Foundation Fighting Blindness Clinical Consortium

Universal Rare Gene Study (Uni-Rare)

Uni-Rare RD Rare Gene List Report

Through 26 Oct 2026

Gene	Common Inheritance Pattern	Variable Inheritance Pattern	Uni-Rare Registry Gene	Uni-Rare NHS Target Gene
ABCC6	Recessive	Yes	Yes	No
ABCD1	X-linked	No	Yes	No
ABHD12	Recessive	No	Yes	No
ACBD5	Recessive	No	Yes	No
ACO2	Recessive	No	Yes	No
ADAM9	Recessive	No	Yes	No
ADAMTS18	Recessive	No	Yes	No
ADGRV1 (aka GRP98)	Recessive	No	Yes	Yes
ADIPOR1	AD/AR	Yes	Yes	No
AFG3L2	Dominant	No	Yes	No
AGBL5	Recessive	No	Yes	No
AHI1	Recessive	No	Yes	No
AHR	Recessive	No	Yes	No
AIPL1	Recessive	Yes	Yes	No
AIRE	Recessive	Yes	Yes	No
ALDH3A2	Recessive	No	Yes	No
ALMS1	Recessive	No	Yes	No
ALPK1	Dominant	No	Yes	No
AMACR	Recessive	No	Yes	No
AP5M1	Recessive	No	Yes	No
AP5Z1	Recessive	No	Yes	No
ARHGEF18	Recessive	No	Yes	No
ARL13B	Recessive	No	Yes	No
ARL2BP	Recessive	No	Yes	No
ARL3	AD/AR	Yes	Yes	No
ARL6	Recessive	No	Yes	No
ARMC9	Recessive	No	Yes	No
ARSG	Recessive	No	Yes	No
ASRGL1	Recessive	No	Yes	No
ATF6	Recessive	No	Yes	No
ATOH7	Recessive	No	Yes	No
ATXN7	Dominant	No	Yes	No

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B9D1	Recessive	No	Yes	No
B9D2	Recessive	No	Yes	No
BBIP1	Recessive	No	Yes	No
BBS1	Recessive	No	Yes	No
BBS10	Recessive	No	Yes	No
BBS12	Recessive	No	Yes	No
BBS2	Recessive	No	Yes	No
BBS4	Recessive	No	Yes	No
BBS5	Recessive	No	Yes	No
BBS7	Recessive	No	Yes	No
BBS9	Recessive	No	Yes	No
BEST1 (aka VMD2)	Dominant	Yes	Yes	No
C12orf65	Recessive	No	Yes	No
C19ORF44	Recessive	No	Yes	No
C1QTNF5	Dominant	No	Yes	No
C21orf2 (aka CFAP410)	Recessive	No	Yes	No
C2CD3	Recessive	No	Yes	No
C2orf71 (aka PCARE)	Recessive	No	Yes	Yes
C5AR2	Dominant	No	Yes	No
C5orf42 (aka CPLANE1)	Recessive	No	Yes	No
C8orf37	Recessive	No	Yes	No
CA4	Dominant	No	Yes	No
CABP4	Recessive	No	Yes	No
CACNA1F (aka iCSNb2)	X-linked	No	Yes	No
CACNA2D4	Recessive	No	Yes	No
CAPN5	Dominant	No	Yes	No
CC2D2A	Recessive	No	Yes	No
CCDC188	Recessive	No	Yes	No
CCT2	Recessive	No	Yes	No
CDH23	Recessive	No	Yes	Yes

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CDH3	Recessive	No	Yes	No
CDHR1	Recessive	No	Yes	Yes
CEP104	Recessive	No	Yes	No
CEP120	Recessive	No	Yes	No
CEP164	Recessive	No	Yes	No
CEP19	Recessive	No	Yes	No
CEP250	Recessive	No	Yes	No
CEP41	Recessive	No	Yes	No
CEP78	Recessive	No	Yes	No
CEP83	Recessive	No	Yes	No
CERKL	Recessive	No	Yes	Yes
CIB2	Recessive	No	Yes	No
CISD2	Recessive	No	Yes	No
CLCC1	Recessive	No	Yes	No
CLCN2	Recessive	No	Yes	No
CLDN19	Recessive	No	Yes	No
CLEC3B	Dominant	No	Yes	No
CLN3	Recessive	No	Yes	No
CLN5	Recessive	No	Yes	No
CLN6	Recessive	No	Yes	No
CLN8	Recessive	No	Yes	No
CLRN1 (aka USH3/USH3A)	Recessive	No	Yes	Yes
CLUAP1	Recessive	No	Yes	No
CNGA1	Recessive	No	Yes	Yes
CNGA3	Recessive	No	Yes	No
CNGB1	Recessive	No	Yes	Yes
CNGB3	Recessive	No	Yes	No
CNNM4	Recessive	No	Yes	No
COL11A1	Dominant	Yes	Yes	No
COL11A2	Dominant	Yes	Yes	No
COL18A1	Recessive	No	Yes	No
COL2A1	Dominant	No	Yes	No

