Lissencephaly

Precision Panel

Overview

Lissencephaly, also known as smooth brain, is a malformation of the cerebral cortex associated with an abnormal neuronal migration and development of cerebral convolutions or gyri. There can be absent gyri (agyria) or abnormally wide gyri (pachygyria) alongside abnormally thick and poorly organized cortex, diffuse neuronal heterotopia, dysmorphic ventricles and often failure of the corpus callosum to develop. Lissencephaly has been associated with several syndromes and so genetic factors play an important role in its etiology. It is a significant cause of neurological morbidity in children worldwide, responsible for many cases of mental retardation, cerebral palsy, and epilepsy. It is inherited in an autosomal recessive pattern in its majority, although there are forms that are inherited in an autosomal dominant and X-linked fashion.

The Igenomix Lissencephaly Precision Panel can be used to make a directed and accurate diagnosis 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 Lissencephaly Precision Panel is indicated for those patients with head imaging findings (ultrasound, computed tomogram (CT), magnetic resonance imaging (MRI)) suggestive of lissencephaly or with the following manifestations:

- Unusual facial appearance
- Difficulty swallowing
- Failure to thrive
- Muscle spasms
- Seizures
- Severe psychomotor retardation
- Deformed hands, feet or toes
- Microcephaly (small head size)

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.

- Early initiation of symptomatic and supportive treatment in the form early referral to a high-risk center, multidisciplinary counselling and close coordination between pediatrics, neurologists and other specialists.
- Risk assessment of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.
- Identification of the genetic basis of these associated disorders for a better insight into the mechanisms of brain development.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ACTB	Baraitser-Winter Syndrome, Juvenile-Onset Dystonia, Developmental Malformations-Deafness-Dystonia Syndrome	AD	100	40 of 40
ACTG1	Baraitser-Winter Syndrome	AD	98.59	55 of 55
ADAMTS3	Hennekam Lymphangiectasia-Lymphedema Syndrome, Hennekam Syndrome	AR	99.97	4 of 4
ADGRG1	Bilateral Frontoparietal Polymicrogyria	AR	100	NA of NA
ANKLE2	Autosomal Recessive Primary Microcephaly	AR	96.08	4 of 4
APC2	Complex Cortical Dysplasia With Other Brain Malformations, Sotos Syndrome	AR	94.97	11 of 11
ARHGAP31	Adams-Oliver Syndrome	AD	100	6 of 6
ARX	Agenesis of Corpus Callosum With Abnormal Genitalia , Early Infantile Epileptic Encephalopathy, Lissencephaly, X-linked Mental Retardation With Or Without Seizures, Partington Syndrome, West Syndrome, X-linked Spasticity-Intellectual Disability-Epilepsy Syndrome	X,XR,G	81.92	NA of NA
ASPM	Autosomal Recessive Primary Microcephaly	AR	99.74	221 of 222
ATP6V0A2	Autosomal Recessive Cutis Laxa Type II, Wrinkly Skin Syndrome	AR	99.99	55 of 55
ATP6V1A	Autosomal Recessive Cutis Laxa Type II, Undetermined Early-Onset Epileptic Encephalopathy	AD,AR	99.98	9 of 9
ATP6V1E1	Autosomal Recessive Cutis Laxa Type II	AR	100	2 of 2
ATR	Familial Cutaneous Telangiectasia And Cancer Syndrome, Seckel Syndrome	AD,AR	99.98	39 of 40
B3GALNT2	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Type A, Autosomal Recessive Non-Syndromic Intellectual Disability , Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	97.14	17 of 17
B4GAT1	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Type A, Walker-Warburg Syndrome	AR	na	na
CASK	Nonspherocytic Hemolytic Anemia Due To G6PD Deficiency, X-linked Mental Retardation With Or Without Nystagmus, Mental Retardation And Microcephaly With Pontine And Cerebellar Hypoplasia, Early Infantile Epileptic Encephalopathy	X,XR,XD,G	99.98	NA of NA
CCBE1	Hennekam Lymphangiectasia-Lymphedema Syndrome, Hennekam Syndrome	AR	100	16 of 16
CCDC88A	Peho-like Syndrome	AR	91.9	3 of 4
CDK5	Lissencephaly With Cerebellar Hypoplasia	AR	100	5 of 5
CDK5RAP2	Autosomal Recessive Primary Microcephaly	AR	100	32 of 32
CDK6	Autosomal Recessive Primary Microcephaly	AR	100	1 of 1
CDKL5	Early Epileptic Epileptic Encephalopathy, Atypical Rett Syndrome, West Syndrome	X,XD,G	99.92	NA of NA
CENPJ	Autosomal Recessive Primary Microcephaly, Seckel Syndrome	AR	99.97	13 of 13
CEP135	Autosomal Recessive Primary Microcephaly	AR	99.48	7 of 8
CEP152	Autosomal Recessive Primary Microcephaly, Seckel Syndrome	AR	97.73	21 of 24
CEP63	Autosomal Recessive Primary Microcephaly, Seckel Sydrome	AR	100	3 of 3
CEP85L	Lissencephaly	AD	99.73	1 of 1
CIT	Autosomal Recessive Primary Microcephaly	AR	99.98	17 of 17
COL4A1	Hereditary Angiopathy With Nephropathy, Aneurysms, And Muscle Cramps, Autosomal Dominant Pontine Microangiopathy And Leukoencephalopathy, Hanac Syndrome, Walker-Warburg Syndrome	AD	99.99	173 of 173
COPB2	Autosomal Recessive Primary Microcephaly	AR	99.64	4 of 4

