

## Coloboma

### WHAT IS A COLOBOMA?

A coloboma is a problem in the eye that happens before birth. Part of the eye or eyelid does not form the right way. In a coloboma, parts of the eye or eyelid may be too small, not fully formed, or missing. This happens when the tissues that should form the eye and eyelids do not grow normally.



**Figure 1:** Coloboma of upper eyelid. An abnormally shaped upper lid is shown in the photo.

### IS COLOBOMA HERITABLE?

Coloboma can run in families (be inherited). It can also happen by chance, even if no one else in the family has it.

### WHAT MEDICAL PROBLEMS ARE SEEN WITH COLOBOMA?

Coloboma can happen by itself or along with other health problems in the body. These may include:

- Heart problems
- Problems with the nose or sinuses
- Slow growth or learning delays
- Urinary or genital problems
- Ear problems (such as in CHARGE syndrome)

Some of these problems are genetic. They can be found with tests using blood or a swab from inside the mouth. A genetics specialist may help check for these genetic conditions.

### WHAT TYPES OF COLOBOMAS OCCUR IN THE EYE?

Colobomas can affect different parts of the eye, including:

- Iris (the colored part)
- Lens (the clear part that helps focus)
- Retina (the back part of the eye that senses light)
- Optic nerve (the nerve that sends vision signals to the brain)

## WHAT IS A COLOBOMA OF THE EYELID?

An eyelid coloboma means part of the eyelid is missing. It can be a small notch or almost the whole eyelid. It most often affects the upper eyelid. This problem develops during early pregnancy.

## WHAT OTHER MEDICAL PROBLEMS ARE SEEN WITH A COLOBOMA OF THE EYELID?

Eyelid coloboma can be linked to other conditions, such as:

- Goldenhar syndrome (which may include eye growths, eye movement problems, ear, or spine problems)
- Treacher Collins syndrome (which may cause small jaw, slanted eyes, and sunken cheeks)

## WHAT PROBLEMS CAN COLOBOMA OF THE EYELID CAUSE TO THE EYE?

If part of the eyelid is missing, the front of the eye (cornea) may not be fully covered. This can cause dryness, infection and scarring.

## HOW IS A COLOBOMA OF THE EYELID TREATED?

Treatment may include:

- Eye drops or ointments to keep the eye surface moist
- Surgery to repair or close the eyelid

## DOES COLOBOMA OF THE IRIS AFFECT VISION?

Sometimes. Vision may be normal if the back of the eye (retina or optic nerve) is not affected. Some children may be sensitive to light because the pupil is larger than normal. Special contact lenses can help with light sensitivity and how the eye looks.

## WHAT IS A LENS COLOBOMA?

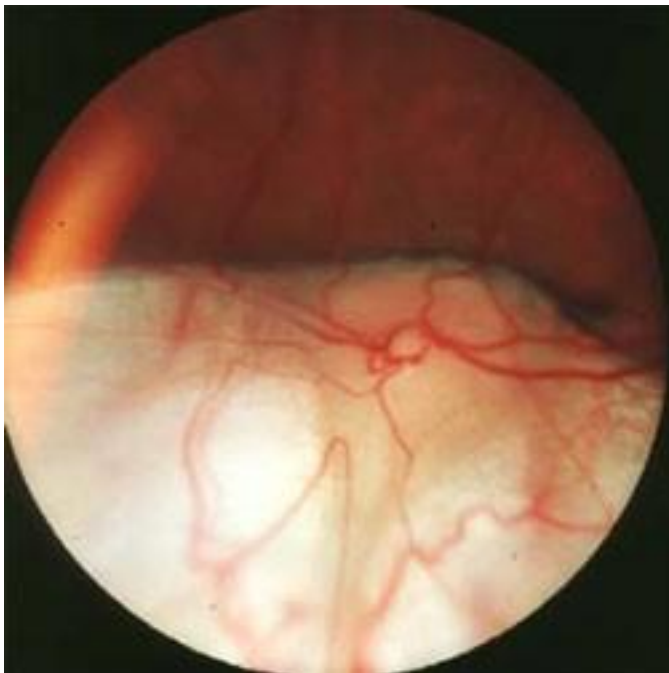
A lens coloboma happens when the parts of the eye that hold the lens in place are not fully developed. This can make the lens an unusual shape and cause:

- Blurry vision due to [refractive error](#) (need for glasses)
- Amblyopia (weakness in vision, lazy eye)

Treatment may include glasses or patching.

