DiGeorge/Velocardiofacial Syndrome

Symptoms

Wide-set eyes

Low-set ears, with notched ear fold

Small jaw

Narrow, short groove in the upper lip

Cleft palate

Recurrent infections (chronic runny nose or multiple bouts of pneumonia, oral thrush, diaper rash or diarrhea)

Cramp-like spasms of the baby's hands and fingers, or twitches in the baby's face, throat or arms

Developmental delaysmost often speech delay

Slow mental development

Lack of appetite/poor weight gain/failure to thrive

(Revised 2012

About the Disorder

DiGeorge syndrome is a disorder present at birth (congenital) caused by problems within a developing embryo that occur due to chromosome defects. It may also be identified as as velocardiofacial syndrome.

Possible complications associated with DiGeorge syndrome are related to the specific medical problems caused by the chromosome defect.

If the parathyroid glands aren't fully developed (hypoparathyroidism), the child may experience stunted growth (short stature), slow mental development (or mental retardation), or cataracts.

If the thymus gland is underdeveloped or absent, severe infection may occur due to a lack of infection-fighting T cells.

Heart defects can result in severe complications including: congestive heart failure, enlargement of the heart (cardiomegaly), high blood pressure in the lungs (pulmonary hypertension), endocarditis or stroke.

Treatment

There is no cure for DiGeorge/Velocardiofacial syndrome, and the long-term outlook for children with this syndrome varies widely, depending on the organs affected. Treatment focuses on correcting or managing the medical problems caused by the chromosome defect. and speech are typically normal, functional mobility is realistic, and individuals are typically able to live productive, independent lives.

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Educational Implications

Developmental delay is a common manifestation in children with VCFS, but it is variable with some children showing almost no early problems and others being quite delayed. The delay in motor milestones has many contributors, including the generalized hypotonia (low muscle tone) that is common in VCFS. Also contributing are factors such as congenital heart disease, multiple hospitalizations and operations, chronic illness, and other factors that can restrict a child's activity. By

school age, the majority of children with VCFS tend to perform

close to the normal range in terms of motors skills, although

they may always be a little more hypotonic or even "clutzier"

than other kids. Speech and language milestones tend to be slightly more delayed, with the average age of onset of the first word at 19 months, but again this is very variable. It would seem that children with VCFS have their own developmental profile that differs from that of other children. In general, most show significant "catch-up" before their fourth birthday.

To date, essentially all individuals with VCFS have shown some type of learning disorder. Although mental retardation can occur in VCFS, the diagnosis of mental retardation is reserved for a very small percentage of cases. Difficulty with abstraction and problem solving tends to interfere most with mathematics and reading comprehension. Concepts are difficult to grasp, even things as seemingly basic as "opposite" or "reverse." Therefore, learning is best done by repetition and breaking things down into the smallest concrete units.

Instructional Strategies & Classroom Accommodations

- Monitor academic, cognitive and speech/language skills, particularly as they related to abstract learning,
- As the child ages, closely monitor student ability with regard to abstract learning, generalized application of newly learned skills, problem solving and organizational thought processes
- Provide support in areas of organizational and independent work skills as needed

Note: Document all accommodations on the student's IEP

Resources

Mayo Clinic
http://www.mayoclinic.com/health/digeorge-syndrome/DS00998

March of Dimes Birth Defects Foundation
1275 Mamaroneck Ave
White Plains, NY 10605
(888)663-4637
Askus@marchofdimes.com
http://www.marchofdimes.com

Velo-Cardio-Facial Syndrome Educational Foundation, Inc.
P.O. Box 874**,** Milltown, NJ 08850
(866) 823-7335

Immune Deficiency Foundation
40 West Chesapeake Ave, Suite 308
Towson, MD 21230
(800)296-4433
idf@primaryimmune.org
http://www.primaryimmune.org

NIH/National Institute on Deafness and Other Communication Disorders (Balance) National Temporal Bone, Hearing and Balance Pathology Resource Registry
Massachusetts Eye & Ear Infirmary
243 Charles Street
Boston, MA 02114-3096
(800) 822-1327
TBRegistry@meei.harvard.edu
http://www.tbregistry.org

NIH/National Institute of Child Health and Human Development
31 Center Dr Building 31
Room 2A32MSC2425
Bethesda, MD 20892
(301) 496-5133
http://www.nih.gov/hichd/

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