



**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"> <li>▪ <b>Developmental:</b> milestones and order they were met in all domains, parental concerns, regressions</li> <li>▪ <b>Perinatal and Prenatal:</b> maternal exposures, birth complications</li> <li>▪ <b>Family:</b> developmental or motor delays, recurrent pregnancy loss, stillbirth, infant death, genetic conditions</li> <li>▪ <b>Medical:</b> feeding, growth, tone, respiratory or swallowing concerns</li> <li>▪ <b>Environmental/Social:</b> abuse, neglect opportunities for movement</li> </ul>	<ul style="list-style-type: none"> <li>▪ <b>Growth and Appearance:</b> weight, height, head circumference, dysmorphic features</li> <li>▪ <b>Head:</b> shape, fontanelles</li> <li>▪ <b>Skin:</b> neurocutaneous stigmata</li> <li>▪ <b>MSK:</b> limb deformities, joint swelling, contractures, range of motion, spine alignment</li> <li>▪ <b>Neuro:</b> 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</li> </ul>

## 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

<b>Cerebral Palsy</b>	Nonprogressive motor impairment from lesions of the brain acquired before, during or after birth	<b>Nutritional deficiencies</b>	Such as: Vitamin D deficiency, Vitamin C deficiency, severe iron deficiency anemia, severe protein malnutrition
<b>Down Syndrome</b>	Genetic disorder from an extra copy of chromosome 21 can cause hypotonia and delayed motor milestones	<b>Spinal Muscular Atrophy</b>	Loss of anterior horn motor neurons in the spinal cord, causing progressive muscle weakness and wasting.
<b>Muscular Dystrophy</b>	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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