The Hypotonic Infant

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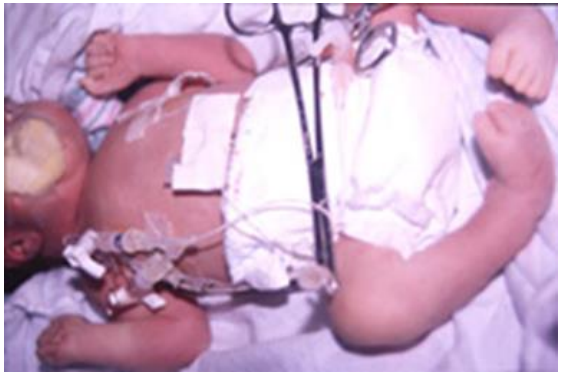
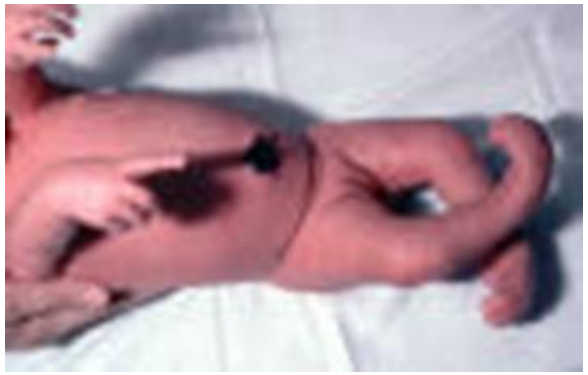
Floppy Infant

- Term reserved for infants with generalized hypotonia presenting at birth
- Decreased resistance to passive movement
- May or may not be associated with muscle weakness

Presenting Features

- Hypotonia in utero
  - Reduced fetal movements
- Problematic delivery
  - Abnormal presentation, failure to progress and frequent C section
- Arthrogryposis





Presenting Features

- Poor respiratory effort
  - Development of pectus excavatum
- Poor suck and swallow
  - Choking
  - Leaking milk

Floppy Infant – History

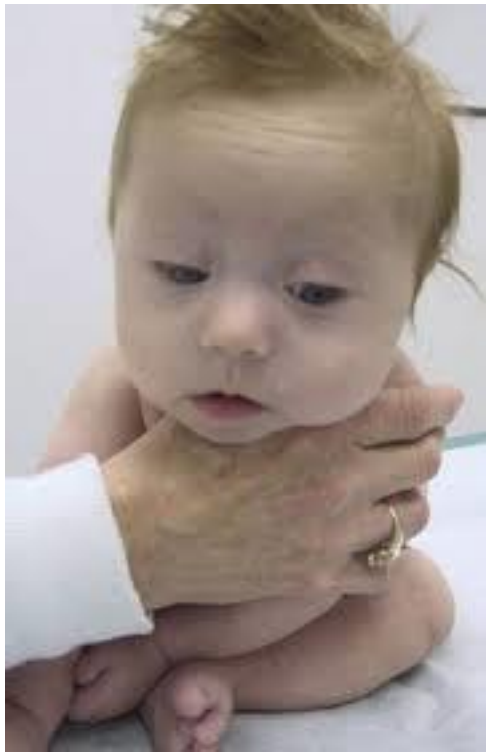
- Perinatal history: polyhydramnios, fetal akinesia, malpresentation, perinatal depression
- Family history: congenital neuromuscular disorders, metabolic disorders

Tone Examination

- The Appearance of Hypotonia
  - Frog like posture
- Clinical examination
  - Traction response
  - Vertical suspension
  - Ventral suspension

Neurologic Examination

- Strength vs tone
  - Antigravity movements
- Motor reflexes
- Eye movements
- Facial movements and tongue fasciculations
- Crying, sucking, and swallowing



