

Supporting patients after retinoblastoma

Patients who have had retinoblastoma in their childhood require ongoing support throughout their lives. Juliette Carter explains the need for genetic counselling, the late effects of treatment and the increased risk of second primary cancers

Retinoblastoma is a rare eye cancer of early childhood. Tumours grow in the retina, the light sensitive lining of the eye. The cancer is often fast growing and in some cases very aggressive; retinoblastoma is very treatable and has one of the most successful survival rates of all the childhood cancers (Table 1). There are, however, lifelong implications for someone who has had retinoblastoma as a young child.

Early recognition of the signs and symptoms of retinoblastoma is important as speedy referral and treatment are necessary to ensure a good outcome (Carter, 2009). In this article, the additional support needs a survivor of retinoblastoma will have throughout his/her life will be highlighted, such as the need for genetic counselling, the late effects of treatment and the increased risks of second primary cancers. These are important as patients will often use primary care as their first port of call to access relevant services.

Incidence

Retinoblastoma may be unilateral or bilateral. There are two forms: genetic and heritable, present in those who carry in their germ cells the genetic mutation in the RB1 gene; and non-genetic/non-heritable. All bilateral disease is heritable, as are about 15% of unilateral cases, but most unilateral cases (85%) have no germ cell mutation. Those with the heritable condition have a 50% chance of passing the mutation to their future children. For some cases of heritable retinoblastoma there is a known family history of the disease but in most affected people the genetic change can start with them.

A personal experience

Stephanie, born in 1983, had retinoblastoma tumours present at birth, and has given permission for her experiences to be shared (Figure 1). Her mother had bilateral retinoblastoma so had been through genetic counselling and was aware of the risks to her future children. Consequently, Stephanie was

Table 1. The facts about retinoblastoma

- Retinoblastoma mostly affects children under the age of 5 years
- Tumours grow in the light sensitive lining of the eye
- It can be unilateral or bilateral
- There can be one or many tumours present
- It has one of the most successful treatment rates of all childhood cancers
- The major signs of retinoblastoma are leukocoria (a white pupillary reflex), a squint and sometimes a decrease in vision
- The main types of current treatment are chemotherapy or enucleation (surgical removal of an eye). Radiotherapy, cryotherapy and laser therapy are used as salvage treatments
- The incidence of retinoblastoma in Great Britain is around 1 in 20000 live births, which accounts for about 40 new cases diagnosed each year

From: Carter, 2009; MacCarthy et al, 2009a.

screened by examination under anaesthetic from birth. Stephanie only had tumours in her right eye initially and had an enucleation (surgical removal of the eye) at 5½ weeks old. At 4 months tumours developed in her left eye—these were treated with cryotherapy and laser but needed radiotherapy when Stephanie was 8 months old.

Between the 1960s and 1980s the mainstay of therapy for children with bilateral retinoblastoma was external beam radiation, often following enucleation of the eye in more advanced tumours. In the later 1980s chemotherapy was often given in addition to radiotherapy in an attempt to reduce the need for enucleation (MacCarthy et al, 2009b).

Due to the aggressive and sometimes unpredictable nature of retinoblastoma, Stephanie continued to be examined, first under anaesthetic until the age of 5 years, and then while awake until 10 years of age, like most children with retinoblastoma. As Stephanie was then free from the risk of developing further tumours she was discharged from the retinoblastoma service. Current practice is to discharge the patient at 16 years of age, with information about their condition, genetic status and treatment



Figure 1. Stephanie had retinoblastoma tumours present at birth

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details. They are also informed about the implications for later life and how to watch out for and manage these. Patients with heritable retinoblastoma also have an annual oncology appointment. Stephanie's generation and those older than her may not have been given any of this information and so are often unaware of the implications and possible risks that retinoblastoma can have into adulthood.

Genetics

It is possible to look at the retinoblastoma gene to identify the change that has caused retinoblastoma in an individual. It can take up to one year to carry out the testing, so for maximum benefit, patients should be referred well before they plan a pregnancy.

Everyone with bilateral retinoblastoma and around 15% of unilateral cases will have heritable retinoblastoma and so have up to a 50% chance of having a child with the condition. The remaining 85% of people with unilateral retinoblastoma will have a low chance of having a child with retinoblastoma. Genetic testing can be used in many families to clarify whether the affected person has the heritable or non-heritable form of retinoblastoma and this information can then be used to assess the risk of a new baby—those at risk will need regular examinations under anaesthetic, those not at risk can safely be excluded from these. However, genetic testing is not helpful in some families; knowing what genetic test to use and interpreting the results is complicated and should only be done by a specialist in genetics.

Screening of children usually starts when the offspring is just a few weeks old. The regular examinations under anaesthetic will last until the age of about 5 years old and will occur between every 2–6 months so that tumours can be identified early and treated promptly while still small. Genetic testing is of obvious benefit to the child as he/she will not need to undergo countless unnecessary anaesthetics. In addition, it costs less for all children to be genetically tested than to have them all screened by regular examination under anaesthetic methods (Richter et al, 2003).

