or friends. Think about how you might feel about the test result and how important the information would be for you and your family. If you are unsure about anything, it can be helpful to discuss it with your healthcare professional.

What happens to the blood sample after NIPD has been done?
In many countries, after the test results have been confirmed no further testing on the blood is done and the sample will be destroyed. However, you should ask your health professional what happens in your own country.

Where can I get further information and support?
A list of national support groups can be found on the EuroGentest website at:
http://www.eurogentest.org/index.php?id=680

EuroGentest has also developed a series of leaflets to provide general information for patients and families about genetics and genetic testing. These can be found here:
http://www.eurogentest.org/index.php?id=226

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Non-invasive prenatal diagnosis for single gene disorders

This leaflet is designed to give you information about a new prenatal test called non-invasive prenatal diagnosis (NIPD) that is now available to women during pregnancy. It can be offered when:

- One or both parents are carriers of a particular genetic condition that might be passed on to the baby, or
- You have had another type of test during pregnancy (such as an ultrasound scan) which has shown there is an increased risk that your baby has a genetic condition.

If you are considering using NIPD, this leaflet may be helpful to you. If anything is unclear or you want to discuss your personal situation, you should discuss the test with your own healthcare professional, such as your midwife, genetic specialist, family doctor or obstetrician.

What is NIPD?
Genetic conditions are caused by changes in the DNA. DNA is in all the cells in our body and codes for our characteristics, such as our eye colour and height. Changes in the DNA can also cause genetic conditions. We now know that the baby’s DNA circulates in the mother’s blood during pregnancy and by testing this DNA, we are able to identify whether or not the baby is affected by a particular genetic condition. The baby’s DNA is lost from the mother’s bloodstream within a few hours of delivery and so testing is specific to the baby in that pregnancy.

For most genetic conditions, there are several potential gene faults or DNA changes that can cause the condition. To ensure the laboratory staff know which one to look for, it is usually necessary to test the DNA of the parents or a previously affected individual before NIPD is done, preferably before pregnancy.

How is the test done?
The test is performed on a sample of the mother’s blood. About 20mls (roughly two tablespoons) is taken from the arm like a normal blood test. The blood is then sent to the laboratory for testing.

When is the test done?
The timing of the test is important as it works best after 7 weeks of pregnancy, but in some cases the doctor will recommend it is done later. You will need to have an ultrasound scan first to find out exactly how many weeks pregnant you are and whether there is more than one baby in the womb (such as twins).

How accurate is the test?
At the moment we can only do this test for a few genetic conditions. NIPD for single gene disorders has been shown to be highly accurate. However, as this is a new development for some conditions your health professional may suggest using invasive testing to confirm a positive NIPD test result. Recommendations will vary depending on the condition being tested and your health professional will discuss the details relevant for your particular situation.

How long does it take to get the result?
It will usually take 1-2 weeks to get the test result, but this might vary from centre to centre so you should ask your health professional when you can expect to receive your result. Occasionally the laboratory is unable to give a result. This might be because there was not enough of the baby’s DNA present in the blood sample to perform the test. If this happens the test can be repeated.

How safe is the test?
The test is a blood test taken from the mother’s arm like a normal blood test. Therefore, the test carries no significant risk to you or your baby. As is the case with all blood tests, there may be some bruising around the area where the blood sample was taken.

Does the test look for all genetic conditions?
No, the test can only be used for specific conditions. The condition tested for will be discussed with you by your health professional before the blood sample is taken.

Do I have to take the test?
It is your choice whether or not you take any test in pregnancy, including NIPD. Before making a decision about NIPD, you may want to take some time to consider the test and discuss it with your partner or other relatives.