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). It is also found in various types of muscle. 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 as part of a protein complex. Myocilin may interact with a number of other proteins including a form of the cytochrome P450 protein, the product of the *CYP1B1* gene. Like myocilin, this protein is found in the trabecular meshwork, ciliary body, and other structures of the eye.

Health Conditions Related to Genetic Changes

Early-onset glaucoma

Approximately 10 percent to 33 percent of people with juvenile open-angle glaucoma have mutations in the *MYOC* gene. *MYOC* gene mutations have also been detected in some people with primary congenital glaucoma. More than 40 *MYOC* gene mutations have been identified.

Mutations in the *MYOC* gene may alter the myocilin protein so that its interactions with other proteins are impeded. Defective myocilin that is not incorporated into protein complexes may accumulate in the trabecular meshwork and ciliary body. The excess protein may prevent sufficient flow of fluid from the eye, resulting in increased intraocular pressure and causing the signs and symptoms of early-onset glaucoma.

Individuals with mutations in both the *MYOC* and *CYP1B1* genes may develop glaucoma at an earlier age than do those with mutations in only one of the genes.

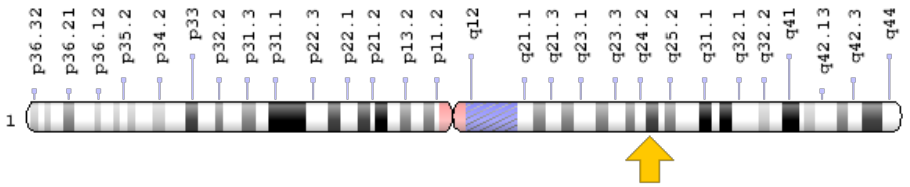
Other disorders

A small percentage of individuals with late-onset primary open-angle glaucoma (POAG), the most common adult form of glaucoma, have mutations in the *MYOC* gene. Most cases of this condition, however, are caused by other diseases, aging, and lifestyle factors such as smoking.

Chromosomal Location

Cytogenetic Location: 1q24.3, which is the long (q) arm of chromosome 1 at position 24.3

Molecular Location: base pairs 171,635,417 to 171,652,688 on chromosome 1 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

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

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28MYOC%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- GLAUCOMA, PRIMARY OPEN ANGLE
  http://omim.org/entry/137760
- MYOCILIN
  http://omim.org/entry/601652

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_MYOC.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MYOC%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:7610
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:4653
- Myocilin Allele-Specific Phenotype Database
  http://www.myocilin.com/
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/4653
- UniProt
  https://www.uniprot.org/uniprot/Q99972

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