

MYOC gene

myocilin

Normal Function

The *MYOC* gene provides instructions for producing a protein called myocilin. Myocilin is found in certain structures of the eye, called the trabecular meshwork and the ciliary body, that regulate the pressure within the eye (intraocular pressure). Myocilin's function is not well understood, but it may help to control the intraocular pressure through its action in the muscle tissue of the ciliary body.

Researchers believe that myocilin functions together with other proteins in the eye as part of the extracellular matrix, which is an intricate lattice that forms in the space between cells and provides structural support. Myocilin may interact with a number of other proteins that also function in the extracellular matrix.

Health Conditions Related to Genetic Changes

Early-onset glaucoma

Approximately 10 percent to 33 percent of people with juvenile open-angle glaucoma have variants (also called mutations) in the *MYOC* gene. People with this condition experience increased pressure within the eye before the age of 40. *MYOC* gene variants have also been detected in some people with primary congenital glaucoma. More than 100 *MYOC* gene variants have been identified.

Variants in the *MYOC* gene may alter the myocilin protein so that its interactions with other proteins are impeded. Defective myocilin that does not get incorporated in the extracellular matrix remains inside the cell. The defective protein may damage the cell, causing insufficient flow or fluid from the eye, resulting in increased intraocular pressure and causing the signs and symptoms of early-onset glaucoma.

Other disorders

A small percentage (3 to 5 percent) of individuals with late-onset primary open-angle glaucoma (POAG), the most common adult form of glaucoma, have variants in the *MYOC* gene.

Other Names for This Gene

- GLC1A
- GPOA
- JOAG
- JOAG1
- MYOC_HUMAN
- myocilin, trabecular meshwork inducible glucocorticoid response
- TIGR
- trabecular meshwork-induced glucocorticoid response protein

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MYOC ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4653\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4653[geneid]))

Scientific Articles on PubMed

- PubMed ([https://pubmed.ncbi.nlm.nih.gov/?term=\(MYOC%5BTIAB%5D\)+AND+\(\(Genes%5BMH%5D\)+OR+\(Genetic+Phenomena%5BMH%5D\)\)+AND+english%5Bla%5D+AND+human%5Bmh%5D\)](https://pubmed.ncbi.nlm.nih.gov/?term=(MYOC%5BTIAB%5D)+AND+((Genes%5BMH%5D)+OR+(Genetic+Phenomena%5BMH%5D))+AND+english%5Bla%5D+AND+human%5Bmh%5D)))

Catalog of Genes and Diseases from OMIM

- GLAUCOMA, PRIMARY OPEN ANGLE; POAG (<https://omim.org/entry/137760>)
- MYOCILIN; MYOC (<https://omim.org/entry/601652>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/4653>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=MYOC\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=MYOC[gene]))

References

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Genomic Location

The *MYOC* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

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