ID YOUR IRD® Testing Panel

The only way to confirm that your patient has an inherited retinal disease (IRD) is with a genetic test. Through the **ID YOUR IRD gene testing initiative**, Spark Therapeutics supports people living with certain IRDs by providing genetic information that can empower their decisions. The ID YOUR IRD panel tests for mutations in **over 325 genes** associated with IRDs.

Visit **IDYOURIRD.com** for more information or to order free* test kits.

ID YOUR IRD®

INHERITED RETINAL DISEASE GENE TESTING INITIATIVE

GENE	DISEASE CATEGORY†
ABCA4	Stargardt disease, autosomal recessive Cone or cone-rod dystrophy, autosomal recessive[1,2]
ABCC6‡	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
ABHD12	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
ACBD5	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
ACO2	Optic atrophy, autosomal recessive Infantile cerebellar-retinal degeneration, autosomal recessive[1,2]
ADAM9	Cone or cone-rod dystrophy, autosomal recessive[1,2]
ADAMTS18	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
ADAMTSL4	Isolated ectopia lentis, autosomal recessive
ADGRA3§	Retinitis pigmentosa, autosomal recessive[1,2]
ADGRV1	Usher syndrome, autosomal recessive[1,2]
ADIPOR1	Retinitis pigmentosa, autosomal dominant Bardet-Biedl syndrome, autosomal recessive[1,2]
AGBL5	Retinitis pigmentosa, autosomal recessive[1,2]
AHI1	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
AHR	Retinitis pigmentosa, autosomal recessive[1,2]
AIPL1	Cone or cone-rod dystrophy, autosomal dominant Leber congenital amaurosis, autosomal recessive[1,2]
ALMS1	Syndromic/systemic diseases with retinopathy, autosomal recessive Alstrom syndrome, autosomal recessive[1,2]
ARHGEF18	Retinitis pigmentosa, autosomal recessive[1,2]
ARL13B	Joubert syndrome, autosomal recessive[2]
ARL2BP	Retinitis pigmentosa, autosomal recessive[1,2]
ARL3	Retinitis pigmentosa, autosomal dominant[1,2]
ARL6	Retinitis pigmentosa, autosomal recessive Bardet-Biedl syndrome, autosomal recessive[1,2]
ARMC9‡	Joubert syndrome, autosomal recessive[2]
ARSG	Usher syndrome, autosomal recessive[1,2]
ASRGL1	Other retinopathy, autosomal recessive[1,2]
ATF6	Cone or cone-rod dystrophy, autosomal recessive[1,2]
ATOH7	Nonsyndromal congenital retinal nonattachment, autosomal recessive (aka *RNANC*)[2]
B9D1	Joubert syndrome, autosomal recessive Meckel syndrome, autosomal recessive[1,2]
BBIP1	Retinitis pigmentosa, autosomal recessive Bardet-Biedl syndrome, autosomal recessive[1,2]
BBS1	Retinitis pigmentosa, autosomal recessive Bardet-Biedl syndrome, autosomal recessive[1,2]
BBS2	Retinitis pigmentosa, autosomal recessive Bardet-Biedl syndrome, autosomal recessive[1,2]
BBS4	Bardet-Biedl syndrome, autosomal recessive[1,2]
BBS5	Bardet-Biedl syndrome, autosomal recessive[1,2]
BBS7	Bardet-Biedl syndrome, autosomal recessive[1,2]
BBS9‡	Bardet-Biedl syndrome, autosomal recessive[1,2]
BBS10	Bardet-Biedl syndrome, autosomal recessive[1,2]
BBS12	Bardet-Biedl syndrome, autosomal recessive[1,2]
BEST1	Vitelliform macular degeneration (Best disease), autosomal dominant and recessive Retinitis pigmentosa, autosomal dominant and recessive[1,2]
C1QTNF5	Macular dystrophy, autosomal dominant[1,2]
C8ORF37	Bardet-Biedl syndrome, autosomal recessive Cone-rod dystrophy, autosomal recessive[1,2]
C12ORF65	Hereditary spastic paraplegia 55, autosomal recessive Combined oxidative phosphorylation deficiency 7, autosomal recessive
CA4	Retinitis pigmentosa, autosomal dominant[1,2]
CABP4	Congenital stationary night blindness, autosomal recessive[1,2]
CACNA1F	Cone or cone-rod dystrophy, X-linked[1,2]
CACNA2D4	Cone or cone-rod dystrophy, autosomal recessive[1,2]
CAPN5	Other retinopathy, autosomal dominant[1,2]
CC2D2A	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
CCT2	Leber congenital amaurosis, autosomal recessive[1,2]
CDH3	Other retinopathy, autosomal recessive[1,2]
CDH23	Usher syndrome, autosomal recessive and digenic recessive[1,2]
CDHR1	Cone or cone-rod dystrophy, autosomal recessive[1,2]
CEP19	Bardet-Biedl syndrome, autosomal recessive[1,2]
CEP41	Joubert syndrome, autosomal recessive[2]
CEP78	Cone or cone-rod dystrophy with hearing loss, autosomal recessive[1,2]
CEP83	Infantile nephronophthisis with occasional retinopathy[2]
CEP164	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
CEP250	Usher syndrome, autosomal recessive[1,2]
CEP290	Leber congenital amaurosis, autosomal recessive Joubert syndrome, autosomal recessive[1,2]
CERKL	Cone or cone-rod dystrophy, autosomal recessive Retinitis pigmentosa, autosomal recessive[1,2]
CFAP410§	Cone or cone-rod dystrophy, autosomal recessive[1,2]
CHM	Choroideremia, X-linked[1,2]
CIB2	Deafness alone or syndromic, autosomal recessive[1,2]
CISD2	Syndromic/systemic diseases with optic atrophy, autosomal recessive Wolfram syndrome, autosomal recessive[1,2]
CLCC1	Retinitis pigmentosa, autosomal recessive[1,2]

