Epileptic Encephalopathy and Early Infantile Epileptic Encephalopathy Precision Panel

Overview

Epileptic Encephalopathy and Early Infantile Epileptic Encephalopathy (EIEE) describes a clinical and genetic heterogeneous group of epilepsy syndromes associated with severe cognitive and behavioral abnormalities. Clinically these disorders vary in their age of onset, developmental outcome, etiologies, neuropsychological deficits, seizure types and prognosis. The difference between these two entities relies in the age of onset, EIEE manifests in the first year of life. Identifiable factors that may influence the course and degree of cognitive and behavioral impairment in these disorders include underlying etiology, age of onset, seizure frequency and severity, cumulative detrimental effect and genetic factors. Genetically these disorders can be caused by de novo mutations, but they can also be inherited in an autosomal dominant, recessive or X-linked pattern.

The Igenomix Epileptic Encephalopathy and Early Infantile Epileptic Encephalopathy (EIEE) Precision Panel can serve as an accurate and directed diagnostic tool as well as for a differential diagnosis for early onset epilepsy 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 Epileptic Encephalopathy and Early Infantile Epileptic Encephalopathy (EIEE) Precision Panel is indicated in patients with a clinical suspicion or diagnosis presenting with the following manifestations:

- Early-onset seizures
- Metabolic abnormalities
- Myoclonic and partial motor seizures
- Infantile spasms

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 care with antiepileptic drugs, corticosteroids, vigilance and monitorization of cognitive status as well as surgical care if needed.
- 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
ACADS	Acyl-Coa Dehydrogenase	AR	100	84 of 84
ACTL6B	Epileptic Encephalopathy, Intellectual Developmental Disorder, Intellectual Disability	AD,AR	100	21 of 21
ACY1	Aminoacylase 1 Deficiency	AR	100	15 of 15
ADAM22	Epileptic Encephalopathy	AR	99.98	4 of 4
ADGRG1	Polymicrogyria	AR	100	NA of NA
ADGRV1	Febrile Convulsions, Usher Syndrome, Generalized Epilepsy	AD,AR	97.53	NA of NA
ADSL	Adenylosuccinase Deficiency	AR	100	59 of 59
ALDH5A1	Succinic Semialdehyde Dehydrogenase Deficiency	AR	95.41	65 of 69
ALDH7A1	Epilepsy	AR	99.98	131 of 134
ALG13	Epileptic Encephalopathy	X,XR,XD,G	99.62	-
AP2M1	Epilepsy	AD	100	1 of 1
AP3B2	Epileptic Encephalopathy	AR	99.95	11 of 12
ARFGEF2	Periventricular Nodular Heterotopia	AR	100	15 of 15
ARHGEF15	Angelman Syndrome, Epileptic Encephalopathy	-	99.89	3 of 3
ARHGEF9	Hyperekplexia And Epilepsy	X,XR,G	100	-
ARV1	Epileptic Encephalopathy	AR	100	3 of 3
ARX	Corpus Callosum, Epileptic Encephalopathy, Lissencephaly, Partington Syndrome, West Syndrome	X,XR,G	81.92	-
ASNS	Asparagine Synthetase Deficiency	AR	99.98	37 of 37
ASPM	Microcephaly	AR	99.74	221 of 222
ATP1A2	Hemiplegia Of Childhood, Migraine	AD	100	108 of 108
ATP1A3	Hemiplegia Of Childhood, Cerebellar Ataxia And Sensorineuralhearing Loss, Dystonia	AD	99.94	138 of 138
ATP6AP2	Congenital Disorder Of Glycosylation, Mental Retardation, Parkinsonism	X,XR,G	100	-
ATRX	Alpha-Thalassemia Myelodysplasia Syndrome, Mental Retardation-Hypotonic Facies Syndrome, Carpenter-Waziri Syndrome, Chudley-Lowry-Hoar Syndrome, Holmes-Gang Syndrome, Juberg-Marsidi Syndrome, Neuroendocrine Tumor Of Stomach, Smith-Fineman-Myers Syndrome	X,XR,XD,G	98.5	-
BCKDK	Branched-Chain Ketoacid Dehydrogenase Kinase Deficiency	-	99.91	6 of 6
BRAT1	Neurodevelopmental Disorder With Cerebellar Atrophy, Rigidity And Multifocal Seizure Syndrome	AR	99.95	29 of 29
CACNA1A	Epileptic Encephalopathy, Ataxia, Migraine, Paroxysmal Torticollis Of Infancy	AD	96.13	249 of 266
CACNA1E	Epileptic Encephalopathy	AD	99.94	25 of 25
CACNA2D2	Cerebellar Atrophy With Seizures And Variable Developmental Delay	AR	94	10 of 10
CACNB4	Epilepsy, Episodic Ataxia	AD	99.87	5 of 5
CASK	Anemia, Fg Syndrome 4, Mental Retardation And Microcephaly, Epileptic Encephalopathy	X,XR,XD,G	99.98	-
CASR	Hyperparathyroidism, Hypocalcemia, Hypocalciuric Hypercalcemia, Hereditary Chronic Pancreatitis	AD,AR	100	445 of 446
CBL	Juvenile Myelomonocytic Leukemia, Noonan Syndrome, Juvenile Myelomonocyticleukemia, Mastocytosis	AD	100	46 of 47