## WHAT IS A RETINA OR OPTIC NERVE COLOBOMA?

Sometimes, a small or large part of the back parts of one or both eyes may not form correctly [See figure 2]. This can involve the retina (inner layer lining the back of the eye that helps us see) and/or the optic nerve (which connects the eye to the brain). An eye with an optic nerve or retina coloboma can be smaller than normal.



**Figure 2: Coloboma involving the retina and optic nerve.**

## WHAT PROBLEMS CAN HAPPEN WITH RETINA AND OPTIC NERVE COLOBOMAS?

Possible but uncommon problems include:

- Retinal detachment (the retina pulls away from the back of the eye)
- Abnormal blood vessels that may bleed inside the eye

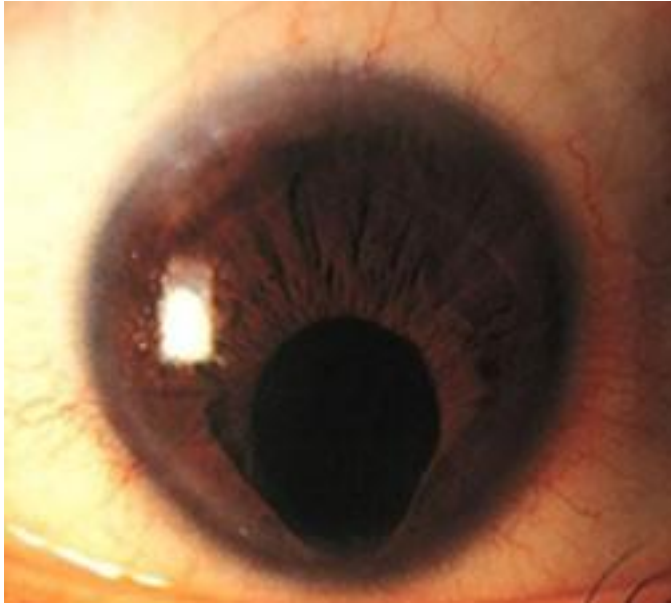
These problems can cause vision loss. Regular eye exams are important to find and treat problems early.

## DOES A COLOBOMA OF THE RETINA OR OPTIC NERVE AFFECT VISION?

Yes, it can. If important areas of the retina or optic nerve are affected, vision may be blurry and may not improve with glasses. In some cases, vision may be very poor.

## WHAT ARE THE SYMPTOMS AND SIGNS OF A COLOBOMA INSIDE THE EYE?

Common signs include a notch in the iris (iris coloboma) [See figure 3.], eye movement problems, and poor vision. A lens coloboma is usually only seen on an eye exam when the eye is dilated (pupil is made larger with special eye drops). A white reflex or glow from the eye ([leukocoria](#)) may be seen with a large coloboma of the retina or optic nerve. However, a full eye exam is usually needed to see a coloboma of the retina or optic nerve.



**Figure 3:** Iris coloboma that looks like a keyhole-shaped notch in the lower part of the iris.

### WHAT IS A COLOBOMATOUS CYST?

This is a severe form of coloboma where the eye does not develop well and cannot see. A cyst (fluid-filled sac) may form outside the eye and attach to a very small eye.

### WHERE CAN I FIND MORE INFORMATION ABOUT COLOBOMA?

- [CHARGE Syndrome Foundation](#)
- <https://eyewiki.org/Coloboma>

### SUMMARY FOR PATIENTS AND CAREGIVERS

A coloboma is a condition a child is born with where part of the eye or eyelid did not form the right way. It can affect different parts of the eye and may or may not change vision. Some children with coloboma also have other health problems, so doctors may suggest extra tests. Treatment depends on the type and how severe it is. It may include eye drops, glasses, patching, or surgery. Regular eye exams are very important to protect vision and find any problems early. If you have questions or concerns, talk to your eye doctor or care team.

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