

Updated Interpretations

GENE	VARIANT	ZYGOSITY	PRIOR VARIANT CLASSIFICATION	NEW VARIANT CLASSIFICATION
USH2A	c.14413G>A (p.Val4805Ile)	heterozygous	Uncertain Significance	Likely Benign



RESULT: POSITIVE

One homozygous Pathogenic variant identified in ABCA4. ABCA4 is associated with autosomal recessive inherited retinal disease.

Additional Variant(s) of Uncertain Significance identified.

GENE	VARIANT	ZYGOSITY	VARIANT CLASSIFICATION
ABCA4	c.5714+5G>A (Intronic)	homozygous	PATHOGENIC
CACNA1F	c.3901A>G (p.Met1301Val)	heterozygous	Uncertain Significance
EYS	c.2027C>T (p.Thr676Met)	heterozygous	Uncertain Significance
KCNV2	c.731G>A (p.Arg244His)	heterozygous	Uncertain Significance
KLHL7	c.1372A>C (p.Thr458Pro)	heterozygous	Uncertain Significance
MKKS	c.1161+3A>G (Intronic)	heterozygous	Uncertain Significance
USH2A	c.14531C>T (p.Thr4844Met)	heterozygous	Uncertain Significance