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Patient Education
Improving health through education

Could calcium and thyroid problems be related to a genetic condition?

UHN

Information for patients and families

Reading this pamphlet can help you:

- Discover how having low calcium levels and thyroid problems 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

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Toronto Rehab

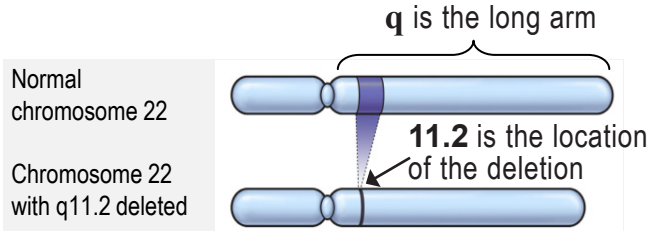
What causes calcium and thyroid problems?

As part of your care, your endocrinologist will be treating your calcium and/or thyroid problems. There are many possible causes of such problems. Some are related to genetic conditions. One genetic condition that can cause calcium and thyroid problems is **22q11.2 deletion syndrome** or **22q11.2DS**. People with 22q11.2DS are missing a tiny piece of one chromosome in each cell of their body. This can cause a wide range of health problems. A blood test can detect a 22q11.2 deletion.

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. Everyone has 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.



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

Syndrome A health condition with many features.

Please call us if you have any questions:

The Dalglish Family
Hearts and Minds Clinic
22q
for Adults with
22q11.2 Deletion Syndrome

The Dalglish Family Hearts and Minds Clinic
For Adults with 22q11.2 Deletion Syndrome
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For more information, please visit our website: **22q.ca**

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

Who will I meet at the Dalglish Clinic?

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

- **Genetics expert in 22q11.2DS**
- **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 22q11.2DS
- 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

What are some of the common features of 22q11.2DS?

- Hypocalcemia (low calcium levels)
- Thyroid problems
- Learning difficulties and/or intellectual and developmental disabilities
- Palate problems and a nasal voice
- Psychiatric issues such as anxiety and/or schizophrenia
- Congenital heart defects (heart defects present from birth)
- Seizures, epilepsy

22q11.2DS is a condition that affects many areas of the body. People with 22q11.2DS 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 22q11.2DS because of the wide range of possible symptoms. Also, many medical professionals may not recognize the symptoms as 22q11.2DS.

What causes 22q11.2DS?

About 1 in 2,000 people are born with 22q11.2DS. Newborns are not tested for this syndrome on standard newborn screening.

Usually the 22q11.2 deletion occurs as a new genetic change in a family. In other words, it is not usually passed down (inherited) from a parent. Only about 1 out of 10 people with the 22q11.2 deletion has 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 22q11.2DS?

Your doctor may order a blood test to look for the 22q11.2 deletion if your calcium level is low, and especially if there is also another feature of 22q11.2DS, such as:

- Learning difficulties and/or intellectual disability*
- Palate problems and a nasal sounding voice
- Congenital heart defects
- Psychiatric issues such as anxiety and/or schizophrenia

If you have features of 22q11.2DS, your doctor can also refer you to The Dalglish Family Hearts and Minds 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 and your doctors 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 22q11.2DS?

Most adults with 22q11.2DS manage well with support and health care tailored to their specific needs.

If you have 22q11.2DS, your doctor can refer you to The Dalglish Family Hearts and Minds 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.