

Velocardiofacial Syndrome (VCFS)

Also known as:

- Shprintzen Syndrome
- De George syndrome
- 22q11.2 syndrome



Etiology

- Gene deletion on chromosome 22q11.2
- May be deletion of another yet-to-be discovered gene
- Occurs sporadically
- Autosomal dominant in affected individuals offspring- 50% recurrence rate

Test for VCFS

- FISH probe to look for deletion on 22q11.2
- Deletion confirms diagnosis
- Lack of deletion does not rule out diagnosis

Variable Expressivity

- Can exhibit many or only a few of the typical characteristics
- Abnormal speech is the most common characteristic
- Tends to become more severe in successive generations

Basic Phenotypic Features

- **Velo:** velopharyngeal dysfunction
- **Cardio:** minor cardiac, vascular anomalies
- **Facial:** dysmorphic facial features
- **Other:** learning disabilities, OM dysfunction, psychological concerns, other medical problems

Velopharyngeal Dysfunction

- cleft of the soft palate
- submucous cleft
- occult submucous cleft
- pharyngeal hypotonia
- Rarely includes cleft lip

Cardiac Anomalies

- ventricular septal defect (VSD)
- atrial septal defect (ASD)
- patent ductus arteriosus (PDA)
- pulmonary stenosis

Vascular Anomalies

- right sided aortic arch
- tetralogy of Fallot
- tortuosity of retinal blood vessels
- medially displaced internal carotid arteries

Facial Features

- long, narrow face with vertical maxillary excess
- broad nasal bridge with narrow alar base and bulbous nasal tip
- narrow palpebral fissures
- flattened malar eminences

Facial Features

- thin upper lip
- micrognathia or retruded mandible, often with Class II malocclusion
- minor auricular anomalies
- abundant scalp hair
- microcephaly

VCFS- Facial Features



change the outcome®

VCFS- Facial Features



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Other Medical Problems

- Pierre Robin sequence (micrognathia, glossoptosis with airway obstruction, cleft palate)
- laryngeal web
- umbilical or inguinal hernias
- Many documented brain anomalies-considered a “neurodevelopmental disorder”

Common Physical Findings

- small stature, usually below the tenth percentile
- Note the 8 year old boy with short stature



Common Physical Findings

- long, slender fingers
- hyperextensibility of the joints



Common Functional Problems

- early feeding problems
- gross and fine motor dysfunction
- conductive or sensori-neural hearing loss
- learning disabilities/mild mental retardation
- concrete thinking, difficulty with abstraction
- outgoing personality with social disinhibition
- risk of onset of psychosis in adolescence

Communication Problems

- hypernasality (VPD)
- misarticulations, often due to verbal apraxia
- hearing loss
- language impairment- receptive problems with abstraction
- high pitched voice

Importance of Identification

- Genetic counseling for family planning
- Allows caregivers to plan for appropriate treatment
- Can allow for planning of realistic goals