 37
MAPK10	Lennox-Gastaut Syndrome	-	99.97	-
MARCHF6	Epilepsy	AD	99.97	-
MARS2	Ataxia, Oxidative Phosphorylation Deficiency, Spastic Ataxia With Leukoencephalopathy	AR	99.94	3 of 3
MBD5	2q23.1 Microdeletion Syndrome	AD	99.99	33 of 35
MBOAT7	Mental Retardation	AR	99.08	11 of 12
MCOLN1	Mucolipidosis	AR	99.99	34 of 36
MCPH1	Microcephaly	AR	99.51	18 of 19
MDH2	Epileptic Encephalopathy, Pheochromocytoma-Paraganglioma	AR	98	11 of 11
ME2	Epilepsy, Li-Fraumenti Syndrome	-	99.99	1 of 1
MECP2	Autism, Encephalopathy, Lubs Mental Retardation Syndrome, Rett Syndrome, Trisomy Xq28	X,XR,XD,MU,G	99.81	-
MED12	Lujan-Fryns Syndrome, Ohdo Syndrome, Opitz-Kaveggia Syndrome, Blepharophimosis-Intellectual Disability Syndrome, Fg Syndrome	X,XR,G	100	-
MED17	Microcephaly	AR	100	9 of 9
MEF2C	Mental Retardation, Stereotypic Movements, Epilepsy, Cerebralmalformations, 5q14.3 Microdeletion Syndrome	AD	99.91	43 of 46
MFSD8	Ceroid Lipofuscinosis, Macular Dystrophy	AR	100	63 of 63
MGAT2	Congenital Disorder Of Glycosylation	AR	97.19	5 of 5
MICAL1	Epilepsy	AD	99.98	3 of 3
MIPEP	Combined Oxidative Phosphorylation Deficiency	AR	99.84	7 of 8
MLC1	Megalencephalic Leukoencephalopathy	AR	100	104 of 106
MOCS1	Molybdenum Cofactor Deficiency	AR	100	36 of 37
MOCS2	Molybdenum Cofactor Deficiency	AR	100	32 of 32
MOGS	Congenital Disorder Of Glycosylation	AR	100	10 of 10
MPDU1	Congenital Disorder Of Glycosylation	AR	100	7 of 7
MRPL44	Oxidative Phosphorylation Deficiency	AR	99.75	2 of 2
MTFMT	Oxidative Phosphorylation Deficiency, Mitochondrial Complex I Deficiency, Leigh Syndrome With Leukodystrophy	AR	99.52	18 of 18
MTHFR	Homocystinuria, Neural Tube Defects, Schizophrenia, Thrombophiliavenous Thromboembolism, Isolated Anencephaly, Isolated Exencephaly	AD,AR	100	122 of 122
MTHFS	Neurodevelopmental Disorder With Microcephaly, Epilepsy, Hypomyelination	AR	100	5 of 5
MTOR	Dysplasia Of Taylor, Smith-Kingsmore Syndrome, Macrocephaly-Intellectual Disability-Neurodevelopmental Disorder-Small Thorax Syndrome	AD	99.98	39 of 39
NACC1	Neurodevelopmental Disorder With Epilepsy, Cataracts, Feeding Difficulties, Delayed Brain Myelination	AD	99.99	3 of 3
NAGLU	Charcot-Marie-Tooth Disease, Mucopolysaccharidosis	AD,AR	93.23	194 of 222
NBEA	Autism, Oxidative Phosphorylation Deficiency	-	99.48	27 of 27
NDE1	Lissencephaly, Microhydranencephaly	AR	86.55	12 of 13
NDST1	Mental Retardation	AR	99.99	11 of 11
NDUFA1	Mitochondrial Complex I Deficiency	X,XR,G	100	-

NDUFA2	Mitochondrial Complex I Deficiency, Leigh Syndrome With Leukodystrophy	AR	99.84	2 of 3
NDUFAF3	Mitochondrial Complex I Deficiency, Leigh Syndrome With Cardiomyopathy	AR	100	9 of 9
NDUFAF5	Mitochondrial Complex I Deficiency, Leigh Syndrome With Leukodystrophy	AR	100	13 of 14
NDUFAF6	Fanconi Renotubular Syndrome, Mitochondrial Complex I Deficiency, Leigh Syndrome With Leukodystrophy	AR	99.4	12 of 13
NDUFS1	Mitochondrial Complex I Deficiency, Leigh Syndrome With Leukodystrophy	AR	99.98	30 of 30
NDUFS2	Mitochondrial Complex I Deficiency, Leber Hereditary Optic Neuropathy, Leigh Syndrome With Cardiomyopathy, Leukodystrophy	AR	100	26 of 26
NDUFS3	Mitochondrial Complex I Deficiency, Leigh Syndrome With Leukodystrophy	AR	93.67	4 of 4
