Ventriculomegaly

Precision Panel

Overview

Ventriculomegaly is the term used to describe ventricular dilation unrelated to increased cerebrospinal fluid pressure, such as dilation due to brain dysgenesis or atrophy. Also known as hydrocephalus, which is described as the pathological dilation of the brain's ventricular system, these terms are used interchangeably. Fetal cerebral ventriculomegaly is a relatively common finding on the second trimester obstetric ultrasound examination, even isolated ventriculomegaly can be a normal variant associated with normal offspring outcome. Ventriculomegaly can be caused by a variety of disorders that result in neurologic, motor and/or cognitive impairment. It is one of the most common abnormal sonographic findings associated with congenital infection, chromosomal and additional structural abnormalities.

The Igenomix Ventriculomegaly Precision Panel can be used to make a directed and accurate diagnosis and correlate an ultrasound finding with potential syndromic associations 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 Ventriculomegaly Precision Panel is indicated for those patients with ultrasound findings suggestive of ventriculomegaly which include:

- Atrial diameter > or equal to 10mm
- Cerebrospinal fluid obstruction

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 ultrasound surveillance as well as perinatal and postnatal follow-up for potential worsening of ventriculomegaly.
- 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**
ACD	Dyskeratosis Congenita, Melanoma, Hoyeraal-Hreidarsson Syndrome	AD,AR	99.89	14 of 14
ACP5	Immunodeficiency, Autoimmunity, Spondyloenchondrodysplasia	AR	100	27 of 28
ACTA2	Aortic Aneurysm, Moyamoya Disease, Multisystemic Smooth Muscle Dysfunction Syndrome, Thoracic Aortic Aneurysm, Aortic Dissection	AD	100	88 of 88
ACTB	Baraitser-Winter Syndrome, Dystonia, Becker Nevus Syndrome, Developmental Malformations, Deafness	AD	100	40 of 40
ACTG1	Baraitser-Winter Syndrome, Deafness	AD	98.59	55 of 55
ADAR	Aicardi-Goutieres Syndrome, Dyschromatosis Symmetrica, Bilateral Striatal Necrosis	AD,AR	99.93	252 of 252
ADGRG1	Polymicrogyria	AR	100	-
ADNP	Helsmoortel-Van Der Aa Syndrome, Adnp Syndrome	AD	99.91	90 of 90
AHCY	Hypermethioninemia, Psychomotor Delay, S-Adenosylhomocysteine Hydrolase Deficiency	AR	100	11 of 11
AHI1	Joubert Syndrome, Joubert Syndrome, Retinitis Pigmentosa	AR	96.79	85 of 97
AKT3	Megalencephaly, Polymicrogyria, Polydactyly, Hydrocephalus	AD	99.9	9 of 11
ALDH7A1	Epilepsy	AR	99.98	131 of 134
AMPD2	Pontocerebellar Hypoplasia, Spastic Paraplegia	AR	99.99	24 of 24
ANKLE2	Microcephaly	AR	96.08	4 of 4
ANKRD11	Kbg Syndrome, 16q24.3 Microdeletion Syndrome	AD	99.6	119 of 124
ANTXR1	Gapo Syndrome, Hemangioma	AD,AR	100	19 of 19
AP1S2	Mental Retardation, Fried Syndrome, Dandy-Walker Malformation, Basal Ganglia Disease, Seizures, Hypotonia, Facial Dysmorphism	X,XR,G	84.15	-
AP4B1	Spastic Paraplegia, Intellectual Disability	AR	99.64	22 of 22
AP4E1	Spastic Paraplegia, Stuttering, Intellectual Disability	AD,AR	99.94	17 of 17
AP4M1	Spastic Paraplegia, Intellectual Disability	AR	100	18 of 18
APC2	Cortical Dysplasia, Sotos Syndrome	AR	94.97	11 of 11
ARHGAP31	Adams-Oliver Syndrome	AD	100	6 of 6
ARID1A	Coffin-Siris Syndrome	AD	95.32	40 of 42
ARID1B	Coffin-Siris Syndrome, 6q25 Microdeletion Syndrome	AD	93.87	226 of 238
ARID2	Coffin-Siris Syndrome	AD	99.97	17 of 17
ARMC9	Joubert Syndrome	AR	99.95	10 of 10
ARX	Corpus Callosum Agenesis, Epileptic Encephalopathy, Mental Retardation, Partington Syndrome, West Syndrome, Lissencephaly, Abnormal Genitalia, Spasticity	X,XR,G	81.92	-
ASNS	Asparagine Synthetase Deficiency	AR	99.98	37 of 37
ASPM	Microcephaly	AR	99.74	221 of 222
ASXL1	Bohring-Opitz Syndrome, Myelodysplastic Syndrome, Systemic Mastocytosis, Hematologic Neoplasm	AD	99.96	41 of 41
ASXL2	Shashi-Pena Syndrome	AD	99.75	6 of 6
ATP1A1	Charcot-Marie-Tooth Disease, Hypomagnesemia, Seizures, Mental Retardation	AD	100	16 of 16
ATP6	Leber Optic Atrophy, Neuropathy, Ataxia, Retinitis Pigmentosa, Bilateral Striatal Necrosis, Leigh Syndrome, Spastic Paraplegia, Narp Syndrome	MI	-	-
ATP6AP2	Congenital Disorder Of Glycosylation, Mental Retardation, Epilepsy, Parkinson Disease, Spasticity	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

ATP6V1E1	Cutis Laxa	AR	100	2 of 2
ATXN1	Spinocerebellar Ataxia	AD	99.93	2 of 2
ATXN2	Parkinson Disease, Spinocerebellar Ataxia, Amyotrophic Lateral Sclerosis	AD	91.78	9 of 10
ATXN3	Machado-Joseph Disease	AD	99.94	-
B3GALNT2	Muscular Dystrophy-Dystroglycanopathy, Intellectual Disability, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	97.14	17 of 17
B3GLCT	Peters-Plus Syndrome	AR	99.96	-
B4GALT1	Congenital Disorder Of Glycosylation	AR	99.97	3 of 3
B4GAT1	Muscular Dystrophy-Dystroglycanopathy, Walker-Warburg Syndrome	AR	-	-
B9D1	Joubert Syndrome, Meckel Syndrome	AR	90.23	11 of 11
B9D2	Meckel Syndrome	AR	84.81	4 of 5
BGN	Meester-Loeys Syndrome, Spondyloepimetaphyseal Dysplasia	X,XR,G	99.87	-
BICD2	Spinal Muscular Atrophy	AD	99.94	39 of 39
BMP2	Brachydactyly, Hemochromatosis, Short Stature, Facial Dysmorphism, Skeletal And Cardiac Anomalies, 20p12.3 Microdeletion Syndrome	AD,AR	99.48	12 of 12
BMP4	Microphthalmia, Cleft Lip/Palate, Brain And Digit Anomalies	AD,MU,P	100	38 of 42
BRCA1	Breast And Ovarian Cancer, Fanconi Anemia, Pancreatic Carcinoma, Peritoneal Carcinoma	AD,AR,MU	98.97	2783 of 2894
BRCA2	Fanconi Anemia, Wilms Tumor, Multiple Cancer Types	AD,AR,MU	98.51	3343 of 3451
BRF1	Cerebellofaciodental Syndrome	AR	99.9	17 of 17
BRIP1	Fanconi Anemia, Breast And Ovarian Cancer	AD,AR	94.97	235 of 237
BUB1	Colorectal Cancer, Mosaic Variegated Aneuploidy Syndrome	AD	99.76	18 of 19
BUB1B	Colorectal Cancer, Mosaic Variegated Aneuploidy Syndrome	AD,AR	99.84	30 of 31
BUB3	Mosaic Variegated Aneuploidy Syndrome	-	99.98	6 of 6
C12ORF57	Craniofacial Dysmorphism, Ocular Coloboma, Absent Corpus Callosum, Aortic Dilatation, Temtamy Syndrome	AR	-	-
C2CD3	Orofaciodigital Syndrome	AR	97.25	18 of 18
CASK	Anemia, Fg Syndrome, Mental Retardation, Microcephaly, Pontine And Cerebellar Hypoplasia, Epileptic Encephalopathy	X,XR,XD,G	99.98	-
CC2D2A	Coach Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.43	98 of 100
CCDC103	Ciliary Dyskinesia	AR	99.92	6 of 6
CCDC174	Hypotonia,Psychomotor Retardation	AR	99.89	1 of 1
CCDC22	Ritscher-Schinzel Syndrome, 3c Syndrome	X,XR,G	99.94	-
CCDC39	Ciliary Dyskinesia	AR	99.56	48 of 52
CCDC40	Ciliary Dyskinesia	AR	98	50 of 50
CCDC65	Ciliary Dyskinesia	AR	99.98	3 of 3
CCDC88A	Peho-Like Syndrome	AR	91.9	3 of 4
CCDC88C	Hydrocephalus, Spinocerebellar Ataxia	AD,AR	99.44	13 of 14
CCND2	Megalencephaly, Polymicrogyria, Polydactyly, Hydrocephalus	AD	99.97	9 of 9
CCNO	Ciliary Dyskinesia	AR	99.94	12 of 12
CDC42	Takenouchi-Kosaki Syndrome, Macrothrombocytopenia, Lymphedema, Developmental Delay, Facial Dysmorphism, Camptodactyly	AD	99.97	10 of 10
CDK5RAP2	Microcephaly	AR	100	32 of 32
CDK6	Microcephaly	AR	100	1 of 1
CDKN1C	Beckwith-Wiedemann Syndrome, Intrauterine Growth Retardation, Metaphyseal Dysplasia, Adrenal Hypoplasiacongenita, Genital Anomalies, Image Syndrome, Diabetes	AD	73.58	55 of 76

