22q11.2 Deletion Syndrome

Information for Educators and Members of School Resource Teams

Reading this pamphlet can help you:

- Learn more about 22q11.2 Deletion Syndrome (“22q”)
- Discover how students with 22q might present in the classroom
- Learn what educators can do to best support students with 22q
- Find out where students with 22q can get specialized help

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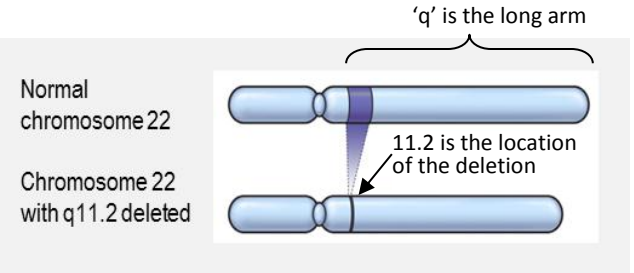
What is 22q?

Individuals with **22q11.2 Deletion Syndrome** or **22q** are missing a tiny piece of one chromosome (genetic material) in each cell of their body. This can cause a wide range of health problems that are highly variable in severity and number of body systems affected.

Most affected individuals experience some degree of intellectual and/or developmental disability, learning and social challenges, and speech and language difficulties.

22q11.2	'**22**' refers to chromosome 22. Chromosome 22 is one of 23 pairs of chromosomes in every cell of the body. Each person 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.

What causes 22q?

About 1 in 2000 people are born with 22q.

Most often, the 22q deletion occurs as a new genetic change in a family, and is not inherited from a parent.

About 1 in 10 people with the 22q deletion will have a parent who is also affected.

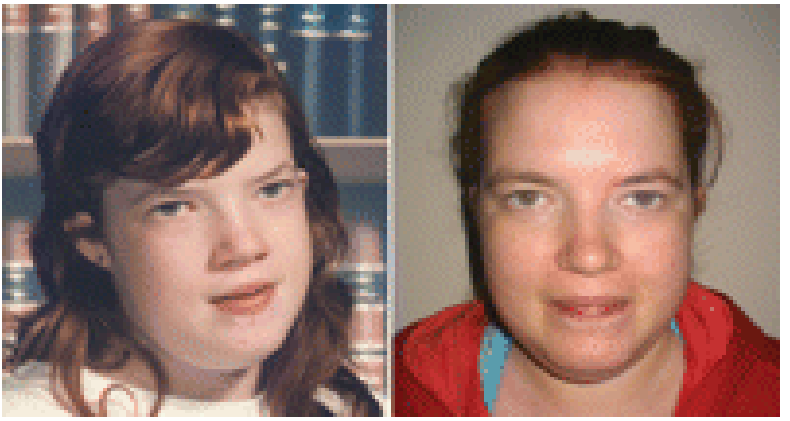
Previously, 22q has been known by many other names, including:

- DiGeorge syndrome
- Velo-cardio-facial syndrome (VCFS)

There are no known causes for the genetic change to occur in 22q. The region where the change occurs on chromosome 22 appears to be naturally susceptible to genetic changes.

22q can affect people of any ethnicity, origin, or family background.

Facial features of a woman with 22q as a child and as an adult



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Kapadia et al, (2008) *CMAJ;* 178 (4): 391-93

Common features of 22q:

Dozens of clinical features have been reported in people with 22q. Some of the more commonly observed features include:

Neurodevelopmental and mental health conditions

- Learning disabilities
- Mild intellectual disability
- Expressive language deficits
- Social deficits
- Mental health conditions such as anxiety, attention deficit disorder, autism spectrum disorder, schizophrenia

Medical conditions

- Palate abnormalities, leading to nasal-sounding speech
- Congenital heart defects, other birth defects
- Recurrent childhood infections
- Low calcium levels
- Thyroid disease
- Seizures/epilepsy

- Features differ between individuals; some have more, some less.

- By the time students with 22q are in school, they may have had several surgeries and hospitalizations due to their medical conditions and often require ongoing monitoring by multiple specialists. Most of these conditions are treatable.

- Speech therapy is a common intervention.

Many youths and adults do not get diagnosed with 22q because of the often subtle features that are difficult to recognize, even by medical professionals.

Strengths and learning challenges in 22q:

Many students with 22q will require some type of specialized educational services as they progress through the school system.