NDUFS4	Mitochondrial Complex I Deficiency, Leigh Syndrome With Leukodystrophy	AR,X,XD,MI,G	100	15 of 15
NDUFS6	Mitochondrial Complex I Deficiency	AR	100	6 of 6
NDUFS7	Mitochondrial Complex I Deficiency, Leigh Syndrome With Leukodystrophy	AR	88	6 of 7
NDUFS8	Mitochondrial Complex I Deficiency, Leigh Syndrome With Leukodystrophy	AR	100	16 of 16
NDUFV1	Mitochondrial Complex I Deficiency, Leigh Syndrome With Leukodystrophy	AR	100	36 of 36
NECAP1	Epileptic Encephalopathy	AR	99.83	2 of 2
NEDD4L	Periventricular Nodular Heterotopia	AD	97.61	10 of 10
NEU1	Neuraminidase Deficiency, Sialidosis	AR	100	68 of 68
NEUROD2	Epileptic Encephalopathy	AD	96.88	2 of 2
NEXMIF	Mental Retardation	X,XR,XD,G	99.74	-
NF1	Juvenile Myelomonocytic Leukemia, Neurofibromatosis-Noonan Syndrome, Watson Syndrome, 17q11.2 Microduplication Syndrome, Pheochromocytoma-Paraganglioma	AD	97.97	3082 of 3166
NFU1	Multiple Mitochondrial Dysfunctions Syndrome	AR	100	13 of 15
NGLY1	Congenital Disorder Of Glycosylation, Alacrimia-Choreoathetosis-Liver Dysfunction Syndrome	AR	99.8	28 of 28
NHLRC1	Lafora Disease	AR	100	71 of 71
NIPBL	Cornelia De Lange Syndrome	AD	99.32	409 of 426
NKX6-2	Spastic Ataxia, Hypomyelinating Leukodystrophy	AR	82.95	8 of 9
NOTCH3	Cerebral Arteriopathy, Leukoencephalopathy, Lateral Meningocele Syndrome, Myofibromatosis, Myofibromatosis	AD	96.31	398 of 399
NPC1	Niemann-Pick Disease	AR	97	503 of 505
NPC2	Niemann-Pick Disease	AR	100	27 of 27
NPHP1	Joubert Syndrome, Nephronophthisis, Senior-Loken Syndrome, Bardet-Biedl Syndrome, Joubert Syndrome With Renal Defect	AR	100	58 of 59
NPRL2	Epilepsy	AD	100	12 of 12
NPRL3	Epilepsy	AD	99.61	18 of 18
NR2F1	Bosch-Boonstra Optic Atrophy Syndrome	AD	89.78	26 of 31
NRXN1	Pitt-Hopkins-Like Syndrome	AR	97.42	33 of 74
NSD1	Sotos Syndrome, 5q35 Microduplication Syndrome, Weaver Syndrome	AD	99.8	451 of 459
NT5C2	Spastic Paraplegia	AR	97.89	6 of 7
NTNG1	Atypical Rett Syndrome	-	99.96	2 of 2
NTRK2	Epileptic Encephalopathy, Obesity, Hyperphagia, Developmental Delay, West Syndrome	AD	100	9 of 9
NUBPL	Mitochondrial Complex I Deficiency	AR	95.2	13 of 13
NUS1	Congenital Disorder Of Glycosylation, Mental Retardation, Epileptic Encephalopathy	AD,AR	99.62	22 of 23
OFD1	Joubert Syndrome, Orofaciodigital Syndrome, Retinitis Pigmentosa, Simpson-Golabi-Behmel Syndrome, Primary Ciliary Dyskinesia	X,XR,XD,G	98.09	-
OPHN1	Mental Retardation, Cerebellar Hypoplasia, Distinctivefacial Appearance	X,XR,G	100	-
P4HTM	Hypotonia, Hyperventilation, Impaired Intellectual Development, Dysautonomia, Epilepsy, Eye Abnormalities	AR	92.81	5 of 5

PACS1	Intellectual Disability-Craniofacial Dysmorphism-Cryptorchidism Syndrome	AD	97.98	3 of 3
PACS2	Epileptic Encephalopathy	AD	99.52	3 of 3
PAFAH1B1	17p13.3 Microduplication Syndrome, Lissencephaly, Miller-Dieker Syndrome	AD	99.95	90 of 92
PAK3	Mental Retardation	X,XR,G	99.96	-
PANK2	Hypoprebetalipoproteinemia, Acanthocytosis, Retinitis Pigmentosa, Pallidal Degeneration, Neurodegeneration, Pantothenate Kinase-Associated Neurodegeneration	AR	98.92	177 of 182
PARS2	Epileptic Encephalopathy	AR	100	7 of 7
PC	Pyruvate Carboxylase Deficiency	AR	100	48 of 48
PCDH19	Epilepsy, Dravet Syndrome	X,G	99.99	-
PCNT	Microcephalic Osteodysplastic Primordial Dwarfism, Seckel Syndrome	AR	99.92	103 of 105