CPT2	Carnitine Palmitoyltransferase II Deficiency	AD,AR	99.99	116 of 116
CRADD	Autosomal Recessive Mental Retardation With Variant Lissencephaly	AR	99.62	6 of 7
CRPPA	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Type A and C, Congenital Muscular Dystrophy Without Intellectual Disability, ISPD-related Limb-Girdle Muscular Dystrophy, Walker-Warburg Syndrome	AR	97.69	NA of NA
CSNK2A1	Okur-Chung Neurodevelopmental Syndrome	AD	99.95	23 of 23
CTNNA2	Complex Cortical Dysplasia With Other Brain Malformations	AR	99.95	8 of 8
CTU2	Microcephaly, Facial Dysmorphism, Renal Agenesis, And Ambiguous Genitalia Syndrome	AR	99.93	6 of 6
DAG1	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Type A and C, Alpha-Dystroglycan-Related Limb-Girdle Muscular Dystrophy, Muscle-Eye-Brain Disease With Bilateral Multicystic Leucodystrophy, Walker-Warburg Syndrome	AR	99.98	9 of 9
DCHS1	Van Maldergem Syndrome, Cerebrofacioarticular Syndrome	AD,AR	99.69	30 of 30
DCX	X-linked Lissencephaly	X,G	100	NA of NA
DHCR24	Desmosterolosis	AR	100	10 of 10
DMXL2	Early Infantile Epileptic Encephalopathy, Polyendocrine-Polyneuropathy Syndrome	AD,AR	99.83	19 of 23
DYNC1H1	Charcot-Marie-Tooth Disease, Axonal, Type 2o, Autosomal Dominant Mental Retardation, Spinal Muscular Atrophy	AD	100	104 of 104
EML1	Band Heterotopia	AR	98.88	7 of 7
ETFA	Multiple Acyl-CoA Dehydrogenase Deficiency	AR	92.33	32 of 32
ETFB	Multiple Acyl-CoA Dehydrogenase Deficiency	AR	100	21 of 21
ETFDH	Multiple Acyl-CoA Dehydrogenase Deficiency	AR	100	221 of 222
FAT4	Hennekam Lymphangiectasia-Lymphedema Syndrome, Van Maldergem Syndrome, Cerebrofacioarticular Syndrome, Hennekam Syndrome	AR	99.8	41 of 41
FIG4	Amyotrophic Lateral Sclerosis, Charcot-Marie-Tooth Disease, Cleidocranial Dysplasia With Micrognathia, Absent Thumbs, And Distal, Polymicrogyria, Bilateral Temporooccipital, Amyotrophic Lateral Sclerosis, Bilateral Parasagittal Parieto-occipital Polymicrogyria, Yunis-Varon Syndrome	AD,AR	99.92	72 of 72
FKRP	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Type A and C, Congenital Muscular Dystrophy With Cerebellar Involvement, Congenital Muscular Dystrophy With Intellectual Disability, Congenital Muscular Dystrophy Without Intellectual Disability, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	99.9	157 of 157
FKTN	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Type A and B, Limb-Girdle Muscular Dystrophy Type 2m, Congenital Muscular Dystrophy Without Intellectual Disability, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	98	54 of 56
FLI1	Platelet-Type Bleeding Disorder, Jacobsen Syndrome, Paris-Trousseau Thrombocytopenia, Peripheral Primitive Neuroectodermal Tumor	AD,AR	100	7 of 7
FOXG1	Rett Syndrome, 14q12 Microdeletion Syndrome, FOXG1 Syndrome	AD	88.71	93 of 109
FTO	Growth Retardation, Developmental Delay, and Coarse Facies	AR	99.91	8 of 8
GFM2	Combined Oxidative Phosphorylation Deficiency Type 39	AR	99.35	5 of 7
GMPPB	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Type A and B, Limb-Girdle Muscular Dystrophy-Dystroglycanopathy Type C, Congenital Muscular Dystrophy With Cerebellar Involvement, Congenital Muscular Dystrophy With Intellectual Disability, Congenital Myasthenic Syndromes With Glycosylation Defect, GMPPB-Related Limb-Girdle Muscular Dystrophy, Muscle-Eye-Brain Disease	AR	99.95	53 of 53
GNAO1	Early Infnatile Epileptic Encephalopathy, Neurodevelopmental Disorder With Involuntary Movements	AD	100	47 of 47
GPX4	Spondylometaphyseal Dysplasia	AR	79.72	3 of 3
HIC1	Miller-Dieker Syndrome	-	97.7	NA of NA
ISCA1	Multiple Mitochondrial Dysfunctions Syndrome	AR	99.86	2 of 2
KATNB1	Lissencephaly With Microcephaly	AR	100	10 of 10
KCNA1	Episodic Ataxia Type 1, Early Infantile Epileptic Encephalopathy, Hereditary Continuous Muscle Fiber Activity, Paroxysmal Kinesigenic Dyskinesia	AD	100	49 of 49
KIAA1109	Alkuraya-Kucinskas Syndrome	AR	99.95	21 of 21
KIF14	Meckel Syndrome, Autosomal Recessive Primary Microcephaly	AR	99.84	18 of 18
KIF2A	Complex Cortical Dysplasia With Other Brain Malformations	AD	99.91	7 of 7
KIFBP	Goldberg-Shprintzen Syndrome	AR	99.27	NA of NA

