CONDITION

Retinoblastoma

Retinoblastoma is a cancer of the light-sensitive retina that usually develops in young children.

WHAT IT IS
Cancer of the retina, usually in young children

BODY SYSTEM
Eyes and vision

MAIN SYMPTOMS
White pupil reflection, crossed eye, red eye, reduced vision

WHAT CAUSES IT
RB1 gene changes, inherited or acquired

HOW IT IS DIAGNOSED
Dilated retinal examination, usually with ultrasound and MRI

MAIN TREATMENT
Eye-directed therapy, chemotherapy, or eye removal surgery

CAN IT BE CURED?
Yes - usually when treated before spread

IS IT AN EMERGENCY?
Sometimes - severe eye pain, swelling, or sudden vision loss

LONG-TERM OUTLOOK
Often excellent survival; vision depends on tumour location and treatment

Overview

Retinoblastoma is a cancer that begins in the retina. The retina is the light-sensitive layer at the back of the eye. It sends visual information to the brain.

The condition almost always develops in babies and young children. It may affect one eye or both eyes. Tumours can sometimes form in more than one part of the retina.

Retinoblastoma matters because an untreated tumour can damage sight and spread outside the eye. Prompt assessment and specialist treatment give the best chance of curing the cancer. Treatment also aims to preserve the eye and useful vision when this can be done safely.

A white reflection from the pupil is the most recognised sign. It may be seen in photographs

taken with a flash. A new crossed or turned eye is another important sign. These findings do not always mean retinoblastoma, but they need prompt medical assessment.

Symptoms

Retinoblastoma may not cause pain or obvious discomfort at first. A parent or caregiver often notices an unusual appearance in the affected eye.

Possible signs include:

Leukocoria may be easiest to see in a photograph. Instead of the usual red-eye reflection, one pupil may look white. Lighting and camera angle can also create harmless reflections, but a repeated white pupil-especially in the same eye-should not be ignored.

Babies cannot describe changes in vision. They may stop following faces or toys, reach inaccurately, or rely more heavily on one eye. These signs can have many causes, so an eye examination is needed to find out what is happening.

- A white, pale, or yellow reflection in the pupil, called leukocoria
- A new crossed, turned, or wandering eye, called strabismus
- Reduced vision or difficulty following objects
- A red or irritated eye that does not improve as expected
- A pupil that looks different from the other pupil
- Eye pain, swelling, or a larger-looking eye in more advanced cases

Causes and risk factors

Most retinoblastomas develop after changes in a gene called RB1 . This gene normally helps control how cells grow. When its protective function is lost in a retinal cell, that cell can grow into a tumour.

There are two broad forms.

In heritable retinoblastoma , an RB1 change is present in many or all of the child's cells. It may have been inherited from a parent, but it can also arise for the first time in the child. This form is more likely to affect both eyes or create several tumours. A person with a heritable RB1 change can pass it to their children.

In non-heritable retinoblastoma , the relevant gene changes occur only within a retinal cell. This form usually affects one eye and is not passed through the family.

Rarely, retinoblastoma develops through a different genetic change. Having no known family history does not rule out a heritable form. Some parents carry an RB1 change without having had a recognised eye tumour, while others have a change in only some of their cells.

Genetic counselling and testing can help clarify whether a child and close relatives need screening.

Diagnosis

A child with suspected retinoblastoma should be assessed promptly by a specialist eye team. The main test is a detailed examination of the retina after drops have widened, or dilated, the pupils.

Young children usually need this examination under a general anaesthetic so the specialist can inspect the entire retina safely and carefully. Photographs or drawings may be used to record each tumour's location and size.

Other tests may include:

A biopsy is not normally taken from a suspected retinoblastoma. Passing a needle through the eye could allow cancer cells to escape from the tumour. Specialists can usually make the diagnosis from the eye examination and imaging.

The team also assesses whether the cancer is contained within the eye. The tumour's size, location, number, and relationship to important structures help guide treatment.

- Eye ultrasound , which uses sound waves to show the tumour and deposits of calcium commonly found within it
- MRI , which uses magnets to examine the eyes, optic nerves, surrounding tissues, and brain
- Genetic testing , usually using a blood sample and sometimes tumour tissue after eye removal

Treatment and management

Treatment is planned by a specialist team experienced in childhood eye cancer. The first goal is to cure the cancer. Preserving the eye and vision is important, but only when this can be done safely.

The plan is based on whether one or both eyes are affected, the number and size of tumours, their location, the child's age, genetic findings, and whether cancer has spread.

Small tumours may be treated directly with:

Chemotherapy may shrink tumours so they can be treated more effectively. It can be given through a vein, delivered into the artery supplying the eye, or injected into the jelly-like material inside the eye. These methods have different uses and risks. They are not suitable for every tumour.

External-beam radiotherapy is used less often than in the past. It may increase the risk of later cancers and affect the growth of nearby tissues, particularly in children with heritable retinoblastoma.

Removal of the eye, called enucleation , may be the safest treatment for a large tumour, an eye with little chance of useful vision, or cancer that cannot be controlled with eye-preserving treatment. An implant can maintain the shape of the eye socket, and a painted artificial eye can later be fitted over it.

