



**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

- **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

### PHYSICAL EXAM

- **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

<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

February 2026

Rachel Gibbs (Medical Student, University of Alberta), Dr. Melanie Lewis (Pediatrician, University of Alberta)  
for [www.pedscases.com](http://www.pedscases.com)