

Preimplantation Genetic Diagnosis Sickle cell or SC disease (2) Supplementary leaflet

This leaflet has been created as an additional source of information, to be read in conjunction with the **Preimplantation Genetic Diagnosis Booklet**. The details within the following pages are specific to you and the reason why you have asked about PGD treatment.

As before, there will be plenty of time to discuss further aspects of treatment during your consultation, but if anything is unclear in the leaflet, please let us know. Our contact details can be found on page 33 of the main booklet.

The **Preimplantation Genetic Diagnosis Booklet** explains what happens up to the stage where a cell is removed from each embryo. This leaflet explains the testing that is done to determine which embryos have the genes that cause sickle cell disease in couples where one partner is affected with sickle or SC disease and the other partner is a carrier.

Testing for Sickle cell disease in PGD

You may remember that Sickle cell disease is caused by an alteration (known as a mutation) in the haemoglobin gene (Hb) on chromosome 11.

A normal copy of the gene is given the letter **A**

A changed copy of the gene is given the letter **S or C**

We all carry two copies of the haemoglobin gene because our genes come in pairs. Therefore as individuals we may have one of the following combinations of haemoglobin genes:

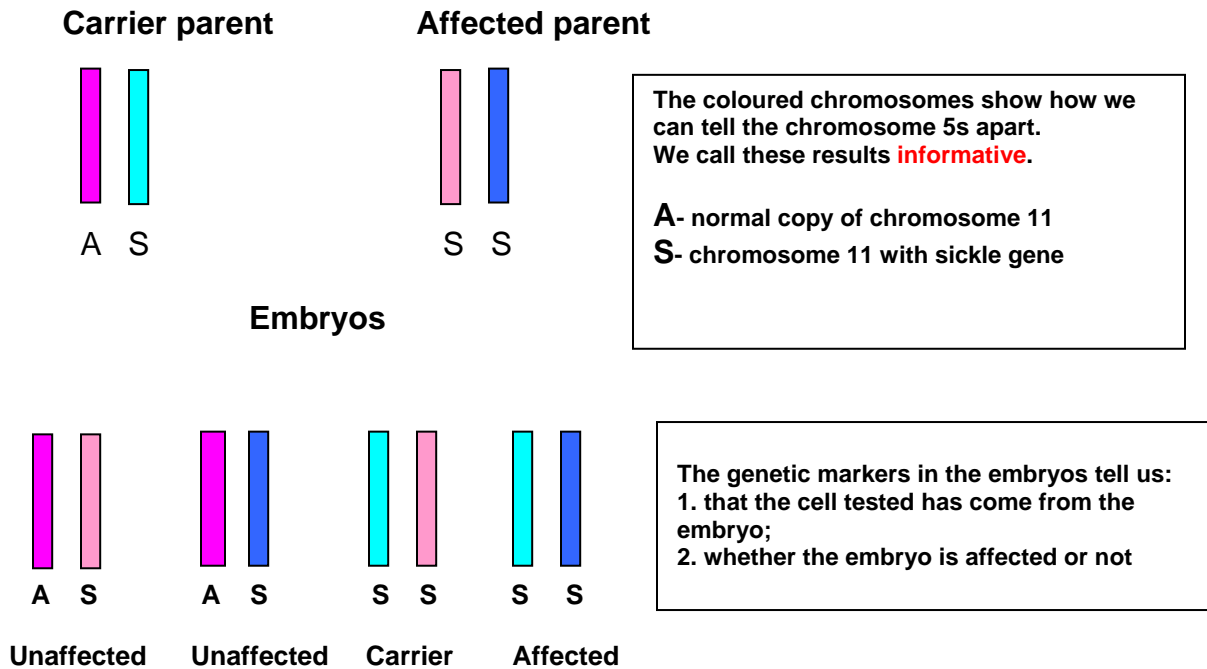
- Those who do not have Sickle cell disease have two normal copies of the gene and are known as being **HbAA**.
- Those who are affected with Sickle cell disease have two changed copies of the gene and are known as **HbSS or HbSC**.
- Those who are carriers of Sickle cell disease have one normal copy and one changed copy of the gene. They are known as **HbAS or HbAC**.

