

PERINATAL HISTORY	MOTOR FUNCTION (cont.)	NEURAL PHYSIOLOGY (cont.)	EYE DEFECTS/VISION (cont.)
Abnormal placenta morphology	Myoclonus	Psychosis	Lacrimal duct stenosis
Abnormality of the umbilical cord	Paraplegia	Seizure	Macular atrophy
Breech presentation	Parkinsonism	Sensorimotor neuropathy	Macular degeneration
Congenital diaphragmatic hernia	Poor suck	Sensory neuropathy	Macular purpura
Failure to thrive	Postural instability	Vertigo	Microphthalmia
Growth delay	Progressive muscle weakness	CRANIOFACIAL/DYSMORPHISM	Myopia
Hydrops fetalis	Proximal muscle weakness	Abnormal facial shape	Nystagmus
Increased nuchal translucency	Spasticity	Abnormality of malar bones	Ophthalmoparesis
Infantile spasms	Tip-toe gait	Anosmia	Optic atrophy
Intrauterine growth retardation	Torticollis	Anteverted nares	Optic neuropathy
Low APGAR score	Tremor	Bifid uvula	Posterior embryotoxon
Nonimmune hydrops fetalis	Waddling gait	Brachycephaly	Proptosis
Oligohydramnios	NEURAL MORPHOLOGY	Choanal atresia	Pterygium
Omphalocele	Abnormal brainstem morphology	Cleft lip	Ptosis
Polyhydramnios	Abnormal corpus callosum morphology	Cleft palate	Reduced visual acuity
Postnatal growth retardation	Abnormal myelination	Coarse facial features	Retinal atrophy
Premature birth	Abnormal periventricular white matter morphology	Craniosynostosis	Retinal degeneration
Prolonged neonatal jaundice	Abnormality of the basal ganglia	Dental enamel pits	Retinal detachment
DEVELOPMENT AND BEHAVIOR	Aplasia/Hypoplasia of the cerebellar vermis	Dental malocclusion	Retinal dystrophy
Absent speech	Aplasia/Hypoplasia of the cerebellum	Depressed nasal bridge	Retinoschisis
Aggressive behavior	Brain stem compression	Depressed nasal tip	Rod-cone dystrophy
Anxiety	Cerebellar atrophy	Dolichocephaly	Strabismus
Apathy	Cerebral cortical atrophy	Facial asymmetry	Telecanthus
Attention deficit hyperactivity disorder	CNS demyelination	Frontal bossing	Upslanted palpebral fissure
Autistic behavior	CNS hypomyelination	High palate	Vitreoretinopathy
Cognitive impairment	Cortical dysplasia	Hypodontia	EAR DEFECTS/HEARING
Delayed speech and language development	Cortical tubers	Macrocephaly	Conductive hearing impairment
Depression	Delayed CNS myelination	Malar flattening	Cupped ear
Developmental regression	Dysgenesis of the basal ganglia	Mandibular prognathia	High-frequency hearing impairment
Dyscalculia	Encephalocele	Microcephaly	Hypoplasia of the semicircular canal
Dyslexia	Gliosis	Micrognathia	Low-frequency hearing loss
Global developmental delay	Gray matter heterotopia	Midface retrusion	Low-set ears
Hyperactivity	Holoprosencephaly	Narrow palate	Microtia
Impaired visuospatial constructive cognition	Hydrocephalus	Retrognathia	Mild hearing impairment
Intellectual disability	Hyperintensity of cerebral white matter on MRI	Short philtrum	Posteriorly rotated ears
Lethargy	Hypointensity of cerebral white matter on MRI	Sloping forehead	Preauricular pit
Memory impairment	Hypoplasia of the corpus callosum	Smooth philtrum	Protruding ear
Obsessive-compulsive behavior	Leukodystrophy	Thick nasal alae	Sensorineural hearing impairment
Polydipsia	Lissencephaly	Thin upper lip vermillion	Unilateral deafness
Restrictive behavior	Pachygryia	Trigonocephaly	NECK AND CHEST
Sleep disturbance	Periventricular leukomalacia	Wide anterior fontanel	Chest pain
Speech articulation difficulties	Periventricular nodular heterotopia	Wide nose	Cystic hygroma
MOTOR FUNCTION	Periventricular white matter hyperintensities	Widely spaced teeth	Gynecomastia
Akinesia	Polymicrogyria	EYE DEFECTS/VISION	Heterotaxy
Areflexia	Tethered cord	Amblyopia	Inverted nipples
Ataxia	Ventriculomegaly	Anophthalmia	Laryngeal stenosis
Athetosis	NEURAL PHYSIOLOGY	Blepharitis	Neck flexor weakness
Cerebral palsy	Abnormal pineal melatonin secretion	Blepharophimosis	Neck muscle weakness
Chorea	Agnosia	Cataract	Neck ptterygia
Dyskinesia	Decreased intracranial pressure	Cerebral visual impairment	Shield chest
Dysmetria	Decreased motor nerve conduction velocity	Coloboma	Short neck
Dystonia	Decreased nerve conduction velocity	Conjunctivitis	Supernumerary nipple
Gait disturbance	Decreased sensory nerve conduction velocity	Corneal dystrophy	Webbed neck
Hemiplegia	Demyelinating peripheral neuropathy	Corneal erosion	Wide intermamillary distance
Hyperreflexia	Distal sensory impairment	Corneal opacity	RESPIRATORY SYSTEM
Hypokinesia	Dysarthria	Developmental cataract	Apnea
Hyporeflexia	Encephalopathy	Downslanted palpebral fissures	Asthma
Inability to walk	Headache	Ectopia lentis	Bronchial wall thickening
Incoordination	Increased intracranial pressure	Epicanthus	Bronchiectasis
Involuntary movements	Lhermitte's sign	Esotropia	Central apnea
Loss of ambulation	Migraine	External ophthalmoplegia	Cyanosis
Motor stereotypy	Mononeuropathy	Foveal hyperpigmentation	Desquamative interstitial pneumonitis
	Optic ataxia	Hyperpigmentation of the fundus	Dyspnea
	Peripheral neuropathy	Hypertelorism	Hyperventilation
	Polyneuropathy	Hypopigmentation of the fundus	Hypoventilation

