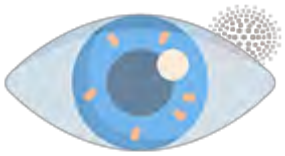
Cone Rod Dystrophy

Precision Panel



Overview

Cone Rod Dystrophies (CRDs) are a clinically and genetically heterogeneous group of inherited retinal diseases characterized by cone photoreceptor degeneration which can lead to rod photoreceptor loss. The main feature of these disorders is progressive loss of central vision, color vision disturbances and light disturbances. There are more than 30 types of cone-rod dystrophies, differentiated by their genetic cause and pattern of inheritance which can be autosomal recessive, autosomal dominant and X-linked. These disorders can occur alone without any other signs and symptoms or they can be associated to a syndrome that affects multiple organs.

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

- Decreased visual acuity
- Photophobia
- Night blindness
- Decreased perception of colors
- Patchy losses of peripheral vision
- Central scotoma

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 to slow down the degenerative process, treating the complications and helping patients to cope with the social and psychological impact of blindness.

- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improve genotype-phenotype correlation associated with these dystrophies.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ABCA4	Cone-Rod Dystrophy, Age-Related Macular Degeneration, Retinitis Pigmentosa, Stargardt Disease	AD,AR	100	1392 of 1430
ABHD12	Polyneuropathy, Hearing Loss, Ataxia, Retinitis Pigmentosa, And Cataract	AR	95.77	21 of 21
ACBD5	Retinal Dystrophy With Leukodystrophy	AR	100	3 of 3
ACOX1	Mitchell Syndrome, Peroxisomal Acyl-CoA Oxidase Deficiency	AD,AR	96.95	22 of 22
ADAM9	Cone Rod Dystrophy	AR	100	10 of 10
ADGRV1	Familial Febrile Convulsions, Usher Syndrome, Generalized Epilepsy With Febrile Seizures-Plus	AD,AR	97.53	-
AHR	Retinitis Pigmentosa	AR	99.91	2 of 2
AIPL1	Leber Congenital Amaurosis, Retinitis Pigmentosa, Cone Rod Dystrophy	AD,AR,X,XR,G	89	82 of 82
ALMS1	Alstrom Syndrome	AR	99.92	302 of 305
AP3B2	Early Infantile Epileptic Encephalopathy	AR	99.95	11 of 12
APOB	Familial Hypobetalipoproteinemia, Homozygous Familial Hypercholesterolemia	AD,AR	99.62	369 of 375
