

Variant: NM\_000350.3(ABCA4):c.5753A>G (p.Asp1918Gly)

Version: 1.0

CA341280424 [↗](#)

1481126 (ClinVar) [↗](#)

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

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

**Inheritance Mode:** Autosomal recessive inheritance

**UUID:** b2e8e9b3-bc76-402c-9403-7ada994befae

**Approved on:** 2026/5/26

**Published on:** 2026/5/26

### HGVS expressions

**NM\_000350.3(ABCA4):c.5753A>G (p.Asp1918Gly)**

**NM\_000350.3:c.5753A>G**

NC\_000001.11:g.94008833T>C

CM000663.2:g.94008833T>C

NC\_000001.10:g.94474389T>C

CM000663.1:g.94474389T>C

NC\_000001.9:g.94246977T>C

NG\_009073.1:g.117317A>G

ENST00000370225.4:c.5753A>G

ENST00000370225.3:c.5753A>G

ENST00000465352.1:n.169A>G

ENST00000536513.5:c.2129A>G

NM\_000350.2:c.5753A>G

**Likely Pathogenic**

**Met criteria codes** 4

PM2\_Supporting

PP3\_Moderate

PM5

PM3\_Supporting

**Not Met criteria codes** 2

PS4

PP4

**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.5753A>G variant in ABCA4 is a missense variant predicted to cause substitution of aspartic acid by glycine at amino acid 1918; p.Asp1918Gly. This variant is absent from gnomAD v4.1.0 meeting PM2\_Supporting. The computational predictor REVEL gives a score of 0.836 which is above the threshold of >0.772 meeting PP3\_Moderate. This variant has been detected in one individual with ABCA4-related retinopathy who is compound heterozygous for the variant and a pathogenic variant suspected in trans (PM3\_Supporting; PMID: 26492201). Another missense variant, c.5753A>T; p.Asp1918Val (ClinVar Variation ID: 871507), in the same codon has been

classified as pathogenic for ABCA4-related retinopathy by the ClinGen ABCA4 VCEP, and Splice AI revealed no expected effects on splicing (PM5). In summary, this variant meets the criteria to be classified as likely pathogenic for ABCA4-related retinopathy based on the ACMG/AMP criteria applied, as specified by the ClinGen ABCA4 VCEP (Specification Version v.1.0.0): PM3\_Supporting, PP3\_Moderate, PM5, PM2\_Supporting.

Met criteria codes

|                |                                                                                   |                                                                                   |                                                                                                                                                                                                                                                                                                    |
|----------------|-----------------------------------------------------------------------------------|-----------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| PM2_Supporting |  |  | This variant is absent from gnomAD v4.1.0 (PM2_Supporting).                                                                                                                                                                                                                                        |
| PP3_Moderate   |  |  | The computational predictor REVEL gives a score of 0.836 which is above the threshold of >0.772, evidence that predicts a damaging effect on ABCA4 function (PP3_Moderate).                                                                                                                        |
| PM5            |  |  | Another missense variant, c.5753A>T, p.Asp1918Val (PMID: 27939946, ClinVar Variation ID 871507), in the same codon has been classified as pathogenic for ABCA4-related retinopathy by the ClinGen ABCA4 VCEP, and Splice AI (score = 0.1) revealed no expected effects on splicing (PM5_Moderate). |
| PM3_Supporting |  |  | This variant has been detected in one individual with ABCA4-related retinopathy who is compound heterozygous for the variant and a pathogenic variant that is suspected to be in trans (PMID: 26492201, PM3_Supporting).                                                                           |

Not Met criteria codes

|     |                                                                                     |                                                                                     |                                                                                                                                                                                                                                                                                                                                                                        |
|-----|-------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| PS4 |   |   | The variant is not found in the Cornelis et al. paper. (PMID:35120629)                                                                                                                                                                                                                                                                                                 |
| PP4 |  |  | PMID: 26492201. Subject 18: Male, onset at 15 years of age, BCVA of 0.82, Full Field Flash electroretinogram normal result (only the macula is affected). Microarray chip detected compound het c.5882G>A and c.5753A>G in ABCA4. Macular flecks on imaging and/or fundus autofluorescence and/or beaten-bronze appearance not specified for this patient. PP4_Not met |

Curation History 

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