VELO-CARDIO-FACIAL SYNDROME  
Specialist Fact Sheet  

Velo-cardio-facial syndrome (VCFS), also known as Shprintzen syndrome, DiGeorge sequence or syndrome, and 22q11 deletion syndrome, 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 findings have a 100% frequency, but all occur with sufficient frequency to warrant assessment. For more information, check the web site of The Velo-Cardio-Facial Syndrome Educational Foundation, Inc. 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  
10. Small teeth (in primary dentition)  
11. Enamel hypoplasia  
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 or hooded 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 (tip appears bifid)  
45. Pinched alar base, narrow nostrils  
46. Narrow nasal passages  

Cardiac 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 aortic arch, 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 vessels  
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. Raynaud's phenomenon  
70. Small veins  
71. Circle of Willis anomalies  

Neurologic and Brain Findings  
72. Reduced total brain volume  
73. Periventricular cysts  
74. Small cerebellar vermis  
75. Cerebellar hypoplasia/dysgenesis  
76. White matter hyperintensities  
77. Generalized hypotonia  
78. Cerebellar ataxia  
79. Seizures  
80. Strokes  
81. Spina bifida/meningomyelocele  
82. Mild developmental delay  
83. Enlarged Sylvian fissure  
84. Cavernous septum pellucidum  
85. Variations in size of various brain segments.  

Pharyngeal/Laryngeal/Airway Findings  
86. Upper airway obstruction in infancy  
87. Absent or small adenoids  
88. Laryngeal web (anterior)  
89. Large pharyngeal airway  
90. Laryngomalacia  
91. Arytenoid hyperplasia  
92. Pharyngeal hypotonia  
93. Asymmetric pharyngeal movement  
94. Thin pharyngeal muscle  
95. Vocal cord paresis/paralysis (usually unilateral)  
96. Structurally asymmetric pharynx  
97. Structurally asymmetric larynx  
98. Reactive airway disease/asthma
Abdominal/Kidney
99. Hypoplastic/aplastic kidney
100. Cystic kidneys
101. Inguinal hernias
102. Umbilical Hernias
103. Malrotation of bowel (uncommon)
104. Diastasis recti
105. Diaphragmatic hernia

Limb Findings
106. Small hands and feet
107. Tapered digits
108. Short nails
109. Rough, red, scaly skin on hands and feet
110. Morphea
111. Contractures (uncommon)
112. Triphalangeal thumbs (uncommon)
113. Polydactyly
114. Soft tissue syndactyly (uncommon)

Problems in Infancy
115. Feeding difficulty, Failure-to-thrive
116. Gastroesophageal reflux (GER/GERD)
117. Nasal regurgitation
118. Irritability
119. Chronic constipation

Genitourinary
120. Hypospadias
121. Cryptorchidism
122. Vesico-urethral reflux
123. Hydrocele

Speech/Language
124. Severe hypernasality
125. Severe articulation impairment (glottal stops)
126. Language impairment (usually mild delay)
127. Velopharyngeal insufficiency (usually severe)
128. High pitched voice
129. Hoarseness

Cognitive/Learning
130. Learning disabilities (math concept, reading comprehension most common)
131. Concrete thinking, executive function deficiency
132. Drop in IQ scores in school years
133. Low IQ, typically 60 – 80, some lower, some higher
134. Attention deficit hyperactivity disorder (ADD/ADHD)

Miscellaneous anomalies
135. Spontaneous oxygen desaturation without apnea
136. Thrombocytopenia
137. Bernard-Soulier syndrome
138. Juvenile rheumatoid arthritis
139. Poor temperature regulation
140. Vaso-motor instability

Psychiatric/Psychological
141. Bipolar affective disorder
142. Manic depressive illness and psychosis
143. Schizophrenia
144. Rapid or ultrarapid cycling of mood disorder
145. Mood disorder, depression
146. Autism spectrum disorder
147. Schizoaffective disorder
148. Impulsiveness
149. Flat affect
150. Dysthymia, Cyclothymia
151. Social Immaturity
152. Obsessive compulsive disorder
153. Generalized anxiety disorder
154. Phobias
155. Separation anxiety
156. Severe startle response

Immunologic
157. Frequent upper respiratory infections
158. Frequent lower airway disease (pneumonia, bronchitis)
159. Reduced T cell populations
160. Reduced thymic hormone

Endocrine
161. Hypocalcemia
162. Hypoparathyroidism
163. Hypothyroidism
164. Auto-immune thyroiditis
165. Mild growth deficiency, relative small stature  (childhood)
166. Absent, hypoplastic thymus
167. Small pituitary gland (rare)

Skeletal/Muscle/Orthopedic
168. Scoliosis
169. Osteopenia
170. Sprengel's anomaly, scapular deformation
171. Talipes equinovarus
172. Small skeletal muscles
173. Joint dislocations
174. Chronic leg pains
175. Flat foot arches
176. Hyperextensible/lax joints
177. Spina bifida occulta
178. Syringa
179. Tethered cord
180. Extra ribs
181. Rib fusion
182. Vertebral anomalies

Skin/Integument
183. Abundant scalp hair
184. Thin appearing skin (venous patterns easily visible)

Secondary sequences/associations
185. Robin sequence
186. DiGeorge sequence
187. Potter sequence
188. Holoprosencephaly

Facts about VCFS
Population prevalence: 1:2000, birth frequency higher
Cause: Deletion of DNA from chromosome 22 at q11.2 band
Most common syndrome of cleft palate
Most common microdeletion syndrome in humans
Most common syndrome expressing conotruncal heart anomalies