Waikato District Health Board

Procedure

Management of the infant with congenital hypotonia

Procedure Responsibilities and Authorisation

Department Responsible for Procedure	NICU
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Target Audience	Consultants, Registrars, NNPs, CNSs, RNs
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Procedure Review History

Version	Updated by	Date Updated	Summary of Changes
3	Sally Overington	January 2021	Protocol Renamed – Management of the infant with congenital hypotonia
3	Sally Overington	January 2021	Major review of protocol

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Management of the infant with congenital hypotonia

1 Overview

1.1 Purpose

To outline the investigations required for neonates with congenital hypotonia.

1.2 Scope

Waikato District Health Board (DHB) staff working in Neonatal Intensive Care Unit (NICU) e.g. medical staff.

1.3 Patient / client group

Neonates in NICU presenting with congenital hypotonia.

1.4 Definitions

APGAR scores	A rapid method of assessing the clinical status of the newborn infant at birth.
Central hypotonia	Due to upper motor neuron defect
CNS	Clinical Nurse Specialist
Congenital	A disease or physical abnormality present from birth
DTRs	Deep tendon reflexes
ECG	Electrocardiogram
EEG	Electroencephalogram
EMG	Electromyography - a diagnostic procedure that evaluates the health condition of muscles and the nerve cells that control them.
EOM	Extra-ocular movements
FISH	Fluorescent in situ hybridization
Hypotonia	A subjective decrease of resistance to a passive range of motion in a newborn and can be due to a defect at any level of the nervous system
Medical staff	In NICU they include Registrars and Paediatricians
MRI	Magnetic resonance imaging
NNP	Neonatal Nurse Practitioner
Peripheral hypotonia	Due to lower motor neuron defect
SMO	Senior Medical Officer
TORCH	Screen for toxoplasmosis, rubella cytomegalovirus, herpes simplex, and HIV.

Weakness	Decreased muscle strength or power

2 Clinical management

2.1 Causes

- Hypotonia is a relatively common diagnosis in the newborn period. It is an important feature that may indicate an underlying systemic illness or neurological problem at the level of the central or peripheral nervous system.
- Central causes – infant hypotonic but has a degree of strength
- Peripheral – infant is hypotonic and weak
- Central causes account for 60-80% of cases and that the diagnosis can usually be made by a careful history and examination. However, there may be a mixed picture. Infants with a peripheral cause for their hypotonia may be at increased risk for problems during labour, delivery and resuscitation and develop hypoxic ischaemic encephalopathy.
- If hypotonia is a new development – consider infection, physical abuse, surgical conditions, adrenal hyperplasia, bilirubin encephalopathy, drug exposure/overdose.

Central (most common)	Hypoxic ischaemic encephalopathy Intracranial haemorrhage Cerebral malformations Chromosomal abnormalities (e.g.Trisomy 21, Prader-Willi syndrome) Congenital infections (TORCH) Acquired infections Peroxisomal disorders Drug effects (e.g. benzodiazepines) Hypothyroidism
Spinal cord	Birth trauma (especially Breech delivery) Syringomyelia
Anterior Horn Cell	Spinal Muscular Atrophy
Peripheral nerves	Hereditary motor and sensory neuropathies
Neuromuscular junction	Myasthenia gravis (transient / congenital) Infantile botulism
Muscle/ Metabolic myopathies	Muscular dystrophies (incl. congenital myotonic dystrophy) Congenital myopathies (e.g. central core disease) Pompe's disease (acid maltase deficiency) Carnitine deficiency Cytochrome–c–oxidase deficiency

- The first goal in diagnosing the source of neonatal hypotonia is to ascertain if it is central or peripheral. This delineation will determine the investigations most likely to yield a diagnosis.

2.2 History

- Family history - consanguinity, stillbirths, childhood deaths,
- Maternal disease – diabetes, epilepsy, neuromuscular disease,
- Pregnancy & delivery history - drug or teratogen exposure, congenital infection, Mg SO4, abnormal presentation, polyhydramnios/oligohydramnios, reduced foetal movements, APGAR scores, resuscitation requirements, cord gases
- Postnatal history – respiratory effort, ability to feed, level of alertness, level of spontaneous activity, character of cry, medication - aminoglycosides)

2.3 Physical examination

A detailed physical examination should be performed assessing muscle tone, any asymmetry, infant strength, deep tendon reflexes (DTR) and any dysmorphic or unusual features.

