

Anophthalmia/Microphthalmia

BIRTH DEFECT RESEARCH FOR CHILDREN



What is Anophthalmia/Microphthalmia (A/M)?

Anophthalmia means that one or both eyes didn't form during the early stages of pregnancy. Congenital anophthalmia may occur alone or with other malformations.

Microphthalmia means that the eye/s started to form during pregnancy but stopped, leaving the baby with eyes that are smaller than normal.

These two terms are sometimes used interchangeably since in most cases CT scans will show some remnants of eye tissue even in cases of anophthalmia.



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Anophthalmia is classified in three ways:

- Primary Anophthalmia – the complete absence of eye tissue
- Secondary Anophthalmia – the eye starts to develop but stops, leaving residual eye tissue that may be detected only by careful examination
- Degenerative Anophthalmia – the eye starts to form, but then degenerates, perhaps due to a lack of blood supply to the developing eye

How often does A/M occur?

Total anophthalmia occurs approximately 1 in 100,000 births, although some reports have found a frequency of 1 in 50,000. Microphthalmia is detected in 1 in 10,000 births.

What causes Anophthalmia/Microphthalmia?

Cases of A/M may result from genetic inheritance, sporadic genetic mutations and chromosomal abnormalities. Some cases of A/M have been linked to prenatal exposure to environmental factors such as drugs, pesticides, radiation, toxins, or some viruses.

How is A/M treated?

An ophthalmologist and an ocularist, who makes and fits prosthetic eyes, should evaluate a newborn with anophthalmia as soon as possible. It is important for the ocular orbits that would normally hold the eyes to develop properly. If the orbital globe is missing or too small, the infant's face will not develop correctly. This can sometimes be corrected with conformers, plastic structures that help support the growth and development of the eye socket and bones in the face. Surgical reconstruction of the eye socket may be necessary. When the eye sockets and facial bones have developed, the ocularist can make prosthetic eyes,

usually from acrylic, but porcelain may be used if a child is allergic to acrylic. The correct time to fit a prosthetic eye/s can vary from child to child. As the child grows, the prosthesis will need to be checked for size, fit, and comfort.

What is the prognosis for children with A/M?

A child with A/M may be blind or only partially sighted. This visual impairment may lead to other problems such as learning difficulties or behavior disorders. Other problems may be involved if A/M is part of a more complex birth defect syndrome.

What conditions may occur with A/M?

- Congenital cataracts
- Optic disk abnormalities
- Congenital cystic eye
- Microcornea
- Persistent hyperplastic primary vitreous
- Nanophthalmia (a heredity condition in which the eye looks normal but the eyeball is significantly shorter)

Fact Sheet by:

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