Tone exam normal.mp4

Tone Exam hypotonia.MP4

The Hypotonic Infant

- Approach to diagnosis
  - Cerebral hypotonia
  - Motor unit hypotonia

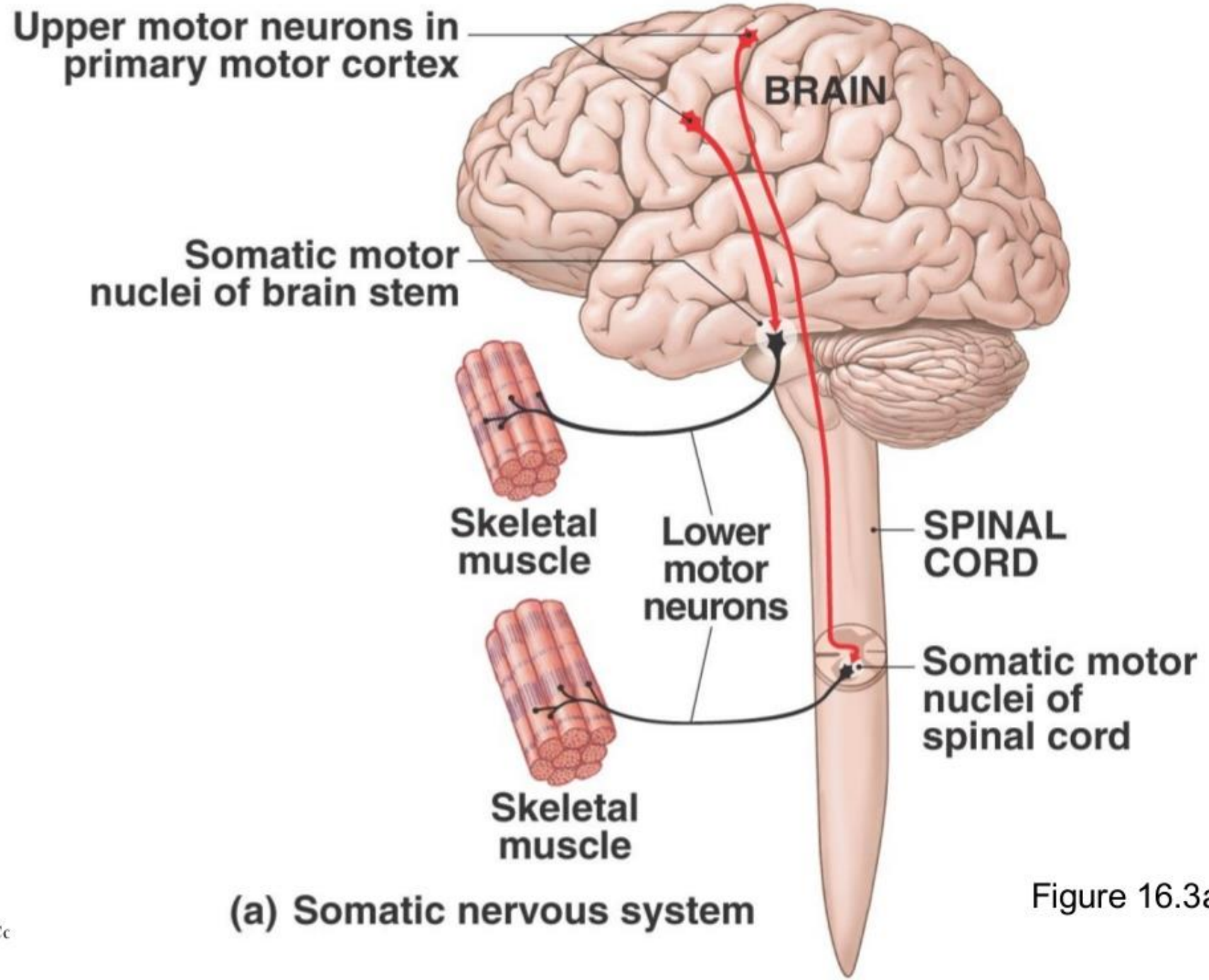


Figure 16.3a

Cc

Cerebral Hypotonia

- Clues to diagnosis
  - Abnormalities of other brain function
  - Dysmorphic features
  - Fisting of the hands
  - Malformations of other organs
  - Movement through postural reflexes
  - Normal or brisk tendon reflexes
  - Scissoring on vertical suspension

Motor Unit Hypotonia

- Absent or depressed tendon reflexes
- Failure of movement on postural reflexes
- Fasciculations
- Muscle atrophy
- No abnormalities of other organs

Tongue fascics.MP4

Cerebral Hypotonia

- CNS infections
  - TORCH, acquired CNS infections
- Chromosomal abnormalities
  - Down syndrome
  - Prader Willi syndrome
- Cerebral malformations
- Hypoxic ischemic encephalopathy
- Inborn errors of intermediary metabolism
- Others

The Hypotonic Infant-DDx

- Spinal cord disorders
  - Hypoxic ischemic myelopathy
  - Spinal cord injury
- Clues to spinal cord injury in the neonate

Neuromuscular Disorders

- Motor unit disorders
  - Anterior horn cell
    - Spinal muscular atrophy
  - Peripheral nerve
    - Peripheral neuropathies rare in the neonatal period
  - Neuromuscular junction
    - Neonatal myasthenia gravis
    - Congenital myasthenic syndromes
  - Congenital myopathies and muscular dystrophies

