Supp. Table 1. Specified transcripts for 105 gene symbols analysed by targeted NGS diagnostic testing.

HGNC symbol	RefSeq Transcript ID	HGNC symbol	RefSeq Transcript ID	HGNC symbol	RefSeq Transcript ID
ABCA4	NM_000350.2	*PRPF31*	NM_015629.3	*IMPG2*	NM_016247.3
ADAM9	NM_003816.2	*PRPF6*	NM_012469.3	*KCNV2*	NM_133497.3
AIPL1	NM_014336.3	*PRPF8*	NM_006445.3	*KLHL7*	NM_001031710.2
ARL6	NM_032146.3	*PRPH2*	NM_000322.4	*IMPG2*	NM_016247.3
BBS1	NM_024649.4	*RAX2*	NM_032753.3	*KCNV2*	NM_133497.3
BBS10	NM_024685.3	*RBP3*	NM_002900.2	*KLHL7*	NM_001031710.2
BBS12	NM_001178007.1	*RD3*	NM_183059.2	*LCA5*	NM_181714.3
BBS2	NM_031885.3	*RDH12*	NM_152443.2	*LRAT*	NM_004744.3
BBS4	NM_033028.3	*RDH5*	NM_001199771.1	*LRP5*	NM_002335.2
BBS5	NM_152384.2	*RGR*	NM_002921.3	*MERTK*	NM_006343.2
BBS7	NM_176824.2	*RGS9*	NM_001165933.1	*MKKS*	NM_018848.2
BBS7	NM_018190.3	*RGS9*	NM_003835.3	*MKS1*	NM_001165927.1
BBS9	NM_198428.2	*RHO*	NM_000539.3	*MKS1*	NM_017777.3
BEST1	NM_004183.3	*RIMS1*	NM_014989.4	*MYO7A*	NM_000260.3
C1QTNF5	NM_015645.3	*RIMS1*	NM_001168407.1	*NDP*	NM_000266.3
C2orf71	NM_001029883.1	*CRB1*	NM_201253.2	*RIMS1*	NM_001168410.1
CA4	NM_000717.3	*CRX*	NM_000554.4	*RLBP1*	NM_000326.4
CACNA2D4	NM_172364.4	*DFNB31*	NM_015404.3	*ROM1*	NM_000327.3
CDH23	NM_022124.5	*DHDDS*	NM_024887.2	*RP1*	NM_006269.1
CDHR1	NM_001171971.1	*EFEMP1*	NM_001039348.2	*RP1L1***	NM_178857.5
CDHR1	NM_033100.2	*ELOVL4*	NM_022726.3	*RP2*	NM_006915.2
*CEP290**	NM_025114.3	*EYS*	NM_001142800.1	*RP9*	NM_203288.1
CERKL	NM_001030311.2	*FAM161A*	NM_001201543.1	*RPE65*	NM_000329.2
CHM	NM_000390.2	*FSCN2*	NM_001077182.2	*RPGR****	NM_001034853.1
CLRN1	NM_052995.2	*FZD4*	NM_012193.3	*RPGRIP1*	NM_020366.3
CLRN1	NM_001195794.1	*GNAT2*	NM_005272.3	*RS1*	NM_000330.3
CNGA1	NM_001142564.1	*GPR98*	NM_032119.3	*SAG*	NM_000541.4
CNGA3	NM_001298.2	*GUCA1A*	NM_000409.3	*SEMA4A*	NM_022367.3
CNGB1	NM_001297.4	*GUCA1B*	NM_002098.5	*SNRNP200*	NM_014014.4
CNGB3	NM_019098.4	*GUCY2D*	NM_000180.3	*SPATA7*	NM_018418.4
NR2E3	NM_014249.2	*IDH3B*	NM_006899.2	*TEAD1*	NM_021961.5
NRL	NM_006177.3	*IDH3B*	NM_174855.1	*TIMP3*	NM_000362.4
OTX2	NM_021728.2	*IMPDH1*	NM_000883.3	*TOPORS*	NM_005802.4
PCDH15	NM_001142763.1	*IMPG2*	NM_016247.3	*TRIM32*	NM_012210.3
PCDH15	NM_001142769.1	*KCNV2*	NM_133497.3	*TTC8*	NM_144596.2
PCDH15	NM_001142771.1	*KLHL7*	NM_001031710.2	*TULP1*	NM_003322.3
PCDH15	NM_001142770.1	*LCA5*	NM_181714.3	*UNC119*	NM_005148.3
PDE6A	NM_000440.2	*LRAT*	NM_004744.3	*UNC119*	NM_054035.2
PDE6B	NM_000283.3	*LRP5*	NM_002335.2	*USH1C*	NM_005709.3
PDE6C	NM_006204.3	*MERTK*	NM_006343.2	*USH1C*	NM_153676.3
PDE6G	NM_002602.3	*MKKS*	NM_018848.2	*USH1G*	NM_173477.2
PITPNM3	NM_031220.3	*MKS1*	NM_001165927.1	*USH2A*	NM_206933.2
PRCD	NM_001077620.2	*MKS1*	NM_017777.3	*ZNF513*	NM_144631.5
PROM1	NM_006017.2	*MYO7A*	NM_000260.3		
PRPF3	NM_004698.2	*NDP*	NM_000266.3		

* Testing of the common intron 26 mutation c.2991+1655A>G in CEP290 is included in this analysis
** Analysis of the coding region of exon 4 of the RP1L1 gene is not included
*** Analysis of the coding region of the final exon (*orf15*) of *RPGR* it is not included *HGNC*, gene symbols approved by the HUGO Gene Nomenclature Committee[1]

1. Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Genenames.org: the HGNC resources in 2015. Nucleic Acids Res 2015;43:D1079-85.

Supp Table 2. Mutation classification criteria.

1 Clearly not pathogenic

- Variant has a frequency >1% in either EVS or dbSNP
- Variant is not present in HGMD
- Variant has no evidence for its pathogenesis

2 Unlikely to be pathogenic

- Variant has a frequency <1% in EVS and <1% in dbSNP,
- Variant is not present in HGMD
- Variant does not have evidence for its pathogenicity (*in-silico*, functional or segregation)

3 Variant of unknown significance

- Variant has a frequency <1% in EVS and <1% in dbSNP,
- Variant is not present in HGMD
- Variant does not have clear evidence for its pathogenicity (functional or segregation)
- Variant has strong and multiple lines of *in-silico* evidence to disrupt protein structure and/or normal translation

(a) Variant is not clinically reported

- Variant is not found in a *disease-causing* state

(b) Variant is clinically reported

- Variant is found in a *disease-causing* state (e.g. homozygous or with another 'clearly pathogenic' heterozygous variant),
- Variant may be found in a gene highly specific to the patient's clinical presentation of IRD,

4 Likely pathogenic

- Variant has a frequency <1% in EVS and <1% in dbSNP,
- Variant may/may not be in HGMD,
- Variant may have been shown to segregate with disease previously in a single family,
- Variant does not have functional evidence for its pathogenicity,
- Variant has strong and multiple lines of *in-silico* evidence to disrupt protein structure and/or normal translation,
- Variant may be in a hotspot for known pathogenic mutations within a gene,
- Variant may be in a gene highly specific to the patient's clinical presentation of IRD,

5 Clearly pathogenic

- Variant has a frequency <1% in EVS and <1% in dbSNP,
- Variant is present in HGMD,
- Variant may have been shown to segregate with disease previously in multiple families,
- Variant may have functional evidence for its pathogenicity,
- Variant has strong and multiple lines of *in-silico* evidence to disrupt protein structure and/or normal translation

Supp Table 3. Clinical indications of individuals referred with inherited retinal disease for diagnostic genomic testing.

Clinical diagnosis	Number of referred cases
***Retinitis pigmentosa* or *rod-cone dystrophy* (RP/RCD)**	250
***Leber congenital amaurosis* or *early onset rod-cone dystrophy* (EORD/LCA)**	78
Other(indication not included in this list, or not defined)	42
***Stargardt disease* or *macular dystrophy* (STGD/MD)**	46
Usher syndrome	38
***Cone-rod dystrophy* (CRD)**	39
***Achromotopsia* or *cone dystrophy* (CD)**	27
Suspected syndromic ciliopathy	8
***Familial Exudative Vitreoretinopathy* (FEVR)**	5
***Choroideremia* (CHM)**	4

Supp. Table S4. Recurrent single nucleotide variants identified by NGS which were determined as false positives after PCR amplification and bidirectional capillary sequencing.

Variant	false positive validations
BBS7 c.166-5T>C	10
BBS7 c.171G>T	9
BBS7 c.181T>A	8
USH2A c.10246T>G	4
ABCA4 c.1356+4A>G	3
RBP3 c.1652A>C	6
RP1L1 c.3981A>G	3
CEP290 c.4006A>T	2
ABCA4 c.4530G>C	6

Supp. Table S5. Genetic variants reported to referring clinicians, including causative, likely causative, and variants of unknown clinical significance.

MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
Patients referred with CD					
12004242	chr8:87656009-87656009	CNGB3 c.1148delC	hom	[1, 2]	-
13017395	chr2:182468670-182468670	CERKL c.375C>G p.(Cys125Trp)	hom	[3, 4]	[5]
13015689	chr2:182413512-182413513	CERKL c.1045_1046delAT	hom	novel	-
13003908	chr2:99012481-99012481	CNGA3 c.848G>A p.(Arg283Gln)	hom	-	[6]
13010246	chr17:7918018-7918018	GUCY2D c.2512C>T p.(Arg838Cys)	het	[7, 8]	[9]
13011432	chr17:7918019-7918019	GUCY2D c.2513G>A p.(Arg838His)	het	[8]	[9]
13010483	chr1:94473287-94473287	ABCA4 c.5908C>T p.(Leu1970Phe)	het	[10, 11]	[12]
	chr1:94471065-94471065	ABCA4 c.6079C>T p.(Leu2027Phe)	het	[12]	[13]
13002658	chr8:87656009-87656009	CNGB3 c.1148delC	hom	[1, 2]	-
13009042	chr2:99013274-99013274	CNGA3 c.1641C>A p.(Phe547Leu)	hom	[14, 15]	[16]
13012218	chr2:99013190-99013190	CNGA3 c.1557G>A p.(Met519Ile)	het	novel	-
	chr2:99012912-99012912	CNGA3 c.1279C>T p.(Arg427Cys)	het	[17-20]	[18]
13000861	chr8:87656009-87656009	CNGB3 c.1148delC	hom	[1, 2]	-
13017391	chr17:7918821-7918821	GUCY2D c.2944+1delG	hom	[21-23]	-
13007934[+]	chr1:110146104-110146104	GNAT2 c.937C>T p.(Arg313Ter)	hom	[24]	-
13002676	chr8:87656009-87656009	CNGB3 c.1148delC	hom	[1, 2]	-
13015850	chr8:87656009-87656009	CNGB3 c.1148delC	hom	[1, 2]	-
12006121	chr8:87656009-87656009	CNGB3 c.1148delC	hom	[1, 2]	-
12001563	chr8:87616320-87616320	CNGB3 c.1781+1G>C	het	[2]	

Supp. Table S5. Genetic variants reported to referring clinicians, including causative, likely causative, and variants of unknown clinical significance.

MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
	chr8:87656009-87656009	CNGB3 c.1148delC	het	[1, 2]	
13001685	chr2:99012912-99012912	CNGA3 c.1279C>T p.(Arg427Cys)	het	[17-20]	[18]
	chr2:99008427-99008427	CNGA3 c.667C>T p.(Arg223Trp)	het	[17, 25]	
12003493	chr10:95405761-95405761	PDE6C c.1892A>G p.(Glu631Gly)	het	novel	-
	chr10:95405763-95405763	PDE6C c.1894A>T p.(Arg632Trp)	het	novel	-
Patients referred with BBS					
12014789	chr4:122749847-122749848	BBS7 c.1712_1713delinsAGA p.(Ser571Ter)	hom	novel	-
12003699[+]	chr11:66293652-66293652	BBS1 c.1169T>G p.(Met390Arg)	hom	[26-29]	[30]
13013491[+]	chr11:66293652-66293652	BBS1 c.1169T>G p.(Met390Arg)	hom	[26-29]	[30]
13000497[+]	chr11:66293652-66293652	BBS1 c.1169T>G p.(Met390Arg)	hom	[26-29]	[30]
10001098[+]	chr4:123664110-123664110	BBS12 c.1063C>T p.(Arg355Ter)	hom	[31]	-
12009772	chr15:73023645-73023645	BBS4 c.712-1G>A	hom	novel	-
13000378	chr12:76741493-76741494	BBS10 c.271dupT	hom	[32]	-
Patients referred with CRD					
12014625[+]	chr1:197403836-197403836	CRB1 c.2843G>A p.(Cys948Tyr)	het	[33-37]	mutation hotspot [34]
	chr1:197396689-197396689	CRB1 c.2234C>T p.(Thr745Met)	het	[33, 35]	mutation hotspot [34]
13011267	chr2:182468670-182468670	CERKL c.375C>G p.(Cys125Trp)	het	[3, 4]	[5]
	chr2:182521541-182521541	CERKL c.193G>T p.(Glu65Ter)	het	novel	-
13005842[+]	chr1:197325978-197325978	CRB1 c.1006T>C p.(Cys336Arg)	het	novel	-

Supp. Table S5. Genetic variants reported to referring clinicians, including causative, likely causative, and variants of unknown clinical significance.

MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
	chr1:197396584-197396584	CRB1 c.2129A>T p.(Glu710Val)	het	[33, 38, 39]	-
12011000	chr19:54621779-54621779	PRPF31 c.121C>G p.(Leu41Val)	het	novel	-
13007236	chr1:94485164-94485164	ABCA4 c.5170T>C p.(Trp1724Arg)	hom	p.Trp1724Cys [40, 41]	-
13010981[+]	chr19:48339520-48339520	CRX c.121C>T p.(Arg41Trp)	het	[42]	[42, 43]
11011507	chr1:94466422-94466422	ABCA4 c.6449G>A p.(Cys2150Tyr)	het	[44, 45]	-
	chr1:94568687-94568687	ABCA4 c.454C>T p.(Arg152Ter)	het	[46]	-
88301	chr4:15985950-15985950	PROM1 c.2309delC	het	novel	-
	chr4:16008260-16008261	PROM1 c.1354dupT	het	[47]	-
82863[+]	chr1:94508984-94508984	ABCA4 c.3098delA	het	novel	-
	chr1:94496666-94496666	ABCA4 c.4139C>T p.(Pro1380Leu)	het	[12, 45, 48, 49]	[50]
990697[+]	chr1:94473278-94473278	ABCA4 c.5917delG	het	[51]	-
	chr1:94463488-94463488	ABCA4 c.6658C>T p.(Gln2220Ter)	het	[52-54]	-
12014678[+]	chr12:88476853-88476854	CEP290 c.4966_4967delGA	het	[55]	-
	chr12:88465636-88465636	CEP290 c.5777G>C p.(Arg1926Pro)	het	[56]	-
11011667[+]	chr2:99013321-99013321	CNGA3 c.1688G>A p.(Arg563His)	het	[17]	[57]
	chr2:98986505-98986505	CNGA3 c.67C>T p.(Arg23Ter)	het	[25]	
13012292	chrX:38158220-38158220	RPGR c.1234C>T p.(Arg412Ter)	het	[58]	-
13016156[+]	chr2:182468670-182468670	CERKL c.375C>G p.(Cys125Trp)	het	[3, 4]	[5]
	chr2:182423344-182423344	CERKL c.847C>T p.(Arg283Ter)	het	[3]	
12013927	chr4:16014893-16014893	PROM1 c.1141+5G>C	hom	novel	-
12009616[+]	chr4:16008260-16008261	PROM1 c.1354dupT	het	[47]	-
	chr4:16000112-16000112	PROM1 c.1579-1G>C	het	novel	-
12010986	chr14:21794230-21794231	RPGRIP1 c.2608_2609insA	hom	novel	-

Supp. Table S5. Genetic variants reported to referring clinicians, including causative, likely causative, and variants of unknown clinical significance.

MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
13008348	chr2:182468670-182468670	CERKL c.375C>G p.(Cys125Trp)	hom	[3, 4]	[5]
11010719	chr2:182423344-182423344	CERKL c.847C>T p.(Arg283Ter)	hom	[3]	
Patients referred with FEVR					
13010229^{+}	chr11:68115344-68115344	LRP5 c.121C>T p.(Arg41Trp)	het	novel	-
	chr11:68153802-68153802	LRP5 c.1034T>A p.(Leu345Gln)	het	novel	-
12006042^{+}	chr11:86662513-86662516	FZD4 c.1282_1285delGACA	het	[59]	-
	chr11:86662837-86662837	FZD4 c.961G>A p.(Val321Ile)	het	novel	-
12000347	chr11:86663485-86663485	FZD4 c.313A>G p.(Met105Val)	het	[60, 61]	-
Patients referred with JOUBERT					
13009772^{+}	chr12:88471040-88471040	CEP290 c.5668G>T p.(Gly1890Ter)	hom	[62]	-
Patients referred with EORD/LCA					
13015227	chr1:68905008-68905008	RPE65 c.726-2A>C	hom	novel	-
83830^{+}	chr17:7919559-7919559	GUCY2D c.3175A>T p.(Arg1059Ter)	het	novel	-
	chr17:7909706-7909706	GUCY2D c.1052A>G p.(Tyr351Cys)	het	[63, 64]	-
12012655^{+}	chr14:68192807-68192807	RDH12 c.383T>G p.(Val128Gly)	het	novel	-
	chr14:68200524-68200524	RDH12 c.910T>C p.(Trp304Arg)	het	novel	-
13009681^{+}	chr1:197404030-197404030	CRB1 c.3037C>T p.(Gln1013Ter)	het	[33]	
	chr1:197396856-197396856	CRB1 c.2401A>T p.(Lys801Ter)	het	[33, 65]	-
13017541	chr17:7918821-7918821	GUCY2D c.2944+1delG	hom	[21-23]	-

Supp. Table S5. Genetic variants reported to referring clinicians, including causative, likely causative, and variants of unknown clinical significance.

MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
13001467	chr12:88478404-88478406	CEP290 c.4661_4663delAAG p.(Glu1554del)	het	[66, 67]	-
	chr12:88532921-88532921	CEP290 c.297+1G>T	het	novel	-
12009612	chr14:21816333-21816333	RPGRIP1 c.3620T>G p.(Leu120Ter)	hom	[68]	-
11011508	chrX:46713068-46713076	RP2 c.260_268delCTAACTGCA p.(Thr87_Cys89del)	hemi	novel	-
13010893	chr12:88465563-88465563	CEP290 c.5850delT	het	[67, 69]	-
	chr12:88477713-88477713	CEP290 c.4723A>T p.(Lys1575Ter)	het	[67, 69]	-
13015209	chr17:7918821-7918821	GUCY2D c.2944+1delG	hom	[21-23]	-
13003353[+]	chr12:88494960-88494960	CEP290 c.2991+1655A>G	het	[70]	-
	chr12:88510853-88510853	CEP290 c.1781T>A p.(Leu594Ter)	het	novel	-
13009682	chr1:197404007-197404007	CRB1 c.3014A>T p.(Asp1005Val)	het	[35]	-
	chr1:197396856-197396856	CRB1 c.2401A>T p.(Lys801Ter)	het	[33, 65]	-
11013818[+]	chr1:197403836-197403836	CRB1 c.2843G>A p.(Cys948Tyr)	hom	[33-37]	mutation hotspot [34]
12003872	chr6:80223010-80223016	LCA5 c.633_639delAGCTAGA	hom	novel	-
13001571[+]	chr1:197403836-197403836	CRB1 c.2843G>A p.(Cys948Tyr)	het	[33-37]	mutation hotspot [34]
	chr1:197298065-197298065	CRB1 c.584G>T p.(Cys195Phe)	het	[34]	-
13013813	chr14:21793021-21793021	RPGRIP1 c.2007delT	hom	novel	-
12012994	chr17:6337250-6337250	AIPL1 c.265T>C p.(Cys89Arg)	hom	[71, 72]	[71]
12013890	chr1:68906634-68906634	RPE65 c.545A>G p.(His182Arg)	hom	[73, 74]	p.His182Tyr, p.His182Asn [74]
13018022	chr14:21792906-21792906	RPGRIP1 c.1892A>G p.(His631Arg)	hom	p.(His631Pro) [75]	[76]

Supp. Table S5. Genetic variants reported to referring clinicians, including causative, likely causative, and variants of unknown clinical significance.

MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
13005787	chr17:7918737-7918737	GUCY2D c.2861T>C p.(Leu954Pro)	hom	[77, 78]	[22]
12015787[+]	chr14:21780621-21780621	RPGRIP1 c.1107delA	hom	[79]	-
12001399	chr1:197404067-197404067	CRB1 c.3074G>A p.(Ser1025Asn)	hom	[33] p.(Ser1025Ile) [63]	-
13002979[+]	chr4:16008260-16008261	PROM1 c.1354dupT	het	[47]	-
	chr4:16025914-16025914	PROM1 c.694+4A>T	het	novel	-
13003494	chr1:68910508-68910508	RPE65 c.304G>T p.(Glu102Ter)	hom	[80-82]	-
12013231[+]	chr19:48342890-48342890	CRX c.566delC	het	other final exon frameshift mutations [83, 84]	-
12012257	chr14:88904207-88904218	SPATA7 c.1241_1252delTCCTGAAAGTAG p.(Val414_Val417del)	het	novel	-
	chr14:88897545-88897545	SPATA7 c.1058dupC	het	novel	-
13005796	chr11:76885842-76885842	MYO7A c.1976C>A p.(Ser659Ter)	het	novel	-
	chr11:76892594-76892594	MYO7A c.2863G>A p.(Gly955Ser)	het	[85, 86]	-
12004275	chr4:155665951-155665951	LRAT c.473G>A p.(Trp158Ter)	hom	novel	-
12013710	chr12:88494960-88494960	CEP290 c.2991+1655A>G	het	[70]	-
	chr12:88474003-88474003	CEP290 c.5182G>T p.(Glu1728Ter)	het	[62, 87]	-
11010486	chr1:197403836-197403836	CRB1 c.2843G>A p.(Cys948Tyr)	het	[33-37]	mutation hotspot [34]
	chr1:197398590-197398590	CRB1 c.2688T>A p.(Cys896Ter)	het	[33, 63, 88]	-
13007873	chr1:197403862-197403862	CRB1 c.2869C>T p.(Gln957Ter)	het	novel	-
	chr1:197403836-197403836	CRB1 c.2843G>A p.(Cys948Tyr)	het	[33-37]	mutation hotspot [34]
12006834[+]	chr12:88494960-88494960	CEP290 c.2991+1655A>G	het	[70]	-

Supp. Table S5. Genetic variants reported to referring clinicians, including causative, likely causative, and variants of unknown clinical significance.

MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
	chr12:88477635-88477635	CEP290 c.4801C>T p.(Gln1601Ter)	het	novel	-
12009433[+]	chr12:88494960-88494960	CEP290 c.2991+1655A>G	het	[70]	-
	chr12:88508195-88508196	CEP290 c.2052+1_2052+2delGT	het	novel	-
13005926	chr12:88519134-88519134	CEP290 c.1078C>T p.(Arg360Ter)	het	[66]	-
	chr12:88477713-88477713	CEP290 c.4723A>T p.(Lys1575Ter)	het	[67, 69]	-
12008985	chr1:68912507-68912507	RPE65 c.131G>A p.(Arg44Gln)	hom	[89]	[90, 91]
13002684	chr17:7918821-7918821	GUCY2D c.2944+1delG	hom	[21-23]	-
13005789	chr14:68193831-68193831	RDH12 c.582C>G p.(Tyr194Ter)	het	[92, 93]	-
	chr14:68196099-68196099	RDH12 c.848+2T>C	het	novel	-
12014872	chr1:197397003-197397003	CRB1 c.2548G>A p.(Gly850Ser)	het	[33, 34]	mutation hotspot [34]
	chr1:197403836-197403836	CRB1 c.2843G>A p.(Cys948Tyr)	het	[33-37]	mutation hotspot [34]
13010086	chr14:21780621-21780621	RPGRIP1 c.1107delA	hom	[79]	-
12004181	chr12:88487674-88487675	CEP290 c.3181_3182delAT	het	novel	-
	chr12:88524986-88524986	CEP290 c.451C>T p.(Arg151Ter)	het	[94]	-
13015422	chr17:6329101-6329101	AIPL1 c.834G>A p.(Trp278Ter)	hom	[95]	[96]
13005791	chr14:68193773-68193773	RDH12 c.524C>T p.(Ser175Leu)	het	[69]	p.(Ser175Pro) [97]
	chr14:68193897-68193927	RDH12 c.648_658+20delGAGGCTCCAAGGTAAGTCTGGAGAAAGAGGA	het	novel	-
13002515	chr14:21786006-21786006	RPGRIP1 c.1303A>T p.(Lys435Ter)	hom	novel	-
12007217[+]	chr12:88494960-88494960	CEP290 c.2991+1655A>G	het	[70]	-
	chr12:88476854-88476854	CEP290 c.4966G>T p.(Glu1656Ter)	het	[70]	-
13012618	chr1:197396856-197396856	CRB1 c.2401A>T p.(Lys801Ter)	het	[33, 65]	-
	chr1:197390570-197390571	CRB1 c.1612_1613insCTTA	het	novel	-

Supp. Table S5. Genetic variants reported to referring clinicians, including causative, likely causative, and variants of unknown clinical significance.

MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
13002689	chr14:21788314-21788314	RPGRIP1 c.1445T>A p.(Leu482Ter)	het	novel	-
	chr14:21793489-21793489	RPGRIP1 c.2314C>T p.(Gln772Ter)	het	novel	-
12007024[+]	chr1:197403835-197403835	CRB1 c.2843-1G>A	het	novel	-
	chr1:197396856-197396856	CRB1 c.2401A>T p.(Lys801Ter)	het	[33, 65]	-
13002649	chr17:7918821-7918821	GUCY2D c.2944+1delG	hom	[21-23]	-
13005797	chr17:7906745-7906745	GUCY2D c.380C>T p.(Pro127Arg)	het	novel	-
	chr17:7918195-7918195	GUCY2D c.2595delG	het	[98]	
Patients referred with OTHER					
12010090	chr2:170336065-170336065	BBS5 c.2T>A p.(Met1?)	hom	[99]	-
13010232[+]	chr12:56115698-56115698	RDH5 c.536A>G p.(Lys179Arg)	hom	novel	mutation hotspot [100]
Patients referred with RP/RCD					
13003141	chr2:96958829-96958829	SNRNP200 c.2041C>T p.(Arg681Cys)	het	[101]	-
12013732[+]	chr8:55538040-55538043	RP1 c.1598_1601delGAAA	het	novel	-
12003878	chr17:1564976-1564976	PRPF8 c.4131C>T p.(Ser1377Ser)	het	novel	-
13001452	chr2:112761560-112761560	MERTK c.1866G>C p.(Lys622Asn)	hom	novel	-
12011770	chr4:16000058-16000058	PROM1 c.1632G>T p.(Gly544Gly)	het	novel	-
	chr4:15995680-15995680	PROM1 c.1697delA	het	novel	-
13015513	chr8:55534803-55534803	RP1 c.742C>T p.(Gln248Ter)	het	novel	location p.500-p.1053 [102]

Supp. Table S5. Genetic variants reported to referring clinicians, including causative, likely causative, and variants of unknown clinical significance.

MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
	chr8:55534065-55534065	RP1 c.539T>G p.(Phe180Cys)	het	novel	-
12015109	chr2:112779023-112779023	MERTK c.2214delT	hom	[103]	-
13002962[+]	chr2:182468729-182468729	CERKL c.316C>T p.(Arg106Cys)	het	p.(Arg106Ser) [104]	-
	chr2:182468670-182468670	CERKL c.375C>G p.(Cys125Trp)	het	[3, 4]	[5]
13015666[+]	chr1:215847812-215847812	USH2A c.13441A>G p.(Arg4481Gly)	het	novel	-
	chr1:216062399-216062399	USH2A c.7595-3C>G	het	[105-107]	[107]
11000824[+]	chr1:197396763-197396763	CRB1 c.2308G>A p.(Gly770Ser)	het	novel	-
	chr1:197396856-197396856	CRB1 c.2401A>T p.(Lys801Ter)	het	[33, 65]	-
11005352	chr19:54631496-54631496	PRPF31 c.994C>T p.(Gln332Ter)	het	novel	-
13003075	chr3:150645894-150645894	CLRN1 c.567T>G p.(Tyr189Ter)	het	[108, 109]	-
	chr3:150659443-150659443	CLRN1 c.359T>A p.(Met120Lys)	het	[109]	[110]
12014873	chr3:100962762-100962763	IMPG2 c.2412_2413delTG	het	novel	-
	chr3:100947720-100947720	IMPG2 c.3634G>T p.(Glu1212Ter)	het	novel	-
13012950[+]	chr15:72103895-72103895	NR2E3 c.191G>T p.(Cys64Phe)	het	novel	-
	chr15:72103821-72103821	NR2E3 c.119-2A>C	het	[111-113]	[114]
12000462	chr1:216424273-216424273	USH2A c.2139C>T p.(Gly713Gly)	het	novel	-
	chr1:216462734-216462734	USH2A c.1859G>T p.(Cys620Phe)	het	[115]	-
13010985[+]	chrX:46737028-46737028	RP2 c.969+3A>G	hemi	[116, 117]	[117]
12002962	chr10:55582113-55582122	PCDH15 c.5385_5394delTCCTCTTCCT	het	novel	-
	chr10:55581921-55581921	PCDH15 c.5586C>A p.(Ala1862Ala)	het	novel	-
13004711	chr8:55538471-55538471	RP1 c.2029C>T p.(Arg677Ter)	het	[118, 119]	-
13012171	chrX:85155656-85155656	CHM c.1408C>T p.(Gln470Ter)	hemi	[120]	-
12012050	chr16:57998062-57998062	CNGB1 c.262C>T p.(Gln88Ter)	het	novel	-
	chr16:57938727-57938728	CNGB1 c.2544dupG	het	novel	-

Supp. Table S5. Genetic variants reported to referring clinicians, including causative, likely causative, and variants of unknown clinical significance.

MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
13010307	chr2:62066572-62066572	FAM161A c.1567C>T p.(Arg523Ter)	het	[121]	-
	chr2:62066830-62066830	FAM161A c.1309A>T p.(Arg437Ter)	het	[122]	-
12010984[+]	chr2:96953706-96953706	SNRNP200 c.3260C>T p.(Ser1087Leu)	het	[101, 123]	[123, 124]
13006766	chr19:54626942-54626942	PRPF31 c.527+3A>G	het	[125, 126]	-
13017314	chrX:38158210-38158211	RPGR c.1243_1244delAG	hemi	[127]	-
13003723	chr1:216221921-216221921	USH2A c.6118T>G p.(Cys2040Gly)	het	novel	-
	chr1:216373373-216373373	USH2A c.3407G>A p.(Ser1136Asn)	het	[115]	-
10003544	chr16:57935275-57935275	CNGB1 c.2957A>T p.(Asn986Ile)	hom	[39]	-
13007749	chr1:216420460-216420460	USH2A c.2276G>T p.(Cys759Phe)	het	[128-132]	-
	chr1:216420437-216420437	USH2A c.2299delG	het	[27, 115, 133-136]	-
12007876	chr3:129251131-129251131	RHO c.568G>A p.(Asp190Asn)	het	[137]	[138]
13013774	chr1:197396745-197396745	CRB1 c.2290C>T p.(Arg764Cys)	het	[33, 35, 36]	-
	chr1:197298065-197298065	CRB1 c.584G>T p.(Cys195Phe)	het	[34]	-
12002958	chr6:64430796-64430796	EYS c.9131G>T p.(Trp3044Leu)	het	[139]	-
	chr6:65016917-65016917	EYS c.6137G>A p.(Trp2046Ter)	het	novel	-
12013881	chr14:68193850-68193850	RDH12 c.601T>C p.(Cys201Arg)	hom	[140]	[140]
13006275	chr9:32541984-32541984	TOPORS c.2539C>T p.(Arg847Ter)	het	[27]	-
12002573	chr6:42141500-42141500	GUCA1A c.149C>T p.(Pro50Leu)	het	[141]	[142]
12016026[+]	chr1:197446947-197446947	CRB1 c.4159G>T p.(Glu1387Ter)	het	novel	-
	chr1:197398590-197398590	CRB1 c.2688T>A p.(Cys896Ter)	het	[33, 63, 88]	-
13002431[+]	chr19:54625894-54625894	PRPF31 c.341T>A p.(Ile114Asn)	het	novel	-
12007088	chr4:658734-658734	PDE6B c.2193+1G>A	het	[143]	-
	chr4:619706-619706	PDE6B c.291C>A p.(Tyr97Ter)	het	novel	-

Supp. Table S5. Genetic variants reported to referring clinicians, including causative, likely causative, and variants of unknown clinical significance.

MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
12005728	chr19:54625953-54625953	PRPF31 c.400delG	het	novel	-
13010253	chr14:24550532-24550573	NRL c.586_627dupGCCCAGCTGGACGCGCTGCGGGCCGAGGTGGCCCGCCTGGCC p.(Ala196_Ala209dup)	het	novel	-
13011161	chrX:38164014-38164014	RPGR c.808C>T p.(Gln270Ter)	hemi	novel	-
13012574	chr3:129252554-129252554	RHO c.1040C>T p.(Pro347Leu)	het	[144, 145]	[146]
13004232[+]	chr4:16014897-16014897	PROM1 c.1141+1G>A	het	novel	-
	chr4:16000058-16000058	PROM1 c.1632G>T p.(Gly544Gly)	het	novel	-
13015639	chr1:94528164-94528164	ABCA4 c.1906C>T p.(Gln636Ter)	het	[147, 148]	-
	chr1:94517254-94517254	ABCA4 c.2588G>C p.(Gly863Ala)	het	[11, 149]	-
12016035	chr1:94506945-94506945	ABCA4 c.3342delC	hom	[49]	
12002868[+]	chr8:55539055-55539055	RP1 c.2613dupA	het	[150]	
12014047[+]	chr1:197396764-197396764	CRB1 c.2309G>A p.(Gly770Asp)	het	novel	-
	chr1:197404010-197404010	CRB1 c.3017C>T p.(Ser1006Phe)	het	novel	-
11006153	chr2:62066675-62066675	FAM161A c.1464G>A p.(Trp488Ter)	hom	novel	-
13011266	chrX:46713315-46713315	RP2 c.507delT	hemi	novel	-
13004716	chr6:65612025-65612026	EYS c.2826_2827delAT	hom	novel	-
12010854	chrX:85302485-85302485	CHM c.49+3A>G	hemi	c.49+5G>T [151]	c.49+5G>T [151]
13000107	chr2:112725802-112725804	MERTK c.933_935delinsTT	het	novel	-
	chr2:112777090-112777090	MERTK c.2180G>A p.(Arg727Gln)	het	novel	-
13002976[+]	chr10:85956379-85956379	CDHR1 c.270dupC	hom	novel	-
13004227	chr8:55538474-55538474	RP1 c.2032C>T p.(Gln678Ter)	het	novel	-

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MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
13000106	chr4:47938971-47938971	CNGA1 c.1747C>T p.(Arg583Ter)	hom	novel	-
13008977	chr3:129252554-129252554	RHO c.1040C>T p.(Pro347Leu)	het	[144, 145]	[146]
10003578	chr1:215955512-215955512	USH2A c.10612C>T p.(Arg3538Ter)	het	novel	-
	chr1:216424331-216424331	USH2A c.2081G>C p.(Cys694Ser)	het	p.Cys694Tyr [38]	-
12015280	chr1:216420460-216420460	USH2A c.2276G>T p.(Cys759Phe)	het	[128-132]	-
	chr1:215848679-215848679	USH2A c.12574C>T p.(Arg4192Cys)	het	p.Arg4192His [152, 153]	-
13010104	chrX:46696638-46696638	RP2 c.102+1G>A	hemi	[154]	-
12002571	chr17:1565308-1565312	PRPF8 c.3910_3914delAACTC	het	novel	-
13004912	chr1:216420460-216420460	USH2A c.2276G>T p.(Cys759Phe)	het	[128-132]	-
	chr1:216420437-216420437	USH2A c.2299delG	het	[27, 115, 133-136]	-
13007240[+]	chr6:65655758-65655758	EYS c.2309A>C p.(Gln770Pro)	hom	[155]	-
13010305	chr19:54629963-54629963	PRPF31 c.916G>A p.(Asp306Asn)	het	novel	location [156]
13002044[+]	chr6:66112400-66112400	EYS c.1155T>A p.(Cys385Ter)	hom	novel	-
12013544[+]	chr2:182468670-182468670	CERKL c.375C>G p.(Cys125Trp)	het	[3, 4]	[5]
	chr2:182521541-182521541	CERKL c.193G>T p.(Glu65Ter)	het	novel	-
12002963	chr6:66204814-66204814	EYS c.490C>T p.(Arg164Ter)	hom	novel	-
12011781	chr1:216420460-216420460	USH2A c.2276G>T p.(Cys759Phe)	het	[128-132]	-
	chr1:215914880-215914880	USH2A c.11549-1G>A	het	novel	-
13016075	chrX:46713161-46713161	RP2 c.353G>A p.(Arg118His)	hemi	[157]	[158]
12011157	chr17:1554143-1554143	PRPF8 c.6961C>T p.(Gln2321Ter)	het	novel	-
13018012[+]	chr1:216420460-216420460	USH2A c.2276G>T p.(Cys759Phe)	het	[128-132]	-