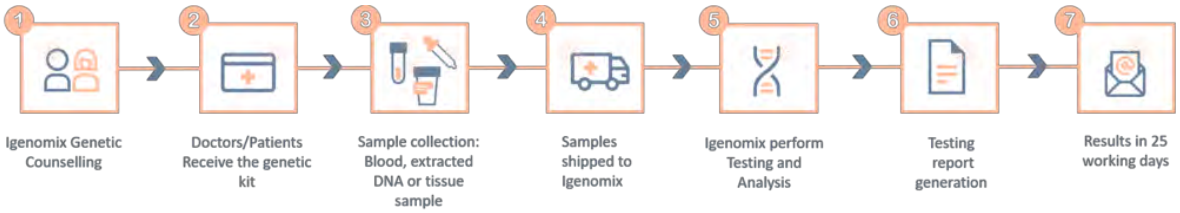
KNL1	Autosomal Recessive Primary Microcephaly	AR	98.91	NA of NA
LAGE3	Galloway-Mowat Syndrome	X,XR,G	91.36	NA of NA
LAMA2	Muscular Dystrophy Congenital Merosin-Deficient, Limb-Girdle Muscular Dystrophy, Laminin Subunit Alpha 2-Related Congenital Muscular Dystrophy	AR	100	363 of 377
LAMB1	Lissencephaly, Cobblestone Lissencephaly Without Muscular Or Ocular Involvement	AR	99.97	8 of 9
LAMC3	Occipital Cortical Malformations	AR	98.72	22 of 22
LARGE1	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Type A, Congenital Muscular Dystrophy Type 1d, Congenital Muscular Dystrophy With Intellectual Disability, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	100	NA of NA
MACF1	Lissencephaly With Complex Brainstem Malformation	AD	99.94	18 of 18
MCPH1	Autosomal Recessive Primary Microcephaly	AR	99.51	18 of 19
METTL5	Intellectual Developmental Disorder, Autosomal Recessive Primary Microcephaly	AR	99.9	4 of 4
MFSD2A	Autosomal Recessive Primary Microcephaly	AR	97.58	6 of 6
MLYCD	Malonyl-CoA Decarboxylase Deficiency	AR	93.84	32 of 40
MPDZ	Autosomal Recessive Nonsyndromic Hydrocephalus	AR	99.44	58 of 58
NCAPD3	Autosomal Recessive Primary Microcephaly	AR	99.97	4 of 5
NDE1	Lissencephaly, Microhydranencephaly, Hydranencephaly	AR	86.55	12 of 13
NEK1	Amyotrophic Lateral Sclerosis, Short Rib-Polydactyly Syndrome Type II, Amyotrophic Lateral Sclerosis, Orofaciodigital Syndrome Type II	AD,AR,MU,D	99.83	73 of 74
NEUROD2	Early Infantile Epileptic Encephalopathy	AD	96.88	2 of 2
NSDHL	Congenital Hemidysplasia With Ichthyosiform Erythroderma And Limb, Ck Syndrome	X,XR,XD,G	100	NA of NA
NUP107	Galloway-Mowat Syndrome, Nephrotic Syndrome	AR	99.91	15 of 15
NUP133	Galloway-Mowat Syndrome, Nephrotic Syndrome	AR	99.94	6 of 6
OCLN	Pseudo-Torch Syndrome, Congenital Intrauterine Infection-like Syndrome	AR	86.89	15 of 17
OSGEP	Galloway-Mowat Syndrome	AR	99.17	19 of 19
PAFAH1B1	Lissencephaly, 17p13.3 Microduplication Syndrome, Miller-Dieker Syndrome	AD	99.95	90 of 92
