Supplementary Information

Whole Exome Sequencing Reveals Mutations in Known Retinal Disease Genes in 33 out of 68 Israeli and Palestinian Families with Inherited Retinopathies

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Supplementary Table S1: A list of 226 known IRD genes that were analyzed in this study by WES.

ABCA4, ABCC6, ABHD12, ACBD5, ADAM9, ADAMTS18, AHI1, AIPL1, ALMS1, ARL2BP, ARL6, ARMS2, ATXN7, BBIP1, BBS1, BBS10, BBS12, BBS17, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, C2, C21orf2, C2orf71, C3, C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDH3, CDHR1, CEP164, CEP250, CEP290, CERKL, CFB, CFH, CHM, CIB2, CLN3, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL2A1, COL9A1, CRB1, CRX, CSPP1, CYP4V2, DFNB31, DHDDS, DHX38, DMD, DTHD1, EFEMP1, ELOVL4, EMC1, ERCC6, EYS, FAM161A, FBLN5, FLVCR1, FSCN2, FZD4, GDF6, GNAT1, GNAT2, GNPTG, GPR125, GPR179, GPR98, GRK1, GRM6, GUCA1A, GUCA1B, GUCY2D, HARS, HK1, HMCN1, HMX1, HTRA1, IDH3B, IFT140, IFT172, IFT27, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, ITM2B, JAG1, KCNJ13, KCNV2, KIAA1549, KIF11, KIZ, KLHL7, LAMA1, LCA5, LRAT, LRIT3, LRP5, LZTFL1, MAK, MERTK, MFRP, MKKS, MKS1, MTTP, MVK, MYO7A, NDP, NEK2, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OAT, OFD1, OPN1LW, OPN1MW, OPN1SW, OTX2, PANK2, PAX2, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PDZD7, PEX1, PEX2, PEX7, PGK1, PHYH, PITPNM3, PLA2G5, PLK4, POC1B, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PXMP3, RAB28, RAX2, RBP3, RBP4, RD3, RDH11, RDH12, RDH5, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, SAG, SDCCAG8, SEMA4A, SLC24A1, SLC7A14, SNRNP200, SPATA7, TEAD1, TIMP3, TLR3, TLR4, TMEM237, TOPORS, TREX1, TRIM32, TRPM1, TSPAN12, TTC8, TTLL5, TTPA, TUB, TULP1, UNC119, USH1C, USH1G, USH2A, VCAN, WDPCP, WDR19, WFS1, ZNF408, ZNF423, ZNF513.

Supplementary Table S2: Primers for all the suspected variants used in this study.

GENE NAME	FORWARD PRIMER	REVERSE PRIMER
ADAM9	GCAAAACATTTTCTTTGCGTTC	CATTTCCTGTTTTCCCAATG
AIPL1	ATGGGGTGAACTGAGTGAGC	AGTTTGCAGGACTGGCTTTG
BBS1	GGGTGTAGACATTGGGTTTCC	CCCTGGAGCCTCCCAAAC
CDHR1	CCTGATACAGACCAAGGACAACC	GGAGCAGAGAGCTTTCTGGGCTG
CNGA1	GCCTTTCCACTAATAAGACTTCTTTC	ATGCATGGGAAAATGTTTGG
CNGA1	AAGCTGGTCTGTTGGTGGAG	TTCCATGAGGTCATCTTTTGAG
CNGB1	TCCATGGAAGGGAGAGAC	CCATCCCTTGAGGAACTC
CNGB1	TGACCTCCCATAACCCTTCC	GAGTTGACTGGGAGCTGGTG
CNNM4	TAATGTTTTGGTTGAGGGAG	AGCTAGGGACACCAAGTTTC
CYP4V2	CTTGATCCACATGTTCTTCTTTG	AGAAACCCACCATCAAGTGG
C2ORF71	AAACAGGACCTCAGAGTCCAGCC	TTCTGGCCGAGCTGGGATTCATAA
FAM161A	TGAGCATGGTGGCACAAG	GCTGACCTACAAGGCAGAGG
MAK	AACGCTCCTTGAGAATGG	ACACCTGGCCTGTTAAGC
NRL	GACCTGGCGCTGACCCGGTTTCTGCATTCT	GCCACCCCCACCAGCCCCCACTACACCACA
NR2E3	GAGGGGAGCGTGCAGCCCTG	CACCCCTCCAGAACCCCTCAG
PDE6A	GGTGACTTTACTGCCAGCATC	CATACATATAATCATTGTTGCT
PRPF3	AAGTGACTTCAAAGACTGATTGTTG	CTCTTGATCCACACTAGGGTCA
PLA2G5	ATTAGGGATGGGGTCAGGAG	CAGGCTGGCTGTTCTCTAGG
RDH5	CAGATGCTCCCAGGAAGAAG	GAGTGGGCTGCTGTTAGTCC
RDH12	AATGCTCTGTCCCCCAGTC	CTCCCATACCCAATTTCCTG
RDH12	GCTAGGGGACTCCTTGCTAAC	AGGCCCCTGGACATTCTC
RPE65	TGCAAATGATGGAGAAAATG	TTTAGATGTGATTCAGATTGAGTGC
RP1	TTCAAGCCTAGGAGGTTGTTG	ATTGAAGCATGGATTTTGCC
SPATA7	AAAAACCAATTTTCTATTTTGTTCC	GCCAAATCTGACCCAATGTC

