

Variant: NM_000350.3(ABCA4):c.6764G>T (p.Ser2255Ile)

Version: 1.0

CA202970 [↗](#)

99494 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 7be97473-8c9c-46e3-9210-68e35172db63

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HGVS expressions

NM_000350.3(ABCA4):c.6764G>T (p.Ser2255Ile)

NM_000350.3:c.6764G>T

NC_000001.11:g.93996161C>A

CM000663.2:g.93996161C>A

NC_000001.10:g.94461717C>A

CM000663.1:g.94461717C>A

NC_000001.9:g.94234305C>A

NG_009073.1:g.129989G>T

ENST00000370225.4:c.6764G>T

ENST00000370225.3:c.6764G>T

ENST00000536513.5:c.3140G>T

NM_000350.2:c.6764G>T

Benign

Met criteria codes 2

BA1

BP4

Not Met criteria codes 2

PS4

PM3

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.6764G>T variant in ABCA4 is a missense variant predicted to cause substitution of serine by isoleucine at amino acid 2255 (p.Ser2255Ile). The GroupMax filtering allele frequency in gnomAD v4.1.0 is 0.4845, which is greater than the ClinGen ABCA4 VCEP's threshold for BA1 (>0.163). The computational predictor REVEL gives a score of 0.2 which is in the range of 0.184-0.290, and the variant has a SpliceAI donor gain score of 0.01, which is below the 0.1 cutoff meeting BP4. In summary, this variant meets the criteria to be classified as benign for ABCA4-related retinopathy based on the ACMG/AMP criteria applied, as specified by the ClinGen ABCA4 VCEP (Specification Version 1.0.0): BA1, BP4.

Met criteria codes

BA1			The GroupMax filtering allele frequency in gnomAD v4.1.0 is 0.4852 (African/African American exomes), which is greater than the ClinGen ABCA4 VCEP's threshold for BA1 (>0.163), and therefore meets this criterion (BA1).
BP4			The variant has a REVEL score of 0.2, which meets the threshold of <0.290 for this code to be applicable as determined by the ABCA4 VCEP. SpliceAI donor gain score of 0.01, which is below the 0.1 cutoff.

Not Met criteria codes

PS4			Variant listed in Cornelis et al. 2022 paper as having OR of 0, and CI of 0.00 to 0.02. Therefore, this variant does not meet criteria for PS4 as specified by the ClinGen ABCA4 VCEP. (PMID: 35120629)
PM3			Reported in 11 year old male patient with Stargardt disease/fundus flavimaculatus with pathogenic co-variant ABCA4:c.5882G>A (PMID: 31618812). However, PM3 will not be applied due to the minor allele frequency being above threshold for disease and BA1 is met.

Curation History



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