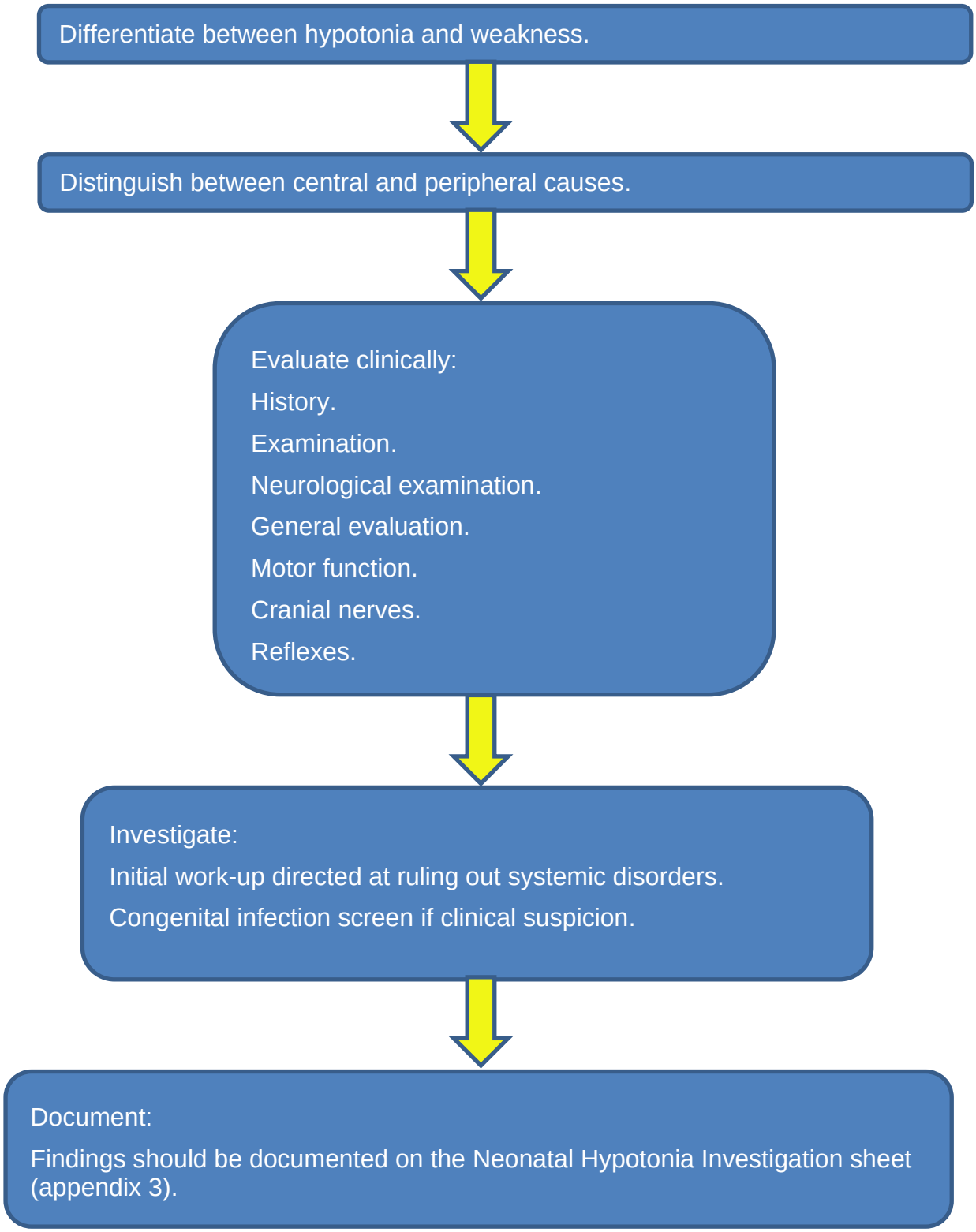
Royal Cornwall Hospitals
NHS Trust

Assessment of Neonatal Hypotonia Clinical Guideline

V4.2

May 2024

Summary


Differentiate between hypotonia and weakness.
Distinguish between central and peripheral causes.
Evaluate clinically:
History.
Examination.
Neurological examination.
General evaluation.
Motor function.
Cranial nerves.
Reflexes.
Investigate:
Initial work-up directed at ruling out systemic disorders.
Congenital infection screen if clinical suspicion.
Document:
Findings should be documented on the Neonatal Hypotonia Investigation sheet (appendix 3).

