

RETINAL DYSTROPHY XPANDED PANEL

GENE LIST (~875 GENES)

Note: Xpanded panel gene lists are regularly updated/improved. This list should be considered an approximation only as the gene list is subject to change at any time. The patient's test report includes a list of genes evaluated and includes a coverage parameter for each gene.

| | | | | | | | |
|-----------------|----------|----------------|----------|----------|-----------------|----------|----------|
| AAAS | ABCA1 | ABCA4 | ABCC6 | ABCD1 | ABHD12 | ACBD5 | ACO2 |
| ACOX1 | ACTB | ACTG1 | ACVR1 | ACVR2B | ADAM9 | ADAMTS10 | ADAMTS17 |
| ADAMTS18 | ADAMTSL4 | ADGRV1 (GPR98) | ADIPOR1 | AGBL1 | AGBL5 | AGK | AGPS |
| AHI1 | AIPL1 | AIRE | AKAP9 | ALDH18A1 | ALDH1A3 | ALDH3A2 | ALG2 |
| ALG6 | ALMS1 | ALX1 | AMACR | AMT | ANKLE2 | ANKS6 | ANTXR1 |
| AP3B1 | AP3D1 | ARL13B | ARL2BP | ARL3 | ARL6 | ARNT2 | ARSB |
| ARX | ASB10 | ASPA | ASPH | ASRGL1 | ATAD3A | ATF4 | ATF6 |
| ATIC | ATM | ATOH7 | ATP13A2 | ATP1A3 | ATXN7 | B3GALNT2 | B3GALTL |
| B4GAT1 (B3GNT1) | B9D1 | B9D2 | BBIP1 | BBS1 | BBS10 | BBS12 | BBS2 |
| BBS4 | BBS5 | BBS7 | BBS9 | BCOR | BCS1L | BEST1 | BFSP1 |
| BFSP2 | BLOC1S3 | BLOC1S6 | BMP1 | BMP4 | BMP7 | BRAF | BRIP1 |
| BTD | BUB1B | C10orf11 | C10orf2 | C12orf57 | C12orf65 | C1orf186 | C1QTNF5 |
| C21orf2 | C2CD3 | C2orf71 | C5orf42 | C8orf37 | CA2 | CA4 | CABP4 |
| CACNA1A | CACNA1F | CACNA1H | CACNA2D4 | CANT1 | CAPN15 | CAPN5 | CASK |
| CAV1 | CBS | CC2D2A | CCDC103 | CCDC114 | CCDC28B | CCDC39 | CCDC40 |
| CD320 | CD96 | CDH23 | CDH3 | CDHR1 | CDKN2A | CDON | CEP164 |
| CEP250 | CEP290 | CEP41 | CEP78 | CEP83 | CERKL | CFAP57 | CHD3 |
| CHD7 | CHM | CHMP4B | CHN1 | CHRD1 | CHST14 | CHST6 | CHSY1 |
| CIB2 | CISD2 | CKAP4 | CLCN7 | CLDN19 | CLN3 | CLN5 | CLN6 |
| CLN8 | CLPB | CLRN1 | CLUAP1 | CNBP | CNGA1 | CNGA3 | CNGB1 |
| CNGB3 | CNNM4 | COL11A1 | COL11A2 | COL17A1 | COL18A1 | COL1A1 | COL1A2 |
| COL2A1 | COL4A1 | COL4A3 | COL4A4 | COL4A5 | COL5A1 | COL5A2 | COL8A2 |
| COL9A1 | COL9A2 | COL9A3 | COLEC11 | COX14 | COX6B1 | COX7B | CP |
| CPAMD8 | CRB1 | CREBBP | CRELD1 | CRIP1 | CRTAP | CRX | CSP1 |
| CTC1 | CTCF | CTDP1 | CTNNA1 | CTNS | CTSA | CTSD | CTSF |
| CWC27 | CYP11B1 | CYP27A1 | CYP4V2 | DAG1 | DBH | DCDC1 | DCN |
| DDB2 | DDX11 | DDX58 | DFNB31 | DHCR7 | DHDDS | DHODH | DHX38 |
| DIAPH1 | DMD | DNAAF1 | DNAAF2 | DNAAF3 | DNAAF5 (HEATR2) | DNAH10 | DNAH11 |
| DNAH5 | DNAH9 | DNAI1 | DNAI2 | DNAJC17 | DNAJC19 | DNAJC5 | DNAL1 |
| DNM2 | DOCK6 | DPAGT1 | DPM1 | DRAM2 | DSE | DTHD1 | DTNBP1 |
| DYNC2H1 | EBP | EDN3 | EDNRA | EFEMP1 | EIF4G1 | ELOVL4 | ELP4 |
| EMC1 | EPG5 | EPHA2 | ERCC1 | ERCC2 | ERCC3 | ERCC4 | ERCC5 |
| ERCC6 | ERCC8 | ESCO2 | EVC | EVC2 | EXOSC2 | EXOSC8 | EYA1 |
| EYS | FAM111A | FAM126A | FAM161A | FANCA | FANCB | FANCC | FANCD2 |
| FANCE | FANCF | FANCG | FANCI | FANCL | FANCM | FAR1 | FASTKD2 |
| FBLN5 | FBN2 | FBXL4 | FIBP | FKBP14 | FKRP | FKTN | FLNB |
| FLVCR1 | FOXC1 | FOXC2 | FOXD1 | FOXO3 | FOXF2 | FOXH1 | FOXO2 |
| FOXRED1 | FRAS1 | FREM1 | FREM2 | FRMD7 | FSCN2 | FTL | FXN |
| FYCO1 | FZD4 | GALE | GALK1 | GALNS | GALT | GBA | GBA2 |
| GCNT2 | GCSH | GDF1 | GDF3 | GDF6 | GFER | GJA1 | GJA8 |
| GJB2 | GJB6 | GJE1 (GJC3) | GLB1 | GLDC | GLIS2 | GLIS3 | GM2A |

