

## 22q = 22q11.2 deletion syndrome

- **22q11.2 deletion syndrome**
- **Multisystem** disorder with a significant burden of illness
- **Under-recognized** due to **variable presentation**
- The most common identifiable cause of **schizophrenia & serious congenital heart defects**
  - About 1 in every 100 people with schizophrenia has 22q
  - About 1 in 8 with tetralogy of Fallot have 22q
- Former names include:
  - **DiGeorge** syndrome
  - **Velocardiofacial** syndrome

*Each patient is affected differently*

## Common Features of 22q

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

## Genetic Testing for 22q

### Genomic SNP microarray (OHIP covered)

- Complete (available on [www.22q.ca](http://www.22q.ca)):
  - **OHIP requisition form** AND
  - **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](mailto: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](http://www.22q.ca)

## 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 easy-to-use referral form, please visit our website: [22q.ca](http://22q.ca).

## 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
- Cardiologist
- Endocrinologist
- Neurologist
- Social Worker
- Registered Dietitian



### The Dalglish Family 22q Clinic

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CANADA

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Do you have  
adult patients  
with

# 22q

in your practice?