ARL2BP	Retinitis Pigmentosa With Or Without Situs Inversus	AR	99.99	7 of 7
ARL3	Joubert Syndrome, Retinitis Pigmentosa	AD,AR	99.99	4 of 4
ARL6	Bardet-Biedl Syndrome 1, Retinitis Pigmentosa	AD,AR,X,XR,G	100	17 of 21
ASPA	Canavan Disease	AR	99.56	93 of 94
ATF6	Achromatopsia, Cone Rod Dystrophy	AR	99.98	16 of 16
ATP6	Leber Optic Atrophy, Neuropathy, Ataxia, And Retinitis Pigmentosa, Familial Infantile Bilateral Striatal Necrosis, Narp Syndrome	MI	-	-
ATXN2	Spinocerebellar Ataxia, Amyotrophic Lateral Sclerosis	AD	91.78	9 of 10
ATXN7	Spinocerebellar Ataxia With Retinal Degeneration, Macular Degeneration And External Ophthalmoplegia	AD	94.99	-
BBIP1	Bardet-Biedl Syndrome	AR	99.88	1 of 1
BBS1	Bardet-Biedl Syndrome	AR	100	102 of 105
BBS10	Bardet-Biedl Syndrome	AR	100	114 of 114
BBS12	Bardet-biedl Syndrome	AR	99.78	61 of 61
BBS2	Bardet-Biedl Syndrome, Retinitis Pigmentosa	AR	100	99 of 100
BBS4	Bardet-Biedl Syndrome	AR	100	45 of 48
BBS5	Bardet-Biedl Syndrome	AR	99.8	30 of 31
BBS7	Bardet-Biedl Syndrome	AR	100	48 of 48
BBS9	Bardet-Biedl Syndrome	AR	99.56	50 of 51
BCS1L	Bjornstad Syndrome, Gracile Syndrome, Leigh Syndrome, Mitochondrial Complex III Deficiency	AR,MI	99.96	40 of 42
BEST1	Bestrophinopathy, Vitelliform Macular Dystrophy, Retinitis Pigmentosa, Vitreoretinochoroidopathy , Adult-Onset Foveomacular Vitelliform Dystrophy	AD,AR	94.35	342 of 344
C1QTNF5	Late-Onset Retinal Degeneration ,	AD	99.97	7 of 7
C8ORF37	Bardet-Biedl Syndrome, Cone-Rod Dystrophy, Retinitis Pigmentosa	AD,AR,X,XR,G	-	-
CA4	Retinitis Pigmentosa	AD	99.97	11 of 11
CACNA1F	Aland Island Eye Disease, Cone-Rod Dystrophy, Congenital Stationary Night Blindness	X,XR,G	100	-
CACNA2D4	Retinal Cone Dystrophy, Cone Rod Dystrophy, Congenital Stationary Night Blindness	AR	99.64	7 of 7
CC2D2A	Coach Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.43	98 of 100
CCDC103	Primary Ciliary Dyskinesia	AR	99.92	6 of 6
CCDC28B	Bardet-Biedl Syndrome	AR	99.83	1 of 1
CCDC39	Primary Ciliary Dyskinesia	AR	99.56	48 of 52
CCDC40	Primary Ciliary Dyskinesia	AR	98	50 of 50
CCDC65	Primary Ciliary Dyskinesia	AR	99.98	3 of 3
CCNO	Primary Ciliary Dyskinesia	AR	99.94	12 of 12

