



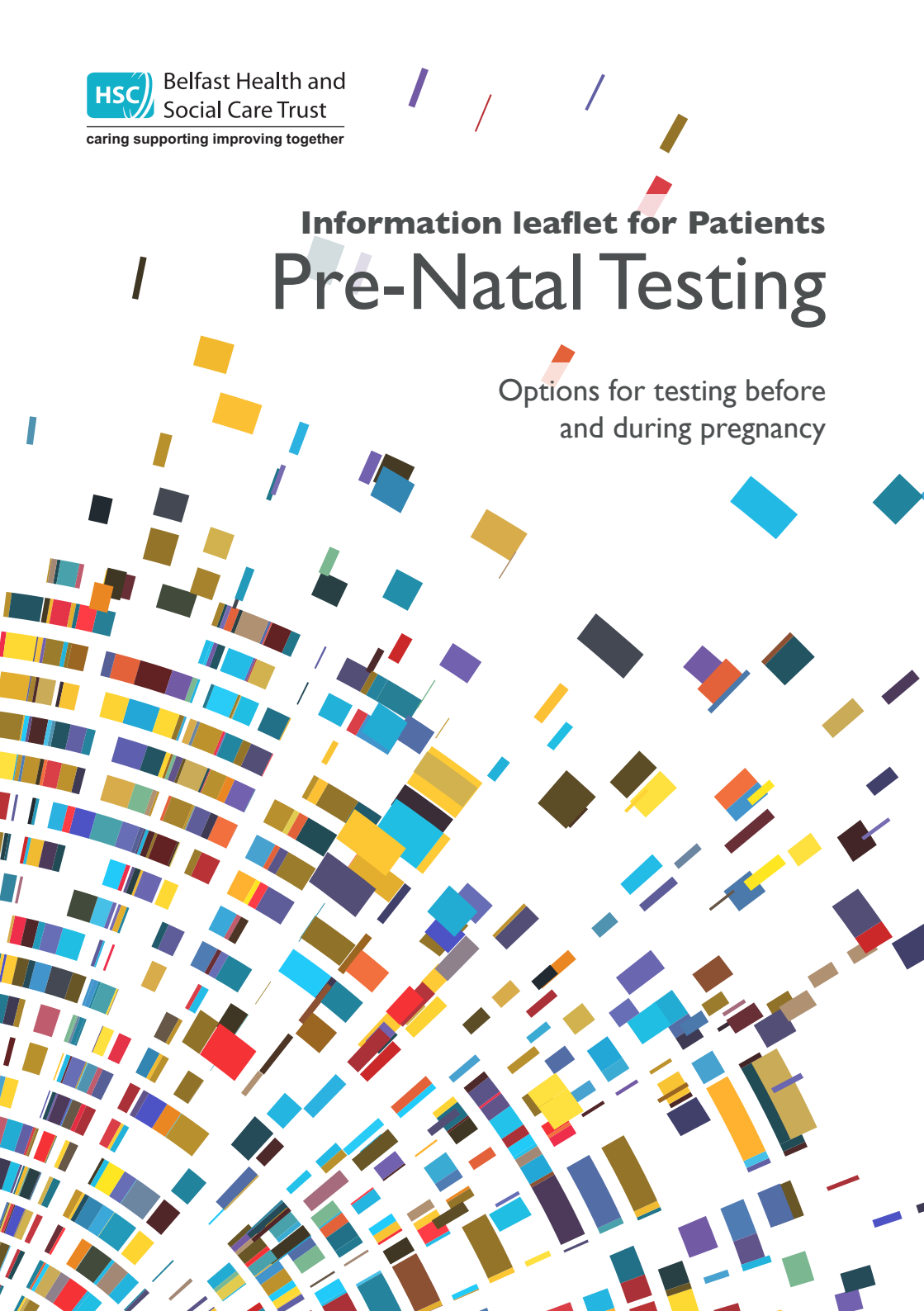
Belfast Health and  
Social Care Trust

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# Information leaflet for Patients

# Pre-Natal Testing

Options for testing before  
and during pregnancy





## Introduction

In this leaflet, you will find information about several different options which may be available to you either before or during pregnancy. Your genetic counsellor/consultant may have given you this leaflet if there is a genetic condition which has been identified in your family. It may help you to think about and decide which course of action would be best for you. Further more detailed discussions about these options can be arranged with your genetic counsellor or fetal medicine team as appropriate.