CENPJ	Microcephaly, Seckel Syndrome	AR	99.97	13 of 13
CEP120	Joubert Syndrome, Short-Rib Thoracic Dysplasia, Polydactyly, Jeune Syndrome	AR	99.8	9 of 9
CEP135	Microcephaly	AR	99.48	7 of 8
CEP152	Microcephaly, Seckel Syndrome	AR	97.73	21 of 24
CEP290	Bardet-Biedl Syndrome, Joubert Syndrome, Leber Congenital Amaurosis, Meckel Syndrome, Senior-Loken Syndrome	AR	96.47	293 of 327
CEP55	Multinucleated Neurons, Anhydramnios, Renal Dysplasia, Cerebellar Hypoplasia, Hydranencephaly, Meckel Syndrome	AR	99.22	3 of 3
CEP57	Mosaic Variegated Aneuploidy Syndrome	AR	99.64	6 of 6
CEP63	Seckel Syndrome, Microcephaly	AR	100	3 of 3
CFAP221	Primary Ciliary Dyskinesia	-	89.78	-
CFAP298	Ciliary Dyskinesia	AR	-	-
CFAP300	Ciliary Dyskinesia	AR	-	-
CHD3	Snijders Blok-Campeau Syndrome	AD	97.93	30 of 30
CHD4	Sifrim-Hitz-Weiss Syndrome	AD	99.65	34 of 34
CHD7	Charge Syndrome, Hypogonadotropic Hypogonadism, Kallmann Syndrome, Omenn Syndrome	AD	96.25	823 of 896
CHST14	Ehlers-Danlos Syndrome	AR	97.7	21 of 22
CILK1	Endocrine-Cerebroosteodysplasia, Epilepsy	AD,AR	100	-
CIT	Microcephaly	AR	99.98	17 of 17
CLCN4	Mental Retardation	X,XR,XD,G	99.69	-
CLP1	Pontocerebellar Hypoplasia	AR	99.89	2 of 2
CNNM2	Hypomagnesemia, Mental Retardation, Seizures, Normocalciuria, Normocalcemia	AD,AR	99.98	9 of 9
COG5	Congenital Disorder Of Glycosylation	AR	100	19 of 19
COG6	Congenital Disorder Of Glycosylation, Shaheen Syndrome, Hypohidrosis, Enamel Hypoplasia, Palmoplantar Keratoderma, Intellectual Disability	AR	100	13 of 13
COG8	Congenital Disorder Of Glycosylation	AR	100	8 of 8
COL18A1	Glaucoma, Knobloch Syndrome	AD,AR	99.76	-
COL3A1	Ehlers-Danlos Syndrome, Polymicrogyria, Acrogeria, Aneurysm	AD,AR	100	676 of 676
COL4A1	Angiopathy, Nephropathy, Aneurysms, Muscle Cramps, Microangiopathy, Leukoencephalopathy, Porencephaly, Hanac Syndrome, Walker-Warburg Syndrome	AD	99.99	173 of 173
COL4A2	Porencephaly	AD	99.93	28 of 28
COPB2	Microcephaly	AR	99.64	4 of 4
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
CRB2	Ventriculomegaly, Cystic Kidney Disease, Focal Segmental Glomerulosclerosis	AR	99.5	26 of 29
CRPPA	Muscular Dystrophy-Dystroglycanopathy, Limb Girdle Muscular Dystrophy, Intellectual Disability, Walker-Warburg Syndrome	AR	97.69	-
CSF1R	Brain Abnormalities, Neurodegeneration, Dysosteosclerosis, Gliosis	AD,AR	100	122 of 124
CSGALNACT1	Skeletal Dysplasia	AR	100	4 of 5
CSPP1	Joubert Syndrome, Jeune Asphyxiating Thoracic Dystrophy, Meckel Syndrome	AR	98.32	29 of 30
CTBP1	Hypotonia, Ataxia, Developmental Delay, Tooth Enamel Defect, Wolf-Hirschhorn Syndrome	AD	98.45	1 of 1
CTCF	Mental Retardation, Feeding Difficulties, Developmental Delay, Microcephaly	AD	96.6	39 of 41
CTDP1	Congenital Cataracts, Facial Dysmorphism, Neuropathy	AR	97.52	0 of 1
CTNNB1	Colorectal Cancer, Exudative Vitreoretinopathy, Hepatocellular Carcinoma, Medulloblastoma, Mental	AD,AR	100	63 of 63

	Retardation, Pilomatrixoma, Craniopharyngioma, Desmoid Tumor, Spastic Diplegia			
CUL4B	Mental Retardation, Short Stature, Small Testes, Musclewasting, Tremor	X,XR,G	99.77	-
CYFIP2	Epileptic Encephalopathy	AD	100	8 of 8
D2HGDH	D-2-Hydroxyglutaric Aciduria	AR	100	42 of 42
DAG1	Muscular Dystrophy-Dystroglycanopathy, Limb-Girdle Muscular Dystrophy, Muscle-Eye-Brain Disease, Bilateral Multicystic Leucodystrophy, Walker-Warburg Syndrome	AR	99.98	9 of 9
DCHS1	Mitral Valve Prolapse, Van Maldergem Syndrome, Cerebrofacioarticular Syndrome	AD,AR	99.69	30 of 30
DCX	Lissencephaly	X,G	100	-
DDX3X	Intellectual Developmental Disorder, Hypotonia	X,XR,XD,G	99.03	-
DEAF1	Dyskinesia, Seizures, Intellectual Developmental Disorder, Mental Retardation, Epilepsy, Extrapyramidal Syndrome, Smith-Magenis Syndrome	AD,AR	93.55	42 of 42
DENND5A	Epileptic Encephalopathy	AR	100	9 of 9
DHCR24	Desmosterolosis	AR	100	10 of 10
DHCR7	Smith-Lemli-Opitz Syndrome	AR	100	217 of 217
DHX30	Neurodevelopmental Disorder, Absent Language	AD	99.98	6 of 6
DISC1	Microcephaly, Polymicrogyria, Corpus Callosum Agenesis	-	97.88	16 of 17
DKC1	Dyskeratosis Congenita, Hoyeraal-Hreidarsson Syndrome	X,XR,G	100	-
DLL1	Neurodevelopmental Disorder, Seizures, Holoprosencephaly	AD	99.83	15 of 15
DMPK	Dystrophia Myotonica, Steinert Myotonic Dystrophy	AD	99.83	3 of 3
DNAAF1	Ciliary Dyskinesia	AR	99.55	36 of 37
DNAAF2	Ciliary Dyskinesia	AR	97.45	7 of 8
DNAAF3	Ciliary Dyskinesia	AR	98.95	13 of 14
DNAAF4	Ciliary Dyskinesia	AD,AR	99.27	-
DNAAF5	Ciliary Dyskinesia	AR	89.27	-
DNAAF6	Ciliary Dyskinesia	X,XR,G	99.63	-
DNAH1	Ciliary Dyskinesia, Spermatogenic Failure	AR	100	58 of 58
DNAH11	Ciliary Dyskinesia	AR	99.27	159 of 169
DNAH5	Ciliary Dyskinesia	AR	100	277 of 278
DNAH9	Ciliary Dyskinesia	AR	98.86	19 of 19
DNAI1	Kartagener Syndrome, Ciliary Dyskinesia	AR	96.91	43 of 43
DNAI2	Ciliary Dyskinesia	AR	98.89	8 of 8
DNAJB13	Ciliary Dyskinesia	AR	99.94	3 of 3
DNAL1	Ciliary Dyskinesia	AR	99.43	5 of 5
DNMT1	Cerebellar Ataxia, Deafness, Narcolepsy, Neuropathy	AD	97.87	30 of 30
DNMT3A	Heyn-Sproul-Jackson Syndrome, Leukemia, Tatton-Brown-Rahman Syndrome, Sporadic Pheochromocytoma, Secreting Paraganglioma, Tall Stature, Intellectual Disability, Facial Dysmorphism	AD	99.95	67 of 68
DOK7	Fetal Akinesia Deformation Sequence, Myasthenia, Limb-Girdle Muscular Dystrophy	AR	99.88	72 of 72
DPF2	Coffin-Siris Syndrome	AD	99.99	10 of 10
DPH1	Developmental Delay, Short Stature, Craniofacial Dysplasia, Intellectual Disability	AR	100	8 of 8
DRC1	Ciliary Dyskinesia	AR	100	9 of 9
DSE	Ehlers-Danlos Syndrome	AR	99.94	3 of 3
DYNC2H1	Short-Rib Thoracic Dysplasia, Polydactyly, Jeune Syndrome	AR,MU,D	99.78	214 of 221
DYNC2I1	Short-Rib Thoracic Dysplasia, Polydactyly, Jeune Syndrome	AR	97.76	14 of 14