CDH23	Usher Syndrome	AD,AR	98	400 of 403
CDH3	Eem Syndrome, Hypotrichosis, Congenital, With Juvenile Macular Dystrophy	AR	95	34 of 36
CDHR1	Cone-Rod Dystrophy, Retinitis Pigmentosa	AR	99.67	55 of 55
CEP250	Cone-Rod Dystrophy And Hearing Loss	AR	99.98	7 of 7
CEP290	Bardet-Biedl Syndrome, Joubert Syndrome, Leber Congenital Amaurosis, Meckel Syndrome, Senior-Loken Syndrome	AR	96.47	293 of 327
CEP78	Cone-Rod Dystrophy And Hearing Loss, Usher Syndrome	AR	99.44	9 of 10
CERKL	Retinitis Pigmentosa	AR	100	46 of 46
CFAP221	Primary Ciliary Dyskinesia	-	89.78	-
CFAP298	Primary Ciliary Dyskinesia	AR	-	-
CFAP300	Primary Ciliary Dyskinesia	AR	-	-
CFAP410	Retinal Dystrophy With Or Without Macular Staphyloma, Cone Rod Dystrophy	AR	-	-
CIB2	Usher Syndrome	AR	99.95	16 of 17
CLDN19	Familial Primary Hypomagnesemia With Hypercalciuria And Nephrocalcinosis With Severe Ocular Involvement	AR	99.96	21 of 21
CLN3	Neuronal Ceroid Lipofuscinosis	AR	99.93	73 of 75
CLRN1	Retinitis Pigmentosa, Usher Syndrome	AD,AR,X,XR,G	99.99	40 of 41
CNGA1	Retinitis Pigmentosa	AD,AR,X,XR,G	99.82	36 of 37
CNGA3	Achromatopsia, Cone Rod Dystrophy	AR	99.97	165 of 165
CNGB1	Retinitis Pigmentosa	AR	100	75 of 75
CNNM4	Cone-Rod Dystrophy And Amelogenesis Imperfecta, Jalili Syndrome	AR	96.86	27 of 27
COQ2	Coenzyme Q10 Deficiency, Multiple System Atrophy, Leigh Syndrome With Nephrotic Syndrome	AD,AR	99.61	37 of 38
CRB1	Leber Congenital Amaurosis, Pigmented Paravenous Chorioretinal Atrophy , Retinitis Pigmentosa	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
CTSD	Neuronal Ceroid Lipofuscinosis	AR	100	18 of 18
CWC27	Retinitis Pigmentosa With Or Without Skeletal Anomalies	AR	99.77	8 of 8
DHDDS	Developmental Delay And Seizures With Or Without Movement Abnormalities, Retinitis Pigmentosa	AD,AR	96.32	8 of 8
DHX38	Retinitis Pigmentosa	AR	100	4 of 4
DNAAF1	Primary Ciliary Dyskinesia	AR	99.55	36 of 37
DNAAF2	Primary Ciliary Dyskinesia	AR	97.45	7 of 8
DNAAF3	Primary Ciliary Dyskinesia	AR	98.95	13 of 14
DNAAF4	Primary Ciliary Dyskinesia	AD,AR	99.27	-
DNAAF5	Primary Ciliary Dyskinesia	AR	89.27	-
DNAAF6	Primary Ciliary Dyskinesia	X,XR,G	99.63	-
DNAH1	Primary Ciliary Dyskinesia	AR	100	58 of 58
DNAH11	Primary Ciliary Dyskinesia	AR	99.27	159 of 169
DNAH5	Primary Ciliary Dyskinesia	AR	100	277 of 278
DNAH9	Primary Ciliary Dyskinesia	AR	98.86	19 of 19
DNAI1	Kartagener Syndrome, Primary Ciliary Dyskinesia	AR	96.91	43 of 43
DNAI2	Primary Ciliary Dyskinesia	AR	98.89	8 of 8
DNAJB13	Primary Ciliary Dyskinesia	AR	99.94	3 of 3
DNAL1	Primary Ciliary Dyskinesia	AR	99.43	5 of 5
DRAM2	Cone Rod Dystrophy	AR	99.87	13 of 13
DRC1	Primary Ciliary Dyskinesia	AR	100	9 of 9
DYNC2I2	Jeune Syndrome, Short Rib-Polydactyly Syndrome	AR	99.54	23 of 23
EXOSC2	Short Stature, Hearing Loss, Retinitis Pigmentosa, And Distinctive Facies	AR	100	3 of 3
EYS	Retinitis Pigmentosa	AR	99.54	358 of 379
FAM161A	Retinitis Pigmentosa	AR	99.74	22 of 23
FDXR	Auditory Neuropathy And Optic Atrophy	AR	99.93	23 of 23
FLVCR1	Posterior Column Ataxia With Retinitis Pigmentosa	AR	99.96	26 of 26
FOXJ1	Primary Ciliary Dyskinesia	AD	99.69	5 of 5
FSCN2	Retinitis Pigmentosa	AD	98.93	16 of 17
GAS2L2	Primary Ciliary Dyskinesia	AR	89	4 of 5
GAS8	Primary Ciliary Dyskinesia	AR	99.98	6 of 6
GATA3	Hypoparathyroidism-Sensorineural Deafness-Renal Disease Syndrome	AD	100	81 of 81
GGCX	Pseudoxanthoma Elasticum-Like Skin Manifestations With Retinitis Pigmentosa	AR	100	62 of 62