GENE	DISEASE CATEGORY†
CLN3	Juvenile neuronal ceroid lipofuscinosis, autosomal recessive[1,2]
CLN5	Neuronal ceroid lipofuscinosis type 5, autosomal recessive
CLN6	Neuronal ceroid lipofuscinosis type 6, autosomal recessive
CLN8	Neuronal ceroid lipofuscinosis type 8, autosomal recessive
CLRN1	Retinitis pigmentosa, autosomal recessive Usher syndrome, autosomal recessive[1,2]
CLUAP1	Leber congenital amaurosis, autosomal recessive[1,2]
CNGA1	Retinitis pigmentosa, autosomal recessive[1,2]
CNGA3	Achromatopsia, autosomal recessive Cone or cone-rod dystrophy, autosomal recessive[1,2]
CNGB1	Retinitis pigmentosa, autosomal recessive[1,2]
CNGB3	Achromatopsia, autosomal recessive Cone or cone-rod dystrophy, autosomal recessive[1,2]
CNNM4	Cone or cone-rod dystrophy, autosomal recessive Jalili syndrome, autosomal recessive[1,2]
COL11A1‡	Stickler syndrome, autosomal dominant Marshall syndrome, autosomal dominant[1,2]
COL11A2	Stickler syndrome, autosomal dominant[1,2]
COL18A1	Knobloch syndrome, autosomal recessive[1,2]
COL2A1	Stickler syndrome, autosomal dominant[1,2]
COL9A1	Stickler syndrome, autosomal recessive[1,2]
COL9A2	Stickler syndrome, autosomal recessive[1,2]
COL9A3	Stickler syndrome, autosomal recessive[1,2]
CPLANE1	Joubert syndrome, autosomal recessive[1,2]
CRB1	Leber congenital amaurosis, autosomal recessive Retinitis pigmentosa, autosomal recessive[1,2]
CRX	Leber congenital amaurosis, autosomal recessive[1,2]
CSPP1	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
CTNNA1	Macular dystrophy, autosomal dominant[1,2]
CTSD	Neuronal ceroid lipofuscinosis type 10, autosomal recessive
CWC27	Retinitis pigmentosa with or without skeletal abnormalities[2]
CYP4V2	Other retinopathy, autosomal recessive[1,2]
DHDDS	Retinitis pigmentosa, autosomal recessive[1,2]
DHX32¶	Inherited retinal disease, autosomal recessive
DHX38	Retinitis pigmentosa, autosomal recessive[1,2]
DNAJC17¶	Retinitis pigmentosa with hypogammaglobulinemia, autosomal recessive
DRAM2	Macular dystrophy, autosomal recessive[1,2]
DSCAML1¶	Retinal disease, autosomal recessive
DTHD1	Leber congenital amaurosis, autosomal recessive[1,2]
EFEMP1	Doyne honeycomb maculopathy (Malattia leventinese), autosomal dominant[1,2]
ELOVL4	Stargardt disease, autosomal dominant[1,2]
EMC1	Retinitis pigmentosa, autosomal recessive[1,2]
ERCC6	Cockayne syndrome B, autosomal recessive Cerebrooculofacioskeletal syndrome, autosomal recessive
EXOSC2	Syndromic retinitis pigmentosa, autosomal recessive[1,2]
EYS	Retinitis pigmentosa, autosomal recessive[1,2]
FAM161A	Retinitis pigmentosa, autosomal recessive[1,2]
FBLN5	Hereditary neuropathy with or without age-related macular degeneration, autosomal dominant Cutis laxa, type 1A, autosomal recessive
FLVCR1	Childhood onset syndromic retinitis pigmentosa, autosomal recessive[1,2]
FRMD7	Infantile nystagmus, X-linked[2]
FSCN2	Macular dystrophy, autosomal dominant[1,2]
FZD4	Other retinopathy, autosomal dominant[1,2]
GDF6	Leber congenital amaurosis, autosomal recessive[1,2]
GNAT1	Congenital stationary night blindness, autosomal dominant[1,2]
GNAT2	Cone or cone-rod dystrophy, autosomal recessive[1,2]
GNB3	Congenital stationary night blindness, autosomal recessive[1,2]
GNPTG	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
GNS	Mucopolysaccharidosis type IIID, also known as Sanfilippo syndrome D, autosomal recessive
GPR45¶	Cone-rod dystrophy
GPR143	Congenital nystagmus, X-linked ocular albinism, X-linked[1,2]
GPR179	Congenital stationary night blindness, autosomal recessive[1,2]
GRM6	Congenital stationary night blindness, autosomal recessive[1,2]
GRN	GRN-related frontotemporal dementia, autosomal dominant Neuronal ceroid lipfuscinosis type 11, autosomal recessive
GUCA1A	Cone or cone-rod dystrophy, autosomal dominant[1,2]
GUCA1B	Macular dystrophy, autosomal dominant[1,2]
GUCY2D	Leber congenital amaurosis, autosomal recessive[1,2]
HARS	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
HGSNAT	Retinitis pigmentosa, autosomal recessive[1,2]
HK1	Retinitis pigmentosa, autosomal dominant[1,2]
HMCN1¶	Age-related macular degeneration, autosomal dominant
HMX1	Oculoauricular syndrome, autosomal recessive[1,2]
IDH3A	Retinitis pigmentosa, autosomal recessive[1,2]
IDH3B	Retinitis pigmentosa, autosomal recessive[1,2]
IFT27	Bardet-Biedl syndrome, autosomal recessive[1,2]
IFT43	Nonsyndromic early onset retinitis pigmentosa, autosomal recessive Sensenbrenner syndrome, autosomal recessive[2]
IFT74	Joubert syndrome, autosomal recessive Bardet-Biedl syndrome,¶ autosomal recessive
IFT80	Syndromic ciliopathy with retinal degeneration, autosomal recessive[2]
IFT81	Syndromic ciliopathy with retinal degeneration, autosomal recessive[1,2]
IFT88¶	Retinal degeneration
IFT140	Leber congenital amaurosis, autosomal recessive[1,2]
IFT172	Bardet-Biedl syndrome, autosomal recessive[1,2]
IMPDH1	Retinitis pigmentosa, autosomal dominant[1,2]
IMPG1	Macular dystrophy, autosomal dominant[1,2]
IMPG2	Retinitis pigmentosa, autosomal recessive[1,2]
INPP5E	Joubert syndrome, mental retardation, truncal obesity, retinal dystrophy, and micropenis (MORM syndrome), autosomal recessive[1,2]
INVS	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
IQCB1	Leber congenital amaurosis, autosomal recessive Senior-Loken syndrome, autosomal recessive (aka *NPHP5*)[1,2]