PDHA1	Pyruvate Decarboxylase Deficiency, Leigh Syndrome With Leukodystrophy	X,XD,G	99.02	-
PDSS2	Coenzyme Q10 Deficiency, Leigh Syndrome With Nephrotic Syndrome	AR	99.99	6 of 6
PEX1	Hearing Loss With Enamel Hypoplasia And Nail Defects, Peroxisome Biogenesis Disorder, Zellweger Syndrome, Refsum Disease, Neonatal Adrenoleukodystrophy	AR	97.02	126 of 134
PEX10	Zellweger Syndrome, Refsum Disease, Neonatal Adrenoleukodystrophy	AR	99.76	29 of 32
PEX12	Zellweger Syndrome, Refsum Disease, Neonatal Adrenoleukodystrophy	AR	100	38 of 38
PEX14	Refsum Disease, Neonatal Adrenoleukodystrophy, Zellweger Syndrome	AR	100	4 of 4
PEX2	Refsum Disease, Neonatal Adrenoleukodystrophy, Zellweger Syndrome	AR	99.89	17 of 17
PEX26	Refsum Disease, Neonatal Adrenoleukodystrophy, Zellweger Syndrome	AR	100	29 of 29
PEX3	Refsum Disease, Neonatal Adrenoleukodystrophy, Zellweger Syndrome	AR	100	9 of 9
PEX5	Adrenoleukodystrophy, Cerebrohepatorenal Syndrome, Rhizomelic Chondrodysplasia Punctata, Refsum Disease, Zellweger Syndrome	AR	100	12 of 12
PEX6	Heimler Syndrome, Spinocerebellar Ataxia-Blindness-Deafness Syndrome, Deafness-Enamel Hypoplasia-Nail Defects Syndrome, Refsum Disease, Neonatal Adrenoleukodystrophy, Zellweger Syndrome	AD,AR	99.94	105 of 108
PEX7	Zellweger Syndrome, Rhizomelic Chondrodysplasia Punctata, Refsum Disease	AR	99.21	47 of 53
PGK1	Phosphoglycerate Kinase 1 Deficiency, Glycogen Storage Disease	X,XR,G	100	-
PHACTR1	Epileptic Encephalopathy, West Syndrome	AD	99.89	5 of 5
PHF6	Borjeson-Forssman-Lehmann Syndrome	X,XR,G	99.93	-
PIGA	Paroxysmal Nocturnal Hemoglobinuria, West Syndrome	X,XR,G	97.98	-
PIGB	Epileptic Encephalopathy	AR	99.97	10 of 10
PIGC	Glycosylphosphatidylinositol Biosynthesis Defect	AR	99.59	4 of 4
PIGG	Epilepsy-Intellectual Disability-Brain Anomalies Syndrome	AR	99.86	6 of 6
PIGN	Fryns Syndrome, Multiple Congenital Anomalies-Hypotonia-Seizures Syndrome	AR	93.97	36 of 39
PIGO	Hyperphosphatasia With Mental Retardation Syndrome	AR	99.93	21 of 21
PIGP	Epileptic Encephalopathy	AR	99.98	2 of 2
PIGQ	Epileptic Encephalopathy	AR	99.99	4 of 4
PIGS	Glycosylphosphatidylinositol Biosynthesis Defect	AR	100	6 of 6
PIGT	Paroxysmal Nocturnal Hemoglobinuria, Intellectual Disability-Seizures-Hypophosphatasia-Ophthalmic-Skeletal Anomalies Syndrome	AD,AR	100	15 of 15
PIGV	Hyperphosphatasia-Intellectual Disability Syndrome	AR	99.99	16 of 16
PIGW	Hyperphosphatasia-Intellectual Disability Syndrome	AR	99.52	6 of 6
PIK3AP1	Bissinosis, Central Nervous System Tuberculosis	-	99.98	5 of 5
PITRM1	Alzheimer Disease, Berylliosis	-	100	3 of 3
PLA2G6	Neuroaxonal Dystrophy, Neurodegeneration With Brain Iron Accumulation, Parkinson Disease	AR	99.94	190 of 191

PLAA	Neurodevelopmental Disorder, Microcephaly, Spasticity	AR	99.41	6 of 6
PLCB1	Epileptic Encephalopathy, West Syndrome	AR	99.92	4 of 6
PLP1	Pelizaeus-Merzbacher Disease, Spastic Paraplegia	X,XR,G	100	-
PLPBP	Epilepsy	AR	100	-
PMM2	Congenital Disorder Of Glycosylation	AR	100	127 of 129
PMPCB	Multiple Mitochondrial Dysfunctions Syndrome	AR	99.46	5 of 5
PNKD	Paroxysmal Nonkinesigenic Dyskinesia	AD	99.98	6 of 6
PNKP	Ataxia-Oculomotor Apraxia, Charcot-Marie-Tooth Disease, Epileptic Encephalopathy	AR	100	36 of 36
PNPO	Pyridoxamine 5-Prime-Phosphate Oxidase Deficiency, Pyridoxal Phosphate-Responsive Seizures	AR	99.99	31 of 31