The leaflet will provide you with an overview of the following options;

- Non Invasive Prenatal Diagnosis (NIPD)
- Chorionic Villus Sampling (CVS)
- Amniocentesis
- Pre-implantation Genetic Diagnosis (PGD)

## Key Points

The table below outlines the key information for each of the tests described in this leaflet. Please read each of the relevant sections in this leaflet for a more detailed description of the available tests. Not all of the tests will be appropriate for you. It can depend on which genetic condition is identified in your family.

## **Non-Invasive Pre-natal diagnosis**

- Non-Invasive test
- Available during early pregnancy
- Involves a blood sample from the mother
- No increased risk of miscarriage
- It is mostly used to detect the sex of the baby where there is a specific medical reason why this information is important. For example in sex linked genetic conditions. In some cases it may be able to tell you whether the baby will be affected by particular genetic condition

## **Chorionic Villus sampling**

- Invasive test
- Available between 11-14 weeks of pregnancy
- Results are usually available within 1-2 weeks
- Can diagnose a known genetic condition
- It increases the risk of miscarriage in 1 in 100 cases (miscarriage risk of 1%)

## **Amniocentesis**

- Invasive test
- Available between 15-20 weeks of pregnancy
- Results are usually available in 2-3 weeks
- Can diagnose a known genetic condition
- It increases the risk of miscarriage in 1 in 100 women (miscarriage risk of 1%)

## **Pre-implantation Genetic Diagnosis**

- Can allow you to conceive a baby which will not be affected by the genetic condition in your family
- You may avoid having any of the above tests during pregnancy or making a decision whether to continue a pregnancy if it is affected by a genetic condition.
- Funding is available from NI health board for agreed conditions-your specialist will advise.
- There are specific criteria a couple must meet to be eligible.
- Can be a lengthy and emotionally stressful process.
- Involves travel to London.

## **Non Invasive Prenatal Diagnosis (NIPD)**

A test which can tell you the sex of the baby early in pregnancy. In some cases this test maybe used to determine whether or not the baby is affected with a genetic condition.

### **Who is this test helpful for?**

Some genetic conditions only occur in males and some others only in females. For families at risk of these sex-linked conditions, it can be useful to know the sex of the baby in early pregnancy. This can help to plan any further genetic testing during pregnancy, or can allow parents to prepare for the future. In some cases, it may also be used to diagnose whether or not a baby is affected with a genetic disorder. This can only be offered in very specific circumstances but is likely to become more common in the future. Often, preparation for this type of test would need to be commenced prior to becoming pregnant. Please discuss this with your genetic counsellor who can provide you with further information and whether this is appropriate or available for you.

### **How does it work?**

To determine fetal sex early in pregnancy, free fetal DNA (ffDNA) testing can be done. DNA from the baby can be detected in the mother's bloodstream during pregnancy. A sample of the mother's blood can therefore be taken to test the baby's DNA. This can tell us whether the baby is male or female. testing can be done. DNA from the baby can be detected in the mother's bloodstream during pregnancy. A sample of the mother's blood can therefore be taken to test the baby's DNA. This can tell us whether the baby is male or female.

## What happens?

A blood sample is taken from the mother early in pregnancy from maternity. The blood sample is usually taken at 9 weeks of pregnancy. A sample can be taken from 7 weeks but as this is very early, a second blood sample is then needed around 1 week later. The genetics laboratory then search this blood sample for genetic material on the Y chromosome which is only found in males. If this genetic material is found in the mother's blood sample, it means the baby is a boy. If this male genetic material is not detected, then the baby is highly likely to be a girl.

## How accurate is it?

Only a very small amount of the total DNA found in a mother's blood comes from the baby (about 3%). It is therefore possible that the laboratory might not be able to detect the male genetic material because there isn't enough DNA from the baby. Sometimes, we need to repeat the blood sample until the laboratory is able to detect enough DNA from the baby to produce an accurate result. The laboratory is unable to produce a result for around 5% of women, even if multiple samples are taken.

