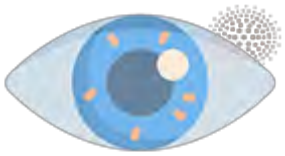
Inherited Retinal Degeneration

Precision Panel



Overview

Inherited Retinal Degeneration (IRD) are a group of rare retinal diseases that ultimately lead to the progressive loss of retinal photoreceptor cells and blindness. These diseases are phenotypically heterogeneous as they can affect individuals of all ages, can progress at different rates and are rare. IRDs are caused by mutations in genes that contribute to proper retinal function, where some genes mutations can cause more severe forms than others. Examples of these diseases include age-related macular degeneration, Leber congenital amaurosis, Stargardt disease and retinitis pigmentosa. Inherited retinal degeneration can be inherited in all modes of inheritance: autosomal dominant, autosomal recessive, X-linked and mitochondria.

The Igenomix Inherited Retinal Degeneration Precision Panel can be used to make an accurate and directed diagnosis as well as a differential diagnosis of progressive blindness 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 Inherited Retinal Degeneration Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- Blurred vision
- Family history of inhertied retinal degeneration
- Difficulties with peripheral vision
- Difficulties with color vision
- Reduction in the sharpness of vision

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 treatment with a multidisciplinary team in the form of stem cell therapy, gene therapy and optogenetic therapy. Surgical care with retinal prosthetics and neuroprotective agents to prevent visual complications.

- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
AARS1	Charcot-Marie-Tooth Disease, Epileptic Encephalopathy	AD,AR	99.07	30 of 30
ABCA4	Cone-Rod Dystrophy, Macular Degeneration, Retinitis Pigmentosa, Stargardt Disease	AD,AR	100	1392 of 1430
ABCC6	Arterial Calcification, Pseudoxanthoma Elasticum	AD,AR	99	346 of 349
ACO2	Cerebellar-Retinal Degeneration, Optic Atrophy	AR	100	33 of 33
ACTL6A	Intellectual Disability	-	99.98	3 of 3
ACTL6B	Epileptic Encephalopathy, Intellectual Developmental Disorder, Speech And Ambulation Defects	AD,AR	100	21 of 21
ACVRL1	Osler-Rendu-Weber Syndrome, Hemorrhagic Telangiectasia	AD	100	457 of 462
ADAMTS18	Microcornea, Myopic Chorioretinal Atrophy, Telecanthus	AR	100	14 of 14
AIPL1	Leber Congenital Amaurosis, Retinitis Pigmentosa, Cone Rod Dystrophy	AD,AR,X,XR,G	89	82 of 82
ALDH3A2	Sjogren-Larsson Syndrome	AR	96	119 of 119
ALMS1	Alstrom Syndrome	AR	99.92	302 of 305
ANO10	Spinocerebellar Ataxia, Cerebellar Ataxia	AR	100	28 of 28
AP3B2	Epileptic Encephalopathy	AR	99.95	11 of 12
APOB	Hypercholesterolemia, Hypobetalipoproteinemia	AD,AR	99.62	369 of 375
APOE	Alzheimer Disease, Lipoprotein Glomerulopathy, Macular Degeneration, Dysbetalipoproteinemia, Sea-Blue Histiocytosis	AD,AR	99.53	65 of 68
ARL2BP	Retinitis Pigmentosa	AR	99.99	7 of 7
ARL6	Bardet-Biedl Syndrome, Retinitis Pigmentosa	AD,AR,X,XR,G	100	17 of 21
ARSG	Usher Syndrome	AR	99.98	2 of 2
ARV1	Epileptic Encephalopathy	AR	100	3 of 3
ASAH1	Farber Lipogranulomatosis, Spinal Muscular Atrophy, Myoclonic Epilepsy, Farber Disease, Spinal Muscular Atrophy	AR	99.98	69 of 70
ATF6	Achromatopsia, Cone Rod Dystrophy	AR	99.98	16 of 16
ATP6V1A	Cutis Laxa, Epileptic Encephalopathy	AD,AR	99.98	9 of 9
ATXN7	Spinocerebellar Ataxia, Retinal Degeneration, Macular Degeneration, Ophthalmoplegia	AD	94.99	-
BBS1	Bardet-Biedl Syndrome	AR	100	102 of 105
BBS2	Bardet-Biedl Syndrome, Retinitis Pigmentosa	AR	100	99 of 100
BBS4	Bardet-Biedl Syndrome	AR	100	45 of 48
BCORL1	Shukla-Vernon Syndrome, Intellectual Disability	X,XR,G	98.77	-
BEST1	Bestrophinopathy, Macular Dystrophy, Retinitis Pigmentosa, Vitreoretinochoroidopathy, Foveomacular Vitelliform Dystrophy, Nanophthalmos	AD,AR	94.35	342 of 344
BPTF	Neurodevelopmental Disorder, Dysmorphic Facies, Distal Limb Anomalies, 17q24.2 Microdeletion Syndrome, Intellectual Disability	AD	94.31	12 of 15
C1QTNF5	Retinal Degeneration	AD	99.97	7 of 7
C8ORF37	Bardet-Biedl Syndrome, Cone-Rod Dystrophy, Retinitis Pigmentosa	AD,AR,X,XR,G	-	-
CACNA1A	Epileptic Encephalopathy, Spinocerebellar Ataxia, Migraine, Paroxysmal Torticollis Of Infancy	AD	96.13	249 of 266
CACNA1B	Neurodevelopmental Disorder, Seizures, Nonepileptic Hyperkinetic Movements, Epileptic Encephalopathy	AR	95.83	7 of 7
CCDC28B	Bardet-Biedl Syndrome	AR	99.83	1 of 1
CDH3	Eem Syndrome, Hypotrichosis, Macular Dystrophy	AR	95	34 of 36

