

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 300 genes** associated with IRDs.

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



INHERITED RETINAL DISEASE
GENE TESTING INITIATIVE

GENE	DISEASE CATEGORY†	GENE	DISEASE CATEGORY†
<i>ABCA4</i>	Stargardt disease, autosomal recessive Cone or cone-rod dystrophy, autosomal recessive ^{1,2}	<i>BBS4</i>	Bardet-Biedl syndrome, autosomal recessive ^{1,2}
<i>ABCC6</i> [†]	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}	<i>BBS5</i>	Bardet-Biedl syndrome, autosomal recessive ^{1,2}
<i>ABHD12</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}	<i>BBS7</i>	Bardet-Biedl syndrome, autosomal recessive ^{1,2}
<i>ACBD5</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}	<i>BBS9</i> [†]	Bardet-Biedl syndrome, autosomal recessive ^{1,2}
<i>ACO2</i>	Optic atrophy, autosomal recessive Infantile cerebellar-retinal degeneration, autosomal recessive ^{1,2}	<i>BBS10</i>	Bardet-Biedl syndrome, autosomal recessive ^{1,2}
<i>ADAM9</i>	Cone or cone-rod dystrophy, autosomal recessive ^{1,2}	<i>BBS12</i>	Bardet-Biedl syndrome, autosomal recessive ^{1,2}
<i>ADAMTS18</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}	<i>BEST1</i>	Vitelliform macular degeneration (Best disease), autosomal dominant and recessive Retinitis pigmentosa, autosomal dominant and recessive ^{1,2}
<i>ADGRA3</i> [§]	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>C10TNF5</i>	Macular dystrophy, autosomal dominant ^{1,2}
<i>ADGRV1</i>	Usher syndrome, autosomal recessive ^{1,2}	<i>C8orf37</i>	Bardet-Biedl syndrome, autosomal recessive Cone-rod dystrophy, autosomal recessive ^{1,2}
<i>ADIPOR1</i>	Retinitis pigmentosa, autosomal dominant Bardet-Biedl syndrome, autosomal recessive ^{1,2}	<i>CA4</i>	Retinitis pigmentosa, autosomal dominant ^{1,2}
<i>AGBL5</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>CABP4</i>	Congenital stationary night blindness, autosomal recessive ^{1,2}
<i>AHI1</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}	<i>CACNA1F</i>	Cone or cone-rod dystrophy, X-linked ^{1,2}
<i>AHR</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>CACNA2D4</i>	Cone or cone-rod dystrophy, autosomal recessive ^{1,2}
<i>AIPL1</i>	Cone or cone-rod dystrophy, autosomal dominant Leber congenital amaurosis, autosomal recessive ^{1,2}	<i>CAPN5</i>	Other retinopathy, autosomal dominant ^{1,2}
<i>ALMS1</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive Alstrom syndrome, autosomal recessive ^{1,2}	<i>CC2D2A</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}
<i>ARHGEF18</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>CCT2</i>	Leber congenital amaurosis, autosomal recessive ^{1,2}
<i>ARL13B</i>	Joubert syndrome, autosomal recessive ²	<i>CDH3</i>	Other retinopathy, autosomal recessive ^{1,2}
<i>ARL2BP</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>CDH23</i>	Usher syndrome, autosomal recessive and digenic recessive ^{1,2}
<i>ARL3</i>	Retinitis pigmentosa, autosomal dominant ^{1,2}	<i>CDHR1</i>	Cone or cone-rod dystrophy, autosomal recessive ^{1,2}
<i>ARL6</i>	Retinitis pigmentosa, autosomal recessive Bardet-Biedl syndrome, autosomal recessive ^{1,2}	<i>CEP19</i>	Bardet-Biedl syndrome, autosomal recessive ^{1,2}
<i>ARMC9</i> [†]	Joubert syndrome, autosomal recessive ²	<i>CEP41</i>	Joubert syndrome, autosomal recessive ²
<i>ARSG</i>	Usher syndrome, autosomal recessive ^{1,2}	<i>CEP78</i>	Cone or cone-rod dystrophy with hearing loss, autosomal recessive ^{1,2}
<i>ASRGL1</i>	Other retinopathy, autosomal recessive ^{1,2}	<i>CEP83</i>	Infantile nephronophthisis with occasional retinopathy ²
<i>ATF6</i>	Cone or cone-rod dystrophy, autosomal recessive ^{1,2}	<i>CEP164</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}