Common areas of relative strength:

- Rote verbal learning and verbal recognition memory
- Reading; decoding and spelling
- Better experiential than auditory learners
- Concrete, sequential learning styles
- Computers/visuals

These relative strengths may mask a student's need for identification for services until later grades when there is a greater focus on developing higher level thinking skills.

Common challenges:

- **Mathematics/arithmetic;** often notably poorer than verbal skills
- Executive functioning; working memory
- Reading comprehension and abstract reasoning
- Attention
- Social and communication abilities

Medical and mental health conditions do not predict learning abilities or disabilities.

Strengths and learning challenges in 22q:

Functioning can vary greatly between individuals, even those with similar full-scale IQ scores.

- Each student with 22q should undergo a full psychoeducational assessment in order to understand his or her specific needs.
- The majority of individuals with 22q have an overall intellectual level that falls in the **borderline** IQ range.
- About one-third of individuals fall in the **mild** intellectual disability range.
- Some individuals may have a moderate to severe disability and some may have intellect in the average range.
- Psychoeducational profiles of children sometimes show significantly higher verbal IQ than performance IQ, **suggestive of a nonverbal learning disorder**. This is a less common profile in older children and adolescents.
- A subset of students with 22q will experience a cognitive decline or plateau around ages 11-13. Repeat assessments are helpful in order to better plan for the transitions from elementary to secondary school.

Repeat assessments after turning 18 years old are important in order to monitor changes, guide educational and vocational options, and to assess eligibility for government financial assistance programs for adults.

Social skills of individuals with 22q:

Students with 22q tend to have significant difficulties in these areas:

- Tending to be literal thinkers and interpret words verbatim.
- Becoming anxious and stressed with changes in routine or structure.
- Speech and language deficits, making it difficult to communicate with peers.
- Social skills acquisition – ability to share, express needs, take turns, etc.
- Determining social norms and responding appropriately to others.
- Frequent target of bullying.
- Poor social judgment leading to increased vulnerability.

Important considerations for students with 22q:

Feature	Suggested Strategies
Delays in functional development Increased frustration and chronic stress with academic activities that are beyond the student's capabilities. Increased risk of the student being taken advantage of by peers.	• Provide accommodations and modifications unique to the student's needs, as for other students requiring additional support (e.g. keyboarding and use of assistive software). • Create a structured environment with clear rules to follow. • Adapt expectations to match the cognitive and social capabilities and limitations of the student.
Decreased learning time due to various medical and specialist appointments.	• Provide extra, one-on-one support while at school, if possible. • Encourage the use of workbooks (e.g. "Jump-Math"), take home books and activities while away.
Executive functioning deficits	• Use visual aids, written reminders and key word cues. • Give concrete examples and use repetition. • Give students one (or maybe two) small tasks rather than multiple directions at once. • Provide templates for complex problems in a clear, concise breakdown of steps.

Important considerations for students with 22q:

Feature	Suggested Strategies
Employability and skills necessary for activities of daily living appear to be areas of relative strength for individuals with 22q.	• **Co-op programs** are often a good way for students to learn useful skills and obtain course credits.
Mental health symptoms including anxiety and other psychiatric illnesses may begin to present during adolescence or earlier. Anxiety disorders, attention deficit disorder and sometimes features similar to autism may be apparent at younger ages. School refusal is a particularly concerning sign.	• Notify caregivers and members of the school resource team involved in the student's care of significant behaviour changes as soon as possible. • Developing a system to document changes in thinking, feelings and behaviour may be helpful. • Work collaboratively with caregivers and associated health professionals, when possible.
Medications or other treatments due to medical and psychiatric conditions can have both beneficial and adverse effects.	• Documentation of changes in behaviour and functioning, for the better or worse, with treatments will be helpful for the family and associated health professionals.
Sensitivity to caffeine as it is a contributory factor to anxiety, irritability and/or tremors.	• Encourage reducing caffeine intake, especially cola, "energy" drinks, and coffee.
Increased need for sleep, which can help reduce irritability and improve learning and daily functioning.	• Encourage regular, early bedtime and more hours of sleep than other same-aged individuals.

Adapted from Table III in Bassett et al, (2011) *J Pediatr*; 159(2): 332–9

How might a student with 22q present in the classroom?

