

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. Platibasias (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