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MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
	chr1:216498841-216498841	USH2A c.949C>A p.(Arg317Arg)	het	[159, 160]	[161]
13005635[+]	chrX:38163914-38163914	RPGR c.908G>A p.(Gly303Glu)	hemi	novel	-
12015561[+]	chr3:129251104-129251104	RHO c.541G>A p.(Glu181Lys)	het	[162-164]	[165]
12002960	chr2:62066830-62066830	FAM161A c.1309A>T p.(Arg437Ter)	hom	[122]	-
12002727[+]	chr22:33255338-33255338	TIMP3 c.610A>T p.(Ser204Cys)	het	[166]	-
12012775	chr2:96959219-96959219	SNRNP200 c.1871G>A p.(Arg624Lys)	het	novel	-
12005771[+]	chr1:197313565-197313565	CRB1 c.807dupA	het	novel	-
	chr1:197398590-197398590	CRB1 c.2688T>A p.(Cys896Ter)	het	[33, 63, 88]	-
13012959	chr16:57935339-57935339	CNGB1 c.2893G>A p.(Gly965Ser)	het	novel	-
	chr16:57965773-57965773	CNGB1 c.1382C>T p.(Thr461Met)	het	novel	-
13007688	chr2:96955598-96955598	SNRNP200 c.2879C>T p.(Ala960Val)	het	novel	-
13007207	chr1:68912520-68912520	RPE65 c.118G>A p.(Gly40Ser)	hom	[91, 167]	-
13004358	chr5:89975426-89975429	GPR98 c.5504_5507delTTCC	hom	novel	-
13015421	chr2:98996670-98996670	CNGA3 c.248G>A p.(Trp83Ter)	het	novel	-
	chr2:99013564-99013564	CNGA3 c.1931T>C p.(Phe644Ser)	het	novel	-
075849[+]	chr4:16002140-16002140	PROM1 c.1557C>A p.(Tyr519Ter)	het	[168]	-
	chr4:16008260-16008261	PROM1 c.1354dupT	het	[47]	-
13008349	chr17:1554982-1554982	PRPF8 c.6470T>A p.(Val2157Glu)	het	novel	-
13018110	chr3:150690352-150690352	CLRN1 c.144T>G p.(Asn48Lys)	hom	[169, 170]	[110]
13009346	chr4:47942803-47942803	CNGA1 c.848A>T p.(Asp283Val)	het	novel	-
	chr4:47938964-47938964	CNGA1 c.1754T>G p.(Met585Arg)	het	novel	-
11006503	chr17:7918017-7918018	GUCY2D c.2511_2512delinsCA p.(Glu837_Arg838delinsAspSer)	het	[8, 171, 172]	-

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MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
13004487[+]	chr6:66112400-66112400	EYS c.1155T>A p.(Cys385Ter)	hom	novel	-
12002965[+]	chr12:88479860-88479860	CEP290 c.4393C>T p.(Arg1465Ter)	het	[173]	-
	chr12:88534765-88534765	CEP290 c.148C>T p.(His50Tyr)	het	novel	-
13002499	chr19:54631469-54631469	PRPF31 c.967G>T p.(Glu323Ter)	het	novel	-
12008423[+]	chr1:216369924-216369924	USH2A c.4222C>T p.(Gln1408Ter)	hom	[134]	-
13012899	chrX:38176607-38176607	RPGR c.581G>A p.(Trp194Ter)	hemi	[174, 175]	-
12011769	chr2:182413512-182413513	CERKL c.1045_1046delAT	hom	novel	-
13005790	chr2:112779111-112779111	MERTK c.2302G>A p.(Ala768Thr)	hom	novel	-
13001457	chr8:55538727-55538731	RP1 c.2285_2289delTAAAT	het	[118]	-
13016341	chr3:129249866-129249866	RHO c.509C>G p.(Pro170Arg)	het	[176, 177]	-
13017339	chr1:197396856-197396856	CRB1 c.2401A>T p.(Lys801Ter)	hom	[33, 65]	-
13018482	chr4:16025917-16025917	PROM1 c.694+1G>A	het	novel	-
	chr4:16000112-16000112	PROM1 c.1579-1G>C	het	novel	-
13009597[+]	chr2:96958828-96958828	SNRNP200 c.2042G>A p.(Arg681Cys)	het	[101]	-
13003909[+]	chr1:197398735-197398735	CRB1 c.2833G>A p.(Gly945Arg)	het	novel	-
	chr1:197403836-197403836	CRB1 c.2843G>A p.(Cys948Tyr)	het	[33-37]	mutation hotspot [34]
13013461	chr1:94463488-94463488	ABCA4 c.6658C>T p.(Gln2220Ter)	hom	[52-54]	-
13018544	chrX:38150211-38150211	RPGR c.1572+1delG	hemi	novel	-
13003354	chr1:197396820-197396822	CRB1 c.2365_2367delAAT p.(Asn789del)	het	[178]	-
	chr1:197403836-197403836	CRB1 c.2843G>A p.(Cys948Tyr)	het	[33-37]	mutation hotspot [34]
12002961	chr3:101039148-101039149	IMPG2 c.68dupA	hom	novel	-

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MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
13013069	chr2:182468670-182468670	CERKL c.375C>G p.(Cys125Trp)	hom	[3, 4]	[5]
13008037[+]	chr1:216221935-216221935	USH2A c.6104G>A p.(Cys2035Tyr)	het	novel	-
	chr1:216420437-216420437	USH2A c.2299delG	het	[27, 115, 133-136]	-
12011625	chr8:55537904-55537904	RP1 c.1462delG	hom	novel	-
12010657[+]	chr14:57268813-57268813	OTX2 c.534C>A p.(Cys178Ter)	het	[179]	-
13000499[+]	chr2:182468670-182468670	CERKL c.375C>G p.(Cys125Trp)	het	[3, 4]	[5]
	chr2:182521541-182521541	CERKL c.193G>T p.(Glu65Ter)	het	novel	-
12008984[+]	chr11:61725751-61725753	BEST1 c.848_850delTTC p.(Phe283Ter)	het	[180]	-
13013377	chr6:66112400-66112400	EYS c.1155T>A p.(Cys385Ter)	hom	novel	-
12002569	chr11:119210284-119210284	C1QTNF5 c.489C>A p.(Ser163Arg)	het	c.489C>G [181]	-
12002956	chr14:88883069-88883069	SPATA7 c.253C>T p.(Arg85Ter)	hom	[182]	-
13012788	chr1:216420460-216420460	USH2A c.2276G>T p.(Cys759Phe)	het	[128-132]	-
	chr1:216380743-216380744	USH2A c.3187_3188delCA	het	[134]	-
11011655[+]	chr19:54631562-54631562	PRPF31 c.1060C>T p.(Arg354Ter)	het	novel	-
13006601	chr3:129251479-129251479	RHO c.800C>T p.(Pro267Leu)	het	[177, 183]	-
12010631[+]	chr8:55538471-55538471	RP1 c.2029C>T p.(Arg677Ter)	het	[118, 119]	-
12015182	chr1:215963510-215963510	USH2A c.10073G>A p.(Cys3358Tyr)	hom	[115, 132, 152, 153, 155]	-
13004890[+]	chr3:100963495-100963495	IMPG2 c.1680T>A p.(Tyr560Ter)	hom	[121]	-
13002513	chr5:149323870-149323870	PDE6A c.367G>T p.(Asp123Tyr)	het	novel	-
	chr5:149263008-149263008	PDE6A c.2119C>T p.(Arg707Trp)	het	novel	-
13000500[+]	chr16:57935275-57935275	CNGB1 c.2957A>T p.(Asn986Ile)	het	[39]	-

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MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
	chr16:57937839-57937839	CNGB1 c.2681G>A p.(Arg894His)	het	novel	-
	chr16:57951242-57951242	CNGB1 c.2096A>G p.(Asp699Gly)	het	novel	-
13007209	chr8:55539191-55539191	RP1 c.2749C>T p.(Gln917Ter)	het	novel	-
12002277	chr1:197297979-197297990	CRB1 c.498_506delAATTGATGGTTA p.(Ile167_Gly169del)	het	[35]	
	chr1:197396584-197396584	CRB1 c.2129A>T p.(Glu710Val)	het	[33, 38, 39]	-
13000103	chrX:38182652-38182652	RPGR c.154G>A p.(Gly52Arg)	hemi	[184, 185]	[186]
Patients referred with MD/STGD					
12008814	chr1:94514477-94514477	ABCA4 c.2690C>T p.(Thr897Ile)	het	[44, 187-190]	-
	chr1:94544977-94544977	ABCA4 c.1140T>A p.(Asn380Lys)	het	[44, 187, 189]	-
13012938[+]	chr17:6329101-6329101	AIPL1 c.834G>A p.(Trp278Ter)	het	[95]	[96]
13017782[+]	chr8:55533647-55533647	RP1 c.121T>C p.(Tyr41His)	het	novel	-
	chr8:55534041-55534041	RP1 c.515T>G p.(Leu172Arg)	het	[155]	-
13002608	chr1:94528164-94528164	ABCA4 c.1906C>T p.(Gln636Ter)	het	[147, 148]	-
	chr1:94496666-94496666	ABCA4 c.4139C>T p.(Pro1380Leu)	het	[12, 45, 48, 49, 191]	[50]
13014675	chr1:94496010-94496010	ABCA4 c.4326C>A p.(Asn1442Lys)	het	[141, 147, 149]	-
	chr1:94471065-94471065	ABCA4 c.6079C>T p.(Leu2027Phe)	het	[12]	[13]
13016023	chr11:61726979-61726979	BEST1 c.877C>A p.(Gln293Lys)	het	[192]	p.(Gln293His) [193]
13008350	chr2:182521541-182521541	CERKL c.193G>T p.(Glu65Ter)	hom	novel	-
13012365	chr1:94473807-94473807	ABCA4 c.5882G>A p.(Gly1961Glu)	het	[11, 49, 194, 195]	[50]
	chr1:94528197-94528197	ABCA4 c.1873C>T p.(Gln625Ter)	het	novel	-

