

# 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	AIP1	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	B3GALT
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
BTBD	BUB1B	C10orf11	C10orf2	C12orf57	C12orf65	C1orf186	C1QTNF5
C21orf2	C2CD3	C2orf71	C5orf42	C8orf37	CA2	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	CHRDL1	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	CRIPT	CRTAP	CRX	CSPP1	CTC1
CTCF	CTDP1	CTNNA1	CTNS	CTSA	CTSD	CTSF	CWC27
CYP1B1	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	FOXL2	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	GMPPA

# 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.

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)	MSM01	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	RDH12

# 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>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>TGFB1</i>	<i>TIMM8A</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>TLL5</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)



207 Perry Parkway

Gaithersburg, MD 20877

T 1 888 729 1206 (Toll-free), 1 301 519 2100 • F 1 201 421 2010

E [zebras@genedx.com](mailto:zebras@genedx.com) • [www.genedx.com](http://www.genedx.com)

