The Leeds Teaching Hospitals NHS Trust

Di George syndrome (22q11 deletion)

Information for patients

Leeds Maternity Care

This leaflet is aimed at pregnant women with a confirmed diagnosis of Di George Syndrome in their baby. The leaflet should only be used to supplement a consultation with an experienced obstetrician.

What is Di George syndrome?

Di George syndrome is a condition that is present from birth that can cause a range of lifelong problems, including heart defects and learning difficulties. The severity of the condition varies. Some children can be severely ill and very occasionally may die from it, but many others may grow up without realising they have it.

Di George syndrome is caused by a problem with a person's genes called 22q11 deletion. This is where a small piece of genetic material is missing from a person's DNA.

In about 9 in 10 (90%) cases, the bit of DNA was missing from the egg or sperm that led to the pregnancy. This can happen by chance when sperm and eggs are made. It isn't as a result of anything you did before or during pregnancy. In these cases, there's usually no family history of Di George syndrome and the risk of it happening again to other children is very small. In around 1 in 10 (10%) cases, the 22q11 deletion is passed on to a child by a parent who has Di George syndrome, although they may not realise they have it if it's mild.

How is in diagnosed?

Di George syndrome is diagnosed antenatally by invasive testing. This is done by either a CVS or amniocentesis. These tests are normally performed because a structural problem has been found on anomaly scanning.

Sometimes it is diagnosed after birth by a blood test, which checks for the genetic fault.

What does it mean for your baby?

Di George syndrome can cause a range of problems, but most people won't have all of these.

Some of the most common issues are:

- **Learning and behavioural problems** - including delays in learning to walk or talk, learning disabilities and problems such as attention deficit hyperactivity disorder (ADHD) or autism
- **Speech and hearing problems** - including temporary hearing loss due to frequent ear infections, being slow to start talking and having a 'nasal-sounding' voice
- **Mouth and feeding problems** - including cleft lip or palate, difficulty feeding and sometimes bringing food back up through the nose
- **Heart problems**
- **Hormone problems** - an underactive parathyroid gland (hypoparathyroidism) is common and can lead to problems such as shaking (tremors) and seizures (fits)

Other possible problems include:

- Short stature
- A higher risk of picking up infections-such as ear infections, oral thrush and chest infections because the immune system is weaker than normal
- Bone, muscle and joint problems-including leg pains that keep coming back, an unusually curved spine (scoliosis) and rheumatoid arthritis
- Mental Health problems- adults are more likely to have problems such as schizophrenia and anxiety disorders

Treatment and outlook for Di George syndrome

There's currently no cure for Di George syndrome. Children and adults with the condition will be closely monitored to check for problems and these can be treated as they occur if needed.

For example, someone with the condition may have:

- Regular hearing tests, blood tests, heart scans and measurements of their height and weight
- An assessment of their development and learning abilities before starting school-if your child has a learning disability, they may need extra support at a mainstream school
- Speech therapy to help with speech problems and dietary changes to help with feeding difficulties
- Physiotherapy for problems with strength and movement
- Surgery for more severe problems, for example, surgery to repair heart defects or an operation to repair a cleft lip or palate

Everyone with Di George syndrome is affected differently and it's difficult to predict how severe the condition will be. Most children survive into adulthood. As someone with Di George syndrome gets older, some symptoms such as heart and speech problems tend to become less of an issue, but behavioural, learning and mental health problems can continue to affect daily life.

Many of those who reach adulthood will have a relatively normal life span, but on going health problems can sometimes mean life expectancy is a bit lower than normal. Adults with Di George syndrome are often able to live independently.

What are the chances of my next child having Di George syndrome?

If neither parent has Di George syndrome, the risk of having another child with it is thought to be less than 1 in 100 (1%). If one parent has the condition, they have a 1 in 2 (50%) chance of passing it onto their child; this applies to each pregnancy. Speak to your GP if you're planning a pregnancy and have a family history of Di George or you have a child with it. They may refer you for genetic counselling to talk to you about your risk and discuss your options. These may include:

- Having a blood test test to check if you and your partner carry the genetic problem that's causes Di George syndrome
- Having tests during pregnancy (CVS or amniocentesis) to check if your baby has the genetic problem that causes the condition-although this can't show how severely your child will be affected

- Pre-implantation genetic diagnosis- a type of IVF where eggs are fertilised in a laboratory and embryos are tested for problems before they are implanted in the womb (this isn't always available on the NHS).

What happens next?

You will have been referred to the Fetal Medicine Unit at Leeds. The Fetal Medicine consultant will discuss and agree the best plan for you and your family, according to your wishes.

You will have the option to continue with the pregnancy and receive the care outlines above or, to end the pregnancy (have a termination).

Where can I get more information & support?

Be sure to ask questions to the doctor supplying you with this leaflet and make a note of any questions you would like to ask at your Fetal Medicine Unit appointment. Your local hospital will also have a specialist midwife who you will be able to contact for further discussion.

Other useful sources of information:

Antenatal Results and Choices

- **www.arc-uk.org**

Supporting families affected by Di George Syndrome

- **www.maxappeal.org.uk**

Questions / Notes

What did you think of your care?

Scan the QR code or visit **bit.ly/nhsleedsfft**

Your views matter

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