PrismGuide IRD Panel Sequencing Report

Order ID: posi_con

Sample Information

Clinical Sample ID	posi_con
Customer-provided Sample Sex	Male

Sequencing Analysis Information

Panel	PrismGuide IRD Panel
Reagent	PrismGuide IRD Panel reagent kit
Analysis Execution Date	31 Jul 2023 14:25:25
On Target Reads (%)	73.86
On And Near Target Reads (%)	82.55
Off Target Reads (%)	17.45
Mapped Reads (%)	99.37
Duplicated Reads (%)	6.33
Reads Mapping Quality 0 (%)	3.53
Average Quality	36.5
Average Insert Size	195.1
Insert size std	55.3
Mean Target Coverage	588.33
Targets Not Covered	4
Evenness	83.83
Uniformity	1.573
Derivative Log Ratio Score for PrismGuide IRD Panel System	0.133
Pipeline-calculated Sample Sex	Male
Low Coverage (>=100x)	98.89
Medium Coverage	65.27
High Coverage	2.14

Data Analysis Information

Analysis Protocol	IRD Protocol v1
Pipeline Version	IRD Pipeline v2.4.3
Enable CNV Calling	Yes
Import SNV Results	Yes
CNV Calling Mode	Yes
Flanking Region (bp)	10
Allele Balance Priors Off	Yes
Minimum Alt Count	5
Minimum Base Quality	20
Minimum Total Read Count	20
Minimum Mapping Quality	30
Max Read Mismatch Fraction	0.04
Threshold for Targets Not Covered	1
Germline Minimum Alt Fraction	0.2
Germline Segmentation p-value Threshold	0.01
Genetic aberration selection criteria (SNV, InDel)	Default SNV Filter
Genetic aberration selection criteria (CNV)	Default CNV Filter
Analysis Program Version	0.3.20

SNV/Indel Information

1	Variant (HGVSc)	NM_000350.3:c.5318C>T
	Gene Symbol	ABCA4
	Variant Type	SNV
	Location	1:94014685-94014685
	Exon Numbers	38/50
	Ref	G
	Alt	A
	Frequency (Read)	0.53289 (721/1353)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000341.2:p.Ala1773Val
	User Classification	Pathogenic
	ACMG Classification	Pathogenic
	ACMG Labels	PS1(ClinVar,HGMD,Custom Variant List),PS3(HGMD,ClinVar),PM2(gnomAD),PP2(ClinVar),PP3(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	7.557E-5
	Allele frequency in gnomAD - East Asian	5.437E-5
	1000 Genomes Minor Allele	-

	1000 Genomes Minor Allele Frequency	-
	SIFT Prediction	deleterious
	dbSNP ID	rs760549861
	ClinVar ID	236129
	ClinVar Pathogenicity	Pathogenic/Likely_pathogenic
	HGMD	CM087708
	GEM-J WGA Allele Frequency	6.586E-5
	Report Inclusion	Prioritized, Shortlisted
2	Variant (HGVSc)	NM_000350.3:c.1760+2T>G
	Gene Symbol	ABCA4
	Variant Type	SNV
	Location	1:94063110-94063110
	Exon Numbers	12/50
	Ref	A
	Alt	C
	Frequency (Read)	0.53697 (610/1136)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Pathogenic
	ACMG Classification	Pathogenic
	ACMG Labels	PVS1(VEP),PS3(HGMD),PM2(gnomAD),PP3(dbscSNV),PP5(Pathogenic Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	splice_donor_variant
	Allele frequency in gnomAD - whole population	-
	Allele frequency in gnomAD - East Asian	-
	1000 Genomes Minor Allele	-
	1000 Genomes Minor Allele Frequency	-
	SIFT Prediction	-
	dbSNP ID	rs61751385
	ClinVar ID	99076
	ClinVar Pathogenicity	Pathogenic
	HGMD	CS024002
	GEM-J WGA Allele Frequency	6.602E-4
	Report Inclusion	Prioritized, Shortlisted
3	Variant (HGVSc)	NM_000329.3:c.1154C>T
	Gene Symbol	RPE65
	Variant Type	SNV
	Location	1:68431560-68431560
	Exon Numbers	11/14
	Ref	G
	Alt	A
	Frequency (Read)	0.53728 (663/1234)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000320.1:p.Thr385Met
	User Classification	Pathogenic
	ACMG Classification	Pathogenic
	ACMG Labels	PS1(HGMD),PS3(HGMD),PM2(gnomAD),PP2(ClinVar),PP3(BLOSUM),BP4(SIFT)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	2.436E-4
	Allele frequency in gnomAD - East Asian	0.002992
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.0012
	SIFT Prediction	tolerated
	dbSNP ID	rs201379753
	ClinVar ID	712719
	ClinVar Pathogenicity	Conflicting_interpretations_of_pathogenicity
	HGMD	CM2015142
	GEM-J WGA Allele Frequency	0.005265
	Report Inclusion	Prioritized, Shortlisted
4	Variant (HGVSc)	NM_001034853.2:c.2838_2839del
	Gene Symbol	RPGR
	Variant Type	Deletion
	Location	X:38286159-38286161
	Exon Numbers	15/15
	Ref	TCC
	Alt	T
	Frequency (Read)	0.98675 (149/151)
	Zygosity	Hemizygous
	Amino Acid Change (HGVSp)	NP_001030025.1:p.Glu947GlyfsTer131

	User Classification	Pathogenic
	ACMG Classification	Pathogenic
	ACMG Labels	PVS1(VEP),PS3(HGMD,ClinVar),PM2(gnomAD)
	Inheritance Pattern	X-linked
	Variant Function	frameshift_variant
	Allele frequency in gnomAD - whole population	-
	Allele frequency in gnomAD - East Asian	-
	1000 Genomes Minor Allele	-
	1000 Genomes Minor Allele Frequency	-
	SIFT Prediction	-
	dbSNP ID	-
	ClinVar ID	975119
	ClinVar Pathogenicity	Pathogenic/Likely_pathogenic
	HGMD	CD070505
	GEM-J WGA Allele Frequency	-
	Report Inclusion	Prioritized, Shortlisted
5	Variant (HGVSc)	NM_025114.4:c.2991+1655A>G
	Gene Symbol	CEP290
	Variant Type	SNV
	Location	12:88101183-88101183
	Exon Numbers	Intronic
	Ref	T
	Alt	C
	Frequency (Read)	0.24345 (158/649)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Likely pathogenic
	ACMG Classification	Likely Pathogenic
	ACMG Labels	PS3(HGMD,ClinVar),PM2(gnomAD)
	Inheritance Pattern	Recessive
	Variant Function	intron_variant
	Allele frequency in gnomAD - whole population	-
	Allele frequency in gnomAD - East Asian	-
	1000 Genomes Minor Allele	-
	1000 Genomes Minor Allele Frequency	-
	SIFT Prediction	-
	dbSNP ID	rs281865192
	ClinVar ID	1337
	ClinVar Pathogenicity	Pathogenic/Likely_pathogenic
	HGMD	CS064383
	GEM-J WGA Allele Frequency	-
	Report Inclusion	Prioritized, Shortlisted
6	Variant (HGVSc)	NM_005183.4:c.1118+6A>G
	Gene Symbol	CACNA1F
	Variant Type	SNV
	Location	X:49228030-49228030
	Exon Numbers	Intronic
	Ref	T
	Alt	C
	Frequency (Read)	1.0 (320/320)
	Zygosity	Hemizygous
	Amino Acid Change (HGVSp)	-
	User Classification	Uncertain significance
	ACMG Classification	Uncertain significance
	ACMG Labels	PM2(gnomAD)
	Inheritance Pattern	X-linked
	Variant Function	splice_donor_region_variant,intron_variant
	Allele frequency in gnomAD - whole population	-
	Allele frequency in gnomAD - East Asian	-
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.6432
	SIFT Prediction	-
	dbSNP ID	rs6609854
	ClinVar ID	-
	ClinVar Pathogenicity	-
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
7	Variant (HGVSc)	NM_006343.3:c.2079+181A>G

	Gene Symbol	MERTK
	Variant Type	SNV
	Location	2:112010247-112010247
	Exon Numbers	Intronic
	Ref	A
	Alt	G
	Frequency (Read)	0.47377 (280/591)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Uncertain significance
	ACMG Classification	Uncertain significance
	ACMG Labels	PM2(gnomAD)
	Inheritance Pattern	Recessive
	Variant Function	intron_variant
	Allele frequency in gnomAD - whole population	-
	Allele frequency in gnomAD - East Asian	-
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.2468
	SIFT Prediction	-
	dbSNP ID	rs77781750
	ClinVar ID	-
	ClinVar Pathogenicity	-
	HGMD	-
	GEM-J WGA Allele Frequency	0.117
	Report Inclusion	None
8	Variant (HGVSc)	NM_001375654.1:c.3816C>T
	Gene Symbol	RP1
	Variant Type	SNV
	Location	8:54852653-54852653
	Exon Numbers	27/30
	Ref	C
	Alt	T
	Frequency (Read)	0.50265 (284/565)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_001362583.1:p.Ile1272=
	User Classification	Uncertain significance
	ACMG Classification	Uncertain significance
	ACMG Labels	PM2(gnomAD),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	-
	Allele frequency in gnomAD - East Asian	-
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	8.0E-4
	SIFT Prediction	-
	dbSNP ID	rs541936347
	ClinVar ID	-
	ClinVar Pathogenicity	-
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
9	Variant (HGVSc)	NM_005183.4:c.3309C>T
	Gene Symbol	CACNA1F
	Variant Type	SNV
	Location	X:49215504-49215504
	Exon Numbers	28/48
	Ref	G
	Alt	A
	Frequency (Read)	1.0 (335/335)
	Zygosity	Hemizygous
	Amino Acid Change (HGVSp)	NP_005174.2:p.His1103=
	User Classification	Likely benign
	ACMG Classification	Likely Benign
	ACMG Labels	PM2(gnomAD),BP6(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	X-linked
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	-
	Allele frequency in gnomAD - East Asian	-
	1000 Genomes Minor Allele	A

	1000 Genomes Minor Allele Frequency	0.4479
	SIFT Prediction	-
	dbSNP ID	rs2075866
	ClinVar ID	769231
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
10	Variant (HGVSc)	NM_005183.4:c.1842T>C
	Gene Symbol	CACNA1F
	Variant Type	SNV
	Location	X:49224829-49224829
	Exon Numbers	14/48
	Ref	A
	Alt	G
	Frequency (Read)	1.0 (433/433)
	Zygosity	Hemizygous
	Amino Acid Change (HGVSp)	NP_005174.2:p.Gly614=
	User Classification	Likely benign
	ACMG Classification	Likely Benign
	ACMG Labels	PM2(gnomAD),BP6(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	X-linked
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	-
	Allele frequency in gnomAD - East Asian	-
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.5595
	SIFT Prediction	-
	dbSNP ID	rs2235127
	ClinVar ID	769230
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
11	Variant (HGVSc)	NM_000260.4:c.2754C>T
	Gene Symbol	MYO7A
	Variant Type	SNV
	Location	11:77181439-77181439
	Exon Numbers	23/49
	Ref	C
	Alt	T
	Frequency (Read)	0.50792 (481/947)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000251.3:p.Ala918=
	User Classification	Likely benign
	ACMG Classification	Likely Benign
	ACMG Labels	PM2(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.003703
	Allele frequency in gnomAD - East Asian	0.0
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.0086
	SIFT Prediction	-
	dbSNP ID	rs78072361
	ClinVar ID	43188
	ClinVar Pathogenicity	Benign/Likely_benign
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
12	Variant (HGVSc)	NM_001375654.1:c.2352G>A
	Gene Symbol	RP1
	Variant Type	SNV
	Location	8:54720269-54720269
	Exon Numbers	16/30
	Ref	G
	Alt	A
	Frequency (Read)	0.43007 (246/572)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_001362583.1:p.Lys784=

	User Classification	Likely benign
	ACMG Classification	Likely Benign
	ACMG Labels	BS1(gnomAD),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.01532
	Allele frequency in gnomAD - East Asian	0.0
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.0232
	SIFT Prediction	-
	dbSNP ID	rs76985026
	ClinVar ID	-
	ClinVar Pathogenicity	-
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
13	Variant (HGVSc)	NM_001375654.1:c.3319A>C
	Gene Symbol	RP1
	Variant Type	SNV
	Location	8:54770134-54770134
	Exon Numbers	24/30
	Ref	A
	Alt	C
	Frequency (Read)	0.44681 (105/235)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_001362583.1:p.Asn1107His
	User Classification	Likely benign
	ACMG Classification	Likely Benign
	ACMG Labels	PM2(gnomAD),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	-
	Allele frequency in gnomAD - East Asian	-
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.0309
	SIFT Prediction	tolerated
	dbSNP ID	rs114997280
	ClinVar ID	-
	ClinVar Pathogenicity	-
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
14	Variant (HGVSc)	NM_178857.6:c.6545C>A
	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10607553-10607553
	Exon Numbers	4/4
	Ref	G
	Alt	T
	Frequency (Read)	0.46746 (395/845)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_849188.4:p.Ala2182Asp
	User Classification	Likely benign
	ACMG Classification	Likely Benign
	ACMG Labels	PP3(BLOSUM),BS1(gnomAD),BP1(ClinVar),BP4(SIFT),BP6(ClinVar)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.007286
	Allele frequency in gnomAD - East Asian	5.562E-5
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.0038
	SIFT Prediction	tolerated
	dbSNP ID	rs183570817
	ClinVar ID	361220
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
15	Variant (HGVSc)	NM_178857.6:c.5837C>A

	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10608261-10608261
	Exon Numbers	4/4
	Ref	G
	Alt	T
	Frequency (Read)	1.0 (803/803)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_849188.4:p.Ala1946Glu
	User Classification	Likely benign
	ACMG Classification	Likely Benign
	ACMG Labels	PM2(gnomAD),PP3(BLOSUM),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	-
	Allele frequency in gnomAD - East Asian	-
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.9197
	SIFT Prediction	tolerated_low_confidence
	dbSNP ID	rs11785822
	ClinVar ID	361236
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.998
	Report Inclusion	None
16	Variant (HGVSc)	NM_178857.6:c.4019_4020delinsGG
	Gene Symbol	RP1L1
	Variant Type	MNP
	Location	8:10610078-10610079
	Exon Numbers	4/4
	Ref	TT
	Alt	CC
	Frequency (Read)	0.27044 (258/954)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_849188.4:p.Glu1340Gly
	User Classification	Likely benign
	ACMG Classification	Likely Benign
	ACMG Labels	PM2(gnomAD),PP3(BLOSUM),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	-
	Allele frequency in gnomAD - East Asian	-
	1000 Genomes Minor Allele	-
	1000 Genomes Minor Allele Frequency	-
	SIFT Prediction	tolerated
	dbSNP ID	rs1554451516
	ClinVar ID	-
	ClinVar Pathogenicity	-
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
17	Variant (HGVSc)	NM_178857.6:c.3956C>G
	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10610142-10610142
	Exon Numbers	4/4
	Ref	G
	Alt	C
	Frequency (Read)	0.87391 (804/920)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_849188.4:p.Ala1319Gly
	User Classification	Likely benign
	ACMG Classification	Likely Benign
	ACMG Labels	PM2(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	-
	Allele frequency in gnomAD - East Asian	-
	1000 Genomes Minor Allele	-

	1000 Genomes Minor Allele Frequency	-
	SIFT Prediction	tolerated
	dbSNP ID	rs4840501
	ClinVar ID	361300
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.489
	Report Inclusion	None
18	Variant (HGVSc)	NM_022367.4:c.2187G>A
	Gene Symbol	SEMA4A
	Variant Type	SNV
	Location	1:156176898-156176898
	Exon Numbers	15/15
	Ref	G
	Alt	A
	Frequency (Read)	0.50693 (439/866)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_071762.2:p.Pro729=
	User Classification	Likely benign
	ACMG Classification	Likely Benign
	ACMG Labels	BS1(gnomAD),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Dominant
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.01542
	Allele frequency in gnomAD - East Asian	0.0
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.0068
	SIFT Prediction	-
	dbSNP ID	rs41265019
	ClinVar ID	292858
	ClinVar Pathogenicity	Conflicting_interpretations_of_pathogenicity
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
19	Variant (HGVSc)	NM_206933.4:c.1434G>C
	Gene Symbol	USH2A
	Variant Type	SNV
	Location	1:216323590-216323590
	Exon Numbers	8/72
	Ref	C
	Alt	G
	Frequency (Read)	0.54383 (304/559)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_996816.3:p.Glu478Asp
	User Classification	Likely benign
	ACMG Classification	Likely Benign
	ACMG Labels	BS1(gnomAD),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.0121
	Allele frequency in gnomAD - East Asian	0.0
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.0048
	SIFT Prediction	tolerated
	dbSNP ID	rs35730265
	ClinVar ID	48437
	ClinVar Pathogenicity	Conflicting_interpretations_of_pathogenicity
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
20	Variant (HGVSc)	NM_000350.3:c.6764G>T
	Gene Symbol	ABCA4
	Variant Type	SNV
	Location	1:93996161-93996161
	Exon Numbers	49/50
	Ref	C
	Alt	A
	Frequency (Read)	0.52402 (349/666)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000341.2:p.Ser2255Ile

