

Variant: *NM_000350.3:c.2488G>T*

Version: 2.0

CA341276980 [↗](#)

4538553 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal recessive inheritance

UUID: 8007486e-4071-4423-8da8-cd003542683e

Approved on: 2026/8/25

Published on: 2026/8/25

HGVS expressions

NM_000350.3:c.2488G>T

NC_000001.11:g.94055210C>A

CM000663.2:g.94055210C>A

NC_000001.10:g.94520766C>A

CM000663.1:g.94520766C>A

NC_000001.9:g.94293354C>A

NG_009073.1:g.70940G>T

ENST00000370225.4:c.2488G>T

ENST00000649773.1:c.2266G>T

ENST00000370225.3:c.2488G>T

ENST00000536513.5:c.-65+7964G>T

NM_000350.2:c.2488G>T

Pathogenic

Met criteria codes **3**

PM3_Supporting

PVS1

PM2_Supporting

Not Met criteria codes **1**

PS4

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

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

ABCA4 VCEP

NM_000350.3(ABCA4):c.2488G>T; p.Glu830Ter variant in ABCA4 is a nonsense variant predicted to cause a premature stop codon in biologically relevant exon 16 of 50 leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1). This variant is absent from gnomAD v4.1.1 (PM2_Supporting). This variant has been reported in at least one proband affected with Stargardt disease meeting PS4_Supporting (PMID: 32619608). 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), as specified by the ClinGen ABCA4 Variant Curation Expert Panel: PVS1, PM3_Supporting, PM2_Supporting.

Met criteria codes

PM3_Supporting			Patient MD-0769: STGD1, onset at 41 years, compound het c.2023G>A p.(Val675Ile) and c.2488G>T p.(Glu830*). No segregation. Phase unconfirmed. PM3 = 0.5 pts.
PVS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2_Supporting			This variant is absent from gnomAD v4.1.0 (PM2_Supporting).

Not Met criteria codes

PS4			This variant is listed in Cornelis et al. 2022 with a confidence interval that contains 1. PS4 is not met.
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Curation History

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