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.1313C>T

Version: 2.0

CA343723835

2429757 (ClinVar)

Gene: MYOC (HGNC:4653)
Condition: open-angle glaucoma (MONDO:0005338)
Inheritance Mode: Autosomal dominant inheritance
UUID: c4cedd64-2ff0-40ae-a6a3-1e50df9ca3f0
Approved on: 2026/1/13
Published on: 2026/1/12

HGVS expressions

NM_000261.2:c.1313C>T
NC_000001.11:g.171636127G>A
CM000663.2:g.171636127G>A
NC_000001.10:g.171605267G>A
CM000663.1:g.171605267G>A
NC_000001.9:g.169871890G>A
NG_008859.1:g.21507C>T
ENST00000037502.11:c.1313C>T
ENST00000637303.1:c.235-2503G>A
ENST00000638471.1:c.*651C>T
ENST00000037502.10:c.1313C>T
ENST00000614688.1:c.*277C>T
NM_000261.1:c.1313C>T

Likely Pathogenic

Met criteria codes 4

PM2_Supporting PP3 PS4_Supporting PP1_Strong

Not Met criteria codes 10

BA1 PM5 PM4 BS1 BS3 PS1 PS2 PS3 BP7 BP4

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.1313C>T variant in MYOC is a missense variant predicted to cause substitution of Threonine by Isoleucine at amino acid 438 (p.Thr438Ile). This variant was not found in any genetic ancestry group of gnomAD (v4.1.0), meeting the ≤ 0.0001 threshold set for PM2_Supporting in a genetic ancestry group of at least 10,000 alleles. The REVEL score = 0.734, which was within the 0.644-0.772 range for PP3, predicting a damaging effect on MYOC function. There was no functional evidence predicting a damaging or benign impact of this variant on MYOC function. 27 segregations in 4 families, with juvenile or primary open angle glaucoma (JOAG or POAG), have been reported

(PMIDs: 12789574, 12872267, 25582056, 34923728), which fulfilled PP1_Strong (≥7 meioses in >1 family). 4 probands with JOAG or POAG have been reported carrying this variant (PMIDs: 12789574, 12872267, 25582056, 34923728), which met PS4_Supporting (≥ 2 probands). In summary, this variant met the criteria to receive a score of 7 and to be classified as likely pathogenic (likely pathogenic classification range 6 to 9, adapted from PMID: 32720330) 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): PP1_Strong, PP3, PS4_Supporting, PM2_Supporting.

Met criteria codes

PM2_Supporting	✔	This variant was not found in any genetic ancestry group of gnomAD (v4.1.0), meeting the ≤ 0.0001 threshold set for PM2_Supporting in a genetic ancestry group of at least 10,000 alleles.
PP3	✔	The REVEL score = 0.734, which was within the 0.644-0.772 range for PP3, predicting a damaging effect on MYOC function.
PS4_Supporting	✔	4 probands with JOAG or POAG have been reported carrying this variant (PMIDs: 12789574, 12872267, 25582056, 34923728), which met PS4_Supporting (≥ 2 probands).
PP1_Strong	✔	27 segregations in 4 families, with juvenile or primary open angle glaucoma (JOAG or POAG), have been reported (PMIDs: 12789574, 12872267, 25582056, 34923728), which fulfilled PP1_Strong (≥7 meioses in >1 family).

Not Met criteria codes

BA1	✖	This criterion was not met as PM2_Supporting has been met.
PM5	✖	No other LP or P missense variants at this amino acid residue have been identified.
PM4	✖	This criterion did not apply to this variant.
BS1	✖	This criterion was not met as PM2_Supporting has been met.
BS3	✖	No functional evidence has been found for this variant.
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.
BP7	✖	This criterion did not apply to this variant.
BP4	✖	This criterion was not met as PP3 has been met.

Curation History

Showing 1 to 2 of 2 rows

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.