(amniocentesis or chorionic villus sampling) to confirm this. If you are considering terminating the pregnancy, it is strongly recommended that you have an invasive test for confirmation because of the small risk of a false positive result. Alternatively you might decide to continue with the pregnancy without any further testing. Your health professional will discuss these options fully with you. You may find it helpful to read the information on amniocentesis and chorionic villus sampling on the EuroGentest website (details can be found at the end of this leaflet).

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

**What happens to the blood sample after NIPT 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?**
European Down Syndrome Association
http://www.down-syndrome.eu/index.html
There are Down Syndrome organisations in most European countries, these are listed on the EDSA website under members at:
A list of other publications on genetic testing including amniocentesis and chorionic villus sampling can be found on the Eurogentest website at:
http://www.eurogentest.org/index.php?id=226

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Non-invasive prenatal testing:  
a new test for Down syndrome

This leaflet is designed to give you information about a new non-invasive prenatal test (NIPT) for Down syndrome. If you are considering using NIPT, please read this leaflet carefully. 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, family doctor or obstetrician.

What is Down syndrome?
Down syndrome is a life-long condition that causes delays in learning and development. It is a variable condition and some people may be more seriously affected than others. For example, it can cause certain medical problems such as heart problems. However, many people with Down syndrome live healthy and satisfying lives. Anyone can have a baby with Down syndrome, but we do know that the risk increases as women get older.

What is NIPT?
DNA is the chemical substance that makes up the chromosomes that are in all of our cells. We now know that the baby’s DNA is present in the mother’s blood from early in pregnancy. Each chromosome has specific DNA sequences associated with it and by counting these sequences in the mother’s blood we are able to determine whether or not the baby is affected by chromosomal conditions like Down syndrome. 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.

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?
There is only enough DNA present in the mother’s blood to conduct the test from around 10 weeks of pregnancy, so the test cannot be done before then. 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 NIPT for Down syndrome?
NIPT for Down syndrome is over 99% accurate. However, there is still a small chance that the result will be incorrect:
- There is a small chance (0.5%) that the test will incorrectly show that the baby has Down syndrome when it does not.
- There is a small chance (less than 1%) that the test will incorrectly show that the baby does not have Down syndrome when it does.

Because of the very small chance of an incorrect result, if you have a positive NIPT result you will be offered an invasive diagnostic test to confirm the result. Further information about invasive tests are provided below.

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.

Can NIPT detect conditions other than Down syndrome?
Down syndrome is the most common condition detected by NIPT. Two rarer chromosome conditions, Edwards syndrome (trisomy 18) and Patau syndrome (trisomy 13) can also be identified using NIPT. These conditions are very serious and many affected babies die before or soon after birth. Different providers offer different tests and your health professional will discuss exactly what conditions are tested for. Like Down syndrome, if the NIPT test is positive for either of these conditions an invasive test is recommended to confirm the result.

What happens if the NIPT result shows the baby has Down syndrome?
If the result shows the baby is affected by a chromosome abnormality you need to decide whether or not to have an invasive test.