RETINAL DYSTROPHY XPANDED PANEL

GENE LIST (~875 GENES)

Note: Xpanded panel gene lists are regularly updated/improved. This list should be considered an approximation only as the gene list is subject to change at any time. The patient's test report includes a list of genes evaluated and includes a coverage parameter for each gene.

| | | | | | | | |
|-----------|---------|---------|----------------|----------|----------------|----------|----------------|
| GMPPA | GMPPB | GNAT1 | GNAT2 | GNB1 | GNB3 | GNPAT | GNPTG |
| GNS | GPC3 | GPHN | GPR125 | GPR143 | GPR179 | GRIP1 | GRK1 |
| GRM6 | GRN | GSN | GSS | GTF2H5 | GUCA1A | GUCA1B | GUCY2D |
| GUSB | HADHA | HADHB | HARS | HCCS | HCN1 | HDAC6 | HESX1 |
| HEXA | HEXB | HGSNAT | HK1 | HLCS | HMCN1 | HMGB3 | HMX1 |
| HNRNPDL | HOXB1 | HPS1 | HPS3 | HPS4 | HPS5 | HPS6 | HSF4 |
| HYLS1 | IARS2 | IBA57 | IDH3B | IDUA | IFIH1 | IFT122 | IFT140 |
| IFT172 | IFT27 | IFT43 | IFT52 | IFT74 | IFT80 | IFT81 | IFT88 |
| IGBP1 | IGFBP7 | IMPDH1 | IMPG1 | IMPG2 | INPP5E | INVS | IQCB1 |
| IRX1 | ISCA2 | ISPD | ITM2B | ITPR1 | JAG1 | JAM3 | KAT6B |
| KCNJ13 | KCNJ3 | KCNV2 | KCTD7 | KDM6A | KERA | KIAA0586 | KIAA1549 |
| KIDINS220 | KIF11 | KIF5A | KIF7 | KIZ | KLHL7 | KMT2D | KRT12 |
| KRT2 | KRT3 | LAMA1 | LAMB1 | LAMB2 | LARGE1 (LARGE) | LCA5 | LCT |
| LEFTY2 | LEMD2 | LHX2 | LIM2 | LMX1B | LONP1 | LOXHD1 | LOXL1 |
| LRAT | LRIT3 | LRP2 | LRP5 | LRPAP1 | LRRC6 | LSS | LTBP2 |
| LTBP3 | LYST | LZTFL1 | MAB21L2 | MAF | MAK | MAN2B1 | MAP3K7 |
| MAPKAPK3 | MBD5 | MBTPS2 | MCEE | MCOLN1 | MECR | MED25 | MERTK |
| MFF | MFN2 | MFRP | MFSD8 | MICALCL | MIP | MIPEP | MKKS |
| MKS1 | MLPH | MMAA | MMAB | MMACHC | MMADHC | MMP1 | MMP19 |
| MOCS1 | MPDU1 | MPLKIP | MRE11 (MRE11A) | MSMO1 | MTTP | MUT | MVK |
| MYH9 | MYO5A | MYO7A | MYOC | NAA10 | NBAS | NDP | NDUFA1 |
| NDUFA10 | NDUFA11 | NDUFA12 | NDUFA2 | NDUFA9 | NDUFAF1 | NDUFAF2 | NDUFAF3 |
| NDUFAF4 | NDUFAF5 | NDUFAF6 | NDUFB3 | NDUFS1 | NDUFS2 | NDUFS3 | NDUFS4 |
| NDUFS6 | NDUFS7 | NDUFS8 | NDUFV1 | NDUFV2 | NEK1 | NEK2 | NEK4 |
| NEK8 | NEU1 | NEUROD1 | NF1 | NHS | NKX2-5 | NLRP1 | NME8 |
| NMNAT1 | NOD2 | NODAL | NOTCH2 | NPC1 | NPHP1 | NPHP3 | NPHP4 |
| NR2E3 | NR2F1 | NRL | NSD1 | NT5C2 | NTF4 | NUB1 | NUS1 |
| NXNL1 | NYX | OAT | OCA2 | OCRL | OFD1 | OPA1 | OPA3 |
| OPN1LW | OPN1SW | OPTN | OSMR | OSTM1 | OTX2 | OVOL2 | P3H2 (LEPREL1) |
| P4HA2 | PANK2 | PAX2 | PAX3 | PAX6 | PCDH15 | PCNA | PCYT1A |
| PDE6A | PDE6B | PDE6C | PDE6D | PDE6G | PDE6H | PDZD7 | PET100 |
| PEX1 | PEX10 | PEX11B | PEX12 | PEX13 | PEX14 | PEX16 | PEX19 |
| PEX2 | PEX26 | PEX3 | PEX5 | PEX6 | PEX7 | PFKM | PGK1 |
| PHGDH | PHYH | PIEZO2 | PIGL | PIGT | PIGY | PIK3R1 | PIKFYVE |
| PITPNM3 | PITX2 | PITX3 | PLA2G5 | PLG | PLK4 | PLOD1 | PLOD3 |
| PLP1 | PMM2 | PNPLA6 | POC1B | POGZ | POLA1 | POMGNT1 | POMGNT2 |
| POMK | POMT1 | PORCN | PPT1 | PQBP1 | PRCD | PRDM13 | PRDM5 |
| PRIMPOL | PRKACA | PRKCG | PROM1 | PRPF3 | PRPF31 | PRPF4 | PRPF6 |
| PRPF8 | PRPH2 | PRPS1 | PRSS56 | PTCH1 | PTF1A | PTPN11 | PVRL3 |
| PXDN | RAB18 | RAB27A | RAB28 | RAB3GAP1 | RAB3GAP2 | RAD50 | RAI1 |
| RARB | RAX | RAX2 | RBP3 | RBP4 | RCBTB1 | RD3 | RDH11 |