CDK19	Epileptic Encephalopathy	AD	99.81	1 of 1
CEP164	Nephronophthisis, Senior-Loken Syndrome	AR	99.98	10 of 10
CEP78	Cone-Rod Dystrophy, Hearing Loss, Usher Syndrome	AR	99.44	9 of 10
CFAP410	Retinal Dystrophy, Macular Staphyloma, Spondylometaphyseal Dysplasia, Amyotrophic Lateral Sclerosis, Cone Rod Dystrophy	AR	-	-
CFH	Basal Laminar Drusen, Complement Factor H Deficiency, Hemolytic Uremic Syndrome, Macular Degeneration, Hellp Syndrome	AD,AR,MU,P	99.94	340 of 342
CFHR1	Hemolytic Uremic Syndrome, Macular Degeneration	AD,AR	88.29	0 of 9
CFHR3	Hemolytic Uremic Syndrome, Macular Degeneration	AD,AR	89.89	0 of 7
CFI	Complement Factor I Deficiency, Hemolytic Uremic Syndrome, Macular Degeneration, Hellp Syndrome	AD,AR	99.93	156 of 158
CHM	Choroideremia	X,XD,G	99.52	-
CLCC1	Retinitis Pigmentosa	AR	97.97	-
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
CLTC	Mental Retardation, Epileptic Encephalopathy	AD	98.81	14 of 14
CNGA3	Achromatopsia, Cone Rod Dystrophy	AR	99.97	165 of 165
CNGB1	Retinitis Pigmentosa	AR	100	75 of 75
CNGB3	Achromatopsia, Cone Dystrophy, Stargardt Disease	AR	99.83	121 of 126
CNKSR2	Mental Retardation, Epileptic Encephalopathy	X,G	99.11	-
COL18A1	Glaucoma, Knobloch Syndrome	AD,AR	99.76	-
COL2A1	Achondrogenesis, Avascular Necrosis Of Femoral Head, Czech Dysplasia, Multiple Epiphyseal Dysplasia, Myopia, Conductive Deafness, Kniest Dysplasia, Legg-Calve-Perthes Disease, Osteoarthritis, Stickler Syndrome, Dysspondyloenchondromatosis	AD,MU	100	583 of 583
COL8A2	Polymorphous Corneal Dystrophy	AD	94.25	10 of 10
CP	Aceruloplasminemia	AR	99.91	58 of 59
CRB1	Leber Congenital Amaurosis, Chorioretinal Atrophy, Retinitis Pigmentosa, Nanophthalmos	AD,AR,X,G	99.84	365 of 371
CRX	Cone-Rod Dystrophy, Leber Congenital Amaurosis, Retinitis Pigmentosa	AD,AR,X,XR,G	99.91	117 of 117
CST3	Amyloidosis, Macular Degeneration	AD,MU,P	95.95	4 of 4
CTNNB1	Colorectal Cancer, Exudative Vitreoretinopathy, Hepatocellular Carcinoma, Medulloblastoma, Mental Retardation, Pilomatrixoma, Craniopharyngioma, Desmoid Tumor, Spastic Diplegia	AD,AR	100	63 of 63
CTSD	Ceroid Lipofuscinosis	AR	100	18 of 18
CWC27	Retinitis Pigmentosa	AR	99.77	8 of 8
CYFIP2	Epileptic Encephalopathy	AD	100	8 of 8
CYP4V2	Bietti Crystalline Corneoretinal Dystrophy	AR	100	112 of 112
DALRD3	Epileptic Encephalopathy	AR	97.17	-
DDX6	Intellectual Developmental Disorder, Impaired Language, Dysmorphic Facies	-	100	5 of 5
DHDDS	Developmental Delay, Seizures, Retinitis Pigmentosa, Epileptic Encephalopathy	AD,AR	96.32	8 of 8
DHX38	Retinitis Pigmentosa	AR	100	4 of 4
DNM1	Epileptic Encephalopathy, Lennox-Gastaut Syndrome	AD	94.8	30 of 30
DOCK3	Neurodevelopmental Disorder, Hypotonia, Ataxia, Mental Retardation	AR	99.94	7 of 8
DPP6	Mental Retardation, Ventricular Fibrillation, Microcephaly	AD	97.03	23 of 28
DRAM2	Cone Rod Dystrophy	AR	99.87	13 of 13
EEF1A2	Epileptic Encephalopathy, Mental Retardation	AD	100	14 of 14

