

Preimplantation Genetic Diagnosis Prader Willi syndrome 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 are affected by Prader Willi syndrome (PWS).

Testing for PWS

There are two steps to obtaining the genetic material (DNA) needed for the test.

1. The DNA is extracted from each single embryo cell and copied a million times (this is called whole genome amplification). This gives us a large sample of DNA to work on.
2. Then the crucial piece of DNA which is near to the PWS deletion is rapidly copied many times again. This process is called PCR (polymerase chain reaction).

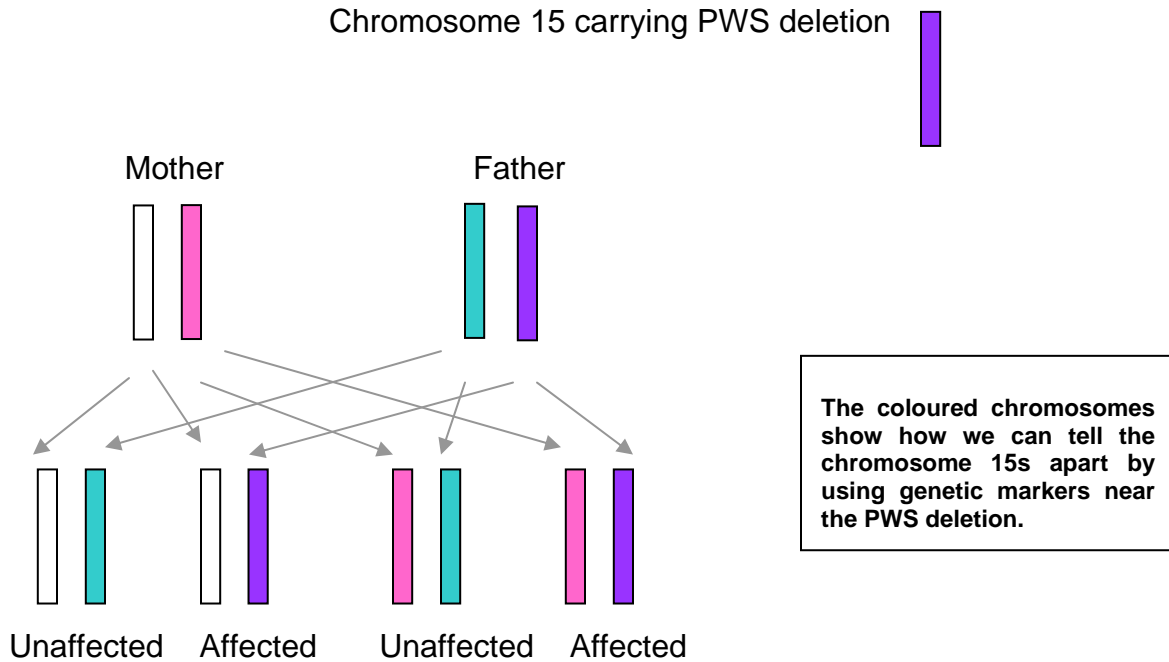
Linkage analysis

The test used for PGD of PWS involves linkage analysis. This is similar to DNA fingerprinting and compares the genetic markers on chromosome 15s with genetic markers on the embryos' chromosome 15s (see diagram below). This tells us 2 pieces of information:

- a. That the cell being tested is definitely a cell from your embryo and not from another source.
- b. That the DNA from both copies of chromosome 15 has shown up on the analysis and that the embryo has inherited one copy from each parent.

In order to do the linkage analysis we will require a blood sample from each of you.

Diagram to show how linkage analysis works in PGD



Outcome of testing

Results in your embryos

It is likely that the results we obtain will be a combination of the following:

- An embryo has two copies of the normal chromosome 15 markers and is unaffected.
- An embryo has one copy of the normal chromosome 15 marker and one copy of the marker near the deletion and is affected.
- The test has failed to produce a result in the embryo. The only embryos that will be considered as suitable for use will be those that are clearly unaffected.

So how accurate is 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 chances of this happening are relatively small, around 1 in 100 (1%).

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.

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 PWS. 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 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

Prader Willi Syndrome Association
125A, London Road,
Derby,
DE1 2QQ

Tel: 01332 365676
www.pwsa.co.uk

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.