Reproductive Options for Cancer Susceptibility Gene Variants (CSGV)

Introduction

When you or your partner, or both, carry a cancer susceptibility gene variant (CSGV), there are several reproductive options available to you. Most of these require that the condition has already been tested for in you and a molecular cause has been identified. To discuss these options in more detail, contact your local Clinical Genetics department for an appointment.

No testing

You may decide not to have any testing in pregnancy and wait for the baby to be born. For conditions that have management implications in childhood, a blood test (cord/venous) can be done after the birth of your baby to see if he/she is affected. For adult-onset conditions, your doctors will recommend that your child waits until early adulthood so they can have the autonomy to decide for themselves if they want to be tested for the CSGV.

Prenatal Diagnosis : CVS and Amniocentesis

If you want to know if your baby is affected with a genetic condition, the most common way is to test the pregnancy. There is a choice of 2 prenatal tests that are available:

Chorionic villus sampling (CVS): This can be done between 11+ and 14 weeks, where a small sample is taken from the edge of the placenta using a needle under ultrasound guidance.

Amniocentesis: This can be done from 15-16 weeks (ideally before 21 weeks), where a small sample of fluid is taken from the amniotic sac also using a needle under ultrasound guidance.

There is a small risk of miscarriage (around 1% for CVS and 0.5% for amniocentesis) related to the procedure.

Pre-Implantation Genetic Testing (PGT-M)

This involves a couple going through IVF treatment to create embryos which are then tested for the genetic condition. Only unaffected embryos are then implanted. For couples who are opposed to termination of pregnancy, for either personal, ethical or religious reasons, PGT-M offers an alternative to prenatal testing.
However, this needs to be balanced against the complexity of the process, including timescales for treatment and likely success rate. NHS funding is available to couples who meet the relevant criteria.

For more information, see: https://www.hfea.gov.uk/treatments/embryo-testing-and-treatments-for-disease/pre-implantation-genetic-testing-for-monogenic-disorders-pgt-m-and-pre-implantation-genetic-testing-for-chromosomal-structural-rearrangements-pgt-sr/

**Non-Invasive tests**

Some CSGVs have a gender bias and therefore couples may find non-invasive fetal sexing useful if they would consider termination of a particular sex of baby, given a higher incidence of CSGV in this group (e.g., females for BRCA1). This involves taking a sample of blood from the mother, from around 9 weeks of pregnancy. Cell-free fetal DNA (cffDNA) can be isolated from the maternal blood stream and tested for the sex of the fetus. This test is NHS-funded for conditions where there is a discrepancy in severity between males and females.

Bespoke NIPD is also possible for CSGV where the male partner carries the condition, or for recessive disorders. This is not currently NHS-funded.

**Sperm or Egg Donation**

Dependent on the inheritance pattern of the CSGV, sperm donation (if the male partner carries the CSGV) or egg donation (if the female partner carries the CSGV) may be an option which would eliminate the risk of the condition being passed on to offspring. Even though a couple may be eligible for NHS treatment, the cost of gamete donation and storage is often not covered by the NHS. For more information, see the Donor Conception Network: www.donor-conception-network.org

**Adoption**

Adoption is an option for couples or single people who are resident in the UK. There is a rigorous application process with medical and social checks to ensure a safe and happy home for a child. To find out more, see: https://www.gov.uk/child-adoption

**Childless Relationship**

Having children is a personal choice and many couples will choose not to have children and have a happy and fulfilling life.

**If you need more information please contact your local genetics department.**

For further support or information about reproductive choices for CSGVs, please visit:

**The Human Fertilisation and Embryology Authority (HFEA)**

The UK independent regulator overseeing the use of gametes and embryos in fertility treatment and research.

Website: www.hfea.gov.uk

**Genetic Alliance UK**

An organisation that works to support people with genetic disorders

Website: www.geneticalliance.org.uk

Telephone: 020 7704 3141

**ARC Antenatal Results and Choices**

A charitable organisation supporting families through antenatal screening and testing.

Website: www.arc-uk.org

Telephone: 0207 713 7486