

# Could schizophrenia be related to a genetic condition?



UHN

## Information for patients and families

Reading this pamphlet can help you:

- Discover how schizophrenia 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

Patient Education

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## What causes schizophrenia?

As part of your care, your psychiatrist will be treating your symptoms of schizophrenia. There are many possible causes of schizophrenia. Some are related to genetic conditions.

One genetic condition that can cause schizophrenia 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. **About 1 in every 100 people with schizophrenia has 22q11.2DS.**

## What does 22q11.2 Deletion Syndrome mean?

<p><b>22q11.2</b></p>	<ul style="list-style-type: none"> <li>• <b>22</b> refers to chromosome 22.</li> <li>• Chromosome 22 is one of 23 pairs of chromosomes in every cell of your body.</li> <li>• You have 2 copies of chromosome 22. One copy is inherited from each parent.</li> <li>• <b>q</b> is the long arm of chromosome 22. Each chromosome has a short arm and a long arm.</li> <li>• <b>11.2</b> is a specific location on chromosome 22, where a tiny piece is missing from one of the two copies.</li> </ul> <div data-bbox="532 1423 1185 1648" style="text-align: center;"> <p>Normal chromosome 22</p> <p>Chromosome 22 with q11.2 deleted</p> <p>q is the long arm</p> <p>11.2 is the location of the deletion</p> </div>
<p><b>Deletion</b></p>	<p>A tiny piece of a chromosome is missing or ‘deleted’.</p>
<p><b>Syndrome</b></p>	<p>A health condition with many features.</p>

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

- 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

**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, medical professionals may not recognize the symptoms as 22q11.2DS.**

## **What causes 22q11.2DS?**

About 1 in 2000 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. It is not usually passed down (inherited) from a parent. Only about 1 out of 10 people with the 22q11.2 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 22q11.2DS?**

Your doctor may order a blood test to look for the 22q11.2 deletion if you have schizophrenia, especially if there is also another feature of 22q11.2DS, such as:

- Learning difficulties and/or intellectual disability
- Nasal sounding voice
- Congenital heart defects
- Low calcium levels

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 an intellectual disability.**

### **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 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 7 for referral information..

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

**Please call us if you have any questions:**

The Dalglish Family  
Hearts and Minds Clinic



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/>**

**Source:**

Bassett A.S. et al. and the International 22q11.2 Deletion Syndrome Consortium. (2011).  
**Practical guidelines for managing patients with 22q11.2 deletion syndrome.**  
*Journal of Pediatrics*. 159(2): 332-339.e1. doi: 10.1016/j.jpeds.2011.02.039