Children's Cancer and Leukaemia Group | the EXPERTS in CHILDHOOD CANCER

Retinoblastoma in children

An information guide for parents and families

The purpose of this guide is to give information about retinoblastoma to help you understand more about the type of cancer your child has.

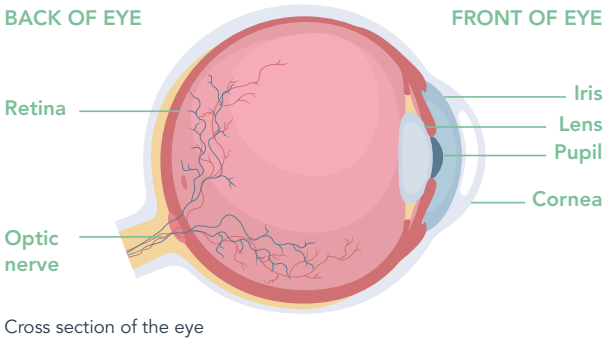
Information in this guide should be used to support professional advice specific to your child's diagnosis. If you have any questions, it is important to ask your child's medical team.

What is retinoblastoma?

Retinoblastoma is cancer of the retina, the light sensitive lining of the eye. It usually affects children under five and about 40 to 50 cases are diagnosed in the UK each year. Treatment is very effective and nearly all (98 out of 100) children with retinoblastoma are cured.

There are two forms of retinoblastoma:

- heritable form - passed on in the family and some children have an affected parent: tumours are often in both eyes (bilateral) or sometimes only in one eye
- non-heritable form - this is not passed on in the family and only one eye is affected



Cross section of the eye

Causes of retinoblastoma

In nearly all cases, retinoblastoma is caused by an abnormality in the retinoblastoma gene (RB1). In the heritable form of retinoblastoma, which accounts for around half of all cases, this abnormal gene is either inherited from a parent or happens for the first time at an early stage of development in the womb.

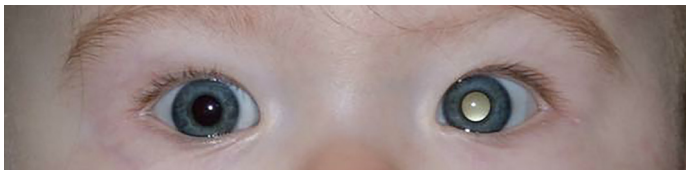
Genetic counselling and support is available for families in which a member has retinoblastoma. Not all children of a parent with retinoblastoma will inherit this gene, but those born into families with a history of retinoblastoma are offered blood testing to look for the abnormal gene. Children found to have inherited the altered RB1 gene from a parent or those known to be at risk will be offered screening. Screening usually starts shortly after birth and is repeated regularly. This means that treatment can be started early if a tumour does develop.

In the non-heritable form of retinoblastoma, the abnormality in the RB1 gene occurs in just one cell on the retina. The reason for this happening is unknown.

Signs and symptoms

In some children with a family history of retinoblastoma, a tumour is picked up by screening before they have any symptoms. If there's no family history of retinoblastoma, the first sign of the condition is often a white pupil that does not reflect light (leucocoria). This may be noticed when a picture of your child is taken using a flash. The

pupil of the affected eye may look white in the photo (see below). Some children may have a squint or, if the tumour is large, they may have a painful red eye.



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How it is diagnosed

A variety of tests and investigations may be needed to diagnose retinoblastoma. Any tests and investigations that your child needs will be explained to you. If the doctors suspect that your child may have retinoblastoma, you will be referred to one of the two specialist centres in the UK for confirmation of the diagnosis and treatment. These centres are in Birmingham and London.

Examination under anaesthetic (EUA) – a specialist (ophthalmologist) examines your child's eye while they're asleep. Retinoblastomas can be diagnosed by their appearance. Your child will have more EUAs during and after treatment.

Your child may have other tests to check the size and position of the tumour, and whether it has spread into surrounding structures. These tests may include:

- ultrasound scan
- lumbar puncture (spinal fluid sample)
- bone marrow sample
- blood test
- MRI scan
- OCT (optical coherence tomography) scan - uses reflected light to create detailed images of the back of the eye
- AFS (aqueous fluid sampling) - obtains DNA from the tumour by taking a small amount of fluid from the front of the eye; this helps doctors understand more about the genetics of the tumour

Staging

The 'stage' of a cancer is a term used to describe its size and whether it has spread beyond its original site. Knowing the type and stage of the cancer helps doctors to decide on the most appropriate treatment.

