

List of Anomalies

This specialist fact sheet was drawn up by the Velo-Cardio-Facial Syndrome Educational Foundation. The list shows the anomalies which have been found to be associated with VCFS. None of the findings have 100% frequency. Thus some patients will exhibit more findings but most people are only going to be affected by a small number of the findings .

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 and/or functionally asymmetric face
- 6 Straight facial profile
- 7 Cleft lip (uncommon)
- 8 Enamel hypoplasia (primary dentition)
- 9 Small teeth
- 10 Congenitally missing teeth
- 11 Hypotonic, flaccid facies
- 12 Downturned oral commissures
- 13 Microcephaly
- 14 Small posterior cranial fossa
- 15 Vertical maxillary excess (long face)

Eye findings

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

Limb findings

- 31 Small hands and feet
- 32 Tapered digits
- 33 Short nails
- 34 Contractures
- 35 Triphalangeal thumbs
- 36 Soft tissue syndactyly
- 37 Rough, red, scaly skin on hands
- 38 Morphea
- 39 Polydactyly (both preaxial and postaxial)

Nasal findings

- 40 Prominent nasal bridge
- 41 Bulbous nasal tip
- 42 Mildly separated nasal domes
- 43 Pinched alar base, narrow nostrils
- 44 Narrow nasal passages

Pharyngeal/laryngeal airway

- 45 Upper airway obstruction in infancy

- 46 Absent or small adenoids
- 47 Laryngeal web (anterior)
- 48 Large pharyngeal airway
- 49 laryngomalacia
- 50 Arytenoid hypoplasia
- 51 Pharyngeal hypotonia
- 52 Asymmetrical pharyngeal movement
- 53 Thin pharyngeal muscle
- 54 Unilateral vocal cord paresis

Skin/integument

- 55 Abundant scalp hair
- 56 Thin appearing skin (venous patterns easily visible)

Skeletal/muscle/orthopaedic/spine

- 57 Scoliosis
- 58 Hemivertebrae
- 59 Spina bifida occulta
- 60 Butterfly vertebrae
- 61 Fused vertebrae (mostly cervical)
- 62 Tethered spinal cord
- 63 Syrinx
- 64 Sprengel's anomaly/scapular deformation
- 65 Small skeletal muscles
- 66 Joint dislocations
- 67 Chronic leg pains
- 68 Flat foot arches
- 69 Hyperextensible/lax joints
- 70 Extra ribs
- 71 Rib fusion
- 72 Talipes equinovarus
- 73 Osteopaenia

Psychiatric/psychological

- 74 Bipolar affective disorder
- 75 Manic depressive illness and psychosis
- 76 Rapid or ultrarapid cycling of mood disorder
- 77 Mood disorder
- 78 Depression
- 79 Hypomania
- 80 Generalised anxiety disorder
- 81 Schizoaffective disorder
- 82 Impulsiveness
- 83 Flat affect
- 84 Dysthymia
- 85 Cyclothymia
- 86 Social immaturity
- 87 Obsessive compulsive disorder
- 88 Phobias

Cardiac findings

- 89 VSD (ventricular septal defect)
- 90 ASD (atrial septal defect)
- 91 Pulmonary atresia or stenosis
- 92 Tetralogy of Fallot
- 93 Right-sided aorta
- 94 Truncus arteriosus
- 95 PDA (patent ductus arteriosus)
- 96 Interrupted aorta
- 97 Coarctation of the aorta
- 98 Aortic valve anomalies
- 99 Aberrant subclavian arteries
- 100 Vascular ring

- 101 Anomalous origin of carotid artery
- 102 Transposition of the great vessels
- 103 Tricuspid atresia

Ear/hearing findings

- 104 Overfolded helix
- 105 Attached lobules
- 106 Protuberant, cup-shaped ears
- 107 Small ears
- 108 Mildly asymmetric ears
- 109 Frequent otitis media
- 110 Mild conductive hearing loss
- 111 Sensorineural hearing loss
- 112 Ear tags or pits (uncommon)
- 113 Narrow external ear canals
- 114

Problems in infancy

- 115 Difficulty in feeding, failure to thrive
- 116 Nasal vomiting
- 117 Gastro-oesophageal reflux
- 118 Nasal regurgitation
- 119 Irritability
- 120 Chronic constipation (not Hirshprung megacolon)

Speech/language

- 121 Severe hypernasality
- 122 Severe articulation impairment
- 123 Language impairment (usually mild delay)
- 124 Dyspraxia
- 125 Velopharyngeal insufficiency (usually severe)
- 126 High-pitched voice
- 127 Hoarseness

Cognitive/Learning

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

Endocrine

- 134 Hypocalcaemia
- 135 Hypoparathyroidism
- 136 Pseudo-hypoparathyroidism
- 137 Hypothyroidism
- 138 Mild growth deficiency, relative small stature
- 139 Absent, hypoplastic thymus
- 140 Hypoplastic pituitary gland

Neurological/brain findings

- 141 Periventricular cysts (mostly anterior horns)
- 142 Small cerebellar vermis
- 143 Cerebellar hypoplasia/dysgenesis
- 144 White matter UBO's (unidentified bright objects)
- 145 Cerebellar ataxia
- 146 Seizures
- 147 Strokes
- 148 Spina bifida/meningomyelocele

- 149 Mild developmental delay
- 150 Enlarged Sylvian fissures
- 151 Generalised hypotonia

Vascular anomalies

- 152 Medially displaced internal carotid arteries
- 153 Tortuous, kinked, absent or accessory internal carotids
- 154 Jugular vein anomalies
- 155 Small veins
- 156 Circle of Willis anomalies
- 157 Absence of vertebral artery (unilateral)
- 158 Low bifurcation of common carotid
- 159 Tortuous or kinked vertebral arteries
- 160 Reynaud's phenomenon

Genito-urinary

- 161 Hypospadias
- 162 G-U reflux
- 163 Cryptorchidism

Immunologic

- 164 Reduced T cell populations
- 165 Frequent lower airway disease (pneumonia, bronchitis)
- 166 Frequent upper respiratory infections
- 167 Reduced thymic hormone
- 168 Reactive airway disease

Abdominal/kidney/gut

- 169 Hypoplastic/aplastic kidney
- 170 Cystic kidneys
- 171 Anal anomalies (displaced, imperforate)
- 172 Inguinal hernias
- 173 Single case of malrotation of bowel
- 174 Hepatoblastoma and diaphragmatic hernia

Associations/sequences

- 175 DiGeorge Sequence
- 176 Pierre Robin Sequence
- 177 Potter Sequence
- 178 CHARGE Association
- 179 Holoprosencephaly (single case)