CYP1B1 Gene

Subjects: Genetics & Heredity
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Cytochrome P450 Family 1 Subfamily B Member 1

genes

1. Normal Function

The *CYP1B1* gene provides instructions for producing an enzyme that is a member of the cytochrome P450 family of enzymes. These enzymes are involved in many processes in the body, such as assisting with reactions that break down drugs and produce certain fats (lipids). The CYP1B1 enzyme participates in biochemical reactions in which an oxygen atom is added to other molecules.

The CYP1B1 enzyme is active in many tissues, including structures of the eye. The function of the CYP1B1 enzyme in the development of the eye is unclear, but it may play a role in forming structures at the front of the eye and may also be involved in a process that regulates the secretion of fluid inside the eye.

2. Health Conditions Related to Genetic Changes

2.1 Early-Onset Glaucoma

More than 140 *CYP1B1* gene mutations have been identified to cause early-onset glaucoma. People with this condition experience increased pressure within the eye before the age of 40. This pressure causes damage to the optic nerves connecting the eyes and the brain. Between 20 percent and 40 percent of people with glaucoma before the age of 5 (primary congenital glaucoma) have mutations in the *CYP1B1* gene. The mutations that cause early-onset glaucoma may result in an enzyme that is unstable or the wrong shape.

It is not well understood how defects in the CYP1B1 enzyme cause signs and symptoms of glaucoma. Recent studies suggest that the defects may interfere with the early development of the trabecular meshwork, which is a network of mesh-like canals that helps drain excess fluid from the eye. If fluid cannot be drained, pressure inside the eye may increase, which is characteristic of glaucoma.

The CYP1B1 enzyme may interact with another protein called myocilin, which is produced from the *MYOC* gene. Individuals with mutations in both the *MYOC* and *CYP1B1* genes may develop glaucoma at an earlier age and have more severe symptoms than do those with mutations in only one of the genes.

2.2 Peters Anomaly

A few mutations in the *CYP1B1* gene have been found to cause Peters anomaly. This condition is characterized by abnormal development of certain structures at the front of the eye and clouding of the clear front surface of the eye (cornea). The mutations that cause Peters anomaly likely impair the normal function of the CYP1B1 enzyme or disrupt enzyme production. As a result, there is a shortage (deficiency) of normal enzyme. A lack of the CYP1B1 enzyme likely disrupts normal development of the eye, although it is unclear exactly how this deficiency leads to the features of Peters anomaly.

3. Other Names for This Gene

- aryl hydrocarbon hydroxylase
- CP1B
- CP1B1_HUMAN
- cytochrome P450, family 1, subfamily B, polypeptide 1
- cytochrome P450, subfamily I (dioxin-inducible), polypeptide 1 (glaucoma 3, primary infantile)
- flavoprotein-linked monooxygenase
- GLC3A
- microsomal monooxygenase
- xenobiotic monooxygenase

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