The International 22q11.2 Foundation Inc.

Does it run in the family:

The 22q11.2 deletion is most often a "de novo" event, meaning that it is not inherited from either parent and does not usually run in a family. Only 10% of children with the deletion have a parent who is also affected.

The 22q11.2 duplication runs in families more often with a de novo rate of 30%.

- For parents who are not affected, the chance that a future child will be affected is very low.
- For parents who are affected, there is a 50% chance of passing it on per pregnancy.

There is nothing that parents do or fail to do that causes the deletion or duplication.

Looking forward: Treatments & therapies

Although there is no cure for the 22q11.2 deletion/duplication syndrome, therapies and medical interventions are available to address associated symptoms. The earlier that symptoms are detected, the more doctors can do to help. Once diagnosed, evaluation is recommended (but not limited to) in the following areas:

- Audiology
- Cardiology
- Child development & psychology
- Cleft palate
- ENT (Ear, nose & throat)
- Endocrinology
- Feeding / nutrition
- Genetics
- Immunology
- Neurology
- Orthopaedics
- Urology

Living with 22q – What next?

Most children and adults with the 22q11.2 deletion/duplication do very well both medically and within their families and communities.

As with anything unexpected, coming to terms the diagnosis is often difficult but gets easier as support, resources and information become available. There are plenty of opportunities for families and individuals to meet and / or network with others through:

- Diagnosis-specific websites
- Family meetings and picnics
- Support networks
- Children's diagnosis-specific camps
- 22q at the Zoo-Worldwide Awareness Day

To learn more about support opportunities, please visit www.22q.org

One Syndrome with many names:

As a result of how our understanding of the 22q11.2 deletion evolved, several different names are used for what we now know to be the same condition. Older terms include:

- DiGeorge Syndrome (DGS)
- Velo-Cardio-Facial Syndrome (VCFS)
- Conotruncal anomaly face syndrome (CTAF)
- Optiz G/BBB syndrome
- Cayler cardiofacial syndrome

Genetically speaking, there is no detectable difference in the microdeletions found in people with VCFS and those with DGS or the other related syndromes. Individuals with these diagnoses have the same underlying condition. Rather than further dividing our small 22q community, the Foundation aims to bring everyone together educating both the public and professionals about the deletion's many names through our Same Name Campaign.

The Syndrome

A syndrome is defined as a recognizable pattern of physical and behavioural features. The 22q11.2 deletion/ duplication syndrome is caused by a missing section or duplication of chromosome 22, a chromosome that is present from birth and generally found in every body cell. It has the potential to affect almost every system in the body and cause a wide range of health problems.

More common than you think

- Almost as common as Down syndrome, a widely-recognized chromosomal disorder
- 1 out of every 2,000 live births
- 1 in 68 children with congenital heart disease
- 5 to 8 percent of children born with cleft palate

No two people manifest the syndrome in exactly the same way. In fact, individuals within the same family who have the syndrome may or may not be similarly affected. For example, a mother could have very mild manifestations while her child may have a severe heart defect requiring surgery immediately after birth.

Though not always present, the key characteristics include combinations and varying degrees of:

- Heart defects
- Feeding & gastrointestinal difficulties
- Immune system deficits
- Growth delay
- Palate differences
- Kidney problems
- Hearing loss
- Low calcium & other endocrine issues
- Cognitive & speech delay
- Behavioural, emotional & psychiatric differences (ADHD, autism, anxiety, etc.)

To read information in Spanish, please see “Recursos”

22qColombia