	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP2(ClinVar),PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.07149
	Allele frequency in gnomAD - East Asian	0.02702
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.1769
	SIFT Prediction	tolerated
	dbSNP ID	rs6666652
	ClinVar ID	99494
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.019
	Report Inclusion	None
21	Variant (HGVSc)	NM_000350.3:c.6730-3T>C
	Gene Symbol	ABCA4
	Variant Type	SNV
	Location	1:93996198-93996198
	Exon Numbers	Intronic
	Ref	A
	Alt	G
	Frequency (Read)	0.53008 (282/532)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(dbscSNV),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	splice_polypyrimidine_tract_variant,splice_region_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.06522
	Allele frequency in gnomAD - East Asian	0.02703
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.1486
	SIFT Prediction	-
	dbSNP ID	rs1800717
	ClinVar ID	99491
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.019
	Report Inclusion	None
22	Variant (HGVSc)	NM_000350.3:c.6285T>C
	Gene Symbol	ABCA4
	Variant Type	SNV
	Location	1:94001103-94001103
	Exon Numbers	46/50
	Ref	A
	Alt	G
	Frequency (Read)	0.48544 (250/515)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000341.2:p.Asp2095=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	splice_region_variant,synonymous_variant
	Allele frequency in gnomAD - whole population	0.1578
	Allele frequency in gnomAD - East Asian	0.1486
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.3011
	SIFT Prediction	-
	dbSNP ID	rs1801555
	ClinVar ID	99444
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.144
	Report Inclusion	None
23	Variant (HGVSc)	NM_000350.3:c.6282+7G>A

	Gene Symbol	ABCA4
	Variant Type	SNV
	Location	1:94001851-94001851
	Exon Numbers	Intronic
	Ref	C
	Alt	T
	Frequency (Read)	0.49858 (350/702)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(dbscSNV)
	Inheritance Pattern	Recessive
	Variant Function	splice_region_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.0766
	Allele frequency in gnomAD - East Asian	0.06161
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.134
	SIFT Prediction	-
	dbSNP ID	rs17110761
	ClinVar ID	99443
	ClinVar Pathogenicity	Benign/Likely_benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.041
	Report Inclusion	None
24	Variant (HGVSc)	NM_000350.3:c.6249C>T
	Gene Symbol	ABCA4
	Variant Type	SNV
	Location	1:94001891-94001891
	Exon Numbers	45/50
	Ref	G
	Alt	A
	Frequency (Read)	0.53635 (450/839)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000341.2:p.Ile2083=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.07668
	Allele frequency in gnomAD - East Asian	0.06145
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.134
	SIFT Prediction	-
	dbSNP ID	rs1801359
	ClinVar ID	99440
	ClinVar Pathogenicity	Benign/Likely_benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.041
	Report Inclusion	None
25	Variant (HGVSc)	NM_000350.3:c.6069T>C
	Gene Symbol	ABCA4
	Variant Type	SNV
	Location	1:94005519-94005519
	Exon Numbers	44/50
	Ref	A
	Alt	G
	Frequency (Read)	0.45991 (304/661)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000341.2:p.Ile2023=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.8979
	Allele frequency in gnomAD - East Asian	0.8852
	1000 Genomes Minor Allele	G

	1000 Genomes Minor Allele Frequency	0.77
	SIFT Prediction	-
	dbSNP ID	rs1762114
	ClinVar ID	136235
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.925
	Report Inclusion	None
26	Variant (HGVSc)	NM_000350.3:c.635G>A
	Gene Symbol	ABCA4
	Variant Type	SNV
	Location	1:94098927-94098927
	Exon Numbers	6/50
	Ref	C
	Alt	T
	Frequency (Read)	0.44207 (393/889)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000341.2:p.Arg212His
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PM5(ClinVar),PP2(ClinVar),PP3(SIFT),BS1(gnomAD),BS3(ClinVar),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.04109
	Allele frequency in gnomAD - East Asian	0.06135
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.0533
	SIFT Prediction	deleterious
	dbSNP ID	rs6657239
	ClinVar ID	99454
	ClinVar Pathogenicity	Benign/Likely_benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.059
	Report Inclusion	None
27	Variant (HGVSc)	NM_000350.3:c.141A>G
	Gene Symbol	ABCA4
	Variant Type	SNV
	Location	1:94112992-94112992
	Exon Numbers	2/50
	Ref	T
	Alt	C
	Frequency (Read)	1.0 (618/618)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_000341.2:p.Pro47=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.9971
	Allele frequency in gnomAD - East Asian	0.9999
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.987
	SIFT Prediction	-
	dbSNP ID	rs4847281
	ClinVar ID	166618
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	1.0
	Report Inclusion	None
28	Variant (HGVSc)	NM_032119.4:c.2367+8C>T
	Gene Symbol	ADGRV1
	Variant Type	SNV
	Location	5:90642770-90642770
	Exon Numbers	Intronic
	Ref	C
	Alt	T
	Frequency (Read)	0.49622 (197/397)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-

	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(dbscSNV)
	Inheritance Pattern	Recessive
	Variant Function	splice_region_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.7328
	Allele frequency in gnomAD - East Asian	0.8072
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.8213
	SIFT Prediction	-
	dbSNP ID	rs2366773
	ClinVar ID	46305
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.837
	Report Inclusion	None
29	Variant (HGVSc)	NM_032119.4:c.3279G>T
	Gene Symbol	ADGRV1
	Variant Type	SNV
	Location	5:90647754-90647754
	Exon Numbers	17/90
	Ref	G
	Alt	T
	Frequency (Read)	0.42105 (184/437)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_115495.3:p.Leu1093Phe
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.7114
	Allele frequency in gnomAD - East Asian	0.8014
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.8103
	SIFT Prediction	tolerated
	dbSNP ID	rs2366777
	ClinVar ID	46313
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.833
	Report Inclusion	None
30	Variant (HGVSc)	NM_032119.4:c.5851G>A
	Gene Symbol	ADGRV1
	Variant Type	SNV
	Location	5:90683772-90683772
	Exon Numbers	28/90
	Ref	G
	Alt	A
	Frequency (Read)	0.49228 (319/648)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_115495.3:p.Val1951Ile
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.7096
	Allele frequency in gnomAD - East Asian	0.8007
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.8037
	SIFT Prediction	tolerated
	dbSNP ID	rs4916684
	ClinVar ID	46343
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.836
	Report Inclusion	None
31	Variant (HGVSc)	NM_032119.4:c.5960C>T

	Gene Symbol	ADGRV1
	Variant Type	SNV
	Location	5:90683881-90683881
	Exon Numbers	28/90
	Ref	C
	Alt	T
	Frequency (Read)	0.51495 (310/602)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_115495.3:p.Pro1987Leu
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.3639
	Allele frequency in gnomAD - East Asian	0.3692
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.3373
	SIFT Prediction	tolerated
	dbSNP ID	rs4916685
	ClinVar ID	46345
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.486
	Report Inclusion	None
32	Variant (HGVSc)	NM_032119.4:c.6695A>G
	Gene Symbol	ADGRV1
	Variant Type	SNV
	Location	5:90690065-90690065
	Exon Numbers	30/90
	Ref	A
	Alt	G
	Frequency (Read)	0.45243 (214/473)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_115495.3:p.Tyr2232Cys
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(SIFT),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.3647
	Allele frequency in gnomAD - East Asian	0.3653
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.3498
	SIFT Prediction	deleterious
	dbSNP ID	rs10037067
	ClinVar ID	46356
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.486
	Report Inclusion	None
33	Variant (HGVSc)	NM_032119.4:c.6952-10G>A
	Gene Symbol	ADGRV1
	Variant Type	SNV
	Location	5:90692595-90692595
	Exon Numbers	Intronic
	Ref	G
	Alt	A
	Frequency (Read)	0.44474 (165/371)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	splice_polypyrimidine_tract_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.3479
	Allele frequency in gnomAD - East Asian	0.3608
	1000 Genomes Minor Allele	A

	1000 Genomes Minor Allele Frequency	0.3221
	SIFT Prediction	-
	dbSNP ID	rs10040165
	ClinVar ID	46358
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.486
	Report Inclusion	None
34	Variant (HGVSc)	NM_032119.4:c.7034A>G
	Gene Symbol	ADGRV1
	Variant Type	SNV
	Location	5:90692687-90692687
	Exon Numbers	32/90
	Ref	A
	Alt	G
	Frequency (Read)	0.46281 (280/605)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_115495.3:p.Asn2345Ser
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.3581
	Allele frequency in gnomAD - East Asian	0.369
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.3223
	SIFT Prediction	tolerated
	dbSNP ID	rs2366926
	ClinVar ID	46360
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.487
	Report Inclusion	None
35	Variant (HGVSc)	NM_032119.4:c.7206G>A
	Gene Symbol	ADGRV1
	Variant Type	SNV
	Location	5:90693962-90693962
	Exon Numbers	33/90
	Ref	G
	Alt	A
	Frequency (Read)	0.49849 (331/664)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_115495.3:p.Glu2402=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.3596
	Allele frequency in gnomAD - East Asian	0.3681
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.3221
	SIFT Prediction	-
	dbSNP ID	rs16876822
	ClinVar ID	46365
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.486
	Report Inclusion	None
36	Variant (HGVSc)	NM_032119.4:c.7751A>G
	Gene Symbol	ADGRV1
	Variant Type	SNV
	Location	5:90694507-90694507
	Exon Numbers	33/90
	Ref	A
	Alt	G
	Frequency (Read)	0.45847 (287/626)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_115495.3:p.Asn2584Ser

	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.7107
	Allele frequency in gnomAD - East Asian	0.8006
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.8085
	SIFT Prediction	tolerated
	dbSNP ID	rs1878878
	ClinVar ID	46379
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.836
	Report Inclusion	None
37	Variant (HGVSc)	NM_032119.4:c.8730+21dup
	Gene Symbol	ADGRV1
	Variant Type	Insertion
	Location	5:90706403-90706403
	Exon Numbers	Intronic
	Ref	C
	Alt	CT
	Frequency (Read)	0.36777 (89/242)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	intron_variant
	Allele frequency in gnomAD - whole population	0.4707
	Allele frequency in gnomAD - East Asian	0.4916
	1000 Genomes Minor Allele	-
	1000 Genomes Minor Allele Frequency	-
	SIFT Prediction	-
	dbSNP ID	rs60522638
	ClinVar ID	197047
	ClinVar Pathogenicity	Benign/Likely_benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.756
	Report Inclusion	None
38	Variant (HGVSc)	NM_032119.4:c.9927T>G
	Gene Symbol	ADGRV1
	Variant Type	SNV
	Location	5:90725106-90725106
	Exon Numbers	47/90
	Ref	T
	Alt	G
	Frequency (Read)	0.49451 (90/182)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_115495.3:p.Pro3309=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.3459
	Allele frequency in gnomAD - East Asian	0.3665
	1000 Genomes Minor Allele	-
	1000 Genomes Minor Allele Frequency	-
	SIFT Prediction	-
	dbSNP ID	rs16869042
	ClinVar ID	46409
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.488
	Report Inclusion	None
39	Variant (HGVSc)	NM_032119.4:c.10411G>A

	Gene Symbol	ADGRV1
	Variant Type	SNV
	Location	5:90728918-90728918
	Exon Numbers	49/90
	Ref	G
	Alt	A
	Frequency (Read)	0.5045 (168/333)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_115495.3:p.Glu3471Lys
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.75
	Allele frequency in gnomAD - East Asian	0.7846
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.8291
	SIFT Prediction	tolerated
	dbSNP ID	rs2366928
	ClinVar ID	46248
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.821
	Report Inclusion	None
40	Variant (HGVSc)	NM_032119.4:c.10872A>G
	Gene Symbol	ADGRV1
	Variant Type	SNV
	Location	5:90745693-90745693
	Exon Numbers	52/90
	Ref	A
	Alt	G
	Frequency (Read)	0.40892 (220/538)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_115495.3:p.Gln3624=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.03118
	Allele frequency in gnomAD - East Asian	5.572E-5
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.0174
	SIFT Prediction	-
	dbSNP ID	rs17624033
	ClinVar ID	46252
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
41	Variant (HGVSc)	NM_032119.4:c.11581-3dup
	Gene Symbol	ADGRV1
	Variant Type	Insertion
	Location	5:90756446-90756446
	Exon Numbers	Intronic
	Ref	T
	Alt	TC
	Frequency (Read)	0.43778 (197/450)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	splice_polypyrimidine_tract_variant,splice_region_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.4024
	Allele frequency in gnomAD - East Asian	0.4074
	1000 Genomes Minor Allele	C

	1000 Genomes Minor Allele Frequency	0.3852
	SIFT Prediction	-
	dbSNP ID	rs34894132
	ClinVar ID	46256
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.551
	Report Inclusion	None
42	Variant (HGVSc)	NM_032119.4:c.11599G>A
	Gene Symbol	ADGRV1
	Variant Type	SNV
	Location	5:90756472-90756472
	Exon Numbers	56/90
	Ref	G
	Alt	A
	Frequency (Read)	0.47312 (264/558)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_115495.3:p.Glu3867Lys
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.3439
	Allele frequency in gnomAD - East Asian	0.4333
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.2961
	SIFT Prediction	tolerated
	dbSNP ID	rs10062026
	ClinVar ID	46257
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.551
	Report Inclusion	None
43	Variant (HGVSc)	NM_032119.4:c.11682C>T
	Gene Symbol	ADGRV1
	Variant Type	SNV
	Location	5:90756555-90756555
	Exon Numbers	56/90
	Ref	C
	Alt	T
	Frequency (Read)	0.4538 (275/606)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_115495.3:p.Pro3894=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.4868
	Allele frequency in gnomAD - East Asian	0.4556
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.5393
	SIFT Prediction	-
	dbSNP ID	rs2438349
	ClinVar ID	46258
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.552
	Report Inclusion	None
44	Variant (HGVSc)	NM_032119.4:c.16031A>G
	Gene Symbol	ADGRV1
	Variant Type	SNV
	Location	5:90811291-90811291
	Exon Numbers	74/90
	Ref	A
	Alt	G
	Frequency (Read)	0.99821 (558/559)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_115495.3:p.Glu5344Gly

	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.9696
	Allele frequency in gnomAD - East Asian	1.0
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.9461
	SIFT Prediction	tolerated
	dbSNP ID	rs2438374
	ClinVar ID	46282
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	1.0
	Report Inclusion	None
45	Variant (HGVSc)	NM_032119.4:c.16279G>A
	Gene Symbol	ADGRV1
	Variant Type	SNV
	Location	5:90823507-90823507
	Exon Numbers	76/90
	Ref	G
	Alt	A
	Frequency (Read)	1.0 (684/684)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_115495.3:p.Val5427Met
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.9931
	Allele frequency in gnomAD - East Asian	1.0
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.9722
	SIFT Prediction	tolerated
	dbSNP ID	rs2438378
	ClinVar ID	46285
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	1.0
	Report Inclusion	None
46	Variant (HGVSc)	NM_032119.4:c.18625-7T>C
	Gene Symbol	ADGRV1
	Variant Type	SNV
	Location	5:91153214-91153214
	Exon Numbers	Intronic
	Ref	T
	Alt	C
	Frequency (Read)	1.0 (555/555)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(dbscSNV)
	Inheritance Pattern	Recessive
	Variant Function	splice_polypyrimidine_tract_variant,splice_region_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.925
	Allele frequency in gnomAD - East Asian	0.8949
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.8984
	SIFT Prediction	-
	dbSNP ID	rs7726023
	ClinVar ID	46299
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.862
	Report Inclusion	None
47	Variant (HGVSc)	NM_004183.4:c.109T>C

	Gene Symbol	BEST1
	Variant Type	SNV
	Location	11:61951915-61951915
	Exon Numbers	2/11
	Ref	T
	Alt	C
	Frequency (Read)	0.48972 (405/827)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_004174.1:p.Leu37=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.4877
	Allele frequency in gnomAD - East Asian	0.912
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.6805
	SIFT Prediction	-
	dbSNP ID	rs1800007
	ClinVar ID	99678
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.956
	Report Inclusion	None
48	Variant (HGVSc)	NM_004183.4:c.1410G>A
	Gene Symbol	BEST1
	Variant Type	SNV
	Location	11:61962564-61962564
	Exon Numbers	10/11
	Ref	G
	Alt	A
	Frequency (Read)	0.52189 (441/845)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_004174.1:p.Thr470=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.2263
	Allele frequency in gnomAD - East Asian	0.001907
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.1122
	SIFT Prediction	-
	dbSNP ID	rs149698
	ClinVar ID	193666
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	5.318E-4
	Report Inclusion	None
49	Variant (HGVSc)	NM_004183.4:c.1557C>T
	Gene Symbol	BEST1
	Variant Type	SNV
	Location	11:61962711-61962711
	Exon Numbers	10/11
	Ref	C
	Alt	T
	Frequency (Read)	0.51945 (414/797)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_004174.1:p.Ser519=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.1748
	Allele frequency in gnomAD - East Asian	0.001253
	1000 Genomes Minor Allele	T

