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What is 22q11.2 Deletion Syndrome?

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The 22q11.2 deletion syndrome is a condition caused by a missing piece of genetic material on chromosome 22 and is present from the time of conception. The 22q11.2 deletion is almost as common as Down syndrome and is present in 1 of every 4,000 live births; in 1 in 68 children with congenital heart disease; and in 5 to 8 percent of children born with cleft palate. This chromosome deletion has the potential to affect almost every system in the body and can cause a wide range of health problems. No two people are ever exactly alike, and this syndrome doesn't affect any two people the same way.

Several different names describe 22q11.2: DiGeorge syndrome (DGS), velo-cardio-facial syndrome (VCFS), conotruncal anomaly face syndrome (CTAF), Opitz G/BBB syndrome, and Cayler cardiofacial syndrome. There are no detectable differences in the deletions of genetic material found in people with VCFS versus those with DGS or other related syndromes. Individuals with these diagnoses all have the same underlying condition of 22q11.2 deletion syndrome.

Some key characteristics of this syndrome include combinations and varying degrees of the following conditions.

- **heart defects**
- **feeding and gastrointestinal difficulties**
- **immune system deficits**
- **growth delay**
- **palate differences**
- **kidney problems**
- **hearing loss**
- **low calcium and other endocrine issues**
- **delays in cognitive language development and speech**
- **behavioral, emotional, and psychiatric differences** (ADHD, autism, anxiety, etc.)

Only about 10% of children with the 22q11.2 deletion have a parent affected by the syndrome. In the majority of individuals, this chromosome deletion happens sporadically without a previous family history, which suggests that 22q11.2 deletion is most often a "de novo" event – **meaning one does not inherit the syndrome** from either parent, and it does not usually "run"in a family.

Therapies for Children with 22q11.2 Chromosome Deletion include the following:

Speech Therapy – Speech therapists (SLPs) begin working with 22q11.2 children at age one, (due to associations with delayed emergence of language), by using alternative communication strategies such as sign language. The SLP also helps children born with a cleft lip or palate with articulation and pronunciation.

Occupational Therapy – Occupational therapists (OTs) work with the small muscle groups used for tying,

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handwriting, buttoning, self-feeding, etc. The OT also addresses problems associated with feeding, swallowing, texture, and other problems related to the refusal of foods.

Physical Therapy – Physical therapists work with the large muscle groups and developmental skills in an effort to meet motor milestones. Play therapy is a big part in early intervening skills.

Most children and adults with the 22q11.2 deletion do quite well. Coming to terms with this diagnosis is difficult at first but becomes easier as more information becomes available. Entire families can benefit from meeting other children and adults with 22q11.2 deletion or speaking with them through diagnosis-specific internet sites. By contacting support networks and groups, families can find lots of information regarding conferences and meetings, family picnics,camps specifically designed for children with 22q11.2 deletion, as well as information about the recent findings of research. The greatest support offered by these networks is that families realize they are not alone in their struggle to cope with this syndrome.

For more information regarding 22q11.2 deletion syndrome, contact The International 22q11.2 Deletion Syndrome Foundation, Inc. - PO Box 532 http://www.22q.org - Matawan, NJ 07747 – USA or request information by email at info@22q.org

Resources:

22q The International 22q11.2 Deletion Syndrome Foundation, Inc. Retrieved April 2012 http://www.22q.org/index.php

22q The International 22q11.2 Deletion Syndrome Foundation, Inc. Frequently asked questions about 22q11.2. http://www.22q.org/index.php/what-is-22q/frequently-asked-questions. Retrieved April 2012.