RESPIRATORY SYSTEM (cont.)		ABDOMEN	URINARY SYSTEM (cont.)	HAIR AND NAIL (cont.)
Hypoxemia		Abdominal pain	Ureteropelvic junction obstruction	Sparse body hair
Interstitial pneumonitis		Ascites	Vesicoureteral reflux	Sparse eyebrow
Intraalveolar phospholipid accumulation		Asplenia	REPRODUCTIVE SYSTEM	Sparse eyelashes
Intralobular septal thickening		Biliary cirrhosis	Ambiguous genitalia	Sparse hair
Neonatal respiratory distress		Cholangitis	Bifid scrotum	Sparse scalp hair
Pleural effusion		Cholelithiasis	Clitoral hypoplasia	Synophrys
Pleuritis		Cholestasis	Cryptorchidism	Thick eyebrow
Pneumothorax		Chronic diarrhea	Gonadal dysgenesis	Trichorrhexis nodosa
Pulmonary fibrosis		Cirrhosis	Hyperplastic labia majora	Woolly hair
Pulmonary hypoplasia		Diastasis recti	Hypoplasia of the uterus	SOFT TISSUE
Recurrent bronchitis		Exocrine pancreatic insufficiency	Hypoplastic labia majora	Abnormal muscle fiber dystrophin expression
Respiratory failure		Hepatic cysts	Inguinal hernia	Achilles tendon contracture
Respiratory insufficiency		Hepatic fibrosis	Micropenis	Antecubital pterygium
HEART		Malformation of the hepatic ductal plate	Precocious puberty	Axillary pterygium
Abnormal cardiac septum morphology		Pancreatic cysts	Shawl scrotum	Bulbar palsy
Abnormal heart morphology		Pancreatic fibrosis	Uterine prolapse	Centrally nucleated skeletal muscle fibers
Aortic root aneurysm		Peritonitis	SKIN	Deposits immunoreactive to β -amyloid protein
Atrial septal defect		Polysplenia	Abnormal blistering of the skin	Distal arthrogryposis
Bicuspid aortic valve		Protuberant abdomen	Acanthosis nigricans	Distal lower limb amyotrophy
Conotruncal defect		Umbilical hernia	Acne inversa	Distal muscle weakness
Dextrocardia		GASTROINTESTINAL SYSTEM	Anhidrosis	Elbow flexion contracture
Dilated cardiomyopathy		Anal atresia	Aplasia cutis congenita	Fatty replacement of skeletal muscle
Double outlet right ventricle		Anal stenosis	Axillary freckling	Flexion contracture
Hypertrophic cardiomyopathy		Constipation	Cafe-au-lait spot	Generalized muscle weakness
Hypoplastic left heart		Diarrhea	Cutis laxa	Gowers sign
Mitral stenosis		Duodenal stenosis	Eczema	Hypertonia
Mitral valve prolapse		Dysphagia	Erythema	Hypotonia
Noncompaction cardiomyopathy	Patent foramen ovale	Esophageal atresia	Fragile skin	Increased variability in muscle fiber diameter
Pericardial effusion		Feeding difficulties	Hyperextensible skin	Joint hypermobility
Pulmonic stenosis		Gastroesophageal reflux	Hyperpigmentation of the skin	Minicore myopathy
Restrictive cardiomyopathy		Hepatomegaly	Hypohidrosis	Muscle fiber cytoplasmatic inclusion bodies
