

Variant: *NM_000350.3(ABCA4):c.4354G>T (p.Glu1452Ter)*

Version: 1.0

CA10602429 [↗](#)

236114 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: c7c4859f-11f7-434c-9ea3-58fb107410f9

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HGVS expressions

NM_000350.3:c.4354G>T

NM_000350.3(ABCA4):c.4354G>T (p.Glu1452Ter)

NC_000001.11:g.94029630C>A

CM000663.2:g.94029630C>A

NC_000001.10:g.94495186C>A

CM000663.1:g.94495186C>A

NC_000001.9:g.94267774C>A

NG_009073.1:g.96520G>T

ENST00000370225.4:c.4354G>T

ENST00000370225.3:c.4354G>T

ENST00000536513.5:c.730G>T

NM_000350.2:c.4354G>T

Pathogenic

Met criteria codes **3**

PVS1

PM3_Supporting

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(ABCA4):c.4354G>T; p.Glu1452Ter variant in ABCA4 is a nonsense variant predicted to cause a premature stop codon in biologically relevant exon 30 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 v4.1.0 (PM2_Supporting). This variant has been detected in at least one individual with ABCA4-related retinopathy who was compound heterozygous for the variant and a pathogenic variant with phase unconfirmed (PM3_Supporting, PMID: 37705246). 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, PM3_Supporting, PM2_Supporting.

Met criteria codes

PVS1			This is a nonsense variant located in exon 30 out of 50, and introduces a premature stop codon between codons 1-2255 (PVS1).
PM3_Supporting			This variant has been detected in at least one individual with ABCA4-related retinopathy was compound heterozygous for the variant and a pathogenic variant with phase unconfirmed (PM3 = 0.5 pts, PM3_Supporting, PMID: 37705246)
PM2_Supporting			This variant is absent from gnomAD v4.1.0 (PM2_Supporting).

Not Met criteria codes

PS4			This variant is not in the Cornelis et al paper. (PMID: 35120629).
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Curation History

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