Disability Awareness

DiGeorge Syndrome

DiGeorge syndrome, also called 22q11.2 deletion syndrome, is a disorder caused by a defect in chromosome 22. It results in the poor development of several body systems.

Medical problems commonly associated with DiGeorge syndrome include heart defects, poor immune system function, a cleft palate, complications related to low levels of calcium in the blood, and delayed development with behavioral and emotional problems.

The number and severity of symptoms associated with DiGeorge syndrome vary greatly. However, almost everyone with DiGeorge syndrome needs treatment from specialists in a variety of fields.

Signs and symptoms of DiGeorge syndrome can vary significantly in type and severity, depending on what body systems are affected and how severe the defects are. Some signs and symptoms may be apparent at birth, but others may not appear until later in infancy or early childhood.

Signs and symptoms may include some combination of the following:

- Bluish skin due to poor circulation of oxygen-rich blood (cyanosis) as a result of a heart defect
- Breathing problems
- Twitching or spasms around the mouth, hands, arms or throat
- Frequent infections
- Certain facial features, such as an underdeveloped chin, low-set ears, wide-set eyes or a narrow groove in the upper lip
- A gap in the roof of the mouth (cleft palate) or other problems with the palate
- Delayed growth
- Difficulty feeding and gastrointestinal problems
- Failure to gain weight
- Poor muscle tone
- Delayed development, such as delays in rolling over, sitting up or other infant milestones
- Delayed speech development
- Learning delays or difficulties and behavior problems

DiGeorge syndrome is caused by the deletion of a portion of chromosome 22. Each person has two copies of chromosome 22, one inherited from each parent.

If a person has DiGeorge syndrome, one copy of chromosome 22 is missing a segment that includes an estimated 30 to 40 genes. Many of these genes haven't been clearly identified and aren't well-understood. The region of chromosome 22 that's deleted in DiGeorge syndrome is known as 22q11.2.

The deletion of genes from chromosome 22 usually occurs as a random event in the father's sperm or in the mother's egg, or it may occur early during fetal development. Rarely, the deletion is an inherited condition passed to a child from a parent who also has deletions in chromosome 22 but may or may not have symptoms.

The portions of chromosome 22 deleted in DiGeorge syndrome play a role in the development of a number of body systems. As a result, the disorder can cause several errors during fetal development. Common problems that occur with DiGeorge syndrome include:

Heart defects. DiGeorge syndrome often causes heart defects that result in an insufficient supply of oxygen-rich blood. These defects may include a hole between the lower chambers of the heart (ventricular septal defect); only one large vessel, rather than two vessels, leading out of the heart (truncus arteriosus); or a combination of four abnormal heart structures (tetralogy of Fallot).

Hypoparathyroidism. The four parathyroid glands in your neck regulate the levels of calcium and phosphorus in your body. DiGeorge syndrome can cause smaller than normal parathyroid glands that secrete too little parathyroid hormone (PTH), leading to a condition called hypoparathyroidism. This condition results in low levels of calcium and high levels of phosphorus in your blood.

(cont. on page 34)

Groups & Activities

GLADD Dances. 3rd Friday of each month, 5:30-7:30 pm, September-June. So. Park Comm. Ctr., 4851 So. Tacoma Way. $4 suggested donation per person. Music & snacks provided. For individuals 18 & over. Call Geri for more info. 253-223-4800. If you need special care or supervision, please bring support.

FrienDS Down Syndrome Play Group
Meets on Saturdays. Contact Susan Jackson at PAVE/Parent to Parent Program for more information. 253-565-2266 x107.

Community Inclusion Program. Activities are held at various locations around the county. Three Pierce County areas monthly. For more information or a schedule of events, please contact PAVE at 253-565-2266 or visit www.communityinclusionprogram.org.

Companionship Program. Please contact Companionship Program at 253-798-2997 or email PCCompanionship@co.pierce.wa.us for program schedule.

(cont. from page 32)

Thymus gland dysfunction. The thymus gland, located beneath the breastbone, is where T cells — a type of white blood cell — mature. Mature T cells are needed to help fight infections. In children with DiGeorge syndrome, the thymus gland may be small or missing, resulting in poor immune function and frequent, severe infections.

Cleft palate. A common condition of DiGeorge syndrome is a cleft palate — an opening (cleft) in the roof of the mouth (palate) — with or without a cleft lip. Other, less visible abnormalities of the palate that may also be present can make it difficult to swallow or produce certain sounds in speech.

Distinct facial features. A number of particular facial features may be present in some people with DiGeorge syndrome. These may include small, low-set ears, wide-set eyes, hooded eyes, a relatively long face, or a short or flattened groove in the upper lip.

Learning, behavioral and mental health problems. The 22q11.2 deletion may cause problems with the development and function of the brain, resulting in learning, social, developmental or behavioral problems. Delays in toddler speech development and learning difficulties are common. A number of children with DiGeorge syndrome develop attention-deficit/hyperactivity disorder (ADHD) or autism spectrum disorder. Later in life people with DiGeorge syndrome are at increased risk of depression, anxiety disorders, schizophrenia and other mental health disorders.

Autoimmune disorders. People who had poor immune function as children, due to a small or missing thymus, may have an increased risk of autoimmune disorders, such as rheumatoid arthritis or Graves' disease.

A large number of medical conditions may be associated with DiGeorge syndrome, such as hearing impairment, poor vision, poor kidney function, and relatively short stature for one's family.

A diagnosis of DiGeorge syndrome is based primarily on a lab test that can detect the deletion in chromosome 22. Your doctor will likely order such a test if your child has:

A combination of medical problems or conditions suggesting DiGeorge syndrome

A heart defect, because certain heart defects are commonly associated with DiGeorge syndrome

In some cases, a child may have a combination of conditions that suggest DiGeorge syndrome, but the lab test doesn't indicate a deletion in chromosome 22. Although these cases present a diagnostic challenge, the coordination of care to address all of the medical, developmental or behavioral problems will likely be similar.

Although there is no cure for DiGeorge syndrome, treatments can usually correct critical problems, such as a heart defect or low calcium levels. Other health issues and developmental, mental health or behavioral problems can be addressed or monitored as needed.

Source:
http://www.mayoclinic.org/diseases-conditions/digeorge-syndrome/basics/definition/con-20031464