

Information on Prenatal Testing 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.

This page gives you information about **prenatal testing** and some specific issues regarding prenatal testing and cavernoma. It leads you to more detailed information 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 Prenatal Testing?

Prenatal testing involves testing a fetus during pregnancy, usually for a disorder or inherited condition.

Prenatal testing is available under the NHS for the genes associated with cavernoma, when the gene mutation has been identified in one of the parents, and the risk to the child of inheriting the gene has been established.

Amniocentesis and Chorionic Villus Sampling (**CVS**) are the diagnostic techniques that can tell you if the baby has inherited a genetic (or chromosomal) condition.

Amniocentesis involves taking out a small amount of amniotic fluid that surrounds the fetus in the womb. This is done by passing a very fine needle through the pregnant woman's skin and abdomen (tummy) to withdraw the fluid. This fluid contains some of the baby's skin cells. The cells can be tested to check the baby's genes or chromosomes. Amniocentesis is usually carried out after 15 weeks in the pregnancy.

Chorionic villi are part of the developing placenta (the organ that forms during pregnancy to join the mother and baby, which is expelled as afterbirth following delivery). **CVS** involves taking a small sample of chorionic villi tissue to test during pregnancy. In the most commonly used method, a fine needle is inserted through the pregnant woman's skin and abdomen (tummy), through the wall of the womb, and into the placenta, to withdraw a small sample of tissue. An ultrasound scan guides the healthcare worker or doctor to the correct location during the procedure. CVS can be done earlier than amniocentesis, usually from 11 to 14 weeks.

What are some of the specific issues around cavernoma and prenatal testing?

Prenatal testing is available on the NHS for the three genes known to be associated with cavernoma - CCM1, CCM2, and CCM3 - when the mutation is present in one of the parents. Everyone has two sets of genes, one from their

mother and one from their father. 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.

If you are thinking about the possibility of prenatal testing, it is important to talk to your doctor or specialist, such as your genetic counsellor or medical geneticist, early on so that any necessary preliminary diagnostic tests can be done early in your pregnancy or before you get pregnant.

If a prenatal test for a CCM gene gives a positive result, it is difficult to interpret what it would mean in the long term for each individual baby once born. The test cannot predict if the child will develop a cavernoma, or how severely the child will be affected if cavernomas develop.

Amniocentesis and CVS carry a small risk of miscarriage, and this can make the decision to have a prenatal test more difficult. The risk of miscarriage is not specific to cavernoma, but a general risk that accompanies the procedures of amniocentesis and CVS. This will need to be considered when thinking about the range of benefits and risks of prenatal testing.

The decision whether to have a prenatal test is personal and individual, but you may want to discuss with your genetic counsellor whether it is right for you.

Many women and couples have prenatal testing because they wish to consider terminating the pregnancy in the event of a positive test for the condition. It is important to understand that if you are given the news that the fetus has inherited a genetic anomaly, continuation of an affected pregnancy is an option. It is your decision about how to proceed with a pregnancy after a positive gene test for familial cavernoma. Whatever path you decide, you may have a mixture of emotions.

Where can I get more information and support about amniocentesis, CVS, and prenatal testing?

ARC, Antenatal Results and Choices, offers further information and support if you wish to talk to someone at any stage:

<http://www.arc-uk.org/tests-explained/diagnostic-tests>

NHS Choices offers information about these tests:

<http://www.nhs.uk/Conditions/Amniocentesis/Pages/Introduction.aspx>

<http://www.nhs.uk/conditions/chorionic-villus-sampling/pages/introduction.aspx>

Preimplantation genetic diagnosis (PGD), testing embryos created by in vitro fertilization, is an option for some families concerned about the risks to

their children of inheriting symptomatic cavernoma. For information see CAUK's article on Preimplantation Genetic Diagnosis (PGD) and Cavernoma.

CAUK's information and booklets and articles are also available by post.
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