ASSESSMENT OF NEONATAL HYPOTONIA CLINICAL GUIDELINE

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

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
   Non-paralytic (floppy but not weak):
   Central nervous system
   - HIE 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, eg. neuronal migration defects

   Connective tissue disorders
   - Congenital laxity of ligaments
   - Ehlers-Danlos
   - Marfans
   - Osteogenesis imperfecta
**Metabolic and endocrine**
- Hypocalcaemia, rickets
- Hypothyroidism
- Renal tubular acidosis
- Organic acidaemias

**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.

**Clues to a central aetiology:**
- Lethargy/reduced conscious level
- Predominantly axial weakness
- Normal strength
- Hyperactive or normal reflexes
- Dystrophic 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

**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.
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 (Apgars, 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

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, midline defects)
- Contractures or laxity of hips or other joints (connective tissue disorders, arthrogryposis multiplex congenita)

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 table below 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)

<table>
<thead>
<tr>
<th>Score</th>
<th>-1</th>
<th>0</th>
<th>1</th>
<th>2</th>
<th>3</th>
<th>4</th>
<th>5</th>
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</thead>
<tbody>
<tr>
<td>Square window (wrist)</td>
<td>![Diagram]</td>
<td>![Diagram]</td>
<td>![Diagram]</td>
<td>![Diagram]</td>
<td>![Diagram]</td>
<td>![Diagram]</td>
<td>![Diagram]</td>
</tr>
<tr>
<td>Heel to ear</td>
<td>![Diagram]</td>
<td>![Diagram]</td>
<td>![Diagram]</td>
<td>![Diagram]</td>
<td>![Diagram]</td>
<td>![Diagram]</td>
<td>![Diagram]</td>
</tr>
</tbody>
</table>
General evaluation:
Observe before handling to determine resting posture, level alertness, presence of involuntary movements and respiratory rate and pattern

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 1-2s when pulled to sit
  - Ventral suspension – measure strength of trunk and neck. Normal term infant holds head in line briefly with flexion of limbs

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.

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

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.

Initial work-up directed at ruling out systemic disorders
Sepsis screen if concerns over infection.
- Consider LP for sepsis and metabolic workup (glycyne and other tests after discussion)
- U+E, bone profile and magnesium
- LFT
- Glucose
- TFT

Congenital infection screen if clinical suspicion
Central hypotonia:
- Karyotype and array CGH, specific DNA testing for Prader-Willi
- 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.

Peripheral hypotonia
- Creatine kinase – elevated in congenital muscular dystrophy, mildly in SMA and normal in many myopathies
- Specific DNA testing for SMA and myotonic dystrophy
- Electrophysiological studies
  - Nerve conduction studies
  - Electromyography
- Muscle biopsy (myopathies, muscular dystrophy, metabolic disorders)

Note that nerve conduction studies, EMG and muscle biopsies are carried out at Bristol Children's Hospital and will require discussion with a tertiary Paediatric Neurologist.

2.6 Documentation
The above findings should be documented on the Neonatal Hypotonia Investigation sheet. Appendix 3
3. Monitoring compliance and effectiveness

<table>
<thead>
<tr>
<th>Element to be monitored</th>
<th>Key changes in practice recommended by guidance</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lead</td>
<td>Paul Munyard. Consultant Paediatrician and Neonatologist</td>
</tr>
<tr>
<td>Tool</td>
<td>Audit. To be included in Neonatal Clinical Audit Programme. Findings reported to the Child Health Directorate Audit Meeting / Governance Meeting</td>
</tr>
<tr>
<td>Frequency</td>
<td>As dictated by Audit Meeting</td>
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<tr>
<td>Reporting arrangements</td>
<td>Child Health Directorate Audit Meeting / Governance Meeting</td>
</tr>
<tr>
<td>Acting on recommendations and Lead(s)</td>
<td>Paul Munyard. Consultant Paediatrician and Neonatologist</td>
</tr>
<tr>
<td>Change in practice and lessons to be shared</td>
<td>Required changes to practice will be identified and actioned within 3 months of audit. 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</td>
</tr>
</tbody>
</table>