EFEMP1	Doyne Honeycomb Retinal Dystrophy	AD	100	7 of 7
ELOVL4	Erythrokeratodermia, Ichthyosis, Spastic Quadriplegia, Mental Retardation, Stargardt Disease, Spinocerebellar Ataxia	AD,AR	100	16 of 17
ERCC2	Cerebrooculofacioskeletal Syndrome, Trichothiodystrophy, Xeroderma Pigmentosum, Cofs Syndrome	AR	100	102 of 102
ERCC3	Trichothiodystrophy, Xeroderma Pigmentosum	AR	99.98	24 of 24
ERCC6	Cerebrooculofacioskeletal Syndrome, Cockayne Syndrome, De Sanctis-Cacchione Syndrome, Lung Cancer, Ovarian Failure, Uv-Sensitive Syndrome, Cofs Syndrome	AD,AR	99.98	127 of 128
ERCC8	Cockayne Syndrome, Uv-Sensitive Syndrome	AR	100	60 of 64
EYS	Retinitis Pigmentosa	AR	99.54	358 of 379
FBLN1	Synpolydactyly, Developmental Delay, Central Nervous System Anomaly, Syndactyly	AD	98.03	4 of 4
FBLN5	Cutis Laxa, Neuropathy, Macular Degeneration	AD,AR	97.43	23 of 23
FBN2	Contractural Arachnodactyly, Macular Degeneration	AD	100	115 of 115
FBXW11	Neurodevelopmental, Jaw, Eye, And Digital Syndrome	AD	99.89	10 of 10
FGF12	Epileptic Encephalopathy	AD	99.98	4 of 6
FKRP	Muscular Dystrophy-Dystroglycanopathy, Limb Girdle Muscular Dystrophy, 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
FSCN2	Retinitis Pigmentosa	AD	98.93	16 of 17
FTL	Basal Ganglia Disease, Hyperferritinemia-Cataract Syndrome, L-Ferritin Deficiency, Neuroferritinopathy	AD,AR	100	21 of 63
GABRA2	Alcohol Dependence, Epileptic Encephalopathy	AD,MU	99.08	3 of 3
GABRA5	Epileptic Encephalopathy	AD	99.94	9 of 9
GABRB2	Epileptic Encephalopathy	AD	99.19	16 of 19
GABRG2	Epileptic Encephalopathy, Dravet Syndrome, Rolandic Epilepsy	AD	99.67	53 of 53
GBA	Dementia, Gaucher Disease, Parkinson Disease	AD,AR	100	469 of 471
GNAT2	Achromatopsia, Cone Dystrophy	AR	100	26 of 26
GNB5	Intellectual Developmental Disorder, Cardiac Arrhythmia, Language Delay, Attention Deficit-Hyperactivity Disorder	AR	100	13 of 13
GNPTAB	Mucolipidosis	AR	100	279 of 280
GRHL2	Corneal Dystrophy, Deafness, Ectodermal Dysplasia	AD,AR	100	8 of 11
GRIN2D	Epileptic Encephalopathy	AD	79.74	17 of 18
GTF2E2	Trichothiodystrophy	AR	99.98	2 of 2
GTF2H5	Trichothiodystrophy	AR	100	8 of 8
GUCA1A	Areolar Choroidal Dystrophy, Cone Rod Dystrophy	AD	99.94	27 of 27
GUCA1B	Retinitis Pigmentosa	AD	100	10 of 10
GUCY2D	Cone Rod Dystrophy, Leber Congenital Amaurosis, Night Blindness, Areolar Choroidal Dystrophy	AD,AR	99.98	248 of 248
HADHA	3-Hydroxyacyl-Coa Dehydrogenase Deficiency, Trifunctional Protein Deficiency	AR	100	75 of 75
HCN1	Epileptic Encephalopathy, Febrile Seizures	AD	98.43	42 of 43
HGSNAT	Mucopolysaccharidosis, Retinitis Pigmentosa	AR	87.91	69 of 73
HK1	Hemolytic Anemia, Neurodevelopmental Disorder, Visual Defects, Brain Anomalies, Neuropathy, Retinitis Pigmentosa, Charcot-Marie-Tooth Disease	AD,AR	100	14 of 17
HLA-A	Birdshot Chorioretinopathy	-	99.72	1 of 1
HMCN1	Macular Degeneration	AD	100	13 of 13
HSD17B10	Hydroxyacyl-Coa Dehydrogenase II Deficiency, Hsd10 Disease	X,XD,G	100	-
IDUA	Hurler-Scheie Syndrome	AR	99.73	287 of 292