GENE	DISEASE CATEGORY[†]
ITM2B	Cerebral amyloid angiopathy, autosomal dominant Retinal dystrophy,[¶] autosomal dominant
JAG1	Syndromic/systemic diseases with retinopathy, autosomal dominant[1,2]
KCNJ13	Leber congenital amaurosis, autosomal recessive[1,2]
KCNV2	Cone or cone-rod dystrophy, autosomal recessive[1,2]
KIAA0586	Joubert syndrome, autosomal recessive[1,2]
KIAA1549	Retinitis pigmentosa, autosomal recessive[1,2]
KIF11	Syndromic/systemic diseases with retinopathy, autosomal dominant[1,2]
KIF7	Joubert syndrome, autosomal recessive[1,2]
KIZ	Retinitis pigmentosa, autosomal recessive[1,2]
KLHL7	Retinitis pigmentosa, autosomal dominant[1,2]
LCA5	Leber congenital amaurosis, autosomal recessive[1,2]
LRAT	Leber congenital amaurosis, autosomal recessive Retinitis pigmentosa, autosomal recessive[1,2]
LRIT3	Congenital stationary night blindness, autosomal recessive[1,2]
LRMDA[§]	Oculocutaneous albinism, type 7, autosomal recessive
LRP2	Syndromic/systemic diseases with retinopathy, autosomal recessive Donnai-Barrow syndrome, autosomal recessive[1,2]
LRP5	Familial exudative vitreoretinopathy, autosomal recessive and dominant[1,2]
LYST	Chediak-Higashi syndrome, autosomal recessive
LZTFL1	Bardet-Biedl syndrome, autosomal recessive[1,2]
MAK	Retinitis pigmentosa, autosomal recessive[1,2]
MAPKAPK3	Other retinopathy, autosomal dominant[1,2]
MERTK	Retinitis pigmentosa, autosomal recessive[1,2]
MFN2	Syndromic/systemic diseases with optic atrophy, autosomal dominant Charcot-Marie-Tooth disease, autosomal recessive and dominant[1,2]
MFRP	Other retinopathy, autosomal recessive[1,2]
MFSD8	Neuronal ceroid lipofuscinosis type 7, autosomal recessive Retinal dystrophy, autosomal recessive
MIR204[¶]	Retinal dystrophy, autosomal dominant Iris coloboma, autosomal dominant
MKKS	Bardet-Biedl syndrome, autosomal recessive[1,2]
MKS1	Joubert syndrome, autosomal recessive Bardet-Biedl syndrome, autosomal recessive[1,2]
MPDZ	Retinitis pigmentosa, autosomal dominant[1,2]
MTPAP	Spastic ataxia 4, autosomal recessive
MTTP	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
MYO7A	Usher syndrome, autosomal recessive[2]
NAGLU	Mucopolysaccharidosis type IIIB, autosomal recessive
NBAS	Infantile liver failure, autosomal recessive Short stature with optic nerve atrophy and Pelger-Huët anomaly syndrome, autosomal recessive
NDP	Norrie disease, X-linked[1,2]
NEK2	Retinitis pigmentosa, autosomal recessive[1,2]
NEUROD1	Retinitis pigmentosa, autosomal recessive[1,2]
NMNAT1	Leber congenital amaurosis, autosomal recessive[1,2]
NPHP1	Bardet-Biedl syndrome, autosomal recessive[1,2]
NPHP3	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
NPHP4	Senior-Loken syndrome, autosomal recessive[1,2]
NR2E3	Retinitis pigmentosa, autosomal recessive Enhanced s-cone syndrome, autosomal recessive[1,2]
