

PERINATAL HISTORY
Abnormal placenta morphology
Abnormality of the umbilical cord
Breech presentation
Congenital diaphragmatic hernia
Failure to thrive
Growth delay
Hydrops fetalis
Increased nuchal translucency
Infantile spasms
Intrauterine growth retardation
Low APGAR score
Nonimmune hydrops fetalis
Oligohydramnios
Omphalocele
Polyhydramnios
Postnatal growth retardation
Premature birth
Prolonged neonatal jaundice
DEVELOPMENT AND BEHAVIOR
Absent speech
Aggressive behavior
Anxiety
Apathy
Attention deficit hyperactivity disorder
Autistic behavior
Cognitive impairment
Delayed speech and language development
Depression
Developmental regression
Dyscalculia
Dyslexia
Global developmental delay
Hyperactivity
Impaired visuospatial constructive cognition
Intellectual disability
Lethargy
Memory impairment
Obsessive-compulsive behavior
Polydipsia
Restrictive behavior
Sleep disturbance
Speech articulation difficulties

MOTOR FUNCTION
Akinesia
Areflexia
Ataxia
Athetosis
Cerebral palsy
Chorea
Dyskinesia
Dysmetria
Dystonia
Gait disturbance
Hemiplegia
Hyperreflexia
Hypokinesia
Hyporeflexia
Inability to walk
Incoordination
Involuntary movements
Loss of ambulation
Motor stereotypy

MOTOR FUNCTION (cont.)
Myoclonus
Paraplegia
Parkinsonism
Poor suck
Postural instability
Progressive muscle weakness
Proximal muscle weakness
Spasticity
Tip-toe gait
Torticollis
Tremor
Waddling gait

NEURAL MORPHOLOGY
Abnormal brainstem morphology
Abnormal corpus callosum morphology
Abnormal myelination
Abnormal periventricular white matter morphology
Abnormality of the basal ganglia
Aplasia/Hypoplasia of the cerebellar vermis
Aplasia/Hypoplasia of the cerebellum
Brain stem compression
Cerebellar atrophy
Cerebral cortical atrophy
CNS demyelination
CNS hypomyelination
Cortical dysplasia
Cortical tubers
Delayed CNS myelination
Dysgenesis of the basal ganglia
Encephalocele
Gliososis
Gray matter heterotopia
Holoprosencephaly
Hydrocephalus
Hyperintensity of cerebral white matter on MRI
Hypointensity of cerebral white matter on MRI
Hypoplasia of the corpus callosum
Leukodystrophy
Lissencephaly
Pachygyria
Periventricular leukomalacia
Periventricular nodular heterotopia
Periventricular white matter hyperintensities
Polymicrogyria
Tethered cord
Ventriculomegaly

NEURAL PHYSIOLOGY
Abnormal pineal melatonin secretion
Agnosia
Decreased intracranial pressure
Decreased motor nerve conduction velocity
Decreased nerve conduction velocity
Decreased sensory nerve conduction velocity
Demyelinating peripheral neuropathy
Distal sensory impairment
Dysarthria
Encephalopathy
Headache
Increased intracranial pressure
Lhermitte's sign
Migraine
Mononeuropathy
Optic ataxia
Peripheral neuropathy
Polyneuropathy

NEURAL PHYSIOLOGY (cont.)
Psychosis
Seizure
Sensorimotor neuropathy
Sensory neuropathy
Vertigo
CRANIOFACIAL/DYSMORPHISM
Abnormal facial shape
Abnormality of malar bones
Anosmia
Anteverted nares
Bifid uvula
Brachycephaly
Choanal atresia
Cleft lip
Cleft palate
Coarse facial features
Craniosynostosis
Dental enamel pits
Dental malocclusion
Depressed nasal bridge
Depressed nasal tip
Dolichocephaly
Facial asymmetry
Frontal bossing
High palate
Hypodontia
Macrocephaly
Malar flattening
Mandibular prognathia
Microcephaly
Micrognathia
Midface retrusion
Narrow palate
Retrognathia
Short philtrum
Sloping forehead
Smooth philtrum
Thick nasal alae
Thin upper lip vermillion
Trigonocephaly
Wide anterior fontanel
Wide nose
Widely spaced teeth