PEX10	Peroxisome Biogenesis Disorder (Zellweger), Autosomal Recessive Ataxia Due To Pex10 Deficiency, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy, Zellweger Syndrome	AR	99.76	29 of 32
PEX13	Peroxisome Biogenesis Disorder (Zellweger), Infantile Refsum Disease, Neonatal Adrenoleukodystrophy, Zellweger Syndrome	AR	99.98	11 of 12
PHC1	Autosomal Recessive Primary Microcephaly	AR	91.73	1 of 1
PHGDH	Neu-Laxova Syndrome, Phosphoglycerate Dehydrogenase Deficiency, 3-Phosphoglycerate Dehydrogenase Deficiency, Infantile/Juvenile Form, Neu-laxova Syndrome	AR	100	26 of 26
PI4KA	Presylvian Polymicrogyria With Cerebellar Hypoplasia And Arthrogryposis, Bilateral Perisylvian Polymicrogyria	AR	99.76	4 of 4
PIGP	Early Infantile Epileptic Encephalopathy	AR	99.98	2 of 2
PIGQ	Early Infantile Epileptic Encephalopathy	AR	99.99	4 of 4
PIK3R2	Megalencephaly-Polymicrogyria-Polydactyly-Hydrocephalus Syndrome	AD	90.81	7 of 7
PNKP	Ataxia-Oculomotor Apraxia, Charcot-Marie-Tooth Disease, Early Infantile Epileptic Encephalopathy	AR	100	36 of 36
POMGNT1	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eye), Muscular Dystrophy-Dystroglycanopathy (Limb-Girdle), Type C, Retinitis Pigmentosa, Congenital Muscular Dystrophy With Cerebellar Involvement, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	99.91	82 of 83
POMGNT2	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Type A, Muscular Dystrophy-Dystroglycanopathy (Limb-Girdle), Type C, Walker-warburg Syndrome	AR	100	10 of 10
POMK	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Type A, Muscular Dystrophy-Dystroglycanopathy (Limb-Girdle), Type C, Congenital Muscular Dystrophy With Cerebellar Involvement, Walker-Warburg Syndrome	AR	99.99	8 of 8
POMT1	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Type A, Muscular Dystrophy-Dystroglycanopathy (Congenital With Mental Retardation), Type B, Limb-Girdle Muscular Dystrophy Type 2k, Congenital Muscular Dystrophy With Cerebellar Involvement, Congenital Muscular Dystrophy With Intellectual Disability, Muscle-Eye-Brain Disease , POMT1-Related Limb-girdle Muscular Dystrophy, Walker-Warburg Syndrome	AR	100	105 of 105

