Fertility Information

Implications for future pregnancies

If a woman is known to be a carrier of Barth syndrome, for any future pregnancy there is a 25% chance of having a boy who is affected by Barth syndrome. This means that in any future pregnancy there is a 75% chance that the baby will **not** have Barth syndrome.

If a carrier female knows that the baby she is carrying is a boy, then the chance that the boy has Barth syndrome is 1 in 2 or 50%. If the baby is a girl, then there is a 1 in 2 or 50% chance that the baby will be a carrier of Barth syndrome but will not have any symptoms of the condition.

Reproductive Options

If a woman has had genetic testing which shows that she is a carrier of Barth syndrome, then there are a number of different options a couple may consider if they wish to have a baby. To fully explore these options, it is recommended that you see or speak to a Genetic Counsellor. Here is a brief summary of the options.

- **The option of conceiving a pregnancy naturally and opting not to have any testing in the pregnancy,** accepting that there would be a 25% chance of an affected boy. Your local foetal medicine unit can be asked to perform additional scanning in a male pregnancy to look for any signs of dilated cardiomyopathy or foetal hydrops (where the foetus develops abnormal fluid collection).

- **Foetal sexing on a blood sample from mum from 7-10 weeks gestation.** Using a blood sample from mum it is possible to look for the presence of Y chromosome material to determine whether or not the pregnancy is male or female. This testing is known as free foetal DNA analysis (ffDNA). The accuracy of the testing should be discussed with the person arranging the test and it is often recommended that the result is confirmed by ultrasound scanning from 16 weeks gestation. If the pregnancy is female, no further testing would be undertaken.

  If the pregnancy is male, then testing for Barth syndrome can be undertaken through the following:

- **Prenatal Testing:** Testing for Barth syndrome in a pregnancy can be offered. There are two main types of tests available. These would be performed in a Specialist Foetal Medicine Unit.

  CVS test (Chorionic Villus Sample) from 11 weeks gestation. CVS testing involves using the guidance of an ultrasound scan to insert a needle through the abdomen to take a sample of the placenta (afterbirth). The placental tissue can be tested to determine whether or not the foetus has inherited the *TAZ* gene change causing Barth syndrome.

  Amniocentesis test from 15-16 weeks gestation. This testing involves using the guidance of an ultrasound scan to insert a needle through the abdomen to take a sample of the amniotic fluid that surrounds the foetus. Cells in the sample can be tested to determine whether or not the foetus has inherited the *TAZ* gene change causing Barth syndrome.

Both CVS and amniocentesis carry a small increased risk of miscarriage in the region of 1 to 2% (1/100 to 1/50) above the usual background risk of miscarriage. In view of this it is important for couples to carefully consider whether they would take any different decisions about the pregnancy if the foetus has inherited Barth syndrome. Of the two tests (CVS and amniocentesis) it is more common for individuals to request a CVS test as it can usually be performed earlier in the pregnancy than an amniocentesis.

PGD or Pre-implantation Genetic Diagnosis for Barth syndrome. This involves a couple going through a process of IVF (In-Vitro Fertilisation) treatment where eggs and sperm are mixed in a test tube. A single cell from the resulting embryos can be tested for Barth syndrome before being re-implanted into womb. Only unaffected embryos would be used. It is important to realise that this process can be very emotionally challenging as there are a number of stages at which the treatment may fail. It is possible to apply for National NHS funding for PGD and if you meet certain criteria the PGD Centres can help with this process.

- **Conceive a pregnancy using a donor egg or embryo** to avoid the risk of passing on the *TAZ* gene change causing Barth syndrome. This would require referral to a Reproductive Medicine clinic for specialist advice.

- **Surrogacy.** This option may be explored if a couple are experiencing fertility issues or if there is a medical reason why it would be necessary to avoid pregnancy for the female. Further information is available at: www.HFEA.gov.uk

- **Some couples may wish to consider the option of adopting a child.** Further information is available at: www.gov.uk/child-adoption

A decision as to how best to proceed is very much a personal decision for each individual couple. The National Barth Syndrome Service has designated Genetic Counsellors who would be able to talk to you further about all the options that would be available to you in more depth. If you would like to arrange to speak to a Genetic Counsellor please visit the Barth Service Website.