The staging system below is commonly used:

- intraocular retinoblastoma – cancer is in one or both eyes, but it has not spread to the surrounding tissues
- extraocular retinoblastoma – the cancer has spread outside the eye into the nerve, surrounding tissue, or to other parts of the body

Intraocular retinoblastoma may be sub-divided into five different grades, depending on the size and position of the cancer and whether there's any damage to the eye. The two most commonly used staging systems are IIRC (staging from A to E) and TNM system (staging from cT1 to cT4).

Treatment

Treatment will depend on the number, position, and size of tumours in your child's eye. The aim of treatment is, firstly, to get rid of the cancer and, secondly, to try to keep as much sight as possible. Your child's doctor will talk to you about the possible risks as well as the advantages of the treatment your child is offered.

Local therapy

For smaller tumours, treatment can be given to the eye while your child is asleep under anaesthetic. This is called local therapy and any combination of the following methods may be used:

- cryotherapy – a freezing treatment applied to the outside of the eye; more than one session may be necessary, usually done at monthly intervals
- laser therapy – uses a laser directed through the pupil to heat the tumour; your child may need a number of sessions at intervals of 3-4 weeks
- plaque – uses a small radioactive disc stitched on the outside part of the eye, overlying the tumour which stays in place for up to four days while the radiation destroys the cancer cells (during this time your child will need to stay in hospital in isolation); this method is used for larger tumours, or when other methods have not been successful
- thermotherapy – uses heat from a laser to destroy the cancer cells and may be combined with chemotherapy or radiotherapy, to improve the effectiveness of these treatments

Chemotherapy

Chemotherapy is the use of anti-cancer drugs to destroy cancer cells and may be used for larger tumours, if the cancer has spread, or if there's a risk that it may spread. It is usually used in combination with local therapy. The drugs most commonly used to treat retinoblastoma are carboplatin, etoposide, and vincristine. Your child's doctor will discuss which is most suitable for your child.

- systemic chemotherapy is given into the veins – it may be given before any local therapies, to help shrink the tumour and make treatment more successful
- intra-arterial chemotherapy (IAC) is injected directly into the blood vessel that supplies the eye – it allows higher doses of chemotherapy to reach the eye with less side effects to the rest of the body than systemic chemotherapy; IAC may be used instead of systemic chemotherapy, or when the tumour has not responded to initial treatment
- intra-vitreal chemotherapy (IViC) is injected into the central gelatinous part of the eye – it allows higher doses of chemotherapy to reach this area and is used to treat retinoblastoma seeds or recurrences on the retina, and is usually combined with either systemic chemotherapy or IAC

Surgery

If the tumour is more advanced and the vision in the eye is lost, your child may need an operation to remove the eye. This operation is called an enucleation. This will only be done if it is absolutely necessary. An artificial eye is then fitted.

Proton beam radiotherapy

Radiotherapy uses high energy rays to destroy the cancer cells. Radiotherapy may be used if there are cancer cells left behind in the orbit or optic nerve after enucleation or to treat retinoblastoma in the eye when other treatments have not been successful. It can be given to the whole eye but does have some effect on the surrounding tissue. Radiotherapy for retinoblastoma is normally only used when other treatments have not worked well.

Side effects of treatment

Treatment for retinoblastoma can cause side effects, and your child's doctor will discuss these with you before the treatment starts. Any side effects will depend on what treatment is being used. Most side effects are short-term and gradually disappear once treatment stops. Side effects can include:

- feeling sick (nausea) and being sick (vomiting)
- temporary hair loss
- tiredness
- low blood count leading to an increased risk of infection and bruising and bleeding (sometimes blood and/or platelet transfusions are needed)
- a sore mouth and tummy
- diarrhoea

Clinical trials

Many children have their treatment as part of a clinical research trial. Trials and studies are carried out to try to improve our understanding of the best way to treat an illness. There are better results for curing children's cancers compared with just a few years ago because of clinical trials. Your child's medical team will talk to you about taking part in a clinical trial and will answer any questions you have. Taking part is completely voluntary, and you'll be given plenty of time to decide if it's right for your child. You may decide not to take part, or you can withdraw from a trial at any stage. Your child will still receive the best standard treatment available.

National treatment guidelines

Sometimes, clinical trials are not available for your child's cancer. In this case, your doctors will offer the most appropriate treatment, using guidelines which have been agreed by experts across the UK. Children's Cancer and Leukaemia Group (CCLG) is an important organisation which helps to produce these guidelines.