POMT2	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Type A, Muscular Dystrophy-Dystroglycanopathy (Congenital With Mental Retardation), Type B, Muscular Dystrophy-dystroglycanopathy (Limb-Girdle), Type C, Congenital Muscular Dystrophy With Cerebellar Involvement, Congenital Muscular Dystrophy With Intellectual Disability, Muscle-Eye-Brain Disease , POMT2-Related Limb-Girdle Muscular Dystrophy, Walker-Warburg Syndrome	AR	100	74 of 74
PRKDC	Immunodeficiency With Or Without Neurologic Abnormalities	AR	99.74	9 of 10
PSAT1	Neu-Laxova Syndrome, Phosphoserine Aminotransferase Deficiency	AR	99.95	9 of 9
PYCR2	Hypomyelinating Leukodystrophy, Autosomal Recessive Primary Microcephaly	AR	98.29	14 of 14
RAB18	Warburg Micro Syndrome	AR	100	4 of 4
RAB3GAP1	Warburg Micro Syndrome, Cataract-Intellectual Disability-Hypogonadism Syndrome	AR	99.94	70 of 70
RAB3GAP2	Martsolf Syndrome, Warburg Micro Syndrome, Autosomal Recessive Spastic Paraplegia Type 69, Cataract-Intellectual Disability-Hypogonadism Syndrome	AR	100	17 of 17
RELN	Familial Temporal Lobe Epilepsy, Lissencephaly	AD,AR	100	70 of 70
RMND1	Combined Oxidative Phosphorylation Deficiency	AR	99.67	15 of 16
RNU4ATAC	Lowry-Wood Syndrome, Microcephalic Osteodysplastic Primordial Dwarfism Type I, Roifman Syndrome	AR	na	na
RTTN	Microcephaly, Short Stature, And Polymicrogyria With Seizures	AR	99.94	28 of 29
RXYLT1	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Type A, Walker-Warburg Syndrome	AR	99.46	NA of NA
SASS6	Autosomal Recessive Primary Microcephaly	AR	99.14	6 of 6
SCN1B	Early Infantile Epileptic Encephalopathy, Generalized Epilepsy With Febrile Seizures Plus Type 1, Dravet Syndrome	AD,AR	99.67	46 of 48
SCN2A	Early Infantile Epileptic Encephalopathy, Episodic Ataxia Type 9, Benign Familial Neonatal-Infantile Seizures, Benign Familial Infantile Epilepsy, Dravet Syndrome, Generalized Epilepsy With Febrile Seizures-Plus, West Syndrome	AD	100	351 of 351
SIK1	Early Infantile Epileptic Encephalopathy, Early Myoclonic Encephalopathy, West Syndrome	AD	99.67	9 of 9
SLC25A19	Microcephaly, Thiamine Metabolism Dysfunction Syndrome (Bilateral Striatal Degenerationand Progressive Polyneuropathy Type), Amish Lethal Microcephaly	AR	97.13	10 of 10
SLC25A22	Early Infantile Epileptic Encephalopathy, Early Myoclonic Encephalopathy	AR	100	16 of 16
SNAP29	Cerebral Dysgenesis, Neuropathy, Ichthyosis, And Palmoplantar Keratoderma Syndrome, Cednik Syndrome	AR	100	13 of 13
SRPX2	Rolandic Epilepsy, Mental Retardation, And Speech Dyspraxia, X-linked, Bilateral Perisylvian Polymicrogyria, Rolandic Epilepsy-Speech Dyspraxia Syndrome	AD	100	NA of NA
STIL	Autosomal Recessive Primary Microcephaly	AR	99.94	18 of 18
STS	Recessive X-linked Ichthyosis	X,XR,G	100	NA of NA
STXBP1	9q33.3q34.11 Microdeletion Syndrome, Atypical Rett Syndrome, Dravet Syndrome, Early Infantile Epileptic Encephalopathy, West Syndrome	AD	100	209 of 215
TAF13	Autosomal Recessive Mental Retardation, Autosomal Recessive Primary Microcephaly	AR	99.97	5 of 5
TBC1D20	Warburg Micro Syndrome	AR	99.94	6 of 6
TBR1	Intellectual Developmental Disorder With Autism And Speech Delay, 2q24 Microdeletion Syndrome	AD	99.04	13 of 13
TCTN1	Joubert Syndrome	AR	94.98	10 of 10
TCTN2	Joubert Syndrome, Meckel Syndrome	AR	100	14 of 14
TMTC3	Lissencephaly, Periventricular Nodular Heterotopia	AR	99.04	10 of 10
TMX2	Neurodevelopmental Disorder With Microcephaly, Cortical Malformations, And Spasticity	AR	99.98	12 of 12
TP53RK	Galloway-Mowat Syndrome	AR	97.68	5 of 5
TPRKB	Galloway-Mowat Syndrome	AR	85.66	2 of 2
TRAPPC14	Autosomal Recessive Primary Microcephaly	AR	na	na
TRIM8	Early Infantile Epileptic Encephalopathy	-	99.5	7 of 7
TUBA1A	Lissencephaly	AD	100	95 of 95
TUBB2B	Complex Cortical Dysplasia, With Other Brain Malformations, Dysequilibrium Syndrome, Polymicrogyria Due To TUBB2B Mutation	AD	84.28	29 of 38
TUBB3	Complex Cortical Dysplasia, With Other Brain Malformations, Congenial Fibrosis Of Extraocular Muscles, Cortical Dysgenesis With Pontocerebellar Hypoplasia Due To TUBB3 Mutation	AD	99.96	30 of 30
TUBG1	Complex Cortical Dysplasia, With Other Brain Malformations	AD	99.94	10 of 10