TSPAN12	TGACAGATATAGCTCTGGGTACAAA	GGAAAATTTCATTGGCATATTG
TULP1	AATCACAGAGCTCCCCAGAG	ACTGTGGTGGGTGCTCTACC
USH2A	AAGTATTGCTGGCAAGTGGC	GATCTTCACTACTACTGGTTTTGGG
USH2A	CACATCAAGAGTGCTTGCTTTC	CACCGCCACTTACTTCTTCC
CDH3	TGCTGTAGTCTCTTTGGCCC	GCTGGTTGGTGGTGGAGTC
CDHR1	TACCCCCATTGTGGTCTCT	CAAGCCTACAGCTCTGGTTC
RPE65	GGGCTGGAAATGAAAATCAC	GGCCCTACTTTGAGGAGGAG
RPE65	CCCTTTATTCTTCATGTTGTGC	AGAGGCAATCAGTGCAGTCC

Supplementary Table S3: A list of published Israeli families with nonsyndromic IRDs

Gene	Retinal phenotype	Number of Families	Inheritance pattern	Reference
ADAM9	CRD	2	AR	[1]
AIPL1	LCA	5	AR	[2]
ARL2BP	RP	1	AR	[3]
BBS1	RP	4	AR	[4]
CDHR1	CRD	1	AR	[5]
		1		[6]
CEP290	LCA	1	AR	[7]
CERKL	RP / CRD	7	AR	[8]
CRB1	Early RP	1	AR	[9]
		1		[10]
		15		[11]
C2ORF71	RP	2	AR	[12]
		2		[13]
C8ORF37	RP	2	AR	[14]
		1		[6]
DHDDS	RP	15	AR	[15]
EYS	RP	10	AR	[16]
		1		[7]
FAM161A	RP	20	AR	[16]
GUCY2D	LCA	4	AR	[2]
	CRD	6	AD	[6]
IMPG2	RP	2	AR	[17]
LCA5	LCA	1	AR	[7]
MAK	RP	1	AR	[18]
MFRP	RP	1	AR	[7]

NR2E3	ESCS	10	AR	[19]
	GFS, RP	2		[7]
PDE6G	Early RP	1	AR	[20]
PDE6B	RP	1	AR	[21]
PRCD	RP	9	AR	[22]
PROM1	CRD	1	AR	[7]
		1		[23]
RAB28	CRD	1	AR	[24]
RDH5	AA	14	AR	[25]
RDH12	Early RP	7	AR	[7]
		1		[10]
RPE65	LCA/RP	10	AR	[2]
RPGR	RP	1	X-lined	[26]
TULP1	Early RP	1	AR	[7]
		2		[27]
USH1C	RP	9	AR	[28]
USH2A	RP	1	AR	[29]

The list includes phenotypes that were studied in the current study (mainly nonsyndromic RP, LCA, and CRD). Syndromic forms (mainly Usher syndrome and Bardet-Biedl syndrome) and maculopathies (mainly Stargardt disease and Best disease) were excluded from this table. Abbreviations: RP- Retinitis pigmentosa, CRD- Cone-rod degeneration, LCA- Leber congenital amaurosis, ESCS- Enhanced S-cone syndrome, GFS- Goldmann-Favre syndrome, AA- Albipunctata albescens.

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Supplementary Figure S1: Pedigrees with identified disease-causing mutations