IFT140	Retinitis Pigmentosa, Short-Rib Thoracic Dysplasia, Polydactyly, Jeune Syndrome, Leber Congenital Amaurosis	AR	99.97	81 of 81
IFT172	Retinitis Pigmentosa, Short-Rib Thoracic Dysplasia, Polydactyly, Bardet-Biedl Syndrome, Jeune Syndrome	AR	100	37 of 37
IMPDH1	Leber Congenital Amaurosis, Retinitis Pigmentosa	AD	99.98	29 of 29
JAG1	Alagille Syndrome, Tetralogy Of Fallot	AD	99.98	640 of 641
KCNA2	Epileptic Encephalopathy	AD	99.86	23 of 23
KCNB1	Epileptic Encephalopathy	AD	99.95	55 of 55
KCNJ13	Leber Congenital Amaurosis, Vitreoretinal Degeneration	AD,AR	99.64	11 of 11
KCNV2	Retinal Cone Dystrophy	AR	99.98	86 of 88
KIAA1549	Retinitis Pigmentosa	AR	96.67	9 of 10
KMT2E	O'donnell-Luria-Rodan Syndrome, Intellectual Disability	AD	99.83	34 of 34
LAMA1	Poretti-Boltshauser Syndrome, Ataxia, Intellectual Disability, Oculomotor Apraxia, Cerebellar Cysts	AR	100	43 of 43
LARGE1	Muscular Dystrophy-Dystroglycanopathy, Limb Girdle Muscular Dystrophy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	100	-
LMNA	Cardiomyopathy, Charcot-Marie-Tooth Disease, Emery-Dreifuss Muscular Dystrophy, Heart-Hand Syndrome, Hutchinson-Gilford Progeria Syndrome, Malouf Syndrome, Mandibuloacral Dysplasia, Werner Syndrome, Lipodystrophic Laminopathy, Hypergonadotropic Hypogonadism, Dermopathy	AD,AR	100	619 of 620
LZTFL1	Bardet-Biedl Syndrome	AR	99.83	4 of 4
MAB21L1	Cerebellar, Ocular, Craniofacial And Genital Syndrome	AR	99.97	6 of 6
MAN2B1	Mannosidosis	AR	100	149 of 149
MAPKAPK3	Macular Dystrophy	AD	99.98	2 of 2
MCOLN1	Mucolipidosis	AR	99.99	34 of 36
MED13	Intellectual Developmental Disorder	AD	97.23	17 of 17
MERTK	Retinitis Pigmentosa	AR	100	99 of 101
MFRP	Microphthalmia, Retinitis Pigmentosa, Foveoschisis, Nanophthalmos	AR	100	36 of 36
MICOS13	Oxidative Phosphorylation Deficiency, 3-Methylglutaconic Aciduria	AR	-	-
MIR204	Retinal Dystrophy, Iris Coloboma, Cataract	AD	-	-
MPLKIP	Trichothiodystrophy	AR	100	13 of 13
MTTP	Abdominal Obesity-Metabolic Syndrome, Abetalipoproteinemia	AD,AR	100	69 of 71
MYT1L	Mental Retardation	AD	99.98	30 of 30
NDE1	Lissencephaly, Hydranencephaly	AR	86.55	12 of 13
NECAP1	Epileptic Encephalopathy	AR	99.83	2 of 2
NR2E3	S-Cone Syndrome, Retinitis Pigmentosa	AD,AR	-	-
NRL	Retinitis Pigmentosa	AD	99.81	25 of 25
NTNG1	Rett Syndrome, Intellectual Disability	-	99.96	2 of 2
NTRK2	Epileptic Encephalopathy, Obesity, Hyperphagia, Developmental Delay, West Syndrome	AD	100	9 of 9
NUS1	Congenital Disorder Of Glycosylation, Mental Retardation, Seizures, Epileptic Encephalopathy	AD,AR	99.62	22 of 23
OAT	Ornithine Aminotransferase Deficiency, Gyrate Atrophy Of Choroid And Retina	AR	100	72 of 73
OPA1	Behr Syndrome, Mitochondrial Dna Depletion Syndrome, Optic Atrophy, Deafness, Ophthalmoplegia, Myopathy, Ataxia, Neuropathy	AD,AR	99.98	397 of 402
OVOL2	Corneal Dystrophy	AD	99.87	0 of 3
P3H2	Myopia, Cataract, Vitreoretinal Degeneration	AR	99.81	-

