Retinoblastoma

By Juliette Carter, 16 April 2009

There are many long-term issues faced by people who have had childhood eye cancer. By Juliette Carter

Retinoblastoma is a rare eye cancer of early childhood. The cancer is often fast growing and in some cases very aggressive.

Retinoblastoma is very treatable and it has one of the most successful survival rates of all the childhood cancers. However, there are lifelong implications for adult survivors.

These patients may have a need for genetic counselling, or require support for the late effects of the treatments they received. Patients with some forms of retinoblastoma also have an increased risk of developing second primary cancers, although many patients are not aware of this risk themselves.

Incidence

Retinoblastoma is rare, affecting 1:20,000 live births, which accounts for approximately 40 cases each year.[1] It may be unilateral or bilateral.

There are two forms of the condition: 'heritable' - those who carry in their germ cells the genetic mutation in the RB1 gene, and 'non-heritable' - those who do not.

All bilateral disease is heritable, as are about 15 per cent of unilateral cases, but most unilateral cases do not have a germ cell mutation. Those with the heritable condition have a 50 per cent chance of passing on the mutation to their children.

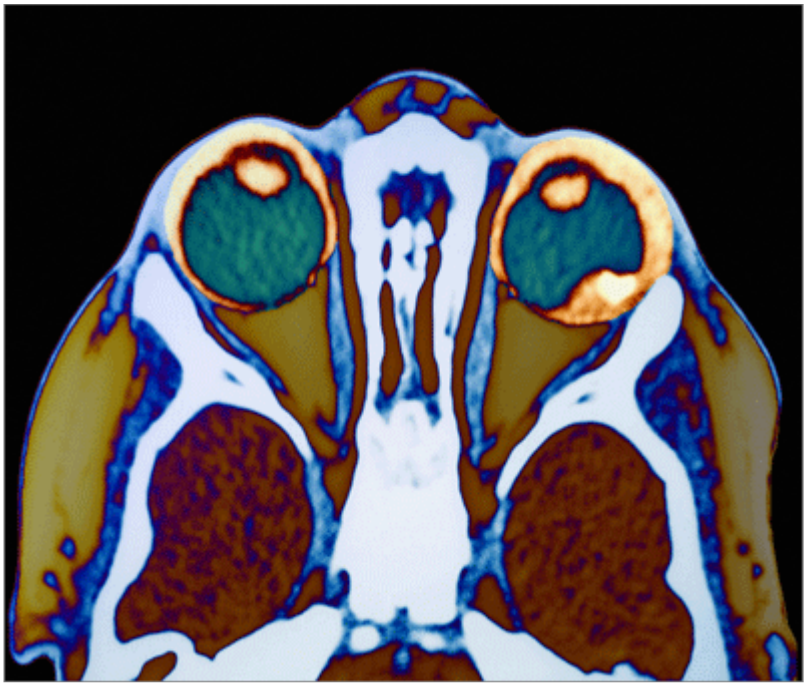
Genetics

Because patients with heritable retinoblastoma have a 50 per cent chance of passing on the affected gene mutation to their offspring they should be offered genetic counselling.

Patients who do not know if they have the heritable form can have this clarified by genetic testing. In both cases this can help a patient find out more about their own condition and provide information about their options prior to conception.

It can take up to a year to carry out the testing to identify any gene mutation that has caused retinoblastoma in an individual, so early referral for genetic counselling is advisable.

Genetic testing may be used in pregnancy to determine whether a child is likely to be affected with retinoblastoma. This can only be done if the genetic change responsible for retinoblastoma in that family has been identified.



Many couples choose not to have antenatal testing, not wishing to consider terminating a pregnancy if the retinoblastoma gene is discovered. However, it is available for couples who feel unable to cope with an affected child, for example if they already have a child undergoing surveillance or treatment.

Pre-implantation genetic diagnosis (PGD), which is carried out during IVF allows genetic testing to take place before an embryo is implanted in the womb. PGD is a relatively new service provision and although accurate, has a relatively low rate of achieving a pregnancy.

Anyone with retinoblastoma can either be referred to a local genetic counsellor or be seen by one of the two specialist retinoblastoma genetics services (see box).

Consequences of treatment

The current treatment of retinoblastoma now includes new chemotherapeutic regimens attempting to minimise tissue damage.

Most recently intra-arterial chemotherapy was introduced. A catheter is inserted into the femoral artery and advanced using fluoroscopic guidance to the ophthalmic artery, the drugs are then injected straight into the ophthalmic artery.

Twenty years ago, the main options were radiotherapy and enucleation. Although they were often very effective in curing the tumours, they both have significant consequences.

Patients living with the late effects of treatments may experience a range of problems including cataracts, dry eye and visual impairments, living with an artificial eye and sometimes facial disfigurement. This is because often in the past no implant was used to improve cosmetic effect and the tissue around the eye socket could drop. Radiotherapy could also alter facial bone development.

Second primary cancers

It has been recognised for some time now that patients with the heritable form of retinoblastoma have an increased risk of developing a second primary cancer.[2]

Second tumours may develop anywhere in the body and many patients are unaware of this associated risk. Some such tumours are malignant melanomas, squamous cell carcinomas and sarcomas.

There is no regular reliable screening for second tumours. Regular X-rays or whole-body CT scans may well be harmful.

Patients with a visual impairment may seek frequent appointments with their GP for a complete body skin check, since they will not be able to see any potentially malignant skin lesions themselves.

Patient resources

Retinoblastoma is a treatable cancer; however, there are many problems that may affect patients later on in life. Patients who had retinoblastoma may not be aware of these problems. They may seek advice on issues of genetics, family planning, late effects of treatment and concerns over second primary cancers.

The Childhood Eye Cancer Trust is funding a project, from October 2008 to 2010, aimed at identifying those potentially at risk of passing on the mutant gene to any offspring and developing second primary cancers by recalling appropriate patients for molecular investigations.

It is important for GPs to be aware of the life-long support needs of someone who has had retinoblastoma because patients will often use primary care as their first port of call to access relevant services.

Juliette Carter is a trained paediatric nurse who now works for the Childhood Eye Cancer Trust as a support worker and information officer (**www.chect.org.uk**)

Genetic and retinoblastoma services
Dr Trevor Cole, consultant geneticist at Birmingham Children's Hospital - **www.bch.org.uk/departments/retinoblastoma.htm**

Dr Elisabeth Rosser, consultant geneticist at Bart's and the London NHS Trust - **www.bartsandthelondon.org.uk/Retinoblastoma_Service/**

The Royal London Hospital, Retinoblastoma Service - **www.bartsandthelondon.nhs.uk/Retinoblastoma_Service/**

Birmingham Children's Hospital, Retinoblastoma Service **www.bch.org.uk/departments/retinoblastoma.htm**

References

1. MacCarthy A, Birch J, Draper G et al. Retinoblastoma: treatment and survival in Great Britain 1963 to 2002. Br J Ophthalmol 2009; 93(1): 38-9.

2. Halford L, Cole T, Kingston J, Onadin Z, Reddy A. Retinoblastoma for life. Focus: The Royal College of Ophthalmologists. Summer 2008 5-6.