

Coloboma

WHAT IS A COLOBOMA?

A coloboma is a congenital defect in the structure of either the eyelid or the eye [See figure 1].



Fig. 1: 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, retardation of growth and development, genital or urinary tract anomalies, or ear malformations (CHARGE syndrome). CHARGE syndrome has been associated with a mutation in the *CHD7* gene.

WHAT TYPES OF COLOBOMAS OCCUR IN THE EYE?

Colobomas 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 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 LENS COLOBOMA?

A lens coloboma occurs when there is a focal defect of the structures that are responsible for holding the lens in place (ciliary body and zonules). The lens shape may be asymmetric, which can cause **refractive error** (please link to appropriate FAQ) and/or **amblyopia** (link), which may require treatment with glasses and/or patching.

WHAT IS A RETINA OR OPTIC NERVE COLOBOMA?

A small or large developmental defect may occur in the deeper structures of one or both eyes[See figure 2]. 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.

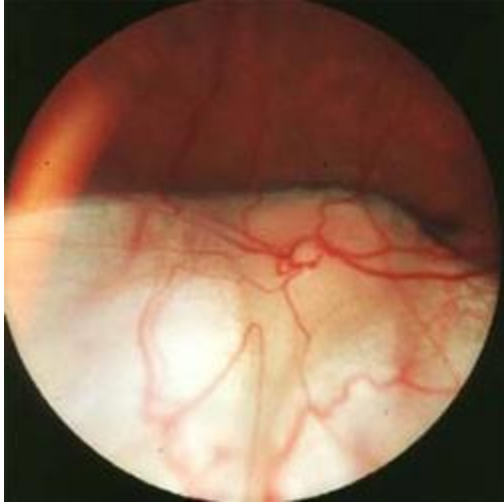


Fig. 2: Coloboma involving the retina and optic nerve

WHAT ARE THE COMPLICATIONS OF RETINA AND OPTIC DISC COLOBOMAS?

Although rare, detachment of the retina can occur any time in life and require surgical correction. Abnormal blood vessels may also grow next to the coloboma and cause bleeding, which can rarely cause vision loss. Regular follow up is important to detect these complications and treat them early.

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

If the defect extends to the central part of the retina or optic nerve, vision can be impaired, sometimes severely. The defect can sometimes cause a **refractive error** ([link](#)) or **amblyopia** ([link](#)), which may require treatment with glasses and/or patching.

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 lens coloboma is usually only visible if the eye is dilated. A white reflex from the eye (**leukocoria**, [please link](#)) may be seen with a large coloboma of the retina or optic nerve. However, an ophthalmoscope is usually required to see a coloboma of the retina or optic nerve.

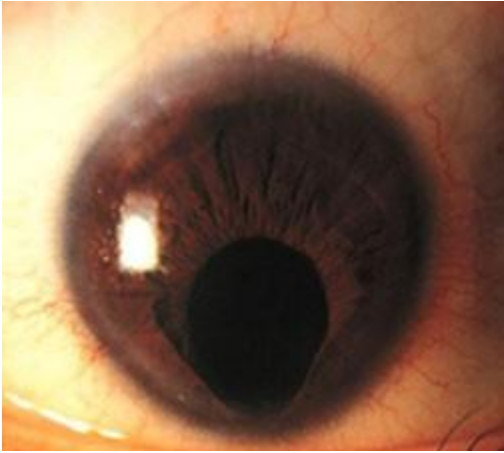


Fig. 3: Iris Coloboma

WHAT IS A COLOBOMATOUS CYST?

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

WHERE CAN I FIND MORE INFORMATION REGARDING COLOBOMA?

- [CHARGE Syndrome Foundation](#)

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