RETINAL DYSTROPHY XPANDED PANEL

GENE LIST (~875 GENES)

Note: Xpanded panel gene lists are regularly updated/improved. This list should be considered an approximation only as the gene list is subject to change at any time. The patient's test report includes a list of genes evaluated and includes a coverage parameter for each gene.

| | | | | | | | |
|----------------|----------------|-----------------|---------------------|-----------------|----------------|-----------------|----------------|
| <i>RDH12</i> | <i>RDH5</i> | <i>RECQL4</i> | <i>REEP6</i> | <i>RERE</i> | <i>RGR</i> | <i>RGS9</i> | <i>RGS9BP</i> |
| <i>RHO</i> | <i>RIMS1</i> | <i>RIPK4</i> | <i>RLBP1</i> | <i>ROM1</i> | <i>RP1</i> | <i>RP1L1</i> | <i>RP2</i> |
| <i>RP9</i> | <i>RPE65</i> | <i>RPGR</i> | <i>RPGRIP1</i> | <i>RPGRIP1L</i> | <i>RPS19</i> | <i>RRM2B</i> | <i>RS1</i> |
| <i>RSPH4A</i> | <i>RSPH9</i> | <i>RSP01</i> | <i>SACS</i> | <i>SAG</i> | <i>SALL2</i> | <i>SALL4</i> | <i>SAMD9</i> |
| <i>SBF2</i> | <i>SC5D</i> | <i>SCARF2</i> | <i>SCN8A</i> | <i>SCO2</i> | <i>SDCCAG8</i> | <i>SDHAF1</i> | <i>SEC23A</i> |
| <i>SEMA3E</i> | <i>SEMA4A</i> | <i>SERPINH1</i> | <i>SH3PXD2B</i> | <i>SHH</i> | <i>SIL1</i> | <i>SIPA1L3</i> | <i>SIX3</i> |
| <i>SIX5</i> | <i>SIX6</i> | <i>SLC16A12</i> | <i>SLC24A1</i> | <i>SLC24A5</i> | <i>SLC25A1</i> | <i>SLC25A46</i> | <i>SLC2A1</i> |
| <i>SLC33A1</i> | <i>SLC38A8</i> | <i>SLC39A5</i> | <i>SLC45A2</i> | <i>SLC4A11</i> | <i>SLC4A4</i> | <i>SLC4A5</i> | <i>SLC4A7</i> |
| <i>SLC52A2</i> | <i>SLC52A3</i> | <i>SLC7A14</i> | <i>SLITRK6</i> | <i>SLX4</i> | <i>SMCHD1</i> | <i>SMG9</i> | <i>SMOC1</i> |
| <i>SMPD1</i> | <i>SNAI2</i> | <i>SNRNP200</i> | <i>SNX10</i> | <i>SNX3</i> | <i>SOD1</i> | <i>SON</i> | <i>SOX2</i> |
| <i>SOX3</i> | <i>SPARC</i> | <i>SPATA7</i> | <i>SPINT2</i> | <i>SPP2</i> | <i>SPTBN2</i> | <i>SRD5A3</i> | <i>SREBF2</i> |
| <i>ST3GAL5</i> | <i>STIM1</i> | <i>STK38L</i> | <i>STN1 (OBFC1)</i> | <i>STRA6</i> | <i>STS</i> | <i>STT3B</i> | <i>SUCLA2</i> |
| <i>SUOX</i> | <i>TACSTD2</i> | <i>TAT</i> | <i>TBC1D20</i> | <i>TBC1D7</i> | <i>TBCE</i> | <i>TBL1XR1</i> | <i>TCF4</i> |
