Table 10-2 Information to Be Sought during the Physical Examination of a Child with Suspected Developmental Disabilities

ITEM	POSSIBLE SIGNIFICANCE
General appearance	May indicate significant delay in development or obvious syndrome
STATURE	
Short stature	Williams syndrome, malnutrition, Turner syndrome; many children with severe retardation have associated short stature.
Obesity	Prader-Willi syndrome
Large stature	Sotos syndrome
HEAD	
Macrocephaly	Alexander syndrome, Sotos syndrome, gangliosidosis, hydrocephalus, mucopolysaccharidosis, subdural effusion
Microcephaly	Virtually any condition that can retard brain growth (e.g., malnutrition, Angelman syndrome, de Lange syndrome, fetal alcohol effects)
FACE	
Coarse, triangular, round, or flat face; hypotelorism or hypertelorism, slanted or short palpebral fissure; unusual nose, maxilla, and mandible	Specific measurements may provide clues to inherited, metabolic, or other diseases such as fetal alcohol syndrome, cri du chat syndrome (5p- syndrome), or Williams syndrome.
EYES	
Prominent	Crouzon syndrome, Seckel syndrome, fragile X syndrome
Cataract	Galactosemia, Lowe syndrome, prenatal rubella, hypothyroidism
Cherry-red spot in macula	Gangliosidosis (GM_1), metachromatic leukodystrophy, mucolipidosis, Tay-Sachs disease, Niemann-Pick disease, Farber lipogranulomatosis, sialidosis III
Chorioretinitis	Congenital infection with cytomegalovirus, toxoplasmosis, or rubella
Corneal cloudiness	Mucopolysaccharidosis I and II, Lowe syndrome, congenital syphilis
EARS	
Pinnae, low set or malformed	Trisomies such as 18, Rubinstein-Taybi syndrome, Down syndrome, CHARGE association, cerebro-oculo-facio-skeletal syndrome, fetal phenytoin effects
Hearing	Loss of acuity in mucopolysaccharidosis; hyperacusis in many encephalopathies
HEART	
Structural anomaly or hypertrophy	CHARGE association, CATCH-22, velocardiofacial syndrome, glycogenosis II, fetal alcohol effects, mucopolysaccharidosis I; chromosomal anomalies such as Down syndrome; maternal phenylketonuria; chronic cyanosis may impair cognitive development.
LIVER	
Hepatomegaly	Fructose intolerance, galactosemia, glycogenosis types I to IV, mucopolysaccharidosis I and II, Niemann-Pick disease, Tay-Sachs disease, Zellweger syndrome, Gaucher disease, ceroid lipofuscinosis, gangliosidosis
GENITALIA	
Macro-orchidism (usually not noted until puberty)	Fragile X syndrome
Hypogenitalism	Prader-Willi syndrome, Klinefelter syndrome, CHARGE association
EXTREMITIES	
Hands, feet, dermatoglyphics, and creases	May indicate specific entity such as Rubinstein-Taybi syndrome or be associated with chromosomal anomaly
Joint contractures	Sign of muscle imbalance around joints (e.g., with meningomyelocele, cerebral palsy, arthrogryposis, muscular dystrophy; also occurs with cartilaginous problems such as mucopolysaccharidosis)
SKIN	
Café au lait spots	Neurofibromatosis, tuberous sclerosis, Bloom syndrome
Eczema	Phenylketonuria, histiocytosis
Hemangiomas and telangiectasia	Sturge-Weber syndrome, Bloom syndrome, ataxia-telangiectasia
Hypopigmented macules, streaks, adenoma sebaceum	Tuberous sclerosis, hypomelanosis of Ito
HAIR	
Hirsutism	de Lange syndrome, mucopolysaccharidosis, fetal phenytoin effects, cerebro-oculo-facio-skeletal syndrome, trisomy 18 syndrome
NEUROLOGIC	
Asymmetry of strength and tone	Focal lesion, cerebral palsy
Hypotonia	Prader-Willi syndrome, Down syndrome, Angelman syndrome, gangliosidosis, early cerebral palsy
Hypertonia	Neurodegenerative conditions involving white matter, cerebral palsy, trisomy 18 syndrome
Ataxia	Ataxia-telangiectasia, metachromatic leukodystrophy, Angelman syndrome

Adapted and updated from Liptak G: Mental retardation and developmental disability. In Kliegman RM, Greenbaum LA, Lye PS, editors: Practical Strategies in Pediatric Diagnosis and Therapy, *ed 2, Philadelphia, 2004, Saunders, p 540.*

CATCH-22, **C**ardiac defects, **a**bnormal face, **t**hymic hypoplasia, **c**left palate, **h**ypocalcemia, defects on chromosome **22**; *CHARGE,* **c**oloboma, **h**eart defects, **a**tresia choanae, **r**etarded growth, **g**enital anomalies, **e**ar anomalies (deafness).