If a result is possible the accuracy of the test is close to 100%. If you have a twin pregnancy or other multiple pregnancy, it is possible that this will affect the reliability of the test.

## Chorionic Villus Sampling (CVS)

This test is used to accurately diagnose a genetic condition in the baby, early in pregnancy.

## What is chorionic villus sampling (CVS)?

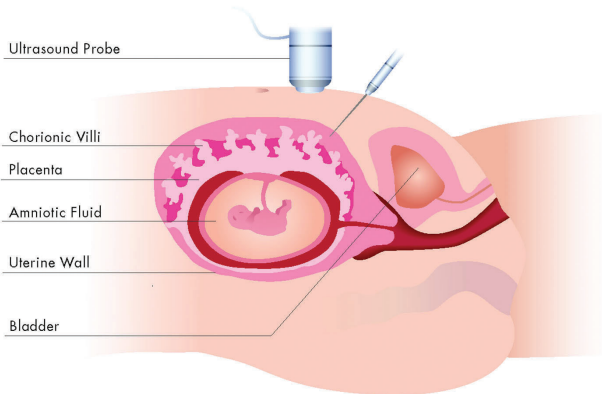
CVS is a test carried out during pregnancy, which involves removing tissue from the placenta. It is used to detect chromosomal and can also be used to check for particular genetic alterations in the baby.

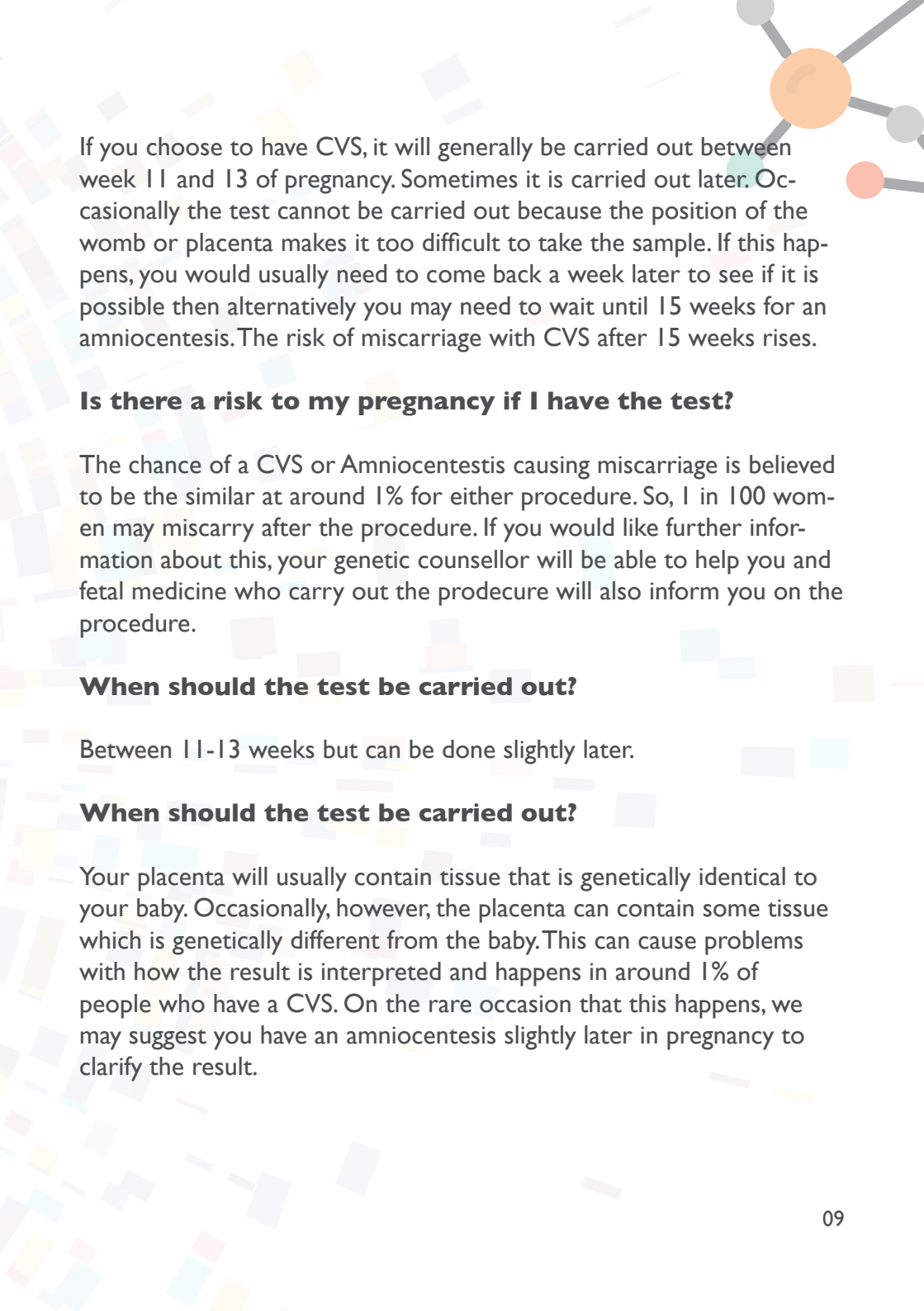
### How is CVS done?

