

Variant: *NM_000350.3:c.885del*

Version: 1.0

CA958684 [↗](#)

438109 (ClinVar) [↗](#)

Gene: ABCA4 ([HGNC:24](#))

Condition: ABCA4-related retinopathy ([MONDO:0800406](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 65bb8769-0196-4a65-9d3c-fe9ef1f9123e

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HGVS expressions

NM_000350.3:c.885del

NC_000001.11:g.94080692del

CM000663.2:g.94080692del

NC_000001.10:g.94546248del

CM000663.1:g.94546248del

NC_000001.9:g.94318836del

NG_009073.1:g.45458del

ENST00000370225.4:c.885del

ENST00000649773.1:c.885del

ENST00000370225.3:c.885del

NM_000350.2:c.885del

Pathogenic

Met criteria codes **3**

PM2_Supporting

PVS1

PS4

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:c.885del (p.Leu296CysfsTer4) variant in ABCA4 is a frameshift variant located in exon 8 of 50 and introduces a premature stop codon between codons 1-2255 (PVS1). The total minor allele frequency in gnomAD v4.1.1 is 0.000008673 (14/1614150 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 75.6 and CI of 22.24 to 397.38, 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

PM2_Supporting			The total minor allele frequency in gnomAD v4.1.1 is 0.000008673 (14/1614150 alleles), which is lower than the ClinGen ABCA4 VCEP's threshold for PM2_Supporting (<0.0001), meeting this criterion (PM2_Supporting).
PVS1			This is a frameshift variant located in exon 8 out of 50, and introduces a premature stop codon between codons 1-2255 (PVS1).
PS4			The prevalence of the variant in affected individuals is significantly increased compared with the prevalence in controls. The OR is 75.6 and the CI is 22.24 to 397.38, which is above the ABCA4 VCEP threshold of ≥ 5 , where the CI does not contain 1 (PS4; PMID: 35120629).

Curation History

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