POLG	Mitochondrial Dna Depletion Syndrome, External Ophthalmoplegia With Mitochondrial Dna Deletions, Sensory Ataxic Neuropathy, Dysarthria, Ophthalmoparesis, Alpers-Huttenlocher Syndrome, Mitochondrial Neurogastrointestinal Encephalomyopathy	AD,AR	99.92	325 of 326
POLR3A	Leukodystrophy, Progeroid Syndrome, Tremor-Ataxia-Central Hypomyelination Syndrome, Wiedemann-Rautenstrauch Syndrome	AR	100	122 of 122
POLR3B	Leukodystrophy, Hypomyelination-Hypogonadotropic Hypogonadism-Hypodontia Syndrome	AR	100	61 of 61
POMGNT1	Muscular Dystrophy-Dystroglycanopathy, Retinitis Pigmentosa, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	99.91	82 of 83
POMT1	Limb-Girdle Muscular Dystrophy-Dystroglycanopathy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	100	105 of 105
POMT2	Limb-Girdle Muscular Dystrophy-Dystroglycanopathy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	100	74 of 74
PPP2CA	Neurodevelopmental Disorder And Language Delay	AD	99.9	14 of 14
PPP3CA	Arthrogryposis, Cleft Palate, Craniosynostosis, Impaired Intellectual Development, Epileptic Encephalopathy	AD	99.98	16 of 16
PPT1	Ceroid Lipofuscinosis	AR	100	81 of 81
PQBP1	Renpenning Syndrome, Hamel Cerebro-Palato-Cardiac Syndrome	X,XR,G	99.99	-
PRDM8	Epilepsy, Lafora Body Disease	AR	89.24	1 of 1
PRICKLE1	Epilepsy, Unverricht-Lundborg Disease	AR	98.41	23 of 23
PRICKLE2	Epilepsy	-	94.92	6 of 6
PRIMA1	Miocardial Infarction, Ichthyosis	-	99.59	1 of 1
PRODH	Hyperprolinemia, Schizophrenia	AD,AR	98.57	5 of 5
PRRT2	Convulsions, Dyskinesia, Epilepsy, Hemiplegic Migraine	AD	99.93	111 of 111
PSAP	Saposin Deficiency, Gaucher Disease, Krabbe Disease, Metachromatic Leukodystrophy, Encephalopathy	AR	100	33 of 33
PTCH1	Basal Cell Carcinoma, Basal Cell Nevus Syndrome, Holoprosencephaly, Gorlin Syndrome, Monosomy 9q22.3	AD	98.89	498 of 502
PTEN	Cowden Disease, Autism, Meningioma, Bannayan-Riley-Ruvalcaba Syndrome, Juvenile Polyposis Of Infancy, Lhermitte-Duclos Disease, Proteus Syndrome, Segmental Outgrowth-Lipomatosis-Arteriovenous Malformation-Epidermal Nevus Syndrome	AD	99.97	609 of 629
PTPN11	Myelomonocytic Leukemia, Leopard Syndrome, Metachondromatosis, Noonan Syndrome	AD	100	150 of 151
PTPN23	Neurodevelopmental Disorder And Structural Brain Anomalies	AR	99.99	17 of 17
PTS	6-Pyruvoyl-Tetrahydropterin Synthase Deficiency	AR	99.97	108 of 112
PUM1	Spinocerebellar Ataxia	AD	99.98	8 of 8
PURA	Mental Retardation	AD	85.36	59 of 65
PYCR2	Leukodystrophy, Leukoencephalopathy	AR	98.29	14 of 14
QARS	Microcephaly	-	100	12 of 12
QDPR	Phenylketonuria, Dihydropteridine Reductase Deficiency	AR	100	66 of 67
RAB39B	Mental Retardation, Parkinsonism	X,XR,G	100	-
RAB3GAP1	Warburg Micro Syndrome, Cataract-Intellectual Disability-Hypogonadism Syndrome	AR	99.94	70 of 70
RAI1	Smith-Magenis Syndrome, 17p11.2 Microduplication Syndrome, Gene Duplication Syndrome	AD	99.91	50 of 53
RALA	Tuberculosos, Myocardial Infarction	-	99.94	7 of 7
RANBP2	Necrotizing Encephalopathy	AD	99.41	9 of 9

RARS1	Leukodystrophy	AR	99.64	28 of 28
RARS2	Pontocerebellar Hypoplasia	AR	99.98	39 of 40
RBFOX1	Epilepsy, Spinocerebellar Ataxia, Developmental Coordination Disorder	-	97.99	4 of 5
RBFOX3	Epilepsy, Ectodermal Dysplasia	-	88.9	1 of 1