DYNC2I2	Short-Rib Thoracic Dysplasia, Polydactyly, Jeune Syndrome,	AR	99.54	23 of 23
DYRK1A	Mental Retardation, Intellectual Disability	AD	99.85	78 of 81
EBP	Chondrodysplasia Punctata, Mend Syndrome	X,XR,XD,G	100	-
EGF	Hypomagnesemia, Normocalciuria, Normocalcemia	AR	99.98	9 of 9
EHMT1	Kleefstra Syndrome	AD	98.58	58 of 75
EIF2S3	Mehmo Syndrome	X,XR,G	98.64	-
EMG1	Bowen-Conradi Syndrome	AR	99.91	1 of 1
EML1	Band Heterotopia	AR	98.88	7 of 7
EOMES	Microcephaly, Polymicrogyria, Corpus Callosum Agenesis	-	98.82	-
ERCC2	Cerebrooculofacioskeletal Syndrome, Trichothiodystrophy, Xeroderma Pigmentosum	AR	100	102 of 102
ERCC3	Trichothiodystrophy, Xeroderma Pigmentosum	AR	99.98	24 of 24
ERCC4	Fanconi Anemia, Xeroderma Pigmentosum, Xfe Progeroid Syndrome, Cockayne Syndrome	AR	99.68	69 of 72
EVC	Ellis-Van Creveld Syndrome, Weyers Acrofacial Dysostosis, Acrofacial Dysostosis	AD,AR	94.04	68 of 73
EVC2	Ellis-Van Creveld Syndrome, Weyers Acrofacial Dysostosis	AD,AR	99.98	75 of 75
EXT1	Chondrosarcoma, Exostoses, Multiple Osteochondromas, Trichorhinophalangeal Syndrome	AD,AR	99.97	518 of 525
EXTL3	Immunoskeletal Dysplasia, Neurodevelopmental Abnormalities, Immunodeficiency	AR	99.99	10 of 10
EZH2	Weaver Syndrome	AD	99.82	40 of 41
FANCA	Fanconi Anemia	AR	95.17	497 of 502
FANCB	Fanconi Anemia, VACTERL, Hydrocephalus	X,XR,G	95.53	-
FANCC	Fanconi Anemia	AR	100	75 of 75
FANCD2	Fanconi Anemia	AR	100	62 of 63
FANCE	Fanconi Anemia	AR	97	17 of 18
FANCF	Fanconi Anemia	AR	99.31	17 of 18
FANCG	Fanconi Anemia	-	100	94 of 94
FANCI	Fanconi Anemia	AR	100	53 of 54
FANCL	Fanconi Anemia	AR	100	25 of 26
FANCM	Ovarian Failure, Spermatogenic Failure, Fanconi Anemia, Male Infertility	AR	99.73	59 of 61
FAR1	Peroxisomal Fatty Acyl-Coa Reductase 1 Disorder	AR	98.77	4 of 4
FARS2	Combined Oxidative Phosphorylation Deficiency, Spastic Paraplegia	AR	99.98	23 of 23
FAT4	Hennekam Lymphangiectasia-Lymphedema Syndrome, Van Maldergem Syndrome, Cerebrofacioarticular Syndrome	AR	99.8	41 of 41
FBN1	Acromicric Dysplasia, Ectopia Lentis, Geleophysic Dysplasia, Marfan Syndrome, Mass Syndrome, Stiff Skin Syndrome, Weill-Marchesani Syndrome, Familial Thoracic Aortic Aneurysm, Aortic Dissection, Glaucoma, Microspherophakia, Shprintzen-Goldberg Syndrome	AD	100	2836 of 2845
FBP1	Fructose-1,6-Bisphosphatase Deficiency	AR	100	47 of 49
FBXW11	Neurodevelopmental, Jaw, Eye, And Digital Syndrome, Intellectual Disability	AD	99.89	10 of 10
FGFR1	Encephalocraniocutaneous Lipomatosis, Hartsfield Syndrome, Jackson-Weiss Syndrome, Kallmann Syndrome, Osteoglophonic Dysplasia, Pfeiffer Syndrome, Trigonocephaly, Holoprosencephaly, Hypogonadotropic Hypogonadism, Oligodontia	AD	100	279 of 280
FGFR2	Antley-Bixler Syndrome, Apert Syndrome, Bent Bone Dysplasia Syndrome, Crouzon Syndrome, Scaphocephaly Syndrome, Gastric Cancer, Jackson-Weiss Syndrome, Lacrimoauriculodentodigital Syndrome, Saethre-Chotzen Syndrome, Cutis Gyrate,	AD	98	140 of 143

	Acanthosis Nigricans, Craniosynostosis, Pfeiffer Syndrome			
FGFR3	Achondroplasia, Bladder Cancer, Camptodactyly, Cervical Cancer, Colorectal Cancer, Crouzon Syndrome, Acanthosis Nigricans, Epidermal Nevus, Hypochondroplasia, Lacrimoauriculodentodigital Syndrome, Muenke Syndrome, Testicular Tumor, Thanatophoric Dysplasia, Tall Stature, Scoliosis, Hearing Loss, Brachycephaly, Plagiocephaly, Saethre-Chotzen Syndrome	AD,AR	99.89	77 of 78
FGFRL1	Wolf-Hirschhorn Syndrome	AD	99.94	1 of 1
FIG4	Amyotrophic Lateral Sclerosis, Charcot-Marie-Tooth Disease, Cleidocranial Dysplasia, Micrognathia, Absent Thumbs, Polymicrogyria, Yunis-Varon Syndrome	AD,AR	99.92	72 of 72
FKRP	Limb Girdle Muscular Dystrophy, Muscular Dystrophy-Dystroglycanopathy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	99.9	157 of 157
FKTN	Cardiomyopathy, Muscular Dystrophy-Dystroglycanopathy, Limb Girdle Muscular Dystrophy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	98	54 of 56
FLI1	Bleeding Disorder, Jacobsen Syndrome, Paris-Trousseau Thrombocytopenia, Neuroectodermal Tumor	AD,AR	100	7 of 7
FLII	Smith-Magenis Syndrome	-	99.98	3 of 3
FLVCR2	Proliferative Vasculopathy, Hydranencephaly-Hydrocephaly Syndrome	AR	99.97	16 of 16
FOXF1	Alveolar Capillary Dysplasia, Misalignment Of Pulmonary Veins	AD	95.93	74 of 96
FOXJ1	Ciliary Dyskinesia	AD	99.69	5 of 5
FOXP3	Immunodysregulation, Polyendocrinopathy, Enteropathy	X,XR,G	99.86	-
FOXRED1	Mitochondrial Complex I Deficiency, Leigh Syndrome, Leukodystrophy	AR	100	13 of 13
FTO	Body Mass Index Quantitative Trait Locus, Developmental Delay	AR	99.91	8 of 8
GABBR2	Epileptic Encephalopathy, Neurodevelopmental Disorder, Rett Syndrome	AD	95.98	7 of 7
GABRD	1p36 Deletion Syndrome, Febrile Seizures, Myoclonic Epilepsy	AD	95.23	3 of 3
GAS2L2	Ciliary Dyskinesia	AR	89	4 of 5
GAS8	Ciliary Dyskinesia	AR	99.98	6 of 6
GBA	Dementia, Gaucher Disease, Parkinson Disease, Ophthalmoplegia, Cardiovascular Calcification	AD,AR	100	469 of 471
GCDH	Glutaric Acidemia, Glutaryl-Coa Dehydrogenase Deficiency	AR	88.74	254 of 254
GFM2	Combined Oxidative Phosphorylation Deficiency	AR	99.35	5 of 7
GLB1	Gangliosidosis, Morquio Syndrome	AR	100	242 of 243
GLI3	Greig Cephalopolysyndactyly Syndrome, Hypothalamic Hamartomascongenital Hypothalamic Hamartoma Syndrome, Polydactyly, Acrocallosal Syndrome, Pallister-Hall Syndrome, Tibial Hemimelia	AD,AR	100	231 of 231
GLUL	Glutamine Deficiency	AR	100	4 of 4
GMPPB	Muscular Dystrophy-Dystroglycanopathy, Myasthenic Syndromes, Glycosylation Defect, Limb-Girdle Muscular Dystrophy, Muscle-Eye-Brain Disease	AR	99.95	53 of 53
GNAO1	Epileptic Encephalopathy, Neurodevelopmental Disorder	AD	100	47 of 47
GPC3	Simpson-Golabi-Behmel Syndrome, Wilms Tumor, Nephroblastoma	AD,X,XR,G	99.84	-
GPC4	Keipert Syndrome, Simpson-Golabi-Behmel Syndrome, Wilms Tumor	AD,X,XR,G	98.43	-
GPSM2	Chudley-Mccullough Syndrome	AR	100	13 of 13
GRM1	Spinocerebellar Ataxia	AD,AR	99.88	20 of 21