CDH13	Attention Deficit-Hyperactivity Disorder, Seminoma	-	99.94	5 of 5
CDH2	Agenesis Of Corpus Callosum, Arrhythmogenic Right Ventricular Dysplasia	AD	99.98	16 of 16
CDKL5	Epileptic Encephalopathy, Atypical Rett Syndrome, West Syndrome	X,XD,G	99.92	-
CENPJ	Microcephaly, Seckel Syndrome	AR	99.97	13 of 13
CHD2	Epileptic Encephalopathy, Lennox-Gastaut Syndrome	AD	98.91	103 of 103
CHRM3	Prune Belly Syndrome	AR	99.8	4 of 4
CHRNA2	Epilepsy	AD	99.91	8 of 8
CHRNA4	Epilepsy	AD	99.8	24 of 24
CHRNA7	15q13.3 Microdeletion Syndrome	AD	82.09	2 of 2
CHRNB2	Epilepsy	AD	100	13 of 13
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, Epilepsy-Intellectual Disability Syndrome	AR	100	44 of 45
CLTC	Mental Retardation, Epileptic Encephalopathy	AD	98.81	14 of 14
CNPY3	Epileptic Encephalopathy, West Syndrome	AR	100	5 of 5
CNTN5	Coffin-Siris Syndrome, 3pter-P25 Deletion Syndrome, Myopathy, Cyclothymic Disorder	-	99.69	3 of 3
CNTNAP2	Pitt-Hopkins-Like Syndrome	AR	99.91	39 of 41
COQ4	Coenzyme Q10 Deficiency	AR	91.05	21 of 21
CPA6	Epilepsy, Febrile Seizures	AD,AR	99.97	9 of 9
CPLX1	Epileptic Encephalopathy, Wolf-Hirschhorn Syndrome	AD,AR	99.81	3 of 3
CSTB	Autosomal Recessive Hypohidrotic Ectodermal Dysplasia, Unverricht-Lundborg Disease	AR	100	14 of 14
CTSD	Ceroid Lipofuscinosis	AR	100	18 of 18
CTSF	Ceroid Lipofuscinosis	AR	92.18	12 of 12
CUX2	Epileptic Encephalopathy, Lennox-Gastaut Syndrome	AD	99.72	2 of 2
CYFIP2	Epileptic Encephalopathy	AD	100	8 of 8
CYTB	Leber Optic Atrophy, Mitochondrial Myopathy, Histiocytoid Cardiomyopathy	MI	98.8	-
DCX	Lissencephaly	X,G	100	-
DDX3X	Intellectual Developmental Disorder	X,XR,XD,G	99.03	-
DENND5A	Epileptic Encephalopathy	AR	100	9 of 9
DEPDC5	Epilepsy	AD	100	127 of 127
DHDDS	Developmental Delay And Seizures, Retinitis Pigmentosa, Epileptic Encephalopathy	AD,AR	96.32	8 of 8
DNAJC5	Ceroid Lipofuscinosis	AD	100	2 of 2
DNAJC6	Parkinson Disease	AR	99.86	13 of 14
DNM1	Epileptic Encephalopathy, Lennox-Gastaut Syndrome	AD	94.8	30 of 30
DOCK7	Epileptic Encephalopathy	AR	99.95	11 of 11
DOK5	Malignant Pheochromocythoma, Scleroderma	-	100	-
DYRK1A	Mental Retardation	AD	99.85	78 of 81
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

