"22q" (22q11.2 Deletion Syndrome)

What is in the name?

"22"	Chromosome 22 (the smallest of the human chromosomes, in every cell of the body)
"q"	The long arm of the chromosome
"11.2"	The position on the chromosome - like the GPS coordinates
"Deletion"	A piece missing from one of the pair of chromosomes 22
"Syndrome"	A collection of features, formerly known as DiGeorge Syndrome or Velo-cardio-facial syndrome (VCFS)

What is it?

A genetic condition people are born with, but one that is usually not inherited
A genetic condition that has many features, some that are obvious at birth, and some that develop later in life
The most common microdeletion syndrome – a type of genetic condition we have known about only for about 20 years

How common is it?

More common than cystic fibrosis or Huntington's disease
About one in every 2,000 babies are born with 22q – boys and girls, from every community
The most common cause of "blue babies" – one in every 8 babies with tetralogy of Fallot has 22q
The second most common genetic cause of intellectual disabilities

How serious is it?

It can cause stillbirths, or babies to die; adults may have early mortality in middle age
It is often disabling
It can cause serious physical and mental health problems

How does it affect families?

Families often search for many years for an answer – the "diagnostic odyssey" – for multiple health problems
Surgeries for babies and children, speech and learning difficulties in school, physical and mental health problems
Multiple appointments, often lack of coordinated care, and uncertainties about work, relationships, long term outcome

Why have we never heard of it?

The piece missing on chromosome 22 is too small to be seen with the standard chromosome test in use for 60 years
Testing only became available in the mid-1990s and not all doctors are familiar with these newer genetic tests
The features and their severity vary from person to person so doctors may not recognize it
The many previous names for 22q can cause confusion (DiGeorge, velocardiofacial, conotruncal anomaly face...)

How important is it?

It is a major cause of heart defects and other birth defects
It is the only testable cause of schizophrenia
It can cause seizures, curvatures of the spine, thyroid problems, low calcium, speech and swallowing problems, early onset Parkinson's disease, hearing problems, anxiety disorders, and other treatable conditions
22q may need "the whole hospital" - potentially every kind of medical specialist and health care clinician
It can affect people from any ethnic or social background

How can we treat it?

Most of the individual features of 22q are treatable – and for some, such as low calcium, prevention is possible
We can monitor for and recognize problems early – and improve outcomes

What does the Dalglish Family Hearts and Minds Clinic at the Toronto General Hospital offer?

The first clinic in the world devoted to adults with 22q
Coordinated care across multiple specialties, a "one-stop-shop" for people with 22q and their caregivers
Transition for youths "graduating" from paediatric care (e.g., at the 22q Deletion Syndrome Clinic, at SickKids)
Telehealth services and peer support groups
Guidance in finding available resources that fit the needs of the individual
Excellence in education about 22q
World leading research to increase our knowledge about 22q, and unlock its secrets to benefit the general population

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