The Leeds Teaching Hospitals NHS Trust

22q11 deletion (DiGeorge Syndrome)

General information

22q11d deletion also known as DiGeorge Syndrome, is a genetic disorder caused by a deletion/small missing segment of chromosome 22.

It is a common chromosomal disorder and can be tested for during pregnancy with either Chorionic Villus Sampling (CVS) or Amniocentesis. Otherwise a diagnosis can be made after birth. In the majority of cases there is no family history. Only 10% of cases are inherited from a parent.

22q11d has the potential to affect almost every system in the body and can cause a wide range of health problems.

Though not always present the key characteristics of this syndrome include combinations and varying degrees of:

- Distinct facial features. (Low set ears, widely set eyes, a relatively long face, Small teeth and lower jaw).
- Congenital Heart Disease.
- Kidney problems.
- Feeding and gastrointestinal difficulties.
- Immune system deficiencies.
- Hearing loss.
- Low calcium and other endocrine issues.
- Cognitive, developmental and speech delays.
- Behavioural, emotional and psychiatric differences (ADHD, ASD, Major Depression, Anxiety, Mood and Psychotic disorders such as Schizophrenia).

Not all children with 22q11d have the same problems, there is a lot of variability. Some children do not experience problems or have very mild symptoms whereas others can have moderate or severe physical, developmental and behavioural problems. This is not something that can predicted before birth.

There is a lot of support available to help children with 22q11d / DiGeorge Syndrome lead healthy and fulfilling lives, these include:

- Access to good healthcare – including a range of different specialists.
- Support for your child's development – including speech and language therapy and physiotherapy.
- Support groups – such as Max Appeal **www.maxappeal.org.uk**

What did you think of your care?

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

Your views matter

Developed by: Marie Wray Fetal/Children's Cardiac Nurse Specialist

Produced by: Medical Illustration Services • MID code: 20240626_014/EP

LN004925
Publication date
07/2024
Review date
07/2027