EIF2B5	Leukoencephalopathy	AR	100	99 of 99
EPM2A	Lafora Disease	AR	89.2	63 of 70
ETHE1	Ethylmalonic Encephalopathy	AR	100	32 of 33
FARS2	Oxidative Phosphorylation Deficiency, Spastic Paraplegia	AR	99.98	23 of 23
FASN	Fatty Liver Disease	-	100	6 of 6
FGF12	Epileptic Encephalopathy	AD	99.98	4 of 6
FLNA	Cardiac Valvular Dysplasia, Fg Syndrome, Frontometaphyseal Dysplasia, Heterotopia, Intestinal Pseudoobstruction, Otopalatodigital Syndrome, Terminal Osseous Dysplasia, Short Bowel Syndrome, Melnick-Needles 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
FOXP2	Speech-Language Disorder, Childhood Apraxia Of Speech	AD	100	17 of 17
FRRS1L	Epileptic Encephalopathy	AR	85.58	7 of 7
GABBR2	Epileptic Encephalopathy, Neurodevelopmental Disorder, Rett Syndrome	AD	95.98	7 of 7
GABRA1	Epileptic Encephalopathy, Dravet Syndrome	AD	100	45 of 46
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
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
GLDC	Glycine Encephalopathy	AR	98.69	359 of 367
GLS	Epileptic Encephalopathy, Global Developmental Delay, Infantile Cataract	AD,AR	97.77	8 of 9
GNAO1	Epileptic Encephalopathy, Neurodevelopmental Disorder With Involuntary Movements	AD	100	47 of 47
GOSR2	Epilepsy	AR	88.39	6 of 6
GPHN	Hyperekplexia, Molybdenum Cofactor Deficiency	AD,AR	99.2	6 of 6
GRIN1	Neurodevelopmental Disorder	AD,AR	100	43 of 43
GRIN2A	Epileptic Encephalopathy, Intellectual Disability, Rolandic Epilepsy	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
GUF1	Epileptic Encephalopathy, West Syndrome	AR	99.88	4 of 4
HCN1	Epileptic Encephalopathy	AD	98.43	42 of 43
HNRNPH1	Myasthenic Syndrome, Dermatopathia Pigmentosa Reticularis	-	100	2 of 2
HNRNPU	Epileptic Encephalopathy, 1q44 Microdeletion Syndrome	AD	99.8	36 of 36
HSPG2	Dyssegmental Dysplasia, Schwartz-Jampel Syndrome	AR	99.41	68 of 69
IER3IP1	Microcephaly	AR	99.97	5 of 5
IL27RA	Crisponi/Cold-Induced Sweating Syndrome	-	99.87	3 of 3
IQSEC1	Intellectual Developmental Disorder With Short Stature And Behavioral Abnormalities	AR	99.92	3 of 3
IQSEC2	Mental Retardation, Microduplication Xp11.22p11.23 Syndrome, Severe Intellectual Disability-Progressive Postnatal Microcephaly-Midline Stereotypic Hand Movements Syndrome, Smith-Magenis Syndrome	X,XR,XD,G	99.73	-
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
KCNB1	Epileptic Encephalopathy	AD	99.95	55 of 55
