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22q11.2 deletion syndrome (22q): Help for people with an under-recognized condition

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In this issue:

What is 22q?

- 22q is a common cause of developmental delays/intellectual disabilities.
- 22q can affect almost every part of the body. Some features are obvious at birth, while others develop later in life. Some people with 22q have a mild form; other presentations can be more severe.
- Many people with 22q live for years with multiple health and other issues without knowing the underlying diagnosis. Their "diagnostic odyssey" is related to lack of awareness about 22q in the health care and social service communities and in the general public.

How common is 22q?

- The **most common** microdeletion syndrome in humans
- Estimated to affect about **one in every 3000** live births
- **The second most common genetic cause of intellectual disability after Down syndrome**

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What's in the Name?	
22	Chromosome 22 (a small chromosome in every cell in the body)
q	The long arm of the chromosome
11.2	The location on the chromosome (like GPS coordinates)
Deletion	A piece missing from one of the pair of chromosomes 22
Syndrome	A collection of features

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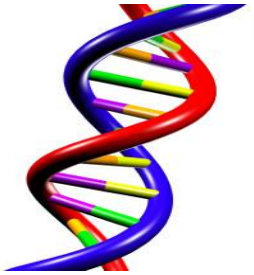
Did you know…

- Genetic testing using a modern method like microarray is recommended for every person with an intellectual disability who does not already have a molecular diagnosis.
- These tests are covered by health insurance plans, and available to any family doctor or medical practitioner.
- Genetic counselling is an important part of testing which means you always have the chance to learn and ask questions about genetic test results.



How do you know if someone has 22q?

The features of 22q are variable from person to person.
Here are some of the more common signs and symptoms:

- Learning difficulties (most common), developmental delay and/or intellectual disabilities (ID)
- Birth defects such as congenital heart disease, palate abnormalities causing nasal speech, scoliosis and others
- Psychiatric issues such as anxiety disorders (most common) or schizophrenia
- Neurological issues such as seizures, epilepsy or movement disorders (e.g. Parkinson's disease)
- Endocrine (hormonal) conditions such as thyroid disorders or low calcium levels

What makes 22q different from other causes of ID?

- One in four people with 22q develop schizophrenia.
- One in two to three has a form of congenital heart defect that needs ongoing care.
- On average, adults with 22q have 9 medical features requiring care and attention, so they may be considered “complex care patients.”
- Fertility is unaffected, but parenting can be a challenge.
- There is a great deal of variability in the presentation of 22q; features can also change over time
- 22q can be a “life-limiting” condition.
- 22q is usually a new genetic change in a family but can be inherited in some cases.

How can you help people with 22q?

Although there is no “cure” for the 22q deletion, each separate condition can be managed with appropriate treatment and support. Each associated condition can affect the individual’s level of functioning. With effective, long-term management of chronic conditions along with individualized, appropriate social supports, and access to suitable programs and services, individuals with 22q can be successful and enjoy a happy, fulfilled life.

1) Get educated about 22q and learn some effective strategies for communication.
2) Advocate for appropriate services and supports, and assist in finding meaningful employment / daily activities.
3) Involve family members and other support staff in collaborative care plans.

How can you help people with 22q?

A molecular diagnosis of 22q can be very helpful to the patient, the family and caregivers. Once the “Diagnostic Odyssey” is resolved and you know the person has 22q, you can…

1) Get educated about 22q and learn some effective strategies for communication.

- Explore available 22q resources and learn about the person’s specific conditions and needs.

Some ways to improve communication with someone who has 22q:

- Use simple, concrete instructions; give one or two instructions/directions at a time.
- Avoid using analogies, probabilities or statistics.
- Clarify understanding—individuals may say, “Yes” or “I understand” though they do not.
- Praise successes and achievements; encourage positive behaviour changes.
- Focus on the present situation (anticipatory anxiety, worrying ahead of time, is common in 22q).
- Written/pictorial communication, e.g., with modern technology, can be helpful.

2) Advocate for appropriate services, supports, and employment / daily activities.

- People with 22q usually function at their best with a structured daily routine.
- People with 22q typically have relative strengths in daily living skills and in rote memory. High level problem-solving is often stressful and challenging so needs extra support.
- People with 22q may be well-suited to structured, hands-on tasks where they can gain mastery.
- Assist with developing and implementing schedules and ensuring essential appointments are attended.
- Provide reminders.

3) Involve family members and other support staff in collaborative care plans.

- Foster a collaborative care approach.
- Assist with directing to appropriate resources for future planning purposes (e.g., lawyers familiar with special needs and disability estate planning).
- Multiple medical and psychiatric conditions often result in the need for multiple medications. People with 22q often do best when medications are provided in blister packs or dossette cases.
- Some level of supervision or oversight to ensure things stay on track is usually essential.

Multiple medical specialists, allied health professionals, support staff and other are often involved in an attempt to "put the puzzle pieces together" for undiagnosed patients.

The Dalglish Family 22q Clinic
Toronto General Hospital

- The world's first Clinic specializing in the care of adults with 22q and their families.
- The Clinic provides multidisciplinary patient-centred care for Canadian patients. Our staff includes 22q experts, psychiatrists, a social worker and a dietitian. Cardiologists from the Toronto Congenital Cardiac Centre for Adults (TCCCA) are an important part of our Clinic team. We also work closely with other health care specialists including those from neurology, endocrinology, ENT, etc.
- For Canadian patients, the Clinic does initial consultations, as well as ongoing monitoring and follow-up care for patients with 22q who have multiple physical and mental health conditions.
- The Clinic provides peer support and welcomes the opportunity for caregiver support groups.
- The Clinic's goal is to improve the quality of life for people with 22q and their families. World-leading research involves over 300 adult patients and their families. There are also multiple educational initiatives for patients and their families, health care professionals, support staff, students at all levels, and for the general public.

For Ontario patients:

If you are working with a person with 22q or suspect that one of your clients may have 22q, you can refer them to the Dalglish Family 22q Clinic at Toronto General Hospital. Please call (416-340-5145) or visit our website, www.22q.ca

The Dalglish Family 22q Clinic

In summary

22q is a multisystem condition that is an under-recognized cause of developmental delay and intellectual disability. Although specific support needs must be tailored according to the individual's needs and interests, some commonalities with 22q are helpful to be aware of. Being informed of the multiple features of 22q will help you meet the individual's physical and psychosocial needs.

Resources

- The Dalglish Family 22q Clinic, **www.22q.ca**
- 22q Fact Sheets from the 22q deletion syndrome Clinic at **SickKids** Hospital
- The International 22q11.2 Foundation, *22q11.2* **www.22q.org**

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Answers to FAQ's about the journal

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