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COL4A3	Recessive	Yes	Yes	No
COL4A4	Recessive	No	Yes	No
COL9A1	Recessive	No	Yes	No
COL9A2	Recessive	No	Yes	No
COL9A3	Recessive	No	Yes	No
COQ2	Recessive	No	Yes	No
COQ8B	Recessive	No	Yes	No
CRB1	Recessive	Yes	Yes	No
CRB2	Recessive	No	Yes	No
CREB3	Recessive	No	Yes	No
CRX	Dominant	Yes	Yes	No
CSPP1	Recessive	No	Yes	No
CTC1	Recessive	No	Yes	No
CTNNA1	Dominant	No	Yes	No
CTNNB1	Dominant	No	Yes	No
CTNND1	Dominant	No	Yes	No
CTSD	Recessive	No	Yes	No
CWC27	Recessive	No	Yes	No
CYP2U1	Recessive	No	Yes	No
CYP4V2	Recessive	No	Yes	No
DACT2	Recessive	No	Yes	No
DDR1	Recessive	No	Yes	No
DFNB31 (aka WHRN)	Recessive	No	Yes	No
DHDDS	Recessive	No	Yes	No
DHX32	Recessive	No	Yes	No
DHX38	Recessive	No	Yes	No
DNAJC17	Recessive	No	Yes	No
DRAM2	Recessive	No	Yes	No
DSCAML1	Recessive	No	Yes	No
DTHD1	Recessive	No	Yes	No
DYNC2H1 (aka WDR34)	Recessive	No	Yes	No
DYRK1A	Dominant	No	Yes	No

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EFEMP1	Dominant	No	Yes	No
ELOVL4	Dominant	Yes	Yes	No
ENSA	Recessive	No	Yes	No
ERCC6	Recessive	No	Yes	No
ERCC8	Recessive	No	Yes	No
ESPN	Recessive	No	Yes	No
ESRRB	Recessive	No	Yes	No
EXOSC2 (aka RRP4)	Recessive	No	Yes	No
FAM161A	Recessive	No	Yes	Yes
FBN2	Dominant	No	Yes	No
FBN3	Recessive	No	Yes	No
FLVCR1	Recessive	No	Yes	No
FSCN2	Dominant	No	Yes	No
FZD4	Dominant	Yes	Yes	No
GDF6	Recessive	No	Yes	No
GNAT1	AD/AR	Yes	Yes	No
GNAT2	Recessive	No	Yes	No
GNB3	Recessive	No	Yes	No
GNPTG	Recessive	No	Yes	No
GPR125 (aka ADGRA3)	Recessive	No	Yes	No
GPR179	Recessive	No	Yes	No
GPR45	Recessive	No	Yes	No
GRK1	Recessive	No	Yes	No
GRM6	Recessive	No	Yes	No
GRN	Recessive	No	Yes	No
GUCA1A	Dominant	No	Yes	No
GUCA1B	Dominant	No	Yes	No
GUCY2D	Dominant	Yes	Yes	No
HARS	Recessive	No	Yes	No
HGSNAT	Recessive	No	Yes	No
HK1	Dominant	Yes	Yes	No
HKDC1	Recessive	No	Yes	No

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Gene	Common Inheritance Pattern	Variable Inheritance Pattern	Uni-Rare Registry Gene	Uni-Rare NHS Target Gene
HMX1	Recessive	No	Yes	No
IDH3A	Recessive	No	Yes	No
IDH3B	Recessive	No	Yes	No
IDH3G	X-linked	No	Yes	No
IFT140	Recessive	No	Yes	No
IFT172	Recessive	No	Yes	No
IFT27	Recessive	No	Yes	No
IFT43	Recessive	No	Yes	No
IFT74	Recessive	No	Yes	No
IFT80	Recessive	No	Yes	No
IFT81	Recessive	No	Yes	No
IFT88	Recessive	No	Yes	No
IKBKG	X-linked	No	Yes	No
IMPDH1	Dominant	No	Yes	Yes
IMPG1	AD/AR	Yes	Yes	Yes
IMPG2	Recessive	Yes	Yes	Yes
INPP5E	Recessive	No	Yes	No
INVS	Recessive	No	Yes	No
IQCB1 (aka NPHP5)	Recessive	No	Yes	No
ITM2B	Dominant	No	Yes	No
JAG1	Dominant	No	Yes	No
KCNJ13	Recessive	Yes	Yes	No
KCNV2	Recessive	No	Yes	Yes
KIAA0556	Recessive	No	Yes	No
KIAA0586	Recessive	No	Yes	No
KIAA0753	Recessive	No	Yes	No
KIAA1549	Recessive	No	Yes	No
KIF11	Dominant	No	Yes	No
KIF3B	Dominant	No	Yes	No
KIF7	Recessive	No	Yes	No
KIZ	Recessive	No	Yes	Yes
KLHL7	Dominant	Yes	Yes	No