GUCA1A	Cone Dystrophy, Central Areolar Choroidal Dystrophy, Cone Rod Dystrophy	AD	99.94	27 of 27
GUCA1B	Retinitis Pigmentosa	AD	100	10 of 10
GUCY2D	Central Areolar Choroidal Dystrophy, Cone-Rod Dystrophy, Leber Congenital Amaurosis, Night Blindness, Congenital Stationary	AD,AR	99.98	248 of 248
HGSNAT	Mucopolysaccharidosis Type IIIc, Retinitis Pigmentosa	AR	87.91	69 of 73
HK1	Neurodevelopmental Disorder With Visual Defects And Brain Anomalies, Retinitis Pigmentosa	AD,AR	100	14 of 17
HMX1	Oculoauricular Syndrome	AR	85.58	2 of 2
HSPD1	Autosomal Recessive Hypomyelinating Leukodystrophy	AD,AR	100	7 of 7
HYDIN	Primary Ciliary Dyskinesia	AR	81.7	45 of 63
IDH3B	Retinitis Pigmentosa	AR	100	5 of 5
IFT140	Retinitis Pigmentosa, Jeune Syndrome, Leber Congenital Amaurosis	AR	99.97	81 of 81
IFT172	Retinitis Pigmentosa, Bardet-Biedl Syndrome, Jeune Syndrome	AR	100	37 of 37
IFT27	Bardet-Biedl Syndrome	AR	100	5 of 5
IFT74	Bardet-Biedl Syndrome	AR	99.95	6 of 6
IMPDH1	Leber Congenital Amaurosis, Retinitis Pigmentosa	AD	99.98	29 of 29
IMPG2	Vitelliform Macular Dystrophy, Retinitis Pigmentosa	AD,AR	99.7	46 of 46
IQCB1	Senior-Loken Syndrome, Leber Congenital Amaurosis	AR	99.98	43 of 43
KCNV2	Retinal Cone Dystrophy	AR	99.98	86 of 88
KIF3B	Retinitis Pigmentosa	AD	99.92	-
KIF5A	Autosomal Dominant Spastic Paraplegia	AD	100	85 of 85
KIZ	Retinitis Pigmentosa	AR	na	-
KLHL7	Retinitis Pigmentosa, Bohring-Opitz Syndrome	AD,AR	98.69	19 of 19
KNTC1	Orbital Plasma Cell Granuloma, Chronic Orbital Inflammation, Mikulicz Disease	-	99.89	-
LRAT	Leber Congenital Amaurosis, Retinitis Pigmentosa	AD,AR,X,XR,G	100	25 of 25
LRRC56	Primary Ciliary Dyskinesia	AR	99.77	5 of 5
LRRC6	Primary Ciliary Dyskinesia	AR	99.88	21 of 21
LZTFL1	Bardet-Biedl Syndrome	AR	99.83	4 of 4
MAK	Retinitis Pigmentosa	AR	100	28 of 28
MAPKAPK3	Macular Dystrophy	AD	99.98	2 of 2
MCIDAS	Primary Ciliary Dyskinesia	AR	99.92	4 of 4
MDH2	Early Infantile Epileptic Encephalopathy	AR	98	11 of 11
MERTK	Retinitis Pigmentosa	AR	100	99 of 101
MFRP	Posterior Microphthalmia With Retinitis Pigmentosa, Foveoschisis And Optic Disc Drusen, Nanophthalmos	AR	100	36 of 36
MKKS	Bardet-Biedl Syndrome, Mckusick-Kaufman Syndrome	AR	89.96	71 of 71
MKS1	Bardet-Biedl Syndrome, Joubert Syndrome, Meckel Syndrome	AR	99.98	49 of 49
MTTP	Abdominal Obesity-Metabolic Syndrome, Abetalipoproteinemia	AD,AR	100	69 of 71
MVK	Mevalonic Aciduriamevalonate Kinase Deficiency	AD,AR	100	180 of 181
MYO6	Autosomal Dominant and Recessive Deafness	AD,AR	100	74 of 75
MYO7A	Autosomal Dominant Deafness, Usher Syndrome	AD,AR	100	579 of 580
ND1	Leber Optic Atrophy, Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Isolated Complex I Deficiency	MI	-	-
ND2	Leber Optic Atrophy, Isolated Complex I Deficiency, Mitochondrial DNA-Associated Leigh Syndrome	MI	85.56	-
ND3	Isolated Complex I Deficiency, Mitochondrial DNA-Associated Leigh Syndrome		99.99	-
ND4	Leber Optic Atrophy, Leber Hereditary Optic Neuropathy, Mitochondrial DNA-Associated Leigh Syndrome	MI	-	-
ND5	Leber Optic Atrophy, Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes	MI	99.89	-
ND6	Leber Optic Atrophy, Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes	MI	100	-
NDUFA9	Mitochondrial Complex I Deficiency, Leigh Syndrome With Leukodystrophy	AR	99.98	3 of 3
NEK10	Primary Ciliary Dyskinesia	AR	99.95	3 of 3
NEK2	Retinitis Pigmentosa	AR	99.94	5 of 5
NGLY1	Congenital Disorder Of Glycosylation Type IV, Alacrimia-Choreoathetosis-Liver Dysfunction Syndrome	AR	99.8	28 of 28
NME8	Primary Ciliary Dyskinesia	AR	99.99	9 of 9
NMNAT1	Cone Rod Dystrophy, Leber Congenital Amaurosis	AR	98.94	72 of 75
NPHP1	Joubert Syndrome, Senior-Loken Syndrome, Bardet-Biedl Syndrome	AR	100	58 of 59
NPHP4	Senior-Loken Syndrome	AR	99.96	118 of 119