<i>ATOH7</i>	Nonsyndromal congenital retinal nonattachment, autosomal recessive (aka <i>RNANC</i>) ²	<i>CEP250</i>	Usher syndrome, autosomal recessive ^{1,2}
<i>B9D1</i>	Joubert syndrome, autosomal recessive Meckel syndrome, autosomal recessive ^{1,2}	<i>CEP290</i>	Leber congenital amaurosis, autosomal recessive Joubert syndrome, autosomal recessive ^{1,2}
<i>BBIP1</i>	Retinitis pigmentosa, autosomal recessive Bardet-Biedl syndrome, autosomal recessive ^{1,2}	<i>CERKL</i>	Cone or cone-rod dystrophy, autosomal recessive Retinitis pigmentosa, autosomal recessive ^{1,2}
<i>BBS1</i>	Retinitis pigmentosa, autosomal recessive Bardet-Biedl syndrome, autosomal recessive ^{1,2}	<i>CFAP410</i> [§]	Cone or cone-rod dystrophy, autosomal recessive ^{1,2}
<i>BBS2</i>	Bardet-Biedl syndrome, autosomal recessive Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>CHM</i>	Choroideremia, X-linked ^{1,2}
		<i>CIB2</i>	Deafness alone or syndromic, autosomal recessive ^{1,2}
		<i>CISD2</i>	Syndromic/systemic diseases with optic atrophy, autosomal recessive Wolfram syndrome, autosomal recessive ^{1,2}
		<i>CLCC1</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}

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<i>CLN3</i>	Juvenile neuronal ceroid lipofuscinosis, autosomal recessive ^{1,2}	<i>GPR143</i>	Congenital nystagmus, X-linked ocular albinism, X-linked ^{1,2}
<i>CLRN1</i>	Retinitis pigmentosa, autosomal recessive Usher syndrome, autosomal recessive ^{1,2}	<i>GPR179</i>	Congenital stationary night blindness, autosomal recessive ^{1,2}
<i>CLUAP1</i>	Leber congenital amaurosis, autosomal recessive ^{1,2}	<i>GRM6</i>	Congenital stationary night blindness, autosomal recessive ^{1,2}
<i>CNGA1</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>GUCA1A</i>	Cone or cone-rod dystrophy, autosomal dominant ^{1,2}
<i>CNGA3</i>	Achromatopsia, autosomal recessive Cone or cone-rod dystrophy, autosomal recessive ^{1,2}	<i>GUCA1B</i>	Macular dystrophy, autosomal dominant ^{1,2}
<i>CNGB1</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>GUCY2D</i>	Leber congenital amaurosis, autosomal recessive ^{1,2}
<i>CNGB3</i>	Achromatopsia, autosomal recessive Cone or cone-rod dystrophy, autosomal recessive ^{1,2}	<i>HARS</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}
<i>CNNM4</i>	Cone or cone-rod dystrophy, autosomal recessive Jalili syndrome, autosomal recessive ^{1,2}	<i>HGSNAT</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}
<i>COL11A1†</i>	Stickler syndrome, autosomal dominant Marshall syndrome, autosomal dominant ^{1,2}	<i>HK1</i>	Retinitis pigmentosa, autosomal dominant ^{1,2}
<i>COL11A2</i>	Stickler syndrome, autosomal dominant ^{1,2}	<i>HMX1</i>	Oculoauricular syndrome, autosomal recessive ^{1,2}
<i>COL18A1</i>	Knobloch syndrome, autosomal recessive ^{1,2}	<i>IDH3A</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}
<i>COL2A1</i>	Stickler syndrome, autosomal dominant ^{1,2}	<i>IDH3B</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}
<i>COL9A1</i>	Stickler syndrome, autosomal recessive ^{1,2}	<i>IFT27</i>	Bardet-Biedl syndrome, autosomal recessive ^{1,2}
<i>COL9A2</i>	Stickler syndrome, autosomal recessive ^{1,2}	<i>IFT43</i>	Nonsyndromic early onset retinitis pigmentosa, autosomal recessive Sensenbrenner syndrome, autosomal recessive ²
<i>COL9A3</i>	Stickler syndrome, autosomal recessive ^{1,2}	<i>IFT80</i>	Syndromic ciliopathy with retinal degeneration, autosomal recessive ²
<i>CPLANE1</i>	Joubert syndrome, autosomal recessive ^{1,2}	<i>IFT81</i>	Syndromic ciliopathy with retinal degeneration, autosomal recessive ^{1,2}