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MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
13003074	chr2:182468670-182468670	CERKL c.375C>G p.(Cys125Trp)	hom	[3, 4]	[5]
13012308	chr19:48342929-48342929	CRX c.605delG	het	novel	-
13013879	chr1:94508386-94508386	ABCA4 c.3259G>A p.(Glu1087Lys)	hom	[10]	-
13001831[+]	chr1:94490567-94490567	ABCA4 c.4577C>T p.(Thr1526Met)	het	[45, 48]	-
	chr1:94486794-94486794	ABCA4 c.5018+2T>C	het	[45, 188]	-
12005906	chr1:94473807-94473807	ABCA4 c.5882G>A p.(Gly1961Glu)	het	[11, 49, 194, 195]	[50]
	chr1:94496666-94496666	ABCA4 c.4139C>T p.(Pro1380Leu)	het	[12, 45, 48, 49, 191]	[50]
13009479	chr6:42672272-42672272	PRPH2 c.659G>A p.(Asn220Gln)	het	p.(Arg220Trp) [196]	-
13012619	chr1:94471037-94471037	ABCA4 c.6107A>G p.(Tyr2036Cys)	het	novel	-
	chr1:94495071-94495071	ABCA4 c.4469G>A p.(Cys1490Tyr)	het	[46, 197, 198]	[197]
13009064[+]	chr19:48337801-48337801	CRX c.100+1G>C	het	novel	-
13017396	chr11:61723244-61723244	BEST1 c.302C>T p.(Pro101Leu)	het	[199] p.(Pro101Thr) [180, 200]	-
	chr11:61723364-61723364	BEST1 c.422G>A p.(Arg141His)	het	[180, 201-205]	[206]
12013892	chr9:2729470-2729470	KCNV2 c.1381G>A p.(Gly461Arg)	hom	[207-209]	-
12001090	chr1:94528819-94528819	ABCA4 c.1609C>T p.(Arg537Cys)	hom	[49]	-
13011741	chr2:182468670-182468670	CERKL c.375C>G p.(Cys125Trp)	hom	[3, 4]	[5]
12004145	chr1:94508969-94508969	ABCA4 c.3113C>T p.(Ala1038Val)	het	[11, 210]	[50]
	chr1:94577135-94577135	ABCA4 c.161G>A p.(Cys54Tyr)	het	[45, 48, 195]	-
13015690	chr1:94473807-94473807	ABCA4 c.5882G>A p.(Gly1961Glu)	het	[11, 49, 194, 195]	[50]
	chr1:94520688-94520688	ABCA4 c.2566T>A p.(Tyr856Asn)	het	novel	-

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MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
13006449	chr2:182468670-182468670	CERKL c.375C>G p.(Cys125Trp)	het	[3, 4]	[5]
	chr2:182521541-182521541	CERKL c.193G>T p.(Glu65Ter)	het	novel	-
Patients referred with USHER					
12015998	chr11:76924019-76924019	MYO7A c.6377delC	hom	[115]	-
12010758[+]	chr1:216420460-216420460	USH2A c.2276G>T p.(Cys759Phe)	het	[128-132]	-
	chr1:215847475-215847475	USH2A c.13778C>T p.(Ser4593Leu)	het	novel	-
13009036	chr1:216373453-216373453	USH2A c.3327C>A p.(Tyr1109Ter)	het	novel	-
	chr1:216420437-216420437	USH2A c.2299delG	het	[27, 115, 133-136]	-
12007911	chr1:216270451-216270451	USH2A c.4732C>T p.(Arg1578Cys)	het	[115]	-
	chr1:216019240-216019240	USH2A c.8981G>A p.(Trp2994Ter)	het	[211]	-
	chr1:216173905-216173905	USH2A c.6326-1G>A	het	novel	-
12000202[+]	chr11:76919517-76919517	MYO7A c.5899C>T p.(Arg1967Ter)	het	novel	-
	chr11:76869378-76869378	MYO7A c.905G>A p.(Arg302His)	het	[212]	minimal effect [213]
	chr11:76853873-76853873	MYO7A c.132+5G>A	het	novel	-
	chr11:76871263-76871263	MYO7A c.1135G>A p.(Gly379Arg)	het	novel	-
13012581[+]	chr1:215963510-215963510	USH2A c.10073G>A p.(Cys3358Tyr)	het	[115, 132, 152, 153, 155]	-
	chr1:215940074-215940074	USH2A c.10996T>G p.(Cys3666Gly)	het	novel	-
13011433[+]	chr1:216420214-216420214	USH2A c.2522C>A p.(Ser841Tyr)	het	[132, 214]	-
	chr1:215853690-215853690	USH2A c.12095G>T p.(Gly4032Val)	het	novel	-
	chr1:216243517-216243517	USH2A c.5975A>G p.(Tyr1992Cys)	het	[132, 152]	-
	chr1:216173760-216173760	USH2A c.6470delG	het	novel	-
12013066[+]	chr1:216040454-216040454	USH2A c.8740C>T p.(Arg2914Ter)	het	[152]	-
	chr1:216380743-216380744	USH2A c.3187_3188delCA	het	[134]	-
12006582	chr11:76900513-76900513	MYO7A c.3628A>T p.(Lys1210Ter)	het	novel	-
	chr11:76916643-76916643	MYO7A c.5617C>T p.(Arg1873Trp)	het	[215, 216]	-

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MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
11012656	chr1:216495311-216495311	USH2A c.1558delT	hom	novel	-
13005697	chr1:216420214-216420214	USH2A c.2522C>A p.(Ser841Tyr)	het	[132, 214]	-
	chr1:215847632-215847632	USH2A c.13621C>T p.(Gln4541Ter)	het	[115]	-
	chr1:215901725-215901725	USH2A c.11713C>T p.(Arg3905Cys)	het	novel	-
13013364	chr1:216062399-216062399	USH2A c.7595-3C>G	het	[105-107]	[107]
	chr1:216420075-216420075	USH2A c.2661C>G p.(Tyr887Ter)	het	novel	-
13001832	chr1:216420460-216420460	USH2A c.2276G>T p.(Cys759Phe)	hom	[128-132]	-
13002679	chr3:150690398-150690398	CLRN1 c.98G>A p.(Trp33Ter)	het	novel	-
	chr3:150645894-150645894	CLRN1 c.567T>G p.(Tyr189Ter)	het	[108, 109]	-
10002008[+]	chr11:76903286-76903286	MYO7A c.4115T>G p.(Val1372Gly)	het	novel	-
	chr11:76919822-76919822	MYO7A c.6025delG	het	[216, 217]	-
12006031[+]	chr1:215972365-215972365	USH2A c.9842G>T p.(Cys3281Phe)	het	[115]	-
	chr1:216420460-216420460	USH2A c.2276G>T p.(Cys759Phe)	het	[128-132]	-
	chr1:215848679-215848679	USH2A c.12574C>T p.(Arg4192Cys)	het	p.Arg4192His [152, 153]	-
	chr1:216373196-216373196	USH2A c.3584G>T p.(Cys1195Phe)	het	novel	-
	chr1:216420480-216420480	USH2A c.2256T>C p.(His752His)	het	[128, 130, 131]	-
12015458	chr9:117169063-117169063	DFNB31 c.1808G>T p.(Gly603Val)	het	novel	-
	chr9:117170303-117170303	DFNB31 c.1627-5T>A	het	novel	-
13008753	chr1:216270538-216270538	USH2A c.4645C>T p.(Arg1549Ter)	het	[132]	-
	chr1:216380775-216380775	USH2A c.3158-2A>G	het	novel	-
13002780[+]	chr1:216420437-216420437	USH2A c.2299delG	hom	[27, 115, 133-136]	-
12015030[+]	chr1:216369894-216369894	USH2A c.4251+1G>A	het	novel	-
	chr1:216420437-216420437	USH2A c.2299delG	het	[27, 115, 133-136]	-
12008422	chr1:216173784-216173784	USH2A c.6446C>A p.(Pro2149Gln)	hom	novel	-

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MCGM ID	Genomic loci *hg19*	Variant details *specified transcripts, Table S1*	Zygosity	Supporting evidence from literature	
				Disease segregation	Functional
13008826	chr1:216498866-216498867	USH2A c.920_923dupGCCA	het	[218, 219]	-
	chr1:215901562-215901563	USH2A c.11875_11876delCA	het	[220]	-
12012730	chr1:216420214-216420214	USH2A c.2522C>A p.(Ser841Tyr)	het	[132, 214]	-
	chr1:215822102-215822102	USH2A c.14350G>T p.(Glu4784Ter)	het	novel	-
	chr1:215823989-215823989	USH2A c.14288G>A p.(Gly4763Glu)	het	p.(Gly4763Arg) [152]	-
12007398	chr1:94480243-94480243	ABCA4 c.5316G>A p.(Trp1772Ter)	het	[49]	-
	chr1:94471055-94471055	ABCA4 c.6089G>A p.(Arg2030Gln)	het	[48, 191, 221]	-
12013349[+]	chr11:17542412-17542412	USH1C c.1210+5G>A	het	novel	c.1210+6T>G [107]
	chr11:17548769-17548769	USH1C c.496+1G>A	het	[115, 222]	-
13009855	chr1:216420437-216420437	USH2A c.2299delG	hom	[27, 115, 133-136]	-
13000951	chr10:73461778-73461778	CDH23 c.2398-1G>T	het	novel	-
	chr10:73565598-73565598	CDH23 c.7908C>G p.(Tyr2636Ter)	het	novel	-

Novel, at the time analysis the reported mutation was determined to not have been previously reported in the literature as a cause of inherited retinal disease, or to have been reported as a normal polymorphism. [+], additional familial segregation analysis has been requested and undertaken.

Supporting *functional* evidence includes biochemical assays for variants of interest, and detailed computational modeling and/or evidence-based discussion of the impact the variant of interest will have on protein structure and/or function.

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Supp. Table S5. Genetic variants reported to referring clinicians, including causative, likely causative, and variants of unknown clinical significance.

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Supp. Table S5. Genetic variants reported to referring clinicians, including causative, likely causative, and variants of unknown clinical significance.

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Supp. Table S5. Genetic variants reported to referring clinicians, including causative, likely causative, and variants of unknown clinical significance.