Transposition of the great arteries		Inflammation of the large intestine	Hypopigmentation of the skin	Muscle hypertrophy of the lower extremities
Tricuspid regurgitation		Intrahepatic biliary atresia	Ichthyosis	Muscle spasm
Ventricular septal defect		Laryngomalacia	Intermittent generalized erythematous papular rash	Myalgia
CARDIOVASCULAR SYSTEM		Nausea	Jaundice	Myopathic facies
Abnormal cardiac test		Pancreatitis	Keloids	Myopathy
Aortic aneurysm		Pyloric stenosis	Lichenoid skin lesion	Nemaline bodies
Aortic dissection		Rectal prolapse	Oral mucosal blisters	Popliteal pterygium
Aortic regurgitation		Splenomegaly	Palmoplantar blistering	Rhabdomyolysis
Aortic valve stenosis		Tracheoesophageal fistula	Palmoplantar hyperkeratosis	Shoulder girdle muscle atrophy
Arrhythmia		Vomiting	Palmoplantar keratoderma	Skeletal muscle atrophy
URINARY SYSTEM			Psoriasisiform dermatitis	Skeletal muscle hypertrophy
Arteriovenous malformation		Cystic renal dysplasia	Pulmonary capillary hemangiomatosis	Type 1 muscle fiber predominance
Atrioventricular block		Decreased numbers of nephrons	Single transverse palmar crease	Type 2 muscle fiber predominance
Bradycardia		Ectopic kidney	Skin rash	Upper limb muscle hypertrophy
Carotid artery calcification		Glomerular sclerosis	Soft skin	Weakness of facial musculature
Carotid artery dissection		Glomerulonephritis	Subepidermal blistering	SKELETAL: THORAX
Coarctation of aorta		Hematuria	Telangiectasia	11 pairs of ribs
Cutis marmorata		Horseshoe kidney	Urticaria	Abnormal clavicle morphology
Hypertension		Hydronephrosis	Vascular skin abnormality	Aplasia/Hypoplasia of the clavicles
Mitral regurgitation		Hypospadias	Vitiligo	Aplasia/Hypoplasia of the ribs
Patent ductus arteriosus		Multiple glomerular cysts	HAIR AND NAIL	Aplasia/Hypoplasia of the sternum
Prolonged QT interval		Nephrolithiasis	Abnormality of the nail	Barrel-shaped chest
Pulmonary arterial hypertension		Polycystic kidney dysplasia	Absent eyelashes	Bifid sternum
Pulmonary venous hypertension		Polyuria	Alopecia	Broad clavicles
Stroke		Proteinuria	Anonychia	Cuffed ribs
Supraventricular arrhythmia		Proximal tubulopathy	Coarse hair	Enlarged thorax
Syncope		Renal agenesis	Hyperconvex fingernails	Flaring of rib cage
Tachycardia		Renal amyloidosis	Hypertrichosis	Pectus carinatum
Tetralogy of Fallot		Renal cortical cysts	Keratosis pilaris	Pectus excavatum
Total anomalous pulmonary venous		Renal dysplasia	Long eyelashes	Prominent sternum
Vascular dilatation		Renal insufficiency	Low anterior hairline	Rib fusion
Ventricular arrhythmia			Lumbosacral hirsutism	Short clavicles
Ventricular tachycardia			Nail dystrophy	Short ribs

