

Variant: *NM_000350.3(ABCA4):c.2879del (p.Ala960fs)*

Version: 1.0

CA2573132716 [↗](#)

1372740 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 3cdd7b12-a832-4d7d-81c0-d8859a606ef7

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HGVS expressions

NM_000350.3(ABCA4):c.2879del (p.Ala960fs)

NM_000350.3:c.2879del

NC_000001.11:g.94046958del

CM000663.2:g.94046958del

NC_000001.10:g.94512514del

CM000663.1:g.94512514del

NC_000001.9:g.94285102del

NG_009073.1:g.79192del

ENST00000370225.4:c.2879del

ENST00000649773.1:c.2657del

ENST00000370225.3:c.2879del

ENST00000536513.5:c.-64-6869del

NM_000350.2:c.2879del

Pathogenic

Met criteria codes **3**

PM3_Supporting

PVS1

PM2_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:c.2879del (p.Ala960AspfsTer17) variant in ABCA4 is a frameshift variant predicted to cause a premature stop codon in biologically relevant exon 19/50 leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1). The total minor allele frequency in gnomAD v4.1.0 is 0.0000006195 (1/1614136 alleles), which is lower than the ClinGen ABCA4 VCEP's threshold for PM2_Supporting (<0.0001). This variant has been detected in at least 1 individual with ABCA4-related retinopathy who was compound heterozygous for the variant and a pathogenic variant (c.6119G>A; p.Arg2040Gln) which was not

confirmed in trans (PM3_Supporting; PMID: 29343940). 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 1): PVS1, PM3_Supporting, PM2_Supporting.

Met criteria codes

PM3_Supporting			This variant has been detected in at least 1 individual with ABCA4-related retinopathy. This individual was compound heterozygous for the variant and a pathogenic variant (c.6119G>A (p.Arg2040Gln)) which was not confirmed in trans (PM3= 0.5 points; PM3_Supporting; PMID: 29343940).
PVS1			This is a frameshift variant located in exon 19 out of 50, and introduces a premature stop codon between codons 1-2255 (PVS1).
PM2_Supporting			The total minor allele frequency in gnomAD v4.1.0 is 0.0000006195 (1/1614136 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

PS4			The prevalence of the variant in affected individuals is not significantly increased compared with the prevalence in controls. The OR is infinity and the CI is 0.31-infinity, which is above the ABCA4 VCEP threshold of ≥ 5 , but the CI does contain 1 (PMID: 35120629).
PP4			At least 1 individual with a diagnosis of Stargardt disease was reported to have this variant, but the phenotype does not meet the phenotype criteria to apply PP4 as specified by the ABCA4 VCEP.

Curation History



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