1. Aim/Purpose of this Guideline

1.1. To provide guidance on the management of hypotonic infants. All involved will benefit from the improvement in service.

1.2. This version supersedes any previous versions of this document.

Data Protection Act 2018 (UK General Data Protection Regulation – GDPR) Legislation.

The Trust has a duty under the Data Protection Act 2018 and UK General Data Protection Regulations 2016/679 to ensure that there is a valid legal basis to process personal and sensitive data. The legal basis for processing must be identified and documented before the processing begins. In many cases we may need consent; this must be explicit, informed, and documented. We cannot rely on opt out, it must be opt in.

Data Protection Act 2018 and UK General Data Protection Regulations 2016/679 is applicable to all staff; this includes those working as contractors and providers of services.

For more information about your obligations under the Data Protection Act 2018 and UK General Data Protection Regulations 2016/679 please see the Information Use Framework Policy or contact the Information Governance Team.

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2. The Guidance

2.1. Introduction

The floppy infant represents a diagnostic challenge with a wide range of possible diagnoses including central or peripheral nervous system abnormalities, myopathies, genetic disorders, endocrinopathies, metabolic diseases and acute illness. Therefore, a systematic approach is required.

It is important to differentiate between hypotonia **and** weakness and these terms are defined below. Weak infants are always hypotonic but hypotonia may exist without weakness.

- **Hypotonia**= reduced resistance to passive range of movement in joints.
- **Weakness**= reduction in the maximum power that can be generated.

When considering aetiology broadly, central causes (acute and chronic) are more common than peripheral (60-80% vs 15-30%).

2.2. Causes

2.2.1. Non-paralytic (floppy but not weak):

2.2.1.1. Central nervous system

- HIE (hypoxic- ischemic encephalopathy) and other encephalopathies.

- Birth trauma (intracerebral and spinal cord).
- Hypotonic Cerebral Palsy.
- Genetic/ chromosomal disorders including Trisomy 21, Prader-Willi Syndrome (PWS), Fragile X.
- Neurometabolic conditions: leukodystrophies, lipidoses, aminoacidurias, Leighs Syndrome.
- Structural malformations, e.g., neuronal migration defects.

2.2.1.2. Connective tissue disorders

- Congenital laxity of ligaments.
- Ehlers-Danlos.
- Marfans.
- Osteogenesis imperfecta.

2.2.1.3. Metabolic and endocrine

- Hypocalcaemia, rickets.
- Hypothyroidism.
- Renal tubular acidosis.
- Organic acidaemias.

2.2.2. Neuromuscular conditions (floppy and weak/paralysed):

- Spinal muscular atrophy (SMA).
- Congenital muscular dystrophy.
- Congenital myotonic dystrophy.
- Neonatal/congenital myasthenia.
- Congenital myopathies (myotubular, nemaline, congenital fibre type disproportion, central core disease).
- Metabolic myopathies (glycogenoses, mitochondrial myopathies, lipid storage disorders, periodic paralysis).
- Neuropathies (Hereditary Sensory Motor Neuropathy, congenital hypomyelination, Polio, Guillain Barre Syndrome, infantile botulism).

Hypoxic-ischaemic encephalopathy, genetic/ chromosomal syndromes and brain anomalies account for 63% of cases (19%, 31% and 13% respectively).

2.3. Distinguishing between central and peripheral causes

Careful clinical assessment may reveal some to clues as to whether the underlying disorder is central or peripheral.

