22q11.2 Deletion Syndrome

22q deletion syndrome has been called by many names:

- **DiGeorge Syndrome**
- **Velocardiofacial Syndrome**
- **Conotruncal Anomaly Face Syndrome**
- **Autosomal Dominant Opitz G/BBB Syndrome**
- **Cayler Cardiofacial Syndrome**
- **Shprintzen Syndrome**

Molecular genetic research has revealed that all syndromes listed above have one common link…. there is a small amount of genetic material missing, termed a microdeletion, on the long arm (referred to as the q arm) of chromosome 22.

1. ***What is 22q11.2 Deletion Syndrome?***

   It is a condition caused by a loss of genetic material on chromosome 22.

2. ***Sign and Symptoms of 22q11.2 Deletion Syndrome?***

   There are a variety of physical and behavioral disorders that have been linked to 22q11.2 deletion syndrome. The syndrome can impact every system in the body and can lead to a wide-range of health issues. Some of the most common problems associated with 22q11.2 include:

   - **Heart defects**
   - **Feeding difficulties**
   - **Gastrointestinal problems**
   - **Immune system problems**
   - **Wounds that don't heal properly**
   - **Growth problems**
   - **Delayed development in speaking, rolling over, sitting up or walking**
   - **Cleft Palate**
   - **Low Calcium levels**
   - **Behavioral, emotional, and psychiatric differences that may cause ADHD, autism, anxiety and more.**

3. ***Can 22q11.2 deletion syndrome be treated?***

   Since 22q11.2 deletion syndrome has the ability to affect every system of the body, it is important that affected children are treated by a team of pediatric specialists who can identify the variety of physical and psychosocial needs these patients may have. Although there is no cure for the 22q11.2 deletion, many therapies and medical interventions

are available to help address its symptoms. The earlier these symptoms are detected, the more that can be done to help. That's why, when diagnosed with this condition, evaluation is recommended in some or all of the following areas:

- **Audiology**
- **Cardiology**
- **Developmental Pediatrics**
- **Cleft Lip and Palate Center**
- **Resonance Disorders Program**
- **Speech and Language Pathology**
- **ENT Services**
- **Endocrinology**
- **Interdisciplinary**
- **Genetics**
- **Immunology**
- **Hematology**
- **Neurology**
- **Neuropsychology**
- **Behavioral Health**
- **Orthopedics**
- **Urology**

4. Can 22q11.2 deletion syndrome be inherited?

The 22q deletion is caused by a missing piece of chromosome 22. A parent with 22q has a 50% chance of passing it on to his or her offspring. However, only 10% of people "inherit" 22q from a parent. The remaining 90% develop 22q as a new mutation, meaning the missing piece of chromosome 22 happens spontaneously. Parents who have a child with this "de novo" form of 22q do not have a higher risk of subsequent children being affected.

5. How common is 22q11.2deletion syndrome?

The 22q deletion is thought to affect 1 in every 4,000 people. This number, however, will likely rise as awareness of the condition and its many presentations come to light.

Chromosome 22q11.2 deletion is almost as common as Trisomy 21, also known as Down syndrome, which is a more widely recognized chromosomal disorder.

Resources:

- http://www.nationwidechildrens.org/22q11-deletion-syndrome
- http://ghr.nlm.nih.gov/condition/22q112-deletion-syndrome
- https://en.wikipedia.org/wiki/DiGeorge_syndrome
- http://www.22q.org/
- http://www.chop.edu/conditions-diseases/chromosome-22q112-deletion#.VaUhYflVhBc
- http://www.mayoclinic.org/diseases-conditions/digeorge-syndrome/basics/definition/con-20031464