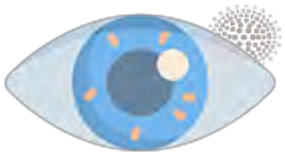
Retinitis Pigmentosa

Precision Panel



Overview

Retinitis Pigmentosa (RP) comprises a complex group of inherited dystrophies characterized by degeneration and dysfunction of the retina, affecting photoreceptor and pigment epithelial function. RP can be an isolated finding or be part of a syndrome that can be inherited in a dominant, recessive or X-linked pattern. This disease presents as progressive loss of night and peripheral vision, leading to a constricted visual field and markedly diminished vision. The clinical presentation of these findings is highly variable, some patients being affected during childhood while others are asymptomatic well into adulthood. There is an increase in mortality rate due to psychiatric comorbidities.

The Igenomix Retinitis Pigmentosa 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 Retinitis Pigmentosa Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- Family history of RP
- Night blindness
- Progressive constriction of the visual field, usually peripheral
- Cataracts
- Sensation of sparking lights (photopsias)
- Headache

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 multidisciplinary treatment in the form of medical care with vitamin A and other antioxidants and surgical care for potential cataract extraction or retinal prosthesis.

- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Detect novel disease-causing genes and novel variant in disease-causing genes.

Genes & Diseases

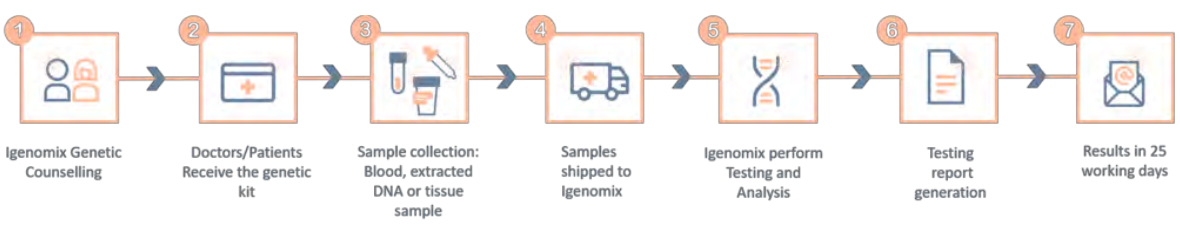
GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ABCA4	Cone-Rod Dystrophy, Macular Degeneration, Retinitis Pigmentosa, Stargardt Disease	AD,AR	100	1392 of 1430
ABHD12	Polyneuropathy, Hearing Loss, Ataxia, Retinitis Pigmentosa, Cataract	AR	95.77	21 of 21
AGBL5	Retinitis Pigmentosa	AR	99.97	9 of 9
AHI1	Joubert Syndrome, Ocular Defect, Retinitis Pigmentosa	AR	96.79	85 of 97
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
AMACR	Alpha-Methylacyl-Coa Racemase Deficiency, Bile Acid Synthesis Defect	AR	100	8 of 8
ARHGEF18	Retinitis Pigmentosa	AR	99.95	6 of 6
ARL2BP	Retinitis Pigmentosa, Situs Inversus	AR	99.99	7 of 7
ARL3	Joubert Syndrome, Retinitis Pigmentosa	AD,AR	99.99	4 of 4
ARL6	Bardet-Biedl Syndrome, Retinitis Pigmentosa	AD,AR,X,XR,G	100	17 of 21
ATP6	Leber Optic Atrophy, Neuropathy, Ataxia, Retinitis Pigmentosa, Bilateral Striatal Necrosis, Leigh Syndrome, Spastic Paraplegia, Narp Syndrome	MI	-	-
BBS2	Bardet-Biedl Syndrome, Retinitis Pigmentosa	AR	100	99 of 100
BEST1	Bestrophinopathy, Macular Dystrophy, Retinitis Pigmentosa, Vitreoretinochoroidopathy, Best Vitelliform Macular Dystrophy, Nanophthalmos	AD,AR	94.35	342 of 344
C8ORF37	Bardet-Biedl Syndrome, Cone-Rod Dystrophy, Retinitis Pigmentosa	AD,AR,X,XR,G	-	-
CA4	Retinitis Pigmentosa	AD	99.97	11 of 11
CDHR1	Cone-Rod Dystrophy, Retinitis Pigmentosa	AR	99.67	55 of 55
CERKL	Retinitis Pigmentosa	AR	100	46 of 46
CLCC1	Retinitis Pigmentosa	AR	97.97	-
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
CNGB1	Retinitis Pigmentosa	AR	100	75 of 75
CRB1	Leber Congenital Amaurosis, Pigmented Paravenous 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
CWC27	Retinitis Pigmentosa, Skeletal Anomalies	AR	99.77	8 of 8
DHDDS	Developmental Delay, Seizures, Movement Abnormalities, Retinitis Pigmentosa, Epileptic Encephalopathy	AD,AR	96.32	8 of 8
DHX38	Retinitis Pigmentosa	AR	100	4 of 4
EXOSC2	Short Stature, Hearing Loss, Retinitis Pigmentosa, 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
FLVCR1	Posterior Column Ataxia, Retinitis Pigmentosa	AR	99.96	26 of 26
FSCN2	Retinitis Pigmentosa	AD	98.93	16 of 17