2.3.1. Clues to a central aetiology:

- Lethargy/ reduced conscious level.
- Predominantly axial weakness.
- Normal strength.
- Hyperactive or normal reflexes.
- Dysmorphic features.
- Fisting of hands, scissoring on vertical suspension.
- Malformations of other organs.
- Seizures, abnormal eye movements.
- Apnoea or exaggerated irregular breathing patterns.
- May evolve into increased tone.

2.3.2. Clues to a peripheral (neuromuscular) aetiology:

- Alert, responds normally to surroundings.
- Normal sleep-wake patterns.
- Profound weakness.
- Hyporeflexia or areflexia.
- Respiratory impairment.
- Feeding difficulties.
- Tongue fasciculations.

2.4. Clinical Evaluation

The following features should be specifically sought or considered in the history and examination of a floppy infant.

2.4.1. History:

- Pregnancy- polyhydramnios, reduced fetal movements and malpresentation frequently occur in infants with neuromuscular conditions.

- Maternal exposure to drugs, toxins, and infections.
- Delivery- birth trauma (breech or cervical presentation), poor condition at birth (Apgar's, resuscitation, cord gases).
- Consanguinity (many neuromuscular or metabolic conditions are autosomal recessive).
- Family history (neuromuscular disorders, metabolic disorders).
- Maternal disease e.g., myotonic dystrophy- shake mum's hand.

2.4.2. Examination (general):

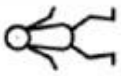
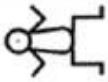
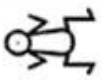
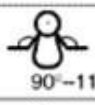
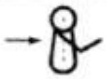
- Assess for acute illness such as sepsis.
- Dysmorphic features and congenital defects.
- Weight, length, and head circumference (plot on growth chart).
- Skin pallor, bruising, or petechiae (trauma).
- Abnormalities of respiratory pattern or diaphragmatic movement (congenital myopathies).
- Evidence of cardiomyopathy (carnitine deficiency, fatty acid oxidation disorders).
- Organomegaly (inborn errors of metabolism, congenital infections).
- Defects of genitalia (PWS (Prader- Willi Syndrome), midline defects).
- Contractures or laxity of hips or other joints (connective tissue disorders, arthrogryposis multiplex congenita).

2.4.3. Neurological examination of the newborn:

It is important to consider the gestational age of the infant, level of alertness at the time of the exam and the experience of the clinician as these may affect the outcome of a neurological examination. The Neuromuscular Maturity score shows neuromuscular maturity at a variety of gestational ages with respect to resting posture and a number of passive manoeuvres (0 = <28 weeks, 1 = 32 weeks, 2 = 34 weeks, 3 = 36 weeks, 4 = 40 weeks)

Please see following page for diagram.

Neuromuscular Maturity

Score	-1	0	1	2	3	4	5
Posture							
Square window (wrist)	 >90°	 90°	 60°	 45°	 30°	 0°	
Arm recoil		 180°	 140°–180°	 110°–140°	 90°–110°	 <90°	
Popliteal angle	 180°	 160°	 140°	 120°	 100°	 90°	 <90°
Scarf sign							
Heel to ear							

2.4.4. General evaluation:

Observe before handling to determine resting posture, level alertness, presence of involuntary movements and respiratory rate and pattern.

2.4.5. Motor function:

- Observe resting posture and measure tone by assessing resistance to passive movements as shown in the table above.
- Assess active muscle function:
  - 32-34/40 infants should have symmetric, smooth, and spontaneous movements in all limbs.
  - Note persistent asymmetry.
  - Sustained tremulousness beyond day 4 may be due to cortical dysfunction.
  - Stepping response in infants >32/40.
  - Vertical suspension measures strength of the shoulder girdle. May also reveal subtle increased tone in legs.
  - Head control- by 40 weeks neck and truncal strength is sufficient to maintain head in line with trunk for 1-2 seconds when pulled to sit.
  - Ventral suspension- measure strength of trunk and neck. Normal term infant holds head in line briefly with flexion of limbs.

2.4.6. Cranial nerves:

You can obtain a fairly full examination by observation of the eyes, facial movements, suck and swallow and tongue and observation of responses to tactile stimuli of the face and to auditory and visual stimuli.