GRN	Ceroid Lipofuscinosis, Frontotemporal Lobar Degeneration, Frontotemporal Dementia, Semantic Dementia	AD,AR	100	220 of 229
GTF2E2	Trichothiodystrophy	AR	99.98	2 of 2
GTF2H5	Trichothiodystrophy	AR	100	8 of 8
H19-ICR	Beckwith-Wiedemann Syndrome, Multiple Cancer Types, Silver-Russell Syndrome	AD	-	-
HDAC8	Cornelia De Lange Syndrome, Wilson-Turner Syndrome	X,XD,G	99.78	-
HECW2	Neurodevelopmental Disorder, Hypotonia, Seizures	AD	99.85	13 of 13
HEPACAM	Megalencephalic Leukoencephalopathy, Subcortical Cysts, Mental Retardation	AD,AR	97.87	30 of 30
HERC1	Macrocephaly, Dysmorphic Facies, Psychomotor Retardation, Megalencephaly, Kyphoscoliosis	AR	99.96	11 of 11
HERC2	Mental Retardation, Prader-Willi Syndrome	AD,AR	98.91	9 of 9
HIBCH	3-Hydroxyisobutyryl-Coa Hydrolase Deficiency, Neurodegeneration	AR	96.47	27 of 27
HK1	Hemolytic Anemia, Neurodevelopmental Disorder, Visual Defects, Neuropathy, Retinitis Pigmentosa, Charcot-Marie-Tooth Disease	AD,AR	100	14 of 17
HNRNPU	Epileptic Encephalopathy, 1q44 Microdeletion Syndrome	AD	99.8	36 of 36
HRAS	Bladder Cancer, Costello Syndrome, Epidermal Nevus, Schimmelpenning-Feuerstein-Mims Syndrome, Thyroid Cancer	AD	100	34 of 34
HS6ST2	Paganini-Miozzo Syndrome	X,XR,G	98.39	-
HSD17B4	D-Bifunctional Protein Deficiency, Perrault Syndrome	AR	99.52	85 of 85
HTRA2	3-Methylglutaconic Aciduria, Parkinson Disease	AD,AR	99.81	18 of 18
HTT	Huntington Disease, Lopes-Maciel-Rodan Syndrome	AD,AR	99	-
HYDIN	Ciliary Dyskinesia	AR	81.7	45 of 63
HYLS1	Hydrolethalus Syndrome, Joubert Syndrome	AR	100	2 of 2
IFIH1	Aicardi-Goutieres Syndrome, Singleton-Merten Syndrome	AD	99.62	26 of 27
IFT172	Retinitis Pigmentosa, Short-Rib Thoracic Dysplasia, Polydactyly, Bardet-Biedl Syndrome, Jeune Syndrome	AR	100	37 of 37
IFT43	Cranioectodermal Dysplasia, Retinitis Pigmentosa, Short-Rib Thoracic Dysplasia, Polydactyly	AR	100	6 of 6
IFT80	Asphyxiating Thoracic Dystrophy, Jeune Syndrome, Short Rib-Polydactyly Syndrome	AR	99.96	16 of 16
IGF2	Beckwith-Wiedemann Syndrome, Growth Restriction, Silver-Russell Syndrome, Wilms Tumor, Hemihyperplasia	AD,X,XR,G	100	9 of 9
INPP5E	Joubert Syndrome, Mental Retardation, Obesity, Retinal Dystrophy, Hepatic Defect	AR	99.89	56 of 56
INTU	Orofaciodigital Syndrome, Short-Rib Throacic Dysplasia, Polydactyly	AR	99.7	9 of 9
IPW	Prader-Willi Syndrome	AD	-	-
IQSEC2	Mental Retardation, Microduplication Xp11.22p11.23 Syndrome, Microcephaly, Smith-Magenis Syndrome	X,XR,XD,G	99.73	-
ISCA1	Multiple Mitochondrial Dysfunctions Syndrome	AR	99.86	2 of 2
JAM2	Basal Ganglia Calcification	AR	99.98	-
JAM3	Hemorrhagic Destruction Of The Brain, Subependymal Calcification, Cataracts	AR	100	4 of 4
KANK1	Cerebral Palsy	AD,ADWMI	99.91	6 of 6
KANSL1	Koolen-De Vries Syndrome	AD	96.03	22 of 27
KAT8	Li-Ghorgani-Weisz-Hubshman Syndrome	AD	99.97	1 of 1
KATNB1	Lissencephaly, Microcephaly	AR	100	10 of 10
KCNAB2	1p36 Deletion Syndrome	-	79	3 of 3
KCNK4	Facial Dysmorphism, Hypertrichosis, Epilepsy, Developmental Delay, Gingival Overgrowth Syndrome	AD	94.93	2 of 2
KCNQ1	Atrial Fibrillation, Beckwith-Wiedemann Syndrome, Jervell And Lange-Nielsen Syndrome, Long Qt	AD,AR	93.23	600 of 624

	Syndrome, Short Qt Syndrome, Romano-Ward Syndrome			
KCNQ1OT1	Beckwith-Wiedemann Syndrome, Hemihyperplasia	AD	-	-
KDM6A	Kabuki Syndrome	AD,X,XD,G	99.98	-
KIAA0586	Joubert Syndrome, Short-Rib Thoracic Dysplasia, Polydactyly, Joubert Syndrome	AR	99.84	31 of 32
KIAA0753	Orofaciodigital Syndrome	AR	97.73	7 of 7
KIAA1109	Alkuraya-Kucinskas Syndrome	AR	99.95	21 of 21
KIDINS220	Spastic Paraplegia, Intellectual Disability, Nystagmus, Obesity	AD	99.83	17 of 17
KIF14	Meckel Syndrome, Microcephaly	AR	99.84	18 of 18
KIF1A	Mental Retardation, Neuropathy, Spastic Paraplegia, Peho Syndrome	AD,AR	100	76 of 76
KIF7	Acrocallosal Syndrome, Hydrolethalus Syndrome, Macrocephaly, Multiple Epiphyseal Dysplasia, Distinctive Facies, Orofaciodigital Syndrome	AR	94.91	47 of 50
KIFBP	Goldberg-Shprintzen Syndrome	AR	99.27	-
KLHL15	Mental Retardation	X,XR,G	99.61	-
KLHL7	Crisponi Sweating Syndrome, Retinitis Pigmentosa, Bohring-Opitz Syndrome	AD,AR	98.69	19 of 19
KMT2A	Hairy Elbows, Short Stature, Facial Dysmorphism, Developmental Delay, Cornelia De Lange Syndrome, Wiedemann-Steiner Syndrome	AD	98.14	144 of 149
KMT2D	Kabuki Syndrome	AD	99.71	839 of 847
KNL1	Microcephaly	AR	98.91	-
KNSTRN	Immunodeficiency, Faciooculoskeletal Anomalies	-	99.98	-
KRAS	Aplasia Cutis Congenita, Arteriovenous Malformation Of The Brain, Cardiofaciocutaneous Syndrome, 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
L1CAM	Corpus Callosum Agenesis, Hydrocephalus, Stenosis Of Aqueduct Of Sylvius, Masa Syndrome	X,XR,G	100	-
LAMA1	Poretti-Boltshauser Syndrome, Ataxia, Intellectual Disability	AR	100	43 of 43
LARGE1	Muscular Dystrophy-Dystroglycanopathy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	100	-
LETM1	Wolf-Hirschhorn Syndrome	AD	98.2	2 of 2
LMNB2	Barraquer-Simons Syndrome, Epilepsy, Lipodystrophy	AD,AR	95.03	5 of 5
LONP1	Codas Syndrome	AR	99.84	21 of 21
LRRC56	Ciliary Dyskinesia	AR	99.77	5 of 5
LRRC6	Ciliary Dyskinesia	AR	99.88	21 of 21
MAB21L1	Cerebellar, Ocular, Craniofacial, And Genital Syndrome	AR	99.97	6 of 6
MAD2L2	Fanconi Anemia	AR	99.91	1 of 1
MAF	Ayme-Gripp Syndrome, Cataract	AD	75.14	23 of 23
MAG	Spastic Paraplegia	AR	99.97	7 of 7
MAGEL2	Prader-Willi Syndrome	AD	99.99	43 of 48
MAST1	Mega-Corpus-Callosum Syndrome, Cerebellar Hypoplasia, Cortical Malformations	AD	98.92	26 of 26
MBTPS2	Ichthyosis Follicularis, Atrichia, Photophobia Syndrome, Keratosis Follicularis Spinulosa Decalvans, Osteogenesis Imperfecta, Palmoplantar Keratoderma, Bresek Syndrome, Alopecia, Periorificial Keratotic Plaques	X,XR,G	100	-
MCIDAS	Ciliary Dyskinesia	AR	99.92	4 of 4
MCPH1	Microcephaly	AR	99.51	18 of 19
MED12	Lujan-Fryns Syndrome, Ohdo Syndrome, Opitz-Kaveggia Syndrome, Blepharophimosis, Intellectual Disability, Fg Syndrome	X,XR,G	100	-

