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Variant: NM_000350.3(ABCA4):c.5917del (p.Gly1972_Val1973insTer)

Version: 1.0

CA227355

99419 (ClinVar)

Gene: ABCA4 (HGNC:24)
Condition: ABCA4-related retinopathy (MONDO:0800406)
Inheritance Mode: Autosomal recessive inheritance
UUID: 637cbdd5-017d-4c22-90d6-8cc2d4c9f77c
Approved on: 2026/7/28
Published on: 2026/7/28

HGVS expressions

NM_000350.3:c.5917del
NM_000350.3(ABCA4):c.5917del (p.Gly1972_Val1973insTer)
NC_000001.11:g.94007722del
CM000663.2:g.94007722del
NC_000001.10:g.94473278del
CM000663.1:g.94473278del
NC_000001.9:g.94245866del
NG_009073.1:g.118428del
ENST00000370225.4:c.5917del
ENST00000370225.3:c.5917del
ENST00000465352.1:n.333del
ENST00000484388.1:n.31del
ENST00000536513.5:c.2293del
NM_000350.2:c.5917del

Pathogenic

Met criteria codes 3

PS4 PVS1 PM2_Supporting

Evidence Links 0

Expert Panel

ABCA4 VCEP

Criteria Specification Information

Criteria Specification: *ClinGen ABCA4 Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for ABCA4 Version 1.0*
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

ABCA4 VCEP

The NM_000350.3(ABCA4):c.5917del; p.Val1973Ter variant in ABCA4 is a nonsense variant predicted to cause a premature stop codon in biologically relevant exon 43 of 50 leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1). The total minor allele frequency in gnomAD v4.1.0 is 0.00001363 (22/1613736 alleles), which is lower than the ClinGen

ABCA4 VCEP's threshold for PM2_Supporting (<0.0001). The prevalence of the variant in affected individuals is significantly increased compared with the prevalence in controls with an OR of 537.2 and CI is 145.66 to 5177.53, which is above the ABCA4 VCEP threshold of ≥5, where the CI does not contain 1 (PS4; PMID: 35120629). In summary, this variant meets the criteria to be classified as pathogenic for ABCA4-related retinopathy based on the ACMG/AMP criteria applied, as specified by the ClinGen ABCA4 VCEP (Specification Version 1.0): PVS1, PS4, PM2_Supporting.

Met criteria codes

PS4	✔	The prevalence of the variant in affected individuals is significantly increased compared with the prevalence in controls. The OR is 537.2 and the CI is 145.66 to 5177.53, which is above the ABCA4 VCEP threshold of ≥5, where the CI does not contain 1 (PS4; PMID: 35120629).
PVS1	✔	This is a frameshift variant located in exon 43 out of 50, and introduces a premature stop codon between codons 1-2255 (PVS1).
PM2_Supporting	✔	The total minor allele frequency in gnomAD v4.1.0 is 0.00001363 (22/1613736 alleles), which is lower than the ClinGen ABCA4 VCEP's threshold for PM2_Supporting (<0.0001), meeting this criterion (PM2_Supporting).

Curation History

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