

MOST AFFECTED SYSTEMS

- Cardio-vascular
- Cleft palate
- Feeding difficulties
- Immunization problems
- Growth hormone deficiencies
- Delayed neurological and psychological developments
- Speech problems
- Renal abnormalities

Anomaly fact sheet of symptoms

Abdominal/kidney/gut

- _____ Hypoplastic/aplastic kidney
- _____ Cystic kidneys
- _____ Anal anomalies (displaced, imperforate)
- _____ Inguinal hernias
- _____ Umbilical hernias
- _____ Single case of malrotation of the bowel
- _____ Hepatoblastoma and diaphragmatic hernia (rare)
- _____ Diastasis recti abdominis

Cardiac findings

- _____ VSD (ventricular septal defect)
- _____ ASD (atrial septal defect)
- _____ Pulmonary atresia or stenosis
- _____ Tetralogy of Fallot
- _____ Right-sided aorta
- _____ Truncus arteriosus
- _____ PDA (patent ductus arteriosus)
- _____ Interrupted aorta
- _____ Coarctation of the aorta
- _____ Aortic valve anomalies
- _____ Aberrant subclavian arteries
- _____ Vascular ring
- _____ Anomalous origin of carotid artery
- _____ Transposition of the great vessels
- _____ Tricuspid atresia

Cognitive/learning

- _____ Learning disabilities (math concept, reading comprehension)
- _____ Concrete thinking, difficulty with abstract thinking
- _____ Drop in IQ scores in school years (test artifact)
- _____ Borderline normal intellect (based on 100% as “normal”)
- _____ Occasional mild mental retardation
- _____ Attention deficit hyperactivity disorder (ADD/ADHD)

Craniofacial/oral findings

- _____ Overt, submucous or occult submucous cleft palate
- _____ Retrognathia (retruded lower jaw)
- _____ Platybasia (flat skull base)
- _____ Asymmetric crying facies in infancy
- _____ Structurally and/or functionally asymmetric face
- _____ Straight facial profile
- _____ Cleft lip (uncommon)
- _____ Enamel hypoplasia on teeth (primary dentition)
- _____ Small teeth
- _____ Congenitally missing teeth
- _____ Hypotonic, flaccid facies
- _____ Downturned oral commissures
- _____ Microcephaly (small head)
- _____ Small posterior cranial fossa
- _____ Vertical maxillary excess (long face)
- _____ Tortuous retinal vessel
- _____ Suborbital congestion “allergic shiners”
- _____ Strabismus
- _____ Narrow palpebral fissures
- _____ Posterior embryotoxin
- _____ Small optic disk
- _____ Prominent corneal nerves
- _____ Cataract
- _____ Iris nodules
- _____ Iris coloboma (uncommon)
- _____ Retinal coloboma (uncommon)
- _____ Small eyes
- _____ Mild orbital hypertelorism
- _____ Mild orbital dystopia
- _____ Puffy eyelids

Ear/hearing findings

- _____ Over-folded helix
- _____ Attached lobules
- _____ Protuberant, cup-shaped ears
- _____ Small ears
- _____ Mild asymmetric ears
- _____ Frequent otitis media
- _____ Mild conductive hearing loss
- _____ Sensorineural hearing loss
- _____ Ear tags or pits (uncommon)
- _____ Narrow external ear canals

Endocrine

- _____ Hypocalcaemia
- _____ Hypoparathyroidism
- _____ Pseudo-hypoparathyroidism
- _____ Hypothyroidism
- _____ Mild growth deficiency, relative small stature
- _____ Absent, hypoplastic thymus
- _____ Hypoplastic pituitary gland
- _____ Poor body temperature regulation

Genito-urinary

- _____ Hypospadias
- _____ G-U reflux
- _____ Cryptorchidism

Immunologic

- _____ Reduced T cell populations
- _____ Frequent lower airway disease (pneumonia, bronchitis)
- _____ Frequent upper respiratory infections
- _____ Reduced thymic hormone

