

Variant: *NM_000261.2(MYOC):c.39T>G (p.Pro13=)*

Version: 2.0

CA1244342 [↗](#)

293724 (ClinVar) [↗](#)

Gene: MYOC ([HGNC:4653](#))

Condition: open-angle glaucoma ([MONDO:0005338](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 795036f1-b135-4d5a-9011-ac67bf704e4f

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HGVS expressions

NM_000261.2(MYOC):c.39T>G (p.Pro13=)

NM_000261.2:c.39T>G

NC_000001.11:g.171652573A>C

CM000663.2:g.171652573A>C

NC_000001.10:g.171621713A>C

CM000663.1:g.171621713A>C

NC_000001.9:g.169888336A>C

NG_008859.1:g.5061T>G

ENST00000037502.11:c.39T>G

ENST000000638471.1:c.39T>G

ENST00000037502.10:c.39T>G

ENST000000614688.1:c.39T>G

NM_000261.1:c.39T>G

Benign

Met criteria codes 3

BP4 BP7 BA1

Not Met criteria codes 11

PM2 PM5 PM4 BS1 BS3
PS1 PS2 PS3 PS4 PP1 PP3

Evidence Links 0

Expert Panel

[Glaucoma VCEP](#) [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen Glaucoma Expert Panel
Specifications to the ACMG/AMP Variant Interpretation
Guidelines for MYOC Version 2.0.0

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

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

Evidence submitted by expert panel

Glaucoma VCEP

The c.39T>G variant in MYOC is a synonymous variant (p.Pro13=). The highest minor allele frequency of this variant was in the African/African-American genetic ancestry group of gnomAD (v4.1.0) = 0.07326, which met the ≥ 0.01 threshold set for BA1 (5,497 alleles out of 75032, meeting the threshold of ≥ 5 of at least 2,000 observed alleles). The SpliceAI score = 0, which met the ≤ 0.1 threshold for BP4, suggesting that the variant does not impact MYOC function. This synonymous variant meets BP4, so BP7 is met. There was no functional evidence predicting a damaging or benign impact of this variant on MYOC function. As BA1 was met, PP1 did not apply and

segregations were not counted. Although probands with juvenile or primary open angle glaucoma have been reported carrying this variant, PM2_Supporting was not met, therefore PS4 did not apply. In summary, this variant was classified as benign (BA1 is a stand-alone criterion for a benign level of pathogenicity) for juvenile open angle glaucoma based on the ACMG/AMP criteria met, as specified by the ClinGen Glaucoma VCEP (v2.0.0, 5 Dec 2024): BA1, BP4, BP7

Met criteria codes

BP4			The SpliceAI score = 0, which met the ≤ 0.1 threshold for BP4, suggesting that the variant does not impact MYOC function.
BP7			This synonymous variant meets BP4, so BP7 is met.
BA1			The highest minor allele frequency of this variant was in the African/African-American genetic ancestry group of gnomAD (v4.1.0) = 0.07326, which met the ≥ 0.01 threshold set for BA1 (5,497 alleles out of 75032, meeting the threshold of ≥ 5 of at least 2,000 observed alleles).

Not Met criteria codes

PM2			This criterion was not met as BA1 has been met.
PM5			This is not a missense variant.
PM4			This variant does not cause a protein length change.
BS1			This criterion was not met as BA1 has been met.
BS3			No functional evidence has been found for this variant.
PS1			This variant does not involve an amino acid change.
PS2			This variant has not been identified de novo.
PS3			No functional evidence has been found for this variant.
PS4			Although probands with juvenile or primary open angle glaucoma have been reported carrying this variant, PM2_Supporting was not met, therefore PS4 did not apply.
PP1			As BA1 was met, PP1 did not apply and segregations were not counted.
PP3			This is not a missense variant.

Curation History

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