<i>CRB1</i>	Leber congenital amaurosis, autosomal recessive Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>IFT140</i>	Leber congenital amaurosis, autosomal recessive ^{1,2}
<i>CRX</i>	Leber congenital amaurosis, autosomal recessive ^{1,2}	<i>IFT172</i>	Bardet-Biedl syndrome, autosomal recessive ^{1,2}
<i>CSPP1</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}	<i>IMPDH1</i>	Retinitis pigmentosa, autosomal dominant ^{1,2}
<i>CTNNA1</i>	Macular dystrophy, autosomal dominant ^{1,2}	<i>IMPG1</i>	Macular dystrophy, autosomal dominant ^{1,2}
<i>CWC27</i>	Retinitis pigmentosa with or without skeletal abnormalities ²	<i>IMPG2</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}
<i>CYP4V2</i>	Other retinopathy, autosomal recessive ^{1,2}	<i>INPP5E</i>	Joubert syndrome, mental retardation, truncal obesity, retinal dystrophy, and micropenis (MORM syndrome), autosomal recessive ^{1,2}
<i>DHDDS</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>INVS</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}
<i>DHX38</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>IQCB1</i>	Leber congenital amaurosis, autosomal recessive Senior-Loken syndrome, autosomal recessive (aka <i>NPHP5</i>) ^{1,2}
<i>DRAM2</i>	Macular dystrophy, autosomal recessive ^{1,2}	<i>JAG1</i>	Syndromic/systemic diseases with retinopathy, autosomal dominant ^{1,2}
<i>DTHD1</i>	Leber congenital amaurosis, autosomal recessive ^{1,2}	<i>KCNJ13</i>	Leber congenital amaurosis, autosomal recessive ^{1,2}
<i>EFEMP1</i>	Doyle honeycomb maculopathy (Malattia leventinese), autosomal dominant ^{1,2}	<i>KCNV2</i>	Cone or cone-rod dystrophy, autosomal recessive ^{1,2}
<i>ELOVL4</i>	Stargardt disease, autosomal dominant ^{1,2}	<i>KIAA0586</i>	Joubert syndrome, autosomal recessive ^{1,2}
<i>EMC1</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>KIAA1549</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}
<i>EXOSC2</i>	Syndromic retinitis pigmentosa, autosomal recessive ^{1,2}	<i>KIF11</i>	Syndromic/systemic diseases with retinopathy, autosomal dominant ^{1,2}
<i>EYS</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>KIF7</i>	Joubert syndrome, autosomal recessive ^{1,2}
<i>FAM161A</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>KIZ</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}
<i>FLVCR1</i>	Childhood onset syndromic retinitis pigmentosa, autosomal recessive ^{1,2}	<i>KLHL7</i>	Retinitis pigmentosa, autosomal dominant ^{1,2}
<i>FRMD7</i>	Infantile nystagmus, X-linked ²	<i>LCA5</i>	Leber congenital amaurosis, autosomal recessive ^{1,2}
<i>FSCN2</i>	Macular dystrophy, autosomal dominant ^{1,2}	<i>LRAT</i>	Leber congenital amaurosis, autosomal recessive Retinitis pigmentosa, autosomal recessive ^{1,2}
<i>FZD4</i>	Other retinopathy, autosomal dominant ^{1,2}	<i>LRIT3</i>	Congenital stationary night blindness, autosomal recessive ^{1,2}
<i>GDF6</i>	Leber congenital amaurosis, autosomal recessive ^{1,2}	<i>LRP2</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive Donnai-Barrow syndrome, autosomal recessive ^{1,2}
<i>GNAT1</i>	Congenital stationary night blindness, autosomal dominant ^{1,2}	<i>LRP5</i>	Familial exudative vitreoretinopathy, autosomal recessive and dominant ^{1,2}
<i>GNAT2</i>	Cone or cone-rod dystrophy, autosomal recessive ^{1,2}	<i>LZTFL1</i>	Bardet-Biedl syndrome, autosomal recessive ^{1,2}
<i>GNB3</i>	Congenital stationary night blindness, autosomal recessive ^{1,2}	<i>MAK</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}
<i>GNPTG</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}	<i>MAPKAPK3</i>	Other retinopathy, autosomal dominant ^{1,2}
		<i>MERTK</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}

