

Preimplantation Genetic Diagnosis Embryo sexing for X Linked conditions 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 the sex of the embryos.

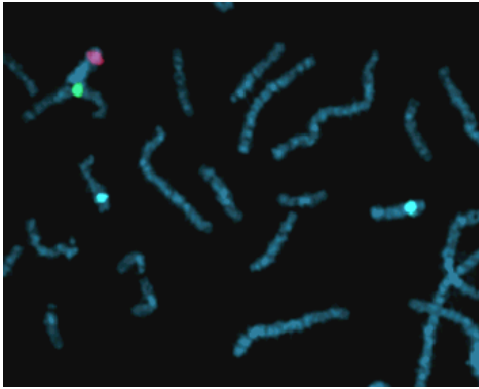
Testing of embryos

You may remember that we all have a set of 46 chromosomes in nearly every cell in our body. The chromosomes are inherited in pairs, one of each pair from our mother, the other from our father. One pair of the chromosomes is known as the sex chromosomes and it is these that determine our sex as individuals: males have one X chromosome and one Y chromosome, whereas females have two X chromosomes.

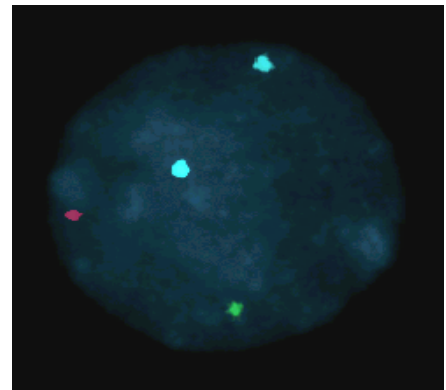
Testing of the single cells to determine the sex of the embryos uses a technique called *fluorescence in situ hybridisation* (**FISH**). FISH technology uses small pieces of artificially made genetic material (DNA) normally found on the X and Y chromosomes. These are called probes and are similar to the X and Y chromosome material. They contain a fluorescent colour dye which enables us to see them under a microscope. When added to the cell taken from each embryo after biopsy, the probes will seek out the corresponding bit of DNA in the sex chromosomes in the embryos and stick (hybridise) to them, and we can distinguish between them by their colours. We use green for X and red for Y. We use another probe (blue) as a way of ensuring that at least one other pair of chromosomes is present in the cell.

The following pictures show how these probes work.

FISH probes highlighted on full chromosome picture- male



FISH probes highlighted in single cell from embryo- male

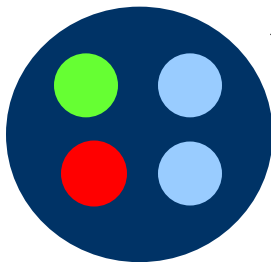


Red probe detecting Y

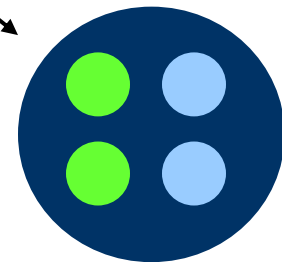
Green probe detecting X

Blue probes acts as a control

MALE- XY



FEMALE - XX



The gene responsible for causing the X linked condition in your family lies on one of the X chromosomes. Women who carry an X linked condition have one X chromosome carrying the abnormal gene and one X chromosome carrying the normal gene. Boys will inherit either the abnormal gene and be affected by the condition or the normal gene and have no related health problems. Girls will inherit either the X chromosome with the abnormal gene and be an unaffected carrier or the normal gene on the other X chromosome and will therefore not be a carrier.

Using this test, it is not possible for us to tell the difference between female embryos that are carriers of the X linked condition and those that are not.

Outcome of embryo testing

Following the testing on embryos from any cycle of PGD we may have a combination of the following results:

- An embryo that is male and at 50% risk of the X linked condition
- An embryo that is female and unaffected by the condition
- The test has failed to give a clear result in the embryo.

The only embryos that will be considered as suitable for use in treatment will be those that are **clearly female**.

Accuracy of the test

Whilst the greatest care is taken to ensure that the diagnosis is as accurate as possible, there is a chance that it could be incorrect. Fortunately the chance of this happening is relatively small. The risk of misdiagnosing a male embryo as female is less than 1% (1 in 100) per embryo.

Confirmation of diagnosis

As PGD is not 100% accurate, we offer couples who become pregnant following treatment, a test to confirm the sex of the baby in an ongoing pregnancy. Most couples opt for a scan to determine the sex of the baby which is reliable after 16 weeks and carries no associated risk of miscarriage. However, if a couple wish to have this information earlier or have other concerns about their pregnancy (see below) then a CVS (chorionic villus sampling) test (carried out at 11 weeks) or an amniocentesis (carried out at 16 weeks) would confirm the sex of the baby in an ongoing pregnancy.

Limitations of testing

Testing the embryos is limited to offering a test to determine the sex of the embryos in X linked conditions. It is not possible to undertake any other testing on the single cells simultaneously, e.g. Down syndrome. The chances of any other problems affecting your embryos would be the same as for other couples in the general population. The incidence of Down syndrome does increase with a woman's age and this may be something for which you may want to have a prenatal test, if you were to become pregnant.

Glossary

Amniocentesis: Test done during pregnancy. A fine needle removes fluid from the amniotic sac 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. 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.

HFEA licence

We have already been granted licences by the HFEA to carry out PGD for many X linked conditions. If the X linked condition affecting your family is new to us, we must first check with the HFEA to find out whether another UK PGD centre already has a licence. If they do then this means that we do not have to apply for a new licence. We must inform the HFEA, but it is a relatively simple procedure.

If no other centres have a relevant licence, then we will need to make a full application to the HFEA. This can take several months.

Licences have been given to us by the HFEA in the past on the basis that we will use PGD to establish the sex of the embryos and use only female embryos for transfer. If you think that a situation could arise whereby you may decide to have male embryos transferred (e.g. the only embryos available for transfer are male), then you must discuss this with us during your consultation.

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

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Guys & St. Thomas NHS Foundation Trust