

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	Upslanted 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	Gliososis	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	Pachygyria	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.)

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
Tracheoesophageal fistula

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
Hepatomegaly
Inguinal hernia
Malformation of the hepatic ductal plate

Pancreatic cysts
Pancreatic fibrosis
Peritonitis
Polysplenia
Protuberant abdomen
Splenomegaly
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
Hypospadias
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

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