Retinoblastoma is an uncommon eye cancer that usually affects children under five, although it can affect children of any age. About 40 to 50 cases of retinoblastoma are diagnosed in the UK each year. Treatment is very effective and nearly all (98 out of 100) children with retinoblastoma are cured.

More children than ever are surviving childhood cancer. There are new and better drugs and treatments, and we can now also work to reduce the after-effects of having had cancer in the past.

It is devastating to hear that your child has cancer, and at times it can feel overwhelming. There are many healthcare professionals and support organisations to help you through this difficult time.

Understanding more about the cancer your child has and the treatments that may be used can often help parents to cope. We hope you find the information here helpful. Your child’s specialist will give you more detailed information and if you have any questions it is important to ask the specialist doctor or nurse who knows your child's individual situation.

This factsheet is published in conjunction with CCLG's booklet entitled ‘Children and Young People with Cancer: A Parent’s Guide’.

Retinoblastoma

Retinoblastoma develops in the light-sensitive lining of the eye (called the retina).

There are two forms of retinoblastoma:
- A heritable form - this is genetic or inherited. There are often tumours in both eyes (bilateral) or sometimes only in one eye.
- A non-heritable form - this is not passed on in the family. There’s one tumour in only one eye (unilateral).

Causes of retinoblastoma

The heritable form of retinoblastoma, which accounts for about two in every five cases, is caused by a genetic abnormality. This means that an abnormal gene allows the tumour to develop. This abnormal gene may either be inherited from a parent or happen 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 children born into families with a history of retinoblastoma are offered blood testing. They are also usually checked (screened) soon after birth for signs of retinoblastoma. This means that treatment can be started early if a tumour does develop.

Screening usually starts shortly after birth and is repeated every few months for five years. An eye specialist examines the eye, while shining a light into it with an opthalmoscope. In younger children this may need to be done under a general anaesthetic.

The genetic abnormality in the heritable form of retinoblastoma is now well-understood. But we don’t know what causes non-heritable retinoblastoma.
**Signs and symptoms**

Some children with a family history of retinoblastoma are 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 picked up when a picture of your child is taken using flash photography. The pupil of the affected eye may look white in the photograph.

Some children may have a squint or, if the tumour is large, they may have a painful red eye.

**How a retinoblastoma is diagnosed**

Tests are likely to involve an examination under anaesthetic (EUA) where an eye specialist (ophthalmologist) examines your child’s eye while they’re asleep. Unlike nearly all other types of cancer, retinoblastomas can be diagnosed just by their appearance. So it’s not usually necessary to take a sample of tissue (a biopsy). Your child will have several more EUAs to check how treatment is progressing.

When a retinoblastoma is diagnosed, your child may have some other tests to check the exact position and size of the tumour, and whether it has begun to spread into surrounding structures. This is known as staging.

They may have some of the following tests:

- An ultrasound scan. This is a painless scan that uses sound waves to examine the eye and the surrounding area.
- An MRI (magnetic resonance imaging) scan. This uses magnetism to build up a detailed picture of the eye and the head.
- A lumbar puncture. The doctor inserts a fine needle between the bones in the lower spine to remove a sample of the fluid from around the brain and spinal cord (cerebrospinal fluid). The fluid is examined under a microscope to check whether there are any cancer cells present.
- A bone marrow sample. This may be taken to check whether there are any cancer cells in the bone marrow (where our blood cells are made). The doctor inserts a needle into the hip bone and draws some of the bone marrow out with a syringe so that it can be checked for any cancer cells.
- A bone scan. This involves taking a series of x-rays to check for signs of any spread to the bones.
- A blood test. This may be taken for genetic testing for the Rb gene. The results of this test can take some months.

The doctor or a specialist nurse will explain more about the tests that your child needs.

**Staging**

The stage of a cancer is a term used to describe its size and whether it has spread from where it first started. Knowing the stage of your child’s retinoblastoma helps the doctors to decide on the most appropriate treatment.

The following staging system is commonly used for retinoblastoma:

**Intraocular retinoblastoma**

There is cancer in one or both eyes, but it has not spread to other parts of the eye or the tissues surrounding the eye. This stage may be sub-divided into five grades (A–E), depending on the size and position of the cancer and on whether there’s any damage to the eye.

**Extraocular retinoblastoma**

The cancer has spread outside the eye into surrounding tissue, or to other parts of the body.

**Treatment**

This depends on the number, position and size of the tumours in the eye. The aim of treatment is firstly to get rid of the cancer and secondly to try to keep the sight in the eye. Some treatments may cause changes to the vision in the affected eye. Your child’s specialist will talk to you about the possible risks as well as the advantages of the treatment.

