What is 22q?

One Condition, Many Names

1/2000

22q11.2 deletion syndrome occurs in an estimated 1/2000 births which makes it almost as common as Down syndrome.

So why haven't you heard of it?

Over the last few decades, this syndrome has had many different names, but all have the same underlying cause.

- DiGeorge Syndrome (DGS)
- Velo-Cardio-Facial Syndrome (VCFS)
- Opitz G/BBB Syndrome
- Conotruncal Anomaly Face Syndrome (CTAF)
- Cayler Cardiofacial Syndrome

22q11.2 Deletion Syndrome

We now know that this genetic condition is caused by a small, missing or "deleted" piece of the 22nd chromosome, and that missing piece can affect every system in the human body.

Heart

75% of individuals with the 22q11.2 deletion have mild to life-threatening heart defects.

Palate and GI System

Differences in the palate, such as a cleft (hole in the roof of the mouth) or nasal speech are very common as are feeding and GI (gastrointestinal) problems.

Immune System

Many individuals have immune system problems leading to trouble with infections or vaccines.

Thyroid and Endocrine System

Low calcium levels and low growth-hormone levels may be present and are treatable.

Kidney

1/3 of people with 22q have renal system differences such as a missing kidney.

Behavior

Learning and behavioral differences, such as ADHD, anxiety, and other mental health issues are also diagnosed in some children and adults with the 22q11.2 deletion.

Inheritance

7% of 22q11.2 deletions are inherited from a parent with the deletion. Once the deletion is present there is a 50% recurrence risk.

The 22q11.2 deletion syndrome can cause many differences, ranging from mild to serious, making detection complex.

Early detection of 22q can lead to earlier interventions and better outcomes for affected individuals.

The International 22q11.2 Foundation Inc.

The mission of the International 22q11.2 Foundation is to improve the quality of life for individuals affected by the 22q11.2 syndromes through family and professional partnerships.

www.22q.org

Sources

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