APPROACH TO HYPOTONIA

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- Tone : is defined as the resistance to passive movement
- Hypotonia is decreased resistance to passive movement
- The term “ floppy” is frequently used to describe hypotonic infants

- It is important to distinguish weakness from hypotonia
- Weak infants always have hypotonia, but hypotonia may exist without weakness.

Evaluation

- The age at which hypotonia is first evident .
- Prenatal history :(fetal movement in utero, fetal presentation, and the amount of amniotic fluid) Maternal exposures to toxins or infections suggest a central cause.
- Hypotonia in utero reduced fetal movement may result in arthogryposis
- Low Apgar scores.
- Newborn who becomes hypotonic after the first few days of life consider sepsis and inborn error of metabolism

- Family history of neurologic or neuromuscular disease
- Associated finding of dysmorphology .
- Seizures .
- Delay in motor development

Physical examination

- Dysmorphic features
- Unusual posture (frog-like position)
- Tongue : fasciculations (difficult to observe in limbs because of abundant subcutaneous tissue
- Bell-shaped chest and pectus excavatum
- Scoliosis
- Ambulatory child may show waddiling gate

- Detailed neurological examination (tone-reflexes)
- Horizontal suspension (U shaped)
- Vertical suspension (slips)
- Headlag

Central vs peripheral hypotonia

- Upper motor neuron unit includes(motor cortex-cortecospinal tract- spinal cord)
- Lower motor neuron unit (anterior horn cell peripheral nerve neuromuscular junction muscle)

Think of central hypotonia

- When
- 1- lethargic , not fixating
- 2- seizures
- 3- dysmorphic features
- 4- fisting of the hands
- 5- scissoring on vertical suspension
- 6- malformation of other organs
- 7- normal or increased reflexes

Think of peripharal hypotnia

- When
- 1- alert responsive
- 2- hyporeflexia or areflexia
- 3- muscle attrophy

Examples of Central causes

- Cerebrovascular accident
- hypoxic-ischemic encephalapophy
- Infection
- Chromosomal abnormality
- Metabolic disorders
- Trauma

Examples of peripheral causes

- spinal muscular atrophy
- Myasthenia gravis
- Congenital myopathy
- Hypothyroidism

Diagnosis

- 1- history and physical examination
- 2- imaging'(brain CT or MRI)
- 3- lab tests (metabolic workup) (CPK)
- 4- genetic testing (karyotype , specific genetic defects)
- 5- EMG-NCS
- 6- muscle biopsy

IMAGING

- Brain calcification eg: TORCH
- Brain malformation eg: structural
- Basal ganglia eg: metabolic disorder
- Cerebellar defects eg : Joubert syndrome

Spinal muscular atrophy type I (SMA I)

- Werding-hoffman disease
- 1/3 of cases of SMA
- Presents < 6 months
- Autosomal recessive inheritance
- Progressive majority die < 1 year

Spinal muscular atrophy type II SMA II

- Majority of SMA
- Autosomal recessive inheritance
- Presents between 6-18 months
- Can sit – motor arrest

Spinal muscular atrophy type III
SMA III

- Mild form
- < 10 % of SMA
- Onset > 18 months of age
- Most are autosomal recessive
- Slow progression ??? Life span

SMA investigations

- Labs Creatine kinase normal (CPK)
- CSF normal
- EMG (neuronopathic)
- WES

SMA treatment

- Multidisciplinary (physical , occupational therapy Nutrition , Pulmonologist)
- Manage GERD
- Genetic counseling
- Specific treatment (expensive)

(disease modifying , gene replacement)

Key points

- – Floppy infant is a clinical sign, not a diagnosis
- – Differentiate central vs. peripheral hypotonia
- – History and physical exam are critical
- – Investigations are guided by findings
- – Early diagnosis can improve outcomes

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THANK YOU

Best of luck