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Supp. Table S6. Individuals suggested for clinical reevaluation after genomic testing.

study id	referral group	gene reported
12007398	USHER	*ABCA4*
12010090	OTHER – atypical STGD	*BBS5*
12010854	RP/RCD	*CHM*
13012171	RP/RCD	*CHM*
13015421	RP/RCD	*CNGA3*
13004358	RP/RCD	*GPR98*
13017391	ACHM/CD	*GUCY2D*
12002962	RP/RCD	*PCDH15*
13017782	STGD/MD	*RP1*

Supp Table S7. The heterogeneity of inherited retinal disease reported in this study.

causal gene	number of individuals	number of disease referrals	disease referrals
ABCA4	20	5	CD (1), CRD (4), RP/RCD (3), STGD/MD (11), USHER (1)
AIPL1	3	2	EORD/LCA (2), STGD/MD (1)
BBS1	3	1	BBS (3)
BBS10	1	1	BBS (1)
BBS12	1	1	BBS (1)
BBS4	1	1	BBS (1)
BBS5	1	1	OTHER (1)
BBS7	1	1	BBS (1)
BEST1	3	2	RP/RCD (1), STGD/MD (2)
C1QTNF5	1	1	RP/RCD (1)
CDH23	1	1	USHER (1)
CDHR1	1	1	RP/RCD (1)
CEP290	12	4	CRD (1), JOUBERT (1), EORD/LCA (9), RP/RCD (1)
CERKL	15	4	CD (2), CRD (4), RP/RCD (5), STGD/MD (4)
CHM	2	1	RP/RCD (2)
CLRN1	3	2	RP/RCD (2), USHER (1)
CNGA1	2	1	RP/RCD (2)
CNGA3	6	3	CD (4), CRD (1), RP/RCD (1)
CNGB1	4	1	RP/RCD (4)
CNGB3	7	1	CD (7)
CRB1	21	3	CRD (2), EORD/LCA (10), RP/RCD (9)
CRX	4	3	CRD (1), EORD/LCA (1), STGD/MD (2)
DFNB31	1	1	USHER (1)
EYS	7	1	RP/RCD (7)
FAM161A	3	1	RP/RCD (1)
FZD4	2	1	FEVR (2)
GNAT2	1	1	CD (1)
GPR98	1	1	RP/RCD (1)
GUCA1A	1	1	RP/RCD (1)
GUCY2D	11	3	CD (3), EORD/LCA (7), RP/RCD (1)
IMPG2	3	1	RP/RCD (1)
KCNV2	1	1	STGD/MD (1)
LCA5	1	1	EORD/LCA (1)
LRAT	1	1	EORD/LCA (1)
LRP5	1	1	FEVR (1)
MERTK	4	1	RP/RCD (4)
MYO7A	5	2	EORD/LCA (1), USHER (4)
NR2E3	1	1	RP/RCD (1)
NRL	1	1	RP/RCD (1)
OTX2	1	1	RP/RCD (1)
PCDH15	1	1	RP/RCD (1)

PDE6A	1	1	RP/RCD (1)
PDE6B	1	1	RP/RCD (1)
PDE6C	1	1	CD (1)
PROM1	8	3	CRD (3), EORD/LCA (1), RP/RCD (4)
PRPF31	8	2	CRD (1), RP/RCD (7)
PRPF8	4	1	RP/RCD (1)
PRPH2	1	1	STGD/MD (1)
RDH12	4	2	EORD/LCA (3), RP/RCD (1)
RDH5	1	1	OTHER (1)
RHO	6	1	RP/RCD (6)
RP1	10	2	RP/RCD (9), STGD/MD (1)
RP2	5	2	EORD/LCA (1), RP/RCD (4)
RPE65	5	2	EORD/LCA (4), RP/RCD (1)
RPGR	7	2	CRD (1), RP/RCD (6)
RPGRIP1	8	2	CRD (1), EORD/LCA (7)
SNRNP200	5	1	RP/RCD (1)
SPATA7	2	2	EORD/LCA (1), RP/RCD (1)
TIMP3	1	1	RP/RCD (1)
TOPORS	1	1	RP/RCD (1)
USH1C	1	1	USHER (1)
USH2A	31	2	RP/RCD (13), USHER (18)

Supp. Table 8. Variants clinically reported as a novel cause of disease.

hg19 genomic co-ordinate	Variant annotation	Patient Study ID	Referral subtype
missense			
chr1:197325978	CRB1 c.1006T>C p.(Cys336Arg)	13005842	CRD
chr1:197396763	CRB1 c.2308G>A p.(Gly770Ser)*	11000824	RP/RCD
chr1:197398735	CRB1 c.2833G>A p.(Gly945Arg)	13003909	RP/RCD
chr1:197396764	CRB1 c.2309G>A p.(Gly770Asp)*	12014047	RP/RCD
chr1:197404010	CRB1 c.3017C>T p.(Ser1006Phe)*	12014047	RP/RCD
chr1:215847475	USH2A c.13778C>T p.(Ser4593Leu)	12010758	USHER
chr1:215847812	USH2A c.13441A>G p.(Arg4481Gly)	13015666	USHER
chr1:215853690	USH2A c.12095G>T p.(Gly4032Val)	13011433	USHER
chr1:215901725	USH2A c.11713C>T p.(Arg3905Cys)*	13005697	USHER
chr1:215940074	USH2A c.10996T>G p.(Cys3666Gly)	13012581	USHER
chr1:216173784	USH2A c.6446C>A p.(Pro2149Gln)	12008422	USHER
chr1:216221921	USH2A c.6118T>G p.(Cys2040Gly)	13003723	RP/RCD
chr1:216221935	USH2A c.6104G>A p.(Cys2035Tyr)	13008037	RP/RCD
chr1:216373196	USH2A c.3584G>T p.(Cys1195Phe)	12006031	USHER
chr1:94471037	ABCA4 c.6107A>G p.(Tyr2036Cys)	13012619	STGD/MD
chr1:94520688	ABCA4 c.2566T>A p.(Tyr856Asn)	13015690	STGD/MD
chr10:95405761	PDE6C c.1892A>G p.(Glu631Gly)	12003493	CD
chr10:95405763	PDE6C c.1894A>T p.(Arg632Trp)	12003493	CD
chr11:68115344	LRP5 c.121C>T p.(Arg41Trp)	13010229	FEVR
chr11:68153802	LRP5 c.1034T>A p.(Leu345Gln)	13010229	FEVR
chr11:76871263	MYO7A c.1135G>A p.(Gly379Arg)	12000202	USHER
chr11:76903286	MYO7A c.4115T>G p.(Val1372Gly)	10002008	USHER
chr11:86662837	FZD4 c.961G>A p.(Val321Ile)	12006042	FEVR
chr12:56115698	RDH5 c.536A>G p.(Lys179Arg)*	13010232	OTHER
chr12:88534765	CEP290 c.148C>T p.(His50Tyr)	12002965	RP/RCD
chr14:68192807	RDH12 c.383T>G p.(Val128Gly)	12012655	EORD/LCA
chr14:68200524	RDH12 c.910T>C p.(Trp304Arg)	12012655	EORD/LCA
chr15:72103895	NR2E3 c.191G>T p.(Cys64Phe)	13012950	RP/RCD
chr16:57937839	CNGB1 c.2681G>A p.(Arg894His)	13000500	RP/RCD
chr16:57951242	CNGB1 c.2096A>G p.(Asp699Gly)	13000500	RP/RCD
chr16:57935339	CNGB1 c.2893G>A p.(Gly965Ser)	13012959	RP/RCD
chr16:57965773	CNGB1 c.1382C>T p.(Thr461Met)	13012959	RP/RCD
chr17:1554982	PRPF8 c.6470T>A p.(Val2157Glu)	13008349	RP/RCD
chr17:7906745	GUCY2D c.380C>T p.(Pro127Arg)	13005797	EORD/LCA
chr19:54621779	PRPF31 c.121C>G p.(Leu41Val)	12011000	CRD
chr19:54625894	PRPF31 c.341T>A p.(Ile114Asn)	13002431	RP/RCD
chr19:54629963	PRPF31 c.916G>A p.(Asp306Asn)	13010305	RP/RCD
chr2:112761560	MERTK c.1866G>C p.(Lys622Asn)	13001452	RP/RCD
chr2:112777090	MERTK c.2180G>A p.(Arg727Gln)*	13000107	RP/RCD
chr2:112779111	MERTK c.2302G>A p.(Ala768Thr)	13005790	RP/RCD
chr2:96955598	SNRNP200 c.2879C>T p.(Ala960Val)	13007688	RP/RCD
chr2:96959219	SNRNP200 c.1871G>A p.(Arg624Lys)	12012775	RP/RCD
chr2:99013190	CNGA3 c.1557G>A p.(Met519Ile)	13012218	CD
chr2:99013564	CNGA3 c.1931T>C p.(Phe644Ser)	13015421	RP/RCD
chr4:47938964	CNGA1 c.1754T>G p.(Met585Arg)	13009346	RP/RCD
chr4:47942803	CNGA1 c.848A>T p.(Asp283Val)	13009346	RP/RCD
chr5:149263008	PDE6A c.2119C>T p.(Arg707Trp)	13002513	RP/RCD