	1000 Genomes Minor Allele Frequency	0.1014
	SIFT Prediction	-
	dbSNP ID	rs1800008
	ClinVar ID	99680
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	2.643E-4
	Report Inclusion	None
50	Variant (HGVSc)	NM_004183.4:c.1608T>C
	Gene Symbol	BEST1
	Variant Type	SNV
	Location	11:61962762-61962762
	Exon Numbers	10/11
	Ref	T
	Alt	C
	Frequency (Read)	0.50532 (380/752)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_004174.1:p.Thr536=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.4599
	Allele frequency in gnomAD - East Asian	0.8607
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.5915
	SIFT Prediction	-
	dbSNP ID	rs1800009
	ClinVar ID	99682
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.941
	Report Inclusion	None
51	Variant (HGVSc)	NM_004183.4:c.1739+188T>C
	Gene Symbol	BEST1
	Variant Type	SNV
	Location	11:61963081-61963081
	Exon Numbers	Intronic
	Ref	T
	Alt	C
	Frequency (Read)	0.50248 (405/806)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BP6(ClinVar)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	intron_variant
	Allele frequency in gnomAD - whole population	0.178
	Allele frequency in gnomAD - East Asian	0.003559
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.1044
	SIFT Prediction	-
	dbSNP ID	rs17185413
	ClinVar ID	1277400
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	1.98E-4
	Report Inclusion	None
52	Variant (HGVSc)	NM_022124.6:c.366T>C
	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71511149-71511149
	Exon Numbers	6/70
	Ref	T
	Alt	C
	Frequency (Read)	0.52393 (405/773)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_071407.4:p.Val122=

	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.6614
	Allele frequency in gnomAD - East Asian	0.7783
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.7346
	SIFT Prediction	-
	dbSNP ID	rs3802720
	ClinVar ID	45928
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.742
	Report Inclusion	None
53	Variant (HGVSc)	NM_022124.6:c.1449+130T>C
	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71646747-71646747
	Exon Numbers	Intronic
	Ref	T
	Alt	C
	Frequency (Read)	0.49706 (338/680)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	intron_variant
	Allele frequency in gnomAD - whole population	0.1436
	Allele frequency in gnomAD - East Asian	0.07479
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.1206
	SIFT Prediction	-
	dbSNP ID	rs2305209
	ClinVar ID	45870
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.081
	Report Inclusion	None
54	Variant (HGVSc)	NM_022124.6:c.1487G>A
	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71675149-71675149
	Exon Numbers	15/70
	Ref	G
	Alt	A
	Frequency (Read)	0.52799 (349/661)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_071407.4:p.Ser496Asn
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.2736
	Allele frequency in gnomAD - East Asian	0.2973
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.2524
	SIFT Prediction	tolerated
	dbSNP ID	rs10999947
	ClinVar ID	45875
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.201
	Report Inclusion	None
55	Variant (HGVSc)	NM_022124.6:c.2316T>C

	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71695444-71695444
	Exon Numbers	22/70
	Ref	T
	Alt	C
	Frequency (Read)	0.42779 (391/914)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_071407.4:p.Asn772=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.6284
	Allele frequency in gnomAD - East Asian	0.5034
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.5545
	SIFT Prediction	-
	dbSNP ID	rs3752752
	ClinVar ID	45893
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.456
	Report Inclusion	None
56	Variant (HGVSc)	NM_022124.6:c.2388T>C
	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71695516-71695516
	Exon Numbers	22/70
	Ref	T
	Alt	C
	Frequency (Read)	0.41463 (340/820)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_071407.4:p.Asp796=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.6282
	Allele frequency in gnomAD - East Asian	0.5008
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.5539
	SIFT Prediction	-
	dbSNP ID	rs3752751
	ClinVar ID	45896
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.452
	Report Inclusion	None
57	Variant (HGVSc)	NM_022124.6:c.3009T>C
	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71706952-71706952
	Exon Numbers	26/70
	Ref	T
	Alt	C
	Frequency (Read)	0.48642 (394/810)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_071407.4:p.Ser1003=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.1034
	Allele frequency in gnomAD - East Asian	0.2544
	1000 Genomes Minor Allele	C

	1000 Genomes Minor Allele Frequency	0.2075
	SIFT Prediction	-
	dbSNP ID	rs10823829
	ClinVar ID	44111
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.314
	Report Inclusion	None
58	Variant (HGVSc)	NM_022124.6:c.3369+298C>G
	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71713111-71713111
	Exon Numbers	Intronic
	Ref	C
	Alt	G
	Frequency (Read)	0.43876 (326/743)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BP6(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	intron_variant
	Allele frequency in gnomAD - whole population	0.317
	Allele frequency in gnomAD - East Asian	0.4475
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.4499
	SIFT Prediction	-
	dbSNP ID	rs2394838
	ClinVar ID	680503
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.412
	Report Inclusion	None
59	Variant (HGVSc)	NM_022124.6:c.3369+312C>T
	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71713125-71713125
	Exon Numbers	Intronic
	Ref	C
	Alt	T
	Frequency (Read)	0.45308 (338/746)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BP6(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	intron_variant
	Allele frequency in gnomAD - whole population	0.3148
	Allele frequency in gnomAD - East Asian	0.4459
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.4413
	SIFT Prediction	-
	dbSNP ID	rs2166631
	ClinVar ID	680504
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.413
	Report Inclusion	None
60	Variant (HGVSc)	NM_022124.6:c.4051A>G
	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71732322-71732322
	Exon Numbers	32/70
	Ref	A
	Alt	G
	Frequency (Read)	0.4908 (507/1033)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_071407.4:p.Asn1351Asp

	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.8069
	Allele frequency in gnomAD - East Asian	0.8465
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.7821
	SIFT Prediction	tolerated
	dbSNP ID	rs1227065
	ClinVar ID	45937
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.817
	Report Inclusion	None
61	Variant (HGVSc)	NM_022124.6:c.4310G>A
	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71738598-71738598
	Exon Numbers	35/70
	Ref	G
	Alt	A
	Frequency (Read)	0.46705 (404/865)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_071407.4:p.Arg1437Gln
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PM5(HGMD,ClinVar),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.03607
	Allele frequency in gnomAD - East Asian	0.0875
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.0521
	SIFT Prediction	tolerated
	dbSNP ID	rs56181447
	ClinVar ID	45944
	ClinVar Pathogenicity	Benign/Likely_benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.118
	Report Inclusion	None
62	Variant (HGVSc)	NM_022124.6:c.4723G>A
	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71741799-71741799
	Exon Numbers	38/70
	Ref	G
	Alt	A
	Frequency (Read)	0.46214 (415/898)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_071407.4:p.Ala1575Thr
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.7742
	Allele frequency in gnomAD - East Asian	0.8323
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.7288
	SIFT Prediction	tolerated
	dbSNP ID	rs1227051
	ClinVar ID	45955
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.815
	Report Inclusion	None
63	Variant (HGVSc)	NM_022124.6:c.5023G>A

	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71777857-71777857
	Exon Numbers	39/70
	Ref	G
	Alt	A
	Frequency (Read)	0.50715 (461/909)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_071407.4:p.Val1675Ile
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.1793
	Allele frequency in gnomAD - East Asian	0.1604
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.1322
	SIFT Prediction	tolerated
	dbSNP ID	rs17712523
	ClinVar ID	45960
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.142
	Report Inclusion	None
64	Variant (HGVSc)	NM_022124.6:c.5100C>T
	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71778221-71778221
	Exon Numbers	40/70
	Ref	C
	Alt	T
	Frequency (Read)	0.50911 (391/768)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_071407.4:p.Tyr1700=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.1868
	Allele frequency in gnomAD - East Asian	0.1864
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.1793
	SIFT Prediction	-
	dbSNP ID	rs10762480
	ClinVar ID	45965
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.239
	Report Inclusion	None
65	Variant (HGVSc)	NM_022124.6:c.5411G>A
	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71784329-71784329
	Exon Numbers	42/70
	Ref	G
	Alt	A
	Frequency (Read)	0.52174 (396/759)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_071407.4:p.Arg1804Gln
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.155
	Allele frequency in gnomAD - East Asian	0.2365
	1000 Genomes Minor Allele	A

	1000 Genomes Minor Allele Frequency	0.139
	SIFT Prediction	tolerated
	dbSNP ID	rs3802711
	ClinVar ID	45978
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.294
	Report Inclusion	None
66	Variant (HGVSc)	NM_022124.6:c.5996C>G
	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71790360-71790360
	Exon Numbers	46/70
	Ref	C
	Alt	G
	Frequency (Read)	0.49035 (432/881)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_071407.4:p.Thr1999Ser
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.4229
	Allele frequency in gnomAD - East Asian	0.1648
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.3452
	SIFT Prediction	tolerated
	dbSNP ID	rs11592462
	ClinVar ID	45997
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.133
	Report Inclusion	None
67	Variant (HGVSc)	NM_022124.6:c.6130G>A
	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71791212-71791212
	Exon Numbers	47/70
	Ref	G
	Alt	A
	Frequency (Read)	0.47732 (463/970)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_071407.4:p.Glu2044Lys
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.3093
	Allele frequency in gnomAD - East Asian	0.5287
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.3327
	SIFT Prediction	tolerated
	dbSNP ID	rs10466026
	ClinVar ID	46002
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.605
	Report Inclusion	None
68	Variant (HGVSc)	NM_022124.6:c.7073G>A
	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71799129-71799129
	Exon Numbers	51/70
	Ref	G
	Alt	A
	Frequency (Read)	0.46009 (317/689)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_071407.4:p.Arg2358Gln

	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.3149
	Allele frequency in gnomAD - East Asian	0.5299
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.3413
	SIFT Prediction	tolerated
	dbSNP ID	rs4747194
	ClinVar ID	46026
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.605
	Report Inclusion	None
69	Variant (HGVSc)	NM_022124.6:c.7139C>T
	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71799195-71799195
	Exon Numbers	51/70
	Ref	C
	Alt	T
	Frequency (Read)	0.48904 (424/867)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_071407.4:p.Pro2380Leu
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(SIFT),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.3081
	Allele frequency in gnomAD - East Asian	0.5296
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.3283
	SIFT Prediction	deleterious
	dbSNP ID	rs4747195
	ClinVar ID	46028
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.605
	Report Inclusion	None
70	Variant (HGVSc)	NM_022124.6:c.7572G>A
	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71802987-71802987
	Exon Numbers	54/70
	Ref	G
	Alt	A
	Frequency (Read)	0.44264 (382/863)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_071407.4:p.Ala2524=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.3106
	Allele frequency in gnomAD - East Asian	0.5268
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.3285
	SIFT Prediction	-
	dbSNP ID	rs10823849
	ClinVar ID	46034
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.606
	Report Inclusion	None
71	Variant (HGVSc)	NM_022124.6:c.9077+8G>A

	Gene Symbol	CDH23
	Variant Type	SNV
	Location	10:71810577-71810577
	Exon Numbers	Intronic
	Ref	G
	Alt	A
	Frequency (Read)	0.5308 (405/763)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(dbscSNV)
	Inheritance Pattern	Recessive
	Variant Function	splice_region_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.1685
	Allele frequency in gnomAD - East Asian	0.2363
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.1486
	SIFT Prediction	-
	dbSNP ID	rs11818398
	ClinVar ID	46068
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.302
	Report Inclusion	None
72	Variant (HGVSc)	NM_033100.4:c.477A>G
	Gene Symbol	CDHR1
	Variant Type	SNV
	Location	10:84200639-84200639
	Exon Numbers	6/17
	Ref	A
	Alt	G
	Frequency (Read)	0.47152 (356/755)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_149091.1:p.Ala159=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.4739
	Allele frequency in gnomAD - East Asian	0.3797
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.516
	SIFT Prediction	-
	dbSNP ID	rs4933975
	ClinVar ID	262215
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.305
	Report Inclusion	None
73	Variant (HGVSc)	NM_033100.4:c.2439T>C
	Gene Symbol	CDHR1
	Variant Type	SNV
	Location	10:84214480-84214480
	Exon Numbers	17/17
	Ref	T
	Alt	C
	Frequency (Read)	0.49097 (408/831)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_149091.1:p.Thr813=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.5321
	Allele frequency in gnomAD - East Asian	0.3927
	1000 Genomes Minor Allele	C

	1000 Genomes Minor Allele Frequency	0.5737
	SIFT Prediction	-
	dbSNP ID	rs3814213
	ClinVar ID	194794
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.311
	Report Inclusion	None
74	Variant (HGVSc)	NM_033100.4:c.*4A>G
	Gene Symbol	CDHR1
	Variant Type	SNV
	Location	10:84214625-84214625
	Exon Numbers	17/17
	Ref	A
	Alt	G
	Frequency (Read)	0.53737 (266/495)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	3_prime_UTR_variant
	Allele frequency in gnomAD - whole population	0.5861
	Allele frequency in gnomAD - East Asian	0.3934
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.6274
	SIFT Prediction	-
	dbSNP ID	rs3814212
	ClinVar ID	194795
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.311
	Report Inclusion	None
75	Variant (HGVSc)	-
	Gene Symbol	CDHR1
	Variant Type	SNV
	Location	10:84219183-84219183
	Exon Numbers	Intronic
	Ref	G
	Alt	A
	Frequency (Read)	0.48636 (321/660)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BP6(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	downstream_gene_variant
	Allele frequency in gnomAD - whole population	0.491
	Allele frequency in gnomAD - East Asian	0.3935
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.5541
	SIFT Prediction	-
	dbSNP ID	rs4244947
	ClinVar ID	1192715
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.311
	Report Inclusion	None
76	Variant (HGVSc)	NM_025114.4:c.3574-9del
	Gene Symbol	CEP290
	Variant Type	Deletion
	Location	12:88089495-88089496
	Exon Numbers	Intronic
	Ref	TA
	Alt	T
	Frequency (Read)	0.98387 (244/248)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	-

	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	splice_polypyrimidine_tract_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.9308
	Allele frequency in gnomAD - East Asian	0.9904
	1000 Genomes Minor Allele	-
	1000 Genomes Minor Allele Frequency	0.8736
	SIFT Prediction	-
	dbSNP ID	rs10717563
	ClinVar ID	96170
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.997
	Report Inclusion	None
77	Variant (HGVSc)	NM_025114.4:c.2268A>G
	Gene Symbol	CEP290
	Variant Type	SNV
	Location	12:88111301-88111301
	Exon Numbers	22/54
	Ref	T
	Alt	C
	Frequency (Read)	1.0 (349/349)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_079390.3:p.Ser756=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.8938
	Allele frequency in gnomAD - East Asian	0.9755
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.7646
	SIFT Prediction	-
	dbSNP ID	rs2468255
	ClinVar ID	126249
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.937
	Report Inclusion	None
78	Variant (HGVSc)	NM_025114.4:c.2055T>C
	Gene Symbol	CEP290
	Variant Type	SNV
	Location	12:88111856-88111856
	Exon Numbers	21/54
	Ref	A
	Alt	G
	Frequency (Read)	0.51323 (97/189)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_079390.3:p.Ala685=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	splice_region_variant,synonymous_variant
	Allele frequency in gnomAD - whole population	0.1382
	Allele frequency in gnomAD - East Asian	8.134E-5
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.0689
	SIFT Prediction	-
	dbSNP ID	rs45465996
	ClinVar ID	96168
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
79	Variant (HGVSc)	NM_001030311.3:c.1584C>T

	Gene Symbol	CERKL
	Variant Type	SNV
	Location	2:181539124-181539124
	Exon Numbers	13/14
	Ref	G
	Alt	A
	Frequency (Read)	1.0 (438/438)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_001025482.1:p.Asp528=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.9979
	Allele frequency in gnomAD - East Asian	1.0
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.9912
	SIFT Prediction	-
	dbSNP ID	rs10180793
	ClinVar ID	166844
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	1.0
	Report Inclusion	None
80	Variant (HGVSc)	NM_001030311.3:c.156C>T
	Gene Symbol	CERKL
	Variant Type	SNV
	Location	2:181656851-181656851
	Exon Numbers	1/14
	Ref	G
	Alt	A
	Frequency (Read)	0.47279 (443/937)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_001025482.1:p.Phe52=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.5276
	Allele frequency in gnomAD - East Asian	0.7109
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.5074
	SIFT Prediction	-
	dbSNP ID	rs1473295
	ClinVar ID	166849
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.742
	Report Inclusion	None
81	Variant (HGVSc)	NM_004928.3:c.449C>T
	Gene Symbol	CFAP410
	Variant Type	SNV
	Location	21:44331939-44331939
	Exon Numbers	5/7
	Ref	G
	Alt	A
	Frequency (Read)	0.48362 (443/916)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_004919.1:p.Thr150Ile
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.02902
	Allele frequency in gnomAD - East Asian	0.05725
	1000 Genomes Minor Allele	A

	1000 Genomes Minor Allele Frequency	0.0373
	SIFT Prediction	tolerated
	dbSNP ID	rs2277809
	ClinVar ID	259587
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.068
	Report Inclusion	None
82	Variant (HGVSc)	NM_004928.3:c.33G>A
	Gene Symbol	CFAP410
	Variant Type	SNV
	Location	21:44339162-44339162
	Exon Numbers	1/7
	Ref	C
	Alt	T
	Frequency (Read)	0.44395 (301/678)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_004919.1:p.Arg11=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.2403
	Allele frequency in gnomAD - East Asian	0.1299
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.3261
	SIFT Prediction	-
	dbSNP ID	rs11870
	ClinVar ID	259586
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.166
	Report Inclusion	None
83	Variant (HGVSc)	NM_000087.5:c.-15+10300C>T
	Gene Symbol	CNGA1
	Variant Type	SNV
	Location	4:47971093-47971093
	Exon Numbers	Intronic
	Ref	G
	Alt	A
	Frequency (Read)	1.0 (240/240)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BP6(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	intron_variant
	Allele frequency in gnomAD - whole population	0.5657
	Allele frequency in gnomAD - East Asian	0.2994
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.4593
	SIFT Prediction	-
	dbSNP ID	rs7693648
	ClinVar ID	1217361
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.266
	Report Inclusion	None
84	Variant (HGVSc)	NM_001298.3:c.72T>C
	Gene Symbol	CNGA3
	Variant Type	SNV
	Location	2:98370047-98370047
	Exon Numbers	2/8
	Ref	T
	Alt	C
	Frequency (Read)	0.48667 (365/750)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_001289.1:p.Asp24=