NR2F1	Bosch-Boonstra-Schaaf optic atrophy syndrome, autosomal dominant[1,2]
NRL	Retinitis pigmentosa, autosomal dominant[1,2]
NYX	Congenital stationary night blindness, X-linked[1,2]
OAT[‡]	Other retinopathy, autosomal recessive[1,2]
OCA2	Oculocutaneous albinism, autosomal recessive[1,2]
OFD1	Joubert syndrome, X-linked recessive Retinitis pigmentosa, X-linked recessive[1,2]
OPA1	Hereditary optic atrophy, autosomal dominant Syndromic/systemic diseases with optic atrophy, autosomal dominant Behr syndrome, autosomal recessive[1,2]
OPA3	Optic atrophy and cataract, autosomal dominant[1,2]
OPN1SW	Other retinopathy, autosomal dominant[1,2]
OR2W3[¶]	Retinitis pigmentosa
OTX2	Leber congenital amaurosis, autosomal dominant[1,2]
P3H2	High myopia with cataract and vitreoretinal degeneration (MCVD), autosomal recessive[2]
PAX2	Syndromic/systemic diseases with optic atrophy and retinopathy, autosomal dominant[1,2]
PAX6	Peters anomaly, autosomal dominant Aniridia, autosomal dominant Optic nerve malformations, autosomal dominant Foveal hypoplasia, autosomal dominant[1,2]
PCARE[§]	Retinitis pigmentosa, autosomal recessive[1,2]
PCDH15	Deafness alone or syndromic, autosomal recessive Usher syndrome, autosomal recessive and digenic recessive[1,2]
PCYT1A	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
PDE6A	Retinitis pigmentosa, autosomal recessive[1,2]
PDE6B	Retinitis pigmentosa, autosomal recessive[1,2]
PDE6C	Cone or cone-rod dystrophy, autosomal recessive[1,2]
PDE6D	Joubert syndrome, autosomal recessive[1,2]
PDE6G	Retinitis pigmentosa, autosomal recessive[1,2]
PDE6H	Cone or cone-rod dystrophy, autosomal recessive[1,2]
PDZD7	Deafness alone or syndromic, autosomal recessive[1,2]
PEX1	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
PEX2	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
PEX3	Peroxisome biogenesis disorder, autosomal recessive[2]
PEX5	Peroxisome biogenesis disorder, autosomal recessive[2]
PEX6	Peroxisome biogenesis disorder, autosomal recessive[2]
PEX7	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
PEX10	Peroxisome biogenesis disorder, autosomal recessive[2]
PEX11B	Peroxisome biogenesis disorder, autosomal recessive[2]
PEX12	Peroxisome biogenesis disorder, autosomal recessive[2]
PEX13	Peroxisome biogenesis disorder, autosomal recessive[2]
PEX14	Peroxisome biogenesis disorder, autosomal recessive[2]
PEX16	Peroxisome biogenesis disorder, autosomal recessive[2]
PEX19	Peroxisome biogenesis disorder, autosomal recessive[2]
PEX26	Peroxisome biogenesis disorder, autosomal recessive[2]
PHYH	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
PITPNM3	Cone or cone-rod dystrophy, autosomal dominant[1,2]
PLA2G5	Other retinopathy, autosomal recessive[1,2]
PLK4	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]