Central	Anterior Horn Cell	Nerve	Neuromuscular Junction	Muscle
Normal strength	Generalised weakness	Weakness, distal>proximal	Weakness, face/eyes/bulbar	Weakness, proximal>distal, face, EOM
Normal/increased DTRs	Decreased/ absent DTR	Decreased/ absent DTRs	Normal DTRs	Decreased DTRs
+/- seizures	Fasciculations	Fasciculations	No fasciculations	
+/- dysmorphic features	Often described as alert		+/- arthrogryposis	+/- contractures

Additional clues may direct to a specific diagnosis –

- Hepatosplenomegaly - storage disorders, congenital infections
- Renal cysts, high forehead, wide fontanelles - Zellweger's syndrome
- Hepatomegaly, retinitis pigmentosa - neonatal adrenoleukodystrophy
- Congenital cataracts, glaucoma - oculocerebrorenal (Lowe) syndrome
- Abnormal odour - metabolic disorders
- Hypopigmentation, undesceded testes - Prader Willi
- Maternal examination - important in suspected cases of congenital myotonic dystrophy or myasthenia gravis

2.4 Investigations

These are guided by the history and examination:

- If the infant is hypotonic but has a degree of strength, a central cause is most likely
- If the infant is hypotonic and weak – a peripheral cause is possible and an early review by the neurology service is warranted.

2.4.1 Investigations for causes – first line

	Investigation	Sample
1	Neuroimaging - Ultrasound	
	MRI – brain if structural abnormality of brain development is suspected and to exclude other abnormalities	
2	EEG – prognostic information as to brain function – if seizures suspected	
3	ECG – short P-R interval in Pompe disease	
4	CXR – cardiomyopathy or thin ribs related to reduced fetal respiratory movements – early indication of neuromuscular disease. Pompe disease	
5	Congenital infection screen – TORCH screen Infection – CBC, CRP, blood cultures, Lumbar puncture - markedly increased protein CSF concentration may indicate peripheral neuropathy or specific degenerative conditions	
6	Metabolic workup – Calcium & magnesium LFTs – AST, ALT may be muscle origin Lactate – mitochondrial myopathies Creatinine kinase – muscle injury/dystrophy Transferrin isoforms – carbohydrate deficient glycoprotein syndrome TFTs Carnitine level	 0.2ml heparinised tube 0.2ml heparinised tube Blood gas 0.2ml heparinise tube 0.5ml plain red tube
7	Inborn errors of metabolism – see associated guideline (2662)	

2.4.2 Investigations for causes – second line

	Investigation	Sample
8	Neurology services review	
9	Genetics –	
	Karyotype	1ml heparinised tube
	FISH – Prader Willi	
	Spinal muscular atrophy (SMA) probe	2ml EDTA tube
	Myotonic dystrophy probe	2ml EDTA tube
	Prader-Willi probe	2ml EDTA tube
	Microarray	2ml EDTA tube
10	EMG - reliable after 32 weeks gestation. SMA/disorders of the neuromuscular junction (myasthaenia gravis)	Starship
11	Nerve conduction studies	Starship
12	Muscle biopsy - considered even with normal electrophysiological studies-	Starship

3 Evidence base

3.1 Bibliography

- Hypotonia in neonates. Retrieved on January 2nd 2021 from https://www.starship.org.nz/guidelines/hypotonia-in-neonates
- Ahmed M., Iqbal M and Hussain N. A Structured Approach to the Assessment of a Floppy Neonate. Journal Pediatric Neurosciences 2016 Jan-Mar; 11 (1): 2-6
- Bodame,O., (2019). Approach to the infant with hypotonia and weakness. In M. Patterson & R. Goddeau (Eds.), UpToDate Retrieved January 11, 2021 https://www.uptodate.com/contents/approach-to-the-infant-with-hypotonia-and-weakness

3.2 Associated Waikato DHB Documents –

- Inborn Errors of Metabolism in Infants - Guidelines for Investigation (Ref. 2662)