An ultrasound probe is used to scan and check the position of your baby. Some local anaesthetic is applied and a fine needle is inserted through your abdomen and into your womb. A tiny sample of tissue is then removed from your placenta.

The test itself takes around 10 minutes. Some women say it is uncomfortable or slightly painful when the needle is inserted and when the sample of tissue is removed.

#### Transabdominal Chorionic Villus Sampling (CVS)





If you choose to have CVS, it will generally be carried out between week 11 and 13 of pregnancy. Sometimes it is carried out later. Occasionally the test cannot be carried out because the position of the womb or placenta makes it too difficult to take the sample. If this happens, you would usually need to come back a week later to see if it is possible then alternatively you may need to wait until 15 weeks for an amniocentesis. The risk of miscarriage with CVS after 15 weeks rises.

### **Is there a risk to my pregnancy if I have the test?**

The chance of a CVS or Amniocentesis causing miscarriage is believed to be the similar at around 1% for either procedure. So, 1 in 100 women may miscarry after the procedure. If you would like further information about this, your genetic counsellor will be able to help you and fetal medicine who carry out the procedure will also inform you on the procedure.

### **When should the test be carried out?**

Between 11-13 weeks but can be done slightly later.

### **When should the test be carried out?**

Your placenta will usually contain tissue that is genetically identical to your baby. Occasionally, however, the placenta can contain some tissue which is genetically different from the baby. This can cause problems with how the result is interpreted and happens in around 1% of people who have a CVS. On the rare occasion that this happens, we may suggest you have an amniocentesis slightly later in pregnancy to clarify the result.

## **What we be tested for?**

Several different tests will usually be done on the sample depending on whether you are having testing due to an increased risk of a chromosome rearrangement or if there is a particular condition they are looking for or your pregnancy is at risk from. It is also possible to detect the sex of the baby.

## **How long will the results take?**

The initial chromosome results whereby the laboratory check chromosomes 13, 18 and 21 are usually available 3-4 working days after the test. The test for the particular condition in your family will often take longer, and you will receive these results at separate times. Results will usually be ready within 1-2 weeks after the CVS.

## **Amniocentesis**

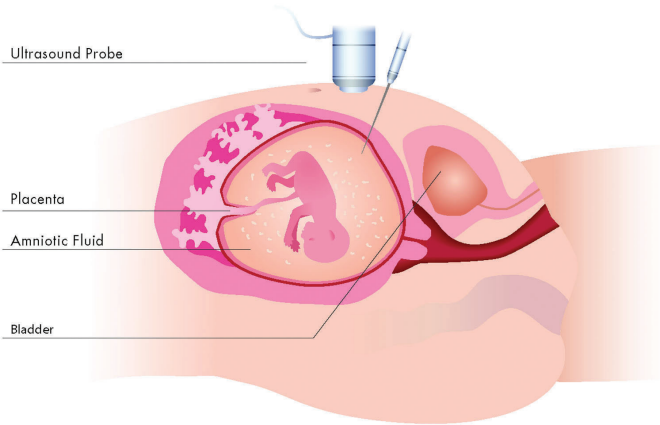
This test is used to accurately diagnose a chromosomal or genetic condition in the baby.

