Disability Awareness: Velo-Cardio-Facial Syndrome

Excerpted from: http://www.vcfsef.org/about_vcfs/general_information.html

What is Velo-cardio-facial syndrome (VCFS)?
Velo-cardio-facial syndrome (VCFS) is a genetic condition that is related to DiGeorge syndrome and involves a similar chromosome abnormality as DiGeorge syndrome. Common conditions include certain heart defects, effects on facial appearance, and lack of or underdeveloped thymus and parathyroid glands. DiGeorge syndrome describes the same clinical features as Velo-cardio-facial syndrome, but an individual must have immune system deficiencies associated with lack of a thymus gland to be considered to have true DiGeorge syndrome.

What causes Velo-cardio-facial syndrome?
90 percent of patients with the features of this syndrome are missing a small part of their chromosome 22 at the q11 region. This region encompasses about 30 individual genes and results in developmental defects in specific structures throughout the body. It is not known why this region of chromosome 22 is prone to become deleted, but this is one of the most frequent chromosome defects in newborns. Deletion 22q11.2 is estimated to occur in one in 3,000 to 4,000 live births. Most of the 22q11.2 deletion cases are new occurrences or sporadic (occurs by chance). However, in about 10 percent of families, the deletion is inherited and other family members are affected or at risk for passing this deletion to their children. The gene is autosomal dominant, therefore, any person who has this deletion has a 50 percent chance of passing the deletion to a child. For this reason, whenever a deletion is diagnosed, both parents are offered the opportunity to have their blood studied to look for this deletion. Approximately 10 percent of individuals who have the features Velo-cardio-facial syndrome (VCFS) do not have a deletion in the chromosome 22q11 region. Other chromosome defects have been associated with these features, as have maternal diabetes, fetal alcohol syndrome, and prenatal exposure to Accutane® (a medication for cystic acne).

What are the features of Velo-cardio-facial syndrome?
The following are the most common features of Velo-cardio-facial syndrome. However, not every child will have every feature of the syndrome and the severity of the features will vary between children. Features may include:

- palatal abnormalities (such as cleft lip and/or palate)
- feeding difficulties
- conotruncal heart defects (e.g., tetralogy of Fallot, interrupted aortic arch, ventricular septal defects, vascular rings)
- hearing loss or abnormal ear exams
- genitourinary anomalies (absent or malformed kidney)
- hypocalcemia (low blood calcium levels)
- microcephaly (small head)
- mental retardation (usually borderline to mild)
- IQs are generally in the 70 to 90 range
- psychiatric disorders in adults (e.g., schizophrenia, bipolar disorder)
- severe immunologic dysfunction (an immune system which does not work properly due to abnormal T-cells, causing frequent infections), these patients have true DiGeorge syndrome.

Facial features of children with Velo-cardio-facial syndrome may include the following:

- small ears with squared upper ear
- hooded eyelids
- cleft lip and/or palate
- asymmetric crying facies
- small mouth, chin, and side areas of the nose tip

How is Velo-cardio-facial syndrome diagnosed?
In addition to a prenatal history, complete medical and family history, and a physical examination, diagnostic procedures for Velo-cardio-facial may include:

- blood tests and tests to examine for immune system problems
- x-ray - a diagnostic test which uses invisible electromagnetic energy beams to produce images of internal tissues, bones, and organs onto film.
- echocardiography - a procedure that evaluates the structure and function of the heart by using sound waves recorded on an electronic sensor that produce a moving picture of the heart and heart valves.
- fluorescence in situ hybridization (FISH) studies - when features of conotruncal heart defects, clefting, other facial features, hypocalcemia, and absent thymus are identified, a blood test is usually ordered to look for a deletion in the chromosome 22q11.2 region. FISH is specifically designed to look for small groups of genes that are deleted. If the FISH test finds no deletion in the 22q11.2 region and the features of VCFS are still strongly suggestive, then a full chromosome study is usually performed to look for other chromosome defects that have been associated with this syndrome.

If a 22q11.2 deletion is detected in a child, then both parents are offered the FISH test to see if this deletion is inherited. In approximately 10 percent of families, the deletion has been inherited from one of the parents. Any individual who has this 22q11.2 deletion has a 50 percent chance, with each pregnancy, of passing it on to a child.