Hypotonia

Definition / Supporting Information

Hypotonia is defined as a reduced resistance to passive movement.

Hypotonia is a common finding in infants and young children during acute, febrile illnesses but in such cases the symptoms and signs of the febrile illness predominate and assessment should be directed towards the underlying febrile illness. Only hypotonia occurring outside the context of an acute, febrile illness is considered here.

Keywords / also known as: floppy baby syndrome, low muscle tone

Essential History

Ask about:

- Onset of hypotonia
  - Review birth and perinatal history:
    - Reduced fetal movements may indicate antenatal onset
    - An association between breech presentation and neonatal hypotonia has been described
    - Perinatal drug administration
    - If premature, factors surrounding prematurity
    - Newborn screening results
- Character of hypotonia
  - Is hypotonia improving or worsening?
  - Is weakness intermittent or constant?
  - Is predominant distribution of weakness proximal, distal, or global?
  - Photographs or videos can be used to document findings
- Medical history
  - Previous infections
  - History of trauma
  - Medication history
  - Feeding difficulties (see Dysphagia)
    - Ability to chew
  - Recent history of constipation
  - Respiratory difficulty
    - Cough
  - Seizures
- Family history / pedigree
  - Genetic conditions

  - Individuals with:
    - Weakness
    - Stiffness
    - Developmental delay
- Developmental history
  - Include a chronology of developmental milestones

‘Red Flag’ Symptoms and Signs

Ask about:

- New onset of symptoms
- Speed of progression
- Delayed or disordered developmental milestones
- Swallowing difficulties (see Dysphagia)

Look for:

- Paradoxical respiration
  - Chest wall moves inward during inspiration instead of expanding
- Respiratory distress
- Hepatomegaly / splenomegaly
- Signs of congestive heart failure
  - Tachycardia
  - Gallop rhythm
  - Hepatomegaly
- Altered mental status (see Altered Conscious Level)
- Signs of trauma (including non-accidental injury)
- Abnormal infant cry (eg, cri-du-chat syndrome)
- Positive Gower’s sign (eg, Duchenne muscular dystrophy)
  - Using the hands to ‘walk’ up the legs when changing from squatting to standing position
- Facial muscle weakness (eg, congenital myopathies and fascioscapulohumeral muscular dystrophy)
- Poor proximal tone
  - Infant ‘slips through’ when held upright and suspended in air
- Hyper-reflexia (upper motor neurone lesions)
- Hyporeflexia (lower motor neurone lesions)
- Abnormal gait

Differential Diagnosis / Conditions

- Central hypotonia (brain disorders or dysgenesis)
  - Hypoxic ischaemic encephalopathy

  - Intracranial haemorrhage
  - Infection
  - Metabolic disorders
    - Hyperkalaemic periodic paralysis
      - Rare autosomal-dominant disorder with onset usually in infancy or early childhood
  - Trauma (including non-accidental injury)
  - Hypoglycaemia
  - Hypothyroidism
  - Hyperbilirubinaemia
  - Chromosomal abnormalities
- Spinal cord injury
  - May be a complication of breech deliveries
- Anterior horn cell disorders
  - Infantile progressive spinal muscular atrophy
    - Striking paucity of spontaneous movement
    - Weakness and muscle wasting:
      - Proximal > distal
      - Legs > arms
    - Paradoxical respiration
    - Tongue fasciculation
  - Poliovirus infection
  - Glycogen storage disease type 2 (Pompe disease)
    - Glycogen deposits may form in anterior horn cells
- Peripheral nerve disorders
  - Rare in infancy and very early childhood
  - Guillain-Barré syndrome
    - Acute onset
    - Ascending polyneuropathy
    - Previously normal individual
    - Gait abnormalities
      - Achilles deep tendon reflexes diminished or absent
  - Charcot-Marie-Tooth disease and variants
    - Chronic onset is most frequently caused by hereditary sensorimotor neuropathies
  - Demyelinating neuropathy (eg, presentation of leukodystrophy)
    - Severe weakness, hypotonia and areflexia
  - Chronic Inflammatory demyelinating polyneuropathy

- Neuromuscular junction disorders
  - Passively acquired autoimmune myasthenia gravis (transient neonatal myasthenia)
    - Perinatal transmission of antibodies to infant from seropositive mother
    - Few patients are symptomatic, and symptoms (if present) are usually mild
    - Weakness is unlikely to develop if not present by 1 week of age
  - Acquired autoimmune myasthenia gravis
    - Juvenile myasthenia
  - Non-autoimmune myasthenic syndromes
    - Congenital myasthenia gravis
  - Infantile botulism
    - Rapid onset, usually around age 3 months
    - Symptoms:
      - Initially - constipation
      - Later - weakness, hypotonia, poor feeding, and diminished activity
  - Medications
    - Diazoxide and Clomethiazole used together during pregnancy
    - Magnesium hydroxide
    - Baclofen
- Myopathies
  - Diverse group of genetic, inflammatory, and metabolic disorders
    - May involve other systems (eg, muscle, brain, heart)
  - Congenital myopathies (frequently hereditary)
    - Early-onset hypotonia, weakness, and respiratory and / or feeding difficulties
    - Variable severity
  - Myotonic dystrophy
    - Clinical or electrical onset: age > 5 years
    - If suspected in neonatal period, examine mother for characteristic features of the disease
  - Muscular dystrophies
    - Duchenne muscular dystrophy (common)
      - As this condition is progressive, patients do not have hypotonia in infancy
    - Merosin-deficient type
      - Intellectual disability
      - Seizures
      - White matter abnormalities