NR2E3	Enhanced S-Cone Syndrome, Retinitis Pigmentosa	AD,AR	-	-
NRL	Retinitis Pigmentosa	AD	99.81	25 of 25
ODAD1	Primary Ciliary Dyskinesia	AR	99.68	10 of 10
ODAD2	Primary Ciliary Dyskinesia	AR	97.3	26 of 28
ODAD3	Primary Ciliary Dyskinesia	AR	95	4 of 4
ODAD4	Primary Ciliary Dyskinesia	AR	-	-
OFD1	Joubert Syndrome, Orofaciodigital Syndrome, Retinitis Pigmentosa, Simpson-Golabi-Behmel Syndrome, Primary Ciliary Dyskinesia	X,XR,XD,G	98.09	-
OPN1LW	Blue Cone Monochromacy, Colorblindness, Cone Rod Dystrophy	X,XR,G	88	-
OPN1MW	Blue Cone Monochromacy, Colorblindness, Cone Rod Dystrophy	X,XR,G	41.73	-
PANK2	Hypoprebetalipoproteinemia, Acanthocytosis, Retinitis Pigmentosa And Pallidal Degeneration , Classic Pantothenate Kinase-Associated Neurodegeneration	AR	98.92	177 of 182
PCARE	Retinitis Pigmentosa	AR	-	-
PCDH15	Usher Syndrome	AR	99.36	152 of 158
PCYT1A	Spondylometaphyseal Dysplasia With Cone-Rod Dystrophy, Leber Congenital Amaurosis	AR	99.98	22 of 22
PDE6A	Retinitis Pigmentosa	AR	100	75 of 75
PDE6B	Retinitis Pigmentosa, Congenital Stationary Night Blindness	AD,AR	100	156 of 156
PDE6C	Cone Dystrophy, Achromatopsia, Progressive Cone Dystrophy	AR	100	63 of 63
PDE6G	Retinitis Pigmentosa	AD,AR,X,XR,G	100	2 of 2
PDE6H	Retinal Cone Dystrophy, Achromatopsia	AD,AR	100	2 of 2
PDZD7	Usher Syndrome	AR	100	28 of 28
PEX1	Hearing Loss, Sensorineural, With Enamel Hypoplasia And Nail Defects, Peroxisome Biogenesis Disorder, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	97.02	126 of 134
PEX10	Peroxisome Biogenesis Disorder, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	99.76	29 of 32
PEX11B	Peroxisome Biogenesis Disorder, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	90.29	7 of 7
PEX12	Peroxisome Biogenesis Disorder, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	100	38 of 38
PEX13	Peroxisome Biogenesis Disorder, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	99.98	11 of 12
PEX14	Peroxisome Biogenesis Disorder, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	100	4 of 4
PEX16	Peroxisome Biogenesis Disorder, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	100	17 of 17
PEX19	Peroxisome Biogenesis Disorder, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	100	5 of 5
PEX2	Peroxisome Biogenesis Disorder, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	99.89	17 of 17
PEX26	Peroxisome Biogenesis Disorder, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	100	29 of 29
PEX3	Peroxisome Biogenesis Disorder, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	100	9 of 9
PEX5	Adrenoleukodystrophy, Cerebrohepatorenal, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AR	100	12 of 12
PEX6	Heimler Syndrome, Peroxisome Biogenesis Disorder, Autosomal Recessive Spinocerebellar Ataxia-Blindness-Deafness Syndrome, Infantile Refsum Disease, Neonatal Adrenoleukodystrophy	AD,AR	99.94	105 of 108
PEX7	Peroxisome Biogenesis Disorder, Refsum Disease	AR	99.21	47 of 53
PHYH	Refsum Disease	AR	100	34 of 34
PITPNM3	Cone-Rod Dystrophy	AD	99.8	7 of 7
PMM2	Congenital Disorder Of Glycosylation Type Ia	AR	100	127 of 129
POC1B	Cone Rod Dystrophy	AR	99.87	10 of 10
POGZ	White-Sutton Syndrome, Intellectual Disability-Microcephaly-Strabismus-Behavioral Abnormalities Syndrome	AD	99.97	85 of 85
PPP2R3C	Gonadal Dysgenesis, Dysmorphic Facies, Retinal Dystrophy, And Myopathy	AD,AR	99.85	3 of 3
PRCD	Retinitis Pigmentosa	AR	100	7 of 7
PROM1	Cone-Rod Dystrophy, Macular Dystrophy, Retinal, Retinitis Pigmentosa, Stargardt Disease, Cone Rod Dystrophy	AD,AR	99.61	90 of 93
PRPF3	Retinitis Pigmentosa	AD	100	8 of 9