	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.1327
	Allele frequency in gnomAD - East Asian	0.006253
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.0899
	SIFT Prediction	-
	dbSNP ID	rs6727412
	ClinVar ID	257964
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.002251
	Report Inclusion	None
85	Variant (HGVSc)	NM_001297.5:c.3462+7T>C
	Gene Symbol	CNGB1
	Variant Type	SNV
	Location	16:57887848-57887848
	Exon Numbers	Intronic
	Ref	A
	Alt	G
	Frequency (Read)	0.48758 (314/644)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(dbscSNV)
	Inheritance Pattern	Recessive
	Variant Function	splice_region_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.3071
	Allele frequency in gnomAD - East Asian	0.3422
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.3702
	SIFT Prediction	-
	dbSNP ID	rs11076207
	ClinVar ID	320059
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.234
	Report Inclusion	None
86	Variant (HGVSc)	NM_001297.5:c.2664C>G
	Gene Symbol	CNGB1
	Variant Type	SNV
	Location	16:57903952-57903952
	Exon Numbers	27/33
	Ref	G
	Alt	C
	Frequency (Read)	0.50357 (353/701)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_001288.3:p.Ala888=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.7653
	Allele frequency in gnomAD - East Asian	0.918
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.7049
	SIFT Prediction	-
	dbSNP ID	rs413562
	ClinVar ID	166894
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.952
	Report Inclusion	None
87	Variant (HGVSc)	NM_001297.5:c.2635-10C>T

	Gene Symbol	CNGB1
	Variant Type	SNV
	Location	16:57903991-57903991
	Exon Numbers	Intronic
	Ref	G
	Alt	A
	Frequency (Read)	0.4912 (279/568)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	splice_polypyrimidine_tract_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.812
	Allele frequency in gnomAD - East Asian	0.6565
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.7484
	SIFT Prediction	-
	dbSNP ID	rs437920
	ClinVar ID	166895
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.636
	Report Inclusion	None
88	Variant (HGVSc)	NM_001297.5:c.327C>T
	Gene Symbol	CNGB1
	Variant Type	SNV
	Location	16:57963028-57963028
	Exon Numbers	5/33
	Ref	G
	Alt	A
	Frequency (Read)	0.49537 (428/864)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_001288.3:p.Gly109=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.4306
	Allele frequency in gnomAD - East Asian	0.3309
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.3141
	SIFT Prediction	-
	dbSNP ID	rs17821448
	ClinVar ID	320109
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.345
	Report Inclusion	None
89	Variant (HGVSc)	NM_001297.5:c.299G>A
	Gene Symbol	CNGB1
	Variant Type	SNV
	Location	16:57963056-57963056
	Exon Numbers	5/33
	Ref	C
	Alt	T
	Frequency (Read)	0.99875 (798/799)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_001288.3:p.Arg100His
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(SIFT),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.7822
	Allele frequency in gnomAD - East Asian	0.844
	1000 Genomes Minor Allele	T

	1000 Genomes Minor Allele Frequency	0.752
	SIFT Prediction	deleterious_low_confidence
	dbSNP ID	rs13336595
	ClinVar ID	320110
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.837
	Report Inclusion	None
90	Variant (HGVSc)	NM_019098.5:c.892A>C
	Gene Symbol	CNGB3
	Variant Type	SNV
	Location	8:86654023-86654023
	Exon Numbers	7/18
	Ref	T
	Alt	G
	Frequency (Read)	1.0 (314/314)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_061971.3:p.Thr298Pro
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.6607
	Allele frequency in gnomAD - East Asian	0.7959
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.6797
	SIFT Prediction	tolerated
	dbSNP ID	rs4961206
	ClinVar ID	95929
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.798
	Report Inclusion	None
91	Variant (HGVSc)	NM_019098.5:c.702T>G
	Gene Symbol	CNGB3
	Variant Type	SNV
	Location	8:86667075-86667075
	Exon Numbers	6/18
	Ref	A
	Alt	C
	Frequency (Read)	0.99848 (656/657)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_061971.3:p.Cys234Trp
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.8925
	Allele frequency in gnomAD - East Asian	0.9998
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.9575
	SIFT Prediction	tolerated
	dbSNP ID	rs6471482
	ClinVar ID	261090
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	1.0
	Report Inclusion	None
92	Variant (HGVSc)	NM_019098.5:c.80A>G
	Gene Symbol	CNGB3
	Variant Type	SNV
	Location	8:86743548-86743548
	Exon Numbers	1/18
	Ref	T
	Alt	C
	Frequency (Read)	0.52196 (309/592)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_061971.3:p.Asn27Ser

	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.01846
	Allele frequency in gnomAD - East Asian	5.437E-5
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.0066
	SIFT Prediction	tolerated
	dbSNP ID	rs35807406
	ClinVar ID	100588
	ClinVar Pathogenicity	Benign/Likely_benign
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
93	Variant (HGVSc)	NM_201253.3:c.1410A>G
	Gene Symbol	CRB1
	Variant Type	SNV
	Location	1:197421238-197421238
	Exon Numbers	6/12
	Ref	A
	Alt	G
	Frequency (Read)	1.0 (705/705)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_957705.1:p.Leu470=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.9818
	Allele frequency in gnomAD - East Asian	0.9998
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.9197
	SIFT Prediction	-
	dbSNP ID	rs3902057
	ClinVar ID	166956
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	1.0
	Report Inclusion	None
94	Variant (HGVSc)	NM_207352.4:c.64C>G
	Gene Symbol	CYP4V2
	Variant Type	SNV
	Location	4:186191887-186191887
	Exon Numbers	1/11
	Ref	C
	Alt	G
	Frequency (Read)	0.50496 (509/1008)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_997235.3:p.Leu22Val
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PM5(HGMD),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.4436
	Allele frequency in gnomAD - East Asian	0.2721
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.4097
	SIFT Prediction	tolerated
	dbSNP ID	rs1055138
	ClinVar ID	39267
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.285
	Report Inclusion	None
95	Variant (HGVSc)	NM_207352.4:c.775C>A

	Gene Symbol	CYP4V2
	Variant Type	SNV
	Location	4:186199057-186199057
	Exon Numbers	6/11
	Ref	C
	Alt	A
	Frequency (Read)	1.0 (392/392)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_997235.3:p.Gln259Lys
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.5734
	Allele frequency in gnomAD - East Asian	0.3935
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.5593
	SIFT Prediction	tolerated
	dbSNP ID	rs13146272
	ClinVar ID	263297
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.396
	Report Inclusion	None
96	Variant (HGVSc)	NM_207352.4:c.802-7C>T
	Gene Symbol	CYP4V2
	Variant Type	SNV
	Location	4:186201150-186201150
	Exon Numbers	Intronic
	Ref	C
	Alt	T
	Frequency (Read)	1.0 (413/413)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(dbscSNV)
	Inheritance Pattern	Recessive
	Variant Function	splice_polypyrimidine_tract_variant,splice_region_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.3338
	Allele frequency in gnomAD - East Asian	0.3334
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.2923
	SIFT Prediction	-
	dbSNP ID	rs3817184
	ClinVar ID	166975
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.332
	Report Inclusion	None
97	Variant (HGVSc)	NM_207352.4:c.810T>G
	Gene Symbol	CYP4V2
	Variant Type	SNV
	Location	4:186201165-186201165
	Exon Numbers	7/11
	Ref	T
	Alt	G
	Frequency (Read)	0.99796 (490/491)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_997235.3:p.Ala270=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.5882
	Allele frequency in gnomAD - East Asian	0.3923
	1000 Genomes Minor Allele	G

	1000 Genomes Minor Allele Frequency	0.5775
	SIFT Prediction	-
	dbSNP ID	rs3736455
	ClinVar ID	166976
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.391
	Report Inclusion	None
98	Variant (HGVSc)	NM_207352.4:c.823G>A
	Gene Symbol	CYP4V2
	Variant Type	SNV
	Location	4:186201178-186201178
	Exon Numbers	7/11
	Ref	G
	Alt	A
	Frequency (Read)	1.0 (562/562)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_997235.3:p.Glu275Lys
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BS1(gnomAD),BS3(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.02622
	Allele frequency in gnomAD - East Asian	1.087E-4
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.0336
	SIFT Prediction	tolerated
	dbSNP ID	rs34745240
	ClinVar ID	263298
	ClinVar Pathogenicity	Benign/Likely_benign
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
99	Variant (HGVSc)	NM_024887.4:c.757G>A
	Gene Symbol	DHDDS
	Variant Type	SNV
	Location	1:26460136-26460136
	Exon Numbers	8/9
	Ref	G
	Alt	A
	Frequency (Read)	0.47341 (365/771)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_079163.2:p.Val253Met
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP2(ClinVar),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.307
	Allele frequency in gnomAD - East Asian	0.7259
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.4553
	SIFT Prediction	tolerated
	dbSNP ID	rs3816539
	ClinVar ID	297075
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.773
	Report Inclusion	None
100	Variant (HGVSc)	NM_178454.6:c.601-9A>G
	Gene Symbol	DRAM2
	Variant Type	SNV
	Location	1:111118906-111118906
	Exon Numbers	Intronic
	Ref	T
	Alt	C
	Frequency (Read)	0.48493 (177/365)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-

	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	splice_polypyrimidine_tract_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.2226
	Allele frequency in gnomAD - East Asian	0.2399
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.3285
	SIFT Prediction	-
	dbSNP ID	rs325917
	ClinVar ID	677168
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.296
	Report Inclusion	None
101	Variant (HGVSc)	NM_001292009.2:c.6977G>A
	Gene Symbol	EYS
	Variant Type	SNV
	Location	6:63984461-63984461
	Exon Numbers	35/44
	Ref	C
	Alt	T
	Frequency (Read)	0.53099 (317/597)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_001278938.1:p.Arg2326Gln
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PM5(ClinVar),PP3(SIFT),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(BLOSUM),BP6(Benign Variants from PMID: 31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.3354
	Allele frequency in gnomAD - East Asian	0.3525
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.3496
	SIFT Prediction	deleterious
	dbSNP ID	rs4710457
	ClinVar ID	93620
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.349
	Report Inclusion	None
102	Variant (HGVSc)	NM_001292009.2:c.6079-4_6079-3del
	Gene Symbol	EYS
	Variant Type	Deletion
	Location	6:64307084-64307086
	Exon Numbers	Intronic
	Ref	TGA
	Alt	T
	Frequency (Read)	0.49333 (148/300)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	splice_polypyrimidine_tract_variant,splice_region_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.5506
	Allele frequency in gnomAD - East Asian	0.5032
	1000 Genomes Minor Allele	-
	1000 Genomes Minor Allele Frequency	0.6741
	SIFT Prediction	-
	dbSNP ID	rs35395170
	ClinVar ID	196616
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.51
	Report Inclusion	None

103	Variant (HGVSc)	NM_001292009.2:c.5705A>T
	Gene Symbol	EYS
	Variant Type	SNV
	Location	6:64439292-64439292
	Exon Numbers	27/44
	Ref	T
	Alt	A
	Frequency (Read)	0.46 (184/400)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_001278938.1:p.Asn1902Ile
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.3047
	Allele frequency in gnomAD - East Asian	0.4896
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.2913
	SIFT Prediction	tolerated
	dbSNP ID	rs9353806
	ClinVar ID	137256
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.444
	Report Inclusion	None
104	Variant (HGVSc)	NM_001292009.2:c.3444-5C>T
	Gene Symbol	EYS
	Variant Type	SNV
	Location	6:64626250-64626250
	Exon Numbers	Intronic
	Ref	G
	Alt	A
	Frequency (Read)	0.44643 (100/224)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(dbscSNV),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	splice_polypyrimidine_tract_variant,splice_region_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.2252
	Allele frequency in gnomAD - East Asian	0.3385
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.1955
	SIFT Prediction	-
	dbSNP ID	rs9445051
	ClinVar ID	357717
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.303
	Report Inclusion	None
105	Variant (HGVSc)	NM_001292009.2:c.2555T>C
	Gene Symbol	EYS
	Variant Type	SNV
	Location	6:64912570-64912570
	Exon Numbers	16/44
	Ref	A
	Alt	G
	Frequency (Read)	0.46196 (255/552)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_001278938.1:p.Leu852Pro
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.593
	Allele frequency in gnomAD - East Asian	0.4017

	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.6464
	SIFT Prediction	tolerated
	dbSNP ID	rs9294631
	ClinVar ID	93605
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.458
	Report Inclusion	None
106	Variant (HGVSc)	NM_001292009.2:c.1891G>A
	Gene Symbol	EYS
	Variant Type	SNV
	Location	6:65295995-65295995
	Exon Numbers	12/44
	Ref	C
	Alt	T
	Frequency (Read)	1.0 (548/548)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_001278938.1:p.Gly631Ser
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.6246
	Allele frequency in gnomAD - East Asian	0.917
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.595
	SIFT Prediction	tolerated
	dbSNP ID	rs9342464
	ClinVar ID	137267
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.893
	Report Inclusion	None
107	Variant (HGVSc)	NM_001292009.2:c.1809C>T
	Gene Symbol	EYS
	Variant Type	SNV
	Location	6:65296077-65296077
	Exon Numbers	12/44
	Ref	G
	Alt	A
	Frequency (Read)	1.0 (358/358)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_001278938.1:p.Val603=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.625
	Allele frequency in gnomAD - East Asian	0.9174
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.5948
	SIFT Prediction	-
	dbSNP ID	rs9345601
	ClinVar ID	137266
	ClinVar Pathogenicity	Benign/Likely_benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.894
	Report Inclusion	None
108	Variant (HGVSc)	NM_001292009.2:c.1146T>C
	Gene Symbol	EYS
	Variant Type	SNV
	Location	6:65402516-65402516
	Exon Numbers	7/44
	Ref	A
	Alt	G
	Frequency (Read)	0.45588 (124/272)
	Zygosity	Heterozygous

	Amino Acid Change (HGVSp)	NP_001278938.1:p.Asn382=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.5195
	Allele frequency in gnomAD - East Asian	0.443
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.506
	SIFT Prediction	-
	dbSNP ID	rs974110
	ClinVar ID	137262
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.446
	Report Inclusion	None
109	Variant (HGVSc)	NM_001292009.2:c.359C>T
	Gene Symbol	EYS
	Variant Type	SNV
	Location	6:65495052-65495052
	Exon Numbers	4/44
	Ref	G
	Alt	A
	Frequency (Read)	0.49912 (284/569)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_001278938.1:p.Thr120Met
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.2086
	Allele frequency in gnomAD - East Asian	0.2468
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.1703
	SIFT Prediction	tolerated_low_confidence
	dbSNP ID	rs12193967
	ClinVar ID	137261
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.255
	Report Inclusion	None
110	Variant (HGVSc)	NM_001201543.2:c.1212T>C
	Gene Symbol	FAM161A
	Variant Type	SNV
	Location	2:61839792-61839792
	Exon Numbers	3/7
	Ref	A
	Alt	G
	Frequency (Read)	1.0 (727/727)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_001188472.1:p.Cys404=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.9756
	Allele frequency in gnomAD - East Asian	0.9964
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.9796
	SIFT Prediction	-
	dbSNP ID	rs4672457
	ClinVar ID	96217
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.994
	Report Inclusion	None

111	Variant (HGVSc)	NM_001201543.2:c.706A>G
	Gene Symbol	FAM161A
	Variant Type	SNV
	Location	2:61840298-61840298
	Exon Numbers	3/7
	Ref	T
	Alt	C
	Frequency (Read)	0.52802 (358/678)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_001188472.1:p.Ile236Val
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.1836
	Allele frequency in gnomAD - East Asian	0.1053
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.1164
	SIFT Prediction	tolerated
	dbSNP ID	rs17513722
	ClinVar ID	336742
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.077
	Report Inclusion	None
112	Variant (HGVSc)	NM_001201543.2:c.165T>G
	Gene Symbol	FAM161A
	Variant Type	SNV
	Location	2:61853877-61853877
	Exon Numbers	1/7
	Ref	A
	Alt	C
	Frequency (Read)	1.0 (867/867)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_001188472.1:p.Ala55=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.9756
	Allele frequency in gnomAD - East Asian	0.9967
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.9794
	SIFT Prediction	-
	dbSNP ID	rs4270331
	ClinVar ID	193466
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.994
	Report Inclusion	None
113	Variant (HGVSc)	NM_000180.4:c.61T>C
	Gene Symbol	GUCY2D
	Variant Type	SNV
	Location	17:8003108-8003108
	Exon Numbers	2/20
	Ref	T
	Alt	C
	Frequency (Read)	0.49822 (419/841)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000171.1:p.Trp21Arg
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP2(ClinVar),PP3(BLOSUM),BS1(gnomAD),BS3(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.04798
	Allele frequency in gnomAD - East Asian	0.02253

