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Variant: NM_000350.3(ABCA4):c.2900C>T (p.Ala967Val)

Version: 1.0

CA341275316

957296 (ClinVar)

Gene: ABCA4 (HGNC:24)
Condition: ABCA4-related retinopathy (MONDO:0800406)
Inheritance Mode: Autosomal recessive inheritance
UUID: 8afe714d-cb35-4495-b0b5-cc3504ced18e
Approved on: 2026/5/26
Published on: 2026/5/26

HGVS expressions

NM_000350.3:c.2900C>T
NM_000350.3(ABCA4):c.2900C>T (p.Ala967Val)
NC_000001.11:g.94046937G>A
CM000663.2:g.94046937G>A
NC_000001.10:g.94512493G>A
CM000663.1:g.94512493G>A
NC_000001.9:g.94285081G>A
NG_009073.1:g.79213C>T
ENST00000370225.4:c.2900C>T
ENST00000649773.1:c.2678C>T
ENST00000370225.3:c.2900C>T
ENST00000536513.5:c.-64-6848C>T
NM_000350.2:c.2900C>T

Likely Pathogenic

Met criteria codes 5

PM2_Supporting PP4 PS4 PP3_Moderate PM3_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 1.0.0*
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

ABCA4 VCEP

The NM_000350.3(ABCA4):c.2900C>T variant in ABCA4 is a missense variant predicted to cause substitution of Alanine by Valine at amino acid 967; p.Ala967Val. This variant is absent from gnomAD v4.1.0 meeting PM2_Supporting. The computational predictor REVEL gives a score of 0.956 which is above the threshold of >0.772 meeting PP3_Moderate. The prevalence of the variant in affected individuals is significantly increased compared with the prevalence in controls with an OR of infinity and CI of 2.24-infinity (PS4; PMID: 35120629). At least one patient with this variant displayed presence of at least two ABCA4 variants, onset under 18 years of age, macular flecks on

imaging, macular atrophy, and decreased central visual acuity, which is highly specific for ABCA4-related retinopathy (PP4; PMID: 31934596). This variant has been detected in at least 1 individual with ABCA4-related retinopathy who was homozygous for the variant (PM3_Supporting; PMID: 31934596). In summary, this variant meets the criteria to be classified as likely pathogenic for ABCA4-related retinopathy based on the ACMG/AMP criteria applied, as specified by the ClinGen ABCA4 VCEP specification version 1.0.0: PS4, PP4, PM3_Supporting, PP3_Moderate, PM2_Supporting.

Met criteria codes

PM2_Supporting	✔	This variant is absent from gnomAD v4.1.0 (PM2_Supporting).
PP4	✔	At least one patient with this variant displayed presence of at least two ABCA4 variants, onset under 18 years of age, macular flecks on imaging, macular atrophy, and decreased central visual acuity, which is highly specific for ABCA4-related retinopathy (PP4; PMID: 31934596).
PS4	✔	The prevalence of the variant in affected individuals is significantly increased compared with the prevalence in controls. The OR is infinity and the CI is 2.24-infinity, which is above the ABCA4 VCEP threshold of ≥5, where the CI does not contain 1 (PS4; PMID: 35120629).
PP3_Moderate	✔	The computational predictor REVEL gives a score of 0.956 which is above the threshold of >0.772, evidence that predicts a damaging effect on ABCA4 function (PP3_Moderate).
PM3_Supporting	✔	This variant has been detected in at least 1 individual with ABCA4-related retinopathy. This individual was homozygous for the variant (0.5 points; PM3_Supporting; PMID: 31934596).

Curation History

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