GENE	DISEASE CATEGORY†
PNPLA6	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
POC1B	Cone or cone-rod dystrophy, autosomal recessive[1,2]
POC5¶	Retinitis pigmentosa
POMGNT1	Retinitis pigmentosa, autosomal recessive[1,2]
PPT1	Neuronal ceroid lipofuscinosis 1, autosomal recessive
PRCD	Retinitis pigmentosa, autosomal recessive[1,2]
PRDM13	Chorioretinal atrophy or degeneration, autosomal dominant[1,2]
PROM1‡	Cone or cone-rod dystrophy, autosomal dominant[1,2]
PRPF3	Retinitis pigmentosa, autosomal dominant[1,2]
PRPF4	Retinitis pigmentosa, autosomal dominant[1,2]
PRPF6	Retinitis pigmentosa, autosomal dominant[1,2]
PRPF8	Retinitis pigmentosa, autosomal dominant[1,2]
PRPF31	Retinitis pigmentosa, autosomal dominant[1,2]
PRPH2	Cone or cone-rod dystrophy, autosomal dominant[1,2]
PRPS1	Syndromic/systemic diseases with optic atrophy, X-linked Charcot-Marie-Tooth disease, X-linked recessive[1,2]
RAB28	Cone or cone-rod dystrophy, autosomal recessive[1,2]
RAX2	Cone or cone-rod dystrophy, autosomal recessive[1,2]
RBP1¶	Leber congenital amaurosis, autosomal dominant
RBP3	Retinitis pigmentosa, autosomal recessive[1,2]
RBP4	Other retinopathy, autosomal recessive[1,2]
RCBTB1	Other retinopathy, autosomal dominant[1,2]
RD3	Leber congenital amaurosis, autosomal recessive[1,2]
RDH5	Cone or cone-rod dystrophy, autosomal recessive[1,2]
RDH11	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
RDH12	Leber congenital amaurosis, autosomal recessive Retinitis pigmentosa, autosomal dominant[1,2]
REEP6	Retinitis pigmentosa, autosomal recessive[1,2]
RGR	Retinitis pigmentosa, autosomal recessive[1,2]
RGS9	Other retinopathy, autosomal recessive[1,2]
RGS9BP	Other retinopathy, autosomal recessive[1,2]
RHO	Retinitis pigmentosa, autosomal dominant[1,2]
RIMS1	Cone or cone-rod dystrophy, autosomal dominant[1,2]
RLBP1	Retinitis pigmentosa, autosomal recessive[1,2]
ROM1	Retinitis pigmentosa, autosomal dominant[1,2]
RP1	Retinitis pigmentosa, autosomal dominant[1,2]
RP9¶	Retinitis pigmentosa
RP1L1‡	Occult macular dystrophy, autosomal dominant Retinitis pigmentosa, autosomal recessive[1,2]
RP2	Retinitis pigmentosa, X-linked[1,2]
RPE65	Retinitis pigmentosa, autosomal recessive Leber congenital amaurosis, autosomal recessive[1,2]
RPGR	Primary ciliary dyskinesia, X-linked Retinitis pigmentosa, X-linked Cone-rod dystrophy, X-linked
RPGRIP1	Cone or cone-rod dystrophy, autosomal recessive[1,2]
RPGRIP1L	Joubert syndrome, autosomal recessive[1,2]
RS1	Retinoschisis, X-linked[1,2]
RTN4IP1	Optic atrophy, autosomal recessive[1,2]

