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. Microthalmia 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.

Genetic Changes

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

• 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

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

General Information from MedlinePlus
• Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html

• Drug Therapy
  https://medlineplus.gov/drugtherapy.html

• Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html

• Palliative Care
  https://medlineplus.gov/palliativecare.html

• Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html
Additional Information & Resources

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
- Disease InfoSearch: Cataract, Congenital, with Microphthalmia
  http://www.diseaseinfosearch.org/Cataract%2C+Congenital%2C+with+Microphthalmia/1155
- Disease InfoSearch: Cataract, Microphthalmia and Nystagmus
  http://www.diseaseinfosearch.org/Cataract%2C+Microphthalmia+and+Nystagmus/1156
- Disease InfoSearch: Microphthalmia, isolated 2
  http://www.diseaseinfosearch.org/Microphthalmia%2C+isolated+2/8873
- Disease InfoSearch: Microphthalmia, isolated 3
  http://www.diseaseinfosearch.org/Microphthalmia%2C+isolated+3/8874
- Disease InfoSearch: Microphthalmia, isolated 4
  http://www.diseaseinfosearch.org/Microphthalmia%2C+isolated+4/8875
- Disease InfoSearch: Microphthalmia, isolated 5
  http://www.diseaseinfosearch.org/Microphthalmia%2C+isolated+5/8876
- Disease InfoSearch: Microphthalmia, isolated 6
  http://www.diseaseinfosearch.org/Microphthalmia%2C+isolated+6/8877
- Disease InfoSearch: Microphthalmia, isolated 7
  http://www.diseaseinfosearch.org/Microphthalmia%2C+isolated+7/8878
- Disease InfoSearch: Microphthalmia, isolated, with cataract 4
  http://www.diseaseinfosearch.org/Microphthalmia%2C+isolated%2C+with+cataract+4/8879
• Disease InfoSearch: Microphthalmia, isolated, with coloboma 3  
  http://www.diseaseinfosearch.org/Microphthalmia%2C+isolated%2C+with+coloboma+3/8880

• Disease InfoSearch: Microphthalmia, isolated, with coloboma 5  
  http://www.diseaseinfosearch.org/Microphthalmia%2C+isolated%2C+with+coloboma+5/8881

• Disease InfoSearch: Microphthalmia, isolated, with coloboma 6  
  http://www.diseaseinfosearch.org/Microphthalmia%2C+isolated%2C+with+coloboma+6/8882

• Disease InfoSearch: Microphthalmia, isolated, with coloboma 7  
  http://www.diseaseinfosearch.org/Microphthalmia%2C+isolated%2C+with+coloboma+7/8883

• Disease InfoSearch: Microphthalmia, isolated, with corectopia  
  http://www.diseaseinfosearch.org/Microphthalmia%2C+isolated%2C+with+corectopia/4795

• MalaCards: microphthalmia  
  http://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: Anophthalmia-microphthalmia syndrome  
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=98555

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

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

• Orphanet: Microphthalmia-cataract syndrome  
  http://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  
  http://macs.org.uk/

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

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

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22microphthalmia%22

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

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 9
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

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  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3059417/

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