Limb findings

- _____ Small hands and feet
- _____ Tapered digits
- _____ Short nails
- _____ Contractures
- _____ Triphalangeal thumbs
- _____ Soft tissue syndactyly
- _____ Rough, red, scaly skin on hands
- _____ Morphea
- _____ Polydactyly (both preaxial and postaxial)

Nasal findings

- _____ Prominent nasal bridge
- _____ Bulbous nasal tip
- _____ Mildly separated nasal domes
- _____ Pinched alar base, narrow nostrils
- _____ Narrow nasal passages

Neurological/brain findings

- _____ Periventricular cysts (mostly anterior horns)
- _____ Small cerebellar vermis
- _____ Cerebellar hypoplasia/dysgenesis
- _____ White matter UBO's (unidentified bright objects)
- _____ Cerebellar ataxia
- _____ Seizures
- _____ Strokes
- _____ Spina bifida/meningomyelocele
- _____ Mild developmental delay
- _____ Enlarged Sylvian fissures
- _____ Generalized hypotonia

Pharyngeal/laryngeal airway

- _____ Upper airway obstruction in infancy
- _____ Absent or small adenoids
- _____ Laryngeal web (anterior)
- _____ Large pharyngeal airway

- _____ Laryngomalacia
- _____ Arytenoid hypoplasia
- _____ Pharyngeal hypotonia
- _____ Asymmetrical pharyngeal movement
- _____ Thin pharyngeal muscle
- _____ Unilateral vocal cord paresis
- _____ Reactive airway disease
- _____ Spontaneous oxygen desaturation without apnea

Problems in infancy

- _____ Difficulty in feeding, failure to thrive
- _____ Nasal vomiting
- _____ Gastro-esophageal reflux
- _____ Nasal regurgitation
- _____ Irritability
- _____ Chronic constipation (not Hirshprung megacolon)

Psychiatric/psychological

- _____ Bipolar affective disorder
- _____ Manic depressive illness and psychosis
- _____ Rapid or ultra-rapid cycling of mood disorder
- _____ Mood disorder
- _____ Depression
- _____ Hypomania
- _____ Generalized anxiety disorder
- _____ Schizoaffective disorder
- _____ Impulsiveness
- _____ Flat affect
- _____ Dysthymia
- _____ Cyclomania
- _____ Social immaturity
- _____ Obsessive compulsive disorder
- _____ Phobias
- _____ Exaggerated startle response

Skeletal/muscle/orthopedic/spine

- _____ Scoliosis
- _____ Hemivertebrae
- _____ Spina bifida occulta
- _____ Butterfly vertebrae
- _____ Fused vertebrae (mostly cervical)
- _____ Tethered spinal cord
- _____ Syrinx
- _____ Sprengel's anomaly/scapular deformation
- _____ Small skeletal muscles
- _____ Joint dislocations
- _____ Chronic leg pains
- _____ Flat foot arches
- _____ Hyperextensible/lax joints
- _____ Extra ribs
- _____ Rib fusion
- _____ Talipes equinovarus (club feet)
- _____ Osteopaenia

_____ Juvenile rheumatoid arthritis (JRA)

Skin/integument

_____ Abundant scalp hair
_____ Thin appearing skin (venous patterns easily visible)
_____ Rosacea

Speech/language

_____ Severe hypernasality
_____ Severe articulation impairment
_____ Language impairment (usually mild delay)
_____ Dyspraxia
_____ Velopharyngeal insufficiency (VPI) (usually severe)
_____ High-pitched voice
_____ Hoarseness

Vascular anomalies

_____ Medially displaced internal carotid artery
_____ Tortuous, kinked, absent or accessory internal carotids
_____ Jugular vein anomalies
_____ Small veins
_____ Circle of Willis anomalies
_____ Absence of vertebral artery (unilateral)
_____ Low bifurcation of common carotid
_____ Tortuous or kinked vertebral arteries
_____ Raynaud's phenomenon
_____ Thrombocytopenia, Bernard-Soulier disease