IRD-associated genes (n=541)

RetiGene

Non-syndromic (n=233)

Syndromic (n=375)

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=33)

AP5B1, AP5Z1, CFH, CLEC3B, CLN5, EFEMP1, ELOVL4, FBLN5, MCDR1 locus, MCDR3 locus, SAMD7, THRB, TIMP3, ZZEF1-ALOX15 locus

CVD (n=10)

GNAT2, OPN1SW, PDE6H

ATF6, CNGA3, CNGB3, OPN1LW, OPN1MW, PDE6C

CD / CRD (n=40)

ABCA4, MFSD8, PROM1

ADAM9, CACNA2D4, DRAM2, KCNV2, POC1B, RAB28, TLCD3B, XXYLT1

CSNB (n=15)

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

GNAT1, PDE6B, RHO, SAG, SLC24A1

CRD

CACNA1F

C1QTNF5

BEST1

CTNNA1

EVR

IMPG1, IMPG2, RP1L1

CYP4V2*, NR2E3, RLBP1

CDHR1, CERKL, EYS, GUCA1A, HK1, RP1, RPGR

CRX, CRB1*, PRPH2

ALMS1, NMNAT1

CFAP418, CREB3, RAX2, PCARE, TTLL5, RCBTB1, UBAP1L

TULP1

GUCY2D

CSNB

RPGRIP1

CVD

OA (n=27)

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, NDUFS1, NDUFS7, NDUFV1, NSUN3, OPA1, OPA3, PPIB, RTN4IP1, SPG7, TMEM126A

RP (n=118)

AGBL5, AHI1, ARHGEF18, ARL2BP, ARL3, ARL6, BBS1, BBS2, BBS9, BCOR, C19orf44, CC2D2A, CEP162, CEP250, CFAP20, CFAP410, CLCC1, CLN3, CLRN1, CLUAP1, CNGA1, CNGB1, COQ2, COQ5, COQ8B, DHDDS, DHX38, DYNC2H1, FAM161A, FLVCR1, FSD1L, HGSNAT, IDH3A, IDH3B, IDH3G, IDUA, IFT172, KIAA1549, KIZ, KLHL7, MAK, MERTK, MVK, NRL, OFD1, PDE6A, PDE6G, PDSS1, POMGNT1, PRCD, PRKCZ, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, RBP3*, REEP6, RNU4-2, RNU6-1, RNU6-2, RNU6-8, RNU6-9, RP17 locus, RP2, SAXO6, SCLT1, SLC37A3, SLC4A7, SLC66A1, SLC7A14, SNRNP200, TBC1D32, TMEM216, TOPORS, TTC8, USH2A, Xq27.1 locus

ZNF408

EVR

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

CEP290, IQCB1, LCA5, PCYT1A, RD3

LCA (n=22)

KCNJ13*

Others (non-syndromic)

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

EVR (n=11)

Others (syndromic) (n=177)

USH / USH-like (n=15)

ADGRV1, AGBL5, ARSG, CDH23, CEP250, CEP78, CLRN1, MYO7A, PCDH15, TUBB4B, USH1C, USH1G, USH2A, WHRN

ABHD12

Syndromic OA (n=131)

AAAS, ACAD9, AFG3L2, AMPD2, ANKRD17, ANTXR1, AP3B2, ATAD3A, ATG7, ATP1A3, ATP6V1A, ATRX, ATXN1, B3GALNT2, BCAP31, BLOC1S1, BOLA3, BORCS5, BORCS8, BRAF, BTD, C19orf12, CASK, CCDC88A, CDC42, CISD2, CLCN3, CRIPT, CYFIP2, DDHD2, DIAPH1, DNAJC19, DNAJC30, DYRK1A, EIF2B4, ELOVL1, EPRS1, FA2H, FASTKD2, FDX2, FSD1L, GATAD2B, GJA1, HECW2, 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, NAGA, NARS2, NBAS, NDUFA1, NDUFA10, NDUFA12, NDUFAF2, NDUFAF3, NDUFS1, NDUFS2, NFIX, 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, SOX5, SPG7, SRD5A3, ST3GAL5, STAMBP, SURF1, TBCE, TFG, TIMM8A, TMEM126A, TMEM53, TRAPPC12, TSFM, UCHL1, WDR73, ZNHIT3, 16p13.3 triplication locus

ATXN2, WFS1

Other syndromic

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

CEP290, INPP5E, RNU4ATAC

NPHP1

MKS1

CEP164, SDCCAG8

IFT172, SCAPER

BBS (n=22)

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

ABCC6, ACBD5, ACO2, AGXT, AIRE, ALDH3A2, ALMS1, ALPK1, AMACR, AP3D1, AP5M1, AP5Z1, ARL2BP, ATXN7, BRAT1, CDH3, 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, DSCAM, DYNC2H1, DYNC2I2, EDNRB, EMC1, ERCC3, ERCC6, ERCC8, EXOSC2, FDXR, FKRP, FLVCR1, GNB1, GPATCH11, GTPBP2, GSS, HADHA, HGSNAT, HK1, HMX1, HPS3, HPS4, HSD17B10, IDS, IDUA, IFT140, IGFBP7, IKBKG, INTS11, JAG1, KIF11, KIF3B, KLHL7, LAMA1, LAMB2, LAMP2, LIG3, LOXL3, LRP2, LRRC32, LRRC8C, LYST, MAB21L2, MAN2B1, MCOLN1, MED12, MFSD8, MITF, MMACHC, MORC2, 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, PEX12, PGK1, PHYH, PISD, PMM2, PNPLA6, POC5, POMGNT1, POMT1, PPP2R3C, PPT1, PRPS1, PYGM, RBP4, RCBTB1, RDH11, RIMS2, RPGR, RXYLT1, SACS, SGSH, SLC19A2, SLC38A3, SLC6A6, SMPD1, SPG11, SPTLC1, SSBP1, STN1, STX3, SUMF1, TBC1D32, TGFBR2, TIMP3, TINF2, TPP1, TREX1, TRNT1, TTC21B, TTLL5, TTPA, TUB, TUBGCP4, TUBGCP6, VPS13B, YARS1, ZFYVE26, ZNF423

WDR19

SLS / NPHP

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

OCA (n=17)

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

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