22q = 22q11.2 deletion syndrome

- Is prevalent
- Affects 1 in every 2148 newborns
- **Multisystem**
- Has a significant burden of illness
- **Variable presentation**, so **under-recognized**
- The most common identifiable cause of **schizophrenia** (~1 in 100) and **congenital heart defects** (~1 in 8 with tetralogy of Fallot)
- Former names include:
  - **DiGeorge** syndrome
  - **Velocardiofacial** syndrome

Common Features of 22q

- Multiple **medical** & **psychiatric** issues at a relatively young age
- Medical complexity, multimorbidity
- **Learning difficulties** and/or intellectual/developmental disabilities
- **Psychiatric** issues such as **anxiety** and/or **schizophrenia**
- **Hypocalcemia**
- **Hypothyroidism**
- Seizures, epilepsy
- Congenital **heart** defect
- Palate problems, e.g., nasal voice
- Scoliosis

Genetic Testing for 22q

Genomic SNP microarray (OHIP covered)

- Complete (available at www.22q.ca):
  - **Genomic SNP microarray requisition form** from the Hospital for Sick Children (HSC)
- Ask your patient to **bring the two completed forms** to a lab operated by Dynacare or Life Labs to have his/her blood drawn **Monday to Wednesday**
- The lab will deliver the blood sample to HSC for testing.
- Questions? Please email 22q@uhn.ca

Referral Criteria

- Adults 17+ years with **confirmed 22q**
- Adults with **multiple features of 22q**
  - We can facilitate genetic diagnosis

Making a Referral

Referral form at: **www.22q.ca**

Or in **Epic** in UHN:

Create an encounter and go "Add order" at the bottom. Select "Outpatient referral to Centre for Mental Health" and click "22q" as the primary reason for referral.

Possible accompanying documents

- **Genetic** test results
- Psychiatric history
- Cardiac history & test results
- Endocrine issues & blood work
- **Intellectual functioning** assessments
- Any other relevant test results

How Our Clinic helps

The adult population of 22q is **increasing**, but healthcare professionals are often **unaware** of this genetic condition.

The Dalglish Family 22q Clinic offers a "**one-stop-shop**", providing anticipatory care from healthcare experts across many specialties.

We **coordinate long term specialized care** through our Clinic and work together with professionals in the patient's local community.

Our clinical team offers **social work services**, **peer and family support groups**, and reinforcement of genetic counselling.

We strive to **reduce barriers** for youth with 22q transitioning to adult healthcare.

We look forward to assisting you with the **long-term management of your patients** with a diagnosis of 22q. For additional information, or to download our referral form, please visit our website: **22q.ca**.

Resources

Updated clinical practice recommendations for managing adults with 22q11.2 deletion syndrome

Clinically relevant genetic considerations for patients with tetralogy of Fallot

The Dalglish Family 22q Clinic

We are the first specialty clinic in the world to focus on adults with 22q11.2 deletion syndrome (22q11.2DS or 22q).

We provide specialized, **patient-centred care** to adults and their families.

We **educate** patients, families, trainees and health care providers.

We perform world class **research** to improve the quality of life for individuals and families affected by the syndrome.

Our team

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

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Clinic Director

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Do you have adult patients with

22q

in your practice?

UHN Toronto General Hospital