        - Fukuyama-type congenital muscular dystrophy (Japan)
            - Severe central nervous system (CNS) abnormalities
        - Facioscapulohumeral muscular dystrophy
            - Linked to high-frequency hearing loss
    - Glycogen storage disease
        - Weakness
        - Marked cardiomegaly
        - Congestive heart failure
        - Hepatomegaly / splenomegaly
    - Mitochondrial disease
        - May exhibit:
            - CNS and muscle involvement
            - Increased serum lactic acid levels
- Benign conditions
    - Benign congenital hypotonia
        - Diagnosis of exclusion
        - Early onset: usually no weakness or developmental delay
        - Frequently associated with joint hyperlaxity
            - May lead to dislocations when adult
- Conditions that may mimic hypotonia
    - Ehlers-Danlos syndrome
        - Hyperlaxity of joints simulates hypotonicity
    - Inflammatory myopathies (eg, dermatomyositis)
        - Rare in very young children
        - Produce weakness, not hypotonia

Investigations

To be undertaken by specialist practitioners (eg, Paediatric / Paediatric Neurology Team(s)):

- Full blood count
- Viral, urine, blood, and cerebrospinal fluid cultures
- Serum glucose measurement
- Urea and electrolyte measurement
- Liver function tests
    - If there is evidence of hepatomegaly
- Creatine kinase – elevated in:
    - Duchenne muscular dystrophy
    - Becker muscular dystrophy
    - Congenital muscular dystrophies

- Consider fractionated bilirubin in a severely jaundiced infant
- Serum lactic acid
  - May be elevated in mitochondrial disorders
- Serum thyroid studies
  - Consider if diagnosis is not obvious on physical examination
  - Hyperthyroidism or hypothyroidism may be associated with acquired autoimmune myasthenia gravis
- Genetic studies
- Karyotype

To be undertaken by a Paediatric Neurology Team:

- Electromyogram and nerve conduction studies
- Consideration to relevant neurometabolic studies
- Genetic studies
  - May confirm genetic disorder suspected on clinical grounds
  - When conclusive, spares patient from painful / invasive diagnostic tests
  - Specific studies may be considered for:
    - Infantile progressive spinal muscular atrophy
    - Myotonic dystrophy
    - Some hereditary sensorimotor neuropathies (demyelinating and X-linked)
    - Some mitochondrial myopathies
    - Duchenne muscular dystrophy
    - Becker muscular dystrophy
    - Hypomyelinating neuropathy
    - Facioscapulohumeral muscular dystrophy
    - Myasthenic syndrome
    - Cri-du-chat syndrome
    - Präder-Willi syndrome

Treatment Approach

- For most causes of hypotonia, treatment is supportive
- Diagnosis is key to outlining the management plan, and likely prognosis
- A multidisciplinary team may coordinate the different interventions

When to Refer

Refer urgently to specialist practitioners (eg, Emergency Department / Paediatric / Paediatric Neurology Team(s)) if:

- Any ‘red flag’ signs or symptoms
- Evidence of respiratory distress (see Dyspnoea)

- Tachycardia
- New onset of neurological signs
- Persistent lack of normal motor development
- Regression of motor development
- Sudden or precipitous worsening of tone or strength
- Swallowing dysfunction (see Dysphagia)

Admit to hospital if:

- Unable to maintain adequate oxygenation
- Symptoms are progressing rapidly
- Mental status changes
- Clinical instability

All other infants and children with hypotonia should be referred non-urgently to Paediatric Team (who will selectively refer on to more highly specialist services), unless the hypotonia is an expected feature of a condition already known to exist in the patient.

‘Safety Netting’ Advice

Advise parent / carer to seek urgent medical advice if any ‘red flag’ signs or symptoms develop.

Patient / Carer Information

Please note: whilst these resources have been developed to a high standard they may not be specific to children.

- Hypotonia - Introduction(Web page), the NHS website
- Hypotonia - Causes(Web page), the NHS website
- Hypotonia - Diagnosis(Web page), the NHS website
- Hypotonia - Treatment(Web page), the NHS website

Resources

National Clinical Guidance

British Thoracic Society Respiratory Management of Children with Neuromuscular Weakness Guideline Group. Guidelines for respiratory management of children with neuromuscular weakness. Thorax 2012;67:1–45.

Suggested Resources

Please note: these resources include links to external websites. These resources may not have national accreditation and therefore PCO UK cannot guarantee the accuracy of the content.

Emergency Guidelines (Web page), British Inherited Metabolic Diseases Group.

D'Amico A, Mercuri E, Tiziano FD et al. Spinal muscular atrophy. Orphanet Journal of Rare Diseases 2011;6:71

Guidelines for the diagnosis of inherited metabolic disease in children with dysmorphic features (pdf), National Metabolic Biochemistry Network.

Stiefel L. Hypotonia in infants. Pediatr Rev 1996;17:104–105. [PubMed]

Zand DJ, Zackai EH. Cytogenetic and molecular diagnoses of hypotonia in the newborn. Neo Reviews 2004;5:e296–300.

Leyenaar J, Camfield P, Camfield C. A schematic approach to hypotonia in infancy. Paediatr Child Health 2005;10(7):397–400. [PubMed]

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