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<i>MFN2</i>	Syndromic/systemic diseases with optic atrophy, autosomal dominant Charcot-Marie-Tooth disease, autosomal recessive and dominant ^{1,2}	<i>PEX2</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}
<i>MFRP</i>	Other retinopathy, autosomal recessive ^{1,2}	<i>PEX3</i>	Peroxisome biogenesis disorder, autosomal recessive ²
<i>MKKS</i>	Bardet-Biedl syndrome, autosomal recessive ^{1,2}	<i>PEX5</i>	Peroxisome biogenesis disorder, autosomal recessive ²
<i>MKS1</i>	Joubert syndrome, autosomal recessive Bardet-Biedl syndrome, autosomal recessive ^{1,2}	<i>PEX6</i>	Peroxisome biogenesis disorder, autosomal recessive ²
<i>MPDZ</i>	Retinitis pigmentosa, autosomal dominant ^{1,2}	<i>PEX7</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}
<i>MTTP</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}	<i>PEX10</i>	Peroxisome biogenesis disorder, autosomal recessive ²
<i>MYO7A</i>	Usher syndrome, autosomal recessive ²	<i>PEX11B</i>	Peroxisome biogenesis disorder, autosomal recessive ²
<i>NDP</i>	Norrie disease, X-linked ^{1,2}	<i>PEX12</i>	Peroxisome biogenesis disorder, autosomal recessive ²
<i>NEK2</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>PEX13</i>	Peroxisome biogenesis disorder, autosomal recessive ²
<i>NEUROD1</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>PEX14</i>	Peroxisome biogenesis disorder, autosomal recessive ²
<i>NMNAT1</i>	Leber congenital amaurosis, autosomal recessive ^{1,2}	<i>PEX16</i>	Peroxisome biogenesis disorder, autosomal recessive ²
<i>NPHP1</i>	Bardet-Biedl syndrome, autosomal recessive ^{1,2}	<i>PEX19</i>	Peroxisome biogenesis disorder, autosomal recessive ²
<i>NPHP3</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}	<i>PEX26</i>	Peroxisome biogenesis disorder, autosomal recessive ²
<i>NPHP4</i>	Senior-Loken syndrome, autosomal recessive ^{1,2}	<i>PHYH</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}
<i>NR2E3</i>	Retinitis pigmentosa, autosomal recessive Enhanced s-cone syndrome, autosomal recessive ^{1,2}	<i>PITPNM3</i>	Cone or cone-rod dystrophy, autosomal dominant ^{1,2}
<i>NR2F1</i>	Bosch-Boonstra-Schaaf optic atrophy syndrome, autosomal dominant ^{1,2}	<i>PLA2G5</i>	Other retinopathy, autosomal recessive ^{1,2}
<i>NRL</i>	Retinitis pigmentosa, autosomal dominant ^{1,2}	<i>PLK4</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}
<i>NYX</i>	Congenital stationary night blindness, X-linked ^{1,2}	<i>PNPLA6</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}
<i>OAT[†]</i>	Other retinopathy, autosomal recessive ^{1,2}	<i>POC1B</i>	Cone or cone-rod dystrophy, autosomal recessive ^{1,2}
<i>OCA2</i>	Oculocutaneous albinism, autosomal recessive ^{1,2}	<i>POMGNT1</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}
<i>OFD1</i>	Joubert syndrome, X-linked recessive Retinitis pigmentosa, X-linked recessive ^{1,2}	<i>PRCD</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}
<i>OPA1</i>	Hereditary optic atrophy, autosomal dominant Syndromic/systemic diseases with optic atrophy, autosomal dominant Behr syndrome, autosomal recessive ^{1,2}	<i>PRDM13</i>	Chorioretinal atrophy or degeneration, autosomal dominant ^{1,2}
<i>OPA3</i>	Optic atrophy and cataract, autosomal dominant ^{1,2}	<i>PROM1[†]</i>	Cone or cone-rod dystrophy, autosomal dominant ^{1,2}
<i>OPN1SW</i>	Other retinopathy, autosomal dominant ^{1,2}	<i>PRPF3</i>	Retinitis pigmentosa, autosomal dominant ^{1,2}
<i>OTX2</i>	Leber congenital amaurosis, autosomal dominant ^{1,2}	<i>PRPF4</i>	Retinitis pigmentosa, autosomal dominant ^{1,2}
<i>P3H2</i>	High myopia with cataract and vitreoretinal degeneration (MCVD), autosomal recessive ²	<i>PRPF6</i>	Retinitis pigmentosa, autosomal dominant ^{1,2}
