PrismGuide IRD Panel Summary Report (Prioritized Variants)

QC Status: Pass

Order ID: posi_con

Sample Information

Clinical Sample ID	posi_con

SNV/Indel Information

#	Gene Name	Inheritance Pattern	Mutant Allele Frequency		Mutation Content		Consequence	
			Frequency (Read)	Zygosity	HGVSc	HGVSp	ACMG Classification	ACMG labels
1	ABCA4	Recessive	53.29% (721/1353)	Heterozygous	c.5318C>T	p.Ala1773Val	Pathogenic	PS1,PS3,PM2,PP2,PP3
2	ABCA4	Recessive	53.70% (610/1136)	Heterozygous	c.1760+2T>G	-	Pathogenic	PVS1,PS3,PM2,PP3,PP5
3	RPE65	Recessive, Dominant	53.73% (663/1234)	Heterozygous	c.1154C>T	p.Thr385Met	Pathogenic	PS1,PS3,PM2,PP2,PP3
4	RPGR	X-linked	98.68% (149/151)	Hemizygous	c.2838_2839del	p.Glu947GlyfsTer131	Pathogenic	PVS1,PS3,PM2
5	CEP290	Recessive	24.35% (158/649)	Heterozygous	c.2991+1655A>G	-	Likely pathogenic	PS3,PM2

SNV/Indel Report:

1. ABCA4のAla1773Valはヘテロ であり、潜性(劣性)の病的変異である。
2. ABCA4のc.1760+2T>Gはヘテロ であり、潜性(劣性)の病的変異である。
3. RPE65のThr385Metはヘテロ であり、潜性(劣性)または顕性(優性)の病的変異である。
4. RPGRのGlu947GlyfsTer131はヘミ であり、X-linkedの病的変異である。
5. CEP290のc.2991+1655A>Gはヘテロ であり、潜性(劣性)の病的変異の可能性がある。

SNV/Indel Notes:

Sample Notes:

Report Date: Confirmation Sign:

Databases/Software Used

Database/Software	Version Used
PFAM	2024-04-02
Ensembl VEP cache 1000 Genomes	phase3
Ensembl VEP cache SIFT	6.2.1
Benign Variants from PMID:31213501	v1.0.0
Ensembl VEP cache dbSNP	156
Ensembl VEP cache RefSeq	GCF_000001405.40-RS_2023_03
freebayes	1.3.2-38-g71a3e1c-dirty
Custom Variant List	v1.0.0
GEMJ	2021-12-10
multiqc	1.8
ClinVar	2024-06-12
Human gene annotation	2023-10-08
fastp	0.20.0
vep	111.0
dbscSNV	2015-04-13
Ensembl VEP cache assembly	GRCh38.p14
samtools	1.9
Ensembl VEP cache	2024-04-01
Repeat Masker database	2022-10-19
BLOSUM62	111
bwa	0.7.17-r1188
Pathogenic Variants from PMID:31213501	v1.0.0
ogtkit	0.8.6
CNV references	v2.0.0
HGMD	v2024.1
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_45

免責事項

本検査に基づく解析レポートに関して、医療機関が自己の責任で適応性、妥当性、適時性などを判断の上、活用するものとする。

シスメックス株式会社

神戸市中央区脇浜海岸通1-5-1 〒651-0073

Appendix 1. 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