MED25	Basel-Vanagaite-Smirin-Yosef Syndrome, Charcot-Marie-Tooth Disease, Congenital Cataract, Microcephaly, Nevus Flammeus Simplex, Intellectual Disability	AR	100	5 of 5
MEF2C	Mental Retardation, Stereotypic Movements, Epilepsy, Cerebral Malformations, 5q14.3 Microdeletion Syndrome	AD	99.91	43 of 46
METTL5	Intellectual Developmental Disorder, Microcephaly	AR	99.9	4 of 4
MFSD2A	Microcephaly	AR	97.58	6 of 6
MKRN3	Prader-Willi Syndrome, Precocious Puberty	AD,ADWMI	99.98	39 of 41
MKRN3-AS1	Prader-Willi Syndrome	AD	-	-
MKS1	Bardet-Biedl Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.98	49 of 49
MOCS1	Molybdenum Cofactor Deficiency	AR	100	36 of 37
MOCS2	Molybdenum Cofactor Deficiency	AR	100	32 of 32
MPDZ	Hydrocephalus	AR	99.44	58 of 58
MPLKIP	Trichothiodystrophy	AR	100	13 of 13
MRE11	Ataxia-Telangiectasia-Like Disorder, Breast And Ovarian Cancer	AR	99.95	-
MRPS16	Oxidative Phosphorylation Deficiency	AR	100	1 of 1
MSL3	Basilicata-Akhtar Syndrome	X,XD,G	99.72	-
MTHFR	Neural Tube Defects, Schizophrenia, Thrombophiliavenous Thromboembolism, Homocystinuria, Methylene Tetrahydrofolate Reductase Deficiency, Anencephaly, Exencephaly	AD,AR	100	122 of 122
MTHFS	Neurodevelopmental Disorder, Microcephaly, Epilepsy, Hypomyelination	AR	100	5 of 5
MTOR	Focal Cortical Dysplasia Of Taylor, Smith-Kingsmore Syndrome, Macrocephaly, Intellectual Disability, Neurodevelopmental Disorder	AD	99.98	39 of 39
MTRR	Homocystinuria, Megaloblastic Anemia, Neural Tube Defects, Methylcobalamin Deficiency	AR	100	39 of 40
MUSK	Fetal Akinesia Deformation Sequence, Myasthenic Syndrome, Acetylcholinereceptor Deficiency	AR	95.58	23 of 25
MYMK	Carey-Fineman-Ziter Syndrome	AR	100	-
MYOD1	Myopathy, Diaphragmatic Defects, Respiratory Insufficiency, Dysmorphic Facies, Fetal Akinesia Deformation Sequence	AR	99.97	6 of 6
MYORG	Basal Ganglia Calcification, Striopallidodentate Calcinosis	AR	100	-
NAA10	Microphthalmia, Ogden Syndrome	X,XR,XD,G	99.86	-
NADK2	2,4-Dienoyl-Coa Reductase Deficiency, Progressive Encephalopathy, Leukodystrophy	AR	95.37	3 of 3
NANS	Spondyloepimetaphyseal Dysplasia	AR	99.97	12 of 12
NCAPD3	Microcephaly	AR	99.97	4 of 5
NDE1	Lissencephaly, Microhydranencephaly, Hydranencephaly	AR	86.55	12 of 13
NDN	Prader-Willi Syndrome	AD	97.41	2 of 2
NDUFA6	Mitochondrial Complex I Deficiency	AR	100	9 of 9
NDUFAF3	Mitochondrial Complex I Deficiency, Leigh Syndrome With Cardiomyopathy	AR	100	9 of 9
NDUFB11	Linear Skin Defects, Microphthalmia, Mitochondrial Complex I Deficiency	X,XD,G	97.48	-
NDUFB8	Mitochondrial Complex I Deficiency, Leigh Syndrome, Cardiomyopathy	AR	100	4 of 4
NDUFS2	Mitochondrial Complex I Deficiency, Leber Optic Neuropathy, Leigh Syndrome, Cardiomyopathy, Leukodystrophy	AR	100	26 of 26
NEK1	Amyotrophic Lateral Sclerosis, Short Rib-Polydactyly Syndrome, Orofaciodigital Syndrome	AD,AR,MU,D	99.83	73 of 74
NEK10	Ciliary Dyskinesia	AR	99.95	3 of 3
NEXMIF	Mental Retardation	X,XR,XD,G	99.74	-

NFIA	Chromosome 1p32-P31 Deletion Syndrome	AD	92.5	7 of 7
NFIX	Marshall-Smith Syndrome, Sotos Syndrome, 19p13.3 Microduplication Syndrome, Malan Overgrowth Syndrome	AD	94.42	75 of 81
NGLY1	Congenital Disorder Of Glycosylation, Alacrimia-Choreoathetosis-Liver Dysfunction Syndrome	AR	99.8	28 of 28
NID1	Isolated Dandy-Walker Malformation	-	100	4 of 4
NIPBL	Cornelia De Lange Syndrome	AD	99.32	409 of 426
NME8	Ciliary Dyskinesia	AR	99.99	9 of 9
NOTCH2NLC	Neuronal Intranuclear Inclusion Disease, Tremor	AD	-	-
NPAP1	Prader-Willi Syndrome	AD	99.82	-
NPHP3	Meckel Syndrome, Nephronophthisis, Renal-Hepatic-Pancreatic Dysplasia, Senior-Loken Syndrome	AR	99.99	84 of 84
NRAS	Epidermal Nevus, Giant Pigmented Hairy Nevus, Neurocutaneous Melanosis, Noonan Syndrome, Autoimmune Lymphoproliferative Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome, Multiple Cancer Types	AD	100	15 of 15
NSD1	Sotos Syndrome, 5q35 Microduplication Syndrome, Weaver Syndrome	AD	99.8	451 of 459
NSD2	Wolf-Hirschhorn Syndrome	AD	99.91	-
NUP188	Sandestig-Stefanova Syndrome	AR	100	6 of 6
NUP88	Fetal Akinesia Deformation Sequence	AR	95.82	3 of 3
OCLN	Pseudo-Torch Syndrome, Intrauterine Infection-Like Syndrome	AR	86.89	15 of 17
OCRL	Dent Disease, Lowe Oculocerebrorenal Syndrome	X,XR,G	100	-
ODAD1	Ciliary Dyskinesia	AR	99.68	10 of 10
ODAD2	Ciliary Dyskinesia	AR	97.3	26 of 28
ODAD3	Ciliary Dyskinesia	AR	95	4 of 4
ODAD4	Ciliary Dyskinesia	AR	-	-
ODC1	Colorectal Cancer, Global Developmental Delay, Alopecia, Macrocephaly, Facial Dysmorphism, Structural Brain Anomalies	AD	100	7 of 7
OFD1	Joubert Syndrome, Orofaciodigital Syndrome, Retinitis Pigmentosa, Simpson-Golabi-Behmel Syndrome, Orofaciodigital Syndrome, Primary Ciliary Dyskinesia	X,XR,XD,G	98.09	-
OPHN1	Mental Retardation, Cerebellar Hypoplasia, Distinctivefacial Appearance	X,XR,G	100	-
OSGEP	Galloway-Mowat Syndrome	AR	99.17	19 of 19
OSTM1	Osteopetrosis, Neuroaxonal Dysplasia	AR	100	8 of 9
OTUD6B	Intellectual Developmental Disorder, Seizures, Distal Limb Anomalies, Facial Dysmorphism, Global Developmental Delay	AR	99.81	7 of 7
PAFAH1B1	Lissencephaly, 17p13.3 Microduplication Syndrome, Miller-Dieker Syndrome	AD	99.95	90 of 92
PALB2	Fanconi Anemia, Breast And Ovarian Cancer	AD,AR	98.78	601 of 617
PARN	Dyskeratosis Congenita, Pulmonary Fibrosis, Bone Marrow Failure, Hoyeraal-Hreidarsson Syndrome	AD,AR	99.98	33 of 33
PAX6	Aniridia, Anterior Segment Dysgenesis, Coloboma Of Optic Nerve, Foveal Hypoplasia, Presenile Cataract, Keratitis, Wilms Tumor, Cerebellar Ataxia, Intellectual Disability, Morning Glory Disc Anomaly, Peters Anomaly, Wagr Syndrome	AD	100	460 of 485
PCNT	Microcephalic Osteodysplastic Primordial Dwarfism, Seckel Syndrome	AR	99.92	103 of 105
PDGFB	Basal Ganglia Calcification, Meningioma, Bilateral Striopallidodentate Calcinosis, Dermatofibrosarcoma Protuberans	AD	100	22 of 22
PDGFRB	Basal Ganglia Calcification, Kosaki Overgrowth Syndrome, Myeloproliferative Disorder, Myofibromatosis, Premature Aging Syndrome	AD	99.64	28 of 28

