

Childrens Services

22q11 deletion syndrome – parent information

This leaflet is for families where there is a suspected diagnosis of 22q11 deletion syndrome. It is intended to be an initial source of useful information.

What is 22q11 deletion?

We each have 23 pairs of chromosomes, with one of each pair coming from the mother and the other from the father. The chromosomes are numbered from 1 to 22 with the 23rd pair, called the sex chromosomes determining whether we are male or female.

In 22q11 deletion syndrome a tiny part is missing from chromosome 22, the q11 indicates exactly where this is.

The condition is also known as DiGeorge Syndrome, Velo-Cardio-Facial Syndrome (VCFS) or Shprintzen Syndrome. It carries multiple names because over the years several researchers with different areas of expertise have described it. Today the preferred term is 22q11, but the other names continue to be used. This is dependent upon which specialists are dealing with your child and what the presenting features were when the condition was first suspected.

Why did it happen?

In about 90 per cent of cases the cause is not known. The condition is a chromosomal abnormality due to a new mutation and there is no family history. The chromosome deletion was present in either the egg or sperm when your child was conceived and there is nothing you could have done to have caused or prevented it from happening.

In approximately 10 per cent of cases the deletion is inherited. An affected parent has a 1 in 2 (50 per cent) chance of passing on the condition, and this applies in each pregnancy. For unaffected parents the chance that they will have a further affected child is 1-2 per cent.

How common is it?

It is the most commonly occurring chromosome deletion. Current estimates as to its frequency vary. Some indicate that around 1 in 4,000 of the population may be affected by the condition (Children's Hospital of Philadelphia), others suggest it might be as many as 1 in 1,800 (Max Appeal, VCFS Educational Foundation).

These estimates differ as it is only recently that the condition has become more recognised and screening undertaken.

In East Anglia, there are currently about 60 children who have this diagnosis.

Despite much progress in its study and recognition, 22q11 is still not that well recognised. Sometimes there are no obvious symptoms or they are very mild, making diagnosis difficult and often resulting in delay. The condition may even go unnoticed for some time.

What are the features?

If you read about 22q11 deletion, you might be overwhelmed by the number of features that are described. You must remember this is a 'syndrome', which means that it is a collection of symptoms that have been seen in different patients. Not all of these problems occur in all children.

It is not possible to know how many of these features your child might have or to what extent they will be affected by them. There may be many problems or there may only be a few and these problems may be very mild or may be more severe. The spectrum is very wide.

The most frequently occurring features are:

- Heart problems
- Palate abnormalities that may be associated with feeding and speech
- Developmental delays and learning difficulties

Less frequently occurring features are:

- Immunity problems
- Hypocalcaemia, (an inability to metabolise calcium)
- Kidney abnormalities
- Leg pains

What happens now?

If you are reading this leaflet because your child is suspected of having 22q11, it is likely that his/her blood will have been taken to test for the deletion and confirm the diagnosis. You will have to wait several weeks for the results.

If it is confirmed that your child has 22q11, it is likely that as parents you will be asked if you want to be tested for the condition. At some point you may also be offered genetic counselling.

What help is available?

Addenbrooke's Hospital is one of the few hospitals that has a multidisciplinary 22q11 clinic with a number of health professionals who can assess your child and decide whether help in certain areas is required.

Max Appeal, a British Charity founded by parents of a child diagnosed with 22q11 also has a very comprehensive website you can access for further information and support.

www.maxappeal.org.uk

These are two further websites recommended by Max Appeal.

www.vcfsef.org

<http://www.chop.edu/consumer/jsp/division/generic.jsp?id=74654>



We are currently working towards a smoke-free site. Smoking is only permitted in the designated smoking areas.

For advice and support in quitting, contact your GP or the free NHS Stop Smoking helpline on 0800 169 0 169

Help with this leaflet:



If you would like this information in another language, large print or audio format, please ask the department to contact Patient Information: 01223 216032 or

patient.information@addenbrookes.nhs.uk



Document history

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