GGCX	Pseudoxanthoma Elasticum-Like Disorder, Coagulation Factor Deficiency, Body Skin Hyperlaxity, Vitamin K-Dependent Coagulation Factor Deficiency, Retinitis Pigmentosa	AR	100	62 of 62
GUCA1B	Retinitis Pigmentosa	AD	100	10 of 10
HGSNAT	Mucopolysaccharidosis, Retinitis Pigmentosa	AR	87.91	69 of 73
HK1	Hemolytic Anemia, Hexokinase Deficiency, Neurodevelopmental Disorder, Visual Defects, Brain Anomalies, Neuropathy, Retinitis Pigmentosa, Charcot-Marie-Tooth Disease	AD,AR	100	14 of 17
IDH3A	Retinitis Pigmentosa	-	100	9 of 9
IDH3B	Retinitis Pigmentosa	AR	100	5 of 5
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
IFT43	Cranioectodermal Dysplasia, Retinitis Pigmentosa, Short-Rib Thoracic Dysplasia, Polydactyly	AR	100	6 of 6
IFT88	Retinitis Pigmentosa	-	99.46	6 of 6
IMPDH1	Leber Congenital Amaurosis, Retinitis Pigmentosa	AD	99.98	29 of 29
IMPG2	Macular Dystrophy, Retinitis Pigmentosa, Foveomacular Vitelliform Dystrophy	AD,AR	99.7	46 of 46
KIAA1549	Retinitis Pigmentosa	AR	96.67	9 of 10
KIF3B	Retinitis Pigmentosa	AD	99.92	-
KIZ	Retinitis Pigmentosa	AR	-	-
KLHL7	Crisponi/Cold-Induced Sweating Syndrome, Retinitis Pigmentosa, Bohring-Opitz Syndrome	AD,AR	98.69	19 of 19
LRAT	Leber Congenital Amaurosis, Retinitis Pigmentosa	AD,AR,X,XR,G	100	25 of 25
MAK	Retinitis Pigmentosa	AR	100	28 of 28
MERTK	Retinitis Pigmentosa	AR	100	99 of 101
MFRP	Microphthalmia, Retinitis Pigmentosa, Foveoschisis, Optic Disc Drusen, Nanophthalmos	AR	100	36 of 36
NEK2	Retinitis Pigmentosa	AR	99.94	5 of 5
NR2E3	S-Cone Syndrome, Retinitis Pigmentosa	AD,AR	-	-
NRL	Retinitis Pigmentosa	AD	99.81	25 of 25
OFD1	Joubert Syndrome, Orofaciodigital Syndrome, Retinitis Pigmentosa, Simpson-Golabi-Behmel Syndrome, Primary Ciliary Dyskinesia	X,XR,XD,G	98.09	NA of NA
PANK2	Hypoprebetalipoproteinemia, Acanthocytosis, Retinitis Pigmentosa, Pallidal Degeneration, Neurodegeneration, Brain Iron Accumulation	AR	98.92	177 of 182
PCARE	Retinitis Pigmentosa	AR	-	-
PDE6A	Retinitis Pigmentosa	AR	100	75 of 75
PDE6B	Night Blindness, Retinitis Pigmentosa	AD,AR	100	156 of 156
PDE6G	Retinitis Pigmentosa	AD,AR,X,XR,G	100	2 of 2
POMGNT1	Limb Girdle Muscular Dystrophy, Retinitis Pigmentosa, Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	99.91	82 of 83
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
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	Choroidal Dystrophy, Retinitis Punctata Albescens, Macular Dystrophy, Retinitis Pigmentosa, Foveomacular Vitelliform Dystrophy, Cone Rod Dystrophy, Stargardt Disease	AD,AR	100	188 of 188
RBP3	Retinitis Pigmentosa	AD,AR,X,XR,G	100	17 of 17
RDH11	Retinal Dystrophy, Retinitis Pigmentosa, Cataract, Short Stature, Intellectual Disability	AR	99.97	3 of 3
RDH12	Leber Congenital Amaurosis, Retinitis Pigmentosa	AD,AR	100	122 of 122
REEP6	Retinitis Pigmentosa	AR	97.59	9 of 9
RGR	Retinitis Pigmentosa	AD,AR	100	9 of 9
RHO	Retinitis Punctata Albescens, Night Blindness, Retinitis Pigmentosa	AD,AR	100	229 of 229
RLBP1	Bothnia Retinal Dystrophy, Cone-Rod Dystrophy, Retinitis Pigmentosa, Retinitis Punctata Albescens	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
RP1L1	Macular Dystrophy, Retinitis Pigmentosa	AD,AR	99.98	56 of 56
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	Macular Degeneration, Retinitis Pigmentosa, Sinorespiratory Infections, Deafness, Achromatopsia, Cone Rod Dystrophy, Dyskinesia	X,XR,G	94	-
SAG	Oguchi Disease, Retinitis Pigmentosa, Night Blindness	AR	100	18 of 18
SCAPER	Intellectual Developmental Disorder, Retinitis Pigmentosa	AR	99.92	17 of 18
SEMA4A	Cone Rod Dystrophy, Colorectal Cancer, Retinitis Pigmentosa	AD,AR	99.94	15 of 15
SLC7A14	Retinitis Pigmentosa	AR	99.97	10 of 10
SNRNP200	Retinitis Pigmentosa	AD	100	40 of 40
SPATA7	Leber Congenital Amaurosis, Retinitis Pigmentosa	AR	97.02	43 of 43
TOPORS	Retinitis Pigmentosa	AD	99.96	24 of 25
TRNT1	Retinitis Pigmentosa, Erythrocytic Microcytosis, Sideroblastic Anemia, B-Cell Immunodeficiency	AR	99.47	22 of 27
TTC8	Bardet-Biedl Syndrome, Retinitis Pigmentosa	AR	99.33	28 of 28
TUB	Retinal Dystrophy, Obesity, Retinitis Pigmentosa	AR	99.91	4 of 4
TULP1	Retinitis Pigmentosa, Leber Congenital Amaurosis	AR	99.9	82 of 82
USH2A	Retinitis Pigmentosa, Usher Syndrome	AR	100	1286 of 1314
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



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