

COLOBOMA means that there is a gap, or cleft, in one of the structures of the eye. This is the result of congenital malformation, with a portion of the eye failing to complete its growth and fuse together, so leaving a gap in one or more of the layers of the eye. Different parts of the eye may be affected, including the lens, choroid and optic nerve, although the iris is most commonly involved, giving a keyhole shaped pupil. Vision may or may not be affected depending on the part of the eye involved.

Coloboma may be unilateral (*affecting one eye*) or bilateral (*affecting both eyes*) and if bilateral, can be quite asymmetrical. It may be an isolated ocular finding or may be associated with other eye defects, such as Microphthalmia (*small eye*) in the same or opposite eye, and extremely rarely Anophthalmia (*absent eye*) in the opposite eye.

We gratefully acknowledge our friends and professionals in the medical field for their assistance in putting this leaflet together.



Unilateral Coloboma

MACS was formed in 1993 by a group of parents, all of whom had children with anophthalmia, microphthalmia and coloboma. Until then most of them didn't know of any other children like their own, and children with these conditions did not have the opportunity to meet each other, support each other and learn from each other. MACS offers emotional and practical support to the children and families living with the daily challenges that accompany the MACS conditions.

We aim to:

- *Raise the profile of these rare conditions.*
- *Encourage families to support each other through: our membership scheme, newsletter, website, social networking sites and importantly via direct contact at our annual Family Weekend, regional events for families and events held specifically for young MACS adults.*
- *Provide beneficial rest for parents, children and extended family members.*
- *Provide financial assistance to MACS families who need to purchase equipment or services to help meet their specific needs.*
- *To become the leading international resource on Anophthalmia, Microphthalmia and Coloboma, for parents and professionals.*

Microphthalmia, Anophthalmia & Coloboma Support

Supporting children born without eyes or with underdeveloped eyes

Tel: 0800 169 8088, **Web:** www.macs.org.uk

Registered charity no: 1161897

Registered address: MACS, Suite 472, Kemp House, 152 City Road, London, EC1V 2NX.

MACS

Microphthalmia, Anophthalmia & Coloboma Support
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An understanding of the conditions



Definition of MACS Eye Conditions

Anophthalmia is a rare developmental abnormality. It is part of a range of abnormalities in which babies are born with no eye in the eye socket (*anophthalmia*) or with a small eye in the eye socket (*microphthalmia*). Coloboma is a related condition where one or more of the layers of the eye fail to fuse properly leaving a small gap.

ANOPHTHALMIA can affect both eyes, in which case the baby will be blind, or in one eye in which case the baby may have normal vision in the other eye.

Anophthalmia or severe **MICROPHTHALMIA** occurs in 3 – 7 babies every 100,000 live births. This means that in England and Wales there are only about 30-35 babies born each year with Anophthalmia or severe Microphthalmia. About half of these babies have other developmental problems in addition to Anophthalmia.



Unilateral Microphthalmia

The condition is likely to occur because the delicate sequence of early developmental steps to form an eye is disrupted in some way. These signals for development in the embryo come from the genes within the developing cells. The exact mechanism is not fully understood, but a disruption of this process can occur through an error in the genes themselves or external factors acting alone or by influencing the function of these genes during pregnancy.

The occurrence of Anophthalmia and Microphthalmia has been related to some illnesses during pregnancy including virus infections, such as rubella (*German Measles*) and Varicella (*Chicken Pox*). It has also been linked to some drugs taken during pregnancy, including recreational drugs and thalidomide. There has been a suggestion that insecticides and fungicides used to spray crops may be related to eye and other congenital anomalies, but to date there is no scientific proof to support this, only indirect evidence from animal studies. Also over the last few years a growing number of genes have been identified that are important in Anophthalmia or the related conditions Microphthalmia and Coloboma.



Unilateral Anophthalmia and Bi-lateral Anophthalmia

An accurate genetic diagnosis can help a family to understand the cause of their child's condition, and the outlook for the future. It will also allow informed genetic counselling regarding recurrence risks in future pregnancies and if prenatal diagnosis can be offered.

It is not possible to restore sight to a baby with Anophthalmia affecting both eyes. However artificial eyes, usually made of acrylic and painted to look like real eyes, are used to help with the cosmetic appearance. Treatment is beneficial for those babies from an early age, and can start with soft expanders shortly after birth. If a baby is born without an eye in the socket, the eye socket does not receive the correct signals to grow properly.

This results in a small socket and it may be difficult to fit artificial eyes later to these children. Therefore it is important that babies born without an eye or with a very small eye are referred for assessment at a specialist centre as soon after the birth as possible.



Use of a Prosthetic Eye, before and after insertion