

Variant: *NM_000350.3(ABCA4):c.6285T>C (p.Asp2095=)*

Version: 1.0

CA285825 [↗](#)

99444 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: ab6237c1-841a-4f44-a1ab-2dcf9c825788

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HGVS expressions

NM_000350.3:c.6285T>C

NM_000350.3(ABCA4):c.6285T>C (p.Asp2095=)

NC_000001.11:g.94001103A>G

CM000663.2:g.94001103A>G

NC_000001.10:g.94466659A>G

CM000663.1:g.94466659A>G

NC_000001.9:g.94239247A>G

NG_009073.1:g.125047T>C

ENST00000370225.4:c.6285T>C

ENST00000370225.3:c.6285T>C

ENST00000536513.5:c.2661T>C

NM_000350.2:c.6285T>C

Benign

Met criteria codes **2**

BP7 **BA1**

Not Met criteria codes **3**

PS4 **BP4** **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.6285T>C variant is a synonymous variant, p.Asp2095=. The GroupMax filtering allele frequency in gnomAD v4.1.0 is 0.6547, which is greater than the ClinGen ABCA4 VCEP's threshold for BA1 (>0.163). The computational splicing predictor SpliceAI gives this synonymous variant a delta score of 0.03 for donor gain, which is below the ClinGen ABCA4 VCEP BP7 threshold of <0.1 and does not strongly predict a splicing defect (BP7_Moderate). 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, BP7_Moderate.

Met criteria codes

BP7		✓	The computational splicing predictor SpliceAI gives this synonymous (silent) variant a delta score of 0.03 for donor gain which is below the ClinGen ABCA4 VCEP BP7 threshold of <0.1 and does not strongly predict a splicing defect. To our knowledge, there is no known conflicting minigene or other functional data available (BP7_Moderate).
BA1		✓	The GroupMax filtering allele frequency in gnomAD v4.1.0 is 0.6547, which is greater than the ClinGen ABCA4 VCEP's threshold for BA1 (>0.163), and therefore meets this criterion (BA1).

Not Met criteria codes

PS4		✗	The prevalence of the variant in affected individuals is not significantly increased compared with the prevalence in controls. The OR is 0 and the CI is 0-0 (PMID: 35120629).
BP4		✗	The computational predictor CADD gives a score of 0.305 which is less than or equal to the threshold of ≤17.3, evidence that does not predict a damaging effect on ABCA4 function (BP4_Moderate). This code will not be applied since BP7 is being applied at the moderate weight instead.
PM3		✗	This variant has been detected in at least 94 individuals with ABCA4-related retinopathy. Of those individuals, 2 were compound heterozygous for the variant and a pathogenic variant (c.6445C>T p.(R2149X); c.5882G>A p.(Gly1961Glu)) and neither of those were confirmed in trans (PM3= 1 point; PMIDs: 20801516, 28002570, 28118664). However, since this variant meets BA1, PM3 is not applicable.

Curation History



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Showing 1 to 1 of 1 rows

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