

Prenatal diagnosis and testing

What is prenatal diagnosis?

In the case of a pregnancy, if one or both parents are 'carriers' of a particular muscle-wasting condition (see the Muscular Dystrophy UK factsheet: *Carrier testing and reproduction: your options*), there is a risk that their unborn child will be affected by that condition.

'Prenatal tests' are carried out during pregnancy to try to find out if the foetus (unborn child) is affected. The tests are only available for some muscle-wasting conditions. You can check on availability by contacting your geneticist.

As a couple, you may want this information for two reasons: either to consider whether or not to continue the pregnancy if the foetus is affected, or to continue with the pregnancy armed with the information to prepare and plan for the future.

Different types of prenatal tests can be carried out at different times during the pregnancy:

- 1) non-invasive prenatal diagnosis (NIPD) – can be carried out from seven weeks' gestation
- 2) chorion villus sampling (CVS) – at around 10-12 weeks
- 3) amniocentesis – at around 14-16 weeks
- 4) placental biopsy and foetal blood sampling – at around 18 weeks
- 5) ultrasound scanning to determine the gender of the baby (some conditions such as Duchenne muscular dystrophy will only affect boys in almost all cases) – at around 20 weeks.

Women/couples need to consider carefully which test to have and discuss this with their genetic counsellor, obstetrician or midwife. Earlier testing would allow early termination of the pregnancy, which would potentially be less traumatic for the couple, however it carries a slightly higher risk of miscarriage than later testing (see below).

To get the maximum benefit from these tests, it is very important that investigations into carrier status should commence BEFORE a pregnancy is started. For this reason, many people choose to be carrier tested in their late teens.

What are these tests and what do they involve?

Non-invasive prenatal diagnosis (NIPD)

It has been known for some time that DNA from the baby can be detected in the mother's blood, early in a pregnancy. NIPD is a new test that has recently been developed, which can extract the baby's DNA directly from a routine blood test taken from the mother. This test cannot always be used to determine whether the foetus is affected by a muscle-wasting condition – but it can be used to find out the sex of the foetus.



Knowing the foetal sex may be important for women who are carriers of types of muscle-wasting conditions that affect mainly males (see our leaflets on *Inheritance and the muscular dystrophies*).

Women who know they are having a boy may then want to have further tests, such as CVS or amniocentesis (see below). Women who know they are having a girl, do not need to proceed to any further prenatal testing that may put the pregnancy at risk.

Women who are considering NIPD should discuss this with their doctor or genetic counsellor. This would be important to establish whether or not this test would be appropriate and available for them, as well as the accuracy of this testing for their particular condition.

What does the test involve?

NIPD is done on a sample of the mother's blood, taken from the arm. NIPD to determine the sex of the foetus can be done from seven weeks into the pregnancy.

How accurate is the test?

NIPD tests to determine the sex of the foetus are around 99 percent accurate. Your geneticist will discuss all these factors with you before any test, and will explain whether or not there is likely to be a risk of error with the test result.

What are the risks of the test?

Because NIPD testing is via a blood sample, there is no risk to the pregnancy.

Chorion villus sampling (CVS)

The chorion is the tissue that surrounds the foetus during early pregnancy, part of which later becomes the placenta (afterbirth). It contains the same DNA as the foetus. A tiny sample of the chorion is obtained at a very early stage of the pregnancy.

Scientists will use 'direct genetic tests' or 'linked genetic markers' (see our factsheet on *Carrier testing and options for reproduction*) to try and discover whether or not the foetus has inherited the genetic mutation. They will be looking for the same genetic mutation as the one already identified in an affected person in the family. The parents will be told the results, and will then decide whether or not to continue with the pregnancy.

What does the test involve?

The CVS test can be performed at about 10 to 12 weeks of pregnancy. An ultrasound scan is performed first, to check how far advanced the pregnancy is, and to determine the position of the placenta. The test involves passing a needle through the abdomen in a similar way to an amniocentesis (see below) to obtain a small piece from the chorion. The test usually takes about 20 to 30 minutes, and can be done in a hospital outpatient department. The woman being tested is awake and aware of what is happening during the test.



Is the test painful?

Most women describe the test as being uncomfortable rather than painful. Local anaesthetic is usually used but most women say that they are aware of a 'pushing' feeling; they may feel some soreness over the area afterwards.

How accurate is the test?

Sometimes it is important to know the sex of the foetus and this can be determined very quickly using this test. Identifying a genetic mutation can depend on several factors, such as the size of the mutation, and therefore how easy it is to find, or whether or not a sample from an affected relative has been obtained, or how accurate 'linked genetic markers' are. Your geneticist or obstetrician can discuss all these factors with you before any test, and explain whether or not there is a risk of error with the test result.

Is the test safe?

There is a very small risk of miscarriage (one to two percent), which is a higher risk than for amniocentesis (see below). As far as the mother's health is concerned, there are thought to be no serious implications, although she should avoid strenuous exercise for a few days following the test.

Amniocentesis

What does the test involve?

First of all, an ultrasound scan is done to check how far advanced the pregnancy is. A fine needle is passed through the abdomen, and a sample of about 20ml (four teaspoons) of the amniotic fluid surrounding the foetus in the uterus (womb) is taken.

This fluid sample contains DNA from the foetus. Amniocentesis can be carried out in a hospital outpatient department, and the woman being tested is awake and aware of what is happening during the test. It usually takes only a few minutes.

Is it painful?

Women who have had the test generally say it is no more painful than having a blood test or an injection.

How accurate is the test?

Amniocentesis results are as accurate as CVS results. The accuracy varies from one condition to another. Your geneticist will discuss this with you before you take the test.

Is the test safe?

There is a slight risk (about half to one percent) of miscarriage. Otherwise it is not thought to harm the baby. Women are advised to avoid strenuous exercise for a few days following the test.

Ultrasound scanning to determine the sex of the baby

For sex-linked conditions, these scans may be performed from about 20 weeks.

Determining the sex of the baby depends on the development of the external genitals of a male. Occasionally these may be under-formed and the sex may not be accurate at this stage.



Other related publications:

- ▶ Inheritance and the muscular dystrophies
- ▶ Pregnancy & fertility
- ▶ Carrier testing and reproduction: your options

We're here for you at the point of diagnosis and at every stage thereafter, and can:

- ▶ give you accurate and up-to-date information about your or your child's muscle-wasting condition, and let you know of progress in research
- ▶ give you tips and advice about day-to-day life, written by people who know exactly what it's like to live with a muscle-wasting condition
- ▶ put you in touch with other families living with the same muscle-wasting condition, who can tell you about their experiences
- ▶ tell you about – and help you get – the services, equipment and support you're entitled to.

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Here for you

The friendly staff in the care and support team at the Muscular Dystrophy UK's London office are available on **0800 652 6352** or **info@muscular dystrophyuk.org** from 8.30am to 6pm Monday to Friday to offer free information and emotional support.

If they can't help you, they are more than happy to signpost you to specialist services close to you, or to other people who can help.

www.muscular dystrophyuk.org