IRD-associated genes (n=519)

RetiGene

Non-syndromic (n=226)

Others (non-syndromic) (n=31)

ADAMTS18, ARR3, ATOH7, CABP4, CAPN5, CHM, FRMD7, GPR143*, LOXL3*, MAPKAPK3, MFRP, MIR204*, PAX6*,PLA2G5, RDH5, RGS9, RGS9BP, RS1, SIX6, SLC38A8, SSBP1, SUMF1, TEAD1,WDR19

MD (n=31)

AP5B1, AP5Z1, CFH, CLEC3B, CLN5, EFEMP1, ELOVL4, FBLN5, MCDR1 locus, MCDR3 locus, SAMD7, THRB, TIMP3

CVD (n=10)

GNAT2, OPN1SW, PDE6H

CD / CRD (n=38)

ATF6, CNGA3, CNGB3, OPN1LW, OPN1MW, PDE6C

ABCA4, MFSD8, PROM1

ADAM9, CACNA2D4, DRAM2, KCNV2, POC1B, RAB28, TLCD3B, TTLL5

CSNB (n=15)

EGFLAM, GNB3, GPR179, GRK1, GRM6, LRIT3, NYX, TRPM1

CRD: CACNA1F

C1QTNF5

EVR: CTNNA1

BEST1

IMPG1, IMPG2, RP1L1

CYP4V2*, NR2E3, RLBP1

CDHR1, CERKL, EYS, GUCA1A, RP1, RPGR

CRX, CRB1*, PRPH2

ALMS1, NMNAT1

GNAT1, PDE6B, RHO, SAG, SLC24A1

RP (n=113)

AGBL5, AHI1, ARHGEF18, ARL2BP, ARL3, ARL6, BBS1, BBS2, BBS9, BCOR, C19orf44, CC2D2A, CEP162, CEP250, CFAP20, CFAP410, CLCC1, CLN3, CLRN1, CNGA1, CNGB1, COQ2, COQ5, COQ8B, DHDDS, DHX38, DYNC2H1, FAM161A, FLVCR1, FSD1L, HGSNAT, HK1, IDH3A, IDH3B, IDH3G, IFT172, KIAA1549, KIZ, KLHL7, MAK, MERTK, MVK, NRL, OFD1, PDE6A, PDE6G, PDSS1, POMGNT1, PRCD, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, RBP3*, REEP6, RNU4-2, RNU6-1, RNU6-2, RNU6-8, RNU6-9, RP17 locus, RP2, SAXO6, SLC37A3, SLC4A7, SLC66A1, SLC7A14, SNRNP200, TBC1D32, TMEM216, TOPORS, TTC8, USH2A, Xq27.1 locus

EVR: ZNF408

CFAP418, CREB3, RAX2, PCARE, RCBTB1, UBAP1L

TULP1

CSNB: GUCY2D

CVD: RPGRIP1

LCA (n=22)

AIPL1, IFT140, IMPDH1, INPP5E, LRAT, RDH12, RPE65, SPATA7

CEP290, IQCB1, LCA5, PCYT1A, RD3

Others (non-syndromic): KCNJ13*

OA (n=26)

ACO2, AFG3L2, DNAJC30, DNM1L, MCAT, MT-CO1, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, NDUFA12, NDUFAF4, NDUFAF8, NDUFS7, NDUFV1, NSUN3, OPA1, OPA3, PPIB, RTN4IP1, SPG7, TMEM126A

EVR (n=11)

CTNNB1, CTNND1, FZD4, KIF11, LRP5, NDP, P3H2*, TSPAN12, VCAN*

Syndromic (n=355)

Joubert (n=21)

AHI1, ARL13B, ARL3, ARMC9, CC2D2A, CEP104, CEP41, CSPP1, KIAA0586, OFD1, PDE6D, TMEM138, TMEM216, TMEM218, TMEM231, TMEM67

SLS / NPHP (n=8)

IQCB1, NPHP4, SCLT1, TRAF3IP1

NPHP1

CEP290, INPP5E, RNU4ATAC

MKS1

CEP164, SDCCAG8

IFT172, SCAPER

BBS (n=22)

ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CFAP418, IFT27, IFT74, LZTFL1, MKKS, TTC8

Others (syndromic) (n=171)

