Retinoblastoma

Retinoblastoma is a type of eye cancer that develops in the light-sensitive lining of the eye, called the retina. Retinoblastoma can occur at any age but mainly occurs in children younger than 5 years of age and most often in those younger than 2.

Retinoblastoma may occur in one or both eyes, but rarely spreads to other parts of the body. Although it is the most common eye tumour in children, it is a rare childhood cancer and accounts for about 3-4% of childhood cancers.

There are 2 Types of Retinoblastoma:

- **Inherited:** Retinoblastoma is sometimes inherited from the parent; this is known as hereditary or germline retinoblastoma and is usually bi-lateral (in both eyes).

- **Non-Inherited:** This type of Retinoblastoma generally occurs in only one eye (unilateral)

The main challenge of treating Retinoblastoma is the prevention of blindness, however approximately 98% of children with retinoblastoma are cured.

Causes of Retinoblastoma

Hereditary or germline retinoblastoma is caused by a gene mutation within the q14 band of chromosome 13. This abnormality can either be passed on from parent to child or it can develop for the first time at an early stage of development in the womb.

Not all children of a parent that has this abnormal gene will inherit it and get retinoblastoma, but all children of such a parent(s) should be screened for signs of retinoblastoma soon after birth. This screening should be repeated every 6 months until the child is 5 years old.

Children with hereditary retinoblastoma have an increased risk for developing pineal tumours in the brain, known as trilateral retinoblastoma and other cancers such as bone or soft tissue sarcoma or melanoma in later years

Although the genetic abnormality in hereditary retinoblastoma is understood, the cause of non-inherited retinoblastoma is unknown.

Signs and Symptoms

Some of the typical signs and symptoms of Retinoblastoma are:

- **White Pupil:** There is a whitish colour behind the pupil that does not reflect light. This can easily be seen in photographs taken with a flash; the pupil will look white instead of dark
- **Crossed Eyes:** A squint or problems with eye movements (crossed eyes)
- **Red Eye:** A red painful irritation may persist if the tumour is large

Tests and Diagnosis

Retinoblastoma can be diagnosed using the following tests and procedures:

- **Physical Exam and History:** The doctor will perform an physical examination, checking your child's general health as well as checking for anything unusual or signs of cancer, and a complete medical history will be taken
- **Eye exam with dilated pupil:** Medicated eye drops will be put into the eye(s) to make the pupils open wide and allow the doctor to look through the lens to the retina. This may be done under general anaesthesia, depending on the age of the child
- **CT scan (CAT scan):** A CAT scan takes a series of detailed pictures of the inside of the body from different angles, using a computer linked to an x-ray machine; a dye may also either be swallowed or injected into a vein to allow the organs and tissues to show up more clearly on the pictures.

Other tests that may be done if retinoblastoma is detected, and may include an MRI (magnetic resonance imaging), a lumbar puncture, bone scan, bone marrow sample, blood test, or an ultrasound.

Treatment options

Treatment options for retinoblastoma differ, depending on the position, size and number of tumours in the eye. The first aim of the treatment is to get rid of the cancer and the second is to retain sight in the eye.

Treatment options consist of:

Smaller Tumours

Smaller tumours are treated locally (in the eye itself) while the child is asleep under general anaesthetic.

- **Cryotherapy:** The tumours are frozen. If more than one session is required they will be done at monthly intervals

- **Laser therapy:** This is the opposite of cryotherapy; the tumour is removed by heating it. This may take several sessions, which will take place with intervals of 3-4 weeks
- **Plaque:** In the case of larger tumours or tumours which could not be successfully treated by other methods, a small radioactive disc is placed over the tumour on the outside of the eye with stitches. This disc must stay in place for up to four days while the tumour is destroyed by radiation.
- **Thermotherapy:** Heat is produced by a laser and used to destroy the cancer cells, usually combined with chemotherapy

Larger tumours

- **Chemotherapy:** The use of a combination of anti-cancer drugs to destroy or shrink cancer cells is the preferred treatment for children, and may be done prior to and after surgery
- **Surgery:** If the tumour is very large and vision ahs already been lost, surgery may be necessary to remove the eye and replace it with an artificial eye. This is only done in the most extreme of circumstances
- **Radiotherapy:** High-energy rays that destroy the cancer cells but do minimal harm to normal cells. Radiotherapy is normally only used for retinoblastoma when other treatments have not worked

Some of the treatment options may result in after-effects such as nausea, vomiting, irritation or soreness of the skin from radiation, hair loss, a risk of infection, fatigue, bruising and bleeding or diarrhoea. The doctor should explain all of this to you, but if they do not, please ask them about side effects.

Awareness Ribbon Colour

The awareness ribbon colour for Retinoblastoma is White

Article researched and written on behalf of Little Fighters Cancer Trust by Billi du Preez of Red Feather Scribes

Academic Editing by Dr Marc Hendricks MBChB (UCT), DIP PEC (SA), DCH (SA), FCPaeds (SA), CMO Paeds (SA) Senior Specialist, Paediatric Oncologist Red Cross War Memorial Children's Hospital