PDHA1	Pyruvate Decarboxylase Deficiency, Leigh Syndrome, Leukodystrophy	X,XD,G	99.02	-
PHACTR1	Epileptic Encephalopathy, West Syndrome	AD	99.89	5 of 5
PHC1	Microcephaly	AR	91.73	1 of 1
PHGDH	Neu-Laxova Syndrome, Phosphoglycerate Dehydrogenase Deficiency	AR	100	26 of 26
PIEZO2	Arthrogryposis, Gordon Syndrome, Marden-Walker Syndrome	AD,AR	96.93	37 of 37
PIGB	Epileptic Encephalopathy	AR	99.97	10 of 10
PIGN	Multiple Congenital Anomalies, Hypotonia, Seizures, Fryns Syndrome	AR	93.97	36 of 39
PIGO	Hyperphosphatasia, Mental Retardation	AR	99.93	21 of 21
PIGQ	Epileptic Encephalopathy	AR	99.99	4 of 4
PIK3CA	Capillary Malformation Of The Lower Lip, Lipomatous Overgrowth, Multiple Cancer Types, Cowden Syndrome, Epidermal Nevus, Keratosis, Macrocephaly, Megalodactyly, Hemihyperplasia, Lynch Syndrome, Megalencephaly, Polymicrogyria	AD	99.58	54 of 58
PIK3CD	Immunodeficiency, Faciooculoskeletal Anomalies	AD	100	23 of 23
PLAA	Neurodevelopmental Disorder, Progressive Microcephaly, Spasticity, Brain Anomalies	AR	99.41	6 of 6
PLG	Plasminogen Deficiency, Hypoplasminogenemia	AR	100	79 of 79
PLPBP	Epilepsy	AR	100	-
PMM2	Congenital Disorder Of Glycosylation	AR	100	127 of 129
PMPCA	Spinocerebellar Ataxia, Cerebelloparenchymal Disorder	AR	99.91	9 of 9
PNKP	Ataxia-Oculomotor Apraxia, Charcot-Marie-Tooth Disease, Epileptic Encephalopathy	AR	100	36 of 36
POLR2A	Neurodevelopmental Disorder, Hypotonia	AD	100	17 of 17
POLR3A	Leukodystrophy, Oligodontia, Hypogonadotropic Hypogonadism, Progeroid Syndrome, Hypomyelination, Hypodontia, Tremor, Ataxia, Wiedemann-Rautenstrauch Syndrome	AR	100	122 of 122
POMGNT1	Muscular Dystrophy-Dystroglycanopathy, Limb Girdle Muscular Dystrophy, Retinitis Pigmentosa, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	99.91	82 of 83
POMGNT2	Muscular Dystrophy-Dystroglycanopathy, Limb Girdle Muscular Dystrophy, Walker-Warburg Syndrome	AR	100	10 of 10
POMK	Muscular Dystrophy-Dystroglycanopathy, Limb Girdle Muscular Dystrophy, Walker-Warburg Syndrome	AR	99.99	8 of 8
POMT1	Muscular Dystrophy-Dystroglycanopathy, Limb Girdle Muscular Dystrophy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	100	105 of 105
POMT2	Muscular Dystrophy-Dystroglycanopathy, Muscle-Eye-Brain Disease, Limb-Girdle Muscular Dystrophy, Walker-Warburg Syndrome	AR	100	74 of 74
PPP1CB	Noonan Syndrome	AD	99.87	12 of 12
PPP2CA	Neurodevelopmental Disorder, Language Delay	AD	99.9	14 of 14
PPP2R1A	Mental Retardation, Microcephaly, Corpus Callosum Hypoplasia	AD	91.99	6 of 6
PPP2R5D	Mental Retardation, Macrocephaly, Hypotonia	AD	100	11 of 11
PRDM16	Left Ventricular Noncompaction, 1p36 Deletion Syndrome, Dilated Cardiomyopathy	AD	98.81	20 of 20
PRNP	Creutzfeldt-Jakob Disease, Insomnia, Gerstmann-Straussler Disease, Huntington Disease, Kuru, Spongiform Encephalopathy, Neuropsychiatric Features, Alzheimer	AD	100	69 of 69
PSAT1	Neu-Laxova Syndrome, Phosphoserine Aminotransferase Deficiency	AR	99.95	9 of 9
PTCH1	Basal Cell Carcinoma, Holoprosencephaly, Gorlin Syndrome, Monosomy 9q22.3	AD	98.89	498 of 502
PTPN23	Neurodevelopmental Disorder, Structural Brain Anomalies, Seizures, Spasticity	AR	99.99	17 of 17

PUF60	Verheij Syndrome, 8q24.3 Microdeletion Syndrome, Intellectual Disability, Cardiac Anomalies, Short Stature, Joint Laxity	AD	100	30 of 30
PUS3	Mental Retardation, Severe Growth Deficiency, Strabismus, Extensive Dermal Melanocytosis	AR	99.01	8 of 9
PWAR1	Prader-Willi Syndrome	AD	-	-
PWRN1	Prader-Willi Syndrome	AD	-	-
PYCR2	Leukodystrophy, Microcephaly	AR	98.29	14 of 14
QARS1	Microcephaly, Cerebral Atrophy	AR	-	-
RAB18	Warburg Micro Syndrome, Micro Syndrome	AR	100	4 of 4
RAC1	Mental Retardation, Microcephaly, Corpus Callosum, Cerebellar Vermis Hypoplasia, Facial Dysmorphism	AD	98.73	9 of 9
RAC3	Neurodevelopmental Disorder, Structural Brain Anomalies, Dysmorphic Facies	AD	94.13	5 of 5
RAD21	Cornelia De Lange Syndrome, Mungan Syndrome	AD,AR	99.8	16 of 17
RAD51	Breast And Ovarian Cancer, Fanconi Anemia, Mirror Movements	AD	99.98	16 of 16
RAD51C	Breast-Ovarian Cancer, Fanconi Anemia	AR	100	130 of 130
RAI1	Smith-Magenis Syndrome, 17p11.2 Microduplication Syndrome	AD	99.91	50 of 53
RAPSN	Fetal Akinesia Deformation Sequence, Myasthenic Syndrome, Acetylcholine Receptor Deficiency	AR	99.98	59 of 61
RERE	Neurodevelopmental Disorder, Anomalies Of The Brain, Eye Or Heart, 1p36 Deletion Syndrome	AD	92.43	21 of 21
RFWD3	Fanconi Anemia	AR	99.99	2 of 2
RHOBTB2	Epileptic Encephalopathy	AD	100	6 of 6
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
RNF113A	Trichothiodystrophy	X,XD,G	99.7	-
RNF125	Tenorio Syndrome	AD	100	3 of 3
RNF213	Moyamoya Disease	-	99.82	109 of 110
ROGDI	Kohlschutter-Tonz Syndrome, Amelocerebrohypohidrotic Syndrome	AR	99.83	10 of 12
RPGR	Cone-Rod Dystrophy, Macular Degeneration, Retinitis Pigmentosa, Deafness, Achromatopsia, Ciliary Dyskinesia	X,XR,G	94	-
RPGRIP1	Cone-Rod Dystrophy, Leber Amaurosis, Meckel Syndrome	AR	99.33	146 of 159
RPGRIP1L	Coach Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.96	52 of 52
RPS6KA3	Coffin-Lowry Syndrome, Mental Retardation	X,XD,G	99.95	-
RRAS2	Noonan Syndrome	AD	99.8	6 of 6
RSPH1	Ciliary Dyskinesia	AR	100	10 of 10
RSPH3	Ciliary Dyskinesia	AR	99.85	5 of 5
RSPH4A	Ciliary Dyskinesia	AR	99.98	27 of 27
RSPH9	Ciliary Dyskinesia	AR	100	13 of 13
RTEL1	Dyskeratosis Congenita, Pulmonary Fibrosis, Bone Marrow Failure, Hoyeraal-Hreidarsson Syndrome	AD,AR	99.73	127 of 131
RTTN	Microcephaly, Polymicrogyria, Seizures, Dwarfism	AR	99.94	28 of 29
RXYLT1	Muscular Dystrophy-Dystroglycanopathy, Walker-Warburg Syndrome	AR	99.46	-
SAMHD1	Aicardi-Goutieres Syndrome, Chilblain Lupus	AD,AR	100	51 of 51
SASS6	Microcephaly	AR	99.14	6 of 6
SCO2	Cardioencephalomyopathy, Cytochrome C Oxidasedeficiency, Myopia, Charcot-Marie-Tooth Disease, Copper Metabolism Defect, Leigh Syndrome	AD,AR	100	38 of 38
SEC31A	Neurodevelopmental Disorder, Spastic Quadriplegia, Optic Atrophy, Seizures	AR	99.92	3 of 3

