



**Microphthalmia, Anophthalmia
& Coloboma Support**

Supporting people born without eyes and with
underdeveloped eyes

Tel: 0800 644 6017 (Enquiries)

Tel: 0800 169 8088 (Members Support)

Web: www.macs.org.uk

Address: MACS (Microphthalmia,
Anophthalmia & Coloboma Support),
71-75 Shelton Street, London, WC2H 9JQ



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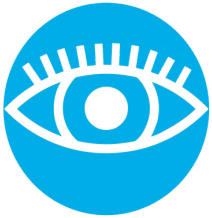
The conditions



Introduction to MAC conditions

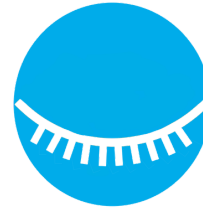
What are they?

Microphthalmia:



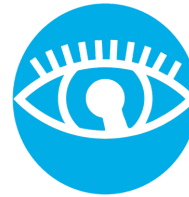
Microphthalmia, meaning "small eye," is a condition where a child may be born with one (unilateral) or both (bilateral) eyes that are smaller than usual and have anatomic malformations. While some children with this condition may be blind, others may have some residual sight or light perception. Microphthalmia can also affect the development of the eye socket, potentially leading to asymmetry in facial growth. To help support socket development and improve appearance, some children wear an orbital conformer or a custom-made cosmetic shell

Anophthalmia:



Anophthalmia means the absence of the eye. A child with anophthalmia may be born without one eye (unilateral) or both eyes (bilateral). In this condition, the eye socket develops differently, which can lead to features like smaller eyelids or changes in the shape of the surrounding area.

Coloboma:



Coloboma is a condition where there's a gap or cleft in one or more parts of the eye, like the iris, retina, or optic disc. It happens when a small gap in the eye, present during early development, doesn't fully close before birth.

The effect on vision varies depending on the size and location of the gap - sometimes vision is unaffected, while in other cases, it may cause challenges. In very young children, it can be tricky to determine how coloboma impacts their vision, but doctors gain more insight as the child grows and more precise tests can be done.

How They Occur

The exact causes of **Microphthalmia** and **Anophthalmia** are not yet fully understood, but they are believed to result from disruptions in the developmental process as the eyes form during pregnancy. These disruptions may stem from changes in specific genes linked to eye development, with researchers continually identifying more genes involved in these conditions.

External factors can also influence how these genes work. For example, illnesses during pregnancy, such as chickenpox or rubella, or certain medications like thalidomide, have been associated with these conditions. Environmental factors, such as exposure to insecticides or fungicides, have also been linked.

Coloboma, on the other hand, occurs when part of the eye doesn't fully form during early pregnancy, leaving a gap or cleft in one or more of its structures.

Statistics

- Approximately **114 children** are born with MACS conditions each year in the UK.
- These conditions can involve **vision loss**, **developmental** challenges, or other congenital differences.
- To date, over **90 different genes** have been linked to various forms of MAC, highlighting the complexity of human eye development during pregnancy. Researchers are still uncovering the roles of many genes involved in this intricate process.