RELN	Epilepsy, Lissencephaly	AD,AR	100	70 of 70
RFT1	Congenital Disorder Of Glycosylation	AR	99.98	18 of 18
RHOBTB2	Epileptic Encephalopathy	AD	100	6 of 6
RMND1	Combined Oxidative Phosphorylation Deficiency	AR	99.67	15 of 16
RNASEH2A	Aicardi-Goutieres Syndrome	AR	100	23 of 23
RNASEH2B	Aicardi-Goutieres Syndrome	AR	99.95	41 of 41
RNASEH2C	Aicardi-Goutieres Syndrome	AR	100	14 of 14
RNASET2	Leukoencephalopathy	AR	100	11 of 13
RNF13	Epileptic Encephalopathy	AD	99.88	2 of 2
RNF216	Cerebellar Ataxia-Hypogonadism Syndrome	AR	99.89	15 of 15
RNR1	MERRF (Myoclonus Epilepsy Associated With Ragged-Red Fibres)	-	-	-
ROGDI	Kohlschutter-Tonz Syndrome, Amelocerebrohypohidrotic Syndrome	AR	99.83	10 of 12
RORA	Intellectual Developmental Disorder, Epilepsy, Cerebellar Ataxia	AD	99.94	12 of 12
RORB	Epilepsy	AD	99.98	4 of 4
RPGRIP1L	Coach Syndrome, Joubert Syndrome, Meckel Syndrome, Joubert Syndrome	AR	99.96	52 of 52
RUBCN	Spinocerebellar And Cerebellar Ataxia	AR	99.96	-
RYR3	Deafness	-	99.98	20 of 20
SAMD12	Epilepsy	AD	99.74	-
SAMHD1	Aicardi-Goutieres Syndrome	AD,AR	100	51 of 51
SATB2	Chromosome 2q32-Q33 Deletion Syndrome	AD	99.87	97 of 124
SCARB2	Action Myoclonus-Renal Failure Syndrome, Gaucher Disease, Unverricht-Lundborg Disease	AR	99.95	29 of 29
SCN10A	Episodic Pain Syndrome, Brugada Syndrome, Paroxysmal Extreme Pain Disorder, Primary Erythromelalgia, Romano-Ward Syndrome	AD	99.89	96 of 96
SCN1A	Epileptic Encephalopathy, Febrile Convulsions, Migraine, Dravet Syndrome, Lennox-Gastaut Syndrome	AD	99.8	1776 of 1797
SCN1B	Atrial Fibrillation, Brugada Syndrome, Epileptic Encephalopathy, Dravet Syndrome, Familial Progressive Cardiac Conduction Defect	AD,AR	99.67	46 of 48
SCN2A	Epileptic Encephalopathy, Episodic Ataxia, Seizures, Dravet Syndrome, West Syndrome	AD	100	351 of 351
SCN3A	Epileptic Encephalopathy	AD	99.98	18 of 18
SCN4A	Hyperkalemic Periodic Paralysis, Myasthenic Syndrome, Myotonia, Paramyotonia Congenita Of Von Eulenburg, Postsynaptic Congenital Myasthenic Syndromes	AD,AR	99.77	136 of 142
SCN5A	Atrial Fibrillation, Brugada Syndrome, Cardiomyopathy, Long Qt Syndrome, Heart Block, Sick Sinus Syndrome, Sudden Infant Death Syndrome, Romano-Ward Syndrome	AD,AR,MU	99.45	929 of 942
SCN8A	Cognitive Impairment With Or Without Cerebellar Ataxia, Epileptic Encephalopathy, Myoclonus, Seizures, Infantile Convulsions And Choreoathetosis	AD	97.85	156 of 172
SCN9A	Erythermalgia, Epilepsy, Indifference To Pain, Neuropathy, Extreme Pain Disorder, Dravet Syndrome, Primary Erythromelalgia	AD,AR	96.25	126 of 137
SCO1	Complex Iv Deficiency	AR,MI	100	6 of 6
SCO2	Cardioencephalomyopathy, Myopia, Charcot-Marie-Tooth Disease, Leigh Syndrome With Cardiomyopathy	AD,AR	100	38 of 38
SDHA	Cardiomyopathy, Leigh Syndrome, Paragangliomas, Pheochromocytoma-Paraganglioma, Succinate-Coq Reductase Deficiency	AD,AR,MI	99.98	103 of 103
SDHAF1	Succinate-Coq Reductase Deficiency	AR	100	6 of 6
SERAC1	Methylglutaconic Aciduria, Deafness, Encephalopathy, Leigh-Like Syndrome	AR	99.93	53 of 53
SERPINI1	Encephalopathy	AD	100	9 of 9

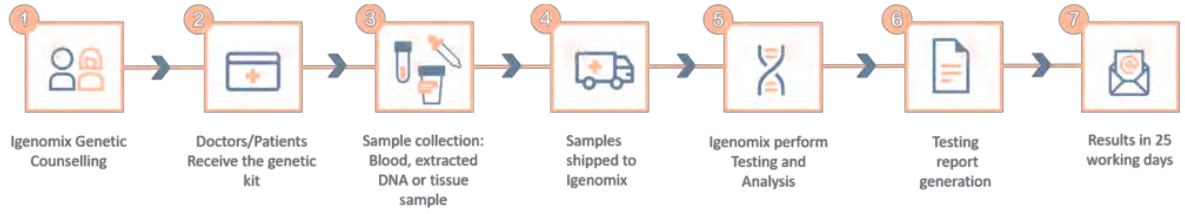
SETBP1	Mental Retardation, Schinzel-Giedion Midface-Retraction Syndrome, Intellectual Disability-Expressive Aphasia-Facial Dysmorphism Syndrome	AD	98.61	43 of 43
