CONDITION

Coloboma of the iris

Learn how iris coloboma affects the pupil, vision, and eye health, including its causes, diagnosis, treatment options, and recommended follow-up.

WHAT IT IS
Congenital opening in the iris

BODY SYSTEM
Eyes and vision

MAIN SYMPTOMS
Keyhole-shaped pupil, light sensitivity, glare, blurred vision

WHAT CAUSES IT
Developmental, sometimes inherited or unknown

HOW IT IS DIAGNOSED
Slit-lamp and dilated eye examinations

MAIN TREATMENT
Vision correction and symptom management

CAN IT BE CURED?
No, iris tissue cannot be replaced

IS IT AN EMERGENCY?
Sometimes, with sudden vision loss

LONG-TERM OUTLOOK
Often stable, with poorer vision if deeper structures are involved

Overview

Coloboma of the iris is an opening or gap in the colored part of the eye. The iris controls how much light enters through the pupil. With an iris coloboma, the pupil may look like a keyhole, notch, or teardrop instead of a circle.

The condition develops before birth. Early in pregnancy, a temporary seam called the optic fissure normally closes as the eye forms. If part of this seam does not close completely, a coloboma can remain. A typical iris coloboma is usually in the lower, inner part of the iris.

Coloboma may affect one eye or both eyes. It may involve only the iris, or it may also affect structures farther back in the eye, such as the retina, choroid, or optic nerve. The retina is the light-sensitive tissue lining the back of the eye. The choroid is the layer of blood vessels beneath it, and the optic nerve carries visual information to the brain.

An isolated iris coloboma may cause few vision problems. However, a larger defect can let extra light into the eye, leading to glare or sensitivity to bright light. Vision can be more seriously affected when other parts of the eye are involved.

An eye specialist can determine which structures are affected. This distinction matters because a person with only an iris coloboma may need different monitoring from someone who also has a retinal or optic nerve coloboma.

Symptoms

The appearance of the pupil is often the most noticeable feature. The pupil may extend downward in a keyhole shape. In some people, the change is subtle and is found during an eye examination.

Possible symptoms include:

A small iris coloboma may not reduce sharp central vision. Significant vision loss is more likely when the coloboma affects the retina, macula, or optic nerve. The macula is the central part of the retina used for detailed vision.

Children may also have refractive errors, meaning the eye does not focus light clearly. These include nearsightedness, farsightedness, and astigmatism. If one eye sees less clearly during early childhood, amblyopia can develop. Amblyopia, sometimes called "lazy eye," occurs when the brain does not build normal vision from that eye.

Other eye differences may occur with coloboma. These can include an unusually small eye, known as microphthalmia, or clouding of the eye's natural lens, known as a cataract. These findings are not present in everyone.

- Sensitivity to bright light, also called photophobia
- Glare or difficulty adjusting between bright and dim settings
- Blurred or reduced vision
- A visible notch or gap in the iris
- Eyes that appear different from each other
- Poor depth perception if the eyes have unequal vision
- Eye strain or difficulty seeing in certain lighting conditions

Causes and risk factors

Iris coloboma results from a difference in eye development during the first weeks of pregnancy. It is congenital, which means it is present at birth, even if it is not recognized until later.

Some colobomas are related to a change in a gene or chromosome. They may occur as part of a genetic condition that also affects hearing, growth, the heart, the nervous system, or other parts of the body. One example is CHD7 disorder, which includes the condition historically called CHARGE syndrome. Many other genetic causes are possible.

In some families, coloboma can be inherited. The pattern of inheritance depends on the gene involved. A parent may have a mild eye finding while a child has more extensive involvement. In other families, the genetic change occurs for the first time in the affected person.

Often, no specific genetic or environmental cause is found. This does not mean that a parent caused the condition through an ordinary activity during pregnancy. Genetic testing can sometimes identify a cause, but a negative result does not rule out a genetic contribution.

Factors that may make a genetic assessment especially useful include:

An opening in the iris can also occur after eye surgery, injury, or inflammation. This is sometimes called an acquired or traumatic coloboma, although it is different from the developmental condition present at birth.

- Coloboma in both eyes
- Coloboma affecting several members of a family
- Small or underdeveloped eyes
- Hearing, heart, kidney, growth, or developmental concerns
- Other differences present from birth

Diagnosis

An ophthalmologist, a medical doctor specializing in eye care, diagnoses iris coloboma through an eye examination. The doctor looks at the shape of the pupil and examines the iris using a microscope called a slit lamp.

A complete assessment usually includes a dilated eye examination. Dilating drops temporarily enlarge the pupil so the doctor can inspect the lens, retina, optic nerve, and other structures. This helps determine whether the coloboma is limited to the iris or extends into the back of the eye.

Vision testing is adapted to the person's age and ability. Refraction checks for

nearsightedness, farsightedness, and astigmatism. In infants and young children, the clinician also looks for unequal vision, abnormal eye alignment, or other signs that could lead to amblyopia.

