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Coloboma of the iris

Coloboma of the iris is a hole or defect of the iris of the eye. Most colobomas are present since birth (congenital).

Considerations

Coloboma of the iris can look like a second pupil or a black notch at the edge of the pupil. This gives the pupil an irregular shape. It can also appear as a split in the iris from the pupil to the edge of the iris.

A small coloboma (especially if it is not attached to the pupil) may allow a second image to focus on the back of the eye. This may cause:

- Blurred vision
- Decreased visual acuity
- Double vision
- Ghost image

If it is congenital, the defect may include the retina, choroid, or optic nerve.

Most colobomas are diagnosed at birth or shortly afterward.

Causes

Most cases of coloboma have no known cause and are not related to other abnormalities. Some are due to a specific genetic defect. A small number of people with coloboma have other inherited developmental problems.

When to Contact a Medical Professional

Contact your health care provider if:

- You notice that your child has what appears to be a hole in the iris or an unusual-shaped pupil.
- Your child's vision becomes blurred or decreased.

In addition to your child, you may also need to see an eye specialist (ophthalmologist).

What to Expect at Your Office Visit

Your provider will take a medical history and do an exam.

Since the problem is most often diagnosed in infants, knowing about the family history is very important.

The provider will do a detailed eye exam that includes looking into the back of the eye while the eye is dilated. An MRI of the brain, eyes, and connecting nerves may be done if other problems are suspected.

Alternative Names

Keyhole pupil; Cat eye syndrome; Iris defect

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