SEMA3E	Charge Syndrome, Hypogonadotropic Hypogonadism	AD,AR	99.81	6 of 7
SETBP1	Mental Retardation, Schinzel-Giedion Syndrome, Expressive Aphasia, Facial Dysmorphism Syndrome	AD	98.61	43 of 43
SETD2	Luscan-Lumish Syndrome, Sotos Syndrome	AD	99.83	19 of 19
SETD5	Mental Retardation, Cornelia De Lange Syndrome	AD	99.77	37 of 37
SH2B1	16p11.2 Microdeletion Syndrome, Obesity, Insulin Resistance Syndrome	-	99.98	25 of 25
SHANK3	Phelan-Mcdermid Syndrome, Schizophrenia, Monosomy 22q13.3	AD,MU,P	96.67	-
SHH	Holoprosencephaly, Microphthalmia, Schizencephaly, Hypoplastic Tibiae, Postaxial Polydactyly, Radial Hemimelia, Syndactyly	AD	99.48	161 of 184
SHPK	Sedoheptulokinase Deficiency	-	99.96	2 of 2
SIM1	6q16 Microdeletion Syndrome, Obesity, Prader-Willi-Like Syndrome	-	99.64	39 of 40
SIN3A	Chromosome 15q24 Deletion Syndrome, Mental Retardation	AD	99.94	18 of 18
SKI	Shprintzen-Goldberg Syndrome, 1p36 Deletion Syndrome	AD	99.66	39 of 39
SLC12A6	Peripheral Neuropathy, Corpus Callosum Agenesis	AR	100	21 of 21
SLC13A5	Epileptic Encephalopathy, Amelocerebrohypohidrotic Syndrome	AR	95.92	24 of 24
SLC18A3	Myasthenic Syndrome, Fetal Akinesia Deformation Sequence	AR	99.97	5 of 5
SLC20A2	Basal Ganglia Calcification, Bilateral Striopallidodentate Calcinosis	AD	99.96	123 of 127
SLC25A1	Hydroxyglutaric Aciduria, Myasthenic Syndrome	AR	90	23 of 25
SLC25A19	Microcephaly, Thiamine Metabolism Dysfunction Syndrome	AR	97.13	10 of 10
SLC35A2	Congenital Disorder Of Glycosylation	X,XD,G	99.97	-
SLC39A8	Congenital Disorder Of Glycosylation	AR	99.89	7 of 7
SLC6A9	Glycine Encephalopathy	AR	99.99	5 of 5
SLC9A6	Mental Retardation, Christianson Syndrome	X,XD,G	98.87	-
SLX4	Fanconi Anemia	AR	99.92	76 of 76
SMARCA4	Coffin-Siris Syndrome, Rhabdoid Tumor Predisposition Syndrome	AD	100	68 of 69
SMARCB1	Coffin-Siris Syndrome, Rhabdoid Tumor Predisposition Syndrome, Schwannomatosis, Meningioma	AD	100	97 of 99
SMARCC2	Coffin-Siris Syndrome	AD	99.49	16 of 16
SMARCD1	Coffin-Siris Syndrome	AD	93.17	7 of 7
SMARCE1	Coffin-Siris Syndrome, Meningioma	AD	98.98	15 of 15
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
SMG9	Heart And Brain Malformation Syndrome	AR	100	4 of 4
SMO	Basal Cell Carcinoma, Craniofacial Malformations, Hypothalamic Hamartomascongenital Hypothalamic Hamartoma Syndrome, Curry-Jones Syndrome, Meningioma	AR	94.03	10 of 10
SNIP1	Psychomotor Retardation, Epilepsy, Craniofacial Dysmorphism	AR	99.68	1 of 1
SNORD115-1	Prader-Willi Syndrome	AD	-	-
SNORD116-1	Prader-Willi Syndrome	AD	-	-
SNRPN	Autism, Prader-Willi Syndrome	AD,MU	100	2 of 2
SON	Zttk Syndrome, Brain Malformations, Musculoskeletal Abnormalities, Facial Dysmorphism, Intellectual Disability	AD	99.27	30 of 32
SOX11	Mental Retardation, Coffin-Siris Syndrome	AD	95.23	11 of 11
SOX4	Coffin-Siris Syndrome	AD	75.52	4 of 4

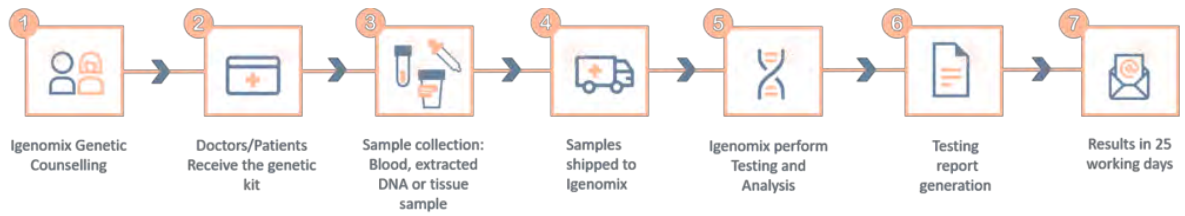
SOX9	Campomelic Dysplasia, Testicular Disorder Of Sex Development, Partial Gonadal Dysgenesis, Pierre Robin Syndrome	AD	97.28	87 of 95
SPAG1	Ciliary Dyskinesia	AR	94.8	11 of 12
SPECC1L	Facial Clefting, Hypertelorism, Opitz Gbbb Syndrome	AD	99.66	14 of 14
SPEF2	Spermatogenic Failure, Ciliary Dyskinesia	AR	99.6	10 of 13
SPG11	Amyotrophic Lateral Sclerosis, Charcot-Marie-Tooth Disease, Spastic Paraplegia	AR	99.93	289 of 297
STAC3	Native American Myopathy	AR	99.98	5 of 5
STIL	Microcephaly	AR	99.94	18 of 18
STK36	Ciliary Dyskinesia	-	100	5 of 5
STRADA	Polyhydramnios, Megalencephaly, Symptomatic Epilepsy	AR	97.95	4 of 6
SUCLA2	Mitochondrial Dna Depletion Syndrome, Methylmalonic Aciduria	AR	100	27 of 27
SUFU	Basal Cell Nevus Syndrome, Joubert Syndrome, Medulloblastoma, Acrocallosal Syndrome, Gorlin Syndrome, Meningioma, Microform Holoprosencephaly	AD,AR	99.99	43 of 43
SUMF1	Multiple Sulfatase Deficiency	AR	100	52 of 52
SURF1	Charcot-Marie-Tooth Disease, Leigh Syndrome, Cardiomyopathy, Leukodystrophy	AR,MI	98.59	117 of 124
TAF1	Dystonia, Mental Retardation, Dystonia, Parkinson Disease, Global Development Delay, Facial Dysmorphism, Sacral Caudal Remnant	X,XR,G	99.74	-
TAF13	Mental Retardation, Microcephaly	AR	99.97	5 of 5
TAPT1	Osteochondrodysplasia	AR	89.49	3 of 3
TARS1	Trichothiodystrophy	AR	99.94	-
TBC1D24	Deafness, Doors Syndrome, Epilepsy, Epileptic Encephalopathy	AD,AR	100	80 of 80
TBCD	Encephalopathy, Brain Atrophy, Thin Corpus Callosum, Microcephaly, Muscle Weakness, Optic Atrophy	AR	94.89	28 of 28
TBCE	Encephalopathy, Amyotrophy, Optic Atrophy, Hypoparathyroidism, Kenny-Caffey Syndrome, Spastic Ataxia, Distal Spinal Muscular Atrophy, Sanjad-Sakati Syndrome	AR	100	8 of 8
TBCK	Hypotonia, Psychomotor Retardation, Intellectual Disability	AR	99.95	15 of 15
TBL1XR1	Mental Retardation, Pierpont Syndrome, Promyelocytic Leukemia	AD	99.78	23 of 23
TCTN2	Joubert Syndrome, Meckel Syndrome	AR	100	14 of 14
TERT	Aplastic Anemia, Dyskeratosis Congenita, Leukemia, Melanoma, Pulmonary Fibrosis, Bone Marrow Failure, Hoyeraal-Hreidarsson Syndrome, Meningioma	AD,AR	99.09	194 of 197
TET3	Beck-Fahrner Syndrome	AD,AR	97.53	1 of 1
TGDS	Catel-Manzke Syndrome	AR	99.99	7 of 7
THOC2	Mental Retardation, Short Stature, Overweight	X,XR,G	96.31	-
THOC6	Microcephaly, Cardiac And Genitourinary Malformations, Developmental Delay, Facial Dysmorphism	AR	100	13 of 13
TINF2	Dyskeratosis Congenita, Revesz Syndrome, Hoyeraal-Hreidarsson Syndrome	AD	99.94	47 of 47
TMCO1	Cerebrofaciothoracic Dysplasia	AR	88	5 of 5
TMEM107	Meckel Syndrome, Orofaciodigital Syndrome	AR	100	3 of 3
TMEM138	Joubert Syndrome, Oculorenal Defect	AR	99.94	9 of 9
TMEM216	Joubert Syndrome, Meckel Syndrome, Orofaciodigital Syndrome	AR	98.74	8 of 8
TMEM231	Joubert Syndrome, Meckel Syndrome, Oculorenal Defect, Orofaciodigital Syndrome	AR	98.63	20 of 21
TMEM237	Joubert Syndrome, Oculorenal Defect	AR	100	11 of 11

