

Variant: NM_000350.3(ABCA4):c.6088C>T (p.Arg2030Ter)

Version: 1.0

CA119140 [↗](#)

7907 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 49a4d768-480e-4162-8cba-a6db3f65a865

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HGVS expressions

NM_000350.3(ABCA4):c.6088C>T (p.Arg2030Ter)

NM_000350.3:c.6088C>T

NC_000001.11:g.94005500G>A

CM000663.2:g.94005500G>A

NC_000001.10:g.94471056G>A

CM000663.1:g.94471056G>A

NC_000001.9:g.94243644G>A

NG_009073.1:g.120650C>T

ENST00000370225.4:c.6088C>T

ENST00000370225.3:c.6088C>T

ENST00000465352.1:n.504C>T

ENST00000484388.1:n.202C>T

ENST00000536513.5:c.2464C>T

NM_000350.2:c.6088C>T

Pathogenic

Met criteria codes **3**

PM2_Supporting

PS4

PVS1

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

NM_000350.3(ABCA4):c.6088C>T; p.Arg2030Ter variant in ABCA4 is a nonsense variant predicted to cause a premature stop codon in biologically relevant exon 44 of 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 v4.1.1 is 0.00001425, which is lower than the threshold (<0.0001) for PM2_Supporting. The prevalence of the variant in affected individuals is significantly increased compared with the prevalence in controls

with an OR of 195.4 and CI of 62.90 to 953.37, 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), as specified by the ClinGen ABCA4 Variant Curation Expert Panel: PVS1, PS4, PM2_Supporting.

Met criteria codes

PM2_Supporting			The total minor allele frequency in gnomAD v4.1.1 is 0.00001425, 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 195.4 and the CI is 62.90 to 953.37, 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 44 out of 50, and introduces a premature stop codon between codons 1-2255 (PVS1).

Curation History



 

 

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