

DiGeorge Syndrome

Definition:

- DiGeorge syndrome is a birth defect that is caused by an abnormality in chromosome 22 which affects the immune system.

Description:

- DiGeorge Syndrome is also known as: 22q 11.2 deletion syndrome, Velocardiofacial Syndrome, and Strong Syndrome
- It is a congenital developmental anomaly characterized by abnormalities of the immune system and congenital heart defects
- The immune system deficiencies are caused by the failure of the thymus gland to develop
- Common conditions include heart defects, effects on facial appearance, and lack of or underdeveloped thymus and parathyroid glands

Characteristics:

- Parathyroid glands (which regulate blood Ca) fail to develop
- Results from abnormality in the development of the 3rd and 4th pharyngeal pouches of the embryo (which forms aorta)
- Infants may exhibit grayish/purple discoloration caused by abnormal amounts of low hemoglobin blood due to cardiac abnormalities
- Congenital heart defects
- Seizures can occur as a result of hypocalcemia
- Failure to Thrive can result and most of these infants will die within a month from severe cardiac abnormalities or infections
- Infants must be isolated once immune deficiency is identified
- Survivors face increased susceptibility to respiratory infections
- Uneven eyes
- Eye slits slanted toward nose
- Uprturned nose
- Palatal abnormalities (such as cleft lip and/or palate)
- Hearing loss or abnormal ear exams (Outer ears may be small, low-set or rotated slightly backward, and malformed)
- Severe immunologic dysfunction (an immune system which does not work properly due to abnormal T -cells, causing frequent infections)
- Psychiatric disorders in adults (e.g., schizophrenia, bipolar disorder)
- Microcephaly (small head)
- Mild to moderate mental retardation
- Genitourinary anomalies (absent or malformed kidney)

Causes (known and speculated):

- In most cases, a deletion in the upper portion of the long arm of chromosome 22 is found
- Believed to be caused by faulty development of the cephalic neural crest
- In about 10% of cases, the disorder is inherited from a parent
- The gene is autosomal dominant which means the parent has a 50% chance of passing down this disorder
- Women who take Accutane once a day in the treatment of severe cystic acne have offspring with features that resemble the DiGeorge syndrome
- Exposure to alcohol in utero may also cause similar features

- May occur in infants of diabetic mothers

Implications for speech and language:

- Delayed development of speech and language skills
- Hypernasal speech (excessively nasal speech) due to velopharyngeal dysfunction (a condition in which the soft palate is unable to consistently elevate to contact the back of the throat; this prevents a speaker from closing off the oral cavity from the nasal cavity during speech)
- Articulation disorders (difficulty producing speech sounds)
- Voice disorders and laryngeal anomalies (e.g., breathy voice, vocal fold paralysis)
- Language impairment (e.g., slow vocabulary growth, difficulties formulating complex sentences)
- Pragmatic and social skills difficulties

Diagnosing this disorder:

(Note: Speech-language pathologists do not diagnose this condition.)

The diagnosis of the DiGeorge Syndrome is usually made on the basis of signs and symptoms that are present at birth or develop soon after birth. Some children may have the facial features that are characteristic of the DiGeorge Syndrome. Affected children may also show signs of low blood calcium levels as a result of their hypoparathyroidism. This may show up as low blood calcium on a routine blood test, or the infant may be “jittery” or have seizures (convulsions) as a result of the low calcium. Affected children may also show signs and symptoms of a heart defect. The severity of heart disease is usually the most important determining factor.

Some children have signs or symptoms at birth or while they are still in the hospital nursery. Others may not show signs or symptoms until they are a few weeks or months older. Some children and adults are diagnosed at a much older age due to speech delay, qualitative speech problems, or feeding problems.

Physical

Coordination and balance issues will begin to emerge after a child is able to walk independently so be consistent with reinforcing your child’s physical skills. Since gross motor skills are more obviously impaired than fine motor skills, encourage your child to develop these skills early.

Language

Children with DiGeorge Syndrome are often very slow in acquiring language skills and most children are nonverbal prior to age 2. Receptive language abilities, such as comprehension, are generally stronger than expressive language abilities. Articulation errors are commonly present in children, so encourage your child to speak as much as possible and help your child enunciate sounds accurately. The speech impairments exhibited are more severe during the younger ages and show a trend of gradual improvement as the child matures.

Cognitive

It is difficult to make broad generalizations about cognitive functioning because children with DiGeorge Syndrome have a tremendous range of neuropsychological abilities. Academically, mathematics is generally a more difficult area whereas reading and spelling abilities are relatively stronger. To enhance your child’s ability to think logically and abstractly, practice simple math skills with your child, such as counting, so that you can avoid these difficulties in the future.

Resources:

Books for Children

- Brown, T. (1995). *Someone Special, Just Like You*. New York: Henry Holt and Co.
- Murrell, D. (2001). *Tobin Learns to Make Friends*. Arlington, TX: Future Horizons.
- Woloson, E. (2003). *My Friend Isabelle*. Bethesda, MD: Woodbine House.

Books for Parents

- Berkow, Robert. (1999). *The Merck Manual of Diagnosis and Therapy*. Rahway, NJ Merck Research Laboratories.
- Gomella, T. (1994). *Neonatology: Management, Procedures, On-Call Problems, Diseases and Drugs*. Norwalk, CT: Appleton & Lange.
- McDonald-McGinne, D. (1999). *22q11 Deletion Syndrome*. Philadelphia: The Children's Hospital of Philadelphia.

Websites

- Immune Deficiency Foundation: www.primaryimmune.org
- The International 22q11.2 Deletion Syndrome Foundation: <http://www.22g.org>
- VCFS Educational Foundation: <http://www.vcfsef.org/index.html>

Support Groups

- Chromosome Deletion Outreach, Inc. (888) 236-6680.
- International DiGeorgeNCF Support Network (607) 753'-1250.
- VCF Group – Suzanne Manthei, Austin, TX, Email: manthei4@juno.com

References:

National Center for Biotechnology Information (US). Genes and Disease [Internet]. Bethesda (MD): National Center for Biotechnology Information (US); 1998-. Retrieved from: <http://www.ncbi.nlm.nih.gov/books/NBK22179/>

Information About the 22q11.2 Deletion, Velocardiofacial Syndrome and DiGeorge Syndrome. Upper Midwest VCFS/DGS Community. Retrieved from: http://www.vcfconnect.org/index.php?option=com_content&task=view&id=12&Itemid=27

DiGeorge Syndrome. Immune Deficiency Foundation. Retrieved from: <http://primaryimmune.org/about-primary-immunodeficiency-diseases/types-of-pidd/digeorge-syndrome>