	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.1226
	SIFT Prediction	tolerated_low_confidence
	dbSNP ID	rs9905402
	ClinVar ID	92581
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.002747
	Report Inclusion	None
114	Variant (HGVSc)	NM_000180.4:c.154G>T
	Gene Symbol	GUCY2D
	Variant Type	SNV
	Location	17:8003201-8003201
	Exon Numbers	2/20
	Ref	G
	Alt	T
	Frequency (Read)	0.5286 (499/944)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000171.1:p.Ala52Ser
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP2(ClinVar),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.4159
	Allele frequency in gnomAD - East Asian	0.6953
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.4159
	SIFT Prediction	tolerated
	dbSNP ID	rs61749665
	ClinVar ID	9353
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.698
	Report Inclusion	None
115	Variant (HGVSc)	NM_000180.4:c.2345T>A
	Gene Symbol	GUCY2D
	Variant Type	SNV
	Location	17:8013961-8013961
	Exon Numbers	12/20
	Ref	T
	Alt	A
	Frequency (Read)	0.51165 (461/901)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000171.1:p.Leu782His
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP2(ClinVar),PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.1228
	Allele frequency in gnomAD - East Asian	7.068E-4
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.1713
	SIFT Prediction	tolerated
	dbSNP ID	rs8069344
	ClinVar ID	255475
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	1.328E-4
	Report Inclusion	None
116	Variant (HGVSc)	NM_006899.5:c.118-3G>C
	Gene Symbol	IDH3B
	Variant Type	SNV
	Location	20:2663761-2663761
	Exon Numbers	Intronic
	Ref	C
	Alt	G
	Frequency (Read)	1.0 (751/751)
	Zygosity	Homozygous

	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	splice_polypyrimidine_tract_variant,splice_region_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.6896
	Allele frequency in gnomAD - East Asian	0.6111
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.613
	SIFT Prediction	-
	dbSNP ID	rs2073193
	ClinVar ID	338028
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.568
	Report Inclusion	None
117	Variant (HGVSc)	NM_000883.4:c.1575G>A
	Gene Symbol	IMPDH1
	Variant Type	SNV
	Location	7:128394575-128394575
	Exon Numbers	15/17
	Ref	C
	Alt	T
	Frequency (Read)	0.4649 (351/755)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000874.2:p.Ala525=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.2715
	Allele frequency in gnomAD - East Asian	0.3682
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.2776
	SIFT Prediction	-
	dbSNP ID	rs2228075
	ClinVar ID	358873
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.346
	Report Inclusion	None
118	Variant (HGVSc)	NM_016247.4:c.3381C>T
	Gene Symbol	IMPG2
	Variant Type	SNV
	Location	3:101230998-101230998
	Exon Numbers	16/19
	Ref	G
	Alt	A
	Frequency (Read)	0.4901 (297/606)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_057331.2:p.Leu1127=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.7909
	Allele frequency in gnomAD - East Asian	0.8145
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.8129
	SIFT Prediction	-
	dbSNP ID	rs348867
	ClinVar ID	342337
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.755
	Report Inclusion	None

119	Variant (HGVSc)	NM_133497.4:c.183C>G
	Gene Symbol	KCNV2
	Variant Type	SNV
	Location	9:2717922-2717922
	Exon Numbers	1/2
	Ref	C
	Alt	G
	Frequency (Read)	0.49948 (478/957)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_598004.1:p.Gly61=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.6181
	Allele frequency in gnomAD - East Asian	0.7809
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.6603
	SIFT Prediction	-
	dbSNP ID	rs10967705
	ClinVar ID	96357
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.839
	Report Inclusion	None
120	Variant (HGVSc)	NM_133497.4:c.795C>G
	Gene Symbol	KCNV2
	Variant Type	SNV
	Location	9:2718534-2718534
	Exon Numbers	1/2
	Ref	C
	Alt	G
	Frequency (Read)	0.47128 (402/853)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_598004.1:p.Ala265=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.4839
	Allele frequency in gnomAD - East Asian	0.7171
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.48
	SIFT Prediction	-
	dbSNP ID	rs12237048
	ClinVar ID	96362
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.75
	Report Inclusion	None
121	Variant (HGVSc)	NM_001242957.3:c.1905T>C
	Gene Symbol	MAK
	Variant Type	SNV
	Location	6:10764494-10764494
	Exon Numbers	15/15
	Ref	A
	Alt	G
	Frequency (Read)	0.51235 (332/648)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_001229886.1:p.His635=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.8135
	Allele frequency in gnomAD - East Asian	0.9249

	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.8656
	SIFT Prediction	-
	dbSNP ID	rs126405
	ClinVar ID	354783
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.919
	Report Inclusion	None
122	Variant (HGVSc)	NM_006343.3:c.1397G>A
	Gene Symbol	MERTK
	Variant Type	SNV
	Location	2:111994351-111994351
	Exon Numbers	9/19
	Ref	G
	Alt	A
	Frequency (Read)	0.47089 (364/773)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_006334.2:p.Arg466Lys
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.5975
	Allele frequency in gnomAD - East Asian	0.2498
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.5615
	SIFT Prediction	tolerated
	dbSNP ID	rs7604639
	ClinVar ID	330754
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.232
	Report Inclusion	None
123	Variant (HGVSc)	NM_006343.3:c.1451-72G>T
	Gene Symbol	MERTK
	Variant Type	SNV
	Location	2:111997251-111997251
	Exon Numbers	Intronic
	Ref	G
	Alt	T
	Frequency (Read)	0.45814 (197/430)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD)
	Inheritance Pattern	Recessive
	Variant Function	intron_variant
	Allele frequency in gnomAD - whole population	0.323
	Allele frequency in gnomAD - East Asian	0.1098
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.3039
	SIFT Prediction	-
	dbSNP ID	rs3811632
	ClinVar ID	-
	ClinVar Pathogenicity	-
	HGMD	-
	GEM-J WGA Allele Frequency	0.121
	Report Inclusion	None
124	Variant (HGVSc)	NM_006343.3:c.1552A>G
	Gene Symbol	MERTK
	Variant Type	SNV
	Location	2:111997424-111997424
	Exon Numbers	10/19
	Ref	A
	Alt	G
	Frequency (Read)	0.5 (338/676)
	Zygosity	Heterozygous

	Amino Acid Change (HGVSp)	NP_006334.2:p.Ile518Val
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.5976
	Allele frequency in gnomAD - East Asian	0.2498
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.5615
	SIFT Prediction	tolerated
	dbSNP ID	rs2230515
	ClinVar ID	330759
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.232
	Report Inclusion	None
125	Variant (HGVSc)	NM_006343.3:c.1881A>G
	Gene Symbol	MERTK
	Variant Type	SNV
	Location	2:112008396-112008396
	Exon Numbers	14/19
	Ref	A
	Alt	G
	Frequency (Read)	0.47841 (277/579)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_006334.2:p.Ser627=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.5853
	Allele frequency in gnomAD - East Asian	0.249
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.5152
	SIFT Prediction	-
	dbSNP ID	rs1131244
	ClinVar ID	330762
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.232
	Report Inclusion	None
126	Variant (HGVSc)	NM_000260.4:c.3503+12_3503+33del
	Gene Symbol	MYO7A
	Variant Type	Deletion
	Location	11:77184725-77184747
	Exon Numbers	Intronic
	Ref	GGGAGGCGGGGACACCAGGGCCT
	Alt	G
	Frequency (Read)	0.42934 (319/743)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	intron_variant
	Allele frequency in gnomAD - whole population	0.3798
	Allele frequency in gnomAD - East Asian	0.2709
	1000 Genomes Minor Allele	-
	1000 Genomes Minor Allele Frequency	-
	SIFT Prediction	-
	dbSNP ID	rs111033223
	ClinVar ID	43207
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.264
	Report Inclusion	None

127	Variant (HGVSc)	NM_000260.4:c.4996A>T
	Gene Symbol	MYO7A
	Variant Type	SNV
	Location	11:77201591-77201591
	Exon Numbers	36/49
	Ref	A
	Alt	T
	Frequency (Read)	0.53571 (450/840)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000251.3:p.Ser1666Cys
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.55
	Allele frequency in gnomAD - East Asian	0.488
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.5517
	SIFT Prediction	tolerated
	dbSNP ID	rs2276288
	ClinVar ID	43269
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.457
	Report Inclusion	None
128	Variant (HGVSc)	NM_000260.4:c.5715A>G
	Gene Symbol	MYO7A
	Variant Type	SNV
	Location	11:77206175-77206175
	Exon Numbers	41/49
	Ref	A
	Alt	G
	Frequency (Read)	0.52081 (388/745)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000251.3:p.Lys1905=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.5163
	Allele frequency in gnomAD - East Asian	0.509
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.5549
	SIFT Prediction	-
	dbSNP ID	rs2276293
	ClinVar ID	43296
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.458
	Report Inclusion	None
129	Variant (HGVSc)	NM_000260.4:c.5857-7A>T
	Gene Symbol	MYO7A
	Variant Type	SNV
	Location	11:77208423-77208423
	Exon Numbers	Intronic
	Ref	A
	Alt	T
	Frequency (Read)	0.47047 (239/508)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(dbscSNV)
	Inheritance Pattern	Recessive
	Variant Function	splice_polypyrimidine_tract_variant,splice_region_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.5621
	Allele frequency in gnomAD - East Asian	0.5047

	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.513
	SIFT Prediction	-
	dbSNP ID	rs1320703
	ClinVar ID	43303
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.457
	Report Inclusion	None
130	Variant (HGVSc)	NM_000260.4:c.5860C>A
	Gene Symbol	MYO7A
	Variant Type	SNV
	Location	11:77208433-77208433
	Exon Numbers	43/49
	Ref	C
	Alt	A
	Frequency (Read)	0.48905 (268/548)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000251.3:p.Leu1954Ile
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.4738
	Allele frequency in gnomAD - East Asian	0.3549
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.499
	SIFT Prediction	tolerated
	dbSNP ID	rs948962
	ClinVar ID	43304
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.295
	Report Inclusion	None
131	Variant (HGVSc)	NM_000260.4:c.6063G>A
	Gene Symbol	MYO7A
	Variant Type	SNV
	Location	11:77211163-77211163
	Exon Numbers	45/49
	Ref	G
	Alt	A
	Frequency (Read)	0.54068 (412/762)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000251.3:p.Lys2021=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.00673
	Allele frequency in gnomAD - East Asian	0.0
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.0309
	SIFT Prediction	-
	dbSNP ID	rs111033209
	ClinVar ID	43317
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	1.323E-4
	Report Inclusion	None
132	Variant (HGVSc)	NM_000283.4:c.958G>A
	Gene Symbol	PDE6B
	Variant Type	SNV
	Location	4:654854-654854
	Exon Numbers	6/22
	Ref	G
	Alt	A
	Frequency (Read)	1.0 (585/585)
	Zygosity	Homozygous

	Amino Acid Change (HGVSp)	NP_000274.3:p.Val320Ile
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.9998
	Allele frequency in gnomAD - East Asian	1.0
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.999
	SIFT Prediction	tolerated
	dbSNP ID	rs10902758
	ClinVar ID	167436
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	1.0
	Report Inclusion	None
133	Variant (HGVSc)	NM_006204.4:c.252G>A
	Gene Symbol	PDE6C
	Variant Type	SNV
	Location	10:93612977-93612977
	Exon Numbers	1/22
	Ref	G
	Alt	A
	Frequency (Read)	0.49032 (380/775)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_006195.3:p.Leu84=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.162
	Allele frequency in gnomAD - East Asian	0.1114
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.1643
	SIFT Prediction	-
	dbSNP ID	rs1131978
	ClinVar ID	95346
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.084
	Report Inclusion	None
134	Variant (HGVSc)	NM_006204.4:c.282C>T
	Gene Symbol	PDE6C
	Variant Type	SNV
	Location	10:93613007-93613007
	Exon Numbers	1/22
	Ref	C
	Alt	T
	Frequency (Read)	0.49486 (385/778)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_006195.3:p.Asp94=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.07892
	Allele frequency in gnomAD - East Asian	3.263E-4
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.0429
	SIFT Prediction	-
	dbSNP ID	rs12781149
	ClinVar ID	95347
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	6.609E-4
	Report Inclusion	None

135	Variant (HGVSc)	NM_006204.4:c.1270-7A>G
	Gene Symbol	PDE6C
	Variant Type	SNV
	Location	10:93635490-93635490
	Exon Numbers	Intronic
	Ref	A
	Alt	G
	Frequency (Read)	1.0 (280/280)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(dbscSNV)
	Inheritance Pattern	Recessive
	Variant Function	splice_region_variant,splice_polypyrimidine_tract_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.7329
	Allele frequency in gnomAD - East Asian	0.6592
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.7288
	SIFT Prediction	-
	dbSNP ID	rs616522
	ClinVar ID	259940
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.643
	Report Inclusion	None
136	Variant (HGVSc)	NM_006204.4:c.1935+10C>A
	Gene Symbol	PDE6C
	Variant Type	SNV
	Location	10:93646057-93646057
	Exon Numbers	Intronic
	Ref	C
	Alt	A
	Frequency (Read)	0.48871 (303/620)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	intron_variant
	Allele frequency in gnomAD - whole population	0.4568
	Allele frequency in gnomAD - East Asian	0.4063
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.4133
	SIFT Prediction	-
	dbSNP ID	rs1409332
	ClinVar ID	259943
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.447
	Report Inclusion	None
137	Variant (HGVSc)	NM_172240.3:c.453-12_453-3del
	Gene Symbol	POC1B
	Variant Type	Deletion
	Location	12:89472277-89472287
	Exon Numbers	Intronic
	Ref	TAGAAAGAAGA
	Alt	T
	Frequency (Read)	0.40476 (68/168)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	splice_polypyrimidine_tract_variant,splice_region_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.4817
	Allele frequency in gnomAD - East Asian	0.5622

	1000 Genomes Minor Allele	-
	1000 Genomes Minor Allele Frequency	0.5399
	SIFT Prediction	-
	dbSNP ID	rs59139895
	ClinVar ID	677291
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.699
	Report Inclusion	None
138	Variant (HGVSc)	NM_006017.3:c.2374-6T>C
	Gene Symbol	PROM1
	Variant Type	SNV
	Location	4:15980543-15980543
	Exon Numbers	Intronic
	Ref	A
	Alt	G
	Frequency (Read)	0.51039 (172/337)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(dbscSNV)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	splice_polypyrimidine_tract_variant,splice_region_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.3905
	Allele frequency in gnomAD - East Asian	0.1737
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.4469
	SIFT Prediction	-
	dbSNP ID	rs6449209
	ClinVar ID	95334
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.225
	Report Inclusion	None
139	Variant (HGVSc)	NM_006017.3:c.303+6G>A
	Gene Symbol	PROM1
	Variant Type	SNV
	Location	4:16035729-16035729
	Exon Numbers	Intronic
	Ref	C
	Alt	T
	Frequency (Read)	0.49471 (234/473)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	splice_donor_region_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.4489
	Allele frequency in gnomAD - East Asian	0.1488
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.2961
	SIFT Prediction	-
	dbSNP ID	rs2078622
	ClinVar ID	259906
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.219
	Report Inclusion	None
140	Variant (HGVSc)	NM_000322.5:c.1013A>G
	Gene Symbol	PRPH2
	Variant Type	SNV
	Location	6:42698323-42698323
	Exon Numbers	3/3
	Ref	T
	Alt	C
	Frequency (Read)	0.51499 (378/734)
	Zygosity	Heterozygous

	Amino Acid Change (HGVSp)	NP_000313.2:p.Asp338Gly
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP2(ClinVar),PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.7761
	Allele frequency in gnomAD - East Asian	0.8616
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.7574
	SIFT Prediction	tolerated
	dbSNP ID	rs434102
	ClinVar ID	138906
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.841
	Report Inclusion	None
141	Variant (HGVSc)	NM_000322.5:c.929G>A
	Gene Symbol	PRPH2
	Variant Type	SNV
	Location	6:42698407-42698407
	Exon Numbers	3/3
	Ref	C
	Alt	T
	Frequency (Read)	0.50628 (484/956)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000313.2:p.Arg310Lys
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP2(ClinVar),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.9123
	Allele frequency in gnomAD - East Asian	0.998
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.9413
	SIFT Prediction	tolerated
	dbSNP ID	rs425876
	ClinVar ID	138905
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.999
	Report Inclusion	None
142	Variant (HGVSc)	NM_000322.5:c.910C>G
	Gene Symbol	PRPH2
	Variant Type	SNV
	Location	6:42698426-42698426
	Exon Numbers	3/3
	Ref	G
	Alt	C
	Frequency (Read)	0.49948 (480/961)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000313.2:p.Gln304Glu
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP2(ClinVar),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.7749
	Allele frequency in gnomAD - East Asian	0.8606
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.7566
	SIFT Prediction	tolerated
	dbSNP ID	rs390659
	ClinVar ID	138904
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.839
	Report Inclusion	None