PRPF31	Retinitis Pigmentosa	AD	100	160 of 166
PRPF4	Retinitis Pigmentosa	AD	99.99	5 of 5
PRPF6	Retinitis Pigmentosa	AD	100	14 of 14
PRPF8	Retinitis Pigmentosa	AD	100	58 of 58
PRPH2	Central Areolar Choroidal Dystrophy, Fundus Albipunctatusretinitis Punctata Albescens, Vitelliform Macular Dystrophy, Patterned Dystrophy Of Retinal Pigment Epithelium, Retinitis Pigmentosa, Adult-Onset Foveomacular Vitelliform Dystrophy, Retinitis Punctata Albescens, Stargardt Disease	AD,AR	100	188 of 188
PRPS1	Arts Syndrome, Phosphoribosylpyrophosphate Synthetase Superactivity, Lethal Ataxia With Deafness And Optic Atrophy	X,XR,G	100	-
RAB28	Cone Rod Dystrophy	AR	100	6 of 6
RAX2	Age-Related Macular Degeneration, Cone Rod Dystrophy	AD	99.89	7 of 9
RBP3	Retinitis Pigmentosa	AD,AR,X,XR,G	100	17 of 17
RDH11	Retinal Dystrophy, Juvenile Cataracts, And Short Stature Syndrome	AR	99.97	3 of 3
REEP6	Retinitis Pigmentosa	AR	97.59	9 of 9
RGR	Retinitis Pigmentosa	AD,AR	100	9 of 9
RHO	Fundus Albipunctatusretinitis Punctata Albescens, Congenital Stationary Night Blindness, Retinitis Pigmentosa	AD,AR	100	229 of 229
RIMS1	Cone Rod Dystrophy	AD	98.2	24 of 24
RLBP1	Bothnia Retinal Dystrophy, Fundus Albipunctatusretinitis Punctata Albescens, Newfoundland Rod-Cone Dystrophy, Retinitis Pigmentosa	AD,AR	100	32 of 33
ROM1	Retinitis Pigmentosa	AD,AR,X,XR,G	100	20 of 20
RP1	Retinitis Pigmentosa	AD,AR	99.95	215 of 218
RP2	Retinitis Pigmentosa	X,G	99.98	-
RP9	Retinitis Pigmentosa	AD	97.78	4 of 4
RPE65	Leber Congenital Amaurosis, Retinitis Pigmentosa, Leber Congenital Amaurosis	AD,AR	100	231 of 231
RPGR	Cone-Rod Dystrophy, X-linked Atrophic Macular Degeneration, Retinitis Pigmentosa, And Sinorespiratory Infections, Withor Without Deafness, Achromatopsia, Cone Rod Dystrophy, Primary Ciliary Dyskinesia	X,XR,G	94	-
RPGRIP1	Cone-Rod Dystrophy, Leber Congenital Amaurosis, Meckel Syndrome	AR	99.33	146 of 159
RPL10	X-linked Intellectual Disability-Cerebellar Hypoplasia-Spondylo-Epiphyseal Dysplasia Syndrome	X,XR,G	100	-
RSPH1	Primary Ciliary Dyskinesia	AR	100	10 of 10
RSPH3	Primary Ciliary Dyskinesia	AR	99.85	5 of 5
RSPH4A	Primary Ciliary Dyskinesia	AR	99.98	27 of 27
RSPH9	Primary Ciliary Dyskinesia	AR	100	13 of 13
SAG	Oguchi Disease, Retinitis Pigmentosa, Congenital Stationary Night Blindness	AR	100	18 of 18
SDCCAG8	Bardet-Biedl Syndrome, Senior-Loken Syndrome	AR	96.29	18 of 19
SEMA4A	Cone-Rod Dystrophy, Retinitis Pigmentosa	AD,AR	99.94	15 of 15
SH2B1	Distal 16p11.2 Microdeletion Syndrome, Proximal 16p11.2 Microdeletion Syndrome	-	99.98	25 of 25
SLC19A2	Thiamine-Responsive Megaloblastic Anemia Syndrome	AR	99.99	67 of 68
SLC35A2	Congenital Disorder Of Glycosylation	X,XD,G	99.97	-
SLC7A14	Retinitis Pigmentosa	AR	99.97	10 of 10
SNRNP200	Retinitis Pigmentosa	AD	100	40 of 40
SPAG1	Primary Ciliary Dyskinesia	AR	94.8	11 of 12
SPEF2	Primary Ciliary Dyskinesia	AR	99.6	10 of 13
SRD5A3	Congenital Disorder Of Glycosylation, Kahrizi Syndrome	AR	100	15 of 15
STK36	Primary Ciliary Dyskinesia		100	5 of 5
TELO2	You-Hoover-Fong Syndrome, Telo2-related Intellectual Disability-neurodevelopmental Disorder	AR	99.98	8 of 8
TMEM67	Bardet-Biedl Syndrome, Coach Syndrome, Joubert Syndrome, Meckel Syndrome, Rhyns Syndrome	AR	96.93	177 of 179
TOPORS	Retinitis Pigmentosa	AD	99.96	24 of 25
TRAF3IP1	Senior-Loken Syndrome	AR	97.54	15 of 15
TRNK	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Myoclonic Epilepsy Associated With Ragged-Red Fibers, Mitochondrial DNA-Associated Leigh Syndrome	MI	-	-
TRNL1	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Myoclonic Epilepsy Associated With Ragged-Red Fibers, Kearns-Sayre Syndrome, Mitochondrial DNA-Associated Leigh Syndrome	MI	-	-