**Smaller tumours**

For smaller tumours, treatment is given to the eye itself (called local therapy), while your child is asleep and under anaesthetic. One of the following methods may be used:

**Cryotherapy**

This is used to freeze the tumours. More than one session may be necessary, in which case they are usually done at monthly intervals.

**Laser therapy**

A laser is used to heat the tumour. Your child may need a number of sessions at intervals of 3-4 weeks.

**Plaque**

A small radioactive disc is stitched over the tumour on the outside of the eye. The disc needs to stay in place for up to four days while the radiation destroys the
cancer cells. This is done for slightly larger tumours, or tumours that have not been successfully treated with other methods.

**Thermotherapy**

This process uses heat to destroy the cancer cells and may be combined with chemotherapy or radiotherapy, as heat can improve the effectiveness of these treatments. The heat is produced by a laser, which is directed at the tumour.

**Larger tumours**

These can be treated in a number of ways, including:

**Chemotherapy**

This is the use of anti-cancer (cytotoxic) drugs to destroy cancer cells. It may be given before the local treatments mentioned above, to help shrink the tumour and make treatment more successful. Chemotherapy can also be used if the cancer has spread to other parts of the body, or if there's a risk that it may spread. The chemotherapy drugs most commonly used to treat retinoblastoma are carboplatin, etoposide, and vincristine.

Newer techniques include giving chemotherapy directly into the artery (the blood vessel) that supplies the eye (intra-arterial chemotherapy) or injecting chemotherapy into the central gelatinous part of the eye (intra-vitreal chemotherapy).

**Surgery**

If the tumour is very large and the vision in the eye is lost, your child is likely to need an operation to remove the eye. The specialist will only do this if it is absolutely necessary. An artificial eye is then fitted.

**Radiotherapy**

Radiotherapy treats cancer by using high energy rays from a machine to destroy the cancer cells, while doing as little harm as possible to normal cells. 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**

The side effects will depend on the treatment your child is having. The immediate side effects will usually improve when treatment finishes. But unfortunately some treatments may cause side effects that develop many years later. The specialist will explain all of this to you before treatment starts, and your child's follow-up will include checking for any long-term effects of treatment.

**Clinical trials**

Many children have their treatment as part of a clinical research trial. Trials aim to improve our understanding of the best way to treat an illness, usually by comparing the standard treatment with a new or modified version. Specialist doctors carry out trials for children's cancers.

If appropriate, your child's medical team will talk to you about taking part in a clinical trial, and will answer any questions you have. Written information is provided to help explain things.

Taking part in a research trial is completely voluntary, and you'll be given plenty of time to decide whether it's right for your child.

**Treatment guidelines**

Sometimes, clinical trials are not available for your child's tumour. This may be because a recent trial has just finished, or because the tumour is very rare. In these cases, you can expect your doctors and nurses to offer treatment which is agreed to be the most appropriate, using guidelines which have been prepared by experts across the country. The Children's Cancer and Leukaemia Group (CCLG) is an important organisation which helps to produce these guidelines.

**Follow-up care**

At least 9 out of every 10 children with retinoblastoma are cured. Following treatment, the eye specialist will frequently examine your child's eye under anaesthetic to check that the retina is healthy, the cancer has not come back, and no new tumours have developed. Follow-up is usually in a clinic for childhood cancers, called a paediatric oncology clinic.

Children with heritable retinoblastoma will be given genetic counselling when they are old enough to understand it.

Having the heritable form of retinoblastoma means 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 get any new symptoms, such as a lump, checked early, and to have a healthy lifestyle to help to reduce cancer risk.
Your feelings

As a parent, the fact that your child has cancer is one of the worst situations you can be faced with. You may have many emotions, such as fear, guilt, sadness, anger and uncertainty. These are all normal reactions and are part of the process that many parents go through at such a difficult time. It’s not possible to address in this factsheet all of the feelings you may have. However, the CCLG booklet ‘Children & Young People’s Cancer; A Parent’s Guide’, 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.

Useful organisations

Children’s Cancer and Leukaemia Group (CCLG)
www.cclg.org.uk
CCLG supports the 1,700 children who develop cancer each year in the UK. It gives support to healthcare professionals involved in caring for children with cancer and is instrumental in the development of high standards of care.

CLIC Sargent
www.clicsargent.org.uk
CLIC Sargent offers practical support to children and young people aged 21 and under, with cancer or leukaemia, and to their families.

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

The Childhood Eye Cancer Trust (CHECT)
www.chect.org.uk
CHECT is a national charity that provides support and information about retinoblastoma.

References

This factsheet has been compiled using a number of reliable sources, including:


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