GENE	DISEASE CATEGORY†
SAG	Congenital stationary night blindness, autosomal recessive Retinitis pigmentosa, autosomal recessive and dominant[1,2]
SAMD11	Retinitis pigmentosa, autosomal recessive[1,2]
SCLT1	Retinitis pigmentosa, autosomal recessive[1,2]
SDCCAG8	Bardet-Biedl syndrome, autosomal recessive[1,2]
SEMA4A	Cone or cone-rod dystrophy, autosomal dominant[1,2]
SGSH	Mucopolysaccharidosis type IIIA, also known as Sanfilippo syndrome A, autosomal recessive
SIX6	Optic disc anomalies with retinal and/or macular dystrophy, autosomal recessive
SLC24A1	Congenital stationary night blindness, autosomal recessive[1,2]
SLC24A5	Oculocutaneous albinism, autosomal recessive
SLC45A2	Oculocutaneous albinism, autosomal recessive[1,2]
SLC7A14	Retinitis pigmentosa, autosomal recessive[1,2]
SNRNP200	Retinitis pigmentosa, autosomal dominant[1,2]
SPATA7	Leber congenital amaurosis, autosomal recessive[1,2]
SPP2	Retinitis pigmentosa, autosomal dominant[1,2]
TCTN1	Joubert syndrome, autosomal recessive[1,2]
TCTN2	Joubert syndrome, autosomal recessive Meckel syndrome, autosomal recessive[1,2]
TCTN3	Joubert syndrome, autosomal recessive[1,2]
TEAD1	Chorioretinal atrophy or degeneration, autosomal dominant[1,2]
TIMM8A	Syndromic/systemic diseases with optic neuropathy, X-linked Mohr-Tranebjaerg syndrome, X-linked recessive[1,2]
TIMP3	Macular dystrophy, autosomal dominant[1,2]
TMED7¶	Retinal dystrophy, autosomal recessive
TMEM67	Joubert syndrome, autosomal recessive Meckel syndrome, autosomal recessive[1,2]
TMEM107	Joubert syndrome, autosomal recessive Meckel syndrome, autosomal recessive[1,2]
TMEM126A	Optic atrophy, autosomal recessive[1,2]
TMEM138	Joubert syndrome, autosomal recessive[1,2]
TMEM216	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
TMEM231	Joubert syndrome, autosomal recessive Meckel syndrome, autosomal recessive[1,2]
TMEM237	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
TOPORS	Retinitis pigmentosa, autosomal dominant[1,2]
TPP1	Infantile neuronal ceroid lipofuscinosis (CLN2), autosomal recessive Spinocerebellar ataxia, autosomal recessive[2]
TRAF3IP1	Senior-Loken syndrome 9 (nephronophthisis and retinal dystrophy), autosomal recessive[2]
TREX1	Syndromic/systemic diseases with retinopathy, autosomal dominant Aicardi-Goutieres syndrome, autosomal dominant and recessive[1,2]
TRIM32	Bardet-Biedl syndrome, autosomal recessive[1,2]
TRNT1	Retinitis pigmentosa, autosomal recessive[1,2]
TRPM1	Congenital stationary night blindness, autosomal recessive[1,2]
TSPAN12	Other retinopathy, autosomal dominant[1,2]
TTC21B	Syndromic/systemic diseases with retinopathy, autosomal recessive Joubert syndrome, autosomal recessive[1,2]
TTC8	Retinitis pigmentosa, autosomal recessive[1,2]
TTLL5	Cone or cone-rod dystrophy, autosomal recessive[1,2]
TTPA	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
TUB	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]

