The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000261.2:c.1160G>A

Version: 2.0

CA1244072

2505287 (ClinVar)

Gene: MYOC (HGNC:4653)
Condition: open-angle glaucoma (MONDO:0005338)
Inheritance Mode: Autosomal dominant inheritance
UUID: b9227514-de3a-44ac-8340-f2653214de03
Approved on: 2026/2/19
Published on: 2026/2/18

HGVS expressions

NM_000261.2:c.1160G>A
NC_000001.11:g.171636280C>T
CM000663.2:g.171636280C>T
NC_000001.10:g.171605420C>T
CM000663.1:g.171605420C>T
NC_000001.9:g.169872043C>T
NG_008859.1:g.21354G>A
ENST00000037502.11:c.1160G>A
ENST00000637303.1:c.235-2350C>T
ENST00000638471.1:c.*498G>A
ENST00000037502.10:c.1160G>A
ENST00000614688.1:c.*124G>A
NM_000261.1:c.1160G>A

Likely Pathogenic

Met criteria codes 3

PM2_Supporting PS4_Supporting PP3_Strong

Not Met criteria codes 11

BS1 BS3 BP7 BP4 PM5 BA1 PM4 PS1 PS2 PS3 PP1

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.1160G>A variant in MYOC is a missense variant predicted to cause substitution of Glycine by Aspartic Acid at amino acid 387 (p.Gly387Asp). The highest minor allele frequency of this variant was in the East Asian genetic ancestry group of gnomAD (v4.1.0) = 0.00004455 (2 alleles out of 44,890), which met the ≤ 0.0001 threshold set for PM2_Supporting in a genetic ancestry group of at least 10,000 alleles. The REVEL score = 0.958, which met the ≥ 0.932 threshold for PP3_Strong, predicting a damaging effect on MYOC function. There was no functional evidence predicting a damaging or benign impact of this variant on MYOC function. 2 probands with primary open

angle glaucoma have been reported carrying this variant (PMID: 24825108), which met PS4_Supporting (≥ 2 probands). In summary, this variant met the criteria to receive a score of 6 and to be classified as likely pathogenic (likely pathogenic classification range 6 to 9, adapted from PMID: 32720330) for primary open angle glaucoma based on the ACMG/AMP criteria met, as specified by the ClinGen Glaucoma VCEP (v2.0.0, 5 Dec 2024): PP3_Strong, PS4_Supporting, PM2_Supporting

Met criteria codes

PM2_Supporting	✔	The highest minor allele frequency of this variant was in the East Asian genetic ancestry group of gnomAD (v4.1.0) = 0.00004455 (2 alleles out of 44,890), which met the ≤ 0.0001 threshold set for PM2_Supporting in a genetic ancestry group of at least 10,000 alleles.
PS4_Supporting	✔	2 probands with primary open angle glaucoma have been reported carrying this variant (PMID: 24825108), which met PS4_Supporting (≥ 2 probands).
PP3_Strong	✔	The REVEL score = 0.958, which met the ≥ 0.932 threshold for PP3_Strong, predicting a damaging effect on MYOC function.

Not Met criteria codes

BS1	✖	This criterion was not met as PM2_Supporting has been met.
BS3	✖	No functional evidence has been found for this variant.
BP7	✖	This is not an intronic, synonymous or non-coding variant.
BP4	✖	This criterion was not met as PP3 has been met.
PM5	✖	No other LP or P missense variants at this amino acid residue have been identified.
BA1	✖	This criterion was not met as PM2_Supporting has been met.
PM4	✖	This variant does not cause a protein length change.
PS1	✖	An established LP or P variant causing this same amino acid change has not been identified.
PS2	✖	This variant has not been identified de novo.
PS3	✖	No functional evidence has been found for this variant.
PP1	✖	No segregations have been reported for this variant.

Curation History

Showing 1 to 2 of 2 rows

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