

## Coloboma<sup>1</sup>

### What is Coloboma?

A coloboma is a congenital defect in the structure of either the eyelid or the eye.



### Is coloboma heritable?

Coloboma can be inherited or can occur spontaneously.

### What medical problems are associated with coloboma?

Coloboma of any eye structure can occur in isolation or it can occur with chromosomal abnormalities that involve other body structures. Coloboma can occur in conjunction with heart defects, choanal atresia, nervous system abnormalities, genital or urinary tract anomalies, or ear malformations (CHARGE syndrome).

### What types of colobomas occur in the eye?

Coloboma may involve the iris, lens, retina, or optic nerve. The embryonic fissure normally closes around the 5th week of gestation (during pregnancy). Improper closure of the fissure causes a defect (coloboma) in one or more of the eye structures.

### What is a coloboma of the eyelid?

It is a defect which varies from a small notch to near total absence of the eyelid. It most commonly affects the upper eyelid.

### What other abnormalities occur with a coloboma of the eyelid?

Eyelid coloboma can be associated with Goldenhar syndrome, which is characterized by a growth on the eye (limbal dermoid), abnormal eye movement (Duane syndrome), ear abnormalities, or vertebral abnormalities. Another syndrome that may be associated with

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<sup>1</sup> <https://aapos.org/glossary/coloboma>

coloboma of the lower lid is Treacher Collins syndrome, which is characterized by depressed cheeks, slanted appearance of eye, and a small mandible.

### **What are the complications of coloboma of the eyelid?**

The lid defect will leave part of the cornea uncovered, which can lead to excessive dryness due to evaporation of the tears.

### **How is a coloboma of the eyelid treated?**

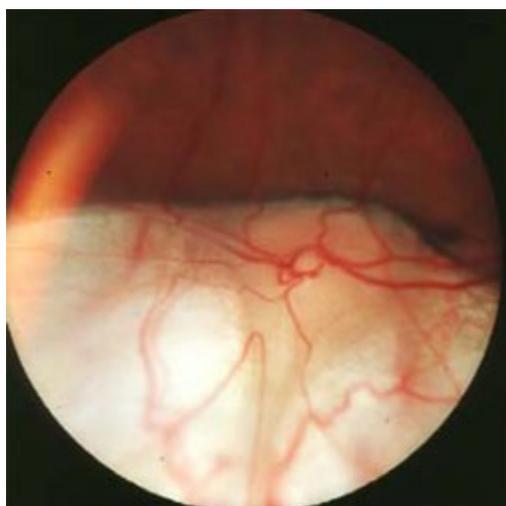
The eye usually requires extra lubrication to prevent drying of the surface, and eventually may require surgery to close the defect.

### **Does coloboma of the iris affect vision?**

If no other abnormalities are present, vision may be normal. Some increase in light sensitivity may occur because of a larger pupil. Contact lenses that change eye color can be used to cover the iris coloboma.

### **What is a fundus or optic nerve coloboma?**

A small or large developmental defect may occur in the deeper structures of one or both eyes. The defect can involve the retina (layer lining the back of the eye responsible for sight) and/or the optic nerve (which connects the eye to the brain). The eye is occasionally smaller than normal.



### **What is the complication of retina and optic disc coloboma?**

Although rare, detachment of the retina can occur any time in life. Regular follow up is important to detect detachment and treat it early.

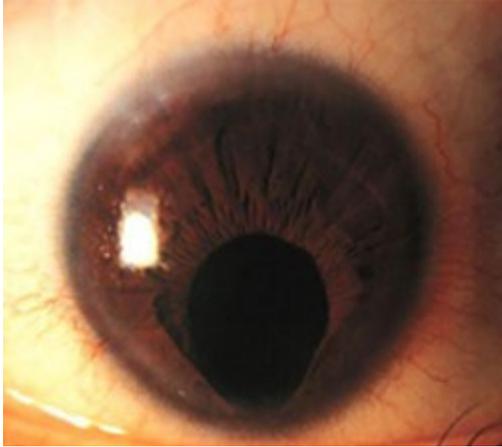
### **Does a coloboma inside the eye affect vision?**

If the defect extends to the central part of the retina or optic nerve, vision can be impaired, sometimes severely.



### **What are the symptoms and signs of a coloboma inside the eye?**

The most common symptom is noticing a defect in the iris (iris coloboma), eye misalignment, or poor vision. A coloboma of the retina or the optic nerve can only be seen when the back of the eye is examined by an ophthalmoscope.



### **What is a colobomatous cyst?**

Incomplete embryonic fissure closure produces a cyst outside the eye, attached to a very small eye.

### **What is an optic nerve pit?**

An optic nerve pit may be related to coloboma and is a small defect near the margin of the optic nerve. Fluid can sometimes collect in the retina adjacent to the defect which may cause reduced vision

Where can I find more information regarding coloboma?

[CHARGE Syndrome Foundation](#)