Genetic testing may be used in pregnancy to determine whether a child is likely to be affected with retinoblastoma. This testing can only be done if the genetic change responsible for retinoblastoma in that family has been identified. Most couples choose not to have prenatal testing but it is a useful option for some couples who feel unable to cope with an

affected child, for example if they have a child undergoing treatment. It is also possible to make sure a child does not have the mutation on the RB1 gene; this process involves pre-implantation genetic diagnosis which is carried out during in vitro fertilization (IVF) and allows genetic testing on an embryo to take place before it is implanted in the womb. Pre-implantation genetic diagnosis is relatively new and has not been performed many times; though accurate, it has a relatively low rate of achieving a pregnancy.

Case study

To return to the personal account of retinoblastoma, Stephanie has recently got married and is thinking about starting a family sometime in the future. She says:

I would go to any length to ensure my child does not have retinoblastoma as it has made my life very challenging and I wouldn't want this for my child.

Through genetic counselling, Stephanie and her partner have been offered information about what genetic tests are available, their options of testing their future children and advising them of the benefits and consequences of carrying out these options so they can make informed decisions about their future. Stephanie had two options for genetic counselling: she could have been referred to her local genetics department or to one of the specialist retinoblastoma genetics services.

Consequences of treatment

As with all treatments a medical team will have to weigh up the benefits of giving a particular treatment with its possible side effects and consequences or late effects. Stephanie's enucleation and radiotherapy cured her retinoblastoma; however both have had an impact on her life. Stephanie says:

I need to put a lot of extra effort in to live my life.

Stephanie has lived with an artificial eye and was registered as partially sighted after her treatment. She went to a nursery and school for visually impaired children up to the age of 7 years, where she was happy. After she turned 7 years old, her parents decided it would be best for her to attend a mainstream school and 'integrate into society'. At the age of 10 years Stephanie had suffered a detached retina in her

KEY POINTS

- Retinoblastoma and its treatment have lifelong implications for patients
- Retinoblastoma patients should be offered genetic counselling and access to specialist services
- Retinoblastoma patients and survivors may have an increased risk of developing a second primary cancer
- Retinoblastoma patients may be unaware of the lifelong implications and the services available to them

Table 2. Risks of second primary cancers

Retinoblastoma patients with an increased risk of second primary cancers should avoid:	<ul style="list-style-type: none"> • Smoking • Ultraviolet (UV) light from the sun or sunbeds • Screening X-rays and computed tomography (CT) scans due to their association with extra radiation
Patients with an increased risk of second primary cancers should:	<ul style="list-style-type: none"> • Try to eat a healthy diet • Be aware that any unusual rashes, lumps or swellings and changes to moles should be taken seriously
What patients and professionals should look out for:	<ul style="list-style-type: none"> • Second tumours may present as lumps, pains, changes to moles or abnormal pigmentation of skin • Second tumours can occur anywhere in the body

remaining eye which she received treatment for; as a result of the detached retina she was registered as blind and to this day only has extremely limited vision. Stephanie remained in mainstream school until she was 16 years old but said:

I have bad memories of it, it's very hard when you're a teenager and just want to fit in, I had very thick glasses and was bullied. I came out with very high exam results but you are not interested in that as a teenager.

For A-Levels Stephanie decided to study at a beacon school for those with a visual impairment. This school fulfilled many of Stephanie's social and psychological needs as well as the academic needs and she had 'the best time of [her] life'. She went straight to university to study psychology but felt she was disadvantaged there because she could not read regular size print, and found university very challenging. Throughout Stephanie's life she has endured many complications as a result of her retinoblastoma and its treatment. She had cataracts from a young age, a detached retina, corneal problems and also 'dry eye' which is getting worse and makes her prone to recurrent infections. She struggled for several years after university especially in the world of work but she now feels she is headed in the right direction and is pursuing a career as a child psychotherapist.

Many people like Stephanie are living with the late effects of treatment—these range from problems like cataracts and dry eye to visual impairments. Other issues people deal with are living with an artificial eye or a disfigurement caused by the radiotherapy affecting bone growth.

Risks of second primary cancers

People with the heritable form of retinoblastoma have an increased risk of developing a

second primary cancer (Halford et al, 2008). Second tumours may develop anywhere in the body. Many affected people and their parents are unaware of these risks (Table 2).

Stephanie had a second primary cancer. Although her mother was aware of the risks and told her about them, Stephanie did not want to listen as a teenager. She did not protect herself in the sun, smoked and used sunbeds. At 21 years old she started to take notice and changed her lifestyle. At 22, she had a malignant melanoma in a mole on her back. This was removed and Stephanie considers herself lucky that it was not worse. There is no regular reliable screening for second tumours. Regular X-rays or computed tomography (CT) scans may be harmful. However, patients with a visual impairment should be offered yearly appointments with the GP or practice nurse for a complete body skin check, since they cannot see melanoma-like lesions themselves. This may only be necessary for those who live alone or have a partner with a visual impairment, but it is good practice to offer it to everyone.

Conclusions

Retinoblastoma is treatable, but it has many implications for survivors later in life. People who had retinoblastoma may or may not be aware of these issues. Often patients' first link into the services they need is through their general practice. They may well seek advice on issues of genetics, family planning, late effects of treatment and concerns over second primary cancers. However, they may not be aware that these services could be of use to them so they should be offered to everyone who has had retinoblastoma.

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