PANK2	Hypoprebetalipoproteinemia, Acanthocytosis, Retinitis Pigmentosa, Pallidal Degeneration, Neurodegeneration, Brain Iron Accumulation	AR	98.92	177 of 182
PARS2	Epileptic Encephalopathy	AR	100	7 of 7
PAX2	Focal Segmental Glomerulosclerosis, Papillorenal Syndrome, Renal Coloboma Syndrome	AD	99.99	100 of 100
PDE6C	Cone Dystrophy, Achromatopsia	AR	100	63 of 63
PDE6H	Retinal Cone Dystrophy, Achromatopsia	AD,AR	100	2 of 2
PEX7	Peroxisome Biogenesis Disorder, Refsum Disease, Rhizomelic Chondrodysplasia Punctata	AR	99.21	47 of 53
PHYH	Refsum Disease	AR	100	34 of 34
PISD	Liberfarb Syndrome	AR	100	4 of 4
PITPNM3	Cone-Rod Dystrophy	AD	99.8	7 of 7
PLK4	Microcephaly, Chorioretinopathy, Seckel Syndrome	AR	99.74	10 of 10
PNPLA6	Boucher-Neuhauser Syndrome, Laurence-Moon Syndrome, Oliver-Mcfarlane Syndrome, Spastic Paraplegia, Cerebellar Ataxia, Hypogonadism	AR	100	65 of 65
POMGNT1	Muscular Dystrophy-Dystroglycanopathy, Limb Girdle Muscular Dystrophy, Muscle-Eye-Brain Disease, Retinitis Pigmentosa, Walker-Warburg Syndrome	AR	99.91	82 of 83
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, Limb Girdle Muscular Dystrophy, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	100	74 of 74
POU3F4	Deafness, Xq21 Microdeletion Syndrome	X,XR,G	99.98	-
PPP3CA	Arthrogryposis, Cleft Palate, Craniosynostosis, Impaired Intellectual Development, Epileptic Encephalopathy	AD	99.98	16 of 16
PPT1	Ceroid Lipofuscinosis	AR	100	81 of 81
PRCD	Retinitis Pigmentosa	AR	100	7 of 7
PROM1	Cone-Rod Dystrophy, Macular Dystrophy, Retinitis Pigmentosa, Stargardt Disease	AD,AR	99.61	90 of 93
PRPF31	Retinitis Pigmentosa	AD	100	160 of 166
PRPF4	Retinitis Pigmentosa	AD	99.99	5 of 5
PRPF8	Retinitis Pigmentosa	AD	100	58 of 58
PRPH2	Choroidal Dystrophy, Retinitis Punctata Albescens, Macular Dystrophy, Retinitis Pigmentosa, Foveomacular Vitelliform Dystrophy, Cone Rod Dystrophy, Stargardt Disease	AD,AR	100	188 of 188
PSMD12	Stankiewicz-Isidor Syndrome, 17q24.2 Microdeletion Syndrome, Intellectual Disability	AD	97.93	3 of 4
RAX2	Cone-Rod Dystrophy, Macular Degeneration	AD	99.89	7 of 9
RBP4	Microphthalmia, Retinal Dystrophy, Iris Coloboma, Comedogenic Acne Syndrome	AD,AR	100	8 of 8
RDH11	Retinal Dystrophy, Juvenile Cataracts, Short Stature Syndrome, Retinitis Pigmentosa, Intellectual Disability	AR	99.97	3 of 3
RDH5	Retinitis Punctata Albescens	AD,AR	100	54 of 54
REEP6	Retinitis Pigmentosa	AR	97.59	9 of 9
RHO	Night Blindness, Retinitis Pigmentosa, Retinitis Punctata Albescens	AD,AR	100	229 of 229
RLBP1	Bothnia Retinal Dystrophy, Cone Rod Dystrophy, Retinitis Pigmentosa, Retinitis Punctata Albescens	AD,AR	100	32 of 33
RLIM	Mental Retardation	X,XR,G	99.52	-
RNF113A	Trichothiodystrophy	X,XD,G	99.7	-
ROM1	Retinitis Pigmentosa	AD,AR,X,XR,G	100	20 of 20
RP2	Retinitis Pigmentosa	X,G	99.98	-
RP9	Retinitis Pigmentosa	AD	97.78	4 of 4

