Approach to a Floppy Neonate

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Introduction

- Hypotonia in a newborn poses a diagnostic challenge for neonatologists and pediatricians, as it is a clinical sign suggestive of both benign and serious conditions.
- The term 'floppy baby or infant' is used to denote an infant with poor muscle tone affecting the limbs, trunk and the cranial–facial musculature.
- The condition is usually evident at birth or is identified during early life as poor muscle tone results in an inability to maintain normal posture during movement and rest.
- It is an important clinical feature that may indicate an underlying systemic illness or neurological problem at the level of the central or peripheral nervous system.
- The differential diagnosis for neonatal hypotonia is extensive and a methodical approach helps in localizing the problem to a specific region of the nervous system and formulating a differential diagnosis.
- The article presents a structured approach highlighting initial assessment, examination, and management of a neonate with generalized hypotonia.

History and Examination

Prenatal, Neonatal and Perinatal assessment

The list of differential diagnosis of hypotonia in neonates is long, but a good history will narrow the possibilities.

A detailed family history:

- A family history of neuromuscular disease needs to be elicited; a history of repeated abortions may suggest a variety of disorders with prenatal onset, developmental delay (a chromosomal abnormality), delayed motor milestones (a congenital myopathy), and premature death (metabolic or muscle disease).

Details of pregnancy, delivery, and postnatal period :

Prenatal risk factors:

- Parent's age - Advanced maternal age – risk of trisomy and advanced paternal age – risk of imprinting disorders
- Consanguinity – Risk of inborn errors of metabolism, neuromuscular disorders etc.
- History of drug or teratogen exposure – Risk of malformations, etc.
- Decreased fetal movements – Possibility of intrauterine hypoxia, syndrome or neuromuscular disorder
- Polyhydramnios – Probability of underlying gastrointestinal obstructions, syndrome, aneuploidy etc.
- An abnormal fetal presentation, arthrogryposis or a shortened umbilical cord indicate poor fetal movement and may point toward neuromuscular disorder.
- Breech presentation – Probability of syndrome or trisomy.
- History fever with or without rash – Probability of congenital infections - toxoplasmosis, rubella, cytomegalovirus, herpes simplex, etc.
- It is also important to evaluate the mother for muscle weakness and myotonia.

Perinatal history:

- Meticulous documentation of birth events, details of neonatal resuscitation (cord blood gas or blood gas within 1 hour of life, APGAR score) and onset, duration and progression of floppiness / weakness.
- A floppy neonate with normal perinatal history – Rule out neuromuscular disease if the neonate is well and inborn errors of metabolism or sepsisif it is sick.
- Repeated extubation failures in absence of significant perinatal history and a relatively normal airway and cardiopulmonary system – Rule out underlying neuromuscular disorder – myopathy, myotonia, etc.

Clinical examination

General examination

- Clinical examination is the key in narrowing the differential diagnosis.
- Assessfor dysmorphism and any neurocutaneous stigmata.
- Tone is defined as resistance of muscle to stretch, therefore babies with hypotonia have decreased resistance on passive stretching. Weakness is decreased muscle strength or power. Lack of spontaneous movement in a baby suggests weakness.

- Define hypotonia in relation to weakness i.e. floppy strong (have good active tone) or floppy weak infants (have poor active tone manifests as limited limb movements).
- Most hypotonic neonates demonstrate a characteristic frog-legged posture-full abduction and external rotation of the legs as well as a flaccid extension of the arms.
- Other signs of hypotonia include head lag, slip-through on vertical suspension, and draping on ventral suspension (180 degree flip test).
- Weakness can be assessed by the cry, facial expressions, sucking and Moro's reflex, antigravity movements, and respiratory effort.
- It is important to evaluate the distribution and progression of weakness in differentiating the causes of neonatal hypotonia.

Physical examination:

Physical examination of parents:

- It provides useful clue as babies with congenital myotonic dystrophy have severe hypotonia and examining mother may reveal the features of myotonia.
- Likewise, neonatal myasthenia may be suspected if mother displays fatigability of eyelids on upward gaze and fatigability of arms with sustained forward extension.

