

Variant: NM_000350.3(ABCA4):c.6601_6602del
(p.Arg2201fs)

Version: 1.0

CA227421 [↗](#)

99472 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 9999a507-2664-439d-b776-c994e5d920cb

Approved on: 2026/2/24

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HGVS expressions

NM_000350.3(ABCA4):c.6601_6602del (p.Arg2201fs)

NM_000350.3:c.6601_6602del

NC_000001.11:g.93997991_93997992del

CM000663.2:g.93997991_93997992del

NC_000001.10:g.94463547_94463548del

CM000663.1:g.94463547_94463548del

NC_000001.9:g.94236135_94236136del

NG_009073.1:g.128161_128162del

ENST00000370225.4:c.6601_6602del

ENST00000370225.3:c.6601_6602del

ENST00000536513.5:c.2977_2978del

NM_000350.2:c.6601_6602del

Pathogenic

Met criteria codes **3**

PS4

PM2_Supporting

PVS1

Not Met criteria codes **2**

PP1

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.6601_6602del (p.Arg2201AlafsTer) variant in ABCA4 is a frameshift variant predicted to cause a premature stop codon in biologically relevant exon 48/50 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 0.0000006196 (1/1614010 alleles), which is lower than the ClinGen ABCA4 VCEP's threshold for PM2_Supporting (<0.0001). The prevalence of the variant in affected individuals is significantly increased compared with the prevalence in controls. The OR is infinity and the CI is 2.24-infinity, which is above the ABCA4 VCEP threshold

of ≥ 5 , where the CI does not contain 1 (PS4; PMID: 35120629). In summary, this variant meets the criteria to be classified as pathogenic for ABCA4-related retinopathy based on the ACMG/AMP criteria applied, as specified by the ClinGen ABCA4 VCEP (Specification Version 1.0.0): PVS1, PS4, PM2_Supporting.

Met criteria codes

PS4			The prevalence of the variant in affected individuals is significantly increased compared with the prevalence in controls. The OR is infinity and the CI is 2.24-infinity, which is above the ABCA4 VCEP threshold of ≥ 5 , where the CI does not contain 1 (PS4; PMID: 35120629).
PM2_Supporting			The total minor allele frequency in gnomAD v4.1.0 is 0.0000006196 (1/1614010 alleles), which is lower than the ClinGen ABCA4 VCEP's threshold for PM2_Supporting (<0.0001), meeting this criterion (PM2_Supporting).
PVS1			This is a frameshift variant located in exon 48 out of 50, and introduces a premature stop codon between codons 1-2255 (PVS1).

Not Met criteria codes

PP1			This variant segregates in 2 family members (siblings), but they do not appear to be affected based on the pedigree.
PP4			This variant has been reported in the literature in several individuals with ABCA4-related retinopathy, but none with phenotypes that meet the requirements of the PP4 code as specified by the ClinGen ABCA4 VCEP (PMIDs: 34440443, 32307445, 20647261, 10958761, 12037008).

Curation History



Showing 1 to 1 of 1 rows

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