TUBGCP2	Pachygyria, Microcephaly, Developmental Delay, And Dysmorphic Facies, With Or Without Seizures	AR	96.78	4 of 4
TUBGCP6	Microcephaly With Chorioretinopathy	AR	99.49	12 of 13
VAC14	Childhood-Onset Striatonigral Degeneration, Yunis-Varon Syndrome	AR	100	11 of 11
VIPAS39	Arthrogryposis, Renal Dysfunction, And Cholestasis	AR	100	15 of 15
VLDLR	Cerebellar Hypoplasia And Mental Retardation With Or Without Quadrupedal Locomotion, Dysequilibrium Syndrome	AR	100	20 of 20
VPS33B	Arthrogryposis, Renal Dysfunction, And Cholestasis	AR	100	62 of 62
WDR26	Skraban-Deardorff Syndrome, Intellectual Disability-Seizures-Abnormal Gait-Facial Dysmorphism Syndrome	AD	99.31	22 of 22
WDR4	Galloway-Mowat Syndrome, Microcephaly, Growth Deficiency, Seizures, And Brain Malformations, Galloway-Mowat Syndrome	AR	99.91	7 of 7
WDR62	Autosomal Recessive Primary Microcephaly	AR	100	60 of 61
WDR73	Galloway-Mowat Syndrome, Camos Syndrome	AR	95.71	14 of 14
YWHAE	17p13.3 Microduplication Syndrome, Miller-Dieker Syndrome	-	98.99	0 of 1
ZNHIT3	Peho Syndrome	AR	73.96	1 of 1

*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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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.

References

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