Coloboma

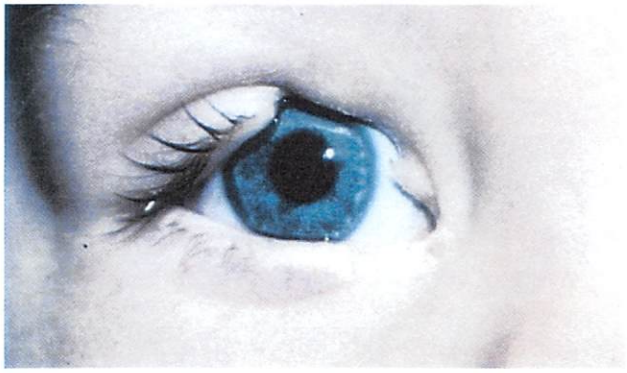
What is a coloboma?

A coloboma describes conditions where normal tissue in or around the eye is missing from birth.

Coloboma comes from the Greek word that means "curtailed." The eye develops quickly during a fetus's first three months of growth. A gap, known as the choroidal fissure, appears at the bottom of the stalks that eventually form the eye. This fissure generally closes by the seventh week of pregnancy. If it does not close, a coloboma or space forms.

A coloboma can affect one or both eyes. If both eyes are involved, it can affect them the same way or differently. There are different types of coloboma, depending on the part of the eye affected:

- **Eyelid coloboma.** A piece of the upper or lower eyelid is missing.
- **Lens coloboma.** A piece of the lens is missing.
- **Macular coloboma.** In this coloboma, the macula fails to develop normally.
- **Optic nerve coloboma.** In this coloboma, the optic nerve is hollowed out, reducing vision.
- **Uveal coloboma.** The uvea is the middle layer of the eye. This coloboma can affect the iris, the colored part of the eye, giving it a distinct keyhole or cat-eye appearance.
- **Chorio-retinal coloboma.** In this coloboma, part of the retina is missing.



Coloboma of the upper eyelid

Eye Words to Know

Retina: Layer of nerve cells lining the back wall inside the eye. This layer senses light and sends signals to the brain so you can see.

Macula: Small but important area in the center of the retina. You need the macula to clearly see details of objects in front of you.

Lens: The clear part of the eye behind the iris that focuses light onto the retina.

Optic nerve: A nerve at the back of your eye that connects to your brain. The optic nerve sends light signals to your brain so you can see.

Uvea: The middle layer of the eye beneath the sclera, or white of the eye. It is made up of the ciliary body, choroid, and iris.

What are symptoms of coloboma?

The symptoms of a coloboma depend on where it occurs. A coloboma of the iris will be visibly noticeable due to the keyhole or cat-eye appearance of the pupil. An eyelid coloboma will also be noticeable due to a notch or defect in the eyelid. People with a coloboma affecting the front of the eye may have problems with vision and should be evaluated in infancy.

Other types of coloboma can also cause vision loss, depending on their size and location. A child with a coloboma affecting the macula or the optic nerve will likely have reduced vision. Children who are missing a part of their retina will have a "field defect," which means that they will not have vision in a specific location (for instance, the upper part of the field of vision). Vision impairment caused by a coloboma may not be noticeable at birth.

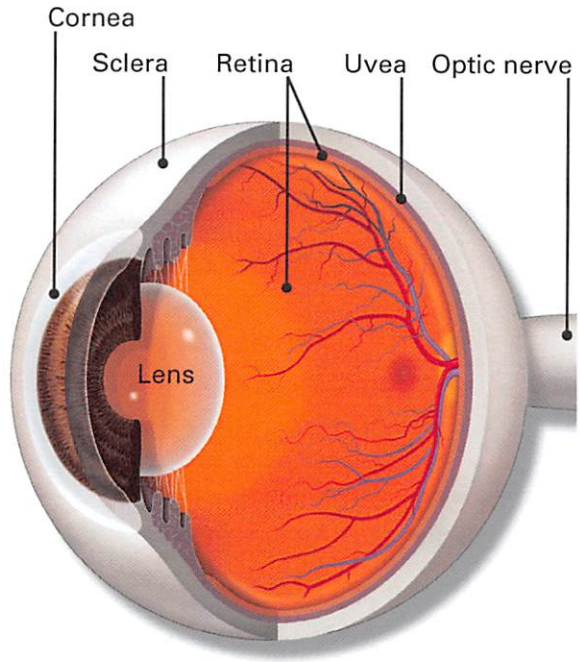
Sometimes children with a coloboma have increased sensitivity to light.

Who is at risk for a coloboma?

A coloboma is believed to be genetic and can be passed along in families.

Sometimes a coloboma is part of a genetic syndrome. For instance, cat eye syndrome, a rare disorder named after the distinctive shape of an iris coloboma, is caused by a specific genetic mutation and occurs along with other physical abnormalities.

However, not all babies born with a coloboma have a family history of this condition, suggesting that the disorder can appear by chance.



How is a coloboma diagnosed?

A coloboma is diagnosed by a thorough eye exam by an ophthalmologist.

The ophthalmologist will use an ophthalmoscope to examine the inside of a baby's eyes. When a child is old enough to express themselves, other tests, such as a visual acuity test, can be conducted to assess how vision may be affected.

How is a coloboma treated?

There is no cure for a coloboma, and treatment varies depending on the type. People with an iris coloboma may wear colored contact lenses to make the iris appear round. Surgery can also correct the appearance of the iris. There are also a number of procedures to repair colobomas of the eyelid.

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For other types of coloboma, helping people adjust to their vision problems, including using low vision devices as needed, is important. Also, your ophthalmologist will help to manage other problems that occur with coloboma, such as cataracts or growth of new blood vessels in the back of the eye as the patient ages.

Your ophthalmologist may recommend certain treatments if your child has a coloboma in only one eye. To make sure that amblyopia (lazy eye) does not develop, the ophthalmologist may want to patch or use special eye drops or glasses in the unaffected eye. Sometimes this treatment can improve vision in eyes even with severe colobomas.

Summary

A coloboma is when normal tissue in or around the eye is missing at birth. A coloboma can affect different parts of the eye. The symptoms of coloboma on the front of the eye will be visually noticeable as the pupil and eyelid will look different. Coloboma inside the eye can cause vision loss. Coloboma is believed to be genetic (passed from parent to child) and can be diagnosed during an eye exam by an ophthalmologist.

Treatment for coloboma depends on the type and location of the coloboma. Treatment can include colored contact lenses and surgery. Low vision devices can help patients with decreased vision due to coloboma.

If you have any questions about your vision, speak with your ophthalmologist. He or she is committed to protecting your sight.

Get more information about coloboma from EyeSmart—provided by the American Academy of Ophthalmology—at **aao.org/coloboma-link**.

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