143	Variant (HGVSc)	NM_000322.5:c.318T>C
	Gene Symbol	PRPH2
	Variant Type	SNV
	Location	6:42722017-42722017
	Exon Numbers	1/3
	Ref	A
	Alt	G
	Frequency (Read)	0.49612 (447/901)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_000313.2:p.Val106=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.5776
	Allele frequency in gnomAD - East Asian	0.5897
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.5845
	SIFT Prediction	-
	dbSNP ID	rs7764439
	ClinVar ID	92846
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.469
	Report Inclusion	None
144	Variant (HGVSc)	NM_002905.5:c.423C>T
	Gene Symbol	RDH5
	Variant Type	SNV
	Location	12:55721801-55721801
	Exon Numbers	3/5
	Ref	C
	Alt	T
	Frequency (Read)	0.51348 (457/890)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_002896.2:p.Ile141=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.1905
	Allele frequency in gnomAD - East Asian	0.1165
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.1528
	SIFT Prediction	-
	dbSNP ID	rs3138142
	ClinVar ID	258857
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.052
	Report Inclusion	None
145	Variant (HGVSc)	NM_207391.3:c.286G>T
	Gene Symbol	RGS9BP
	Variant Type	SNV
	Location	19:32676549-32676549
	Exon Numbers	1/1
	Ref	G
	Alt	T
	Frequency (Read)	0.48469 (459/947)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_997274.2:p.Ala96Ser
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.5934
	Allele frequency in gnomAD - East Asian	0.3218

	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.4349
	SIFT Prediction	tolerated
	dbSNP ID	rs259290
	ClinVar ID	96676
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.278
	Report Inclusion	None
146	Variant (HGVSc)	NM_207391.3:c.*2G>A
	Gene Symbol	RGS9BP
	Variant Type	SNV
	Location	19:32676973-32676973
	Exon Numbers	1/1
	Ref	G
	Alt	A
	Frequency (Read)	0.46315 (421/909)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BP6(ClinVar)
	Inheritance Pattern	Recessive
	Variant Function	3_prime_UTR_variant
	Allele frequency in gnomAD - whole population	0.6032
	Allele frequency in gnomAD - East Asian	0.3122
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.5373
	SIFT Prediction	-
	dbSNP ID	rs259291
	ClinVar ID	167597
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.362
	Report Inclusion	None
147	Variant (HGVSc)	NM_000539.3:c.696+4C>T
	Gene Symbol	RHO
	Variant Type	SNV
	Location	3:129532420-129532420
	Exon Numbers	Intronic
	Ref	C
	Alt	T
	Frequency (Read)	0.47076 (330/701)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	splice_donor_region_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.08151
	Allele frequency in gnomAD - East Asian	0.06336
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.0857
	SIFT Prediction	-
	dbSNP ID	rs56340615
	ClinVar ID	256384
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.039
	Report Inclusion	None
148	Variant (HGVSc)	NM_000327.4:c.353G>C
	Gene Symbol	ROM1
	Variant Type	SNV
	Location	11:62613634-62613634
	Exon Numbers	1/3
	Ref	G
	Alt	C
	Frequency (Read)	1.0 (1121/1121)
	Zygosity	Homozygous

	Amino Acid Change (HGVSp)	NP_000318.2:p.Gly118Ala
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(SIFT)
	Inheritance Pattern	Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.9847
	Allele frequency in gnomAD - East Asian	0.9994
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.9353
	SIFT Prediction	tolerated
	dbSNP ID	rs1799959
	ClinVar ID	167600
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	1.0
	Report Inclusion	None
149	Variant (HGVSc)	NM_006269.2:c.2615G>A
	Gene Symbol	RP1
	Variant Type	SNV
	Location	8:54626497-54626497
	Exon Numbers	4/4
	Ref	G
	Alt	A
	Frequency (Read)	0.44828 (195/435)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_006260.1:p.Arg872His
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(SIFT),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.2761
	Allele frequency in gnomAD - East Asian	0.4408
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.2722
	SIFT Prediction	deleterious
	dbSNP ID	rs444772
	ClinVar ID	167602
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.391
	Report Inclusion	None
150	Variant (HGVSc)	NM_006269.2:c.2953A>T
	Gene Symbol	RP1
	Variant Type	SNV
	Location	8:54626835-54626835
	Exon Numbers	4/4
	Ref	A
	Alt	T
	Frequency (Read)	0.52941 (216/408)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_006260.1:p.Asn985Tyr
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(SIFT),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.3433
	Allele frequency in gnomAD - East Asian	0.06062
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.25
	SIFT Prediction	deleterious
	dbSNP ID	rs2293869
	ClinVar ID	5969
	ClinVar Pathogenicity	Benign/Likely_benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.107
	Report Inclusion	None

151	Variant (HGVSc)	NM_006269.2:c.3699C>T
	Gene Symbol	RP1
	Variant Type	SNV
	Location	8:54627581-54627581
	Exon Numbers	4/4
	Ref	C
	Alt	T
	Frequency (Read)	0.44444 (304/684)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_006260.1:p.Ser1233=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.01618
	Allele frequency in gnomAD - East Asian	0.0
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.0234
	SIFT Prediction	-
	dbSNP ID	rs114557304
	ClinVar ID	363293
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
152	Variant (HGVSc)	NM_006269.2:c.4784G>A
	Gene Symbol	RP1
	Variant Type	SNV
	Location	8:54628666-54628666
	Exon Numbers	4/4
	Ref	G
	Alt	A
	Frequency (Read)	0.45597 (233/511)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_006260.1:p.Arg1595Gln
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(SIFT),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(BLOSUM)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.0164
	Allele frequency in gnomAD - East Asian	1.088E-4
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.0242
	SIFT Prediction	deleterious
	dbSNP ID	rs35084330
	ClinVar ID	363303
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	6.581E-5
	Report Inclusion	None
153	Variant (HGVSc)	NM_006269.2:c.5071T>C
	Gene Symbol	RP1
	Variant Type	SNV
	Location	8:54628953-54628953
	Exon Numbers	4/4
	Ref	T
	Alt	C
	Frequency (Read)	0.46374 (275/593)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_006260.1:p.Ser1691Pro
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.2775
	Allele frequency in gnomAD - East Asian	0.4413

	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.279
	SIFT Prediction	tolerated
	dbSNP ID	rs414352
	ClinVar ID	95354
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.391
	Report Inclusion	None
154	Variant (HGVSc)	NM_006269.2:c.6098G>A
	Gene Symbol	RP1
	Variant Type	SNV
	Location	8:54629980-54629980
	Exon Numbers	4/4
	Ref	G
	Alt	A
	Frequency (Read)	0.43155 (145/336)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_006260.1:p.Cys2033Tyr
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.3354
	Allele frequency in gnomAD - East Asian	0.06062
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.2157
	SIFT Prediction	tolerated
	dbSNP ID	rs61739567
	ClinVar ID	95357
	ClinVar Pathogenicity	Benign/Likely_benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.107
	Report Inclusion	None
155	Variant (HGVSc)	NM_178857.6:c.6853G>A
	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10607245-10607245
	Exon Numbers	4/4
	Ref	C
	Alt	T
	Frequency (Read)	0.50185 (408/813)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_849188.4:p.Gly2285Arg
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(SIFT),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.4784
	Allele frequency in gnomAD - East Asian	0.5622
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.472
	SIFT Prediction	deleterious
	dbSNP ID	rs55642448
	ClinVar ID	361209
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.526
	Report Inclusion	None
156	Variant (HGVSc)	NM_178857.6:c.6723A>G
	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10607375-10607375
	Exon Numbers	4/4
	Ref	T
	Alt	C
	Frequency (Read)	1.0 (870/870)
	Zygosity	Homozygous

	Amino Acid Change (HGVSp)	NP_849188.4:p.Ser2241=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.6191
	Allele frequency in gnomAD - East Asian	0.9777
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.651
	SIFT Prediction	-
	dbSNP ID	rs56382513
	ClinVar ID	361212
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.916
	Report Inclusion	None
157	Variant (HGVSc)	NM_178857.6:c.6418G>A
	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10607680-10607680
	Exon Numbers	4/4
	Ref	C
	Alt	T
	Frequency (Read)	0.44262 (378/854)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_849188.4:p.Glu2140Lys
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(SIFT),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(BLOSUM)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.138
	Allele frequency in gnomAD - East Asian	0.413
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.1699
	SIFT Prediction	deleterious_low_confidence
	dbSNP ID	rs72494282
	ClinVar ID	361223
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.335
	Report Inclusion	None
158	Variant (HGVSc)	NM_178857.6:c.6264G>T
	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10607834-10607834
	Exon Numbers	4/4
	Ref	C
	Alt	A
	Frequency (Read)	0.45199 (386/854)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_849188.4:p.Gln2088His
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.1317
	Allele frequency in gnomAD - East Asian	0.395
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.1641
	SIFT Prediction	tolerated_low_confidence
	dbSNP ID	rs11778341
	ClinVar ID	361228
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.338
	Report Inclusion	None

159	Variant (HGVSc)	NM_178857.6:c.6209A>T
	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10607889-10607889
	Exon Numbers	4/4
	Ref	T
	Alt	A
	Frequency (Read)	0.45574 (381/836)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_849188.4:p.Glu2070Val
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.1314
	Allele frequency in gnomAD - East Asian	0.4029
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.1613
	SIFT Prediction	tolerated_low_confidence
	dbSNP ID	rs11782670
	ClinVar ID	361229
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.363
	Report Inclusion	None
160	Variant (HGVSc)	NM_178857.6:c.5860A>G
	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10608238-10608238
	Exon Numbers	4/4
	Ref	T
	Alt	C
	Frequency (Read)	1.0 (799/799)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_849188.4:p.Thr1954Ala
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.614
	Allele frequency in gnomAD - East Asian	0.9778
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.6494
	SIFT Prediction	tolerated_low_confidence
	dbSNP ID	rs11783478
	ClinVar ID	361235
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.918
	Report Inclusion	None
161	Variant (HGVSc)	NM_178857.6:c.5666A>T
	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10608432-10608432
	Exon Numbers	4/4
	Ref	T
	Alt	A
	Frequency (Read)	0.46002 (374/813)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_849188.4:p.Asp1889Val
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.2591
	Allele frequency in gnomAD - East Asian	0.4091

	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.2374
	SIFT Prediction	tolerated
	dbSNP ID	rs28446662
	ClinVar ID	361242
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.37
	Report Inclusion	None
162	Variant (HGVSc)	NM_178857.6:c.5126C>T
	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10608972-10608972
	Exon Numbers	4/4
	Ref	G
	Alt	A
	Frequency (Read)	0.52658 (515/978)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_849188.4:p.Ala1709Val
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501,ClinVar)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.3744
	Allele frequency in gnomAD - East Asian	0.02192
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.3107
	SIFT Prediction	tolerated
	dbSNP ID	rs13267180
	ClinVar ID	361259
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.084
	Report Inclusion	None
163	Variant (HGVSc)	NM_178857.6:c.4484C>G
	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10609614-10609614
	Exon Numbers	4/4
	Ref	G
	Alt	C
	Frequency (Read)	0.51653 (453/877)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_849188.4:p.Pro1495Arg
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.2983
	Allele frequency in gnomAD - East Asian	0.417
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.2817
	SIFT Prediction	tolerated
	dbSNP ID	rs4841399
	ClinVar ID	361276
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.373
	Report Inclusion	None
164	Variant (HGVSc)	NM_178857.6:c.4401G>T
	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10609697-10609697
	Exon Numbers	4/4
	Ref	C
	Alt	A
	Frequency (Read)	0.99775 (888/890)
	Zygosity	Homozygous

	Amino Acid Change (HGVSp)	NP_849188.4:p.Arg1467Ser
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.9842
	Allele frequency in gnomAD - East Asian	0.9999
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.9409
	SIFT Prediction	tolerated
	dbSNP ID	rs4840498
	ClinVar ID	361282
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	1.0
	Report Inclusion	None
165	Variant (HGVSc)	NM_178857.6:c.4032A>G
	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10610066-10610066
	Exon Numbers	4/4
	Ref	T
	Alt	C
	Frequency (Read)	0.43601 (293/672)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_849188.4:p.Thr1344=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.3331
	Allele frequency in gnomAD - East Asian	0.6662
	1000 Genomes Minor Allele	-
	1000 Genomes Minor Allele Frequency	-
	SIFT Prediction	-
	dbSNP ID	rs4840499
	ClinVar ID	361288
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.591
	Report Inclusion	None
166	Variant (HGVSc)	NM_178857.6:c.3436C>T
	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10610662-10610662
	Exon Numbers	4/4
	Ref	G
	Alt	A
	Frequency (Read)	1.0 (850/850)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_849188.4:p.Arg1146Trp
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(SIFT),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.7332
	Allele frequency in gnomAD - East Asian	0.6261
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.7081
	SIFT Prediction	deleterious
	dbSNP ID	rs4840502
	ClinVar ID	361309
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.63
	Report Inclusion	None

167	Variant (HGVSc)	NM_178857.6:c.2375T>C
	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10611723-10611723
	Exon Numbers	4/4
	Ref	A
	Alt	G
	Frequency (Read)	0.52149 (449/861)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_849188.4:p.Leu792Pro
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.416
	Allele frequency in gnomAD - East Asian	0.02226
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.3149
	SIFT Prediction	tolerated
	dbSNP ID	rs35602868
	ClinVar ID	361349
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.084
	Report Inclusion	None
168	Variant (HGVSc)	NM_178857.6:c.2316G>A
	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10611782-10611782
	Exon Numbers	4/4
	Ref	C
	Alt	T
	Frequency (Read)	0.47843 (377/788)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_849188.4:p.Ser772=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BP6(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.06764
	Allele frequency in gnomAD - East Asian	1.67E-4
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.0303
	SIFT Prediction	-
	dbSNP ID	rs62490857
	ClinVar ID	361352
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
169	Variant (HGVSc)	NM_178857.6:c.1791C>T
	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10612307-10612307
	Exon Numbers	4/4
	Ref	G
	Alt	A
	Frequency (Read)	0.49143 (459/934)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_849188.4:p.Gly597=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.8441
	Allele frequency in gnomAD - East Asian	0.636

	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.7973
	SIFT Prediction	-
	dbSNP ID	rs6996950
	ClinVar ID	361367
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.635
	Report Inclusion	None
170	Variant (HGVSc)	NM_178857.6:c.665A>C
	Gene Symbol	RP1L1
	Variant Type	SNV
	Location	8:10616532-10616532
	Exon Numbers	3/4
	Ref	T
	Alt	G
	Frequency (Read)	0.50809 (408/803)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_849188.4:p.His222Pro
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(SIFT),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.1861
	Allele frequency in gnomAD - East Asian	0.4443
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.2167
	SIFT Prediction	deleterious
	dbSNP ID	rs4388421
	ClinVar ID	361399
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.515
	Report Inclusion	None
171	Variant (HGVSc)	NM_203288.2:c.314-9C>T
	Gene Symbol	RP9
	Variant Type	SNV
	Location	7:33097371-33097371
	Exon Numbers	Intronic
	Ref	G
	Alt	A
	Frequency (Read)	1.0 (388/388)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	-
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar)
	Inheritance Pattern	Dominant
	Variant Function	splice_polypyrimidine_tract_variant,intron_variant
	Allele frequency in gnomAD - whole population	0.9936
	Allele frequency in gnomAD - East Asian	0.9993
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.9966
	SIFT Prediction	-
	dbSNP ID	rs6462460
	ClinVar ID	360045
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	1.0
	Report Inclusion	None
172	Variant (HGVSc)	NM_020366.4:c.287C>A
	Gene Symbol	RPGRIP1
	Variant Type	SNV
	Location	14:21301034-21301034
	Exon Numbers	4/25
	Ref	C
	Alt	A
	Frequency (Read)	0.49773 (439/882)
	Zygosity	Heterozygous

	Amino Acid Change (HGVSp)	NP_065099.3:p.Pro96Gln
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.07088
	Allele frequency in gnomAD - East Asian	0.03546
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.0583
	SIFT Prediction	tolerated
	dbSNP ID	rs1040904
	ClinVar ID	99819
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.024
	Report Inclusion	None
173	Variant (HGVSc)	NM_020366.4:c.574A>G
	Gene Symbol	RPGRIP1
	Variant Type	SNV
	Location	14:21302571-21302571
	Exon Numbers	5/25
	Ref	A
	Alt	G
	Frequency (Read)	1.0 (614/614)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_065099.3:p.Lys192Glu
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.5032
	Allele frequency in gnomAD - East Asian	0.3267
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.4772
	SIFT Prediction	tolerated
	dbSNP ID	rs6571751
	ClinVar ID	99827
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.292
	Report Inclusion	None
174	Variant (HGVSc)	NM_000541.5:c.1207G>A
	Gene Symbol	SAG
	Variant Type	SNV
	Location	2:233346901-233346901
	Exon Numbers	16/16
	Ref	G
	Alt	A
	Frequency (Read)	1.0 (597/597)
	Zygosity	Homozygous
	Amino Acid Change (HGVSp)	NP_000532.2:p.Val403Ile
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive, Dominant
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.3669
	Allele frequency in gnomAD - East Asian	0.4122
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.3029
	SIFT Prediction	tolerated_low_confidence
	dbSNP ID	rs1046974
	ClinVar ID	194606
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.295
	Report Inclusion	None

175	Variant (HGVSc)	NM_014014.5:c.3550T>C
	Gene Symbol	SNRNP200
	Variant Type	SNV
	Location	2:96287095-96287095
	Exon Numbers	27/45
	Ref	A
	Alt	G
	Frequency (Read)	0.50126 (399/796)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_054733.2:p.Leu1184=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Dominant
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.3152
	Allele frequency in gnomAD - East Asian	0.1768
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.2841
	SIFT Prediction	-
	dbSNP ID	rs3171927
	ClinVar ID	337548
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.279
	Report Inclusion	None
176	Variant (HGVSc)	NM_018418.5:c.220G>A
	Gene Symbol	SPATA7
	Variant Type	SNV
	Location	14:88396185-88396185
	Exon Numbers	4/12
	Ref	G
	Alt	A
	Frequency (Read)	0.39338 (107/272)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_060888.2:p.Val74Met
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.3368
	Allele frequency in gnomAD - East Asian	0.3198
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.4075
	SIFT Prediction	tolerated
	dbSNP ID	rs3179969
	ClinVar ID	286866
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.285
	Report Inclusion	None
177	Variant (HGVSc)	NM_005802.5:c.2319T>C
	Gene Symbol	TOPORS
	Variant Type	SNV
	Location	9:32542206-32542206
	Exon Numbers	3/3
	Ref	A
	Alt	G
	Frequency (Read)	0.47067 (337/716)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_005793.2:p.Ser773=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Dominant
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.2085
	Allele frequency in gnomAD - East Asian	0.07645

	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.1791
	SIFT Prediction	-
	dbSNP ID	rs10971019
	ClinVar ID	95313
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.085
	Report Inclusion	None
178	Variant (HGVSc)	NM_003322.6:c.783G>C
	Gene Symbol	TULP1
	Variant Type	SNV
	Location	6:35509248-35509248
	Exon Numbers	8/15
	Ref	C
	Alt	G
	Frequency (Read)	0.5221 (378/724)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_003313.3:p.Lys261Asn
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.8228
	Allele frequency in gnomAD - East Asian	0.8404
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.8385
	SIFT Prediction	tolerated
	dbSNP ID	rs2064318
	ClinVar ID	94127
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.864
	Report Inclusion	None
179	Variant (HGVSc)	NM_003322.6:c.776T>C
	Gene Symbol	TULP1
	Variant Type	SNV
	Location	6:35509255-35509255
	Exon Numbers	8/15
	Ref	A
	Alt	G
	Frequency (Read)	0.51582 (375/727)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_003313.3:p.Ile259Thr
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.3811
	Allele frequency in gnomAD - East Asian	0.493
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.394
	SIFT Prediction	tolerated
	dbSNP ID	rs2064317
	ClinVar ID	99671
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.392
	Report Inclusion	None
180	Variant (HGVSc)	NM_003322.6:c.200C>G
	Gene Symbol	TULP1
	Variant Type	SNV
	Location	6:35511797-35511797
	Exon Numbers	4/15
	Ref	G
	Alt	C
	Frequency (Read)	0.49074 (371/756)
	Zygosity	Heterozygous

	Amino Acid Change (HGVSp)	NP_003313.3:p.Thr67Arg
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.851
	Allele frequency in gnomAD - East Asian	0.8428
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.8744
	SIFT Prediction	tolerated_low_confidence
	dbSNP ID	rs7764472
	ClinVar ID	286865
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.865
	Report Inclusion	None
181	Variant (HGVSc)	NM_206933.4:c.13297G>T
	Gene Symbol	USH2A
	Variant Type	SNV
	Location	1:215674614-215674614
	Exon Numbers	63/72
	Ref	C
	Alt	A
	Frequency (Read)	0.54096 (350/647)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_996816.3:p.Val4433Leu
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.01914
	Allele frequency in gnomAD - East Asian	0.0
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.005
	SIFT Prediction	tolerated
	dbSNP ID	rs111033381
	ClinVar ID	48415
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
182	Variant (HGVSc)	NM_206933.4:c.13191G>A
	Gene Symbol	USH2A
	Variant Type	SNV
	Location	1:215674720-215674720
	Exon Numbers	63/72
	Ref	C
	Alt	T
	Frequency (Read)	0.45203 (245/542)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_996816.3:p.Glu4397=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.2029
	Allele frequency in gnomAD - East Asian	0.3522
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.2035
	SIFT Prediction	-
	dbSNP ID	rs2009923
	ClinVar ID	48413
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.382
	Report Inclusion	None

183	Variant (HGVSc)	NM_206933.4:c.12612A>G
	Gene Symbol	USH2A
	Variant Type	SNV
	Location	1:215675299-215675299
	Exon Numbers	63/72
	Ref	T
	Alt	C
	Frequency (Read)	0.51903 (300/578)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_996816.3:p.Thr4204=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.7669
	Allele frequency in gnomAD - East Asian	0.6474
	1000 Genomes Minor Allele	C
	1000 Genomes Minor Allele Frequency	0.6897
	SIFT Prediction	-
	dbSNP ID	rs2797235
	ClinVar ID	177993
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.613
	Report Inclusion	None
184	Variant (HGVSc)	NM_206933.4:c.10232A>C
	Gene Symbol	USH2A
	Variant Type	SNV
	Location	1:215786825-215786825
	Exon Numbers	52/72
	Ref	T
	Alt	G
	Frequency (Read)	0.52161 (362/694)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_996816.3:p.Glu3411Ala
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.5482
	Allele frequency in gnomAD - East Asian	0.6342
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.5887
	SIFT Prediction	tolerated
	dbSNP ID	rs10864198
	ClinVar ID	48344
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.619
	Report Inclusion	None
185	Variant (HGVSc)	NM_206933.4:c.6506T>C
	Gene Symbol	USH2A
	Variant Type	SNV
	Location	1:215999038-215999038
	Exon Numbers	34/72
	Ref	A
	Alt	G
	Frequency (Read)	0.50519 (146/289)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_996816.3:p.Ile2169Thr
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.5628
	Allele frequency in gnomAD - East Asian	0.8405

	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.5935
	SIFT Prediction	tolerated
	dbSNP ID	rs10864219
	ClinVar ID	48562
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.886
	Report Inclusion	None
186	Variant (HGVSc)	NM_206933.4:c.6317T>C
	Gene Symbol	USH2A
	Variant Type	SNV
	Location	1:216046439-216046439
	Exon Numbers	32/72
	Ref	A
	Alt	G
	Frequency (Read)	0.52826 (243/460)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_996816.3:p.Ile2106Thr
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.5943
	Allele frequency in gnomAD - East Asian	0.4408
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.5956
	SIFT Prediction	tolerated
	dbSNP ID	rs6657250
	ClinVar ID	167815
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.441
	Report Inclusion	None
187	Variant (HGVSc)	NM_206933.4:c.4994T>C
	Gene Symbol	USH2A
	Variant Type	SNV
	Location	1:216084871-216084871
	Exon Numbers	25/72
	Ref	A
	Alt	G
	Frequency (Read)	0.52159 (157/301)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_996816.3:p.Ile1665Thr
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.1104
	Allele frequency in gnomAD - East Asian	0.0209
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.0627
	SIFT Prediction	tolerated
	dbSNP ID	rs56222536
	ClinVar ID	48525
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.019
	Report Inclusion	None
188	Variant (HGVSc)	NM_206933.4:c.4457G>A
	Gene Symbol	USH2A
	Variant Type	SNV
	Location	1:216175422-216175422
	Exon Numbers	21/72
	Ref	C
	Alt	T
	Frequency (Read)	0.51211 (296/578)
	Zygosity	Heterozygous

	Amino Acid Change (HGVSp)	NP_996816.3:p.Arg1486Lys
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT),BP6(Benign Variants from PMID:31213501)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.6605
	Allele frequency in gnomAD - East Asian	0.7813
	1000 Genomes Minor Allele	T
	1000 Genomes Minor Allele Frequency	0.6623
	SIFT Prediction	tolerated
	dbSNP ID	rs1805049
	ClinVar ID	177992
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.818
	Report Inclusion	None
189	Variant (HGVSc)	NM_206933.4:c.2109T>C
	Gene Symbol	USH2A
	Variant Type	SNV
	Location	1:216250961-216250961
	Exon Numbers	12/72
	Ref	A
	Alt	G
	Frequency (Read)	0.49721 (356/716)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_996816.3:p.Asp703=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.01698
	Allele frequency in gnomAD - East Asian	0.0
	1000 Genomes Minor Allele	G
	1000 Genomes Minor Allele Frequency	0.0074
	SIFT Prediction	-
	dbSNP ID	rs45555435
	ClinVar ID	48483
	ClinVar Pathogenicity	Benign/Likely_benign
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None
190	Variant (HGVSc)	NM_206933.4:c.1931A>T
	Gene Symbol	USH2A
	Variant Type	SNV
	Location	1:216289320-216289320
	Exon Numbers	11/72
	Ref	T
	Alt	A
	Frequency (Read)	0.53846 (308/572)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_996816.3:p.Asp644Val
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	PP3(BLOSUM),BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP1(ClinVar),BP4(SIFT)
	Inheritance Pattern	Recessive
	Variant Function	missense_variant
	Allele frequency in gnomAD - whole population	0.0581
	Allele frequency in gnomAD - East Asian	5.989E-4
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.0345
	SIFT Prediction	tolerated
	dbSNP ID	rs1805048
	ClinVar ID	48478
	ClinVar Pathogenicity	Benign/Likely_benign
	HGMD	-
	GEM-J WGA Allele Frequency	-
	Report Inclusion	None

191	Variant (HGVSc)	NM_206933.4:c.1419C>T
	Gene Symbol	USH2A
	Variant Type	SNV
	Location	1:216323605-216323605
	Exon Numbers	8/72
	Ref	G
	Alt	A
	Frequency (Read)	0.46082 (247/536)
	Zygosity	Heterozygous
	Amino Acid Change (HGVSp)	NP_996816.3:p.Thr473=
	User Classification	Benign
	ACMG Classification	Benign
	ACMG Labels	BA1(gnomAD),BS1(gnomAD),BS3(ClinVar),BP7(VEP,dbscSNV,GERP)
	Inheritance Pattern	Recessive
	Variant Function	synonymous_variant
	Allele frequency in gnomAD - whole population	0.2499
	Allele frequency in gnomAD - East Asian	0.1337
	1000 Genomes Minor Allele	A
	1000 Genomes Minor Allele Frequency	0.263
	SIFT Prediction	-
	dbSNP ID	rs1805050
	ClinVar ID	48431
	ClinVar Pathogenicity	Benign
	HGMD	-
	GEM-J WGA Allele Frequency	0.124
	Report Inclusion	None

CNV Information (as Reference)

1	Variant (ISCN)	seq[GRCh38] 1q31.3(195134497_197328497)x1
	Variant Type	Deletion
	Location	1:195134497-197328497
	Copy Number	1
	Log Ratio	-0.617
	Genes	**CRB1 (exons 1-2/12)**
	User Classification	Unclassified
2	Variant (ISCN)	seq[GRCh38] 2q13q13(111996876_111997026)x1
	Variant Type	Deletion
	Location	2:111996876-111997026
	Copy Number	1
	Log Ratio	-1.055
	Genes	**MERTK (intronic)**
	User Classification	Unclassified
3	Variant (ISCN)	seq[GRCh38] 4p15.32p15.32(15979817_15979967)x1
	Variant Type	Deletion
	Location	4:15979817-15979967
	Copy Number	1
	Log Ratio	-1.425
	Genes	**PROM1 (exon 25/28)**
	User Classification	Unclassified
4	Variant (ISCN)	seq[GRCh38] 5q14.3q14.3(90724940_90725233)x1
	Variant Type	Deletion
	Location	5:90724940-90725233
	Copy Number	1
	Log Ratio	-0.942
	Genes	**ADGRV1 (exons 46-47/90)**
	User Classification	Unclassified
5	Variant (ISCN)	seq[GRCh38] 6q12q13(65553302_70093966)x1
	Variant Type	Deletion
	Location	6:65553302-70093966
	Copy Number	1
	Log Ratio	-0.664
	Genes	**EYS (exons 1-2/43)**, **EYS (exons 1-2/44)**
	User Classification	Unclassified
6	Variant (ISCN)	seq[GRCh38] 8q21.3q21.3(86644578_86654111)x1
	Variant Type	Deletion
	Location	8:86644578-86654111
	Copy Number	1
	Log Ratio	-0.6
	Genes	**CNGB3 (exons 7-9/18)**
	User Classification	Unclassified
7	Variant (ISCN)	seq[GRCh38] 12q21.32q21.32(88087770_88087953)x1

	Variant Type	Deletion
	Location	12:88087770-88087953
	Copy Number	1
	Log Ratio	-0.835
	Genes	**CEP290 (exon 32/54)**
	User Classification	Unclassified
8	Variant (ISCN)	seq[GRCh38] 14q31.3q31.3(88841394_88846711)x1
	Variant Type	Deletion
	Location	14:88841394-88846711
	Copy Number	1
	Log Ratio	-0.62
	Genes	**TTC8 (exons 6-7/15)**
	User Classification	Unclassified
9	Variant (ISCN)	seq[GRCh38] Xp11.4p11.4(38290916_38292816)x1
	Variant Type	Deletion
	Location	X:38290916-38292816
	Copy Number	1
	Log Ratio	-0.663
	Genes	**RPGR (exons 12-13/19)**, **RPGR (exons 12-13/15)**
	User Classification	Unclassified

Appendix 1: Disease Category Referred From RetNet

Gene	Associated Diseases
ABCA4	Cone or cone-rod dystrophy, autosomal recessive; Macular degeneration, autosomal recessive; Retinitis pigmentosa, autosomal recessive
ADGRV1	Usher syndrome, autosomal recessive
AIPL1	Cone or cone-rod dystrophy, autosomal dominant; Leber congenital amaurosis, autosomal recessive
BEST1	Macular degeneration, autosomal dominant; Retinitis pigmentosa, autosomal dominant; Retinitis pigmentosa, autosomal recessive; Other retinopathy, autosomal dominant; Other retinopathy, autosomal recessive
C8orf37	Bardet-Biedl syndrome, autosomal recessive; Cone or cone-rod dystrophy, autosomal recessive; Retinitis pigmentosa, autosomal recessive
CA4	Retinitis pigmentosa, autosomal dominant
CACNA1F	Cone or cone-rod dystrophy, X-linked; Congenital stationary night blindness, X-linked; Other retinopathy, X-linked
CDH23	Deafness alone or syndromic, autosomal recessive; Usher syndrome, autosomal recessive
CDHR1	Cone or cone-rod dystrophy, autosomal recessive
CEP290	Bardet-Biedl syndrome, autosomal recessive; Leber congenital amaurosis, autosomal recessive; Syndromic/systemic diseases with retinopathy, autosomal recessive
CERKL	Cone or cone-rod dystrophy, autosomal recessive; Retinitis pigmentosa, autosomal recessive
CFAP410	Cone or cone-rod dystrophy, autosomal recessive
CHM	Other retinopathy, X-linked
CLRN1	Retinitis pigmentosa, autosomal recessive; Usher syndrome, autosomal recessive
CNGA1	Retinitis pigmentosa, autosomal recessive
CNGA3	Cone or cone-rod dystrophy, autosomal recessive; Other retinopathy, autosomal recessive
CNGB1	Retinitis pigmentosa, autosomal recessive
CNGB3	Cone or cone-rod dystrophy, autosomal recessive; Other retinopathy, autosomal recessive
CRB1	Leber congenital amaurosis, autosomal recessive; Retinitis pigmentosa, autosomal recessive; Other retinopathy, autosomal dominant
CRX	Cone or cone-rod dystrophy, autosomal dominant; Leber congenital amaurosis, autosomal dominant; Leber congenital amaurosis, autosomal recessive; Retinitis pigmentosa, autosomal dominant
CYP4V2	Retinitis pigmentosa, autosomal recessive; Other retinopathy, autosomal recessive
DHDDS	Retinitis pigmentosa, autosomal recessive
DRAM2	Macular degeneration, autosomal recessive
EYS	Retinitis pigmentosa, autosomal recessive
FAM161A	Retinitis pigmentosa, autosomal recessive
FSCN2	Macular degeneration, autosomal dominant; Retinitis pigmentosa, autosomal dominant
GNAT2	Cone or cone-rod dystrophy, autosomal recessive
GRK1	Congenital stationary night blindness, autosomal recessive
GUCA1A	Cone or cone-rod dystrophy, autosomal dominant
GUCA1B	Macular degeneration, autosomal dominant; Retinitis pigmentosa, autosomal dominant
GUCY2D	Cone or cone-rod dystrophy, autosomal dominant; Leber congenital amaurosis, autosomal recessive
IDH3B	Retinitis pigmentosa, autosomal recessive
IMPDH1	Leber congenital amaurosis, autosomal dominant; Retinitis pigmentosa, autosomal dominant
IMPG2	Retinitis pigmentosa, autosomal recessive
IQCB1	Leber congenital amaurosis, autosomal recessive; Syndromic/systemic diseases with retinopathy, autosomal recessive
KCNV2	Cone or cone-rod dystrophy, autosomal recessive
KLHL7	Retinitis pigmentosa, autosomal dominant
LRAT	Leber congenital amaurosis, autosomal recessive; Retinitis pigmentosa, autosomal recessive
MAK	Retinitis pigmentosa, autosomal recessive
MERTK	Retinitis pigmentosa, autosomal recessive
MYO7A	Deafness alone or syndromic, autosomal recessive; Usher syndrome, autosomal recessive
NMNAT1	Leber congenital amaurosis, autosomal recessive
NR2E3	Retinitis pigmentosa, autosomal dominant; Retinitis pigmentosa, autosomal recessive; Other retinopathy, autosomal recessive
NRL	Retinitis pigmentosa, autosomal dominant; Retinitis pigmentosa, autosomal recessive
NYX	Congenital stationary night blindness, X-linked
PCARE	Retinitis pigmentosa, autosomal recessive
PDE6A	Retinitis pigmentosa, autosomal recessive

