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 **approximately 250 genes** that are known to cause 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]
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]
ADGRA3††	Retinitis pigmentosa, autosomal recessive (aka *GPR125*)[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]
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]
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	Bardet-Biedl syndrome, autosomal recessive Retinitis pigmentosa, 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]
C21orf2††	Cone or cone-rod dystrophy, autosomal recessive[1,2]
C2orf71	Retinitis pigmentosa, autosomal recessive[1,2]
C8orf37††	Bardet-Biedl syndrome, autosomal recessive Cone-rod dystrophy, autosomal recessive[1,2]
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]
CDH23	Usher syndrome, autosomal recessive and digenic recessive[1,2]
CDH3	Other retinopathy, autosomal 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]

GENE	DISEASE CATEGORY†
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]
CHM	Choroideremia, X-linked[1,2]
CIB2	Deafness alone or syndromic, autosomal recessive[1,2]
CLCC1††	Retinitis pigmentosa, autosomal recessive[1,2]
CLN3††	Juvenile neuronal ceroid lipofuscinosis, autosomal recessive[1,2]
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]
COL2A1	Stickler syndrome, autosomal dominant[1,2]
COL9A1	Stickler 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]
CWC27††	Retinitis pigmentosa with or without skeletal abnormalities[2]
CYP4V2††	Other retinopathy, autosomal recessive[1,2]
DHDDS	Retinitis pigmentosa, autosomal recessive[1,2]
DHX38	Retinitis pigmentosa, autosomal recessive[1,2]
DRAM2††	Macular dystrophy, autosomal recessive[1,2]
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]
EXOSC2	Syndromic retinitis pigmentosa, autosomal recessive[1,2]
EYS	Retinitis pigmentosa, autosomal recessive[1,2]
FAM161A	Retinitis pigmentosa, autosomal recessive[1,2]
FLVCR1	Childhood onset syndromic retinitis pigmentosa, autosomal recessive[1,2]
FRMD7	Infantile nystagmus, X-linked[2]

GENE	DISEASE CATEGORY†
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]
GRM6	Congenital stationary night blindness, autosomal recessive[1,2]
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]
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]
IFT80††	Syndromic ciliopathy with retinal degeneration, autosomal recessive[2]
IFT81	Syndromic ciliopathy with retinal degeneration, autosomal recessive[1,2]
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]
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]
KIAA1549††	Retinitis pigmentosa, autosomal recessive[1,2]
KIF11	Syndromic/systemic diseases with retinopathy, autosomal dominant[1,2]

GENE	DISEASE CATEGORY†
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]
LRP5††	Familial exudative vitreoretinopathy, autosomal recessive and dominant[1,2]
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]
MFRP	Other retinopathy, autosomal recessive[1,2]
MKKS	Bardet-Biedl syndrome, autosomal recessive[1,2]
MKS1	Joubert syndrome, autosomal recessive Bardet-Biedl syndrome, autosomal recessive[1,2]
MTTP	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
MYO7A	Usher syndrome, autosomal recessive[2]
NDP	Norrie disease, X-linked[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]
NRL	Retinitis pigmentosa, autosomal dominant[1,2]
NYX	Congenital stationary night blindness, X-linked[1,2]
OAT††	Other retinopathy, autosomal recessive[1,2]
OPN1SW	Other retinopathy, autosomal dominant[1,2]
OTX2	Leber congenital amaurosis, autosomal dominant[1,2]
P3H2††	High myopia with cataract and vitreoretinal degeneration (MCVD), autosomal recessive[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]
PDE6G	Retinitis pigmentosa, autosomal recessive[1,2]
PDE6H	Cone or cone-rod dystrophy, autosomal recessive[1,2]

GENE	DISEASE CATEGORY†
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]
PNPLA6	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
POC1B††	Cone or cone-rod dystrophy, autosomal recessive[1,2]
POMGNT1	Retinitis pigmentosa, autosomal recessive[1,2]
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]
PRPH2	Cone or cone-rod dystrophy, autosomal dominant[1,2]
RAB28††	Cone or cone-rod dystrophy, autosomal recessive[1,2]
RAX2	Cone or cone-rod dystrophy, autosomal recessive[1,2]
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]

GENE	DISEASE CATEGORY†
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]
RP2	Retinitis pigmentosa, X-linked[1,2]
RPE65	Retinitis pigmentosa, autosomal recessive Leber congenital amaurosis, autosomal recessive[1,2]
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]
SAG††	Congenital stationary night blindness, autosomal recessive Retinitis pigmentosa, autosomal recessive and dominant[1,2]
SAMD11††	Retinitis pigmentosa, autosomal recessive[1,2]
SDCCAG8	Bardet-Biedl syndrome, autosomal recessive[1,2]
SEMA4A	Cone or cone-rod dystrophy, autosomal dominant[1,2]
SLC24A1††	Congenital stationary night blindness, 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]
TEAD1	Chorioretinal atrophy or degeneration, autosomal dominant[1,2]
TIMP3	Macular dystrophy, autosomal dominant[1,2]
TMEM126A	Optic atrophy, autosomal recessive[1,2]
TMEM216	Syndromic/systemic diseases with retinopathy, 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]
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]
TTC8	Retinitis pigmentosa, autosomal recessive[1,2]
TTLL5	Cone or cone-rod dystrophy, autosomal recessive[1,2]
TUB	Syndromic/systemic diseases with retinopathy, autosomal recessive[1,2]
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]
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]
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.

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

References: 1. Daiger SP, Sullivan LS, Bowne SJ. RetNet™ Retinal Information Network. https://sph.uth.edu/retnet/home.htm. Updated May 21, 2019. Accessed May 22, 2019. **2.** OMIM®. An Online Catalog of Human Genes and Genetic Disorders. Updated May 24, 2019. Accessed May 28, 2019.

Spark THERAPEUTICS

N-IDIRD-US-660003 Printed in the USA. June 2019