## **What is amniocentesis?**

Amniocentesis is a test carried out during pregnancy which involves using a fine needle to remove a small amount of the amniotic fluid around your unborn baby. This fluid contains cells from the baby. These cells develop in the laboratory and the DNA from the baby can be tested in the same way as a CVS.

# How is the Amniocentesis done?

## Amniocentesis



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CENTER

A fine needle is inserted through your skin, through your abdomen and into your womb. The needle is used to remove a small sample of the amniotic fluid surrounding your baby.

## When should the test be carried out?

If you choose to have an amniocentesis, it will usually be carried out between your 15th and 20th week of pregnancy. However, the test can be carried out later in pregnancy but options may become more limited after this time.

## Is there a risk to my pregnancy if I have the test?

Because amniocentesis is an invasive procedure, it increases the risk of miscarriage in 1 in 100 women (1%).

### **How accurate is it?**

The amniocentesis test is thought to be very accurate. There is a lower chance of the result being difficult to interpret than with a CVS. This is because the amniotic fluid contains cells which have come directly from the baby.

### **What will you test for?**

Several different tests will usually be done on the sample dependant on whether you are having testing due to an increased risk of a chromosome rearrangement or if there is a particular condition they are looking for or your pregnancy is at risk from. It is also possible to detect the sex of the baby.

### **How long will the results take?**

The initial chromosome results whereby the laboratory check chromosomes 13, 18 and 21 are usually available 3-4 working days after the test. The test for the particular condition in your family will often take longer, and you will receive these results at separate times. Your results will usually be ready within 2-3 weeks after the amniocentesis. This is slightly longer than for the CVS test because the cells need to develop in the laboratory for a few days before they can be tested. Your genetic counsellor/ obstetrician will discuss with you how you would like to receive your results. This will usually be by telephone.

### **Pre-implantation genetic diagnosis (PGD)**

Pre-implantation genetic diagnosis is a technique designed to help couples, who are at risk of having a child with a serious genetic condition have a healthy child without the familial genetic condition.

## **What is PGD?**

It is a technique used to create embryos and test them for the familial genetic condition. It involves contact with your local genetic department for the genetic testing, funding application and referral to a UK PGD centre such as Guys hospital in London. You will then attend assisted conception unit (ACU) attached to the PGD unit for the IVF part of the treatment. You may also attend the regional fertility centre in Belfast for some scans locally before implantation of unaffected embryo.

## **How successful is it?**

Approximately one in three cycles of PGD will result in a baby. The whole process can take 8-18 months dependant on the condition and the complexities of the PGD process.

## **Who can have PGD?**

It is not available to everyone and is only used for certain approved genetic conditions. There must be a licence in place for PGD to be used for the genetic condition in your family. Your doctor or genetic counsellor will be able to tell you whether PGD might be an option for you and you can apply for a licence for new conditions if necessary. Currently if the couples fit the below criteria, they will generally be offered up to three cycles of PGD on the NHS. This funding ceases if they have a healthy child, are not responding to treatment or no longer fit the eligibility criteria.

## The funding criteria include;

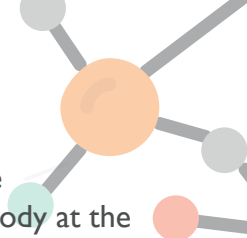
Treatment to start before the female partner is 40 yrs of age	Female partner must have a BMI (Body Mass Index) of more than 19 and less than 30	Both partner must be non-smokers	Testing for condition occurred in an accredited genetic laboratory	Couples have had genetic counselling locally	Couples in a year long relationship and co-habiting
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The reasons for these criteria are based on the success rates for the process. For couples who are not eligible for NHS funding, self-funded treatment is possible. Couples will be given a specific price but could likely range up to £12,000. In a few exceptional circumstances it may be possible to apply for funding for couples who have a health condition that prevents them from fulfilling funding criteria.

