

Variant: *NM_000350.3(ABCA4):c.1A>G (p.Met1Val)*

Version: 1.0

CA226968 [↗](#)

99110 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: d3db3e58-a4b9-4a6b-890c-f977fdf6b7e2

Approved on: 2025/12/5

Published on: 2025/12/23

HGVS expressions

NM_000350.3:c.1A>G

NM_000350.3(ABCA4):c.1A>G (p.Met1Val)

NC_000001.11:g.94121045T>C

CM000663.2:g.94121045T>C

NC_000001.10:g.94586601T>C

CM000663.1:g.94586601T>C

NC_000001.9:g.94359189T>C

NG_009073.1:g.5105A>G

ENST00000370225.4:c.1A>G

ENST00000649773.1:c.1A>G

ENST00000370225.3:c.1A>G

NM_000350.2:c.1A>G

Pathogenic

Met criteria codes **3**

PVS1

PM2_Supporting

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 1.0.0*

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

ABCA4 VCEP

The NM_000350.3:c.1A>G (p.Met1Val) variant in ABCA4 may cause a truncated or absent protein by altering the start codon of the coding sequence and is predicted to lead to the omission of a critical region of the protein (PVS1). The total minor allele frequency in gnomAD v4.1.0 is 0.000004957 (8/1613996 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 131.2 and the CI is 19.05-5442.73, 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): PVS1, PS4, PM2_Supporting.

Met criteria codes

PVS1			The NM_000350.3:c.1A>G (p.Met1Val) variant in ABCA4 may cause a truncated or absent protein by altering the start codon of the coding sequence and is predicted to lead to the omission of a critical region of the protein (PVS1).
PM2_Supporting			The total minor allele frequency in gnomAD v4.1.0 is 0.000004957 (8/1613996 alleles), which is lower than the ClinGen ABCA4 VCEP’s threshold for PM2_Supporting (<0.0001), meeting this criterion (PM2_Supporting).
PS4			The prevalence of the variant in affected individuals is significantly increased compared with the prevalence in controls. The OR is 131.2 and the CI is 19.05-5442.73, 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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