Patient Name:
Date of Birth:
Gender:
Accession ID:
Cross Reference:

Specimen Type:

Receive Date:
Collection Date:
Report Date:

Client Name:

Mailing Address:

Phone Number:

Client Name:

Test Performed: Whole Exome Sequencing and Deletion/Duplication Analysis, Trio

No pathogenic sequence variant(s) in gene related to reported phenotype detected .

Relevant Findings and Interpretation

Sequence variants related to phenotype:

Nr.	Classification	Gene	Exon/ Intron	SNP ID	DNA change	Protein change	Chromosomal location	Zygosity	Inheri-tance	Disease Groups	Associated Disease(s)
1	Benign/Likely benign	RPE65	1	rs12145904	c.1056G>A	p.Glu352=	Chr1:68903942	heterozygous	AR;AD	Neurological diseases;Ophthalmological conditions;Prenatal screening panel	Recessive;Retinitis pigmentosa;Leber congenital amaurosis 2;Leber congenital amaurosis

Gene Name: **RPE65**. SNPs in Gene: **rs12145904**. Associated diseases: Recessive;Retinitis pigmentosa;Leber congenital amaurosis 2;Leber congenital amaurosis;Leber congenital amaurosis;retinitis pigmentosa
Description:
(Mut.Nbr. 1):**RPE65 c.1056G>A (p.Glu352=) - Retinitis pigmentosa** This variant was observed in the ICSL laboratory as part of a predisposition screen in an ostensibly healthy population. It had not been previously curated by ICSL or reported in the Human Gene Mutation Database (HGMD: prior to June 1st, 2018), and was therefore a candidate for classification through an automated scoring system. Utilizing variant allele frequency, disease prevalence and penetrance estimates, and inheritance mode, an automated score was calculated to assess if this variant is too frequent to cause the disease. Based on the score and internal cut-off values, a variant classified as benign is not then subjected to further curation. The score for this variant resulted in a classification of benign for this disease.
(Mut.Nbr. 1):**RPE65 c.1056G>A (p.Glu352=) - Leber congenital amaurosis 2** This variant was observed in the ICSL laboratory as part of a predisposition screen in an ostensibly healthy population. It had not been previously curated by ICSL or reported in the Human Gene Mutation Database (HGMD: prior to June 1st, 2018), and was therefore a candidate for classification through an automated scoring system. Utilizing variant allele frequency, disease prevalence and penetrance estimates, and inheritance mode, an automated score was calculated to assess if this variant is too frequent to cause the disease. Based on the score and internal cut-off values, a variant classified as benign is not then subjected to further curation. The score for this variant resulted in a classification of benign for this disease.
(Mut.Nbr. 1):**RPE65 c.1056G>A (p.Glu352=) - Leber congenital amaurosis** This variant was observed in the ICSL laboratory as part of a predisposition screen in an ostensibly healthy population. It had not been previously curated by ICSL or reported in the Human Gene Mutation Database (HGMD: prior to June 1st, 2018), and was therefore a candidate for classification through an automated scoring system. Utilizing variant allele frequency, disease prevalence and penetrance estimates, and inheritance mode, an automated score was calculated to assess if this variant is too frequent to cause the disease. Based on the score and internal cut-off values, a variant classified as benign is not then subjected to further curation. The score for this variant resulted in a classification of benign for this disease.

Variant Statistics:

Mutation Number	Gene	DNA change	Transcript ID	AF in patient	Nbr. Reads	Quality	Frequency globally	Maximal AF globally	Population with maximal AF
1	RPE65	c.1056G>A	ENST00000262340.5	63.64%	33	53	15.33%	40.39%	East Asian

Note: Quality Score: <10 is low quality; 10-20 is medium quality; >20 is high quality

Relevant Articles:

Number	Name of Mutation	Pubmed ID	Title
1	RPE65 c.1056G>A (p.Glu352=)	22509104	Novel RPE65 mutations associated with Leber congenital amaurosis in Chinese patients.
1	RPE65 c.1056G>A (p.Glu352=)	25383945	A novel mutation in the RPE65 gene causing Leber congenital amaurosis and its transcriptional expression in vitro.
1	RPE65 c.1056G>A (p.Glu352=)	25741868	Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology.