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 carry an unbalanced translocation.

**Testing for a reciprocal translocation**

As you may already know, a reciprocal translocation is caused by segments from two different chromosomes swapping places.

This shows two normal chromosomes: A and B

This shows how segments of the two chromosomes have swapped places to form translocated chromosomes
In a PGD cycle, these translocations are tested for by using a technique called *fluorescence in situ hybridisation (FISH)*. FISH technology uses small pieces of artificially made genetic material (DNA). These are called probes and are made of similar DNA to the chromosomes being investigated. They contain a fluorescent colour dye which enables us to see them under a microscope.

When added to the embryo cell after biopsy, the probes will seek out the two chromosomes being tested and hybridise (stick) to them. We can distinguish between them by their colours. We usually use green for one of the chromosomes and red for the other. We use another probe (blue) as a way of ensuring that there is further proof that the chromosomes in the cell do not show an unbalanced translocation. This means we can be sure that the embryo has developed as we would expect it to do at this early stage.

**How do these probes work?**

Prior to the start of a PGD treatment cycle the probes need to be tested out in blood samples from both partners. This ensures that PGD can be carried out accurately. This procedure may take around 3 months to make sure that the test we offer you is as accurate as possible.

When the cells are examined under a microscope, we would expect to see two spots of each colour (red and green). If not, this indicates an abnormality.

The following picture shows what the FISH probes would look like if we looked at all the chromosomes in one cell from a translocation carrier:

**Diagram showing how FISH works in a cell from a translocation carrier**

![Diagram showing how FISH works in a cell from a translocation carrier](image)

Although in the above picture we can see all the chromosomes in a cell, it takes several days to get this result. The results on the embryos must be available within 24 hours of embryo biopsy and looking at all the chromosomes in the cell takes too long. So when we test cells from the embryos, we can only look at the chromosomes involved in the translocation and will obtain the results below.
Outcome of embryo testing

Following any cycle of PGD it is possible to obtain a combination of any of the following results in the embryos:

**a) Normal embryo**- The following picture shows how a normal cell would appear under a microscope. This embryo is unaffected. This embryo will be either a balanced translocation carrier or have normal chromosomes.

![Single cell from biopsied embryo]

- 2 signals from chromosome A (red)
- 2 signals from chromosome B (green)
- 2 signals from the control (blue)

**b) Abnormal embryos**- The following pictures show examples of cells from embryos with an unbalanced chromosome complement. This embryo is affected and will have an unbalanced translocation.

The following diagrams show examples of cells from an embryo with an unbalanced chromosome complement.

- 3 signals from chromosome A (red)
- 1 signal from chromosome B (green)
- 2 signals from the control (blue)

- 1 signal from chromosome A (red)
- 3 signals from chromosome B (green)
- 2 signals from the control (blue)

**c) 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 unaffected**.
It is not possible to distinguish between chromosomally normal embryos and those with a balanced translocation. So the embryos recommended for transfer may be carriers of balanced translocations. This will be discussed with you when you come to see us in the clinic.

### 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 the result in the embryo analysed, could be incorrect. Fortunately the chance of this happening is relatively small and differs according to the translocation involved.

The actual risks related to your translocation will be discussed in detail with you at a later stage when we have the results of your blood tests.

### Confirmation of diagnosis

As PGD is not 100% reliable, we offer couples that become pregnant following treatment a prenatal test (test in pregnancy) to confirm the diagnosis. This may be a CVS (chorionic villus sampling) done at 11 weeks of pregnancy or an amniocentesis done at 16 weeks. Some couples may opt for detailed ultrasound scanning instead. Abnormalities in pregnancies affected by unbalanced chromosomes may sometimes be detected by scan.

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. Arrangements will be made to contact you with this result.

### Limitations of testing

Testing the embryos is limited to offering a test for the chromosomes involved in the translocation. It is not possible to undertake any other testing on the single cells simultaneously, e.g. Down syndrome (unless chromosome 21 is one of the chromosomes involved in your translocation). The chances of any other problems affecting your embryos would be the same as for any other couple 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.

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

**Other useful contacts**

**UNIQUE**  
PO Box 2189  
Caterham  
Surrey  
CR3 5GN  
Tel/ Fax: 01883 330766  
E-mail: info@rarechromo.org  
Website: www.rarechromo.org

**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.*

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

Guys & St Thomas NHS Foundation Trust  
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