SETD2	Luscan-Lumish Syndrome, Sotos Syndrome	AD	99.83	19 of 19
SGCE	Myoclonic Dystonia	AD	99.46	94 of 98
SGSH	Mucopolysaccharidosis	AR	97.7	151 of 151
SHH	Holoprosencephaly, Microphthalmia, Schizencephaly, Hypoplastic Tibiae-Postaxial Polydactyly Syndrome, Radial Hemimelia, Syndactyly	AD	99.48	161 of 184
SHOC2	Noonan Syndrome	AD	99.98	8 of 8
SIK1	Myoclonic And Epileptic Encephalopathy, West Syndrome	AD	99.67	9 of 9
SIX3	Holoprosencephaly, Schizencephaly	AD	99.79	79 of 80
SLC12A5	Epileptic Encephalopathy	AD,AR	100	19 of 19
SLC13A5	Epileptic Encephalopathy, Amelocerebrohypohidrotic Syndrome	AR	95.92	24 of 24
SLC17A5	Infantile Sialic Acid Storage Disorder, Sialuria	AR	99.91	49 of 49
SLC19A3	Basal Ganglia Disease, Leigh Syndrome, Leukodystrophy	AR	100	38 of 39
SLC1A2	Epileptic Encephalopathy	AD	100	7 of 7
SLC1A3	Episodic Ataxia	AD	100	13 of 13
SLC1A4	Spastic Tetraplegia, Microcephaly	AR	99.76	8 of 9
SLC25A1	D-2- And L-2-Hydroxyglutaric Aciduria, Myasthenic Syndrome	AR	90	23 of 25
SLC25A12	Epileptic Encephalopathy	AR	100	7 of 7
SLC25A15	Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	AR	100	41 of 41
SLC25A19	Microcephaly, Thiamine Metabolism Dysfunction Syndrome	AR	97.13	10 of 10
SLC25A22	Epileptic Encephalopathy, Myoclonic Encephalopathy	AR	100	16 of 16
SLC25A42	Metabolic Crises, Encephalomyopathy	AR	99.91	2 of 2
SLC2A1	Choreoathetosis, Epilepsy, Glucose Transport Defect, Stomatin-Deficient Cryohydrocytosis	AD,AR	99.99	301 of 304
SLC35A1	Congenital Disorder Of Glycosylation	AR	100	6 of 6
SLC35A2	Congenital Disorder Of Glycosylation	X,XD,G	99.97	-
SLC35A3	Arthrogryposis, Autism Spectrum Disorder-Epilepsy-Arthrogryposis Syndrome	AR	99.94	5 of 5
SLC35C1	Congenital Disorder Of Glycosylation	AR	99.73	8 of 8
SLC39A8	Congenital Disorder Of Glycosylation	AR	99.89	7 of 7
SLC46A1	Folate Malabsorption	AR	99.8	21 of 21
SLC4A10	Epilepsy, Corneal Dystrophy	-	99.94	4 of 4
SLC6A1	Myoclonic-Astastic Epilepsy	AD	100	55 of 55
SLC6A5	Hyperekplexia	AD,AR	100	37 of 37
SLC6A8	Creatine Deficiency Syndrome	X,XR,G	99.87	-
SLC6A9	Glycine Encephalopathy	AR	99.99	5 of 5
SLC9A6	Christianson Syndrome	X,XD,G	98.87	-
SMARCA2	Nicolaides-Baraitser Syndrome, Intellectual Disability-Sparse Hair-Brachydactyly Syndrome	AD	97.99	80 of 81
SMC1A	Cornelia De Lange Syndrome, Semilobar Holoprosencephaly, Wiedemann-Steiner Syndrome	X,XR,XD,G	100	-
SMC3	Cornelia De Lange Syndrome	AD	100	30 of 30
SMS	Mental Retardation	X,XR,G	84.04	-
SNAP25	Myasthenic Syndrome	AD	100	6 of 6
SNIP1	Psychomotor Retardation, Craniofacial Dysmorphism	AR	99.68	1 of 1
SNORD118	Leukoencephalopathy	AR	-	-
SNX27	Epilepsy, Parkinson Disease	-	99.52	1 of 1
SOX10	Waardenburg Syndrome, Kallmann Syndrome	AD	99.74	139 of 147
SPATA5	Epilepsy, Deafness	AR	99.83	30 of 30
SPR	Dystonia, Sepiapterin Reductase Deficiency	AD,AR	99.89	27 of 27
SPRED1	Legius Syndrome	AD	100	84 of 84
SPTAN1	Epileptic Encephalopathy, West Syndrome	AD	100	52 of 53
SPTBN4	Myopathy, Deafness	AR	99.26	10 of 10
SRGAP2	Epileptic Encephalopathy, Pilocytic Astrocytoma, West Syndrome, Chromosome 3pter-P25 Deletion Syndrome	-	96.8	1 of 1
SRPX2	Rolandic Epilepsy, Speech Dyspraxia, Bilateral Perisylvian Polymicrogyria	AD	100	-