Additional tests may be recommended when the back of the eye is involved. These may include retinal photographs, optical coherence tomography, or eye ultrasound. Optical coherence tomography uses light waves to create detailed images of the retina and optic nerve. Ultrasound can show eye structures when they are difficult to see directly.

When coloboma affects both eyes, involves several eye structures, runs in the family, or occurs with other health differences, the care team may suggest a genetics evaluation. This can include a review of the family history, a physical examination, and genetic testing. Hearing, heart, kidney, or developmental evaluations are considered according to the person's findings rather than being required for everyone.

Treatment and management

There is no medicine that can replace the iris tissue that did not form. Treatment focuses on improving comfort, supporting vision, and managing related eye conditions.

Tinted glasses or sunglasses may reduce glare and light sensitivity. A hat with a brim can also help in bright settings. Glasses should provide appropriate ultraviolet protection, as recommended for general eye health.

A specially designed contact lens can make the pupil appear rounder and reduce the amount of extra light entering through the iris gap. These are sometimes called cosmetic, prosthetic, or iris-print contact lenses. They require professional fitting and careful cleaning because poorly fitted or contaminated lenses can injure the cornea or cause infection.

Glasses or contact lenses can correct refractive errors. In children, early correction is important because clear, balanced vision supports normal visual development. If amblyopia is present, treatment may include patching the stronger eye or another clinician-directed method that encourages use of the weaker eye. A child's eye specialist should guide this treatment and monitor progress.

Surgery may be considered when glare is severe, appearance is a major concern, or another eye operation is already needed. Depending on the anatomy, a surgeon may reshape the existing iris, close part of the gap, or consider an artificial iris device. These procedures have potential risks, including inflammation, pressure changes, infection, corneal damage, and reduced vision. They are not necessary or suitable for every person.

If vision remains limited despite standard glasses or contacts, low-vision services can help.

These services may provide magnifiers, electronic tools, lighting strategies, classroom support, or workplace adjustments.

Treatment may also be needed for associated problems such as cataract, glaucoma, eye misalignment, or retinal disease. Management depends on the structures involved and should be planned with an ophthalmologist.

Complications

An isolated iris coloboma often remains stable. The iris gap does not usually enlarge over time. The main ongoing concerns may be glare, light sensitivity, appearance, or a refractive error.

Possible complications are more varied when other parts of the eye are affected. They can include:

A retinal detachment occurs when the retina pulls away from the tissue supporting it. This risk is mainly linked to coloboma involving the back of the eye, not simply to the visible iris notch. The ophthalmologist can explain whether an individual eye has this risk.

Emotional and social effects also matter. Some people are comfortable with the appearance of the pupil, while others may feel self-conscious or receive unwanted questions. Cosmetic contact lenses, counseling, or peer support may be helpful when these concerns affect daily life.

- Amblyopia caused by unequal vision during childhood
- Reduced visual acuity from retinal, macular, or optic nerve involvement
- Cataract
- Glaucoma, which is damage to the optic nerve often associated with abnormal eye pressure
- Eye misalignment, also called strabismus
- Retinal detachment if a retinal or choroidal coloboma is present

Follow up

Follow-up depends on age, vision, and whether the coloboma affects structures beyond the iris. Routine visits may include vision testing, an updated glasses prescription, eye-pressure measurement, and a dilated retinal examination.

Children generally need regular monitoring while vision is developing. Appointments are

important even if the child does not report symptoms, because refractive errors and amblyopia may not be obvious at home. Parents and caregivers should also follow recommendations for glasses, patching, or school accommodations.

Adults with a stable, isolated iris coloboma may need only routine eye care. More frequent monitoring may be advised for glaucoma, cataract, retinal involvement, or previous eye surgery.

Arrange prompt eye care if there is a new change in vision, increasing glare, eye redness, discharge, or discomfort. Contact lens wearers should remove the lens and seek prompt advice for pain, marked redness, light sensitivity, or blurred vision.

Sudden loss of vision, a curtain or shadow across the visual field, a sudden burst of new floaters, or repeated flashes of light requires urgent same-day eye assessment. Severe eye pain, major eye trauma, or a chemical injury also needs emergency care. These symptoms are not expected from a stable iris coloboma and may signal a separate problem that needs rapid treatment.

Sources

- National Eye Institute. "Coloboma." U.S. National Institutes of Health. Reviewed November 15, 2023.
- MedlinePlus Genetics. "Coloboma." National Library of Medicine, National Institutes of Health.
- Lingam G, Sen AC, Lingam V, Bhende M, Padhi TR, Xinyi S. "Ocular coloboma-a comprehensive review for the clinician." Eye . 2021;35:2086-2109.
- Gregory-Evans CY, Williams MJ, Halford S, Gregory-Evans K. "Ocular coloboma: a reassessment in the age of molecular neuroscience." Journal of Medical Genetics . 2004;41(12):881-891.