EYE DEFECTS/VISION
Amblyopia
Anphthalmia
Blepharitis
Blepharophimosis
Cataract
Cerebral visual impairment
Coloboma
Conjunctivitis
Corneal dystrophy
Corneal erosion
Corneal opacity
Developmental cataract
Downslanted palpebral fissures
Ectopia lentis
Epicanthus
Esotropia
External ophthalmoplegia
Foveal hyperpigmentation
Hyperpigmentation of the fundus
Hypertelorism
Hypopigmentation of the fundus

EYE DEFECTS/VISION (cont.)
Lacrimal duct stenosis
Macular atrophy
Macular degeneration
Macular purpura
Microphthalmia
Myopia
Nystagmus
Ophthalmoparesis
Optic atrophy
Optic neuropathy
Posterior embryotoxon
Proptosis
Pterygium
Ptosis
Reduced visual acuity
Retinal atrophy
Retinal degeneration
Retinal detachment
Retinal dystrophy
Retinoschisis
Rod-cone dystrophy
Strabismus
Telecanthus
Upslanted palpebral fissure
Vitreoretinopathy

EAR DEFECTS/HEARING
Conductive hearing impairment
Cupped ear
High-frequency hearing impairment
Hypoplasia of the semicircular canal
Low-frequency hearing loss
Low-set ears
Microtia
Mild hearing impairment
Posteriorly rotated ears
Preauricular pit
Protruding ear
Sensorineural hearing impairment
Unilateral deafness

NECK AND CHEST
Chest pain
Cystic hygroma
Gynecomastia
Heterotaxy
Inverted nipples
Laryngeal stenosis
Neck flexor weakness
Neck muscle weakness
Neck pterygia
Shield chest
Short neck
Supernumerary nipple
Webbed neck
Wide intermamillary distance

RESPIRATORY SYSTEM
Apnea
Asthma
Bronchial wall thickening
Bronchiectasis
Central apnea
Cyanosis
Desquamative interstitial pneumonitis
Dyspnea
Hyperventilation
Hypoventilation

RESPIRATORY SYSTEM (cont.)

Hypoxemia
Interstitial pneumonitis
Intraalveolar phospholipid accumulation
Intralobular septal thickening
Neonatal respiratory distress
Pleural effusion
Pleuritis
Pneumothorax
Pulmonary fibrosis
Pulmonary hypoplasia
Recurrent bronchitis
Respiratory failure
Respiratory insufficiency

HEART

Abnormal cardiac septum morphology
Abnormal heart morphology
Aortic root aneurysm
Atrial septal defect
Bicuspid aortic valve
Conotruncal defect
Dextrocardia
Dilated cardiomyopathy
Double outlet right ventricle
Hypertrophic cardiomyopathy
Hypoplastic left heart
Mitral stenosis
Mitral valve prolapse
Noncompaction cardiomyopathy
Patent foramen ovale
Pericardial effusion
Pulmonic stenosis
Restrictive cardiomyopathy
Transposition of the great arteries
Tricuspid regurgitation
Ventricular septal defect

CARDIOVASCULAR SYSTEM

Abnormal cardiac test
Aortic aneurysm
Aortic dissection
Aortic regurgitation
Aortic valve stenosis
Arrhythmia
Arteriovenous malformation
Atrioventricular block
Bradycardia
Carotid artery calcification
Carotid artery dissection
Coarctation of aorta
Cutis marmorata
Hypertension
Mitral regurgitation
Patent ductus arteriosus
Prolonged QT interval
Pulmonary arterial hypertension
Pulmonary venous hypertension
Stroke
Supraventricular arrhythmia
Syncope
Tachycardia
Tetralogy of Fallot
Total anomalous pulmonary venous
Vascular dilatation
Ventricular arrhythmia
Ventricular tachycardia

ABDOMEN

Abdominal pain
Ascites
Asplenia
Biliary cirrhosis
Cholangitis
Cholelithiasis
Cholestasis
Chronic diarrhea
Cirrhosis
Diastasis recti
Exocrine pancreatic insufficiency
Hepatic cysts
Hepatic fibrosis
Malformation of the hepatic ductal plate
Pancreatic cysts
Pancreatic fibrosis
Peritonitis
Polysplenia
Protuberant abdomen
Umbilical hernia