RPE65	Leber Congenital Amaurosis, Retinitis Pigmentosa	AD,AR	100	231 of 231
RPGR	Cone-Rod Dystrophy, Macular Degeneration, Retinitis Pigmentosa, Deafness, Achromatopsia, Ciliary Dyskinesia	X,XR,G	94	-
RPGRIP1	Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome	AR	99.33	146 of 159
RS1	Retinoschisis	X,XR,G	100	-
SAG	Oguchi Disease, Retinitis Pigmentosa, Night Blindness	AR	100	18 of 18
SCAPER	Intellectual Developmental Disorder, Retinitis Pigmentosa	AR	99.92	17 of 18
SCN3A	Epilepsy, Epileptic Encephalopathy	AD	99.98	18 of 18
SCN8A	Cognitive Impairment, Cerebellar Ataxia, Epileptic Encephalopathy, Myoclonus, Seizures, Infantile Convulsions, Choreoathetosis	AD	97.85	156 of 172
SDCCAG8	Bardet-Biedl Syndrome, Senior-Loken Syndrome	AR	96.29	18 of 19
SEMA4A	Cone Rod Dystrophy, Retinitis Pigmentosa, Colorectal Cancer	AD,AR	99.94	15 of 15
SH3BP2	Cherubism	AD	92	16 of 16
SIX6	Microphthalmia, Optic Disc Anomalies, Retinal Dystrophy, Macular Dystrophy	AD,AR	99.96	8 of 8
SLC13A5	Epileptic Encephalopathy, Amelocerebrohypohidrotic Syndrome	AR	95.92	24 of 24
SLC19A2	Thiamine-Responsive Megaloblastic Anemia Syndrome	AR	99.99	67 of 68
SLC1A2	Epileptic Encephalopathy	AD	100	7 of 7
SLC25A15	Hyperornithinemia, Hyperammonemia, Homocitrullinuria	AR	100	41 of 41
SLC7A14	Retinitis Pigmentosa	AR	99.97	10 of 10
SNRNP200	Retinitis Pigmentosa	AD	100	40 of 40
SPG11	Amyotrophic Lateral Sclerosis, Charcot-Marie-Tooth Disease, Spastic Paraplegia	AR	99.93	289 of 297
STUB1	Spinocerebellar Ataxia	AD,AR	99.93	36 of 36
STXBP1	Epileptic Encephalopathy, 9q33.3q34.11 Microdeletion Syndrome, Atypical Rett Syndrome, Dravet Syndrome, West Syndrome	AD	100	209 of 215
SUMF1	Multiple Sulfatase Deficiency	AR	100	52 of 52
SVBP	Neurodevelopmental Disorder, Ataxia, Hypotonia, And Microcephaly, Intellectual Disability	AR	100	-
SYNGAP1	Mental Retardation, Epileptic Encephalopathy	AD	99.46	168 of 171
SYNJ1	Epileptic Encephalopathy, Parkinson Disease	AR	99.81	30 of 32
SZT2	Epileptic Encephalopathy	AR	99.98	39 of 39
TANC2	Intellectual Developmental Disorder, Autism, Language Delay, Seizures	AD	97.81	21 of 21
TARS1	Trichothiodystrophy	AR	99.94	-
TCF20	Developmental Delay, Intellectual Impairment, Behavioral Abnormalities	AD	100	73 of 73
TEAD1	Sveinsson Chorioretinal Atrophy	AD	100	3 of 3
TMEM67	Bardet-Biedl Syndrome, Coach Syndrome, Joubert Syndrome, Meckel Syndrome, Nephronophthisis, Rhyns Syndrome	AR	96.93	177 of 179
TNFRSF11B	Paget Disease Of Bone, Calcium Pyrophosphate Deposition	AR	99.98	16 of 16
TOPORS	Retinitis Pigmentosa	AD	99.96	24 of 25
TPP1	Ceroid Lipofuscinosis, Spinocerebellar Ataxia	AR	100	147 of 147
TRAF3IP1	Senior-Loken Syndrome	AR	97.54	15 of 15
TRAK1	Epileptic Encephalopathy	AR	99.28	7 of 7
TRAPPC4	Neurodevelopmental Disorder, Epilepsy, Spasticity, Brain Atrophy	AR	100	-
TRMT1	Intellectual Developmental Disorder	AR	99.97	5 of 5
TRNT1	Retinitis Pigmentosa, Erythrocytic Microcytosis, Sideroblastic Anemia, B-Cell Immunodeficiency, Periodic Fevers, Developmental Delay	AR	99.47	22 of 27
TTC21B	Thoracic Dystrophy, Nephronophthisis, Jeune Syndrome, Joubert Syndrome	AD,AR	100	67 of 67

