Microphthalmia

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

Frequency

Microphthalmia occurs in approximately 1 in 10,000 individuals.

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).
**Inheritance Pattern**

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.

**Other Names for This Condition**

- microphthalmos

**Diagnosis & Management**

**Genetic Testing Information**

- What is genetic testing? /primer/testing/genetictesting
• Genetic Testing Registry: Microphthalmia, isolated 6
• Genetic Testing Registry: Microphthalmia, isolated 7
• Genetic Testing Registry: Microphthalmia, isolated 8
• Genetic Testing Registry: Microphthalmia, isolated, with coloboma 1
• Genetic Testing Registry: Microphthalmia, isolated, with coloboma 2
• Genetic Testing Registry: Microphthalmia, isolated, with coloboma 3
• Genetic Testing Registry: Microphthalmia, isolated, with coloboma 4
• Genetic Testing Registry: Microphthalmia, isolated, with coloboma 5
• Genetic Testing Registry: Microphthalmia, isolated, with coloboma 6
• Genetic Testing Registry: Microphthalmia, isolated, with corectopia

Research Studies from ClinicalTrials.gov
• ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22microphthalmia%22

Other Diagnosis and Management Resources
• GeneReview: Microphthalmia/Anophthalmia/Coloboma Spectrum
  https://www.ncbi.nlm.nih.gov/books/NBK1378

Additional Information & Resources
Health Information from MedlinePlus
• Encyclopedia: Coloboma of the Iris
  https://medlineplus.gov/ency/article/003318.htm
• Encyclopedia: Congenital Cataract
  https://medlineplus.gov/ency/article/001615.htm
• Health Topic: Eye Diseases
  https://medlineplus.gov/eyediseases.html
Genetic and Rare Diseases Information Center

- Microphthalmia
  https://rarediseases.info.nih.gov/diseases/12085/microphthalmia

Additional NIH Resources

- National Eye Institute: Facts About Anophthalmia and Microphthalmia
  https://nei.nih.gov/health/anoph/anophthalmia

Educational Resources

- MalaCards: microphthalmia
  https://www.malacards.org/card/microphthalmia

- Minnesota Department of Health: Anophthalmia and Microphthalmia Fact Sheet
  http://www.health.state.mn.us/divs/cfh/topic/diseasesconds/anophthalmia.cfm

- Orphanet: Colobomatous microphthalmia
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=98938

- Orphanet: Isolated microphthalmia-anophthalmia-coloboma
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=2542

- Orphanet: Microphthalmia-anophthalmia-coloboma
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=98555

- Orphanet: OBSOLETE: Microphthalmia-cataract syndrome
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=2543

- Scottish Sensory Centre
  http://www.ssc.education.ed.ac.uk/resources/vi&multi/eyeconds/Micro.html

Patient Support and Advocacy Resources

- American Foundation for the Blind
  https://www.afb.org/default.aspx

- Microphthalmia, Anophthalmia and Coloboma Support
  https://macs.org.uk/

- University of Kansas Genetics Education Center
  http://www.kumc.edu/gec/support/anopthal.html

Clinical Information from GeneReviews

- Microphthalmia/Anophthalmia/Coloboma Spectrum
  https://www.ncbi.nlm.nih.gov/books/NBK1378
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28microphthalmia%5BTI%5D %29+OR+%28anophthalmia%5BTI%5D%29%29+AND+english%5Bla%5D+AND +human%5Bmh%5D

Catalog of Genes and Diseases from OMIM

- MICROPHTHALMIA, ISOLATED 1
  http://omim.org/entry/251600
- MICROPHTHALMIA, ISOLATED 2
  http://omim.org/entry/610093
- MICROPHTHALMIA, ISOLATED 3
  http://omim.org/entry/611038
- MICROPHTHALMIA, ISOLATED 4
  http://omim.org/entry/613094
- MICROPHTHALMIA, ISOLATED 5
  http://omim.org/entry/611040
- MICROPHTHALMIA, ISOLATED 6
  http://omim.org/entry/613517
- MICROPHTHALMIA, ISOLATED 7
  http://omim.org/entry/613704
- MICROPHTHALMIA, ISOLATED 8
  http://omim.org/entry/615113
- MICROPHTHALMIA, ISOLATED, WITH CATARACT 1
  http://omim.org/entry/156850
- MICROPHTHALMIA, ISOLATED, WITH COLOBOMA 1
  http://omim.org/entry/300345
- MICROPHTHALMIA, ISOLATED, WITH COLOBOMA 2
  http://omim.org/entry/605738
- MICROPHTHALMIA, ISOLATED, WITH COLOBOMA 3
  http://omim.org/entry/610092
- MICROPHTHALMIA, ISOLATED, WITH COLOBOMA 4
  http://omim.org/entry/251505
- MICROPHTHALMIA, ISOLATED, WITH COLOBOMA 5
  http://omim.org/entry/611638
- MICROPHTHALMIA, ISOLATED, WITH COLOBOMA 6
  http://omim.org/entry/613703
• MICROPHTHALMIA, ISOLATED, WITH COLOBOMA
  http://omim.org/entry/615145
• MICROPHTHALMIA, ISOLATED, WITH CORECTOPIA
  http://omim.org/entry/156900
• OPTIC DISC ANOMALIES WITH RETINAL AND/OR MACULAR DYSTROPHY
  http://omim.org/entry/212550

Sources for This Summary


  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17914432

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12503095

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20574025

• Verma AS, Fitzpatrick DR. Anophthalmia and microphthalmia. Orphanet J Rare Dis. 2007 Nov 26;2:47. Review.  
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18039390  
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2246098/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19864492

Reprinted from Genetics Home Reference:  

Reviewed: November 2011  
Published: November 7, 2018

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services