APPROACH TO A FLOPPY INFANT.

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Abstract : The floppy infant syndrome is a well-recognized entity for Paediatricians and neonatologists and refers to an infant with generalized hypotonia presenting at birth or in early life. An organized approach is essential when evaluating a floppy infant, as the causes are numerous. Muscle tone is generated by unconscious, continuous, partial contraction of muscle. A child with hypotonia has muscles that are slow to initiate a contraction against an outside force, and also cannot sustain a muscle contraction as long. Hypotonia is the most common neurological abnormality of tone. Floppy infant exhibit poor control of movement, delayed motor skills, and hypotonic motor movement patterns. Weak infants always have hypotonia, but hypotonia may exist without weakness. Some indications of CNS abnormality are because of poor state of alertness, lack of response to visual and auditory stimuli, inability to manage co-ordinated functions like swallowing and sucking. There are two categories - Central and peripheral disorders. Several studies have shown that central causes account for 60% to 80% of hypotonia cases and that peripheral causes occur in 15% to 30%. Detailed neurologic assessment – tone, strength, and reflexes. Assessment of tone – begin by examining posture, and movement. A floppy infant often lies with limbs abducted and extended. Treatment of the infant who has hypotonia must be tailored to the specific responsible condition. In general, therapy is supportive.

Keywords : Floppy, Hypotonia, Developmental delay.

Introduction : Muscle tone can be defined as the tension in a relaxed muscle due to involuntary contractions of its motor units. Muscle tone is generated by unconscious, continuous, partial contraction of muscle. It is important to distinguish weakness from hypotonia. True muscle tone is the inherent ability of the muscle to respond to a stretch. The child with low tone has muscles that are slow to initiate a muscle contraction, contract very slowly in response to a stimulus, and cannot maintain a contraction for as long as normal peers. Because these low-toned muscles do not fully contract before they again relax (muscle accommodates to the stimulus and so shuts down again), they remain loose and very stretchy, never realizing their full potential of maintaining muscle contraction overtime. Hypotonia is described as reduced resistance to passive range of motion in joints. Weakness is reduction in the maximum power that can be generated. A useful definition of hypotonia is an impairment of the ability to sustain postural control and movement against gravity.[1]

A child with hypotonia has muscles that are slow to initiate a contraction against an outside force, and also cannot sustain a muscle contraction as long. At 28 week of gestation there is minimal resistance to passive movements and extremeties are extended.

In a normal term infant there is flexion of all 4 limbs with resistance to passive movement. floppy infants exhibit poor control of movement, delayed motor skills, and hypotonic motor movement patterns. Indications of CNS abnormality are because of poor state of alertness, lack of response to visual and auditory stimuli, inability to manage co-ordinated functions like swallowing and sucking noted that the earlier the onset, the more severe and precipitus the course. [2]

Etiology : There are two categories - Central and peripheral disorders.

Several studies have shown that central causes account for 60% to 80% of hypotonia cases and that peripheral causes occur in 15% to 30%.

A] Some indications of involvement of central nervous system:

1. Extensor plantar response.
2. Sustained ankle clonus.
3. Global developmental delay.
4. Microcephaly or suboptimal head growth.
5. Convulsions.
6. Axial weakness a significant feature [3]

The central nervous system causes are :

a. Cerebral insult -Hypoxic ischemic encephalopathy, Intracranial haemorrhage.

b. Brain malformations.

c. Chromosomal disorders – Prader willi syndrome, Down syndrome.

d. Peroxisomal disorders – cerebrohepatorenal syndrome (Zellweger's syndrome), Neonatal Adrenoleukodystrophy.

e. Genetic defects - familial dysautonomia, oculocerebrorenal syndrome (Lowe syndrome), Joubert syndrome.

f. Metabolic disorders – Acid maltase deficiency, Infantile GM1 gangliosidosis

g. Drug effects (ex Maternal Benzodiazepines)

h. Benign congenital hypotonia.

i. Cerebellar ataxia. [4]

B] Indications of involvement of peripheral nervous system:

1. Hypo- to areflexia.
2. Selective motor delay.
3. Normal head circumference and growth.
4. Preserved social interaction.
5. Weakness of antigravitational limb muscles.
6. Low pitched weak cry.
7. Tongue fasciculations. [5]

The peripheral nervous system causes are:

a. Due to anterior horn cell
Spinal muscular atrophy (Wernig Hoffman syndrome)
Traumatic myelopathy (esp following breech delivery)
Hypoxic ischemic myelopathy.
Pompe's disease.
Polio

b. Peripheral nerves:
GBS
Hereditary sensory and motor neuropathy.
Peripheral neuropathy.

c. Due to neuromuscular junction
Myasthenia gravis (Transient acquired neonatal myasthenia ,congenital myasthenia)
Infantile botulism.
Magnesium toxicity.
Aminoglycoside toxicity.

d. Myopathies
Congenital myopathy
Nemaline myopathy
Central core disease
Myotubular myopathy

e. Muscular dystrophies
Congenital muscular dystrophy.
Dystrophinopathies
Walker Warburg disease
Fukuyama disease
Congenital muscular dystrophy with cerebellar atrophy/ hypoplasia

Congenital muscular dystrophy with occipital Agyria
Early infantile facioscapulohumeral Dystrophy

f. Metabolic and multisystem disease
Disorders of glycogen metabolism (ex Acid maltase deficiency)
Severe neonatal phosphofructokinase deficiency
Severe neonatal phophorylase deficiency
Primary carnitine deficiency
Peroxisomal disorders
Neonatal adrenoleukodystrophy
Cerebrohepatorenal syndrome (zellweger)
Disorders of creatine metabolism
Cytochrome c oxidase deficiency

g. Nutritional :
PEM
Rickets.
scurvy.

