

Prenatal Diagnosis

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Most babies are born healthy, but a small number (about 1 in 30) are born with a problem. Some problems are more severe than others and about 1 in 50 (2%) babies in the UK are born with a serious disability. Examples of serious disability include Down syndrome and spina bifida, and there are many others. Some of the problems are genetic disorders. Genetic disorders are caused by a chromosome abnormality, or by a gene with a mutation or change. Many genetic disorders affect more than one member of a family. Sometimes, a baby may be the first person in a family to be affected. Examples of genetic disorders include cystic fibrosis, thalassaemia, sickle cell anaemia, Down syndrome, Duchenne muscular dystrophy. Babies may also have problems that do not have a simple genetic cause, such as spina bifida.

Prenatal diagnosis can be used to find out whether an unborn baby has a serious problem. It is important to note that not all conditions can be detected. If a prenatal test is possible in your family, this does not mean you must have the test. Prenatal diagnosis involves testing an unborn baby for a genetic condition. A baby's genes or chromosomes can be checked. The decision whether or not to go ahead with prenatal diagnosis is a very personal one. You will be supported by medical staff in whatever decision you make.

What are genes and chromosomes?

Our bodies are made up of millions of cells. Each cell contains a complete set of genes. We have thousands of genes. We each inherit two copies of most genes, one copy from our mother and one copy from our father. Genes act like a set of instructions, controlling our growth and how our bodies work. Any alteration in these instructions is called a mutation (or change). Mutations (or changes) can stop a gene from working properly. A mutation (change) in a gene can cause a genetic disorder. Genes are responsible for many of our characteristics, such as our eye colour, blood type or height.

Genes are carried on thread-like structures called chromosomes. Each of us has 46 chromosomes in every cell. We inherit our chromosomes from our parents, one set of 23

chromosomes from our mother and one set of 23 chromosomes from our father. So we have two sets of 23 chromosomes, or 23 pairs. One pair of chromosomes are called the sex chromosomes. It is possible to see chromosomes under a microscope. Picture 1 shows what they look like.

Picture 1. Chromosomes as they appear under a microscope



Inheritance of Genetic disorders

Genetic disorders can be inherited in several ways.

- For some disorders, an affected person has an affected parent. One copy of a gene with a change (mutation) is enough to cause the disorder.
- For other disorders, an affected person inherits one copy of a gene with a change (mutation) from each parent. In this type of inheritance, the parents may be healthy carriers of a single changed gene. Carrier parents do not have any sign of the disorder.
- If a genetic disorder is caused by a mutation (change) in a gene on the X chromosome (one of the sex chromosomes), it is said to be a sex-linked or X-linked disorder. These genetic disorders usually affect only boys. Women who have the gene with the mutation (change) are usually unaffected carriers, but they can pass the condition on to their sons.
- In chromosome disorders, a chromosome is affected, rather than a single gene. For instance, part of a chromosome may be missing or rearranged, affecting many genes.

Prenatal testing is available for some genetic disorders, but not for all of them.

The precise method used for testing depends on the disorder for which a baby is being tested. If you decide to have your baby tested for a particular genetic disorder, you will be told how that specific test is done.

How accurate are tests for genetic disorders?

If you decide to have your baby tested for a genetic disorder, including a chromosome abnormality, you will be told how accurate the result of that test is expected to be. It is important to remember that if a baby is being tested for a genetic disorder, the test result will only be about that one disorder. There is no general test for all genetic disorders.

The following information describes the different ways testing can be done.

Amniocentesis

What is an amniocentesis and how is it done?

Amniocentesis is a test on an unborn baby's genes or chromosomes, done while the baby is in the womb. Amniocentesis is usually done between 15 and 16 weeks of pregnancy but it may be done earlier. Amniocentesis is an outpatient test; you do not have to be admitted to hospital. An ultrasound scan is done first, to check the position of the baby and the placenta (or after-birth). A fine needle is passed through the skin and the abdomen (the tummy or belly) into the womb, and a syringe is used to remove a sample (about 15mls or 3 teaspoonfuls) of the fluid that surrounds the baby. The fluid surrounding the baby contains some of the baby's skin cells and these can be examined in the laboratory to check the baby's genes and chromosomes.

