DiGeorge

BIRTH DEFECT RESEARCH FOR CHILDREN

What is DiGeorge Syndrome?

DiGeorge Syndrome (DGS), also referred to as Velo-Cardio-Facial Syndrome (VCFS), is an immunodeficiency disorder characterized by various congenital abnormalities. Proper functioning of the immune system relies on the thymus gland. In DGS, the thymus and parathyroid glands are either not fully developed or completely absent. The parathyroid glands are responsible for regulating calcium levels in the blood. The symptoms of DGS depend on the extent to which these glands are missing.

DiGeorge Syndrome

Some individuals with DiGeorge Syndrome are nearly asymptomatic while others are affected more severely. DGS can have up to 180 different symptoms, many of which are minor and seen throughout the general population. The most common symptoms are recurrent infections, hypocalcemia (low blood calcium), heart defects, and palate abnormalities. Some of the characteristic facial features are hypertelorism (wide-set eyes), down-slanting eyes, low-set auricles (portion of the ears), prominent nose with squared nasal root, and micrognathia (small size of the lower jaw). Children with DGS can be uninhibited and impulsive, yet they are often very affectionate and able to function socially.

How many children have DiGeorge Syndrome?

DGS is a rare disorder affecting males and females equally and occurring in one of every 3,000-4,000 births.

What causes DiGeorge Syndrome?

Ninety percent of individuals with DGS are missing a piece of genetic information on chromosome 22 at the q11 region, referred to as a deletion on chromosome 22. Most of the 22q11 deletions are new occurrences or sporadic. About 6-10% of the deletions are inherited. The gene is autosomal dominant, meaning each child born to a person with the gene has a 50% chance of receiving the gene and manifesting the syndrome.

The remaining 10% of individuals with DGS do not have a deletion in the chromosome 22q11 region. These cases have been associated with fetal alcohol syndrome, maternal diabetes, prenatal exposure to Accutane, and other chromosome defects.

How can you help your child with DiGeorge Syndrome?

It is nearly impossible to predict which symptoms a child will develop. Cardiac problems may be treated surgically and speech difficulties with therapy. Cosmetic surgery is available to correct facial abnormalities. Hypocalcemia is treated through the use of calcium supplements and 1,25-cholecalciferol. Combating the loss of immune system T-cells is more challenging, though some children have benefited from a thymus transplant. If a baby has palate problems, he or she may have difficulty eating large quantities at one time and may not gain weight sufficiently. Smaller, more frequent feedings may help; however, the child will probably always be small for his/her age. Children with DGS usually fall between the second and twenty-fifth percentile in size. Learning disabilities may become apparent when a child with DiGeorge enters school. Often the child will need some type of special class during the early years of school, but eventually special classes may no longer be necessary. Some people with DGS have become college graduates.

Can DiGeorge Syndrome Be Prevented?

Preventing DiGeorge Syndrome is difficult since it is often a sporadic occurrence. However, parents can be screened for the 22q11 deletion to see if they are carriers of DGS. Fetal alcohol syndrome and prenatal exposure to Accutane have been associated with DGS, so as always, women should avoid alcohol consumption and Accutane use during pregnancy.

Fact Sheet by:

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