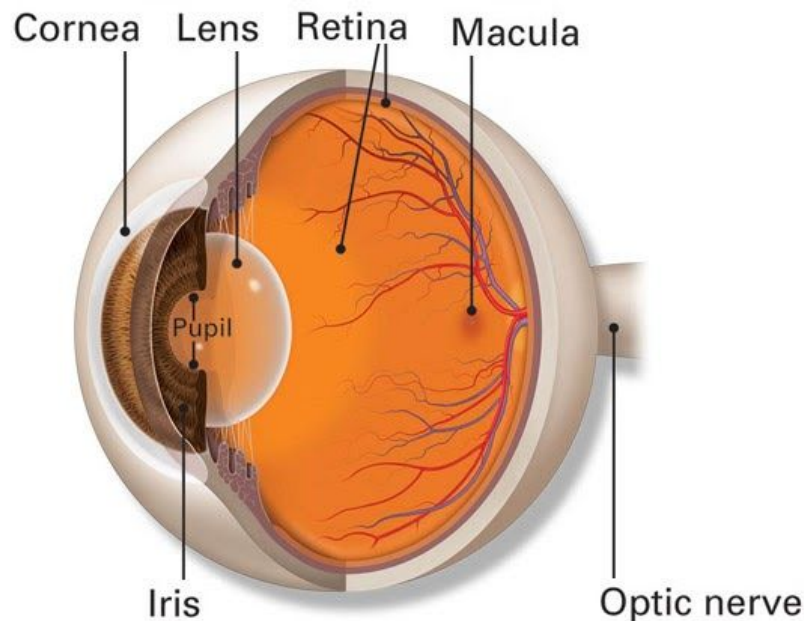


What Is a Coloboma?



A [coloboma](#) describes conditions where normal tissue in or around the eye is missing at birth.

Coloboma comes from the Greek word that means "curtailed." The eye develops quickly during a fetus' first three months of growth. A gap, known as the choroidal fissure, appears at the bottom of the stalks that eventually forms 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 Symptoms

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 or a particular syndrome, suggesting that the disorder can appear by chance.

Coloboma Diagnosis

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.

Coloboma Treatment

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](#).

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.

Sources:

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