

HYPOTONIA (FLOPPY BABY)

RECOGNITION AND ASSESSMENT

Hypotonia can be a relatively common finding in the newborn period and may be transient in the majority of cases, but if it is severe and/or persistent then further investigation indicated

Definition

- Subjective decrease in resistance to passive 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**
- central hypotonia is most common (70–80%)

Signs

- Reduced activity/movement
- Reduced level of consciousness/alertness
- High pitched, weak or fatigable cry
- Increased or reduced respiratory effort
- Feeding difficulties/choking/pooling of secretions
- Seizures/abnormal movements
- **Note:** Look for syndrome-associated dysmorphic features

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 e.g. antidepressants
- Myotonic dystrophy
- Myasthenia gravis

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
- Medication e.g. aminoglycoside toxicity

PHYSICAL EXAMINATION

Mother

- Examine for signs of myotonic dystrophy

Baby

- Full neurological assessment
- Level of alertness
- Abnormal posture
- Degree of hypotonia
- pull to sit – significant head-lag
- scarf sign
- shoulder suspension – ‘slipping through hands’
- ventral suspension
- frog-leg posture
- 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
- Arthrogyposis
- Serial neurological examination for progression of hypotonia or weakness (see **Table 1**)

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, including face/eyes/bulbar	Weakness (proximal>distal), including 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	
+/- Dymorphic features, reduced alertness	Often described as alert		+/- Arthrogyposis	+/- 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
 - hypopigmentation, undescended testes – Prader-Willi syndrome
 - hepatomegaly, retinitis pigmentosa – neonatal adrenoleukodystrophy
 - renal cysts, high forehead, wide fontanelle – Zellweger syndrome
 - congenital cataracts, glaucoma, proteinuria – oculocerebrorenal (Lowe) syndrome
 - abnormal odour – metabolic disorders

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 baby

	Investigation
Infection screen	<ul style="list-style-type: none"> • FBC • CRP • Blood culture • CSF for microscopy, culture and sensitivity • CSF for protein and glucose • Congenital infection screen (TORCH) <ul style="list-style-type: none"> • serum (toxoplasmosis/herpes simplex/rubella) • urine (CMV)

Metabolic screen	<ul style="list-style-type: none"> • Blood glucose • Blood gas • Serum lactate • Serum ammonia • Serum amino acids • Carnitine/acylcarnitine • Very long chain fatty acids • Plasma glycine • Urinary organic and amino acids • Urinary glycosaminoglycans (GAGs) • CSF lactate and glycine
Endocrine screen	<ul style="list-style-type: none"> • Thyroid function (TSH and T4) • U&Es • Calcium • Magnesium (hyper-/hypo- e.g hypermagnesaemia after treatment for maternal eclampsia)
Genetic screen	<ul style="list-style-type: none"> • Karyotype and microarray • 'Hypotonia panel' – may include: <ul style="list-style-type: none"> • DNA for Prader-Willi, Zellweger syndrome • SMA gene (SMA-RD – if respiratory weakness) • DMPK gene (Myotonic Dystrophy gene) • whole exome sequencing (discuss with geneticist) • Other specific genetic test guided by family history/phenotype
Other	<ul style="list-style-type: none"> • Cranial ultrasound scan • MRI brain +/- spinal cord • aEEG (if features of encephalopathy or metabolic condition suspected) • EEG (especially if seizures, but consider even if no clinical seizure) • Creatinine kinase (muscular dystrophy) <ul style="list-style-type: none"> • may be elevated in first few days after birth • if abnormal, repeat after aged 72 hr • if persistently elevated, refer to neurologist and consider muscle biopsy* • Nerve conduction studies • If features of maternal myasthenia gravis: <ul style="list-style-type: none"> • acetylcholine receptor antibodies • tensilon test • EMG • If cardiomyopathy suspected: <ul style="list-style-type: none"> • ECG • chest X-ray • echocardiography • Muscle biopsy*

****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
 - nasogastric tube feeds

Hypotonia (floppy baby) 2025–28

- early speech and language team involvement (where available)
- Skin and developmental care
- regular position changes to avoid pressure sores, reduce risk of contractures and optimise neurodevelopment (see **Developmental care** guideline)
- Physiotherapy
- refer to neonatal/paediatric physiotherapy (while inpatient)
- physiotherapist will:
 - advise on **specific** handling and positioning to optimise neurodevelopmental outcomes
 - assess for symmetry and risk of joint contractures/positional deformity and advise on management
- on discharge refer to community paediatric physiotherapy services
- Early involvement of neurologist, and other specialist teams as indicated