2.4.7. Reflexes:

Tendon reflexes can be difficult to elicit in newborn and are most useful when consistently absent or asymmetric. Can be elicited after 33 weeks gestation and will help localise to upper motor neurone (UMN), lower motor neurone (LMN) or determine level of spinal cord lesion.

- Jaw, biceps, supinator, knee, ankle (triceps difficult due to strong flexion at elbows).
- Superficial reflexes:
  - Abdominal- gentle stroke in all 4 quadrant elicits contraction of abdominal wall.
  - Cremasteric reflex in males.
  - Anal wink.
  - Corneal reflex.
  - Babinski- extensor response may be normal but not if associated with flexor response on other side.

2.5. Investigations

2.5.1. Laboratory, radiological and electrophysiological investigations should be guided by the clinical picture and not all will be required in every case. **See appendix 3 for details.**

2.5.2. Initial work-up directed at ruling out systemic disorders

Sepsis screen if concerns over infection.

- Consider LP (lumbar puncture) for sepsis and metabolic workup (glycyine and other tests after discussion).
- Urea and electrolytes, bone profile and magnesium.
- LFT (liver function test).
- Glucose.
- TFT (thyroid function test).

2.5.3. Congenital infection screen if clinical suspicion

2.5.3.1. Central hypotonia:

- Karyotype and array CGH (comparative genomic hybridisation).
- If no other clinical features send R69 panel.

- If dysmorphic features or other associated signs, then send R27 and R69.

- Screening for inborn errors of metabolism- blood gas, lactate, plasma amino acids and urine organic acids, ammonia, acylcarnitine profile, urine mucopolysaccharides (MPS).

- Neuroimaging for structural abnormalities and metabolic disease- cranial ultrasound scan (USS) and MRI (magnetic resonance imaging).

2.5.3.2. Peripheral hypotonia

- Creatine kinase- elevated in congenital muscular dystrophy, mildly in SMA and normal in many myopathies.

- ECG (electrocardiogram) and echocardiogram.

- CXR (chest x-ray).

- Karyotype and array CGH.

- If no other clinical features send R69 panel.

- If dysmorphic features or other associated signs, then send R27 and R69.

After discussion with neurologist:

- Electrophysiological studies.

- Nerve conduction studies.

- Electromyography.

- Muscle biopsy (myopathies, muscular dystrophy, metabolic disorders).

2.7. Review by other services

- **Ophthalmology**: Infants with hypotonia should be referred to Ophthalmology for review to exclude ocular associations of systemic disease- the Neonatal Consultant should discuss with the Consultant Paediatric Ophthalmologist about the appropriate location for review. No infant should be added to the ROP book without prior consultant level discussion with Ophthalmology.

- **Paediatric Neurology**: Patients with significant or unexplained hypotonia should be discussed with the Paediatric Neurology Consultant at Bristol Children's Hospital.

- **Clinical Genetics**: Patients with unexplained hypotonia or with a family history of hypotonia should be referred to the Clinical Genetics service via rde-tr.pcgreferrals@nhs.net.

2.8. Documentation

The above findings should be documented on the Neonatal Hypotonia Investigation sheet. Appendix 3

3. Monitoring compliance and effectiveness

Information Category	Detail of process and methodology for monitoring compliance
Element to be monitored	Key changes to practice.
Lead	Dr Chris Bell; Neonatal Consultant.
Tool	Audit using a Word /Excel template.
Frequency	As dictated by audit findings.
Reporting arrangements	Neonatal Audit and Guidelines group.
Acting on recommendations and Lead(s)	Dr Chris Bell; Neonatal Consultant.
Change in practice and lessons to be shared	Required changes to practice will be identified and actioned within 3 months. A lead member of the team will be identified to take each change forward where appropriate. Lessons will be shared with all the relevant stakeholders.

4. Equality and Diversity

4.1. This document complies with the Royal Cornwall Hospitals NHS Trust service Equality and Diversity statement which can be found in the Equality Diversity And Inclusion Policy or the Equality and Diversity website.