Linkage analysis

The test we use is called linkage analysis. It uses the process of DNA fingerprinting and compares the genetic markers in your DNA samples with genetic markers in the embryos' DNA. To do this part of the test, we will need to look at blood samples from you **and other members of your family**. This gives us two pieces of information:

- It tells us that the cell being tested is definitely a cell from your embryo and not from another source;
- It tells us which embryos are affected and unaffected by sickle cell disease.

Diagram showing how linkage analysis works



Important information

- It is not always possible to use linkage analysis. Sometimes we are not able to distinguish between the genetic markers carried by a couple because all the markers are the same.
- Only embryos that are clearly unaffected, will be considered suitable for use. This means that we will only use carrier embryos for transfer.

Accuracy of the test

We take great care to ensure that the diagnosis is as accurate as possible. There is a chance however, that the result in the embryo analysed could be incorrect. Fortunately the chance of this happening is relatively small and is likely to be less than 1% (1 chance in 100).

Confirmation of diagnosis

As PGD is not 100% accurate, we offer a prenatal test (test in pregnancy) to women who become pregnant following treatment. This test will confirm the diagnosis. It may be a CVS (chorionic villus sampling) done at 11 weeks of pregnancy or an amniocentesis test done at 16 weeks.

We appreciate that after going through a procedure such as PGD this can be a difficult decision to make. If you decide against confirmatory prenatal testing then we could arrange for a blood sample to be taken from the baby's umbilical cord at birth. The blood sample will be sent to our laboratory and confirmation of the PGD should be available within a week. We will make arrangements to contact you with this result.

Limitations of testing

Testing the embryos is limited to offering a test for sickle cell disease. It is not possible to carry out any other tests on the single cells at the same time, for example Down syndrome. The chances of any other problems affecting your embryos are the same as for any other couple in the general population. The incidence of Down syndrome does increase with a woman's age. You may wish to have a prenatal test for this if you do become pregnant.

Welfare of the child

Our Centre is licensed by the Human Fertilisation and Embryology Authority (HFEA). They ensure that we carry out PGD in accordance with the Human Fertilisation and Embryology Act and the HFEA Code of Practice. As part of the Act we are required to consider the welfare of any child who may be born when offering general fertility treatment or PGD to a couple. This means that you need to consider what the effects will be on you and your child if you become unwell with sickle cell disease.

We appreciate that you may well have considered this already, but you may need to think about how this may affect the care of your child. You may wish to make provision for this and some people find it helpful for plans to be put in place for a child in the future. To help with this further, we will also discuss this issue with you in more detail, usually during your second PGD appointment. We have fertility counsellors who are available should you wish to speak to them about any issue arising from PGD. This is available for you as a couple or for each of you individually.

There will plenty of time to discuss the issues above and those in the **Preimplantation Genetic Diagnosis** booklet when you attend the clinic. In the meantime, if you have other questions please ring us on the contact numbers given in the main booklet.

Other useful contacts

Sickle Cell Society

54 Station Road

London

NW10 4UA

Tel: 020 8961 7795

E-mail: info@sicklecellsociety.org Website: www.sicklecellsociety.org

Sickle and Thalassaemia Association of Counsellors (STAC)

South East London Sickle Cell and Thalassaemia Centre

Wooden Spoon House

5 Dugard Way

Kennington

London

SE11 4TH

Tel: 020 7414 1363 E-mail: info@stac.org Website: www.stacuk.org

NHS Sickle Cell and Thalassaemia Screening Programme

Website: www.kcl-phs.org.uk/haemscreening

Glossary

Amniocentesis: Test done during pregnancy. A fine needle removes fluid from the amniotic sac (water that surrounds the fetus in the uterus) at about 16 weeks of pregnancy. This test is usually performed to check for abnormalities in the fetus.

Chorionic villus sampling (CVS): Test done during pregnancy. A fine needle removes some tissue from the placenta (afterbirth) at about 11 weeks of pregnancy. This test is usually performed to check for abnormalities in the fetus.

We have leaflets on amniocentesis and CVS. If you would like copies, please ask a member of staff.

Factual information presented within this communication is based on accurate contemporaneous peer reviewed literature. Evidence of sources can be provided on request.