GENE	DISEASE CATEGORY†
TUBGCP4	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
TUBGCP6	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
TULP1	Leber congenital amaurosis, autosomal recessive Retinitis pigmentosa, autosomal recessive[1,2]
TYR‡	Oculocutaneous albinism, autosomal recessive[1,2]
TYRP1	Oculocutaneous albinism, autosomal recessive[1,2]
UNC119	Cone or cone-rod dystrophy, autosomal dominant[1,2]
USH1C‡	Usher syndrome, autosomal recessive[1,2]
USH1G	Usher syndrome, autosomal recessive[1,2]
USH2A	Usher syndrome, autosomal recessive Retinitis pigmentosa, autosomal recessive[1,2]
VCAN	Wagner syndrome, autosomal dominant[1,2]
VPS13B	Cohen syndrome, autosomal recessive[2]
WDPCP	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
WDR19	Syndromic/systemic diseases with retinopathy, autosomal recessive[2]
WDR34	Short-rib thoracic dysplasia 11, autosomal recessive Retinitis pigmentosa, also known as rod-cone dystrophy,¶ autosomal recessive
WFS1	Syndromic/systemic diseases with optic atrophy, autosomal recessive and dominant Wolfram syndrome, autosomal recessive[1,2]
WHRN	Usher syndrome, autosomal recessive[2]
ZNF408	Retinitis pigmentosa, autosomal recessive[1,2]
ZNF423	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
ZNF513	Retinitis pigmentosa, autosomal recessive[1,2]

*This initiative is open to U.S. residents only, subject to the Terms and Conditions of the program.

†Disease category is not an all-inclusive list.

‡Denotes limitation.

§Nomenclature has been updated from *c21orf2* to *CFAP410*, *GPR125* to *ADGRA3*, *c2orf71* to *PCARE*, and *C10orf11* to *LRMDA* per HUGO Gene Nomenclature Committee (HGNC) guidelines.

¶Shows preliminary evidence supporting a correlation with the disease.

Visit **IDYOURIRD.com** for more information or to order free* test kits.

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Spark™ THERAPEUTICS