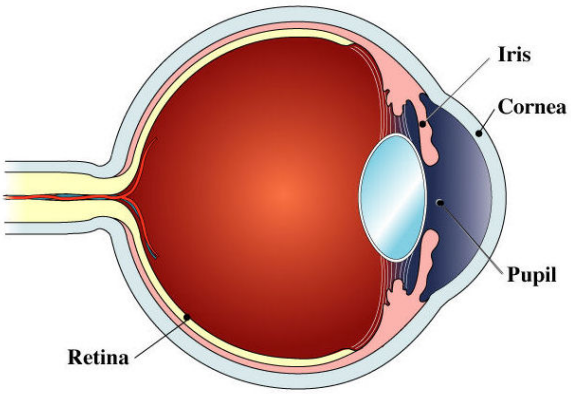
Coloboma

A coloboma is a hole in one of the parts of the eye or eyes. It can affect the lens, eyelid, retina, or any other part. Most commonly, the colored part of the front of the eye (*iris*) is affected. Colobomas may range from small missing parts of the eye, to a totally absent eye (*enophthalmos*). Congenital coloboma is present at birth.

With coloboma of the iris, part of the iris structure is missing. This results in the pupil being oval instead of round. For this reason, an iris coloboma is often called a “Cat’s Eye” or “Keyhole Iris.”



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When a coloboma occurs in the back of the eye, large areas of the back of the eye are missing or malformed.

SYMPTOMS

The problems depend on what part of the eye has been affected. If there is only a small hole in the iris, vision may be normal. If the iris defect is large, there may be light sensitivity or difficulty focusing on near objects, such as reading materials. If a large part of the retina or optic nerve is missing, vision may be partially affected, or totally absent. Portions of peripheral vision may also be dim or absent in eyes with retinal colobomas.

CAUSES

Isolated colobomas may be inherited, often in a pattern that is “dominant.” This means that if one parent has this type of coloboma, each child has a 50% chance of having one, too.

Some colobomas are inherited in a “recessive” pattern. This means that you can only be affected if both parents are “carriers” of the gene. The parents may not have a coloboma, but if they are carriers, 25% of their children may have a coloboma and 50% may be carriers of the gene.

Some people with colobomas have abnormalities in other organs. When part of the eye is destroyed by an injury, a “traumatic coloboma” may result.

DIAGNOSIS

Diagnosis is made by an eye specialist, during a complete eye exam.

TREATMENT

There is no cure for coloboma. Different visual aids may help. Small colobomas with normal vision may require no treatment. Sunglasses help combat the sensitivity to light. If the pupil is significantly distorted by an iris coloboma, special contact lenses may be used to create an artificially round pupil. In some cases, surgery can be an alternative way to create a rounder pupil. Genetic counseling is important for people with colobomas who are considering having children.

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