Microphthalmia

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Microphthalmia is an eye abnormality that arises before birth. In this condition, one or both eyeballs are abnormally small.

Keywords: genetic conditions

1. Introduction

In some affected individuals, the eyeball may appear to be completely missing; however, even in these cases some remaining eye tissue is generally present. Such severe microphthalmia should be distinguished from another condition called anophthalmia, in which no eyeball forms at all. However, the terms anophthalmia and severe microphthalmia are often used interchangeably. Microphthalmia may or may not result in significant vision loss.

People with microphthalmia may also have a condition called coloboma. Colobomas are missing pieces of tissue in structures that form the eye. They may appear as notches or gaps in the colored part of the eye called the iris; the retina, which is the specialized light-sensitive tissue that lines the back of the eye; the blood vessel layer under the retina called the choroid; or in the optic nerves, which carry information from the eyes to the brain. Colobomas may be present in one or both eyes and, depending on their size and location, can affect a person's vision.

People with microphthalmia may also have other eye abnormalities, including clouding of the lens of the eye (cataract) and a narrowed opening of the eye (narrowed palpebral fissure). Additionally, affected individuals may have an abnormality called microcornea, in which the clear front covering of the eye (cornea) is small and abnormally curved.

Between one-third and one-half of affected individuals have microphthalmia as part of a syndrome that affects other organs and tissues in the body. These forms of the condition are described as syndromic. When microphthalmia occurs by itself, it is described as nonsyndromic or isolated.

2. Frequency

Microphthalmia occurs in approximately 1 in 10,000 individuals.

3. Causes

Microphthalmia may be caused by changes in many genes involved in the early development of the eye, most of which have not been identified. The condition may also result from a chromosomal abnormality affecting one or more genes. Most genetic changes associated with isolated microphthalmia have been identified only in very small numbers of affected individuals.

Microphthalmia may also be caused by environmental factors that affect early development, such as a shortage of certain vitamins during pregnancy, radiation, infections such as rubella, or exposure to substances that cause birth defects (teratogens).

3.1. The Genes Associated with Microphthalmia

- BCOR
- GDF3
- GDF6
- OTX2
- PAX6
- SHH
- SOX2

3.1.1. Additional Information from NCBI Gene

- BMP4
- MFRP
- PRSS56
- RAX
- SIX6
- STRA6
- VSX2

4. Inheritance

Isolated microphthalmia is sometimes inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition. In some cases, parents of affected individuals have less severe eye abnormalities.

When microphthalmia occurs as a feature of a genetic syndrome or chromosomal abnormality, it may cluster in families according to the inheritance pattern for that condition, which may be autosomal recessive or other patterns.

Often microphthalmia is not inherited, and there is only one affected individual in a family.

5. Other Names for This Condition

- microphthalmos

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