ST3GAL3	Epileptic Encephalopathy, Mental Retardation, West Syndrome	AR	100	5 of 5
ST3GAL5	Epilepsy Syndrome	AR	99.17	6 of 6
STAG1	Mental Retardation, Facial Dysmorphism, Gastroesophageal Reflux	AD	99.98	16 of 22
STARD7	Epilepsy	AD	98.13	1 of 1
STIL	Microcephaly	AR	99.94	18 of 18
STRADA	Polyhydramnios, Megalencephaly, Epilepsy	AR	97.95	4 of 6
STX1B	Generalized Epilepsy, Febrile Seizures	AD	100	24 of 24
STXBP1	Epileptic Encephalopathy, 9q33.3q34.11 Microdeletion Syndrome, Rett Syndrome, Dravet Syndrome, West Syndrome	AD	100	209 of 215
SUMF1	Multiple Sulfatase Deficiency	AR	100	52 of 52
SUOX	Sulfocysteinuria	AR	99.98	28 of 28
SURF1	Charcot-Marie-Tooth Disease, Leigh Syndrome, Leukodystrophy	AR,MI	98.59	117 of 124
SYN1	Epilepsy, Learning Disabilities	X,XR,XD,G	91.7	-
SYNGAP1	Mental Retardation, Epileptic Encephalopathy	AD	99.46	168 of 171
SYNJ1	Epileptic Encephalopathy, Parkinson Disease	AR	99.81	30 of 32
SYP	Mental Retardation	X,XR,G	99.98	-
SZT2	Epileptic Encephalopathy	AR	99.98	39 of 39
TACO1	Mitochondrial Complex Iv Deficiency, Leigh Syndrome, Leukodystrophy	AR,MI	100	3 of 3
TAF1	Dystonia, Mental Retardation, Parkinsonism	X,XR,G	99.74	-
TBC1D20	Warburg Micro Syndrome	AR	99.94	6 of 6
TBC1D24	Deafness, Doors Syndrome, Epileptic Encephalopathy, Myoclonic Epilepsy, Dystonia	AD,AR	100	80 of 80
TBCD	Encephalopathy, Diffuse Brain Atrophy, Microcephaly	AR	94.89	28 of 28
TBCE	Epileptic Encephalopathy, Hypoparathyroidism-Retardation-Dysmorphism Syndrome, Kenny-Caffey Syndrome, Spastic Ataxia, Sanjad-Sakati Syndrome	AR	100	8 of 8
TBCK	Hypotonia, Intellectual Disability	AR	99.95	15 of 15
TBL1XR1	Mental Retardation, Pierpont Syndrome, Promyelocytic Leukemia	AD	99.78	23 of 23
TBX1	Conotruncal Heart Malformations, Digeorge Syndrome, Tetralogy Of Fallot, Velocardiofacial Syndrome, 22q11.2 Deletion Syndrome, 22q11.2 Microduplication Syndrome	AD,AR	88.7	35 of 42
TCF4	Corneal Dystrophy, Pitt-Hopkins Syndrome, Sclerosing Cholangitis	AD	98.91	124 of 124
TDP2	Spinocerebellar Ataxia	AR	99.93	8 of 8
TIMM50	3-Methylglutaconic Aciduria	AR	91	7 of 7
TMEM67	Bardet-Biedl Syndrome, Coach Syndrome, Joubert Syndrome, Meckel Syndrome, Nephronophthisis, Rhyns Syndrome	AR	96.93	177 of 179
TMEM70	Mitochondrial Complex V Deficiency, Encephalo-Cardiomyopathy	AR	100	22 of 24
TNK2	Gastric Adenocarcinoma, Epilepsy	-	99.72	6 of 7
TPK1	Thiamine Metabolism Dysfunction Syndrome	AR	99.81	15 of 15
TPP1	Ceroid Lipofuscinosis, Spinocerebellar Ataxia	AR	100	147 of 147
TRAK1	Epileptic Encephalopathy	AR	99.28	7 of 7
TRAPPC6B	Neurodevelopmental Disorder, Microcephaly, Epilepsy, Brain Atrophy	AR	100	4 of 4
TREX1	Aicardi-Goutieres Syndrome, Chilblain Lupus, Systemic Lupus Erythematosus, Vasculopathy, Cerebral Leukodystrophy	AD,AR	100	75 of 75
TRIM8	Epileptic Encephalopathy	-	99.5	7 of 7
TRNF	Mitochondrial Myopathy, Epileptic Encephalopathy, MELAS (Myoclonic Encephalopathy, Lactic Acidosis, And Stroke-Like Episodes), MERRF (Myoclonus Epilepsy Associated With Ragged-Red Fibres)	MI	-	-
TRNH	MELAS (Myoclonic Encephalopathy, Lactic Acidosis, And Stroke-Like Episodes), MERRF (Myoclonus Epilepsy Associated With Ragged-Red Fibres)	-	-	-
TRNI	MERRF (Myoclonus Epilepsy Associated With Ragged-Red Fibres)	MI	-	-
