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(ABCA4):c.6213C>G (p.Tyr2071Ter)

Version: 1.0

CA341278268

866298 (ClinVar)

Gene: ABCA4 (HGNC:24)
Condition: ABCA4-related retinopathy (MONDO:0800406)
Inheritance Mode: Autosomal recessive inheritance
UUID: a7775518-d499-407b-8f43-e50d653b4edf
Approved on: 2026/5/26
Published on: 2026/5/26

HGVS expressions

NM_000350.3(ABCA4):c.6213C>G (p.Tyr2071Ter)
NM_000350.3:c.6213C>G
NC_000001.11:g.94001927G>C
CM000663.2:g.94001927G>C
NC_000001.10:g.94467483G>C
CM000663.1:g.94467483G>C
NC_000001.9:g.94240071G>C
NG_009073.1:g.124223C>G
ENST00000370225.4:c.6213C>G
ENST00000370225.3:c.6213C>G
ENST00000465352.1:n.629C>G
ENST00000536513.5:c.2589C>G
NM_000350.2:c.6213C>G

Pathogenic

Met criteria codes 3

PM3 PVS1 PM2_Supporting

Not Met criteria codes 1

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.6213C>G; p.p.Tyr2071Ter variant in ABCA4 is a nonsense variant predicted to cause a premature stop codon in biologically relevant exon 45/50 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 0.0000006195 (1/1614190 alleles), which is lower than the ClinGen ABCA4 VCEP's threshold for PM2_Supporting (<0.0001). This variant has been detected in at least 1 individual with ABCA4-related retinopathy. This individual was compound heterozygous for the variant and another pathogenic variant and was confirmed in trans

(c.6319C>T and c.6213C>G; PMID: 26377081; PM3). 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 1.0.0): PVS1, PM2_Supporting, PM3.

Met criteria codes

PM3	✔	This variant has been detected in at least 1 individual with ABCA4-related retinopathy. This individual was compound heterozygous for the variant and another pathogenic variant and was confirmed in trans (participant 90 has c.6319C>T and c.6213C>G; PM3 +1; PMID: 26377081).
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	✔	The total minor allele frequency in gnomAD v4.1.0 is 0.0000006195 (1/1614190 alleles), which is lower than the ClinGen ABCA4 VCEP's threshold for PM2_Supporting (<0.0001).

Not Met criteria codes

PS4	✖	The prevalence of the variant in affected individuals is increased compared with the prevalence in controls. The OR is infinity, but the CI is [0.31, infinity], which does contain 1, so PS4 is not met (PMID: 35120629).

Curation History

Showing 1 to 1 of 1 rows

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.

ClinGen Terms of Use.
¤ Powered by BCM's Genboree.