Velocardiofacial Syndrome (VCFS)

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Velocardiofacial Syndrome (VCFS) is also known as Shprintzen Syndrome, DiGeorge syndrome, or 22q11.2 Syndrome. VCFS is found in patients with hypernasality, often of unknown etiology. In addition to the characteristic hypernasality, the individual often demonstrates language and learning problems, an articulation disorder and hearing loss. Therefore, it is the speech pathologist that may be the first to detect the problem and refer for further medical assessment and intervention.

Basic Characteristics

- **Velo:** velopharyngeal dysfunction causing hypernasality. Usually secondary an occult submucous cleft or pharyngeal hypotonia. Can be due to an overt cleft palate or submucous cleft.
- **Cardio:** minor cardiac and vascular anomalies including ventriculoseptal deviation (VSD); atrial septal defect (ASD); patent ductus arteriosis (PDA); pulmonary stenosis; tetralogy of Fallot; right sided aortic arch; medially displaced internal carotid arteries; and tortuosity of the retinal arteries. Parents often report a history of heart murmur at birth.
- **Facial:** microcephaly, long face with vertical maxillary excess; micrognathia (small jaw) or retruded mandible, often with a Class II malocclusion; nasal anomalies including wide nasal bridge, narrow alar base and bulbous nasal tip; narrow palpebral fissures (slit-like eyes); malar flatness; thin upper lip; minor auricular anomalies; abundant scalp hair and others.
- **Learning and Cognitive Problems:** learning disabilities; mild to moderate mental retardation in about 40% of the cases; concrete thinking; language disorders.
- **Communication Problems:** hypernasality due to velopharyngeal insufficiency and pharyngeal hypotonia; misarticulations, often due to verbal apraxia; conductive and/or sensorineural hearing loss; language disorders with learning problems; and high pitched voice. Abnormal speech is the most common characteristic.
- **Other Common Physical and Medical Characteristics:** long slender digits, hyperextensibility of the joints; short stature, usually below the 10%ile; Robin malformation sequence (cleft palate, micrognathia, glossoptosis with airway obstruction); umbilical and inguinal hernias; and laryngeal web.
- **Other Common Functional Problems:** early feeding problems; gross and fine motor delays; social disinhibition; and risk of onset of psychosis in adolescence.

Etiology

- Autosomal dominant
- Deletion on chromosome 22q11.2
- Variable expressivity
- Can exhibit many of the typical characteristics or only a few
- Abnormal speech is the most common characteristic

Importance of Identification

- This syndrome is an autosomal dominant genetic condition, so genetic counseling is very important for family planning.
- Identification can allow caregivers to plan for appropriate treatment of medical and speech–language problems.
- Knowledge of the syndrome can allow for the planning of realistic goals.