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 & Human Rights 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

<table>
<thead>
<tr>
<th>Document Title</th>
<th>Assessment of Neonatal Hypotonia – Clinical Guideline</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Date Issued/Approved:</strong></td>
<td>November 2015</td>
</tr>
<tr>
<td><strong>Date Valid From:</strong></td>
<td>November 2015</td>
</tr>
<tr>
<td><strong>Date Valid To:</strong></td>
<td>November 2018</td>
</tr>
<tr>
<td><strong>Directorate / Department responsible (author/owner):</strong></td>
<td>Paul Munyard. Consultant Paediatrician and Neonatologist. Neonatal. Women and Child Health Directorate</td>
</tr>
<tr>
<td><strong>Contact details:</strong></td>
<td>(01872) 253293</td>
</tr>
<tr>
<td><strong>Brief summary of contents</strong></td>
<td>This guideline is designed to ensure the implementation of a standardised approach to the assessment of infants diagnosed with neonatal hypotonia</td>
</tr>
<tr>
<td><strong>Suggested Keywords:</strong></td>
<td>Neonatal. Hypotonia. Assessment</td>
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<tr>
<td><strong>Target Audience</strong></td>
<td>RCHT PCH CFT KCCG</td>
</tr>
<tr>
<td><strong>Executive Director responsible for Policy:</strong></td>
<td>Executive Director</td>
</tr>
<tr>
<td><strong>Date revised:</strong></td>
<td>November 2014</td>
</tr>
<tr>
<td><strong>This document replaces (exact title of previous version):</strong></td>
<td>Assessment of Neonatal Hypotonia</td>
</tr>
<tr>
<td><strong>Approval route (names of committees)/consultation:</strong></td>
<td>Paediatric Consultants. Child Health Audit and Guidelines Meeting</td>
</tr>
<tr>
<td><strong>Divisional Manager confirming approval processes</strong></td>
<td>Helen Ross McGill</td>
</tr>
<tr>
<td><strong>Name and Post Title of additional signatories</strong></td>
<td>Not Required</td>
</tr>
<tr>
<td><strong>Signature of Executive Director giving approval</strong></td>
<td>{Original Copy Signed}</td>
</tr>
<tr>
<td><strong>Publication Location (refer to Policy on Policies – Approvals and Ratification):</strong></td>
<td>Internet &amp; Intranet Intranet Only</td>
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<td><strong>Document Library Folder/Sub Folder</strong></td>
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</tr>
<tr>
<td><strong>Links to key external standards</strong></td>
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Version Control Table

<table>
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<th>Date</th>
<th>Version No</th>
<th>Summary of Changes</th>
<th>Changes Made by (Name and Job Title)</th>
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<tr>
<td>November 2011</td>
<td>V1.0</td>
<td>Initial Issue</td>
<td>Paul Munyard. Consultant Paediatrician and Neonatologist.</td>
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This document is to be retained for 10 years from the date of expiry.

This document is only valid on the day of printing

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## Appendix 2. Initial Equality Impact Assessment Form

<table>
<thead>
<tr>
<th>Name of the strategy:</th>
<th>Assessment of Neonatal Hypotonia - Clinical Guideline</th>
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<tbody>
<tr>
<td>Directorate and service area:</td>
<td>Neonatal. Child Health Directorate</td>
</tr>
<tr>
<td>Is this a new or existing Policy?</td>
<td>Existing</td>
</tr>
<tr>
<td>Name of individual completing assessment:</td>
<td>Paul Munyard</td>
</tr>
<tr>
<td>Telephone:</td>
<td>(01872) 253293</td>
</tr>
</tbody>
</table>

1. **Policy Aim***
   - Who is the strategy / policy / proposal / service function aimed at?
   - To provide guidance on the assessment of hypotonic infants.
   - The guideline is aimed at hospital based staff.

