Having a baby after retinoblastoma

Planning for a family can pose additional challenges for people affected by retinoblastoma (Rb). We take a look at the considerations involved and the options available.

If you had Rb as a child, you may have an increased chance of having a child with Rb. The chances of a child being affected can vary from no greater than the general population to 50%. It is important to know whether you had the heritable form Rb. Genetic testing may also be helpful.

It is sensible for anyone who has had Rb to see a geneticist before they plan a family, to discuss what tests have been performed and to clarify what their chances of having a child with Rb will be. This also gives the chance to have tests performed before a pregnancy – the tests can take several months to get a result.

No right or wrong choice

Couples will then make their decision based on different factors such as the chances of having a child with Rb, the type of treatment and surveillance that is now available, their own experience of the condition and whether they have other children. These decisions are very personal and there is no right or wrong choice. In addition, a couple's choice may change with time or their circumstances. In general, the choices fall into three groups:

Deciding to have a child without any testing in pregnancy

All children of a parent who has had Rb should be regarded as being at risk of developing Rb and should have regular eye checks, unless genetic testing reduces the baby's chances of developing Rb to population levels.

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.

We suggest that all babies in this situation also have a provisional appointment made for an eye check, so that if there is a delay in getting the results, then there is no delay in checking the baby.

If the results show that the baby does not need eye checks then you can cancel that appointment. The appointment for the first eye check would be made as soon as possible after the birth and within seven days, depending on the family history.

Testing for Rb in pregnancy

Testing in a pregnancy for Rb can only be done if genetic testing has previously been carried out in the family, and the results mean that a test in pregnancy would be possible.

This is usually if a parent, or previous child, has the 'genetic' (or heritable) form of Rb. Tests in pregnancy have traditionally been done in two ways, either by chorionic villus

sampling (CVS) at 11 weeks of pregnancy or by amniocentesis at 16 weeks of pregnancy. Both tests carry a small risk of causing a miscarriage so it is generally recommended that these tests are only done if a couple feel sure that they would not want to continue a pregnancy if the baby would have a high risk of developing Rb.

From 2022, a new test termed Non-Invasive Prenatal Diagnosis (NIPD) has been developed and is available for some, though not all, families affected by the heritable (genetic) form of Rb. This test may be performed from around eight weeks of pregnancy and removes the risk of miscarriage. As such, eligible couples may consider this option not to guide the outcome of the pregnancy, but to plan the management from birth, should the baby be found to have a high risk of developing Rb. You can read more about NIPD here chect.org.uk/new-pregnancy-testing-technology-now-available/.

Anyone considering prenatal testing should be seen by a geneticist before they plan a pregnancy, to discuss the options available to them and allow sufficient time to complete the necessary background laboratory work.

Pre-implantation genetic testing for monogenic disorders (PGT-M)

This technique is available in only a few centres in the UK – and not all of those centres offer testing for Rb. If you would like to consider this option, ask your GP to refer you to your local service.

In brief, PGT-M is a combination of IVF (fertility) treatment and genetic testing. IVF is used to create embryos and these embryos undergo genetic testing at a very early stage. Unaffected embryos are then used to try to achieve a pregnancy.

PGT-M has obvious advantages over testing in pregnancy in that it does not involve a termination of the pregnancy. However, several months of background work are needed before the procedure can be done. It will not be possible for every couple and the success rate (ie babies born after the procedure) is around 30% for each attempt.

This is an option considered by some couples, who may have difficulties in getting pregnant or who feel that they do not want to have a child with Rb, but would not want to have a termination of pregnancy for the condition. It is a useful option but is not an easy process to go through.

It is expensive, costing around £10,000 to £15,000 for each cycle. Couples living in England who fit the criteria are offered up to three cycles of PGT-M (or until an unaffected baby is born) on the NHS. For people living in Scotland, Wales and Northern Ireland, funding needs to be applied for. Getting funding usually

takes several months, and is only offered to couples who do not have any unaffected children.

In summary, most people who have had Rb have an increased chance of having a child with Rb, compared to the general population.

There are different options available to ensure that a child is given the best treatment. Choosing the right option is a very personal decision. Anyone who has had Rb and who is thinking of having a baby should be seen by a geneticist to discuss the chances that a child would have Rb, arrange genetic testing and decide what options are best for them.

To find out more about seeing a geneticist and arranging genetic testing contact the CHECT support team on 020 7377 5578 or email support@chect.org.uk

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