GASTROINTESTINAL SYSTEM

Anal atresia
Anal stenosis
Constipation
Diarrhea
Duodenal stenosis
Dysphagia
Esophageal atresia
Feeding difficulties
Gastroesophageal reflux
Hepatomegaly
Inflammation of the large intestine
Intrahepatic biliary atresia
Laryngomalacia
Nausea
Pancreatitis
Pyloric stenosis
Rectal prolapse
Splenomegaly
Tracheoesophageal fistula
Vomiting

URINARY SYSTEM

Cystic renal dysplasia
Decreased numbers of nephrons
Ectopic kidney
Glomerular sclerosis
Glomerulonephritis
Hematuria
Horseshoe kidney
Hydronephrosis
Hypospadias
Multiple glomerular cysts
Nephrolithiasis
Polycystic kidney dysplasia
Polyuria
Proteinuria
Proximal tubulopathy
Renal agenesis
Renal amyloidosis
Renal cortical cysts
Renal dysplasia
Renal insufficiency

URINARY SYSTEM (cont.)

Ureteropelvic junction obstruction
Vesicoureteral reflux
REPRODUCTIVE SYSTEM
Ambiguous genitalia
Bifid scrotum
Clitoral hypoplasia
Cryptorchidism
Gonadal dysgenesis
Hyperplastic labia majora
Hypoplasia of the uterus
Hypoplastic labia majora
Inguinal hernia
Micropenis
Precocious puberty
Shawl scrotum
Uterine prolapse

SKIN

Abnormal blistering of the skin
Acanthosis nigricans
Acne inversa
Anhidrosis
Aplasia cutis congenita
Axillary freckling
Cafe-au-lait spot
Cutis laxa
Eczema
Erythema
Fragile skin
Hyperextensible skin
Hyperpigmentation of the skin
Hypohidrosis
Hypopigmentation of the skin
Ichthyosis
Intermittent generalized erythematous papular rash
Jaundice
Keloids
Lichenoid skin lesion
Oral mucosal blisters
Palmoplantar blistering
Palmoplantar hyperkeratosis
Palmoplantar keratoderma
Psoriasiform dermatitis
Pulmonary capillary hemangiomatosis
Single transverse palmar crease
Skin rash
Soft skin
Subepidermal blistering
Telangiectasia
Urticaria
Vascular skin abnormality
Vitiligo

HAIR AND NAIL

Abnormality of the nail
Absent eyelashes
Alopecia
Anonychia
Coarse hair
Hyperconvex fingernails
Hypertrichosis
Keratosis pilaris
Long eyelashes
Low anterior hairline
Lumbosacral hirsutism
Nail dystrophy

HAIR AND NAIL (cont.)

Sparse body hair
Sparse eyebrow
Sparse eyelashes
Sparse hair
Sparse scalp hair
Synophrys
Thick eyebrow
Trichorrhexis nodosa
Woolly hair

SOFT TISSUE

Abnormal muscle fiber dystrophin expression
Achilles tendon contracture
Antecubital pterygium
Axillary pterygium
Bulbar palsy
Centrally nucleated skeletal muscle fibers
Deposits immunoreactive to β -amyloid protein
Distal arthrogryposis
Distal lower limb amyotrophy
Distal muscle weakness
Elbow flexion contracture
Fatty replacement of skeletal muscle
Flexion contracture
Generalized muscle weakness
Gowers sign
Hypertonia
Hypotonia
Increased variability in muscle fiber diameter
Joint hypermobility
Minicore myopathy
Muscle fiber cytoplasmatic inclusion bodies
Muscle hypertrophy of the lower extremities
Muscle spasm
Myalgia
Myopathic facies
Myopathy
Nemaline bodies
Popliteal pterygium
Rhabdomyolysis
Shoulder girdle muscle atrophy
Skeletal muscle atrophy
Skeletal muscle hypertrophy
Type 1 muscle fiber predominance
Type 2 muscle fiber predominance
Upper limb muscle hypertrophy
Weakness of facial musculature
SKELETAL: THORAX
11 pairs of ribs
Abnormal clavicle morphology
Aplasia/Hypoplasia of the clavicles
Aplasia/Hypoplasia of the ribs
Aplasia/Hypoplasia of the sternum
Barrel-shaped chest
Bifid sternum
Broad clavicles
Cupped ribs
Enlarged thorax
Flaring of rib cage
Pectus carinatum
Pectus excavatum
Prominent sternum
Rib fusion
Short clavicles
Short ribs
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 equinovaglus

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

Type I diabetes mellitus

Type II diabetes mellitus

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

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