

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
Cystic hygroma	Postural instability	Vertigo	Microphthalmia
Encephalocele	Progressive muscle weakness	CRANIOFACIAL/DYSMORPHISM	Myopia
Failure to thrive	Proximal muscle weakness	Abnormal facial shape	Nystagmus
Growth delay	Spasticity	Abnormality of malar bones	Ophthalmoparesis
Hydrops fetalis	Tip-toe gait	Anosmia	Optic atrophy
Increased nuchal translucency	Torticollis	Anteverted nares	Optic neuropathy
Infantile spasms	Tremor	Bifid uvula	Posterior embryotoxon
Intrauterine growth retardation	Waddling gait	Brachycephaly	Proptosis
Low APGAR score	NEURAL MORPHOLOGY	Choanal atresia	Pterygium
Nonimmune hydrops fetalis	Abnormal brainstem morphology	Cleft lip	Ptosis
Oligohydramnios	Abnormal corpus callosum morphology	Cleft palate	Reduced visual acuity
Omphalocele	Abnormal myelination	Coarse facial features	Retinal atrophy
Polyhydramnios	Abnormal periventricular white matter morphology	Craniosynostosis	Retinal degeneration
Postnatal growth retardation	Abnormality of the basal ganglia	Dental enamel pits	Retinal detachment
Premature birth	Aplasia/Hypoplasia of the cerebellar vermis	Dental malocclusion	Retinal dystrophy
Prolonged neonatal jaundice	Aplasia/Hypoplasia of the cerebellum	Depressed nasal bridge	Retinoschisis
DEVELOPMENT AND BEHAVIOR	Brain stem compression	Depressed nasal tip	Rod-cone dystrophy
Absent speech	Cerebellar atrophy	Dolichocephaly	Strabismus
Aggressive behavior	Cerebral cortical atrophy	Facial asymmetry	Telecanthus
Anxiety	CNS demyelination	Frontal bossing	Upplanted palpebral fissure
Apathy	CNS hypomyelination	High palate	Vitreoretinopathy
Attention deficit hyperactivity disorder	Cortical dysplasia	Hypodontia	EAR DEFECTS/HEARING
Autistic behavior	Cortical tubers	Macrocephaly	Conductive hearing impairment
Cognitive impairment	Delayed CNS myelination	Malar flattening	Cupped ear
Delayed speech and language development	Dysgenesis of the basal ganglia	Mandibular prognathia	High-frequency hearing impairment
Depression	Encephalocele	Microcephaly	Hypoplasia of the semicircular canal
Developmental regression	Gliosis	Micrognathia	Low-frequency hearing loss
Dysarthria	Gray matter heterotopia	Midface retrusion	Low-set ears
Dyscalculia	Holoprosencephaly	Narrow palate	Microtia
Dyslexia	Hydrocephalus	Retrognathia	Mild hearing impairment
Global developmental delay	Hyperintensity of cerebral white matter on MRI	Short philtrum	Posteriorly rotated ears
Hyperactivity	Hypointensity of cerebral white matter on MRI	Sloping forehead	Preauricular pit
Impaired visuospatial constructive cognition	Hypoplasia of the corpus callosum	Smooth philtrum	Protruding ear
Intellectual disability	Leukodystrophy	Thick nasal alae	Sensorineural hearing impairment
Lethargy	Lissencephaly	Thin upper lip vermillion	Unilateral deafness
Memory impairment	Pachygryia	Trigonocephaly	NECK AND CHEST
Obsessive-compulsive behavior	Periventricular leukomalacia	Wide anterior fontanel	Chest pain
Polydipsia	Periventricular nodular heterotopia	Wide nose	Cystic hygroma
Restrictive behavior	Periventricular white matter hyperintensities	Widely spaced teeth	Gynecomastia
Sleep disturbance	Polymicrogyria	EYE DEFECTS/VISION	Heterotaxy
Speech articulation difficulties	Tethered cord	Amblyopia	Inverted nipples
MOTOR FUNCTION	Ventriculomegaly	Anophthalmia	Laryngeal stenosis
Akinesia	NEURAL PHYSIOLOGY	Blepharitis	Neck flexor weakness
Areflexia	Abnormal pineal melatonin secretion	Blepharophimosis	Neck muscle weakness
Ataxia	Agnosia	Cataract	Neck pterygia
Athetosis	Decreased intracranial pressure	Cerebral visual impairment	Shield chest
Cerebral palsy	Decreased motor nerve conduction velocity	Coloboma	Short neck
Chorea	Decreased nerve conduction velocity	Conjunctivitis	Supernumerary nipple
Dyskinesia	Decreased sensory nerve conduction velocity	Corneal dystrophy	Webbed neck
Dysmetria	Demyelinating peripheral neuropathy	Corneal erosion	Wide intermamillary distance
Dystonia	Distal sensory impairment	Corneal opacity	RESPIRATORY SYSTEM
Gait disturbance	Dysarthria	Developmental cataract	Apnea
Hemiplegia	Encephalopathy	Downslanted palpebral fissures	Asthma
Hyperreflexia	Headache	Ectopia lentis	Bronchial wall thickening
Hypokinesia	Increased intracranial pressure	Epicanthus	Bronchiectasis
Hyporeflexia	Lhermitte's sign	Esotropia	Central apnea
Inability to walk	Migraine	External ophthalmoplegia	Cyanosis
Incoordination	Mononeuropathy	Foveal hyperpigmentation	Desquamative interstitial pneumonitis
Involuntary movements	Optic ataxia	Hyperpigmentation of the fundus	Dyspnea
Loss of ambulation	Peripheral neuropathy	Hypertelorism	Hyperventilation