Behavioural characteristics that may be seen in students with 22q (regardless of cognitive functioning) include:

- Quiet, shy, socially isolated and/or awkward.
- Dependent, eager to please.
- Seeming younger than their chronological age.
- Perseverative behaviours (e.g. repetitive speech/questions, difficulty “letting go”).
- Poor self-regulation, but often not particularly disruptive students.
- Behaviours similar to those described on the autism spectrum.
- Anxieties (e.g., to be on time, excessive worrying about upcoming events, extra distress with any changes, public speaking, taking tests in a room with others).

What can the educational team do if there is a suspicion of 22q in a student?

- Discuss the specific concerns with members of the school resource team involved in the student's care.

- If a parent inquires about some of these issues, they can be directed to their family doctor or paediatrician who can conduct a more detailed examination and assessment.

If a diagnosis of 22q is suspected, a doctor can order a standard blood test to look for the 22q deletion. The test is called a chromosomal microarray and is covered by OHIP.

A confirmed diagnosis may allow for targeted medical monitoring, preventative measures and appropriate referrals and supports.

Knowing the diagnosis can benefit the student, their family, and educators:

- ✓ Having an explanation of the student's struggles can help to provide a sense of relief and better understanding of expectations and prognosis.

- ✓ Genetic counselling and accessibility to specific resources to learn more about the condition.

- ✓ Accurate information helps the school to provide the best possible supports.

- ✓ Health care professionals and educators will be better able to plan for physical and mental health related issues before they happen, and employ preventative strategies.

- ✓ Appropriate academic, social and financial support can be provided to the student.

- ✓ Future planning, (e.g. exploring resources, post-secondary programs and vocational options) for the student can help ease stress and anxiety and help the student reach their full potential.

On a case by case basis, members of our team can be made available for consultation with members of your school resource team.

Where can students with a diagnosis of 22q get specialized help?

Students **under the age of 17** can be referred to the **SickKids 22q Deletion Syndrome Clinic for multidisciplinary care and support by a team of paediatric professionals.**

Students **aged 17 and older** can be referred to **The Dalglish Family Hearts and Minds Clinic**, which is located at Toronto General Hospital. This is a specialty clinic with a team of health care professionals who are experts in 22q.

Visit the SickKids' and Dalglish Clinics' websites for more information.

Both the paediatric and adult clinics offer multidisciplinary care from a team of health care professionals. These may include:

- **Genetics expert in 22q**
- **Cardiologist**
- **Speech language pathologist**
- **Psychiatrist**
- **Social worker**
- **Registered dietitian**
- **Endocrinologist**
- **Immunologist**

The health care providers involved will:

- Provide timely and effective care for students with 22q
- Provide regular and careful monitoring of their health.
- Connect them with local resources and peer support as needed.
- Help co-ordinate care.

Sources, references and some helpful resources

Publications

- 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.

- Butcher NJ et al. (2012). **Functional outcomes of adults with 22q11.2 deletion syndrome.** Genetics in Medicine. 14:836–843.

- Costain, G et al. (2011). **Caregiver and adult patient perspectives on the importance of a diagnosis of 22q11.2 deletion syndrome.** Journal of Intellectual Disability Research. 56(6): 641-651.

- De Smedt, B. et al. (2009). **Mathematical learning disabilities in children with 22q11.2 deletion syndrome: a review.** Developmental disabilities research reviews. 15(1): 4-10.

- Gerdes, M. et al. (2001). **Taking advantage of early diagnosis: Preschool children with 22q11.2 deletion.** Genetics in Medicine. 3(1), 40-44.

- Kapadia, R. K et al. (2008) **Recognizing a common genetic syndrome: 22q11.2 deletion syndrome.** CMAJ. 178(4): 391-393.

- Swillen, A. & McDonald-McGinn D. (2015). **Developmental trajectories in 22q11.2 deletion.** Am J Med Genet Part C 9999: 1-10. doi: 10.1002/ajmg.c.31435.

Websites

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

- 22q Clinic at The Hospital for Sick Children - http://www.sickkids.ca/cgenetics/what-we-do/22q-deletion-syndrome-clinic/index.html

- The International 22q11.2 Foundation Inc. - http://www.22q.org/

- The Dempster Family Foundation - http://dempsterfamilyfoundation.org/
  - http://dempsterfamilyfoundation.org/sites/default/files/22q-for-teachers.pdf

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The Dalglish Family
Hearts and Minds Clinic

22q

for Adults with
22q11.2 Deletion Syndrome

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

http://www.sickkids.ca/CGenetics/What-we-do/22q-deletion-syndrome-clinic/

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