22q11 deletion DiGeorge syndrome

Information for children, families and carers

We have given you this factsheet because your child has been diagnosed with 22q11 deletion (also known as DiGeorge syndrome). It explains what 22q11 deletion is, what the common symptoms of the condition are and the outlook for your child.

We hope it will help to answer some of the questions you may have. If you have any further questions or concerns, please speak to a member of your child's healthcare team.

What is 22q11 deletion?

22q11 deletion (also known as DiGeorge syndrome) is a condition present from birth that can cause a wide range of health problems, including heart defects and learning difficulties.

What causes 22q11 deletion?

22q11 deletion is a genetic condition. It is caused when a small piece of genetic material is missing from a person's DNA (chromosome 22).

In most cases (90%), the condition is not passed on to a child by their parents. In these cases, the risk of future children also having this condition is very small.

How is 22q11 deletion diagnosed?

22q11 deletion is a common chromosomal disorder that can be tested for in pregnancy, either by:

- an amniocentesis (a diagnostic test which involves removing a small sample of amniotic fluid (the fluid which surrounds your baby) and testing it for certain chromosome differences)
- chorionic villus sampling (a diagnostic test which involves removing and testing a tiny sample of cells from your placenta)

A diagnosis can also be made after your child's birth with a blood test.

What are the symptoms?

22q11 deletion can cause a range of symptoms and the severity of these symptoms will vary for each child. Some children can be severely ill, but many others will grow up without realising they have the condition.

Some of the most common characteristics of the condition are:

- distinct facial features (low set ears, widely set eyes, a long face and a small lower jaw)
- congenital (from birth) heart disease
- kidney problems
- a cleft palate or lip (a gap in the top of the mouth or lip)
- feeding and gastrointestinal difficulties
- immune system deficiencies
- hearing loss
- low calcium and other endocrine issues (when the endocrine system, which produces
- hormones, does not function correctly)
- cognitive, developmental and speech delays
- behavioural, emotional and psychiatric differences, particularly bipolar disorder and schizophrenia.

How is 22q11 deletion treated?

There's currently no cure for 22q11 deletion. However, your child will be closely monitored as they grow up to check for and treat any problems that arise.

For example, your child may have:

- regular hearing tests, blood tests, heart scans and measurements of their height and weight
- an assessment of their development and learning ability before starting school (if your child has
- a learning disability, they may need extra support at a mainstream school, or they may benefit from attending a special school)
- speech therapy to help with speech problems and dietary changes
- physiotherapy for problems with strength and movement
- treatment from a podiatrist for foot and leg problems, and devices such as shoe inserts
- (orthoses) for leg pain
- surgery for more severe problems (for example, surgery to repair heart defects or an operation to repair a cleft palate)

What is the outlook for my child?

Everyone with 22q11 deletion is affected differently and it's difficult to predict how severe your child's condition will be. However, most children survive into adulthood.

As a person with 22q11 deletion gets older, some symptoms, such as heart and speech problems, tend to become less of an issue. Behavioural, learning, and mental health problems can continue to affect their daily life. Although the majority of people with 22q11 deletion do not develop schizophrenia or bipolar disorder, the risk for psychiatric illness is 25 times higher for people with 22q11 deletion than the general population.

Many people with 22q11 deletion who reach adulthood will have a relatively normal life span, but ongoing health problems can sometimes mean their life expectancy is a bit lower than usual. It's important that your child attends regular check-ups so that any problems can be found and treated early. Adults with 22q11 deletion are often able to live independently.

Contact us

If you have any quetions or concerns about your child's condition, please contact us.
Children's cardiac nurse specialist team
Telephone: **023 8120 4659**

Useful links

NHS UK
www.nhs.uk/conditions/digeorge-syndrome

The groups below may also be a good source of support:

Max Appeal
Website: **www.maxappeal.org.uk**

The 22q Family Foundation
Website: **www.22qfamilyfoundation.org**

Support for Disorders of Chromosome 22
Website: www.c22c.org

Text adapted from NHS UK www.nhs.uk/conditions/digeorge-syndrome/

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