Motor stereotypy	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	Hypospadias	Achilles tendon contracture
Respiratory insufficiency		Hepatic fibrosis	Inguinal hernia	Antecubital pterygium
Tracheoesophageal fistula		Hepatomegaly	Micropenis	Axillary pterygium
HEART		Ingual hernia	Precocious puberty	Bulbar palsy
Abnormal cardiac septum morphology		Malformation of the hepatic ductal plate	Shawl scrotum	Centrally nucleated skeletal muscle fibers
Abnormal heart morphology		Pancreatic cysts	Uterine prolapse	Deposits immunoreactive to β -amyloid protein
Aortic root aneurysm		Pancreatic fibrosis	SKIN	Distal arthrogryposis
Atrial septal defect		Peritonitis	Abnormal blistering of the skin	Distal lower limb amyotrophy
Bicuspid aortic valve		Polysplenia	Acanthosis nigricans	Distal muscle weakness
Conotruncal defect		Protuberant abdomen	Acne inversa	Elbow flexion contracture
Dextrocardia		Splenomegaly	Anhidrosis	Fatty replacement of skeletal muscle
Dilated cardiomyopathy		Umbilical hernia	Aplasia cutis congenita	Flexion contracture
Double outlet right ventricle		GASTROINTESTINAL SYSTEM	Axillary freckling	Generalized muscle weakness
Hypertrophic cardiomyopathy		Anal atresia	Cafe-au-lait spot	Gowers sign
Hypoplastic left heart		Anal stenosis	Cutis laxa	Hypertonia
Mitral stenosis		Constipation	Eczema	Hypotonia
Mitral valve prolapse		Diarrhea	Erythema	Increased variability in muscle fiber diameter
Noncompaction cardiomyopathy		Duodenal stenosis	Fragile skin	Joint hypermobility
Patent foramen ovale		Dysphagia	Hyperextensible skin	Minicore myopathy
Pericardial effusion		Esophageal atresia	Hyperpigmentation of the skin	Muscle fiber cytoplasmatic inclusion bodies
Pulmonic stenosis		Feeding difficulties	Hypohidrosis	Muscle hypertrophy of the lower extremities
Restrictive cardiomyopathy		Gastroesophageal reflux	Hypopigmentation of the skin	Muscle spasm
Transposition of the great arteries		Hepatomegaly	Ichthyosis	Myalgia
Tricuspid regurgitation		Inflammation of the large intestine	Intermittent generalized erythematous papular rash	Myopathic facies
Ventricular septal defect		Intrahepatic biliary atresia	Jaundice	Myopathy
CARDIOVASCULAR SYSTEM		Laryngomalacia	Keloids	Nemaline bodies
Abnormal cardiac test		Nausea	Lichenoid skin lesion	Popliteal pterygium
Aortic aneurysm		Pancreatitis	Oral mucosal blisters	Rhabdomyolysis
Aortic dissection		Pyloric stenosis	Palmoplantar blistering	Shoulder girdle muscle atrophy
Aortic regurgitation		Rectal prolapse	Palmoplantar hyperkeratosis	Skeletal muscle atrophy
Aortic valve stenosis		Splenomegaly	Palmoplantar keratoderma	Skeletal muscle hypertrophy
Arrhythmia		Tracheoesophageal fistula	Psoriasiform dermatitis	Type 1 muscle fiber predominance
Arteriovenous malformation		Vomiting	Pulmonary capillary hemangiomatosis	Type 2 muscle fiber predominance
Atrioventricular block		URINARY SYSTEM	Single transverse palmar crease	Upper limb muscle hypertrophy
Bradycardia		Cystic renal dysplasia	Skin rash	Weakness of facial musculature
Carotid artery calcification		Decreased numbers of nephrons	Soft skin	SKELETAL: THORAX
Carotid artery dissection		Ectopic kidney	Subepidermal blistering	11 pairs of ribs
Coarctation of aorta		Glomerular sclerosis	Telangiectasia	Abnormal clavicle morphology
Cutis marmorata		Glomerulonephritis	Urticaria	Aplasia/Hypoplasia of the clavicles
Hypertension		Hematuria	Vascular skin abnormality	Aplasia/Hypoplasia of the ribs
Mitral regurgitation		Horseshoe kidney	Vitiligo	Aplasia/Hypoplasia of the sternum
Patent ductus arteriosus		Hydronephrosis	HAIR AND NAIL	Barrel-shaped chest
Prolonged QT interval		Hypospadias	Abnormality of the nail	Bifid sternum
Pulmonary arterial hypertension		Multiple glomerular cysts	Absent eyelashes	Broad clavicles
Pulmonary venous hypertension		Nephrolithiasis	Alopecia	Cuffed ribs
Stroke		Polycystic kidney dysplasia	Anonychia	Enlarged thorax
Supraventricular arrhythmia		Polyuria	Coarse hair	Flaring of rib cage
Syncope		Proteinuria	Hyperconvex fingernails	Pectus carinatum
Tachycardia		Proximal tubulopathy	Hypertrichosis	Pectus excavatum
Tetralogy of Fallot		Renal agenesis	Keratosis pilaris	Prominent sternum
Total anomalous pulmonary venous		Renal amyloidosis	Long eyelashes	Rib fusion
Vascular dilatation		Renal cortical cysts	Low anterior hairline	Short clavicles
Ventricular arrhythmia		Renal dysplasia	Lumbosacral hirsutism	Short ribs
Ventricular tachycardia		Renal insufficiency	Nail dystrophy	Short sternum

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
Eczema
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
Seborrheic 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