| <i>TCIRG1</i> | <i>TCOF1</i> | <i>TCTN1</i> | <i>TCTN2</i> | <i>TCTN3</i> | <i>TDRD7</i> | <i>TEAD1</i> | <i>TEK</i> |
| <i>TENM1</i> | <i>TENM3</i> | <i>TFAP2A</i> | <i>TGFB1</i> | <i>TGFBI</i> | <i>TMM8A</i> | <i>TIMP3</i> | <i>TINF2</i> |
| <i>TLR3</i> | <i>TLR4</i> | <i>TMEM107</i> | <i>TMEM114</i> | <i>TMEM126A</i> | <i>TMEM138</i> | <i>TMEM216</i> | <i>TMEM231</i> |
| <i>TMEM237</i> | <i>TMEM5</i> | <i>TMEM67</i> | <i>TMEM70</i> | <i>TMEM98</i> | <i>TMTC3</i> | <i>TNFSF11</i> | <i>TNPO1</i> |
| <i>TOPORS</i> | <i>TPP1</i> | <i>TRAF3IP1</i> | <i>TREX1</i> | <i>TRIM32</i> | <i>TRIM37</i> | <i>TRIM44</i> | <i>TRNT1</i> |
| <i>TRPC1</i> | <i>TRPM1</i> | <i>TRPM2</i> | <i>TSPAN12</i> | <i>TTC21B</i> | <i>TTC8</i> | <i>TTL5</i> | <i>TTPA</i> |
| <i>TTR</i> | <i>TUB</i> | <i>TUBA8</i> | <i>TUBB</i> | <i>TUBGCP4</i> | <i>TUBGCP6</i> | <i>TULP1</i> | <i>TYR</i> |
| <i>TYRP1</i> | <i>UBE3B</i> | <i>UBIAD1</i> | <i>UBR1</i> | <i>UCHL1</i> | <i>UNC119</i> | <i>UNC45B</i> | <i>USH1C</i> |
| <i>USH1G</i> | <i>USH2A</i> | <i>USP9X</i> | <i>VAX1</i> | <i>VCAN</i> | <i>VIM</i> | <i>VPS13B</i> | <i>VSX1</i> |
| <i>VSX2</i> | <i>WAC</i> | <i>WASF3</i> | <i>WDPCP</i> | <i>WDR19</i> | <i>WDR34</i> | <i>WDR35</i> | <i>WDR36</i> |
| <i>WFS1</i> | <i>WRN</i> | <i>XPA</i> | <i>XPC</i> | <i>XPNPEP3</i> | <i>YAP1</i> | <i>YME1L1</i> | <i>ZEB1</i> |
| <i>ZEB2</i> | <i>ZIC3</i> | <i>ZNF408</i> | <i>ZNF423</i> | <i>ZNF469</i> | <i>ZNF513</i> | <i>ZNF592</i> | <i>ZNF644</i> |

Also Includes:

- The ORF15 region of the *RPGR* gene is included. However, this region has inherent sequence properties that yield suboptimal data and pathogenic variants in this region may not be reliably detected.
- Analysis of the recurrent c.2991+1655A>G variant in the *CEP290* gene
- Analysis of deep intronic variants of *ABCA4* previously reported in the literature in association with Stargardt disease (5196+1216C>A, 5196+1159G>A, 5196+1137G>A, 5196+1136C>A, 5196+1056A>G, 570+1798A>G, 1938-619A>G, 2160+584A>G, 3050+370C>T, 4540-2036C>A, 4539+2064C>T, 4539+2028C>T, 4539+2001G>A, 4539+1729G>T)