

MYOC Gene

Subjects: Genetics & Heredity

Contributor: Lily Guo

myocilin

Keywords: genes

1. Introduction

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.

2. Health Conditions Related to Genetic Changes

2.1. 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.

2.2. 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.

3. Other Names for This Gene

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

- TIGR
- trabecular meshwork-induced glucocorticoid response protein

References

1. Bayat B, Yazdani S, Alavi A, Chiani M, Chitsazian F, Tusi BK, Suri F, Narooie-Nejhad M, Sanati MH, Elahi E. Contributions of MYOC and CYP1B1 mutations to JOAG. *Mol Vis*. 2008 Mar 13;14:508-17.
2. Chen Y, Jiang D, Yu L, Katz B, Zhang K, Wan B, Sun X. CYP1B1 and MYOC mutations in 116 Chinese patients with primary congenital glaucoma. *Arch Ophthalmol*. 2008 Oct;126(10):1443-7. doi: 10.1001/archophth.126.10.1443.
3. Joe MK, Sohn S, Hur W, Moon Y, Choi YR, Kee C. Accumulation of mutant myocilins in ER leads to ER stress and potential cytotoxicity in human trabecular meshwork cells. *Biochem Biophys Res Commun*. 2003 Dec 19;312(3):592-600.
4. Kanagavalli J, Pandaranayaka E, Krishnadas SR, Krishnaswamy S, Sundaresan P. A review of genetic and structural understanding of the role of myocilin in primary open angle glaucoma. *Indian J Ophthalmol*. 2004 Dec;52(4):271-80. Review.
5. Kaur K, Reddy AB, Mukhopadhyay A, Mandal AK, Hasnain SE, Ray K, Thomas R, Balasubramanian D, Chakrabarti S. Myocilin gene implicated in primary congenital glaucoma. *Clin Genet*. 2005 Apr;67(4):335-40.
6. Kong TH. Post-transcriptional modification of the gene genetically linked to juvenile open-angle glaucoma; novel transcripts in human ocular tissues. *Gene*. 2001 Dec 12;280(1-2):115-22.
7. Polansky JR, Fauss DJ, Zimmerman CC. Regulation of TIGR/MYOC gene expression in human trabecular meshwork cells. *Eye (Lond)*. 2000 Jun;14 (Pt 3B):503-14.
8. Polansky JR. Current perspectives on the TIGR/MYOC gene (Myocilin) and glaucoma. *Ophthalmol Clin North Am*. 2003 Dec;16(4):515-27, v-vi. Review.
9. Ray K, Mukhopadhyay A, Acharya M. Recent advances in molecular genetics of glaucoma. *Mol Cell Biochem*. 2003 Nov;253(1-2):223-31. Review.
10. Vincent AL, Billingsley G, Buys Y, Levin AV, Priston M, Trope G, Williams-Lyn D, Héon E. Digenic inheritance of early-onset glaucoma: CYP1B1, a potential modifier gene. *Am J Hum Genet*. 2002 Feb;70(2):448-60.
11. Weisschuh N, Schiefer U. Progress in the genetics of glaucoma. *Dev Ophthalmol*. 2003;37:83-93. Review.
12. Wu Dunn D. Genetic basis of glaucoma. *Curr Opin Ophthalmol*. 2002 Apr;13(2):55-60. Review.

Retrieved from <https://encyclopedia.pub/entry/history/show/12683>