KCND2	Autism, Epileptic Encephalopathy	-	100	4 of 4
KCNH1	Temple-Baraitser Syndrome, Zimmermann-Laband Syndrome	AD	99.69	15 of 15
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, Dend Syndrome	AD,AR	100	190 of 191
KCNMA1	Cerebellar Atrophy, Liang-Wang Syndrome, Generalized Epilepsy-Paroxysmal Dyskinesia 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
KCNT1	Epilepsy	AD	95.98	64 of 64
KCTD7	Epilepsy	AR	99.99	40 of 40
KDM6A	Kabuki Syndrome	AD,X,XD,G	99.98	-
LAMA2	Limb-Girdle Muscular Dystrophy	AR	100	363 of 377
LGI1	Epilepsy	AD	99.94	54 of 54
LIAS	Pyruvate Dehydrogenase Lipoic Acid Synthetase Deficiency	AR	99.82	8 of 8
MAGI2	Nephrotic Syndrome	AR	93.82	7 of 9
MBD5	Mental Retardation, 2q23.1 Microdeletion Syndrome	AD	99.99	33 of 35
MCPH1	Microcephaly	AR	99.51	18 of 19
MDGA2	Autism, Ring Dermoid Of Cornea	-	99.89	0 of 1
MECP2	Autism, Encephalopathy, Mental Retardation, Rett Syndrome, Trisomy Xq28	X,XR,XD,MU,G	99.81	-
MEF2C	Mental Retardation, Epilepsy, 5q14.3 Microdeletion Syndrome	AD	99.91	43 of 46
MFSD8	Ceroid Lipofuscinosis, Macular Dystrophy With Central Cone Involvement	AR	100	63 of 63
MTHFR	Homocystinuria, Neural Tube Defects, Schizophrenia, Thrombophiliavenous Thromboembolism, Isolated Anencephaly And Exencephaly	AD,AR	100	122 of 122
MTOR	Focal Cortical Dysplasia Of Taylor, Smith-Kingsmore Syndrome, Macrocephaly-Intellectual Disability-Neurodevelopmental Disorder-Small Thorax Syndrome	AD	99.98	39 of 39
MYO9B	Colitis, Celiac Disease, Inflammatory Bowel Disease	-	97.93	5 of 5
NALCN	Congenital Contractures Of The Limbs And Face, Hypotonia, Digitotalar Dysmorphism, Freeman-Sheldon Syndrome, Sheldon-Hall Syndrome	AD,AR	99.97	69 of 69
NDE1	Lissencephaly 4, Microhydranencephaly , Hydranencephaly	AR	86.55	12 of 13
NDUFA1	Complex I Deficiency	X,XR,G	100	-
NECAP1	Epileptic Encephalopathy	AR	99.83	2 of 2
NEDD4L	Nodular Heterotopia	AD	97.61	10 of 10
NEXMIF	Mental Retardation	X,XR,XD,G	99.74	-
NGLY1	Congenital Disorder Of Glycosylation, Alacrimia-Choreoathetosis-Liver Dysfunction Syndrome	AR	99.8	28 of 28
NHLRC1	Lafora Disease	AR	100	71 of 71
NPC1L1	Niemann-Pick Disease, Sitosterolemia	-	100	31 of 31
NPRL2	Epilepsy	AD	100	12 of 12
NPRL3	Epilepsy	AD	99.61	18 of 18
NR2F1	Bosch-Boonstra Optic Atrophy Syndrome, Optic Atrophy-Intellectual Disability Syndrome	AD	89.78	26 of 31

