VELO-CARDIO-FACIAL SYNDROME
Specialist Fact Sheet

Velo-cardio-facial syndrome (VCFS is caused by a deletion of a small segment of the long arm of chromosome 22. It is one of the most common genetic disorders in humans. The following list shows the anomalies that have been found in VCFS. No features are found in 100% of cases, but all occur with sufficient frequency to warrant assessment. If you have any questions, or if you would like to learn more about VCFS, you may reach **The Velo-Cardio-Facial Syndrome Educational Foundation** by telephone at **315-464-6590**, by fax at **315-464-5321**, or by email at **vcfsef@mail.upstate.edu**. The Foundation maintains a web site at www.vcfsef.org

Craniofacial/Oral Findings

- 1. Overt, submucous or occult submucous cleft palate
- 2. Retrognathia (retruded lower jaw)
- 3. Platybasia (flat skull base)
- 4. Asymmetric crying facies in infancy
- 5. Structurally asymmetric face
- 6. Functionally asymmetric face
- 7. Vertical maxillary excess (long face)
- 8. Straight facial profile
- 9. Congenitally missing teeth (one or several)
- 10. Small teeth
- 11. Enamel hypoplasia (primary dentition)
- 12. Hypotonic, flaccid facies
- 13. Downturned oral commissures
- 14. Cleft lip (uncommon)
- 15. Microcephaly
- 16. Small posterior cranial fossa

Eye Findings

- 17. Tortuous retinal vessels
- 18. Suborbital congestion ("allergic shiners")
- 19. Strabismus
- 20. Narrow palpebral fissures
- 21. Posterior embryotoxon
- 22. Small optic disk
- 23. Prominent corneal nerves
- 24. Cataract
- 25. Iris nodules
- 26. Iris coloboma (uncommon)
- 27. Retinal coloboma (uncommon)
- 28. Small eyes
- 29. Mild orbital hypertelorism
- 30. Mild vertical orbital dystopia
- 31. Puffy upper eyelids

Ear/Hearing Findings

- 32. Overfolded helix
- 33. Attached lobules
- 34. Protuberant, cup-shaped ears
- 35. Small ears
- 36. Mildly asymmetric ears
- 37. Frequent otitis media
- 38. Mild conductive hearing loss
- 39. Sensori-neural hearing loss (often unilateral)
- 40. Ear tags or pits (uncommon)
- 41. Narrow external ear canals

Nasal Findings

- 42. Prominent nasal bridge
- 43. Bulbous nasal tip
- 44. Mildly separated nasal domes (nasal tip appears bifid)
- 45. Pinched alar base, narrow nostrils
- 46. Narrow nasal passages

Cardiac and Thoracic Vascular Findings

- 47. VSD (Ventricular septal defect)
- 48. ASD (Atrial septal defect)
- 49. Pulmonic atresia or stenosis
- 50. Tetralogy of Fallot
- 51. Right sided aorta
- 52. Truncus arteriosus
- 53. PDA (patent ductus arteriosus)
- 54. Interrupted aorta, type B
- 55. Coarctation of the aorta
- 56. Aortic valve anomalies
- 57. Aberrant subclavian arteries
- 58. Vascular ring
- 59. Anomalous origin of carotid artery
- 60. Transposition of the great vessles
- 61. Tricuspid atresia

Vascular Anomalies

- 62. Medially displaced internal carotid arteries
- 63. Tortuous or kinked internal carotids
- 64. Jugular vein anomalies
- 65. Absence of internal carotid artery (unilateral)
- 66. Absence of vertebral artery (unilateral)
- 67. Low bifurcation of common carotid
- 68. Tortuous or kinked vertebral arteries
- 69. Reynaud's phenomenon
- 70. Small veins
- 71. Circle of Willis anomalies

Neurologic, Brain, and MR Findings

- 72. Periventricular cysts (mostly at anterior horns)
- 73. Small cerebellar vermis
- 74. Cerebellar hypoplasia/dysgenesis
- 75. White matter UBOs (unidentified bright objects)
- 76. Generalized hypotonia
- 77. Cerebellar ataxia
- 78. Seizures
- 79. Strokes
- 80. Spina bifida/meningomyelocele
- 81. Mild developmental delay
- 82. Enlarged Sylvian fissure

Pharyngeal/Laryngeal/Airway Findings

- 83. Upper airway obstruction in infancy
- 84. Absent or small adenoids
- 85. Laryngeal web (anterior)
- 86. Large pharyngeal airway
- 87. Laryngomalacia
- 88. Arytenoid hyperplasia
- 89. Pharyngeal hypotonia
- 90. Asymmetric pharyngeal movement
- 91. Thin pharyngeal muscle
- 92. Unilateral vocal cord paresis
- 93. Reactive airway disease
- 94. Asthma