TRNK	MELAS (Myoclonic Encephalopathy, Lactic Acidosis, And Stroke-Like Episodes), MERRF (Myoclonus Epilepsy Associated With Ragged-Red Fibres), Leigh Syndrome, Cardiomyopathy, Deafness	MI	-	-

TRNL1	MELAS (Myoclonic Encephalopathy, Lactic Acidosis, And Stroke-Like Episodes), MERRF (Myoclonus Epilepsy Associated With Ragged-Red Fibres), Kearns-Sayre Syndrome, Deafness, Leigh Syndrome, Ophthalmoplegia	MI	-	-
TRNP	MERRF (Myoclonus Epilepsy Associated With Ragged-Red Fibres)	MI	-	-
TRNQ	MELAS (Myoclonic Encephalopathy, Lactic Acidosis, And Stroke-Like Episodes), MERRF (Myoclonus Epilepsy Associated With Ragged-Red Fibres)	MI	-	-
TRNS1	Mitochondrial Complex Iv Deficiency, MELAS (Myoclonic Encephalopathy, Lactic Acidosis, And Stroke-Like Episodes), MERRF (Myoclonus Epilepsy Associated With Ragged-Red Fibres), Ophthalmoplegia, Palmoplantar Keratoderma-Deafness Syndrome	AR,MI	-	-
TRNS2	MELAS (Myoclonic Encephalopathy, Lactic Acidosis, And Stroke-Like Episodes), MERRF (Myoclonus Epilepsy Associated With Ragged-Red Fibres), Usher Syndrome	MI	-	-
TSC1	Dysplasia Of Taylor, Lymphangioleiomyomatosis, Tuberous Sclerosis	AD	99.86	390 of 406
TSC2	Dysplasia Of Taylor, Lymphangioleiomyomatosis, Tuberous Sclerosis	AD	100	1157 of 1159
TSEN2	Pontocerebellar Hypoplasia	AR	95.47	4 of 5
TSEN34	Pontocerebellar Hypoplasia	AR	100	1 of 1
TSEN54	Encephalopathy, Olivopontocerebellar Hypoplasia	AR	96.94	20 of 22
TTC19	Mitochondrial Complex Iii Deficiency	AR	95.3	10 of 12
TUBA1A	Lissencephaly	AD	100	95 of 95
TUBA8	Polymicrogyria With Optic Nerve Hypoplasia	AR	80.97	5 of 5
TUBB2A	Cortical Dysplasia	AD	81.71	5 of 7
TUBB2B	Cortical Dysplasia, Dysequilibrium Syndrome, Polymicrogyria	AD	84.28	29 of 38
TUBB4A	Dystonia, Leukodystrophy	AD	89.81	44 of 44
TWNK	Spinocerebellar Ataxia, Perrault Syndrome, Ophthalmoplegia, Dysarthria	AD,AR	-	-
UBA5	Epileptic Encephalopathy, Spinocerebellar Ataxia	AR	99.98	19 of 19
UBE2A	Mental Retardation	X,XR,G	99.99	-
UBE3A	Angelman Syndrome, 15q11q13 Microduplication Syndrome	AD	99.98	208 of 211
UNC80	Hypotonia, Speech Impairment	AR	99.95	39 of 39
VAMP2	Neurodevelopmental Disorder, Hypotonia, Hyperkinetic Movements	AD	99.62	5 of 5
VARS1	Neurodevelopmental Disorder, Microcephaly, Cortical Atrophy	AR	97.86	19 of 20
VPS13A	Choreoacanthocytosis	AR	99.37	120 of 122
VPS13B	Cohen Syndrome	AR	99.98	182 of 190
WARS2	Neurodevelopmental Disorder, Lactic Acidosis, Oxidative Phosphorylation Defect	AR	99.95	14 of 15
WASF1	Neurodevelopmental Disorder, Seizures	AD	97.03	3 of 3
WDR26	Skraban-Deardorff Syndrome, Intellectual Disability, Seizures, Facial Dysmorphism	AD	99.31	22 of 22
WDR45	Neurodegeneration, Brain Iron Accumulation, West Syndrome	X,XD,G	100	-
WWOX	Epileptic Encephalopathy, Esophageal Cancer, Spinocerebellar Ataxia, Gonadal Dysgenesis, Squamous Cell Carcinoma Of The Esophagus	AR	99.94	44 of 44
YEATS2	Myoclonic Epilepsy	AD	99.98	1 of 1
YWHAG	Epileptic Encephalopathy	AD	99.94	5 of 5
ZDHHC9	Mental Retardation	X,G	100	-
ZEB2	Mowat-Wilson Syndrome	AD	98.95	253 of 254
ZFYVE26	Spastic Paraplegia	AR	99.95	48 of 48
ZIC2	Holoprosencephaly	AD	84.47	86 of 112

*Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial.
**Number of clinically relevant mutations according to HGMD

Methodology



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