

# Coloboma

## WHAT IS A COLOBOMA?

A coloboma is a problem with the formation of either the eyelid or the eyeball that is present since birth [See figure 1]. In a coloboma, parts of the eyeball or the eyelid may be small, underdeveloped or missing. A coloboma happens when tissues that are supposed to become the eyeball and the eyelids do not connect or form well.



**Fig. 1:** Coloboma is a problem with the formation of either the eyelid or the eye. See In the photo there is a funny shape to the upper eyelid.

## IS COLOBOMA HERITABLE?

Coloboma can be inherited (run in families) or can occur spontaneously (where no one else in the family is affected).

## WHAT MEDICAL PROBLEMS ARE SEEN WITH COLOBOMA?

Coloboma of any part of the eyeball or eyelid can happen on its own or it can occur with other problems that affect the rest of the body. Coloboma can be seen with heart problems, problems with development of the nose and sinuses, slow growth and development, genital or urinary tract problems, or problems with ear development (CHARGE syndrome). CHARGE syndrome has been linked with a mutation in the CHD7 gene, which can show on a blood test. There are many other syndromes and medical problems that can be seen with colobomas of the eye. A referral to a genetics specialist may be helpful for those with coloboma.



## WHAT TYPES OF COLOBOMAS OCCUR IN THE EYE?

Colobomas may be seen in the iris (the colored part of the eye), lens (clear focusing part in the middle of the eye), retina (inner lining of the back wall of the eye), or optic nerve (nerve that connects the eye to the brain).

## WHAT IS A COLOBOMA OF THE EYELID?

It is when all or part of the eyelid is missing [see figure 1]. A coloboma in the eyelid can vary from a small notch to near total absence of the eyelid. It most commonly affects the upper eyelid. Eyelid colobomas can occur during the third or fourth month or during the sixth or seventh month of pregnancy.

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

Eyelid coloboma can be seen with Goldenhar syndrome, which is a group of medical problems that includes: a growth on the eye (limbal [dermoid](#)), problems with eye movement ([Duane syndrome](#)), ear problems, or spine problems. A syndrome that may be seen with coloboma of the lower lid is called Treacher Collins syndrome. In this syndrome patients have sunken cheeks, slanted appearance of eye, and a small lower jaw.

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

The opening in the eyelid will leave part of the cornea (clear front part of the eye) uncovered, which can cause severe dryness, infections, and scarring.

## HOW IS A COLOBOMA OF THE EYELID TREATED?

The eye is often treated with extra tear drops and tear ointments to prevent drying, and may require surgery to close the opening in the eyelid.

## DOES COLOBOMA OF THE IRIS AFFECT VISION?

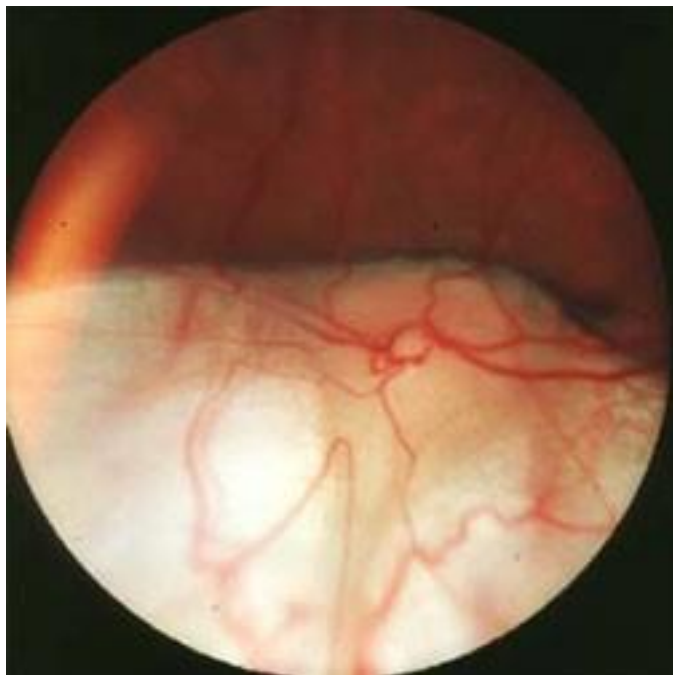
Many times the vision may be normal in those with an iris coloboma if there are no other eye problems. Some will have light sensitivity because of a larger pupil (dark spot in the center of the colored part of the eye). Contact lenses that change eye color may be used to cover the iris coloboma and help with how it looks and problems with light sensitivity.

## WHAT IS A LENS COLOBOMA?

A lens coloboma is when the parts of the eye that hold the lens in place (the ciliary body and zonules) are small or missing in places. This may cause the lens to have a funny shape, which can cause [refractive error](#) (problems focusing due to a need for glasses) and/or [amblyopia](#) (weakness in vision, lazy eye). These problems may need treatment with glasses and/or patching.

## WHAT IS A RETINA OR OPTIC NERVE COLOBOMA?

A small or large problem with development may occur in the deeper parts of one or both eyes [See figure 2]. This can involve the retina (inner layer lining the back of the eye responsible for sight) 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.



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

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

Although rare, detachment of the retina can occur any time in life in those with retina and optic nerve colobomas. If the retina detaches, vision can be lost and it may require surgical treatment. Abnormal blood vessels may also grow next to the retina or optic nerve coloboma

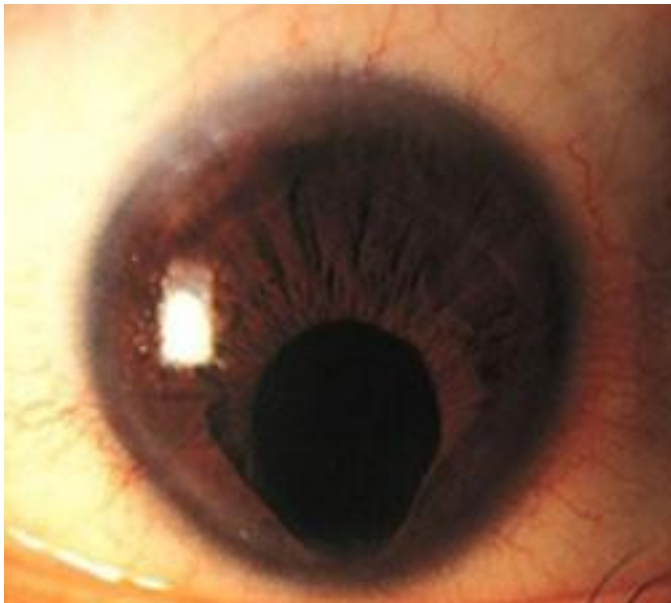
and cause bleeding in the eye, which can also cause vision loss. Regular follow up eye exams are important to pick up these problems and treat them early.

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

It can. If the area of missing or small retina or optic nerve affects the central part of the retina or a large part of the optic nerve, vision can be blurry in a way that does not get better with glasses or other treatments. Sometimes the vision is very very poor in those with coloboma of the retina or optic nerve.

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

The most common symptoms or signs are noticing a funny shape in the iris (iris coloboma), eye movement problems, or 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.



**Fig. 3:** Iris Coloboma

## WHAT IS A COLOBOMATOUS CYST?



This is when the development of the eye has lots of problems. Usually there is a cyst outside the eye, attached to a very small eye.

## WHERE CAN I FIND MORE INFORMATION ABOUT COLOBOMA?

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

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