TTC8	Retinitis Pigmentosa, Bardet-Biedl Syndrome	AR	99.33	28 of 28
TUB	Retinal Dystrophy, Obesity, Retinitis Pigmentosa	AR	99.91	4 of 4
TUBB4B	Leber Congenital Amaurosis, Deafness	AD	100	3 of 3
TULP1	Leber Congenital Amaurosis, Retinitis Pigmentosa	AR	99.9	82 of 82
UBA5	Epileptic Encephalopathy, Spinocerebellar Ataxia	AR	99.98	19 of 19
USP45	Leber Congenital Amaurosis	AR	99.08	4 of 5
VCAN	Wagner Syndrome	AD	99.91	11 of 21
VSX1	Craniofacial Anomalies, Keratoconus, Corneal Dystrophy	AD	94.56	25 of 25
WDFY3	Microcephaly, Intellectual Disability	AD	99.95	60 of 60
WDR19	Thoracic Dystrophy, Cranioectodermal Dysplasia, Nephronophthisis, Senior-Loken Syndrome, Jeune Syndrome	AR	99.96	47 of 49
WRN	Werner Syndrome	AR	99.97	107 of 109
WWOX	Epileptic Encephalopathy, Esophageal Cancer, Spinocerebellar Ataxia, Gonadal Dysgenesis, Cerebellar Ataxia	AR	99.94	44 of 44
XYLT1	Desbuquois Dysplasia, Pseudoxanthoma Elasticum, Congenital Disorder Of Glycosylation	AR	92.61	19 of 23
XYLT2	Pseudoxanthoma Elasticum, Spondyloocular Syndrome	AR	99.98	12 of 12
YWHAG	Epileptic Encephalopathy	AD	99.94	5 of 5
ZEB1	Corneal Dystrophy	AD	89.95	63 of 65
ZFYVE26	Spastic Paraplegia	AR	99.95	48 of 48
ZMIZ1	Neurodevelopmental Disorder, Dysmorphic Facies, Distal Skeletal Anomalies	AD	98.87	13 of 13
ZNF408	Exudative Vitreoretinopathy, Retinitis Pigmentosa	AD,AR	99.98	26 of 26
ZNF423	Nephronophthisis, Joubert Syndrome, Oculorenal Defect	AD,AR	100	10 of 10

*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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