4.2. Equality Impact Assessment

The Initial Equality Impact Assessment Screening Form is at Appendix 2.

Appendix 1. Governance Information

Information Category	Detailed Information
Document Title:	Assessment of Neonatal Hypotonia Clinical Guideline V4.2
This document replaces (exact title of previous version):	Assessment o Neonatal Hypotonia Clinical Guideline V4.1
Date Issued/Approved:	May 2024
Date Valid From:	May 2024
Date Valid To:	August 2025
Directorate / Department responsible (author/owner):	Dr. Chris Bell; Consultant Paediatrician
Contact details:	01872 252667
Brief summary of contents:	This guideline is designed to ensure a standardised approach to the assessment of infants diagnosed with neonatal hypotonia.
Suggested Keywords:	Neonate. Hypotonia. Assessment.
Target Audience:	**RCHT:** Yes **CFT:** No **CIOS ICB:** No
Executive Director responsible for Policy:	Chief Medical Officer
Approval route for consultation and ratification:	Neonatal Audit and Guidelines Meeting
General Manager confirming approval processes:	Caroline Chappell
Name of Governance Lead confirming approval by specialty and care group management meetings:	Tamara Thirlby
Links to key external standards:	None required
Related Documents:	Curran A, Jardine P. The floppy infant. Current Paediatrics 1998;8:37-42 Dubowitz. The floppy infant syndrome. Chapter 12 in Muscle Disorders in Childhood.

Information Category	Detailed Information
	https://www.clinicalguidelines.scot.nhs.uk/nhsggc-guidelines/nhsggc-guidelines/neonatology/evaluation-of-the-floppy-infant/
Training Need Identified?	No
Publication Location (refer to Policy on Policies – Approvals and Ratification):	Internet and Intranet
Document Library Folder/Sub Folder:	Clinical/ Neonatal

Version Control Table

Date	Version Number	Summary of Changes	Changes Made by
November 2011	V1.0	Initial Issue.	Paul Munyard. Consultant Paediatrician
November 2015	V2.0	Reviewed and reformatted.	Reviewer: Paul Munyard. Consultant Paediatrician and Neonatologist. Formatted by: Kim Smith. Staff Nurse
July 2019	V3.0	Full review- no changes other than updated formatting.	Neonatal Guidelines Group
July 2022	V4.0	Additional testing within section 2.5. Echocardiogram and Ophthalmology review added to list of investigations (section 2.6).	Dr Chris Bell; Consultant Paediatrician.
April 2024	V4.1	Genetic testing updated to reflect new genomic testing (2.5.3.1 and 2.5.3.2).	Dr Chris Bell; Consultant Paediatrician.
May 2024	V4.2	Amendment to wording in appendix 3.	Dr Chris Bell; Consultant Paediatrician.

All or part of this document can be released under the Freedom of Information Act 2000.

All Policies, Strategies and Operating Procedures, including Business Plans, are to be kept for the lifetime of the organisation plus 6 years.

This document is only valid on the day of printing.

Controlled Document.

This document has been created following the Royal Cornwall Hospitals NHS Trust The Policy on Policies (Development and Management of Knowledge Procedural and Web Documents Policy). It should not be altered in any way without the express permission of the author or their Line Manager.

Appendix 2. Equality Impact Assessment

Section 1: Equality Impact Assessment (EIA) Form

The EIA process allows the Trust to identify where a policy or service may have a negative impact on an individual or particular group of people.

For guidance please refer to the Equality Impact Assessment Policy (available from the document library) or contact the Equality, Diversity and Inclusion Team rcht.inclusion@nhs.net

Information Category	Detailed Information
Name of the strategy / policy / proposal / service function to be assessed:	Assessment of Neonatal Hypotonia Clinical Guideline V4.2
Directorate and service area:	Neonatal
Is this a new or existing Policy?	Existing
Name of individual completing EIA (Should be completed by an individual with a good understanding of the Service/Policy):	Neonatal Audit and Guidelines Group
Contact details:	01872 252667

