

Variant: *NM_000350.3(ABCA4):c.5406C>T (p.Ile1802=)*

Version: 1.0

CA957287 [↗](#)

298228 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 40900869-df13-45d5-bb67-5fe479e54595

Approved on: 2025/10/28

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HGVS expressions

NM_000350.3(ABCA4):c.5406C>T (p.Ile1802=)

NM_000350.3:c.5406C>T

NC_000001.11:g.94014597G>A

CM000663.2:g.94014597G>A

NC_000001.10:g.94480153G>A

CM000663.1:g.94480153G>A

NC_000001.9:g.94252741G>A

NG_009073.1:g.111553C>T

ENST00000370225.4:c.5406C>T

ENST00000370225.3:c.5406C>T

ENST00000536513.5:c.1782C>T

NM_000350.2:c.5406C>T

Uncertain Significance

Met criteria codes **1**

PM2_Supporting

Not Met criteria codes **4**

BP4

BP7

PP4

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.5406C>T is a synonymous variant (p.Ile1802=). The total minor allele frequency in gnomAD v4.1.0 is 0.000039652 (64/1614042 alleles), which is lower than the ClinGen ABCA4 VCEP's threshold for PM2_Supporting (<0.0001). The SpliceAI predictor score for this variant is 0.13, which is greater than 0.1, the ABCA4 VCEP's threshold for BP7 or BP4 criterion. To our knowledge, this variant has not been reported in the literature in any individuals with ABCA4-related retinopathy. In summary, this variant meets the criteria to be classified as a variant of uncertain significance for ABCA4-related retinopathy based on the ACMG/AMP criteria applied, as specified by the ClinGen ABCA4 VCEP (Specification Version 1.0.0): PM2_Supporting.

Met criteria codes

PM2_Supporting

i

✓

The total minor allele frequency in gnomAD v4.1.0 is [0.000039652] (64/1614042 alleles), which is lower than the ClinGen ABCA4 VCEP’s threshold for PM2_Supporting (<0.0001), meeting this criterion (PM2_Supporting).

Not Met criteria codes

BP4

i

✗

Splice AI is greater than 0.1. BP4 not met.

BP7

i

✗

This variant has a SpliceAI score of greater than 0.1, which is above ABCA4 VCEP's threshold for BP7.

PP4

i

✗

To our knowledge, this variant has not been reported in the literature in any individuals with ABCA4-related retinopathy.

PS4

i

✗

This variant is not found in the Cornelis et al. paper (PMID: 35120629), so an OR has not been calculated. PS4 is not met.

Curation History

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