chr5:149323870	PDE6A c.367G>T p.(Asp123Tyr)	13002513	RP/RCD
chr8:55533647	RP1 c.121T>C p.(Tyr41His)	13017782	STGD/MD
chr8:55534065	RP1 c.539T>G p.(Phe180Cys)	13015513	RP/RCD
chr9:117169063	DFNB31 c.1808G>T p.(Gly603Val)	12015458	USHER
chrX:38163914	RPGR c.908G>A p.(Gly303Glu)	13005635	RP/RCD
nonsense			
chr8:55534803	RP1 c.742C>T p.(Gln248Ter)	13015513	RP/RCD
chr1:197403862	CRB1 c.2869C>T p.(Gln957Ter)	13007873	EORD/LCA
chr1:197446947	CRB1 c.4159G>T p.(Glu1387Ter)	12016026	RP/RCD
chr1:215822102	USH2A c.14350G>T p.(Glu4784Ter)	12012730	USHER
chr1:215955512	USH2A c.10612C>T p.(Arg3538Ter)	10003578	RP/RCD
chr1:216373453	USH2A c.3327C>A p.(Tyr1109Ter)	13009036	USHER
chr1:216420075	USH2A c.2661C>G p.(Tyr887Ter)	13013364	USHER
chr1:94528197	ABCA4 c.1873C>T p.(Gln625Ter)	13012365	STGD/MD
chr10:73565598	CDH23 c.7908C>G p.(Tyr2636Ter)	13000951	USHER
chr11:76885842	MYO7A c.1976C>A p.(Ser659Ter)	13005796	EORD/LCA
chr11:76900513	MYO7A c.3628A>T p.(Lys1210Ter)	12006582	USHER
chr11:76919517	MYO7A c.5899C>T p.(Arg1967Ter)*	12000202	USHER
chr12:88477635	CEP290 c.4801C>T p.(Gln1601Ter)	12006834	EORD/LCA
chr12:88510853	CEP290 c.1781T>A p.(Leu594Ter)	13003353	EORD/LCA
chr14:21786006	RPGRIP1 c.1303A>T p.(Lys435Ter)	13002515	EORD/LCA
chr14:21788314	RPGRIP1 c.1445T>A p.(Leu482Ter)	13002689	EORD/LCA
chr14:21793489	RPGRIP1 c.2314C>T p.(Gln772Ter)	13002689	EORD/LCA
chr16:57998062	CNGB1 c.262C>T p.(Gln88Ter)	12012050	RP/RCD
chr17:7919559	GUCY2D c.3175A>T p.(Arg1059Ter)	083830	EORD/LCA
chr19:54631469	PRPF31 c.967G>T p.(Glu323Ter)	13002499	RP/RCD
chr19:54631496	PRPF31 c.994C>T p.(Gln332Ter)	11005352	RP/RCD
chr19:54631562	PRPF31 c.1060C>T p.(Arg354Ter)*	11011655	RP/RCD
		13006449;	STGD/MD;
		12013544;	RP/RCD;
chr2:182521541	CERKL c.193G>T p.(Glu65Ter)	13011267;	CRD;
		13008350;	STGD/MD;
		13000499	RP/RCD
chr2:62066675	FAM161A c.1464G>A p.(Trp488Ter)	11006153	RP/RCD
chr2:98996670	CNGA3 c.248G>A p.(Trp83Ter)	13015421	RP/RCD
chr3:100947720	IMPG2 c.3634G>T p.(Glu1212Ter)	12014873	RP/RCD
chr3:150690398	CLRN1 c.98G>A p.(Trp33Ter)	13002679	USHER
chr4:155665951	LRAT c.473G>A p.(Trp158Ter)	12004275	EORD/LCA
chr4:47938971	CNGA1 c.1747C>T p.(Arg583Ter)	13000106	RP/RCD
chr4:619706	PDE6B c.291C>A p.(Tyr97Ter)	12007088	RP/RCD
chr6:65016917	EYS c.6137G>A p.(Trp2046Ter)	12002958	RP/RCD
		13013377;	RP/RCD;
chr6:66112400	EYS c.1155T>A p.(Cys385Ter)	13002044;	RP/RCD;
		13004487	RP/RCD
chr8:55538474	RP1 c.2032C>T p.(Gln678Ter)	13004227	RP/RCD
chr8:55539191	RP1 c.2749C>T p.(Gln917Ter)	13007209	RP/RCD
chrX:38164014	RPGR c.808C>T p.(Gln270Ter)	13011161	RP/RCD
out-of-frame indels			
chr4:122749847-122749848	BBS7 c.1712_1713delinsAGA p.(Ser571Ter)	12014789	BBS
chr1:197390570-197390571	CRB1 c.1612_1613insCTTA	13012618	EORD/LCA

chr1:216173760	USH2A c.6470delG	13011433	USHER
chr1:216495311	USH2A c.1558delT	11012656	USHER
chr1:94508984	ABCA4 c.3098delA	082863	CRD
chr10:55582113-55582122	PCDH15 c.5385_5394delTCCTCTTCCT	12002962	RP/RCD
chr12:88487674-88487675	CEP290 c.3181_3182delAT	12004181	EORD/LCA
chr12:88508195-88508196	CEP290 c.2052+1_2052+2delGT*	12009433	EORD/LCA
chr14:21793021	RPGRIP1 c.2007delT	13013813	EORD/LCA
chr14:21794230-21794231	RPGRIP1 c.2608_2609insA*	12010986	CRD
chr14:68193897-68193927	RDH12 c.648_658+20del31	13005791	EORD/LCA
chr17:1565308-1565312	PRPF8 c.3910_3914delAACTC	12002571	RP/RCD
chr19:48342929	CRX c.605delG*	13012308	STGD/MD
chr19:54625953	PRPF31 c.400delG	12005728	RP/RCD
chr2:112725802-112725804	MERTK c.933_935delinsTT	13000107	RP/RCD
chr2:182413512-182413513	CERKL c.1045_1046delAT	12011769; 13015689	RP/RCD; CD
chr3:100962762-100962763	IMPG2 c.2412_2413delTG	12014873	RP/RCD
chr4:15985950	PROM1 c.2309delC	088301	CRD
chr4:15995680	PROM1 c.1697delA	12011770	RP/RCD
chr5:89975426-89975429	GPR98 c.5504_5507delTTCC	13004358	RP/RCD
chr6:65612025-65612026	EYS c.2826_2827delAT	13004716	RP/RCD
chr6:80223010-80223016	LCA5 c.633_639delAGCTAGA	12003872	EORD/LCA
chr8:55537904	RP1 c.1462delG	12011625	RP/RCD
chr8:55538040-55538043	RP1 c.1598_1601delGAAA	12013732	RP/RCD
chrX:38150211-38150211	RPGR c.1572+1delG	13018544	RP/RCD
chrX:46713315	RP2 c.507delT	13011266	RP/RCD
chr1:197313565-197313566	CRB1 c.807dupA	12005771	RP/RCD
chr10:85956379-85956380	CDHR1 c.270dupC	13002976	RP/RCD
chr14:88897545-88897546	SPATA7 c.1058dupC	12012257	EORD/LCA
chr16:57938727-57938728	CNGB1 c.2544dupG	12012050	RP/RCD
chr3:101039148-101039149	IMPG2 c.68dupA	12002961	RP/RCD
canonical splice			
chr1:197403835	CRB1 c.2843-1G>A	12007024	EORD/LCA
chr1:215914880	USH2A c.11549-1G>A	12011781	RP/RCD
chr1:216173905	USH2A c.6326-1G>A	12007911	USHER
chr1:216369894	USH2A c.4251+1G>A	12015030	USHER
chr1:216380775	USH2A c.3158-2A>G	13008753	USHER
chr1:68905008	RPE65 c.726-2A>C	13015227	EORD/LCA
chr10:73461778	CDH23 c.2398-1G>T	13000951	USHER
chr12:88532921	CEP290 c.297+1G>T*	13001467	EORD/LCA
chr14:68196099	RDH12 c.848+2T>C	13005789	EORD/LCA
chr15:73023645	BBS4 c.712-1G>A	12009772	BBS
chr19:48337801	CRX c.100+1G>C	13009064	STGD/MD
chr4:16000112	PROM1 c.1579-1G>C*	13018482; 12009616	RP/RCD; CRD
chr4:16014897	PROM1 c.1141+1G>A	13004232	RP/RCD
chr4:16025917	PROM1 c.694+1G>A	13018482	RP/RCD
synonymous			
chr1:216424273	USH2A c.2139C>T p.(Gly713Gly)	12000462	RP/RCD
chr10:55581921	PCDH15 c.5586C>A p.(Ala1862Ala)	12002962	RP/RCD
chr17:1564976	PRPF8 c.4131C>T p.(Ser1377Ser)	12003878	RP/RCD

chr4:16000058	PROM1 c.1632G>T p.(Gly544Gly)	12011770; 13004232	RP/RCD; RP/RCD
splice region			
chr11:17542412	USH1C c.1210+5G>A	12013349	USHER
chr11:76853873	MYO7A c.132+5G>A	12000202	USHER
chr4:16014893	PROM1 c.1141+5G>C	12013927	CRD
chr4:16025914	PROM1 c.694+4A>T	13002979	EORD/LCA
chr9:117170303	DFNB31 c.1627-5T>A	12015458	USHER
in-frame indels			
chrX:46713068	RP2 c.260_268del p.(Thr87_Cys89del)	11011508	EORD/LCA
chr14:24550532	NRL c.586_627dup p.(Ala196_Ala209dup)	13010253	RP/RCD
chr14:88904207-88904218	SPATA7 c.1241_1252del12 p.(Val414_Val417del)	12012257	EORD/LCA

* indicates variants which have been described in the literature since the variant was clinically reported as a novel cause of a disease: *CRB1* c.2308G>A p.(Gly770Ser)[1]; *CRB1* c.2309G>A p.(Gly770Asp)[2]; *CRB1* c.3017C>T p.(Ser1006Phe)[3]; *USH2A* c.11713C>T p.(Arg3905Cys)[4]; *RDH5* c.536A>G p.(Lys179Arg)[5]; *MERTK* c.2180G>A p.(Arg727Gln)[6]; *CEP290* c.297+1G>T[7]; *PROM1* c.1579-1G>C[8]; *MYO7A* c.5899C>T p.(Arg1967Ter)[9]; *PRPF31* c.1060C>T p.(Arg354Ter)[10]; *CEP290* c.2052+1_2052+2delGT[11]; *RPGRIP1* c.2608_2609insA[12]; *CRX* c.605delG[13]

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