SKELETAL: THORAX (contd.)

Supernumerary ribs
Thin clavicles
SKELETAL: SPINE AND PELVIS
Cervical spina bifida
Coronal cleft vertebrae
Hip dislocation
Hyperlordosis
Hypoplastic pubic rami
Kyphoscoliosis
Kyphosis
Lumbar hyperlordosis
Myelomeningocele
Narrow iliac wing
Platyspondyly
Scoliosis
Shallow acetabular fossae
Spina bifida
Spina bifida occulta
Spinal canal stenosis
Spinal rigidity
Spondyloepiphyseal dysplasia
Vertebral fusion
Vertebral segmentation defect

SKELETAL: LIMBS

Bifid femur
Bowing of the legs
Coxa valga
Disproportionate short-limb short stature
Fibular aplasia
Fibular hypoplasia
Fibular overgrowth
Flat capital femoral epiphysis
Genu valgum
Hypoplasia of the ulna
Mesomelia
Mesomelic arm shortening
Mesomelic leg shortening
Mesomelic short stature
Patellar hypoplasia
Radioulnar synostosis
Rhizomelia
Rhizomelic arm shortening
Rhizomelic leg shortening
Short femoral neck
Short humerus
Short tibia
Stippled calcification proximal humeral epiphyses
Tibial bowing
Widened proximal tibial metaphyses

SKELETAL: HANDS AND FEET

Aplasia/Hypoplasia of metatarsal bones
Aplasia/Hypoplasia of the hallux
Aplasia/Hypoplasia of the radius
Brachydactyly
Broad hallux
Broad thumb
Carpal bone hypoplasia
Clubbing of fingers
Clubbing of toes
Drumstick terminal phalanges
Foot monodactyly
Hammertoe
Hand monodactyly
Large carpal bones
Large hands

SKELETAL: HANDS AND FEET (cont.)

Metatarsus adductus
Patellar dislocation
Pes planus
Polydactyly
Postaxial hand polydactyly
Prominent fingertip pads
Proximal placement of thumb
Short distal phalanx of finger
Short metacarpal
Short metatarsal
Short phalanx of finger
Split foot
Split hand
Syndactyly
Synostosis of metacarpals/metatarsals
Talipes equinovalgus
Talipes equinovarus
Tapered finger
Triphalangeal thumb

ENDOCRINE SYSTEM

Decreased response to growth hormone stimulation test
Delayed puberty
Diabetes insipidus
Diabetes mellitus
Hypercalcemia
Hypergonadotropic hypogonadism
Hyperparathyroidism
Hyperphosphatemia
Hyperthyroidism
Hypocalcemia
Hypoparathyroidism
Hypophosphatemia
Hypothyroidism
Lymphedema
Maturity-onset diabetes of the young
Rickets

HEMATOPOIETIC SYSTEM

Abnormal bleeding
Anemia
Anisocytosis
Bone marrow hypercellularity
Bruising susceptibility
Epistaxis
Gastrointestinal hemorrhage
Hemolytic anemia
Hypochromic anemia
Hypoplastic anemia
Macrocytic anemia
Megaloblastic anemia
Menorrhagia
Microangiopathic hemolytic anemia
Microcytic anemia
Pancytopenia
Poikilocytosis
Prolonged bleeding time
Sideroblastic anemia
Stomatocytosis
Thrombocytopenia

IMMUNE SYSTEM

Abnormal T cell morphology
Agammaglobulinemia
Allergic rhinitis
B lymphocytopenia

IMMUNE SYSTEM (cont.)

Decreased circulating antibody level
Decreased circulating IgA level
Decreased circulating IgG level
Decreased circulating total IgM
Decreased proportion of CD4-positive helper T cells
Eczematoid dermatitis
Folliculitis
Hashimoto thyroiditis
Hemophagocytosis
Immunodeficiency
Leukopenia
Lymphadenopathy
Lymphopenia
Myelokathexis
Neutropenia
Panhypogammaglobulinemia
Predominantly dermal neutrophilic infiltrate
Pyoderma gangrenosum
Rectal abscess
Recurrent bacterial infections
Recurrent enteroviral infections
Recurrent infections
Recurrent otitis media
Recurrent pneumonia
Recurrent respiratory infections
Recurrent sinusitis
Reticulocytosis
Schistocytosis
Seborheic dermatitis
Verrucae
METABOLISM
Abnormal activity of mitochondrial respiratory chain
Abnormality of mitochondrial metabolism
Amyloidosis
Conjugated hyperbilirubinemia
Decreased adenosylcobalamin
Decreased methylcobalamin
Elevated circulating creatine kinase concentration
Elevated circulating creatinine concentration
Elevated circulating glutaric acid concentration
Elevated hepatic transaminase
Elevated plasma branched chain amino acids
Ethylmalonic aciduria
Generalized aminoaciduria
Glutaric aciduria
Glycosuria
Homocystinuria
Hyperammonemia
Hyperbilirubinemia
Hypercholesterolemia
Hyperglycemia
Hyperglycinemia
Hyperinsulinemia
Hyperketonemia
Hypertriglyceridemia
Hyperuricemia
Hypoammonemia
Hypoglycemia
Increased blood urea nitrogen
Increased serum pyruvate
Ketonuria
Lactic acidosis
Metabolic acidosis
Metabolic ketoacidosis