Gene	Common Inheritance Pattern	Variable Inheritance Pattern	Uni-Rare Registry Gene	Uni-Rare NHS Target Gene
LAMA1	Recessive	No	Yes	No
LAMP2	Dominant	No	Yes	No
LCA5	Recessive	No	Yes	No
LRAT	Recessive	No	Yes	No
LRIT3	Recessive	No	Yes	No
LRP5	AD/AR	Yes	Yes	No
LRRTM4	Dominant	No	Yes	No
LZTFL1	Recessive	No	Yes	No
MAK	Recessive	No	Yes	Yes
MAN2B1	Recessive	No	Yes	No
MAPKAPK3	Dominant	No	Yes	No
MCOLN1	Recessive	No	Yes	No
MED12	X-linked	No	Yes	No
MERTK	Recessive	No	Yes	No
MFRP	Recessive	No	Yes	No
MFSD8	Recessive	No	Yes	No
MIR204	Dominant	No	Yes	No
MKKS	Recessive	No	Yes	No
MKS1	Recessive	No	Yes	No
MMACHC	Recessive	No	Yes	No
MORC2	Dominant	No	Yes	No
MPDZ	Dominant	No	Yes	No
MSTO1	Recessive	No	Yes	No
MT-ATP6	Mitochondrial	No	Yes	No
MT-ATP8	Mitochondrial	No	Yes	No
MT-CO1	Mitochondrial	No	Yes	No
MT-CO2	Mitochondrial	No	Yes	No
MT-CO3	Mitochondrial	No	Yes	No
MT-CYB	Mitochondrial	No	Yes	No
MT-ND2	Mitochondrial	No	Yes	No
MT-ND3	Mitochondrial	No	Yes	No
MT-RNR1	Mitochondrial	No	Yes	No
MT-RNR2	Mitochondrial	No	Yes	No

Gene	Common Inheritance Pattern	Variable Inheritance Pattern	Uni-Rare Registry Gene	Uni-Rare NHS Target Gene
MT-TA	Mitochondrial	No	Yes	No
MT-TC	Mitochondrial	No	Yes	No
MT-TD	Mitochondrial	No	Yes	No
MT-TE	Mitochondrial	No	Yes	No
MT-TF	Mitochondrial	No	Yes	No
MT-TG	Mitochondrial	No	Yes	No
MT-TH	Mitochondrial	No	Yes	No
MT-TI	Mitochondrial	No	Yes	No
MT-TK	Mitochondrial	No	Yes	No
MT-TL1	Mitochondrial	No	Yes	No
MT-TL2	Mitochondrial	No	Yes	No
MT-TM	Mitochondrial	No	Yes	No
MT-TN	Mitochondrial	No	Yes	No
MT-TP (Not MTTP)	Mitochondrial	No	Yes	No
MT-TQ	Mitochondrial	No	Yes	No
MT-TR	Mitochondrial	No	Yes	No
MT-TS1	Mitochondrial	No	Yes	No
MT-TS2	Mitochondrial	No	Yes	No
MT-TT	Mitochondrial	No	Yes	No
MT-TV	Mitochondrial	No	Yes	No
MT-TW	Mitochondrial	No	Yes	No
MT-TY	Mitochondrial	No	Yes	No
mtDNA deletion (ie. Kearns-Sayre Syndrome)	Mitochondrial	No	Yes	No
MTTP (not MT-TP)	Recessive	No	Yes	No
MVK	Recessive	No	Yes	No
MYO7A	Recessive	No	Yes	Yes
NAGLU	Recessive	No	Yes	No
NBAS	Recessive	No	Yes	No
ND1 (MT-ND1)	Mitochondrial	No	Yes	No
ND5 (MT-ND5)	Mitochondrial	No	Yes	No
ND6 (MT-ND6)	Mitochondrial	No	Yes	No