TRNT1	Retinitis Pigmentosa And Erythrocytic Microcytosis	AR	99.47	22 of 27
TRNV	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Mitochondrial DNA-Associated Leigh Syndrome	MI	-	-
TRNW	Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-like Episodes, Mitochondrial Myopathy, Episodic, With Optic Atrophy And Reversible Leukoencephalopathy, Mitochondrial DNA-Associated Leigh Syndrome	AR,MI	-	-
TTC12	Primary Ciliary Dyskinesia	AR	99.97	-
TTC8	Bardet-Biedl Syndrome, Retinitis Pigmentosa	AR	99.33	28 of 28
TTLL5	Cone Rod Dystrophy	AR	99.95	15 of 15
TULP1	Leber Congenital Amaurosis, Retinitis Pigmentosa	AR	99.9	82 of 82
UNC119	Immunodeficiency, Cone Rod Dystrophy	AD	100	6 of 6
USH1C	Deafness, Neurosensory, Autosomal Recessive, Usher Syndrome	AR	99.97	79 of 79
USH1G	Usher Syndrome	AR	100	35 of 35
USH2A	Retinitis Pigmentosa, Usher Syndrome, Retinitis Pigmentosa	AR	100	1286 of 1314
WARS2	Neurodevelopmental Disorder, Mitochondrial, With Abnormal Movements And Lactic Acidosis, With Or Without Seizures, Wars2-Related Combined Oxidative Phosphorylation Defect	AR	99.95	14 of 15
WDR19	Senior-Loken Syndrome, Jeune Syndrome	AR	99.96	47 of 49
WHRN	Usher Syndrome	AR	99.94	NA-
ZMYND10	Primary Ciliary Dyskinesia	AR	99.98	16 of 16
ZNF408	Exudative Vitreoretinopathy, Retinitis Pigmentosa	AD,AR	99.98	26 of 26
ZNF513	Retinitis Pigmentosa	AR	99.97	3 of 3

*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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