

Information on

Preimplantation Genetic Diagnosis (PGD) and Cavernoma

This information is for people affected by cavernoma, particularly those who are concerned that they, and their family members, might be affected by the genetic form of cavernoma. It leads you to further information about PGD on other websites. The information is designed to be used alongside discussions you have with your medical providers.

CAUK's information booklet 'The Genetics of Brain Cavernoma' has general information about familial cavernoma and genetic testing that you may find useful.

What is PGD?

Preimplantation genetic diagnosis (PGD) is a technique for testing embryos created by in vitro fertilization (IVF) for specific genetic disorders before an embryo is transferred into a woman's womb. A few cells are removed from an early stage of the IVF embryo, and then tested for the genetic condition that affects the family. PGD gives people with a specific inherited condition a way to avoid having children with the condition by checking the specific gene change in the embryos before they become pregnant.

This area of assisted conception is regulated in the UK by the Human Fertilisation and Embryology Authority (HFEA). There are 3 genes associated with cavernoma, CCM1, CCM2, and CCM3. CCM1 and CCM3 are on the approved list of conditions licensed for PGD, but CCM2 is not. The reason for the difference has to do with the way that the HFEA approaches the licensing process. A fertility clinic that can carry out PGD must apply to the HFEA if they wish to test for a new (unlicensed) condition. An application is often initiated by a clinic when a couple affected by the condition are referred there, usually by the couple's medical geneticist or genetic counsellor.

Couples who wish to consider PGD are referred to a PGD centre usually by their clinical geneticist or genetic counsellor, who will ensure that all the required preliminary clinical tests have been completed and that one of the partners has had a positive gene test for the condition.

PGD is a complex process and can take time to prepare. It is likely to take a number of months to start a treatment cycle. There are specific criteria for eligibility, and in order to have the test it is necessary to know the genetic change causing familial cavernoma. It may also be necessary to have samples available from other affected family members. Your local clinical genetics service can discuss this with you and work out if you are eligible for treatment.

National NHS funding for PGD is now available in England to a couple provided they meet the acceptance criteria. Scotland, Northern Ireland and Wales each have their own funding systems. PGD centres will help those wishing to pursue PGD to find out if funding is available.

Where can I get more information about PGD?

Information for Patients on PGD appears on the Genetic Alliance website. Further detailed patient information appears on the HFEA, and at Guy's and St Thomas' hospital PGD unit websites. Go to:

<http://www.geneticalliance.org.uk/aboutpgd.htm>

<https://www.hfea.gov.uk/treatments/embryo-testing-and-treatments-for-disease/pre-implantation-genetic-diagnosis-pgd/>

<http://www.pgd.org.uk/home.aspx> (Guy's and St Thomas')

The known treatment risks of PGD are similar to those for IVF. The incidence of abnormality in babies conceived through PGD is similar to that seen in other forms of assisted reproduction, and there are no known major abnormalities associated with PGD.

Success rates of having a baby with PGD are difficult to generalize, but information can be found on the Human Fertilisation & Embryology Authority (HFEA) website. Success rates depend on many factors including reason for PGD, age of female partner and health of couple. It is estimated that chances of having a baby with PGD are about the same as for IVF. If PGD results in a pregnancy, the patient is offered a prenatal genetic test to confirm the results of the embryo testing. This is because PGD is never 100% accurate, although for most people testing is 98-99% accurate, and dependent on the condition and the individual test results. Couples can make their own decision about whether or not they wish to have a prenatal test.

What are the specific issues around cavernoma and PGD testing?

Everyone has two sets of genes, one from their mother and one from their father. Someone who has the genetic form of cavernoma has a change in one copy of the gene associated with cavernoma. If one of the parents has a CCM gene, each child they conceive has a 50% chance of inheriting the faulty CCM gene. This is an example of an autosomal dominant pattern of inheritance.

Someone who has a CCM gene but who does not have a detectable cavernoma, or does not experience symptoms, has the same chance of passing on the CCM gene as someone with a CCM gene who is symptomatic.

If a PGD test for a CCM gene gives a positive result, it cannot predict if a child born from the embryo will develop a cavernoma, or how severely the child will be affected if cavernomas develop. This may not be a central issue in PGD, as the purpose of PGD is to select embryos without the gene mutation.

When someone has multiple cavernomas, it is more likely that they will have the genetic form of cavernoma. However, if no genetic change is identified, then it will not be possible to offer PGD.

The risks of developing symptoms from cavernoma are higher for people with multiple cavernomas, and the familial form of cavernoma often leads to multiple cavernomas.

Individuals and families will have their own experiences to bring to discussions and decisions about genetic testing and having children. Clinical geneticists and genetic counsellors assess each case individually, and will advise their patients individually. Testing and decision-making is personal, and often shared and discussed with others.

Prenatal testing (testing in pregnancy) for cavernoma is also possible and available under the NHS. For information see CAUK's article Prenatal Testing and Cavernoma.

CAUK's information and booklets and articles are also available by post.
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Last Updated 11 June 2017.