Anatomic level of pathology

The major task in the evaluation of a hypotonic neonate is to determine the anatomic level of the pathology; whether it is central or peripheral in origin.

The pattern of weakness and muscle involvement may help to localize the involved region in the nervous system [Table 1].

Central vs peripheral hypotonia

- Neonates with central hypotonia generally are hypotonic with good limb movements and those with peripheral hypotonia have relatively poor limb movements.
- The clinical distinction between an upper motor neuron and LMN lesion provides a rationale for investigations based on the localization of lesion in the pathway of motor control**.**

Clinical Features of Central Hypotonia

- These hypotonic neonates show signs of abnormal consciousness, seizures, apneas, abnormal posturing, and feeding difficulties.
- Muscle power is relatively preserved and axial weakness is a significant clinical feature.
- The tendon reflexes are normal or hyperactive, and there is no evidence of muscle fasciculations.
- Diminished or absent tendon reflexes point toward lower motor neuron lesion and brisk reflexes indicate CNS dysfunctions.
- Tight fisting of the hands scissoring when held in vertical suspension are signs of spasticity.
- Postural reflexes are generally preserved in infants with cerebral hypotonia despite a paucity of spontaneous movements.

Clinical Features of Peripheral Hypotonia

- Babies with anterior horn cell disease usually have sparing of extra-ocular muscles while the disorders of neuromuscular junctions may have ptosis and extra-ocular muscle weakness. These infants appear more alert in comparison to those with CNS involvement.
- There is weakness in the antigravity limb muscles along with diminished or absent reflexes. They can have deformities of bones or joints (arthrogryposis).
- Fasciculations, often observed in the tongue, are often very difficult to distinguish from normal random tongue movements.
- Postural reflexes are absent or diminished, and limbs that lack voluntary movement also cannot move reflexively.

Investigations

Appropriate use of investigations is necessary to establish a specific etiologic diagnosis and should be guided by the history and physical examination. We suggest a systematic approach based on the tests currently utilized in the evaluation of infants with hypotonia. Clinical details and relevant diagnostic tests are discussed in conjunction with specific disorders.

The evaluation of the hypotonic infant includes several steps:

- Detail prenatal, natal and postnatal history (includes history of polyhydramnios, decreased fetal movements, prenatal sonography markers showing probability of syndrome, arthrogryposis, fever with rash (TORCH group of infection), pointers of birth asphyxia, and detail onset, duration and progression of hypotonia)
- History of consanguinity and family history of similar illness (e.g. myotonic dystrophy. Myasthenia gravis etc.)
- Physical examination – differentiate between central hypotonia and peripheral hypotonia.
- Laboratory tests, including creatine phosphokinase (CPK), lactate dehydrogenase (LDH),
- and transaminases;
- Electrophysiological studies including nerve conduction studies(NCV), electromyography(EMG) and repetitive nerve conduction studies(RNS).
- Neuroimaging of brain including MRI brain and/or spine, or USG brain, muscle and nerve biopsy.
- Specific genetic testing, including FISH, PCR and genetic panels, clinical exome or whole exome studies

Table. 1. Clinical manifestations based on anatomical localization

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Anatomical site	Clinical features	Predominant helpful investigation	Odd findings
Spinal cord(compressive or noncompressive)	Sensory level,bladder symptoms	MRI spine	Altered consciousness, cranial nerves affection
Anterior horn cell disease (polio/nonpolio viruses)	Lower motor neuron (LMN) type weakness, areflexia	EMG/NCV (if suspected polio, EMG to be avoided in acute stae)	Sensory involvement
Nerve root	Lower motor neuron weakness, areflexia. No sensory loss, follows dermatomal pattern, pain might be present	MRI spine contrast,EMG/NCV	Altered consciousness
Peripheral nerve	Sensory loss, glove and stocking sensory involvement, absent reflexes	EMG/NCV, nerve biopsy	Altered consciousness
Neuromuscular junction	Bulbar/facial weakness >peripheral,waxing-waning weakness	EMG/NCV/RNS	Altered consciousness
muscles	LMN weakness but reflexes may be preserved	CPK high/ EMG/NCV	Sensory affection/ bladder involvement

Fig. 1 Approach to a floppy neonate