Could Congenital Heart Defects be Related to a Genetic Condition?

Information for patients and families

Reading this pamphlet can help you:

- Discover how congenital heart defects may be related to a genetic condition called 22q11.2 deletion syndrome
- Learn more about 22q11.2 deletion syndrome:
  - what it is
  - how best to care for it

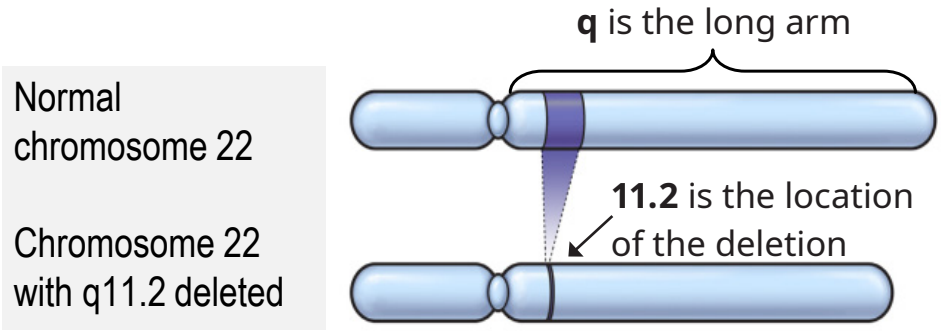
What causes congenital heart defects?

As part of your care, your doctor will try to find the cause of the heart defect you were born with. There are many possible causes of heart defects. Many are related to genetic conditions. One genetic condition that can affect the heart and other parts of the body is **22q11.2 deletion syndrome (22q11.2DS** or **22q)**. People with 22q are missing a tiny piece of one chromosome in each cell of their body. This can cause a wide range of health problems. **22q is the second most common cause of heart defects.**

What does 22q11.2 deletion syndrome mean?

22q11.2

- **22** refers to chromosome 22.
- Chromosome 22 is one of 23 pairs of chromosomes in every cell of your body.
- You have 2 copies of chromosome 22. One copy is inherited from each parent.
- **q** is the long arm of chromosome 22. Each chromosome has a short arm and a long arm.
- **11.2** is a specific location on chromosome 22, where a tiny piece is missing from one of the two copies.



Deletion

A tiny piece of a chromosome is missing or 'deleted'.

Syndrome

A health condition with many features.

What are some of the common features of 22q in adults?

- Learning difficulties and/or intellectual disability
- A nasal voice
- Anxiety or schizophrenia
- Congenital heart defects (heart defects present from birth)
- Low calcium levels
- Thyroid problems
- Seizures or epilepsy
- Movement disorders such as early-onset Parkinson's disease
- Having many medical conditions and needing to take many medications

22q is a condition that affects many areas of the body. People with 22q are affected in different ways, even if they are from the same family. The number and severity of features vary from person to person.

Many adults have not yet been diagnosed with 22q because of the wide range of possible symptoms. Also, medical professionals may not recognize the symptoms as 22q.

What causes 22q?

About 1 in 3000 people are born with 22q. Newborns are not tested for this syndrome on standard newborn screening.

Usually the 22q deletion occurs as a new genetic change in a family. It is NOT usually passed down (inherited) from a parent. Only about 1 out of 10 people with the 22q deletion have a parent who is also affected. In either case, nothing the parent did or did not do caused the deletion to happen.

How can I find out if I have 22q?

Your doctor may order a blood test to look for the 22q deletion if you have a congenital heart defect, especially if there is also another feature of 22q, such as:

- learning difficulties and/or intellectual disability
- nasal sounding voice
- anxiety or schizophrenia
- low calcium levels

If you have features of 22q, your doctor can also refer you to The Dalglish Family 22q Clinic to be diagnosed. **Genetic testing is recommended for everyone with intellectual and developmental disabilities.**

Knowing the diagnosis can benefit you in these ways:

- ✓ Your doctors will have more information when they take care of you.
- ✓ You may be able to better plan for health related issues before they happen.
- ✓ You can get the help that you need, such as social and/or financial support.
- ✓ You can have genetic counselling and get access to specific resources to learn about this diagnosis and the chances of having children who are also affected.

What happens if I have 22q?

Most adults with 22q manage well with support and health care tailored to their specific needs.

If you have 22q, your doctor can refer you to The Dalglish Family 22q Clinic, which is located at the Toronto General Hospital. This is a specialty clinic with a team of health care providers who are familiar with the syndrome. See page 6 for referral information.

Depending on your needs, you may have appointments with one or more of these health care providers:

- Genetics expert in 22q
- Psychiatrist
- Endocrinologist
- Neurologist
- Social Worker
- Registered Dietitian

Cardiologists from the **Toronto Congenital Cardiac Centre for Adults (TCCCA)** are an important part of the Dalglish Clinic team.

The health care providers involved in your care will:

- provide timely and effective care for 22q
- provide regular and careful monitoring of your health
- give you information and support
- connect you with local resources and peer support as needed
- help coordinate care
- provide recommendations to your local clinicians

Please call us if you have any questions:

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For more information, please visit our website: 22q.ca

For referral information, please visit the website:
http://22q.ca/medicalprofessionals/referral-information

Sources:

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