Most women consider amniocentesis to be uncomfortable but not really painful. It is usually over in a few minutes. If your blood type is Rhesus negative, you will be given an injection of anti-D after the procedure, to prevent complications related to your blood type. Some women get a tightening feeling in the womb afterwards, or may feel a little soreness for one day. This is not unusual, and women are advised to relax for two days and to avoid any heavy lifting or strenuous exercise. Women who have had the test should be driven home by someone else, and should then rest for the day. Occasionally (1 in 100 samples) there is not enough material obtained from the amniocentesis for the test to be done on that sample. When this happens, it is usually obvious in the laboratory, about a week to 10 days after the amniocentesis. If this happens, you will be notified and offered another test. If you have abdominal discomfort which lasts longer than 24 hours, or if you have fever, or if you have any unusual vaginal discharge or vaginal bleeding, please contact the unit where the amniocentesis was done, to get advice.

What are the risks of amniocentesis?

Up to 1 woman in 100 will have a miscarriage as a result of having an amniocentesis. This means that 99 out of 100 pregnancies should continue normally.

When and how will you get the results of an amniocentesis?

If an amniocentesis is done to test for a chromosome abnormality, the result is usually ready in 2-3 weeks. If an amniocentesis is done to test for a mutation (change) in a gene, you will be told when the result will be available. Specialist genetic counsellors are usually involved in giving the results of prenatal diagnostic tests. Normally, you will be contacted by telephone with the results. If an abnormality is found, it will be explained and you can then decide whether or not to continue with the pregnancy. Termination of pregnancy at this stage usually involves going through early labour.

Chorionic Villus Sampling

What is Chorionic Villus Sampling and how is it done?

Chorionic Villus Sampling involves obtaining a small number of cells from the developing placenta, which has the same genes as the baby. It is done between 10 and 12 weeks of pregnancy. The test is done as an outpatient procedure. It is advisable that you come for Chorionic Villus Sampling with your partner or a friend. An ultrasound scan is done first to check the position of the baby and the placenta. There are two possible ways to obtain a sample. It can be taken in much the same way as a smear test is done, through the vagina. Most women say that this feels like a smear test and is not painful. The sample can also be obtained using a needle inserted through the skin and the abdomen and into the placenta, in a similar way to an amniocentesis. If your blood type is Rhesus negative, you will be given an injection of anti-D after the procedure, to prevent complications related to your blood type. You should be driven home afterwards by someone else, and you should rest for the day. Some women who have Chorionic Villus Sampling done vaginally have some spotting (spots of blood) and some period-like cramping afterwards; this is normal. If you have heavier bleeding, you should let your own doctor (GP) or Obstetrician know. You should not have sexual intercourse until after any bleeding has stopped.

What are the risks of Chorionic Villus Sampling?

In general about 1-2 women in 100 will have a miscarriage as a result of having Chorionic Villus Sampling. Individual obstetric centres usually have their own risk figures, which may be slightly lower than this.

When and how will you get the results of Chorionic Villus Sampling?

If the sample is being tested for a chromosome disorder, usually, the final result will be available in 2-3 weeks. If the result takes longer than this, it does not mean that the result is abnormal, just that the cells in the sample are taking a longer time to grow. About 1 time in 100 the chromosome result is not clear. In this case another test has to be done, such as amniocentesis, or a blood test on the baby, to obtain a definite answer. If the sample is being tested for a gene change (mutation), you will be advised as to when the result will be available. Specialist genetic counsellors are usually involved in giving the results of prenatal diagnostic tests. You will normally be contacted by telephone with the results. If an abnormality is found, it will be explained and you can then decide whether or not to continue with the pregnancy. If a termination is decided on at this stage, it can usually be done under a general anaesthetic.

Who should consider having an amniocentesis or Chorionic Villus Sampling?

- Women who have had a “high risk” result from a nuchal translucency test or a blood test designed to find out the risk of having a baby with Down syndrome.
- Women who have a potential problem found on an ultrasound scan, which may suggest a chromosome abnormality.
- Women who have had a termination of a previous pregnancy for a genetic disorder.
- Parents who have had a baby with a genetic disorder or those who have a family history of genetic disorders, including rare inherited diseases.
- Women over 36 years old who have missed the opportunity to have a Down syndrome screening test.

