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. Cavum 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. Syrinx
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