

## **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
- \_\_\_\_\_ Cyclothymia
- \_\_\_\_\_ 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