Abdominal/Kidney/Gut

- 95. Hypoplastic/aplastic kidney
- 96. Cystic kidneys
- 97. Inguinal hernias
- 98. Umbilical Hernias
- 99. Malrotation of bowel
- 100. Diastasis recti
- 101. Diaphragmatic hernia (uncommon)
- 102. Hirschsprung megacolon (rare)

Limb Findings

- 103. Small hands and feet
- 104. Tapered digits
- 105. Short nails
- 106. Rough, red, scaly skin on hands and feet
- 107. Morphea
- 108. Contractures
- 109. Triphalangeal thumbs
- 110. Polydactyly, both pre- and postaxial (uncommon)
- 111. Soft tissue syndactyly

Problems in Infancy

- 112. Feeding difficulty, Failure-to-thrive
- 113. Nasal vomiting
- 114. Gastroesophageal reflux
- 115. Irritability
- 116. Chronic constipation (not Hirschsprung megacolon)

Genitourinary

- 117. Hypospadias
- 118. Cryptorchidism
- 119. Vesico-ureteral reflux

Speech/Language

- 120. Severe hypernasality
- 121. Severe articulation impairment (glottal stops)
- 122. Language impairment (usually mild delay)
- 123. Velopharyngeal insufficiency (usually severe)
- 124. High pitched voice
- 125. Hoarseness

Cognitive/Learning

- 126. Learning disabilities (math concept, reading comprehension)
- 127. Concrete thinking, difficulty with abstraction
- 128. Drop in IQ scores in school years (test artifact)
- 129. Borderline normal intellect
- 130. Occasional mild mental retardation
- 131. Attention deficit hyperactivity disorder

Miscellaneous anomalies

- 132. Spontaneous oxygen desaturation without apnea
- 133. Thrombocytopenia, Bernard-Soulier disease
- 134. Juvenile rheumatoid arthritis
- 135. Poor body temperature regulation

Psychiatric/Psychological

- 136. Bipolar affective disorder
- 137. Manic depressive illness and psychosis
- 138. Rapid or ultrarapid cycling of mood disorder
- 139. Mood disorder
- 140. Depression
- 141. Hypomania
- 142. Schizoaffective disorder
- 143. Schizophrenia
- 144. Impulsiveness
- 145. Flat affect
- 146. Dysthymia
- 147. Cyclothymia
- 148. Social immaturity
- 149. Obsessive compulsive disorder
- 150. Generalized anxiety disorder
- 151. Phobias
- 152. Severe startle response

Immunologic

- 153. Frequent upper respiratory infections
- 154. Frequent lower airway disease (pneumonia, bronchitis)
- 155. Reduced T cell populations
- 156. Reduced thymic hormone

Endocrine

- 157. Hypocalcemia
- 158. Hypoparathyroidism
- 159. Hypothyroidism
- 160. Mild growth deficiency, relative small stature
- 161. Absent, hypoplastic thymus
- 162. Small pituitary gland (rare)

Skeletal/Muscle/Orthopedic

- 163. Scoliosis
- 164. Spina bifida occulta
- 165. Hemivertebrae
- 166. Butterfly vertebrae
- 167. Fused vertebrae (usually cervical)
- 168. Osteopenia
- 169. Sprengel's anomaly, scapular deformation
- 170. Talipes equinovarus
- 171. Small skeletal muscles

❛172. Joint dislocations
❛173. Chronic leg pains
❛174. Flat foot arches
❛175. Hyperextensible/lax joints
❛176. Rib fusion
❛177. Extra ribs
❛178. Tethered cord
❛179. Syrinx

Skin/Integument
❛180. Abundant scalp hair
❛181. Thin appearing skin (venous patterns easily visible)

Secondary sequences/associations
❛182. Robin sequence
❛183. DiGeorge sequence
❛184. Potter sequence
❛185. CHARGE association
❛186. Holoprosencephaly (single case)

Some other facts about the syndrome:
Population prevalence (estimated): 1:2,000 people
Birth incidence (estimated): 1:1,800 births
Prevalence in infants with conotruncal heart anomalies:10-30%
Prevalence in cleft palate (without cleft lip): 8%

Future meetings of the Foundation:
2002: Northampton, England
2003: San Diego, CA
2004: Atlanta, GA
2005: Syracuse, NY