

# Coloboma

BIRTH DEFECT RESEARCH FOR CHILDREN



## What is Coloboma?

Coloboma is the absence or defect of ocular (eye) tissue, usually a gap or cleft in one of the structures of the eye. Different parts of the eye can be affected, but the iris (colored part of eye) is the most commonly involved and is visible as a hole, split, or cleft in the iris. This usually occurs at the bottom of the eye. Coloboma can occur in various degrees of severity ranging from a small pit in the optic disk to extensive defects in various parts of the eye. Complete Coloboma occurs when all structures of the eye are involved. Depending on the severity and part of the eye involved, vision may or may not be affected. Blurred vision or decreased visual acuity is common.



# Coloboma



How do you know if your child has Coloboma?

During pregnancy high-resolution ultrasound scanning can provide details of the fetal eye which may show Coloboma. In an infant, Coloboma is usually apparent because the pupil is irregularly shaped, often appearing as a “cat eye” or “keyhole” shape. A complete medical history and physical examination will help diagnose Coloboma. Diagnostic tests may include eye examination, refraction testing, and slit-lamp examination.

What causes Coloboma?

Coloboma results from the imperfect closure of the fetal optic fissure which normally closes before birth. No specific pattern of occurrence has been identified. There appears to be a strong hereditary factor which is sometimes linked to chromosomal disorders. Coloboma can occur alone or be associated with other anomalies. It can also be part of a syndrome such as CHARGE or Cat-Eye Syndrome. Genetic counseling is recommended.

How can you help a child with Coloboma?

There is no cure for Coloboma. Because of an increased risk of detached retina and glaucoma, children should be monitored by an ophthalmologist. If the child’s eye is small or badly formed, an artificial eye or lens can be fitted to promote socket growth or to improve the child’s appearance. Eyeglasses can help improve vision. Children with Coloboma are often bothered by strong light because the pupil is unable to react to light in a normal way; therefore tinted glasses can be helpful.

What’s in the future for a child with Coloboma?

The quality of life for a child with Coloboma will depend on the severity of eye defect and whether other anomalies are present. Some vision problems associated with Coloboma can be improved with treatment. Children with Coloboma without other anomalies usually live a normal lifespan.

Fact Sheet by:

Birth Defect Research Children,  
Inc.

[www.birthdefects.org](http://www.birthdefects.org)