You should only have an amniocentesis or Chorionic Villus Sampling if you and your partner

- feel that it is important for you to have the information which the test can provide
- do not feel the risks are too great for you to take.

Where can I get more information about prenatal diagnosis?

This is only a brief guide to prenatal diagnosis. More information can be obtained from your local regional genetics centre or from these addresses:

Antenatal Results and Choices

73 Charlotte St., London, W1T 4PN

Tel: 020 7631 0285

Email: info@arc-uk.org

Web: www.arc-uk.org

The Genetic Interest Group

Unit 4D, Leroy House, 436 Essex Rd., London, N1 3QP

Telephone: 020 7704 3141

Email: mail@gig.org.uk

Web: www.gig.org.uk

Contact a Family

209-211 City Rd., London, EC1V 1JN

Telephone: 020 7608 8700

FAX: 020 7608 8701

Helpline 0808 808 3555 or Textphone 0808 808 3556

(Freephone for parents and families, 10am-4pm, Mon-Fri)

Email: info@cafamily.org.uk

Web: www.cafamily.org.uk

This edition prepared in July 2005

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Glossary (difficult words and their meanings): Prenatal Diagnosis

This glossary is intended only to explain terms used in the information: **Prenatal diagnosis**. Words shown in **bold** are defined elsewhere in the glossary.

abdomen or tummy/belly The part of the body between the chest and the legs. It contains the gut or digestive system, and in pregnancy, it contains the growing baby.

amniocentesis. A test on an unborn baby's **genes** or **chromosomes**. The baby is surrounded by fluid in the **womb**. The fluid contains a few **cells**. A small sample of the fluid is taken with a thin needle, through the skin of the mother's **abdomen** (tummy or belly). The fluid is sent to a laboratory for testing.

anti-D. An injection for women whose blood type is **Rhesus negative**, given to prevent potential complications in the baby. This is needed after an **amniocentesis** or **chorionic villus sampling**.

carrier. Carrier is used in the descriptions of two different types of inheritance in this information. These are two different ways of being a carrier:

1. A carrier is someone who has one normal copy of a **gene** and who has a change (**mutation**) in the other copy of that **gene**. The presence of the normal **gene** usually ensures that a carrier is not affected by the corresponding **genetic** disorder. This applies for the type of **genetic** disorder in which affected people have changes (**mutations**) in both copies of a **gene**.
2. A carrier is a woman who has one normal copy of a gene on the **X chromosome** and who has a change (**mutation**) in the other copy of that **gene** on her other **X chromosome**. The presence of the normal **gene** usually ensures that she will not be affected. Carriers are not usually affected by the corresponding **X-linked genetic disorder**.

cell. The human body is made up of millions of cells, which are like building blocks. There are many specialised types of cells. These include skin cells, brain cells, and blood cells. Cells in different parts of the body look different and do different things. Every cell (except for eggs in women and sperm in men) contains all the body's **genes**.

chorionic villus sampling, CVS. A test on an unborn baby's **genes** or **chromosomes**. This test can be done earlier than **amniocentesis**. A small number of **cells** is taken from the developing **placenta** and sent to a laboratory for testing.

chromosomes. Thread-like structures which can be seen under the microscope and contain the **genes**. Usually people have 46 chromosomes in every **cell**. There are two sex chromosomes and 22 other pairs of chromosomes. Twenty-three chromosomes come from the mother, and twenty-three come from the father. One chromosome of each pair comes from each parent. (As an analogy: a chromosome is like a book; a **gene** is like a story in the book).

cystic fibrosis. A **genetic** disorder caused by having a change (**mutation**) on both copies of the cystic fibrosis **gene**. Most children with cystic fibrosis have problems with their lungs and with their digestive system.

Down syndrome. A **genetic** disorder caused by having an extra **chromosome 21**. Most children with Down syndrome have learning disabilities and they share a typical facial appearance. They may have other medical problems.