Gene	Common Inheritance Pattern	Variable Inheritance Pattern	Uni-Rare Registry Gene	Uni-Rare NHS Target Gene
NDP	X-linked	No	Yes	No
NEK2	Recessive	No	Yes	No
NEUROD1	Recessive	No	Yes	No
NMNAT1 (aka LCA9)	Recessive	No	Yes	No
NPHP1	Recessive	No	Yes	No
NPHP3	Recessive	No	Yes	No
NPHP4	Recessive	No	Yes	No
NR2E3	Recessive	Yes	Yes	No
NRL	AD/AR	Yes	Yes	No
NYX	X-linked	No	Yes	No
OFD1	X-linked	No	Yes	No
OPN1LW and OPN1MW (and/or Locus Control Region of these genes)	X-linked	No	Yes	No
OPN1LW only	X-linked	No	Yes	No
OPN1MW only	X-linked	No	Yes	No
OPN1SW	Dominant	No	Yes	No
OTX2	Dominant	No	Yes	No
P3H2	Recessive	No	Yes	No
PANK2	Recessive	No	Yes	No
PAX2	Dominant	No	Yes	No
PAX6	Dominant	No	Yes	No
PCYT1A	Recessive	No	Yes	No
PDE6A	Recessive	No	Yes	No
PDE6B	Recessive	No	Yes	No
PDE6C	Recessive	No	Yes	No
PDE6D	Recessive	No	Yes	No
PDE6G	Recessive	No	Yes	No
PDE6H	Recessive	No	Yes	No
PDSS1	Recessive	No	Yes	No
PEX1	Recessive	No	Yes	No
PEX10	Recessive	No	Yes	No

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PEX11B	Recessive	No	Yes	No
PEX12	Recessive	No	Yes	No
PEX13	Recessive	No	Yes	No
PEX14	Recessive	No	Yes	No
PEX16	Recessive	No	Yes	No
PEX19	Recessive	No	Yes	No
PEX2	Recessive	No	Yes	No
PEX26	Recessive	No	Yes	No
PEX3	Recessive	No	Yes	No
PEX5	Recessive	No	Yes	No
PEX6	Recessive	No	Yes	No
PEX7	Recessive	No	Yes	No
PGK1	X-linked	No	Yes	No
PHYH	Recessive	No	Yes	No
PISD	Recessive	No	Yes	No
PITPNM3	Dominant	No	Yes	No
PLA2G5	Recessive	No	Yes	No
PLK4	Recessive	No	Yes	No
PNPLA6	Recessive	No	Yes	No
POC1B	Recessive	No	Yes	No
POC5	Recessive	No	Yes	No
POMGNT1	Recessive	No	Yes	No
PPP2R3C	Recessive	No	Yes	No
PPT1	Recessive	No	Yes	No
PRCD	Recessive	No	Yes	No
PRDM13	Dominant	No	Yes	No
PROM1	AD/AR	Yes	Yes	No
PROS1	Recessive	No	Yes	No
PRPF3	Dominant	No	Yes	Yes
PRPF31	Dominant	No	Yes	Yes
PRPF4	Dominant	No	Yes	No
PRPF6	Dominant	No	Yes	No
PRPF8	Dominant	No	Yes	Yes

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Gene	Common Inheritance Pattern	Variable Inheritance Pattern	Uni-Rare Registry Gene	Uni-Rare NHS Target Gene
PRPH2	Dominant	Yes	Yes	Yes
PRPS1	X-linked	No	Yes	No
PSEN1	Recessive	No	Yes	No
PYGM	Recessive	No	Yes	No
RAB28	Recessive	No	Yes	No
RAX2	AD/AR	Yes	Yes	No
RBP3	Recessive	No	Yes	No
RBP4	Recessive	Yes	Yes	No
RCBTB1	Recessive	No	Yes	No
RD3	Recessive	No	Yes	No
RDH11	Recessive	No	Yes	No
RDH12	Recessive	Yes	Yes	Yes
RDH5	Recessive	No	Yes	No
REEP6	Recessive	No	Yes	No
RGR	AD/AR	Yes	Yes	No
RGS9	Recessive	No	Yes	No
RGS9BP	Recessive	No	Yes	No
RHBDD2	Recessive	No	Yes	No
RHO	Dominant	Yes	Yes	Yes
RIMS1	Dominant	No	Yes	No
RIMS2	Recessive	No	Yes	No
RLBP1	Recessive	No	Yes	No
RNU4-2	Dominant	No	Yes	No
RNU4ATAC	Recessive	No	Yes	No
RNU6-1	Dominant	No	Yes	No
RNU6-2	Dominant	No	Yes	No
RNU6-8	Dominant	No	Yes	No
RNU6-9	Dominant	No	Yes	No
ROM1	AD/AR	Yes	Yes	No
RP1	Dominant	Yes	Yes	No
RP1L1	AD/AR	Yes	Yes	No
RP2	X-linked	No	Yes	Yes
RP9	Dominant	No	Yes	No