Donating to a tissue bank

Retinoblastoma is a rare disease and more research is needed to help doctors develop better treatment for the future. Your child's hospital team will offer you the opportunity to anonymously donate tissue left over from tests carried out, for example, a biopsy or bone marrow test, to the tissue bank. This sample can then be used by scientists to learn more about retinoblastoma and how best to treat it. This is voluntary, and you will have plenty of time to decide if you wish to take part.

Late side effects

Months or years later some children may develop late side effects from the treatment they have had. These may include a change in vision, a change in the way the heart, lungs and kidneys work, reduction in bone growth, a risk of infertility and an increased risk of developing another cancer in later life. It is important to understand that not all side effects will happen to all patients. For more information, please visit www.cclg.org.uk/living-beyond-cancer

Your child's doctor or nurse will talk to you about any possible late side effects and will keep a close eye on possible long-term side effects in follow-up clinics.

Follow-up care

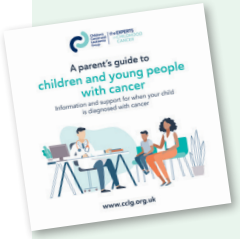
At least 9 out of every 10 children with retinoblastoma are cured. Once treatment has finished, the doctors will monitor your child closely with regular examinations under anaesthetic to check the retina is healthy, the cancer has not come back, and no new tumours have developed. Your child's general health and any long-term effects will also be monitored. After a while, you will not need to visit the clinic so often.

Children with heritable retinoblastoma will be given genetic counselling when they are old enough to understand it. Having the heritable form of retinoblastoma is associated with an increased risk of developing other types of cancer later in life. Your child will be followed up closely into adulthood. They will be encouraged to report any new symptoms, such as a lump, early, and to have a healthy lifestyle to help to reduce cancer risk.

Your feelings

It's devastating to hear that your child has cancer and you may feel overwhelmed but there are many professionals and organisations to help you through this difficult time. You may have many feelings, such as fear, guilt, sadness, anger, and uncertainty. These are all normal reactions and are part of the process that many parents go through.

It's not possible to address in this guide all of the feelings you may have. However, the CCLG booklet 'A parent's guide to children and young people with cancer', talks about the emotional impact of caring for a child with cancer and suggests sources of help and support. Your child may have a variety of powerful emotions throughout their experience of cancer. The Parent's Guide discusses these further and talks about how you can support your child.



The CCLG booklet 'A parent's guide to children and young people with cancer' is available FREE of charge from your child's hospital

All CCLG booklets and leaflets can be downloaded or ordered from our website:

www.cclg.org.uk/publications

USEFUL ORGANISATIONS

Children's Cancer and Leukaemia Group (CCLG) publishes a variety of free resources to order or download.
www.cclg.org.uk

Young Lives vs Cancer offers practical support to children and young people with cancer and to their families
www.younglivesvscancer.org.uk

Macmillan Cancer Support offers support and advice to those affected by cancer.
www.macmillan.org.uk

The Childhood Eye Cancer Trust (CHECT) provides support and information about retinoblastoma.
www.chect.org.uk

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With thanks to Dr Helen Jenkinson, Consultant Paediatric Oncologist at Birmingham Children's Hospital, who reviewed this factsheet on behalf of the CCLG Information Advisory Group, comprising multi-professional experts in the field of children's cancer.

Children's Cancer and Leukaemia Group (CCLG) is a leading national charity and expert voice for all childhood cancers.

Each week in the UK and Ireland, more than 30 children are diagnosed with cancer. Our network of dedicated professional members work together in treatment, care and research to help shape a future where all children with cancer survive and live happy, healthy and independent lives.

We fund and support innovative world-class research and collaborate, both nationally and internationally,to drive forward improvements in childhood cancer. Our award-winning information resources help lessen the anxiety, stress and loneliness commonly felt by families, giving support throughout the cancer journey.

Our work is funded by donations. If you would like to help, text 'CCLG' to 70085 to donate £3. This will cost £3 plus a standard rate message.

We are grateful to all those who have contributed to this publication. We make every effort to ensure that this information is accurate and up to date at the time of printing. CCLG does not accept any responsibility for information provided by third parties including those referred to or signposted to in this publication. Information in this publication should be used to supplement appropriate professional or other advice specific to your circumstances.

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