Supporting adults who have had retinoblastoma

The effects of retinoblastoma on a patient can be lifelong. Anyone who has had retinoblastoma (Rb) should be monitored to detect and manage any long-term problems caused by the disease or treatment in order to ensure they are picked up as quickly as possible.

Screening

Rb is heritable in 45% of cases and a child who inherits the altered Rb gene has a 90% chance of developing retinoblastoma, so screening from birth at an Rb treatment centre is vital.

Genetics

Adults who had Rb as a child may wish to consider genetic screening. For more information on the genetic implications of retinoblastoma visit the "Genetics" page of our website. In cases where the Rb is heritable (all bilateral and some unilateral cases), patients of childbearing age should be seen by a geneticist who will explain the implications and considerations. Information on the genetics of Rb was scarce 30 years ago therefore many patients who carry the altered gene may not be aware of the risks associated with this, including malignant melanoma, and cancer of the bladder, uterus or lung.

Patients planning a family may wish to consider testing in pregnancy, which can include non-invasive prenatal diagnosis (NIPD), from eight weeks of pregnancy; chorionic villus sampling at 11 weeks of pregnancy; or amniocentesis at 16 weeks of pregnancy. Alternatively, they may wish to consider pre-implantation genetic testing for monogenic disorders (PGT-M).

Anyone considering having testing in a pregnancy should be seen by a geneticist before they plan a pregnancy so that the necessary background work can be done.

If doctors have obtained results that mean they can offer an accurate test to a baby then they arrange for a blood sample to be taken from the umbilical cord when the baby is delivered. The testing can then be done quickly so that the results take less than two weeks.

Second cancers

There is an increased risk of second primary cancers both to those who have undergone radiation treatment for their Rb, and to those with the heritable form of Rb, but in general there should be a low threshold for referral for suspected cancer for this group. Patients surviving this form of Rb have an increased risk of developing sarcomas within about 5-25 years of treatment. They are also at an increased risk of developing other forms of cancer into later life. The level of risk depends also on the treatment given for the Rb.

- Patients should be monitored for any worrying or persistent problems such as unexplained lumps or pains, new moles or changes to an existing mole.

• Adults with heritable Rb should be seen regularly in an adult late effects clinic. The referral letter should detail the treatment the patient had as a child – if available. If for any reason this is not possible, contact the relevant Rb team (details below) or the Childhood Eye Cancer Trust (CHECT) will be able to point you in the right direction.

Effects of treatment:

• Late effects of chemotherapy for Rb can include hearing and kidney problems.

• Late effects of radiotherapy can include second malignancies, cataracts, dry eye, facial asymmetry and retinal detachment.

Artificial eyes and vision:

• Some patients undergo enucleation. People in the UK can visit the NHS National Artificial Eye Service (NAES) or a private ocularist about their artificial eye. GPs can refer to these services.

• Socket problems can be encountered following enucleation.

• Fewer socket problems occur if the patient is under the care of a prosthetist.

If problems persist a referral to a local oculoplastic/orbital surgeon via the GP is important as reconstruction of the socket may be possible or necessary.

• Some patients will suffer severe vision loss as a result of their retinoblastoma. This may be in one or both eyes.

Other issues

CHECT can provide emotional, pratical and social support for any issues relating to the cancer experience, including treatment, late effects and living with an artificial eye.

Any medical issues related to Rb and follow-up can be directed to the specialist teams at the Royal London Hospital or Birmingham Children's Hospital.

Royal London Hospital retinoblastoma service - 020 3594 1419

Birmingham Children's Hospital retinoblastoma service - 0121 333 9475

For support and info you can contact the Childhood Eye Cancer Trust.

CHECT continues to support adults affected by their previous Rb in many ways, but can also connect you with other support organisations. Contact our support team to find out more on 020 7377 5578 or email support@chect.org.uk

www.chect.org.uk

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Fighting Retinoblastoma Childhood Eye Cancer Trust