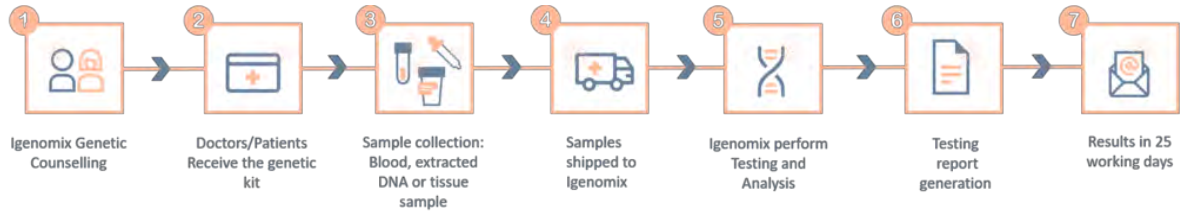
NRXN1	Pitt-Hopkins-Like Syndrome	AR	97.42	33 of 74
OPHN1	Mental Retardation	X,XR,G	100	-
PACS1	Intellectual Disability-Craniofacial Dysmorphism-Cryptorchidism Syndrome	AD	97.98	3 of 3
PAFAH1B1	Lissencephaly, 17p13.3 Microduplication Syndrome, Miller-Dieker Syndrome	AD	99.95	90 of 92
PCDH19	Epilepsy, Dravet Syndrome	X,G	99.99	-
PHF6	Borjeson-Forssman-Lehmann Syndrome	X,XR,G	99.93	-
PIGA	Multiple Congenital Anomalies-Hypotonia-Seizures Syndrome, Paroxysmal Nocturnal Hemoglobinuria, West Syndrome	X,XR,G	97.98	-
PIGN	Multiple Congenital Anomalies-Hypotonia-Seizures Syndrome, Fryns Syndrome	AR	93.97	36 of 39
PIGO	Hyperphosphatasia-Intellectual Disability Syndrome	AR	99.93	21 of 21
PIGV	Hyperphosphatasia-Intellectual Disability Syndrome	AR	99.99	16 of 16
PIK3AP1	Byssinosis, Central Nervous System Tuberculosis	-	99.98	5 of 5
PLCB1	Epileptic Encephalopathy,West Syndrome	AR	99.92	4 of 6
PNKP	Ataxia-Oculomotor Apraxia, Charcot-Marie-Tooth Disease, Epileptic Encephalopathy	AR	100	36 of 36
PNPO	Pyridoxamine 5-Prime-Phosphate Oxidase Deficiency	AR	99.99	31 of 31
POLG	DNA Depletion Syndrome, Sensory Ataxic Neuropathy, Alpers-Huttenlocher Syndrome, Ophthalmoplegia, Neurogastrointestinal Encephalomyopathy	AD,AR	99.92	325 of 326
POLG2	DNA Depletion Syndrome, Ophthalmoplegia	AD,AR	99.97	13 of 13
PPP2R5D	Mental Retardation, Intellectual Disability-Macrocephaly-Hypotonia-Behavioral Abnormalities Syndrome	AD	100	11 of 11
PPT1	Ceroid Lipofuscinosis	AR	100	81 of 81
PRICKLE1	Epilepsy, Unverricht-Lundborg Disease	AR	98.41	23 of 23
PRICKLE2	Epilepsy, Neural Tube Defects, Sensory Ataxic Neuropathy	-	94.92	6 of 6
PRRT2	Kinesigenic Dyskinesia, Epilepsy, Hemiplegic Migraine, Convulsions And Choreoathetosis	AD	99.93	111 of 111
PTEN	Cowden Disease, Macrocephaly/Autism Syndrome, Meningioma, Bannayan-Riley-Ruvalcaba Syndrome, Breast And Ovarian Cancer Syndrome, Polyposis Of Infancy, Lhermitte-Duclos Disease, Proteus Syndrome,Segmental Outgrowth-Lipomatosis-Arteriovenous Malformation-Epidermal Nevus Syndrome	AD	99.97	609 of 629
PURA	Mental Retardation	AD	85.36	59 of 65
QARS	Microcephaly, Strabismus	-	100	12 of 12
QARS1	Microcephaly With Seizures	AR	-	-
RANBP2	Necrotizing Encephalopathy	AD	99.41	9 of 9
RANGAP1	Parkinson Disease, Dermatopathia Pigmentosa Reticularis	-	99.98	2 of 2
RBFOX1	Arial Septal Defect, Spinocerebellar Ataxia, Epilepsy	-	97.99	4 of 5
RELN	Epilepsy, Lissencephaly	AD,AR	100	70 of 70
ROGDI	Kohlschutter-Tonz Syndrome, Amelocerebrohypohidrotic Syndrome	AR	99.83	10 of 12
RYR3	Central Core Myopathy, Neuroleptic Malignant Syndrome, Capillary Malformations	-	99.98	20 of 20
SCARB2	Action Myoclonus-Renal Failure Syndrome, Gaucher Disease, Unverricht-Lundborg Disease	AR	99.95	29 of 29
SCN1A	Febrile Convulsions, Migraine, Dravet Syndrome, Hemiplegic Migraine, Epilepsy, Lennox-Gastaut Syndrome	AD	99.8	1776 of 1797
SCN1B	Atrial Fibrillation, Brugada Syndrome, Epileptic Encephalopathy, Dravet Syndrome, 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
SCN8A	Cognitive Impairment, Epileptic Encephalopathy, Myoclonus, Seizures	AD	97.85	156 of 172

