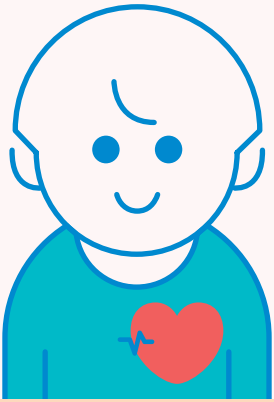
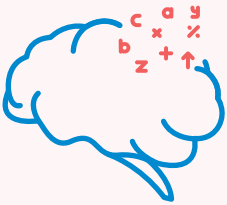
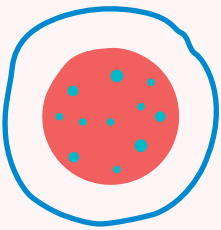
SingHealth DukeNUS
ACADEMIC MEDICAL CENTRE

Genomic
Medicine Centre

22q11.2 Deletion Syndrome







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What is 22q11.2 deletion syndrome?

22q11.2 deletion syndrome (22q11.2DS) is a genetic disorder that can affect many parts of the body, including the heart, immune system and development.

It may present at birth, in childhood or older. Common symptoms include congenital heart defects, palatal abnormalities, an impaired immune system, characteristic facial features and learning difficulties, but these vary widely.

How common is 22q11.2DS?

About **one in 6,000 people** are born with 22q11.2DS. Occurrences of 22q11.2DS are present across all racial groups and it affects both genders equally.

What are the symptoms of 22q11.2DS?

Although 22q11.2DS is a condition one is born with, some symptoms develop gradually over a number of years. The severity of symptoms varies from person to person.

The main features include:

Distinctive facial features. These may include an underdeveloped chin, hooded eyelids, wide-set eyes, low-set ears or a narrow groove in the upper lip.

Abnormal palate. A cleft palate (a gap in the roof of the mouth) and other palatal abnormalities that can cause difficulty in swallowing or producing certain sounds in speech.

Congenital heart defects. A number of heart defects are associated with 22q11.2DS, such as:

- A hole between the lower chambers of the heart (ventricular septal defect)
- Having only one large vessel, instead of two, leading out of the heart (truncus arteriosus)
- A combination of four abnormal heart structures (tetralogy of Fallot)

Weaker immune system function. This makes it difficult for the body to fight infections.

Hypoparathyroidism. The four parathyroid glands located in the neck regulate the calcium level in the body. Smaller-than-normal parathyroid glands can be seen in 22q11.2DS. This causes low levels of the parathyroid hormone resulting in low levels of calcium and high levels of phosphate in the blood.

Learning difficulties, and behavioural and mental health issues. Delays in speech development and learning difficulties can be seen. Attention-deficit/hyperactivity disorder (ADHD) or autism spectrum disorder may develop in some children. There is also a greater risk of depression, schizophrenia and anxiety disorders later in life.

Other medical concerns

The following concerns can also be seen in 22q11.2DS and need to be watched for and treated if necessary:

- Difficulty feeding
- Delayed growth or short stature
- Gastrointestinal anomalies
- Genitourinary tract anomalies including renal anomalies
- Impaired hearing and vision
- Middle ear infections
- Bone and muscle issues – hypotonia, scoliosis, arthritis
- Autoimmune disorders

The features of 22q11.2DS can be very different even amongst people with the condition in the same family.

Can 22q11.2DS be cured?

While it is not possible to cure 22q11.2DS, much can be done to make sure your child has the best possible outcome.

How is 22q11.2DS managed?

The multidisciplinary medical team looking after your child will be able to address specific medical concerns and routine 22q11.2DS-related healthcare issues. The team of specialists may consist of the following:

- Paediatrician
- Geneticist
- Immunologist
- Endocrinologist
- Plastic surgeon
- Psychiatrist
- Occupational therapist
- Cardiologist and/or cardiothoracic surgeon

- Developmental physician
- Speech therapist
- Physiotherapist
- Ear, nose and throat (ENT) specialist

Below are some related medical issues and how they can be managed:

Medical issue	Treatment
Developmental delay	Early intervention therapies
Hypocalcaemia/ hypoparathyroidism	Calcium and vitamin D supplementation
Heart defect	Medications and/or surgery
Cleft palate	Speech therapist, surgery
Anxiety, mood disorders, behavioural issues	Therapy, counselling and/or medication
Immune deficiency	Treatment of infections and/or irradiated blood products

The outlook for persons with 22q11.2DS varies depending on the severity of their congenital disabilities. Some of these conditions can be life-threatening.

However, with ongoing treatment and support, many people with 22q11.2DS live active, fulfilling lives.

What causes 22q11.2DS?

Most individuals with 22q11.2DS are missing a small part of chromosome 22 known as 22q11.2.

A human cell normally contains 46 chromosomes (23 from each parent). Chromosomes are thread-like structures found in every cell of the body, which carry hundreds of genes each. As a result of this deletion at chromosome 22q11.2, an estimated 40 to 60 genes are missing.

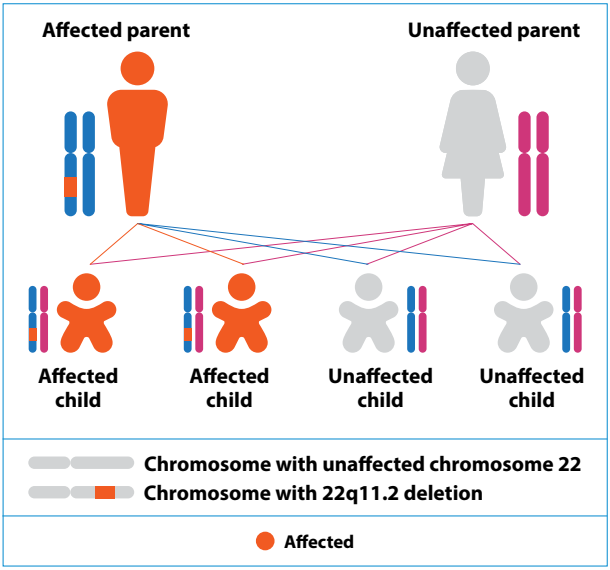
Other single-gene disorders and chromosome disorders can also cause features similar to those seen in 22q11.2DS.

How is 22q11.2DS diagnosed?

Testing for 22q11.2DS is done through a blood test – either a fluorescence in situ hybridisation (FISH) test or chromosomal microarray analysis (CMA).

Is 22q11.2DS inherited?

If your child is found to have 22q11.2DS, both parents can be tested to check if it is hereditary. In 90% of cases, neither parent has the chromosome 22q11.2 deletion.



Everyone has two copies of each chromosome in their body's cells, one copy from each parent. 22q11.2DS follows a **dominant inheritance pattern**. This means that having one chromosome 22 with the deletion can cause features of 22q11.2DS.

A parent with a chromosome 22q11.2 deletion has a 50% chance of passing it down to their offspring.

How likely will I have another child with 22q11.2DS if neither my spouse nor I have it?

In about 90% of cases, 22q11.2DS is the result of a spontaneous change (mutation) in the genetic material of the sperm or egg at conception. In such cases, the risk of having another child with 22q11.2DS is low but slightly greater than that of the general population (≤ 1-2%).

22q11.2DS is a lifelong condition. Should you require financial assistance or emotional support, please approach your doctor for referral to a medical social worker.

Support Group

Club Rainbow Singapore
Club Rainbow Singapore supports and empowers children with chronic illnesses and their families by providing relevant compassionate services in their journey.
Tel: 6377 1789
Email: contact@clubrainbow.org
www.clubrainbow.org

Acknowledgements
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