

# CONGENITAL HYPOTONIA • 1/4

## RECOGNITION AND ASSESSMENT

### Definition

- Subjective decrease in resistance to passive range of movement
- Separate from weakness, which refers to lack of muscle strength
- Important to differentiate between central (upper motor neurone), and peripheral (lower motor neurone) hypotonia – may be a mixed picture. See **Table 1** below
- central hypotonia is most common (70–80%)
- Hypotonia
  - relatively common finding in newborn period
  - transient in majority of cases
  - if severe/persistent investigate further

### Symptoms and signs

- Reduced activity/movement
- Reduced level of consciousness/alertness
- Dysmorphic features
- High pitched, weak or fatigable cry
- Increased or reduced respiratory effort
- Feeding difficulties/choking/pooling of secretions
- Seizures/abnormal movements

## DIFFERENTIAL DIAGNOSIS

- Causes of hypotonia in the newborn baby are numerous, not all are listed here
- Benign congenital hypotonia is a diagnosis of exclusion

### Central

- Hypoxic ischaemic encephalopathy (HIE)
- Intracranial haemorrhage
- Structural brain malformation
- Chromosomal abnormalities e.g. trisomy 21, Prader-Willi syndrome
- Congenital infection e.g. TORCH
- Acquired infection e.g. group B *streptococcus*
- Endocrine e.g. congenital hypothyroidism
- Metabolic disorders e.g. acid maltase deficiency (Pompe's disease), carnitine deficiency, mucopolysaccharidosis, peroxisome biogenesis disorders e.g. Zellweger syndrome
- Drug effects e.g. benzodiazepines

### Peripheral

- Spinal cord e.g. birth trauma (especially breech delivery), syringomyelia
- Anterior horn cell e.g. spinal muscular atrophy (SMA)
- Neuromuscular junction e.g. myasthenia gravis, transitory myasthenia
- Peripheral nerves e.g. hereditary motor and sensory neuropathies e.g. Charcot Marie-Tooth disease
- Muscle disorders e.g. muscular dystrophy, congenital myopathy

## HISTORY

### Family

- Affected parents/siblings
- Consanguinity
- Previous miscarriage/stillbirth
- Metabolic/genetic disease
- Premature death

### Maternal

- Diabetes
- Infection
- Medications
- Myotonic dystrophy
- Myasthenia gravis

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## Antenatal

- TORCH infections
- Drug/alcohol exposure
- Fetal movements
- Liquor volume

## Birth

- Gestational age
- Delivery complications
- Malpresentation
- Instrumental delivery
- APGAR score/resuscitation at birth
- Cord gases

## Neonatal

- Respiratory distress
- Feeding issues
- Level of alertness
- Level of spontaneous movement
- Seizures
- Hypoglycaemia
- Weak cry

## PHYSICAL EXAMINATION

### Mother

- Examine for signs of myotonic dystrophy

### Baby

- Full neurological assessment
- Level of alertness
- Abnormal posture
- Degree of hypotonia
  - pull to sit
  - scarf sign
  - shoulder suspension
  - ventral suspension
- Asymmetry
- Strength
- Deep tendon reflexes
- Primitive reflexes
- Gag and suck
- Fasciculations (including tongue)
- Abnormal eye movements
- Ptosis
- Cataracts
- Dysmorphic features/abnormal facies
- Respiratory effort
- Hepatosplenomegaly
- Undescended testicles
- Contractures
- Arthrogyriposis

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Table 1: Summary of typical findings according to cause

CENTRAL HYPOTONIA	PERIPHERAL HYPOTONIA			
	Anterior horn cell	Nerve	Neuromuscular junction	Muscle
Normal strength	Generalised weakness	Weakness, distal>proximal	Weakness, face/eyes/bulbar	Weakness, proximal>distal, face, extraocular muscles
Normal/ increased deep tendon reflexes (DTRs) Clonus	Decreased/ absent DTRs	Decreased/absent DTRs	Normal DTRs	Decreased DTRs
+/- Seizures	Fasciculations	+/- fasciculations	No fasciculations	
+/- Dysmorphic features, reduced alertness	Often described as alert		+/- Arthrogryposis	+/- Contractures

