

## Will it happen again?

In around 10% of cases the 22q11 deletion is inherited from one parent. Both parents will be offered a blood test to find out whether one of them carries the deletion. If both parents' chromosomes are normal the chance of having a further baby with the deletion is low (less than 1%). If one parent has the 22q11 deletion then that parent has a 50% (1 in 2) chance of passing on the deletion to each of his or her children.

## Prenatal diagnosis

The FISH test can be carried out during pregnancy to test an unborn baby. The FISH test shows whether the baby has the deletion, but cannot predict how severely affected the individual will be. There may be multiple problems or no problems.

Detailed ultrasound scanning will be able to detect certain defects such as cleft palate. A fetal heart scan (fetal echo) at 14 and 20 weeks can detect most heart defects.

## For more information

If you need more advice about any aspect of 22q11 deletions, you are welcome to contact:

### CLINICAL GENETICS DEPARTMENTS:

#### Northern Scotland (main base Aberdeen)

Tel: 01224 552120 / Fax: 01224 559390  
(Aberdeenshire, Moray, Highland, Western & Northern Isles)

#### Tayside (main base Dundee)

Tel: 01382 632035 / Fax: 01382 496382  
(Perth & Kinross, Angus, North East Fife)

#### South East Scotland (main base Edinburgh)

Tel: 0131 537 1116 / Fax: 0131 537 1153  
(Borders, Lothian, South West Fife)

#### West of Scotland (main base Glasgow)

Tel: 0141 354 9200 / Fax: 0141 232 7986  
(Glasgow, Argyll & Bute, **AYRSHIRE**, Dumfries & Galloway, Stirling, Lanarkshire, Falkirk)

If you would like further information and the opportunity to talk to other parents you can contact:

Max Appeal  
15 Meriden Avenue, Wollaston, Starbridge  
West Midlands  
DY8 4QN  
0800 389 1049 (Freephone)  
[www.maxappeal.org.uk](http://www.maxappeal.org.uk)

Seen in clinic by .....

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# 22q11 Deletions



General Information

## What is 22q11 deletion?

Genes are the blueprint of the body. When one or more genes are lost the blueprint is changed, frequently leading to problems from birth and learning difficulties. Genes are made of a chemical called DNA and contained inside larger structures called chromosomes which are found in every cell in the body.

Most people have 23 pairs of chromosomes (46 in total). One of each pair comes from the mother and the other from the father. Chromosomes are numbered 1 (the largest pair) to 22 (the smallest pair), the 23rd pair are called sex chromosomes because they determine whether a person is male or female. Each chromosome has a "q" arm (which is long) and a "p" arm (which is short). The genes are arranged in clusters along these arms.

Someone with a 22q11 deletion has a very small piece missing from the "q" arm of one of their chromosome 22s.

## How is it detected?

The piece of missing chromosome is so small that it cannot be seen under a microscope. A special test called the FISH test (fluorescence in situ hybridization) is used to show whether or not this region of chromosome 22 is present.

## How common is it?

22q11 deletion is one of the commonest chromosomal abnormalities. It is thought to be present in 1 in 3000 - 4000 births. It may be even more common than this as some people who carry the deletion never come to medical attention.

## Why did it happen?

The chromosome deletion was present in either the egg or sperm from which the baby was conceived.

DNA copying errors frequently occur when the eggs and sperm are made. The process of copying the DNA when the eggs and sperm are made is never completely perfect. There is nothing that either parent could have done before or during the pregnancy to have prevented the deletion from happening or that could have been done to cause the deletion to occur. It is important to understand that this is no one's fault.

## How will my child be affected?

22q11 deletion is associated with at least 2 different syndromes. A "syndrome" is a recognised collection of symptoms or findings/abnormalities which commonly occur together. Syndromes are commonly called after the doctor who first described them. 22q11 deletion is found in both DiGeorge Syndrome and Shprintzen (velo-cardio-facial) syndrome. However many children with 22q11 deletions do not have all or even any of the problems associated with either of these syndromes.

The effect of this deletion is extremely variable and very difficult to predict. Over 100 different features are found more commonly in individuals with 22q11 deletions than in the general population. The chance of any one child with a 22q11 deletion having any one of these is small. Once a child has been diagnosed a medical assessment is undertaken to make sure any potential problems are checked for and the child is referred to relevant specialist.

## Medical Problems associated with 22q11 deletion

### Heart problems

Many children with 22q11 deletion are born with a severe heart defect. Even if your child does not have a known heart defect, assessment by a heart specialist (cardiologist) and a heart scan will be offered.

The chances of finding a heart defect in this situation is very low but it is important to find out, as effective treatment can be given if a heart problem is detected.

### Developmental delay, learning difficulties

Many children have some degree of delayed development. It is important that they are monitored regularly to identify problems early and provide appropriate support. Some children may develop behavioural or psychological problems.

### Palate

Cleft palate or weakness of the palate is common and may lead to feeding difficulties and particularly to speech problems. Children will be referred to a plastic surgeon or ENT specialist and a speech therapist.

### Hearing, tonsils and throat problems

Some children have repeated ear infections and hearing impairment.

### Kidneys

Some children will have kidney abnormalities which can be detected on ultrasound scan.

### Immune system

Some children with 22q11 deletions are prone to infections because they have a problem with their immune system. Most children will grow out of this by their first birthday, but a few will continue to have problems. If the diagnosis is made when the child is older, a blood count to check the number of white cells in the blood, will be done.

### Endocrine problems

Some newborn babies will have low calcium levels and require supplements. It is very unusual to have problems later in childhood. Some children may have short stature and growth problems which may require referral to a growth/hormone specialist.

### Neurology

Occasionally children will have symptoms such as fits or problems with balance. Advice from a neurology specialist may be helpful in these cases.