Information Category	Detailed Information
1. Policy Aim - Who is the Policy aimed at? (The Policy is the Strategy, Policy, Proposal or Service Change to be assessed)	To provide guidance on the assessment of hypotonic infants.
2. Policy Objectives	As above.
3. Policy Intended Outcomes	Evidence based and standardised practice.
4. How will you measure each outcome?	Audit.
5. Who is intended to benefit from the policy?	Neonatal Medical and Nursing staff neonatal patients.
6a. Who did you consult with? (Please select Yes or No for each category)	• Workforce: Yes • Patients/ visitors: No • Local groups/ system partners: No • External organisations: No • Other: No

Information Category	Detailed Information
6b. Please list the individuals/groups who have been consulted about this policy.	**Please record specific names of individuals/ groups:** Neonatal Audit and Guidelines Group
6c. What was the outcome of the consultation?	Approved.
6d. Have you used any of the following to assist your assessment?	**National or local statistics, audits, activity reports, process maps, complaints, staff or patient surveys:** No

7. The Impact

Following consultation with key groups, has a negative impact been identified for any protected characteristic? Please note that a rationale is required for each one.

Where a negative impact is identified without rationale, the key groups will need to be consulted again.

Protected Characteristic	(Yes or No)	Rationale
Age	No	
Sex (male or female)		
Gender reassignment (Transgender, non-binary, gender fluid etc.)	No	
Race	No	Any information provided should be in an accessible format for the parent/ carer/ needs- i.e., available in different languages if required/access to an interpreter if required.
Disability (e.g. physical or cognitive impairment, mental health, long term conditions etc.)	No	Those parent/ carer with any identified additional needs will be referred for additional support as appropriate- i.e. to the Liaison Team or for specialised equipment. Written information will be provided in a format to meet the family's needs e.g., easy read, audio etc.
Religion or belief	No	All staff should be aware of any beliefs that may impact on the decision to treat and should respond accordingly.

Protected Characteristic	(Yes or No)	Rationale
Marriage and civil partnership	No	All staff should be aware of any marital arrangements that may have an impact on care (for example: separated parents, domestic abuse).
Pregnancy and maternity	No	
Sexual orientation (e.g. gay, straight, bisexual, lesbian etc.)	No	

A robust rationale must be in place for all protected characteristics. If a negative impact has been identified, please complete section 2. If no negative impact has been identified and if this is not a major service change, you can end the assessment here.

I am confident that section 2 of this EIA does not need completing as there are no highlighted risks of negative impact occurring because of this policy.

Name of person confirming result of initial impact assessment: Neonatal Audit and Guidelines Group

If a negative impact has been identified above OR this is a major service change, you will need to complete section 2 of the EIA form available here:
Section 2. Full Equality Analysis

Appendix 3. Neonatal Hypotonia Investigation Sheet (1 of 2)

Infant's addressograph label.

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History:	
Family history.	
Consanguinity.	
Miscarriages.	
Compare facial features of parents.	
Shake parents hand to confirm grip.	
Pregnancy. Illness, drugs, alcohol, fetal movements, muscular Tone.	
Delivery method/ risk for brachial plexus injury.	
Examination:	
Dysmorphic.	
Weak and floppy.	
Spontaneous movements.	
Anti-gravity movements.	
Facial weakness.	
Tongue fasciculation.	
Ocular muscle weakness.	
Contractures.	
Axial hypotonia in excess of limbs.	
Tendon reflexes increased or decreased.	
Skin elasticity.	
Blue sclera.	

Neonatal Hypotonia Investigation Sheet (2 of 2)

Infant's addressograph label

Blood:	
Urea and electrolytes.	
Bone biochemistry.	
Blood gas.	
Lactate.	
T4 (thyroxine), T3 (triiodothyronine) TSH (thyroid stimulating hormone).	
CK (creatinine kinase).	
Karyotype.	
Gene panel.	
CGH array.	
Urine:	
Organic and amino acids	
MPS (mucopolysaccharidoses) .	
Other tests:	
CXR.	
Cranial USS.	
MRI head.	
Echo.	
ECG.	
Ophthalmology Review	