Funding is applied for from the Health board usually by your genetic counsellor, if you are granted approval this will cover the cost of treatment, travel and accommodation costs.

## What else is there to consider?

- Having PGD is a relatively lengthy and complicated process. There are times of uncertainty and many highs and lows throughout the process.
- The timing of PGD will depend on a number of factors and can vary for each couple.
- Couples will likely attend 4-5 appointments split between Guys Hospital and Regional Fertility Centre at the Royal Victoria hospital in Belfast for each cycle.

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- Women having PGD take extra hormones to control the development of eggs so they can be retrieved from the body at the appropriate time. There is an increased risk of multiple pregnancies as well as a small chance of ovarian hyper-stimulation syndrome (OHSS). These risks will be discussed at your appointments in detail before proceeding with PGD.

## **Continuing support and care during pre- natal testing**

For people whose results confirm a diagnosis, we will give you information and support to help you make a decision about your pregnancy.

- Some parents will want to prepare themselves for the birth knowing their baby may be at risk or affected.
- Some parents may decide not to continue with the pregnancy.

These are very personal and often difficult decisions and you will be given time and information to help you make the decision that is right for you. Whatever your decision, your healthcare professionals will do their best to support you. For individuals who decide not to have pre-natal testing gene testing is possible for the baby at birth, this can be discussed with your genetic counsellor or fetal medicine specialist. We are happy to co-ordinate testing or nay assessment of babies at birth.

## **Advanced Planning and Preparation**

With NIPD (Maternal blood) and PGD it is important to prepare in advance of falling pregnant.

With CVS and AMNIO you must inform your GP as soon as possible of your early pregnancy test and arrange early booking scan and your local maternity unit to confirm the pregnancy. You will then be referred to Fetal Medicine for the CVS or AMNIO procedure then remainder of your care will be back to your local maternity unit and your obstetrician.

The information on this booklet has been developed for the Northern Ireland Regional Genetics Service at Belfast City Hospital. It is based on text produced by the Genetics Unit at Birmingham Women's Hospital, NHS Fetal Anomaly Screening Programme and also text from Guy's Hospital, London.

**If you have any further questions please ask your genetic counsellor or consultant;**

Northern Ireland Regional Genetics Service  
Belfast City Hospital  
A Floor Tower Block  
Lisburn Rd, Belfast, BT9 7AB  
Telephone: 028 950 46426  
E mail [bs0.nigeneticcounsellors@nhs.net](mailto:bs0.nigeneticcounsellors@nhs.net)

Seen in clinic by (Doctor):

(Genetic Counsellor):

Family reference number:

**Useful websites and Contacts**

PGD Team, Clinical Genetics, 7th Floor, Borough Wing, Guy's Hospital,  
London SE1 9RT  
Telephone: 020 7188 1364  
E mail: [PGDGenetics@gstt.nhs.uk](mailto:PGDGenetics@gstt.nhs.uk)  
w: [www.guysandstthomas.nhs.uk/our-services/pgd/](http://www.guysandstthomas.nhs.uk/our-services/pgd/)

**Antenatal Results and Choices (ARC)**

support and information for women during the antenatal testing process Provides  
Helpline: 0845 077 2290 or 0207 713 7486  
Email: [info@arc-uk.org](mailto:info@arc-uk.org)  
Website: <http://www.arc-uk.org/>  
[www.gov.uk](http://www.gov.uk) 'Screening in pregnancy-CVS & Amniocentesis information for parents'.

Regional Fertility Clinic, Royal Jubilee Maternity Service, Belfast HSC Trust, Grosvenor Rd, Belfast, BT12 6BA  
Phone Option 1 028 9615 9600 9.30-12.30 Mon to Friday

Fetal Medicine Unit, Royal Jubilee Maternity Hospital, Belfast HSC Trust, Grosvenor Rd, Belfast, BT12 6BA  
Phone: 028 9615 1173