

# Velocardiofacial Syndrome

## What is velocardiofacial syndrome (VCFS)?

Velocardiofacial Syndrome (VCFS) is a genetic syndrome that can affect speech, hearing, language and learning. It can also cause certain physical characteristics and some medical conditions. VCFS is a genetic syndrome that is also known as Shprintzen syndrome, DiGeorge syndrome, or 22q11.2 syndrome.

## What causes velocardiofacial syndrome?

Velocardiofacial syndrome is caused by deletion of genes on chromosome 22q11.2. VCFS can occur for the first time in a family for unknown reasons. However, each individual with the diagnosis of VCFS has a 50% chance of passing on this syndrome to each of his or her children. In other words, half of the affected individual's children are likely to also have the syndrome.

## What are the characteristics of velocardiofacial syndrome?

The basic characteristics of velocardiofacial syndrome involved the soft palate (also called "velum" and thus "velo"), heart ("cardio") and facial features.

- **Velo:** A condition called "velopharyngeal dysfunction (insufficiency or incompetence)" can cause hypernasal speech. There may also be a cleft of the soft palate or a submucous (under the skin surface) cleft.
- **Cardio:** The individual with VCFS may have a history of a heart murmur or minor cardiac anomalies. These may include ventriculoseptal deviation (VSD), atrial septal defect (ASD), or patent ductus arteriosus (PDA). There may also be some vascular anomalies, such as pulmonary stenosis, tetralogy of Fallot, right sided aortic arch, displaced internal carotid arteries, and abnormal retinal arteries.
- **Facial:** There are often facial characteristics, such as narrow eye openings, a long face, a bulbous nasal tip, a small jaw, or minor ear anomalies.
- **Learning and Cognitive Problems:** Some individuals with VCFS have learning disabilities, or mild to moderate mental retardation.
- **Communication Problems:** Hypernasality and abnormal voice quality is common with VCFS. In addition, there may be misarticulations (often due to a motor speech disorder called "apraxia"), language disorder, and hearing loss.
- **Other Common Physical and Medical Characteristics:** Some individuals with VCFS have long slender digits, short stature (usually below the 10%ile), and hernias.
- **Other Common Functional Problems:** VCFS can cause early feeding problems, gross and fine motor delays, social disinhibition, and some psychiatric problems in adolescence.

Although there are many characteristics of this syndrome, there is a great deal of variability in what characteristics are expressed. Some individuals have many of the typical characteristics and others have only a few. Abnormal speech is the most common characteristic of this syndrome, however.

### **What is the treatment for velocardiofacial syndrome?**

Because velocardiofacial syndrome has characteristics that affect different parts of the body and different functions, a team approach to management is very important. A cleft palate team, with the help of other professionals, is usually best to be able to manage these characteristics. Treatment of speech may include a combination of surgery and speech therapy.



### **Website on velocardiofacial syndrome:**

- Cleft Palate Foundation: <http://www.cleftline.org/>
- American Cleft Palate – Craniofacial Association: [www.acpa-cpf.org](http://www.acpa-cpf.org)

For more information, please contact the Division of Speech Pathology at (513) 636-4341 or visit our website at [www.cincinnatichildrens.org/speech](http://www.cincinnatichildrens.org/speech).