TMEM67	Bardet-Biedl Syndrome, Coach Syndrome, Joubert Syndrome, Meckel Syndrome, Nephronophthisis, Rhyns Syndrome, Hepatic Defect	AR	96.93	177 of 179
TMTC3	Lissencephaly, Periventricular Nodular Heterotopia	AR	99.04	10 of 10
TMX2	Neurodevelopmental Disorder, Microcephaly, Cortical Malformations, Spasticity	AR	99.98	12 of 12
TNFRSF11A	Osteopetrosis, Paget Disease Of Bone, Polyostotic Osteolytic Dysplasia, Dysosteosclerosis	AD,AR	96.37	17 of 22
TNNI3	Cardiomyopathy	AD,AR	100	139 of 139
TPRKB	Galloway-Mowat Syndrome	AR	85.66	2 of 2
TRAF7	Cardiac, Facial, And Digital Anomalies, Developmental Delay, Meningioma	AD	100	5 of 5
TRAIP	Seckel Syndrome	AR	100	2 of 2
TRAPPC12	Encephalopathy, Brain Atrophy, Spasticity, Hearing Loss, Pons Hypoplasia, Brain Atrophy	AR	99.98	3 of 3
TRAPPC14	Microcephaly	AR	-	-
TRAPPC6B	Neurodevelopmental Disorder, Microcephaly, Epilepsy, Brain Atrophy	AR	100	4 of 4
TREM2	Polycystic Lipomembranous Osteodysplasia, Sclerosing Leukoencephalopathy, Amyotrophic Lateral Sclerosis, Frontotemporal And Semantic Dementia, Alzheimer Disease, Nasu-Hakola Disease, Non-Fluent Aphasia	AD	100	55 of 55
TREX1	Aicardi-Goutieres Syndrome, Lupus Erythematosus, Vasculopathy, Leukodystrophy	AD,AR	100	75 of 75
TRIM37	Mulibrey Nanism	AR	97	20 of 22
TRIM71	Hydrocephalus	AD	97.46	2 of 2
TRIP13	Mosaic Variegated Aneuploidy Syndrome, Nephroblastoma	AR	98.14	2 of 2
TRPS1	Trichorhinophalangeal Syndrome	AD	99.45	108 of 112
TSEN2	Pontocerebellar Hypoplasia	AR	95.47	4 of 5
TSFM	Oxidative Phosphorylation Deficiency	AR	93.35	11 of 14
TTC12	Ciliary Dyskinesia	AR	99.97	-
TUBA1A	Lissencephaly	AD	100	95 of 95
TUBB	Cortical Dysplasia	AD	100	8 of 8
TUBB2A	Cortical Dysplasia	AD	81.71	5 of 7
TUBB2B	Cortical Dysplasia, Dysequilibrium Syndrome, Polymicrogyria	AD	84.28	29 of 38
TUBB3	Cortical Dysplasia, Fibrosis Of Extraocular Muscles, Cortical Dysgenesis, Pontocerebellar Hypoplasia	AD	99.96	30 of 30
TYROBP	Polycystic Lipomembranous Osteodysplasia, Sclerosing Leukoencephalopathy, Nasu-Hakola Disease	AR	100	12 of 13
UBE2T	Fanconi Anemia	AR	100	4 of 4
UBE3B	Kaufman Oculocerebrofacial Syndrome	AR	100	28 of 28
UBTF	Neurodegeneration, Motor And Cognitive Regression Syndrome, Extrapyramidal Movement Disorder	AD	99.99	2 of 2
UGDH	Epileptic Encephalopathy	AR	99.98	2 of 2
USP18	Pseudo-Torch Syndrome	AR	95.84	1 of 1
USP7	16p13.2 Microdeletion Syndrome	AD	99.98	18 of 18
USP9X	Facial Dysmorphism, Short Stature, Intellectual Disability	X,XR,XD,G	98.61	-
VPS11	Leukodystrophy	AR	100	2 of 2
VPS13A	Choreoacanthocytosis	AR	99.37	120 of 122
VPS37A	Spastic Paraplegia	AR	99.95	2 of 2
VPS51	Pontocerebellar Hypoplasia	AR	99.98	1 of 1
WAC	Desanto-Shinawi Syndrome, Facial Dysmorphism, Developmental Delay, Behavioral Abnormalities	AD	98.98	35 of 35
WARS2	Neurodevelopmental Disorder, Lactic Acidosis, Oxidative Phosphorylation Defect	AR	99.95	14 of 15
WASF1	Neurodevelopmental Disorder, Absent Language, Seizures	AD	97.03	3 of 3

WASHC5	Dandy-Walker Malformation, Atrioventricular Septal Defect, Spastic Paraplegia, 3c Syndrome	AD,AR	99.99	-
WDPCP	Bardet-Biedl Syndrome, Congenital Heart Defects, Hamartomas Of Tongue, Polysyndactyly, Heart Defect, Tongue Hamartoma, Meckel Syndrome	AR	99.3	8 of 8
WDR26	Skraban-Deardorff Syndrome, Intellectual Disability, Seizures, Facial Dysmorphism	AD	99.31	22 of 22
WDR35	Cranioectodermal Dysplasia, Short-Rib Thoracic Dysplasia, Polydactyly	AR	100	31 of 33
WDR45B	Neurodevelopmental Disorder, Spastic Quadriplegia, Brain Abnormalities, Seizures	AR	99	4 of 4
WDR62	Microcephaly	AR	100	60 of 61
WDR73	Galloway-Mowat Syndrome, Camos Syndrome	AR	95.71	14 of 14
WDR81	Cerebellar Hypoplasia, Mental Retardation, Hydrocephalus, Brain Anomalies, Dysequilibrium Syndrome	AR	99.94	19 of 19
WHCR	Wolf-Hirschhorn Syndrome	AD	na	-
XPR1	Basal Ganglia Calcification, Bilateral Striopallidodentate Calcinosis	AD	99.88	14 of 14
XRCC2	Fanconi Anemia, Male Infertility	AR	98.39	28 of 28
XRCC4	Short Stature, Microcephaly, Endocrine Dysfunction, Lig4 Syndrome, Dwarfism	AR	99.73	10 of 10
YWHAE	17p13.3 Microduplication Syndrome, Miller-Dieker Syndrome	-	98.99	0 of 1
YY1	Gabriele-De Vries Syndrome	AD	99.89	13 of 13
ZBTB11	Intellectual Developmental Disorder	AR	99.56	2 of 2
ZC4H2	Wieacker-Wolff Syndrome, Intellectual Disability, Developmental Delay	X,XR,XD,G	99.69	-
ZEB2	Mowat-Wilson Syndrome	AD	98.95	253 of 254
ZIC1	Craniosynostosis, Structural Brain Anomalies, Impaired Intellectual Development, Brachycephaly, Oxycephaly, Plagiocephaly	AD	100	7 of 7
ZMIZ1	Neurodevelopmental Disorder, Dysmorphic Facies, Distal Skeletal Anomalies	AD	98.87	13 of 13
ZMYND10	Ciliary Dyskinesia	AR	99.98	16 of 16
ZNF148	Global Developmental Delay	AD	99.82	4 of 4
ZNF335	Microcephaly, Dwarfism	AR	99.83	20 of 20
ZNF462	Weiss-Kruszka Syndrome	AD	100	21 of 21
ZNHIT3	Peho Syndrome	AR	73.96	1 of 1
ZSWIM6	Acromelic Frontonasal Dysostosis, Neurodevelopmental Disorder, Movement Abnormalities, Abnormal Gait, And Autistic Features	AD	91.16	2 of 2

*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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