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**Babies with profound central hypotonia may have absent deep tendon reflexes; this sign may not reliably rule out a central cause of hypotonia in first few days of life**

- Weakness uncommon in central hypotonia – except in acute stages
- points to lower motor neurone disorder
- Clinical findings which may direct to a specific diagnosis:
  - hepatosplenomegaly – storage disorders, congenital infections
  - renal cysts, high forehead, wide fontanelle – Zellweger syndrome
  - abnormal odour – metabolic disorders
  - hypopigmentation, undescended testes – Prader-Willi syndrome

## INVESTIGATIONS

- Guided by detailed history and clinical examination
- If hypotonic with a degree of strength, central cause is most likely
- If hypotonic and weak, peripheral cause is possible. Discuss with neurologist
- Involve relevant specialist team early

Table 2: Investigation of the hypotonic infant

	Investigation
Infection screen	<ul style="list-style-type: none"> <li>• FBC</li> <li>• CRP</li> <li>• Blood culture</li> <li>• CSF for microscopy, culture and sensitivity</li> <li>• Congenital infection screen (CMV/toxoplasmosis/herpes simplex/rubella)                             <ul style="list-style-type: none"> <li>• serum</li> <li>• urine</li> </ul> </li> </ul>
Metabolic screen	<ul style="list-style-type: none"> <li>• Blood glucose</li> <li>• Blood gas</li> <li>• Serum lactate</li> <li>• Serum ammonia</li> <li>• Serum amino acids</li> <li>• Carnitine/acylcarnitine</li> <li>• Very long chain fatty acids</li> <li>• Plasma glycine</li> <li>• Urinary organic and amino acids</li> <li>• Urinary glycosaminoglycans (GAGs)</li> <li>• CSF lactate and glycine</li> </ul>
Endocrine screen	<ul style="list-style-type: none"> <li>• Thyroid function (TSH and T<sub>4</sub>)</li> </ul>

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	<ul style="list-style-type: none"> <li>• U&amp;Es</li> <li>• Calcium</li> <li>• Magnesium (e.g hypermagnesaemia after treatment for maternal eclampsia)</li> </ul>
Genetic screen	<ul style="list-style-type: none"> <li>• Karyotype and microarray</li> <li>• DNA for Prader-Willi, Zellweger syndrome</li> <li>• Spinal muscular atrophy (SMA) gene (SMA-RD – if respiratory weakness)</li> <li>• Dystrophin myotonic protein kinase (DMPK gene for myotonic dystrophy)</li> <li>• Other specific genetic test guided by family history/phenotype</li> </ul>
Other	<ul style="list-style-type: none"> <li>• Cranial ultrasound scan</li> <li>• MRI brain +/- spinal cord</li> <li>• EEG (especially if seizures)</li> <li>• CFM (if features of encephalopathy)</li> <li>• Creatinine kinase (muscular dystrophy) <ul style="list-style-type: none"> <li>• may be elevated in first few days after birth</li> <li>• if abnormal repeat after aged 72 hr</li> <li>• if persistently elevated refer to neurologist and consider muscle biopsy</li> </ul> </li> <li>• Nerve conduction studies</li> <li>• If features of maternal myasthenia gravis <ul style="list-style-type: none"> <li>• acetylcholine receptor antibodies</li> <li>• tensilon test</li> <li>• EMG</li> </ul> </li> <li>• If cardiomyopathy suspected <ul style="list-style-type: none"> <li>• ECG</li> <li>• CXR</li> <li>• echocardiography</li> </ul> </li> </ul>

***Muscle biopsy may be delayed until aged 6 months, as neonatal results are difficult to interpret***

## MANAGEMENT

- Specific management determined by individual condition and presentation
- Airway and breathing
  - may need resuscitation at birth
  - airway positioning/Guedel airway
  - intubation and ongoing respiratory support
  - suction of respiratory secretions
- Feeding
  - specialised bottles/teats
  - NG tube feeds
- Skin care
  - regular positional change to avoid pressure sores
- Physiotherapy
  - prevent joint contractures
  - improve muscle strength and co-ordination