<i>PAX2</i>	Syndromic/systemic diseases with optic atrophy and retinopathy, autosomal dominant ^{1,2}	<i>PRPF8</i>	Retinitis pigmentosa, autosomal dominant ^{1,2}
<i>PAX6</i>	Peters anomaly, autosomal dominant Aniridia, autosomal dominant Optic nerve malformations, autosomal dominant Foveal hypoplasia, autosomal dominant ^{1,2}	<i>PRPF31</i>	Retinitis pigmentosa, autosomal dominant ^{1,2}
<i>PCARE[§]</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>PRPH2</i>	Cone or cone-rod dystrophy, autosomal dominant ^{1,2}
<i>PCDH15</i>	Deafness alone or syndromic, autosomal recessive Usher syndrome, autosomal recessive and digenic recessive ^{1,2}	<i>PRPS1</i>	Syndromic/systemic diseases with optic atrophy, X-linked Charcot-Marie-Tooth disease, X-linked recessive ^{1,2}
<i>PCYT1A</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}	<i>RAB28</i>	Cone or cone-rod dystrophy, autosomal recessive ^{1,2}
<i>PDE6A</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>RAX2</i>	Cone or cone-rod dystrophy, autosomal recessive ^{1,2}
<i>PDE6B</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>RBP3</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}
<i>PDE6C</i>	Cone or cone-rod dystrophy, autosomal recessive ^{1,2}	<i>RBP4</i>	Other retinopathy, autosomal recessive ^{1,2}
<i>PDE6D</i>	Joubert syndrome, autosomal recessive ^{1,2}	<i>RCBTB1</i>	Other retinopathy, autosomal dominant ^{1,2}
<i>PDE6G</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>RD3</i>	Leber congenital amaurosis, autosomal recessive ^{1,2}
<i>PDE6H</i>	Cone or cone-rod dystrophy, autosomal recessive ^{1,2}	<i>RDH5</i>	Cone or cone-rod dystrophy, autosomal recessive ^{1,2}
<i>PDZD7</i>	Deafness alone or syndromic, autosomal recessive ^{1,2}	<i>RDH11</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}
<i>PEX1</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}	<i>RDH12</i>	Leber congenital amaurosis, autosomal recessive Retinitis pigmentosa, autosomal dominant ^{1,2}
		<i>REEP6</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}
		<i>RGR</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}
		<i>RGS9</i>	Other retinopathy, autosomal recessive ^{1,2}
		<i>RGS9BP</i>	Other retinopathy, autosomal recessive ^{1,2}
		<i>RHO</i>	Retinitis pigmentosa, autosomal dominant ^{1,2}

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<i>RIMS1</i>	Cone or cone-rod dystrophy, autosomal dominant ^{1,2}	<i>TRNT1</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}
<i>RLBP1</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>TRPM1</i>	Congenital stationary night blindness, autosomal recessive ^{1,2}
<i>ROM1</i>	Retinitis pigmentosa, autosomal dominant ^{1,2}	<i>TSPAN12</i>	Other retinopathy, autosomal dominant ^{1,2}
<i>RP1</i>	Retinitis pigmentosa, autosomal dominant ^{1,2}	<i>TTC21B</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive Joubert syndrome, autosomal recessive ^{1,2}
<i>RP1L1</i> [†]	Occult macular dystrophy, autosomal dominant Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>TTC8</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}
<i>RP2</i>	Retinitis pigmentosa, X-linked ^{1,2}	<i>TLL5</i>	Cone or cone-rod dystrophy, autosomal recessive ^{1,2}
<i>RPE65</i>	Retinitis pigmentosa, autosomal recessive Leber congenital amaurosis, autosomal recessive ^{1,2}	<i>TTPA</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}
<i>RPGRIP1</i>	Cone or cone-rod dystrophy, autosomal recessive ^{1,2}	<i>TUB</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}
<i>RPGRIP1L</i>	Joubert syndrome, autosomal recessive ^{1,2}	<i>TUBGCP4</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}
<i>RS1</i>	Retinoschisis, X-linked ^{1,2}	<i>TUBGCP6</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}