SCN9A	Erythermalgia, Generalized Epilepsy With Febrile Seizures, Indifference To Pain, Neuropathy, Paroxysmal Extreme Pain Disorder, Dravet Syndrome, Hereditary Sensory And Autonomic Neuropathy, Paroxysmal Extreme Pain Disorder	AD,AR	96.25	126 of 137
SEMA5B	Diffuse Glomerulonephritis, Kallmann Syndrome	-	99.91	1 of 1
SHANK3	Phelan-Mcdermid Syndrome, Schizophrenia, Monosomy 22q13.3	AD,MU,P	96.67	-
SIK1	Epileptic Encephalopathy, West Syndrome	AD	99.67	9 of 9
SLC12A5	Epileptic Encephalopathy	AD,AR	100	19 of 19
SLC13A5	Epileptic Encephalopathy, Amelocerebrohypohidrotic Syndrome	AR	95.92	24 of 24
SLC19A3	Basal Ganglia Disease, Leigh Syndrome With Leukodystrophy	AR	100	38 of 39
SLC25A12	Epileptic Encephalopathy	AR	100	7 of 7
SLC25A19	Microcephaly, Thiamine Metabolism Dysfunction Syndrome, Amish Lethal Microcephaly	AR	97.13	10 of 10
SLC25A22	Epileptic Encephalopath	AR	100	16 of 16
SLC26A1	Nephrolithiasis	AR	99.94	4 of 4
SLC2A1	Choreoathetosis/Spasticity, Epilepsy, Glucose Transport Defect, Stomatin-Deficient Cryohydrocytosis With Neurologic Defects, Exertion-Induced Dyskinesia	AD,AR	99.99	301 of 304
SLC35A2	Congenital Disorder Of Glycosylation	X,XD,G	99.97	-
SLC6A1	Myoclonic-Astastic Epilepsy	AD	100	55 of 55
SLC6A8	Creatine Deficiency Syndrome	X,XR,G	99.87	-
SLC9A6	Christianson Syndrome	X,XD,G	98.87	-
SMC1A	Cornelia De Lange Syndrome, Semilobar Holoprosencephaly, Wiedemann-Steiner Syndrome	X,XR,XD,G	100	-
SPATA5	Epilepsy, Hearing Loss, Mental Retardation	AR	99.83	30 of 30
SPTAN1	Epileptic Encephalopathy, West Syndrome	AD	100	52 of 53
SQSTM1	Frontotemporal Dementia, Myopathy, Neurodegeneration With Ataxia, Dystonia, Paget Disease Of Bone, Amyotrophic Lateral Sclerosis	AD,AR	99.25	105 of 107
SRPX2	Mental Retardation, Speech Dyspraxia, Bilateral Perisylvian Polymicrogyria, Rolandic Epilepsy	AD	100	-
ST3GAL3	Epileptic Encephalopathy, Mental Retardation, West Syndrome	AR	100	5 of 5
ST3GAL5	Amish Infantile Epilepsy Syndrome	AR	99.17	6 of 6
STIL	Microcephaly	AR	99.94	18 of 18
STX1B	Generalized Epilepsy With 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
SYN1	Epilepsy With Variable Learning Disabilities And Behavior Disorders	X,XR,XD,G	91.7	-
SYNGAP1	Mental Retardation, Developmental And Epileptic Encephalopathy	AD	99.46	168 of 171
SYT2	Myasthenic Syndrome With Or Without Motorneuropathy	AD	99.98	4 of 4
SZT2	Epileptic Encephalopathy	AR	99.98	39 of 39
TBC1D24	Deafness, Doors Syndrome, Epileptic Encephalopathy	AD,AR	100	80 of 80
TBL1XR1	Mental Retardation, Pierpont Syndrome, Acute Promyelocytic Leukemia	AD	99.78	23 of 23
TCF4	Corneal Dystrophy, Pitt-Hopkins Syndrome, Primary Sclerosing Cholangitis	AD	98.91	124 of 124
TNK2	Epilepsy With Variable Learning Disabilities,	-	99.72	6 of 7
TPP1	Ceroid Lipofuscinosis, Spinocerebellar Ataxia	AR	100	147 of 147
TSC1	Dysplasia Of Taylor, Tuberous Sclerosis, Lymphangioleiomyomatosis	AD	99.86	390 of 406
TSC2	Dysplasia Of Taylor, Lymphangioleiomyomatosis, Tuberous Sclerosis	AD	100	1157 of 1159
TSEN54	Encephalopathy, Pontocerebellar Hypoplasia	AR	96.94	20 of 22

UBE3A	Angelman Syndrome, 15q11q13 Microduplication Syndrome	AD	99.98	208 of 211
VRK2	Fanconi Anemia, Pontocerebellar Hypoplasia, Epileptic Encephalopathy	-	99.77	3 of 3
WDR45	Neurodegeneration With Brain Iron Accumulation, West Syndrome	X,XD,G	100	-
WDR62	Microcephaly	AR	100	60 of 61
WWOX	Epileptic Encephalopathy, Esophageal Cancer, Spinocerebellar Ataxia, Gonadal Dysgenesis, Cerebellar Ataxia	AR	99.94	44 of 44
ZEB2	Mowat-Wilson Syndrome	AD	98.95	253 of 254

*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



Contact us

Call +34 963 905 310 or send an email to supportspain@igenomix.com for any of the following objectives:

- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

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