The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000350.3:c.6166A>T

Version: 1.0

CA227371

99432 (ClinVar)

Gene: ABCA4 (HGNC:24)
Condition: ABCA4-related retinopathy (MONDO:0800406)
Inheritance Mode: Autosomal recessive inheritance
UUID: 4d2347da-34b7-4d23-b835-9b9b20da864f
Approved on: 2026/7/28
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HGVS expressions

NM_000350.3:c.6166A>T
NC_000001.11:g.94001974T>A
CM000663.2:g.94001974T>A
NC_000001.10:g.94467530T>A
CM000663.1:g.94467530T>A
NC_000001.9:g.94240118T>A
NG_009073.1:g.124176A>T
ENST00000370225.4:c.6166A>T
ENST00000370225.3:c.6166A>T
ENST00000465352.1:n.582A>T
ENST00000536513.5:c.2542A>T
NM_000350.2:c.6166A>T

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.6166A>T, p.Lys2056Ter variant in ABCA4 is a nonsense variant predicted to cause a premature stop codon in biologically relevant exon 45 of 50 leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1). This variant is absent from gnomAD v4.1.1 (PM2_Supporting). 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 10.92 to Infinity, 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

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 10.92 to Infinity, which is above the ABCA4 VCEP threshold of ≥5, where the CI does not contain 1 (PS4; PMID: 35120629).
PVS1	✔	This is a nonsense variant located in exon 45 out of 50, and introduces a premature stop codon between codons 1-2255 (PVS1).
PM2_Supporting	✔	This variant is absent from gnomAD v4.1.1 (PM2_Supporting).

Curation History

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