2. **Policy Objectives***
   - As above

3. **Policy – intended Outcomes***
   - Evidence based and standardised practice.

4. **How will you measure the outcome?**
   - Audit

5. **Who is intended to benefit from the policy?**
   - Neonatal Medical and Nursing staff
   - Neonatal patients

6a) **Is consultation required with the workforce, equality groups, local interest groups etc. around this policy?**
   - No. Neonatal Guidelines Group consultant approved guideline

   b) **If yes, have these groups been consulted?**

   c) **Please list any groups who have been consulted about this procedure.**

7. **The Impact**
   - Please complete the following table.

<table>
<thead>
<tr>
<th>Are there concerns that the policy could have differential impact on:</th>
<th>Equality Strands:</th>
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</thead>
<tbody>
<tr>
<td></td>
<td>Yes</td>
</tr>
<tr>
<td><strong>Age</strong></td>
<td>X</td>
</tr>
</tbody>
</table>

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Assessment of Neonatal Hypotonia – Clinical Guideline

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| Sex (male, female, transgender / gender reassignment) | X |
| Race / Ethnic communities / groups | X |
| Disability - learning disability, physical disability, sensory impairment and mental health problems | X |
| Religion / other beliefs | X |
| Marriage and civil partnership | X |
| Pregnancy and maternity | X |
| Sexual Orientation, Bisexual, Gay, heterosexual, Lesbian | X |

You will need to continue to a full Equality Impact Assessment if the following have been highlighted:

- You have ticked “Yes” in any column above and
- No consultation or evidence of there being consultation- this excludes any policies which have been identified as not requiring consultation. or
- Major service redesign or development

8. Please indicate if a full equality analysis is recommended. No

9. If you are not recommending a Full Impact assessment please explain why.

No area indicated

Signature of policy developer / lead manager / director
Paul Munyard 12th November 2014

Names and signatures of members carrying out the Screening Assessment

Keep one copy and send a copy to the Human Rights, Equality and Inclusion Lead, c/o Royal Cornwall Hospitals NHS Trust, Human Resources Department, Knowledge Spa, Truro, Cornwall, TR1 3HD

A summary of the results will be published on the Trust’s web site.

Signed _______ Kim Smith_________

Date _______ 12th November 2015_________
# Neonatal Hypotonia Investigation Sheet (1 of 2)
(For inclusion in the medical notes)

## History:
- Family History
- Consanguinity
- Miscarriages
- Look at parents
- Shake mothers hand
- Pregnancy. Illness, drugs, alcohol, fetal movements, muscular tone
- Birth Trauma

## Examination:
- Dysmorphic
- Weak and Floppy
- Spontaneous movements
- Anti-gravity movements
- Facial weakness
- Tongue fasciculation
- Occular 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)**
(For inclusion in the medical notes)

<table>
<thead>
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<th><strong>Blood:</strong></th>
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<tbody>
<tr>
<td>U&amp;E creatinine</td>
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<tr>
<td>Bone biochemistry</td>
<td></td>
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<tr>
<td>Blood gas</td>
<td></td>
</tr>
<tr>
<td>Lactate</td>
<td></td>
</tr>
<tr>
<td>T4 TSH</td>
<td></td>
</tr>
<tr>
<td>CK</td>
<td></td>
</tr>
<tr>
<td>Chromosome T21</td>
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<td>DNA studies: SMA</td>
<td>(5q13) PWS</td>
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</table>

<table>
<thead>
<tr>
<th><strong>Urine:</strong></th>
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</thead>
<tbody>
<tr>
<td>Organic &amp; amino acids</td>
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</tr>
<tr>
<td>MPS</td>
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</table>

<table>
<thead>
<tr>
<th><strong>Other tests:</strong></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Cerebral Imaging</td>
<td></td>
</tr>
<tr>
<td>EMG &amp; nerve conduction</td>
<td></td>
</tr>
<tr>
<td>Muscle biopsy</td>
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</table>