



Gross motor delay: slower than expected progression in large-muscle motor skills—such as rolling, sitting, standing, and walking, with normal development in other domains (fine motor, speech/language, cognitive, and social)

Regression of motor milestones
Not sitting up by 9 months
Not walking by 18 months
Persistent hyper/hypotonia
Early hand preference
Fasciculations
Gowers sign



PRESENTATION

HISTORY	PHYSICAL EXAM
<ul style="list-style-type: none"> ▪ Developmental: milestones and order they were met in all domains, parental concerns, regressions ▪ Perinatal and Prenatal: maternal exposures, birth complications ▪ Family: developmental or motor delays, recurrent pregnancy loss, stillbirth, infant death, genetic conditions ▪ Medical: feeding, growth, tone, respiratory or swallowing concerns ▪ Environmental/Social: abuse, neglect opportunities for movement 	<ul style="list-style-type: none"> ▪ Growth and Appearance: weight, height, head circumference, dysmorphic features ▪ Head: shape, fontanelles ▪ Skin: neurocutaneous stigmata ▪ MSK: limb deformities, joint swelling, contractures, range of motion, spine alignment ▪ Neuro: fundoscopy, tone, strength, reflexes, cranial nerves, sensation, balance, coordination, corroboration of gross motor skills from history, may include: gait, posture, and ability to rise from floor

INITIAL EVALUATION

- ❑ **History and physical exam**
- ❑ Screening for delays in all developmental categories

INVESTIGATIONS as informed by Hx and PE may include:

- **CBC, electrolytes, TSH, ferritin** – screen for anemia, nutritional deficiencies and thyroid function, **Creatine Kinase** – elevated in muscular dystrophy
- **Brain imaging U/S or MRI** – consider for evaluation for cerebral palsy
- **Genetic testing** – Start with chromosomal microarray for unexplained developmental delay to detect chromosomal abnormalities
- **Nerve conduction studies / EMG** – consider for spinal muscular atrophy

IMPORTANT CAUSES

Cerebral Palsy	Nonprogressive motor impairment from lesions of the brain acquired before, during or after birth	Nutritional deficiencies	Such as: Vitamin D deficiency, Vitamin C deficiency, severe iron deficiency anemia, severe protein malnutrition
Down Syndrome	Genetic disorder from an extra copy of chromosome 21 can cause hypotonia and delayed motor milestones	Spinal Muscular Atrophy	Loss of anterior horn motor neurons in the spinal cord, causing progressive muscle weakness and wasting.
Muscular Dystrophy	Genetic condition that cause muscles to slowly break down and be replaced by fibrous tissue. Many subtypes that vary in severity, the muscles affected and age at onset		

MANAGEMENT

- **General:** Physical and occupational therapy to improve strength and mobility, orthotic devices, ensure adequate nutrition, support for family, regular follow-up with primary health provider and specialists
- **Condition Specific:** corticosteroids for muscular dystrophy, and **gene therapy** for spinal muscular atrophy

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