Spinal Muscular Atrophy

- Primary degeneration of the anterior horn cells of the spinal cord
  - Autosomal recessive, deletion or mutation in survival motor neuron (SMN1) gene, chromosome 5q 11.2-q 13.3
- Severity and phenotypic expression related to a modifying gene, SMN2, which partially compensates for loss of SMN1 protein

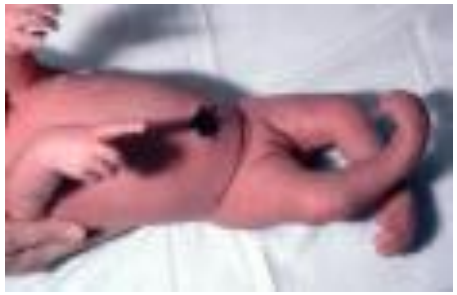
SMA – Classification

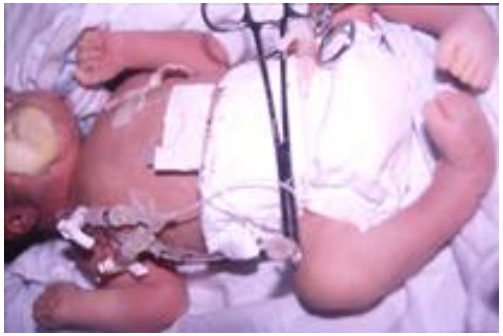
- Many classification systems exist
  - According to age of onset
    - Proximal SMA type I (severe form)
    - Proximal SMA type II (intermediate form)
    - Proximal SMA type III (mild form)
  - According to site of involvement
    - Proximal, distal, upper limb predominance, bulbospinal, scapuloperoneal, …

SMA –Clinical Features

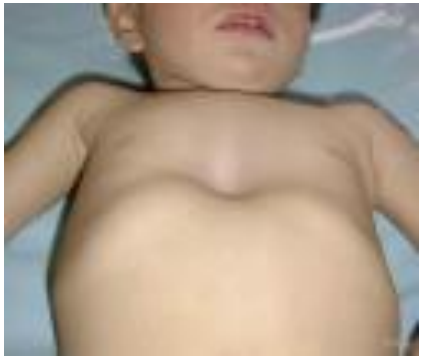
- A lower motor neuron picture:
  - Hypotonia and flaccid weakness
  - Decreased or absent deep tendon reflexes
  - Fasciculations
  - Atrophy, some may have pseudohypertrophy
- Arthrogryposis (multiple cogenital contactures) in the neonate, not common
- Pectus excavatum in long standing cases
- Dysphagia and dysarthria

Arthrogryposis Multiplex





Pectus Excavatum



SMA Type I

- Acute infantile severe form (Werdnig-Hoffman disease)
  - 35% of all SMA cases
  - Presents from birth to 6 months
  - Inherited as autosomal recessive
  - “Floppy baby”
  - Symptoms progress rapidly, the majority die before one year of respiratory failure


Hypotonia
(decreased
muscle tone)
A.D.A.M.


08/31/2002

SMA Type II

- Largest SMA group
- Autosomal recessive inheritance
- Symptoms appear before 18 months of age, usually normal till age 6 months
- Proximal muscle weakness and areflexia
- Motor arrest
- Life span from 2 years to the 3rd decade

SMA Type III

- Mild form
- 8% of SMA of this type
- Onset of symptoms after 18 months of age
- Most are autosomal recessive, some are autosomal dominant or X linked
- Symetrical muscle weakness, atrophy and depressed reflexes
- Slow progression with normal lifespan

SMA - Investigations

- Family history
- Creatine Kinase is usually normal, maybe mildly elevated
- Cerebrospinal fluid normal
- Genetic testing now available and can be done prenatally
- Electromyography: neuronopathic picture

SMA - Treatment

- Specific disease modifying treatments (SMN restoration therapies):
  - Modifying splicing of SMN2: intrathecal, oral
  - Replacing SMN1 gene: intravenous
- Multidisciplinary: physical therapy, occupational therapy, speech therapy,…
- Genetic counseling

Communication Issues

- Nonjudgmental evaluation
  - Avoid showing surprise towards degree of hypotonia and weakness
  - Parents may have feelings of guilt/denial
  - Assess support systems for parents in cases of lengthy admission

Summary

- Hypotonia is a nonspecific sign of neurological disease in infants, most commonly cerebral disease
- Upper vs lower motor neuron hypotonia
- Breathing and feeding issues
- Timely diagnosis is essential in the weak infant