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RPGRIP1	Recessive	No	Yes	No
RPGRIP1L	Recessive	No	Yes	No
RRM2B	Recessive	Yes	Yes	No
RS1	X-linked	No	Yes	No
RTN4IP1	Recessive	No	Yes	No
SAG	Dominant	Yes	Yes	No
SAMD11	Recessive	No	Yes	No
SAMD7	Recessive	No	Yes	No
SCAPER	Recessive	No	Yes	No
SCLT1	Recessive	No	Yes	No
SDCCAG8 (aka BBS16)	Recessive	No	Yes	No
SEMA4A	Recessive	No	Yes	No
SGSH	Recessive	No	Yes	No
SIX6	Recessive	No	Yes	No
SLC24A1	Recessive	No	Yes	No
SLC25A46	Recessive	No	Yes	No
SLC37A3	Recessive	No	Yes	No
SLC39A12	Recessive	No	Yes	No
SLC4A7	Recessive	No	Yes	No
SLC66A1	Recessive	No	Yes	No
SLC6A6	Recessive	No	Yes	No
SLC7A14	Recessive	No	Yes	No
SMARCA4	Dominant	No	Yes	No
SNRNP200 (aka ASCC3L1)	Dominant	Yes	Yes	Yes
SPATA7	Recessive	No	Yes	No
SPG11	Recessive	No	Yes	No
SPP2	Dominant	No	Yes	No
SRD5A3	Recessive	No	Yes	No
SSBP1	Dominant	No	Yes	No
STN1	Recessive	No	Yes	No
STX3	Recessive	No	Yes	No
SUMF1	Recessive	No	Yes	No

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TBC1D32	Recessive	No	Yes	No
TCTN1	Recessive	No	Yes	No
TCTN2	Recessive	No	Yes	No
TCTN3	Recessive	No	Yes	No
TEAD1	Dominant	No	Yes	No
THRB	Dominant	No	Yes	No
TIMP3	Dominant	No	Yes	No
TINF2	Dominant	No	Yes	No
TLCD3B	Recessive	No	Yes	No
TMED7	Recessive	No	Yes	No
TMEM107	Recessive	No	Yes	No
TMEM138	Recessive	No	Yes	No
TMEM216	Recessive	No	Yes	No
TMEM218	Recessive	No	Yes	No
TMEM231	Recessive	No	Yes	No
TMEM237	Recessive	No	Yes	No
TMEM67	Recessive	No	Yes	No
TOPORS	Dominant	No	Yes	No
TPP1 (aka CLN2)	Recessive	No	Yes	No
TRAF3IP1	Recessive	No	Yes	No
TREX1	AD/AR	Yes	Yes	No
TRIM32	Recessive	No	Yes	No
TRNT1	Recessive	No	Yes	No
TRPM1	Recessive	No	Yes	No
TSPAN12	Dominant	Yes	Yes	No
TTC21B	Recessive	No	Yes	No
TTC8	Recessive	No	Yes	No
TTLL5	Recessive	No	Yes	No
TTPA	Recessive	No	Yes	No
TUB	Recessive	No	Yes	No
TUBB4B	Dominant	No	Yes	No
TUBGCP4	Recessive	No	Yes	No
TUBGCP6	Dominant	No	Yes	No

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TULP1	Recessive	No	Yes	No
UBAP1L	Recessive	No	Yes	No
UNC119	Dominant	No	Yes	No
USH1C	Recessive	Yes	Yes	No
USH1G	Recessive	No	Yes	No
USP45	Recessive	No	Yes	No
VCAN	Dominant	No	Yes	No
VPS13B	Recessive	No	Yes	No
VSX2	Recessive	No	Yes	No
WDPCP	Recessive	No	Yes	No
WDR19	Recessive	No	Yes	No
WFS1	Recessive	Yes	Yes	No
ZFYVE26	Recessive	No	Yes	No
ZNF408	AD/AR	Yes	Yes	No
ZNF423	AD/AR	Yes	Yes	No
ZNF513	Recessive	No	Yes	No