Children need repeated eye examinations during and after treatment. Some examinations require anaesthesia until the child is old enough to cooperate. MRI or other tests may be used when there is concern about spread or an inherited genetic risk.

- Laser treatment , which damages the tumour or its blood supply
- Cryotherapy , which freezes tumour tissue
- Plaque radiotherapy , which places a small radioactive disc against the outside of the eye for a limited time

Complications

Retinoblastoma and its treatment can affect vision. A tumour near the central retina or optic nerve may cause lasting sight loss even when the eye is saved. Bleeding, retinal detachment, or increased pressure within the eye can also reduce vision.

Cancer can spread along the optic nerve or into tissues around the eye. It can then reach other parts of the body, although this is much less likely when the condition is found and treated early.

Treatment can cause temporary or lasting effects. These may include irritation, scarring, cataracts, changes in eye growth, hearing problems from some chemotherapy medicines, or infection risk during chemotherapy. The specialist team monitors for effects linked to the treatments used.

Children with heritable retinoblastoma have an increased lifetime risk of developing other cancers. The risk is higher in tissues exposed to radiotherapy, but it also exists without

radiation. Long-term follow-up and attention to new, persistent symptoms are important.

Losing an eye may affect depth perception and side vision. Most children adapt well with support, although protecting the remaining sight becomes especially important.

Prevention

There is no proven way to prevent the genetic changes that cause most retinoblastomas. Parents do not cause the condition through anything they did during pregnancy or early childhood.

For families with a known RB1 change, genetic counselling can explain inheritance and family-planning options. Testing relatives may identify babies who need eye examinations from birth. Screening does not prevent a tumour, but it can find one when it is small and easier to treat.

Children who rely on one eye should use suitable protective eyewear for activities with a risk of eye injury. This protects remaining vision but does not prevent retinoblastoma.

Living with it and what to expect

Treatment and close monitoring often continue for months. Eye examinations may be frequent at first because new tumours can appear and treated tumours can become active again. Appointments usually become less frequent as the child grows and the eyes remain stable.

When retinoblastoma is contained within the eye and specialist treatment is available, the chance of cure is very high. The visual outcome is less predictable. It depends on where the tumours developed, how large they were, and which treatments were needed.

Some children keep useful vision in both eyes. Others have reduced sight in one eye or need an eye removed. Children with limited vision can often learn, play, and attend school with practical adjustments. Vision services can help with mobility, reading, classroom access, and developmental support.

An artificial eye does not provide sight, but it can give a natural appearance. It needs routine review as the child grows. The eye socket and implant are also checked for healthy development.

Follow-up may continue into adulthood, especially for people with a heritable RB1 change. It can include cancer-risk advice, genetic counselling, vision care, and support with the emotional effects of treatment.

When to get medical help

Arrange an urgent medical assessment if you notice:

These signs often have causes other than retinoblastoma. They still need prompt examination rather than waiting for a routine sight test.

Seek emergency care for sudden major vision loss, severe eye pain, rapid swelling around the eye, or an eye that is pushed forward. A child receiving chemotherapy also needs urgent help for fever or signs of infection, following the emergency instructions provided by their cancer team.

During follow-up, contact the specialist team promptly about a new white reflection, a new change in eye position, worsening vision, or persistent problems with an implant or artificial eye. Stable vision needs, routine prosthesis care, and planned screening can usually be discussed at scheduled appointments.

- A repeated white or pale pupil reflection
- A newly crossed or turned eye
- A persistent red or painful eye without a clear explanation
- An eye that looks enlarged, cloudy, or different from the other eye
- A baby who stops following faces or objects

Common questions

Is retinoblastoma always cancer?

Yes. Retinoblastoma is a cancer of retinal cells. However, not every white pupil, crossed eye, or retinal growth is retinoblastoma. A specialist eye examination is needed to identify the cause.

Can a camera flash really show retinoblastoma?

Sometimes. A tumour can reflect light and make the pupil look white in a photograph. Camera angle and lighting can also cause harmless pale reflections. A repeated white reflection in the same eye needs prompt assessment, even if the eye sometimes looks normal.

Will my child have to lose the eye?

Not always. Small and medium-sized tumours can often be treated while keeping the eye. Removal may be recommended when a tumour is large, vision cannot realistically be saved, or eye-preserving treatment would be less safe. The priority is curing the cancer.

Is retinoblastoma inherited?

Some cases are heritable and others are not. Retinoblastoma in both eyes, several tumours, or a family history makes a heritable RB1 change more likely. Genetic testing can provide more reliable information for the child and family.

Can retinoblastoma come back after treatment?

A treated tumour can become active again, and children with a heritable form can develop new retinal tumours while they are young. This is why repeated examinations are important. The likelihood generally falls as the retina matures, but long-term follow-up may still be needed.

Will my child's other eye be affected?

A child with a heritable RB1 change may develop tumours in both eyes, even if only one eye is affected at diagnosis. Regular examinations can detect small tumours early. In a confirmed non-heritable case affecting one eye, the other eye is much less likely to develop retinoblastoma.

Sources

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