Duchenne muscular dystrophy. An **X-linked genetic** disorder caused by a change (**mutation**) in a **gene** on the **X chromosome**. This disease of the muscles affects boys from early childhood, and progresses. Boys who have one copy of the **gene** with the **mutation** (change) are affected. Girls with one copy of the **gene** with the **mutation** (change) are **carriers** and are usually not affected.

gene. Information needed for the body to work, stored in a chemical form on **chromosomes**. Changes or **mutations** in genes alter the information and this can change how the body works. Most genes are in pairs, one from the mother and one from the father. The two genes of a pair are at matching places on a pair of **chromosomes**. Genes on the **X chromosome** and the **Y chromosome** of males are not paired with each other; they do not match. (As an analogy: a gene is like a story in a book, changes in genes are like changes in a story).

general anaesthetic. Treatment given to remove sensation while a procedure (such as an operation) takes place. The patient is not awake under general anaesthetic.

genetic. Caused by **genes**, concerning **genes**.

genetic counsellor. A specialist who gives information and support to people who have a history of a **genetic** condition in their family, or who are concerned about their risk for inheriting or passing on a **genetic** condition.

miscarriage. Early end to a pregnancy, before the baby can survive outside the womb.

mutation. A change in a **gene**. Some mutations are not harmful. Sometimes when a **gene** is changed, its information is altered so it does not work properly. (As an analogy: a change or mutation in a **gene** is like a missing or extra letter in a word in a story).

nuchal translucency test. An **ultrasound scan** of the back of the baby's neck. If there is a problem (such as **Down syndrome**), the result may be abnormal.

obstetrician. A doctor who specialises in pregnancy and childbirth.

placenta (or after-birth). The placenta lies against the wall of the **womb** in a pregnant woman. The baby gets its nourishment from the placenta. The placenta has the same **genes** as the baby.

prenatal diagnosis. Test during a pregnancy for the presence or absence of a **genetic** disorder in the baby.

rhesus negative, RhD negative. A particular blood type. If a rhesus negative mother has a test involving a needle in the **womb**, she will be given an injection of **anti-D**, to prevent problems in the baby. This is not needed if the mother is rhesus positive.

sex chromosomes. The **X chromosome** and the **Y chromosome**. The sex chromosomes control whether a person is male or female. Females usually have two **X chromosomes**. Males usually have one **X chromosome** and one **Y chromosome**.

sex-linked condition. See **X-linked** condition.

sickle cell anaemia. A **genetic** disorder caused by having a change (**mutation**) on both copies of the sickle cell **gene**. People with sickle cell anaemia can have attacks of severe pain or sudden life-threatening infections. The risk of inheriting a sickle cell **gene** with a change (**mutation**) depends on the ancestry of the person. Most affected people are born to parents who are both healthy **carriers** and have no affected relatives.

smear test. A test recommended for all women, to check for abnormality in the **cells** at the opening of the **womb**.

spina bifida. A disorder of the spine. Severely affected people may have considerable physical disability. This disorder does not have a simple **genetic** cause.

spine. The backbone. The series of bones in the middle of the back.

termination. Early ending of a pregnancy by doctors.

thalassaemia. A group of **genetic** disorders affecting the blood. The effects of thalassaemia depend on the particular thalassaemia **gene** which has a change (**mutation**), and on how many changed thalassaemia **genes** a person has. The risk of inheriting a thalassaemia **gene** with a change (**mutation**) depends on the ancestry of the person. Most affected people are born to parents who are both healthy **carriers** and have no affected relatives.

ultrasound scan. An examination of the growing baby during the mother's pregnancy. This is painless. It is done from outside the body, using a microphone on the skin of the **abdomen** (tummy/belly).

vagina. The connection from the **womb** to the outside of a woman, the birth canal.

womb. The part of a woman's body in which a baby grows during pregnancy.

X chromosome. One of the **sex chromosomes**. Females have two X **chromosomes** Males have one X **chromosome** and one **Y chromosome**.

X-linked. Describes a gene on the **X chromosome**. An X-linked **genetic** disorder is one caused by a **mutation** (change) in a **gene** on the **X chromosome**.

Y chromosome. One of the **sex chromosomes**. Males have one Y **chromosome** and one **X chromosome**. Females have two **X chromosomes**.

This glossary is intended only for use by patients and families, with the genetic information to which it refers.

This edition prepared in July 2005

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