PDE6B	Congenital stationary night blindness, autosomal dominant; Retinitis pigmentosa, autosomal recessive
PDE6C	Cone or cone-rod dystrophy, autosomal recessive
PDE6G	Retinitis pigmentosa, autosomal recessive
POC1B	Cone or cone-rod dystrophy, autosomal recessive; Syndromic/systemic diseases with retinopathy, autosomal recessive
PRCD	Retinitis pigmentosa, autosomal recessive
PROM1	Cone or cone-rod dystrophy, autosomal dominant; Macular degeneration, autosomal dominant; Retinitis pigmentosa, autosomal recessive; Other retinopathy, autosomal recessive
PRPF3	Retinitis pigmentosa, autosomal dominant
PRPF31	Retinitis pigmentosa, autosomal dominant
PRPF6	Retinitis pigmentosa, autosomal dominant
PRPF8	Retinitis pigmentosa, autosomal dominant
PRPH2	Cone or cone-rod dystrophy, autosomal dominant; Leber congenital amaurosis, autosomal recessive; Macular degeneration, autosomal dominant; Retinitis pigmentosa, autosomal dominant
RBP3	Retinitis pigmentosa, autosomal recessive
RDH12	Leber congenital amaurosis, autosomal recessive; Retinitis pigmentosa, autosomal dominant
RDH5	Cone or cone-rod dystrophy, autosomal recessive; Congenital stationary night blindness, autosomal recessive
RGR	Chorioretinal atrophy or degeneration, autosomal dominant; Retinitis pigmentosa, autosomal recessive
RGS9BP	Other retinopathy, autosomal recessive
RHO	Congenital stationary night blindness, autosomal dominant; Retinitis pigmentosa, autosomal dominant; Retinitis pigmentosa, autosomal recessive
RLBP1	Retinitis pigmentosa, autosomal recessive; Other retinopathy, autosomal recessive
ROM1	Retinitis pigmentosa, autosomal dominant
RP1	Retinitis pigmentosa, autosomal dominant; Retinitis pigmentosa, autosomal recessive
RP1L1	Macular degeneration, autosomal dominant; Retinitis pigmentosa, autosomal recessive
RP2	Retinitis pigmentosa, X-linked
RP9	Retinitis pigmentosa, autosomal dominant
RPE65	Leber congenital amaurosis, autosomal recessive; Retinitis pigmentosa, autosomal dominant; Retinitis pigmentosa, autosomal recessive
RPGR	Cone or cone-rod dystrophy, X-linked; Macular degeneration, X-linked; Retinitis pigmentosa, X-linked
RPGRIP1	Cone or cone-rod dystrophy, autosomal recessive; Leber congenital amaurosis, autosomal recessive
RS1	Other retinopathy, X-linked
SAG	Congenital stationary night blindness, autosomal recessive; Retinitis pigmentosa, autosomal dominant; Retinitis pigmentosa, autosomal recessive
SEMA4A	Cone or cone-rod dystrophy, autosomal dominant; Retinitis pigmentosa, autosomal dominant
SNRNP200	Retinitis pigmentosa, autosomal dominant
SPATA7	Leber congenital amaurosis, autosomal recessive; Retinitis pigmentosa, autosomal recessive
TOPORS	Retinitis pigmentosa, autosomal dominant
TTC8	Bardet-Biedl syndrome, autosomal recessive; Retinitis pigmentosa, autosomal recessive
TULP1	Leber congenital amaurosis, autosomal recessive; Retinitis pigmentosa, autosomal recessive
USH2A	Retinitis pigmentosa, autosomal recessive; Usher syndrome, autosomal recessive
ZNF513	Retinitis pigmentosa, autosomal recessive

Appendix 2: Criteria for classifying pathogenic and benign variants based on ACMG guidelines

Very strong evidence of pathogenicity	
PVS1	Null variant (nonsense, frameshift, canonical ±1 or 2 splice sites, initiation codon) in a gene where loss of function (LOF) is a known mechanism of disease. SOURCE=VEP Impact
Strong evidence of pathogenicity	
PS1	Same amino acid change as a previously established pathogenic variant regardless of nucleotide change. SOURCE=ClinVar, HGMD, Custom Variant List
PS3	Well-established in vitro or in vivo functional studies supportive of a damaging effect on the gene or gene product. SOURCE= ClinVar, HGMD
Moderate evidence of pathogenicity	
PM1	Located in a mutational hot spot and/or critical and well-established functional domain without benign variation. SOURCE=ClinVar, PFAM
PM2	Absent from controls if dominant or with a frequency <= 0.5% if recessive in gnomAD database. SOURCE=gnomAD Allele Frequency
PM4	Protein length changes due to in-frame deletions/insertions in a non-repeat region or stop-loss variants. SOURCE=VEP consequence, Repeat Masker
PM5	Novel missense change at an amino acid residue where a different missense change determined to be pathogenic and has been seen before. SOURCE= ClinVar, HGMD, Custom Variant List
Supporting evidence of pathogenicity	
PP2	Missense variant in a gene that has a low rate of benign missense variation and where missense variants are a common mechanism of disease. SOURCE=ClinVar
PP3	Multiple lines of computational evidence support a deleterious effect on the gene or gene product (conservation, evolutionary, splicing impact, etc.). SOURCE= SIFT, BLOSUM62, dbscSNV, GERP
PP5	Reputable source recently reports variant as pathogenic but the evidence is not available to the laboratory to perform an independent evaluation. SOURCE=ClinVar, HGMD, Pathogenic Variants from PMID:31213501
Stand-Alone evidence of benign impact	
BA1	Allele frequency is above 5% in gnomAD database. SOURCE=gnomAD Allele Frequency
Strong evidence of benign impact	
BS1	Allele frequency is greater than 0.5% for disorder. SOURCE=gnomAD Allele Frequency
BS3	Well-established in vitro or in vivo functional studies show no damaging effect on protein function or splicing. SOURCE=ClinVar
Supporting evidence of benign impact	
BP1	Missense variant in a gene for which primarily truncating variants are known to cause disease. SOURCE=ClinVar
BP3	In-frame deletions/insertions in a repetitive region without a known function. SOURCE=VEP consequence, Repeat Masker, PFAM
BP4	Multiple lines of computational evidence suggest no impact on gene or gene product (conservation, evolutionary, splicing impact, etc.). SOURCE=SIFT, BLOSUM62, dbscSNV, GERP
BP6	Reputable source recently reports variant as benign but the evidence is not available to the laboratory to perform an independent evaluation. SOURCE=ClinVar, Custom Variant List, Benign Variants from PMID:31213501
BP7	A synonymous (silent) variant for which splicing prediction algorithms predict no impact to the splice consensus sequence nor the creation of a new splice site AND the nucleotide is not highly conserved. SOURCE=VEP Consequence, dbscSNV, GERP

ACMG criteria not implemented: PS2, PS4, PM3, PM6, PP1, PP4, BS2, BS4, BP2, BP5

Appendix 3: Rules for combining criteria to classify variants based on ACMG guidelines

- Pathogenic

1. 1 Very Strong (PVS1) AND
    1. ≥1 Strong (PS1,PS3) OR
    2. ≥2 Moderate (PM1,PM2,PM4,PM5) OR
    3. 1 Moderate (PM1,PM2,PM4,PM5) and 1 Supporting (PP2,PP3,PP5) OR
    4. ≥2 Supporting (PP2,PP3,PP5)
2. ≥2 Strong (PS1,PS3) OR
3. 1 Strong (PS1,PS3) AND
    1. ≥3 Moderate (PM1,PM2,PM4,PM5) OR
    2. 2 Moderate (PM1,PM2,PM4,PM5) AND ≥2 Supporting (PP2,PP3,PP5) OR
    3. 1 Moderate (PM1,PM2,PM4,PM5) AND ≥3 Supporting (PP2,PP3,PP5)

- Likely Pathogenic
    1. 1 Very Strong (PVS1) AND 1 Moderate (PM1,PM2,PM4,PM5) OR
    2. 1 Strong (PS1,PS3) AND 1-2 Moderate (PM1,PM2,PM4,PM5) OR
    3. 1 Strong (PS1,PS3) AND ≥2 (PP2,PP3,PP5) OR
    4. ≥3 Moderate (PM1,PM2,PM4,PM5) OR
    5. 2 Moderate (PM1,PM2,PM4,PM5) AND ≥2 Supporting (PP2,PP3,PP5) OR
    6. 1 Moderate (PM1,PM2,PM4,PM5) and ≥3 Supporting (PP2,PP3,PP5)
- Benign
    1. 1 Stand-Alone (BA1) OR
    2. ≥2 Strong (BS2,BS3)
- Likely Benign
    1. 1 Strong (BS1, BS3) AND 1 Supporting (BP1,BP3,BP4,BP6,BP7) OR
    2. ≥2 Supporting (BP1,BP3,BP4,BP6,BP7)
- Uncertain Significance
    1. Variants that do not meet the above criteria
    2. Conflicting classification labels (eg Likely Pathogenic, Likely Benign) which are listed

Appendix 4: Databases/Software Used

Database/Software	Version Used
PFAM	2022-05-15
Ensembl VEP cache 1000 Genomes	phase3
Ensembl VEP cache SIFT	sift5.2.2
Benign Variants from PMID:31213501	v1.0.0
Ensembl VEP cache dbSNP	154
Ensembl VEP cache RefSeq	110
freebayes	1.3.2-38-g71a3e1c-dirty
Custom Variant List	v1.0.0
GEMJ	v1.0.0
multiqc	1.8
ClinVar	2023-01-09
Human gene annotation	2022-10-04
fastp	0.20.0
vep	108.2
dbscSNV	2015-04-13
Ensembl VEP cache assembly	GRCh38.p13
samtools	1.9
Ensembl VEP cache	2022-10-07
Repeat Masker database	2022-10-19
BLOSUM62	108
bwa	0.7.17-r1188
Pathogenic Variants from PMID:31213501	v1.0.0
ogtkit	0.8.6
CNV references	v2.0.0
HGMD	v2022.3
sambamba	0.7.1
bedtools	2.29.2
Ensembl VEP cache gnomADe	r2.1.1
GERP	v1.0.0
Human genome reference	2014-01-11
vt	0.5
Ensembl VEP cache GENCODE	GENCODE_42

Appendix 5: Targeted Genes

Gene Symbol	Transcript	Protein	HGNC ID
ABCA4	NM_000350.3	NP_000341.2	34
ADGRV1	NM_032119.4	NP_115495.3	17416
AIPL1	NM_014336.5	NP_055151.3	359
BEST1	NM_004183.4	NP_004174.1	12703
C8orf37	NM_177965.4	NP_808880.1	27232
CA4	NM_000717.5	NP_000708.1	1375
CACNA1F	NM_005183.4	NP_005174.2	1393
CDH23	NM_022124.6	NP_071407.4	13733
CDHR1	NM_033100.4	NP_149091.1	14550
CEP290	NM_025114.4	NP_079390.3	29021
CERKL	NM_001030311.3	NP_001025482.1	21699

CFAP410	NM_004928.3	NP_004919.1	1260
CHM	NM_000390.4	NP_000381.1	1940
CLRN1	NM_174878.3	NP_777367.1	12605
CNGA1	NM_000087.5	NP_000078.3	2148
CNGA3	NM_001298.3	NP_001289.1	2150
CNGB1	NM_001297.5	NP_001288.3	2151
CNGB3	NM_019098.5	NP_061971.3	2153
CRB1	NM_201253.3	NP_957705.1	2343
CRX	NM_000554.6	NP_000545.1	2383
CYP4V2	NM_207352.4	NP_997235.3	23198
DHDDS	NM_024887.4	NP_079163.2	20603
DRAM2	NM_178454.6	NP_848549.3	28769
EYS	NM_001292009.2	NP_001278938.1	21555
EYS	NM_001142800.2	NP_001136272.1	21555
FAM161A	NM_001201543.2	NP_001188472.1	25808
FSCN2	NM_001077182.3	NP_001070650.1	3960
GNAT2	NM_005272.5	NP_005263.1	4394
GRK1	NM_002929.3	NP_002920.1	10013
GUCA1A	NM_000409.5	NP_000400.2	4678
GUCY2D	NM_000180.4	NP_000171.1	4689
IDH3B	NM_006899.5	NP_008830.2	5385
IMPDH1	NM_000883.4	NP_000874.2	6052
IMPG2	NM_016247.4	NP_057331.2	18362
IQCB1	NM_001023570.4	NP_001018864.2	28949
KCNV2	NM_133497.4	NP_598004.1	19698
KLHL7	NM_001031710.3	NP_001026880.2	15646
LRAT	NM_004744.5	NP_004735.2	6685
MAK	NM_001242957.3	NP_001229886.1	6816
MERTK	NM_006343.3	NP_006334.2	7027
MYO7A	NM_000260.4	NP_000251.3	7606
NMNAT1	NM_022787.4	NP_073624.2	17877
NR2E3	NM_014249.4	NP_055064.1	7974
NRL	NM_006177.5	NP_006168.1	8002
NYX	NM_022567.3	NP_072089.2	8082
PCARE	NM_001029883.3	NP_001025054.1	34383
PDE6A	NM_000440.3	NP_000431.2	8785
PDE6B	NM_000283.4	NP_000274.3	8786
PDE6C	NM_006204.4	NP_006195.3	8787
PDE6G	NM_002602.4	NP_002593.1	8789
POC1B	NM_172240.3	NP_758440.1	30836
PRCD	NM_001077620.3	NP_001071088.1	32528
PROM1	NM_006017.3	NP_006008.1	9454
PRPF3	NM_004698.4	NP_004689.1	17348
PRPF31	NM_015629.4	NP_056444.3	15446
PRPF6	NM_012469.4	NP_036601.2	15860
PRPF8	NM_006445.4	NP_006436.3	17340
PRPH2	NM_000322.5	NP_000313.2	9942
RBP3	NM_002900.3	NP_002891.1	9921
RDH12	NM_152443.3	NP_689656.2	19977
RDH5	NM_002905.5	NP_002896.2	9940
RGR	NM_001012720.2	NP_001012738.1	9990
RGS9BP	NM_207391.3	NP_997274.2	30304
RHO	NM_000539.3	NP_000530.1	10012
RLBP1	NM_000326.5	NP_000317.1	10024
ROM1	NM_000327.4	NP_000318.2	10254
RP1	NM_006269.2	NP_006260.1	10263
RP1L1	NM_178857.6	NP_849188.4	15946
RP2	NM_006915.3	NP_008846.2	10274
RP9	NM_203288.2	NP_976033.1	10288
RPE65	NM_000329.3	NP_000320.1	10294
RPGR	NM_001034853.2	NP_001030025.1	10295
RPGR	NM_000328.3	NP_000319.1	10295
RPGRIP1	NM_020366.4	NP_065099.3	13436
RS1	NM_000330.4	NP_000321.1	10457
SAG	NM_000541.5	NP_000532.2	10521
SEMA4A	NM_022367.4	NP_071762.2	10729
SNRNP200	NM_014014.5	NP_054733.2	30859
SPATA7	NM_018418.5	NP_060888.2	20423
TOPORS	NM_005802.5	NP_005793.2	21653
TTC8	NM_198309.3	NP_938051.1	20087
TULP1	NM_003322.6	NP_003313.3	12423

USH2A	NM_206933.4	NP_996816.3	12601
ZNF513	NM_144631.6	NP_653232.3	26498

Appendix 6: QC Metrics

Name	Definition
Mapped Reads (%)	The percentage of total reads that are aligned to the reference genome
Duplicated Reads (%)	The percentage of total reads that are marked duplicated
Mean Target Coverage	The mean target coverage is defined as the sum of base counts (coverage) within the targeted regions of the panel divided by the total length of these regions. All base counts are calculated from filtered, mapped and de-duplicated reads
Low Coverage (>=100x)	The percentage of bases that have a coverage above or equal to 100
Medium Coverage	The percentage of bases that have a coverage above or equal to 500
High Coverage	The percentage of bases that have a coverage above or equal to 1000
Evenness	The evenness score represents the fraction of whole-sequencing throughput that is correctly distributed over the targeted regions of the panel
Average Quality	The average base Phred quality score of all reads
Average Insert Size	The average absolute template length for paired and mapped reads
Insert Size std	The standard deviation of the average template length distribution
Reads Mapping Quality 0 (%)	The percentage of total reads that have a mapping quality equal to 0
Targets Not Covered	The number of targeted regions of the panel that have coverage below the specified value of 1
On Target Reads (%)	The percentage of filtered aligned reads that are overlapping on all regions defined in the panel
On And Near Target Reads (%)	The percentage of filtered aligned reads that are overlapping on all regions defined in the panel by 250bp in both directions
Off Target Reads (%)	The percentage of filtered aligned reads that are not overlapping on-target or near-on-target regions defined in the panel
Uniformity	The fold of additional sequencing required to ensure that 80% of the target bases achieve the mean coverage
Pipeline-Calculated Sample Sex	The chromosomal sex predicted based on the coverage of targeted regions from sex chromosomes
Customer-Provided Sex	Sex of the sample as provided by the customer
Derivative Log Ratio Score for IRD System	Derivative spread of the log2 ratios of comparative read depth values used for CNV calling

Appendix 7: QC Thresholds

Name	Pass
Mapped Reads (%)*	>=95.0
Mean Target Coverage	>=240.0
Low Coverage (>=100x)	>=95.0
Evenness	>=75.0
Targets Not Covered*	<=30.0
On Target Reads (%)	>=60.0

* Metric not included in sample pass/fail criteria

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