<i>RTN4IP1</i>	Optic atrophy, autosomal recessive ^{1,2}	<i>TULP1</i>	Leber congenital amaurosis, autosomal recessive Retinitis pigmentosa, autosomal recessive ^{1,2}
<i>SAG</i>	Congenital stationary night blindness, autosomal recessive Retinitis pigmentosa, autosomal recessive and dominant ^{1,2}	<i>TYR</i> [†]	Oculocutaneous albinism, autosomal recessive ^{1,2}
<i>SAMD11</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>TYRP1</i>	Oculocutaneous albinism, autosomal recessive ^{1,2}
<i>SCLT1</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>UNC119</i>	Cone or cone-rod dystrophy, autosomal dominant ^{1,2}
<i>SDCCAG8</i>	Bardet-Biedl syndrome, autosomal recessive ^{1,2}	<i>USH1C</i> [†]	Usher syndrome, autosomal recessive ^{1,2}
<i>SEMA4A</i>	Cone or cone-rod dystrophy, autosomal dominant ^{1,2}	<i>USH1G</i>	Usher syndrome, autosomal recessive ^{1,2}
<i>SLC24A1</i>	Congenital stationary night blindness, autosomal recessive ^{1,2}	<i>USH2A</i>	Usher syndrome, autosomal recessive Retinitis pigmentosa, autosomal recessive ^{1,2}
<i>SLC45A2</i>	Oculocutaneous albinism, autosomal recessive ^{1,2}	<i>VCAN</i>	Wagner syndrome, autosomal dominant ^{1,2}
<i>SLC7A14</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}	<i>VPS13B</i>	Cohen syndrome, autosomal recessive ²
<i>SNRNP200</i>	Retinitis pigmentosa, autosomal dominant ^{1,2}	<i>WDPCP</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}
<i>SPATA7</i>	Leber congenital amaurosis, autosomal recessive ^{1,2}	<i>WDR19</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ²
<i>SPP2</i>	Retinitis pigmentosa, autosomal dominant ^{1,2}	<i>WFS1</i>	Syndromic/systemic diseases with optic atrophy, autosomal recessive and dominant Wolfram syndrome, autosomal recessive ^{1,2}
<i>TCTN1</i>	Joubert syndrome, autosomal recessive ^{1,2}	<i>WHRN</i>	Usher syndrome, autosomal recessive ²
<i>TCTN2</i>	Joubert syndrome, autosomal recessive Meckel syndrome, autosomal recessive ^{1,2}	<i>ZNF408</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}
<i>TCTN3</i>	Joubert syndrome, autosomal recessive ^{1,2}	<i>ZNF423</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}
<i>TEAD1</i>	Chorioretinal atrophy or degeneration, autosomal dominant ^{1,2}	<i>ZNF513</i>	Retinitis pigmentosa, autosomal recessive ^{1,2}
<i>TIMM8A</i>	Syndromic/systemic diseases with optic neuropathy, X-linked Mohr-Tranebjaerg syndrome, X-linked recessive ^{1,2}		
<i>TIMP3</i>	Macular dystrophy, autosomal dominant ^{1,2}		
<i>TMEM67</i>	Joubert syndrome, autosomal recessive Meckel syndrome, autosomal recessive ^{1,2}		
<i>TMEM107</i>	Joubert syndrome, autosomal recessive Meckel syndrome, autosomal recessive ^{1,2}		
<i>TMEM126A</i>	Optic atrophy, autosomal recessive ^{1,2}		
<i>TMEM138</i>	Joubert syndrome, autosomal recessive ^{1,2}		
<i>TMEM216</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}		
<i>TMEM231</i>	Joubert syndrome, autosomal recessive Meckel syndrome, autosomal recessive ^{1,2}		
<i>TMEM237</i>	Syndromic/systemic diseases with retinopathy, autosomal recessive ^{1,2}		
<i>TOPORS</i>	Retinitis pigmentosa, autosomal dominant ^{1,2}		
<i>TTP1</i>	Infantile neuronal ceroid lipofuscinosis (CLN2), autosomal recessive Spinocerebellar ataxia, autosomal recessive ²		
<i>TRAF3IP1</i>	Senior-Loken syndrome 9 (nephronophthisis and retinal dystrophy), autosomal recessive ²		
<i>TREX1</i>	Syndromic/systemic diseases with retinopathy, autosomal dominant Aicardi-Goutieres syndrome, autosomal dominant and recessive ^{1,2}		
<i>TRIM32</i>	Bardet-Biedl syndrome, 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*, and *c2orf71* to *PCARE* per HUGO Gene Nomenclature Committee (HGNC) guidelines.

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