h. Endocrine:
Hypothyroidism.
Hyperparathyroidism.[6]

Clinical features : A detailed birth history, Family history, Developmental milestones and head to toe examination are must. Detailed neurologic assessment – tone, strength, and reflexes. Assessment of tone – begin by examining posture, and movement. A floppy infant often lies with limbs abducted and extended. There are two types of floppy infants:

Floppy strong and weak. [7]

Floppy strong	Floppy weak
Increased tendon reflexes	Hypo- to areflexia
Extensor plantar response	Selective motor delay
Sustained ankle clonus	Normal head circumference and growth
Global developmental delay	Preserved social interaction
Microcephaly or suboptimal head growth	Weakness of antigravitational limb muscles
Obtundation convulsions	Tongue fasciculations
Axial weakness	Low pitched weak cry
Central hypotonia	Paradoxical chest wall Movement

1) Floppy babies in early infancy may present with abnormal posturing of limbs and body, Diminished resistance of limbs to passive movement, abnormal range of joint movement

2) Frog-like posture and quality of spontaneous movements, excessive head lag will be evident on 'pull to sit. Typical ragged doll appearance.

3) Delayed motor milestones.

4) Examine the tongue for size and fasciculation. Fasciculation's, irregular twitching movements, generally indicate an abnormality of the anterior horn cells.

5) Paradoxical breathing pattern(intercostals muscles paralyzed with intact diaphragm)

6) The presence of a typical 'myopathic' faces and paucity of facial expression are common in hypotonic infants.

7) A high arched palate is often noted in infants with neuromuscular disorders.

8) Examination of eye movements may provide clues to the presence of ptosis and external ophthalmoplegia may suggest a Myasthenia gravis.[8]


Frog like posture


Ragged doll appearence

Investigations:

A] In Central hypotonia :

CT / MRI : to rule out any congenital anomaly, HIE, intracerebral haemorrhage.

Karyotyping : For syndromes like Down's syndrome and Lowe's syndrome.

Urine aminoacids and Lactate levels : To rule out inborn errors of metabolism.

Carnitine levels : In carnitine deficiency syndrome.

B] In Peripheral causes:

- EMG (electromyography)
- NCS (nerve conduction test)
- Muscle biopsy.
- Creatine kinase.
- Autoantibodies.

- The most common of the neuromuscular disorders, spinal muscular atrophy (SMA), is now diagnosable by molecular genetic analysis (PCR).

-Spinal cord transection or Haemangioma: tuft of hair in midline.MRI spinal cord is diagnostic. Syringomyelia or other forms of spinal dysraphism: Evidence of bladder or bowel dysfunction .Mixed deep tendon reflexes with absent abdominal and anal reflexes.

-Spinal muscular atrophy : Tongue atrophy and fasciculations, Paradoxical breathing pattern ,Severe proximal muscle weakness with absent tendon reflexes. EMG: Baseline fasciculations , diagnosis by PCR.[9]

-Peripheral neuropathy: Weakness predominantly distal In most cases absent deep tendon reflexes. Diagnosis: Sural nerve biopsy.Myasthenia gravis :Greater involvement of oculomotor and bulbar muscles. True congenital myasthenia due to receptor defects is rare. Exclude transient neonatal form from maternal history.Response to acetylcholine esterase inhibitors and Serum antibodies to from acetylcholine receptors. Infantile botulism :Acute onset descending weakness, cranial neuropathies, ptosis, unreactive pupils, dysphagia. Isolation of organism from stool culture and presence of toxin in the stool.Glycogen storage disease: Pompe's disease characterised by enlarged heart in a very floppy weak newborn, Unexplained cardiac failure, Tongue may appear large.

C]Rule out sepsis:

- Complete blood count.
- blood culture.
- Urine culture.
- CSF culture.
- ESR, Electrolytes [10]

Course of illness : A term infant who is born healthy but develops floppiness after 12 to 24 hours – suspect inborn error of metabolism . Infants suffering central injury usually develop increased tone and deep tendon reflexes. Central congenital hypotonia does not worsen with time but may become more readily apparent. [11]

Management : Treatment of the infant who has hypotonia must be tailored to the specific responsible condition. In general, therapy is supportive. Nutrition is of primary importance to maintain ideal body weight for the age and sex which is often achieved through the nasogastric route or percutaneous gastrostomy. Respiratory problems are a primary cause of morbidity and/or hospital admission particularly in young children. There is an increased prevalence of sleep-related upper airway obstruction and lower airway disease. A large number of children may simply require oxygen, even if they have large airway problems. Non-invasive ventilation is relatively uncommon.[12]

Physiotherapy is a important part of management to prevent further progress and improving strength of muscles. Anticonvulsants to control the convulsions

in case on central cause of hypotonia associated with seizures. Body weight supported treadmill training has shown marked improvement in the gross motor Function. Neuromuscular Electrical Stimulation (NMES) can also be used to "activate hypotonic muscles, improve strength, and generate movement in paralyzed limbs while preventing disuse atrophy. Occupational therapy for promoting independent living of the children with hypotonia. Hydrotherapy has been claimed to positively impact the motor development of young children (Stein, 2004). It uses the beneficial effect of buoyancy which puts less mechanical stress on the joints than in land based exercises (Kent, 2003; White, 1895). It is hypothesized that therapeutic horseback riding benefits children with motor disorders e.g. cerebral palsy, because of the rhythmic, three dimensional movement of the horse's walking which replicates the movement of a human pelvis during walking, thus providing a normal sensorimotor experience Rebound therapy may provide constant opportunity for sensory integration of kinesthetic, visual, and vestibular input. The vestibular sensory system, which responds to changes in head position, body movement and the pull of gravity, is heightened in rebound therapy because of the vertical motions of the body on the trampoline. [12]

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