ABCC6, ACBD5, ACO2, AGBL5, AGXT, AIRE, ALDH3A2, ALMS1, ALPK1, AMACR, AP3D1, AP5M1, AP5Z1, ARL2BP, ATXN7, BRAT1, CDH3, CEP78, CEP83, CFAP410, CHD7, CLCN2, CLN3, CLN5, CLN8, CNNM4, COG4, COL11A1, COL18A1, COL2A1, COL4A1, COL4A4, COL4A5, COL9A1, COL9A2, COL9A3, COQ2, COQ8B, CRPPA, CTC1, CTNNB1, CTNS, CTSD, CWC27, CYP2R1, CYP2U1, DHX16, DOCK6, DPM1, DYNC2H1, DYNC2I2, EDNRB, EMC1, ERCC3, ERCC6, ERCC8, EXOSC2, FDXR, FKRP, FLVCR1, GNB1, GPATCH11, GTPBP2, HADHA, HGSNAT, HK1, HMX1, HPS3, HPS4, HSD17B10, IDS, IFT140, IGFBP7, IKBKG, INTS11, JAG1, KIF11, KIF3B, KLHL7, LAMA1, LAMB2, LAMP2, LIG3, LRP2, LRRC32, LRRC8C, LYST, MAB21L2, MAN2B1, MCOLN1, MED12, MFSD8, MITF, MMACHC, MSTO1, MT-TE, MT-TL1, MT-TP, MT-TS2, MT-TV, MVK, NDP, NEUROD1, NGLY1, NMNAT1, NOTCH2NLC, OAT, OTX2, PANK2, PAX2, PCYT1A, PDSS1, PEX1, PEX2, PEX6, PEX7, PGK1, PHYH, PISD, PMM2, PNPLA6, POC5, POMGNT1, POMT1, PPT1, PRPS1, PYGM, RBP4, RCBTB1, RIMS2, RPGR, RXYLT1, SACS, SGSH, SLC19A2, SLC38A3, SLC6A6, SMPD1, SPG11, SPTLC1, SSBP1, STN1, STX3, SUMF1, TBC1D32, TGFBR2, TIMP3, TINF2, TPP1, TREX1, TRNT1, TTLL5, TTPA, TUB, TUBB4B, TUBGCP4, TUBGCP6, VPS13B, YARS1, ZFYVE26, ZNF423

SLS / NPHP: WDR19

USH (n=12)

ADGRV1, ARSG, CDH23, CEP250, CLRN1, MYO7A, PCDH15, USH1C, USH1G, USH2A, WHRN

ABHD12

Syndromic OA (n=120)

AFG3L2, AMPD2, ANKRD17, ANTXR1, AP3B2, ATAD3A, ATG7, ATP1A3, ATP6V1A, ATRX, ATXN1, B3GALNT2, BCAP31, BLOC1S1, BOLA3, BRAF, BTD, C19orf12, CCDC88A, CDC42, CISD2, CLCN3, CRIPT, CYFIP2, DDHD2, DIAPH1, DNAJC19, DNAJC30, DYRK1A, EIF2B4, ELOVL1, EPRS1, FA2H, FASTKD2, FDX2, GATAD2B, GJA1, HESX1, HIKESHI, HNRNPK, IBA57, ISCA2, KIF1A, KLC2, LAMB1, LETM1, LYRM7, MAG, MECR, MFF, MFN2, MINPP1, MPZ, MT-ATP6, MT-CO2, MT-ND3, MT-ND5, MT-ND6, MT-TH, MTO1, MTRFR, NADK2, NARS2, NBAS, NDUFA1, NDUFA10, NDUFA12, NDUFAF2, NDUFAF3, NDUFS2, NR2F1, NSUN3, OPA1, OPA3, PARS2, PCCB, PCLO, PDXK, PLA2G6, PNPT1, POLG, POLR3A, POLR3B, PPP1R21, PROKR2, PTCD3, PTPN23, PUF60, RAB18, RAB3GAP1, RERE, RNF170, RTN4IP1, SDHA, SDHB, SERAC1, SLC19A3, SLC25A46, SLC44A1, SLC52A2, SNF8, SNX10, SOX2, SPG7, SRD5A3, ST3GAL5, STAMBP, SURF1, TFG, TIMM8A, TMEM126A, TMEM53, TRAPPC12, TSFM, UCHL1, WDR73, ZNHIT3,16p13.3 triplication locus

Other syndromic: ATXN2, WFS1

OCA (n=17)

AHR, AP3B1, BLOC1S3, BLOC1S5, BLOC1S6, DCT, DTNBP1, EPG5, HPS1, HPS5, HPS6, LRMDA, OCA2, SLC24A5, SLC45A2, TYR,TYRP1

Underlined genes are linked to both non-syndromic and syndromic phenotypes
Asterisks point to genes that can also be involved in non-retinal ocular diseases