**Guideline for the Management of the Floppy Infant**

**Definition:**
Subjective decrease in resistance to passive movement due to a problem at any level of the central nervous system (central or peripheral). Pragmatically 'floppy' can mean a decrease in muscle tone (hypotonia), decrease in muscle power (weakness) or a ligamentous laxity with increased range of joint mobility.

**History:**
*Family history:* affected parents/siblings, consanguinity, stillbirths, childhood deaths.
*Mental disease:* myotonic dystrophy (hand shake), epilepsy, diabetes.
*Pregnancy:* drug exposure, fetal movements, polyhydramnios or oligohydramnios, abnormal fetal presentation in-utero.

**Examination:**
A detailed examination should be performed. Look for dysmorphic features and asymmetry. Assess muscle tone, the infant's strength and deep tendon reflexes (DTR). Other symptoms and signs include: arthrogryposis, talipes, feeding difficulties, weak cry, respiratory failure, hepatosplenomagaly (storage disease, congenital infections), undescended testes (Prader-Willi).

**Diagnostic approach:**
Aim to establish whether the floppiness is an **upper motor neurone type** (central) or a **lower motor neurone type** (anterior horn, nerve, neuromuscular junction, muscle). As this will target investigations and underlying aetiology. It can be difficult to distinguish between the two. Up to 60% of cases can be diagnosed from the history and clinical picture. Central causes account for ⅔ of cases with HIE being the most common. Peripheral hypotonia is most commonly caused by muscle disease, particularly myotonic dystrophy.

Remember babies with sepsis, jaundice and hypoglycaemia can also present with floppiness.

**Table1:** clinical findings to help differentiate upper and lower motor neurone type hypotonia

<table>
<thead>
<tr>
<th>Central</th>
<th>Peripheral</th>
</tr>
</thead>
<tbody>
<tr>
<td>Anterior Horn Cell</td>
<td>Neuromuscular Junction</td>
</tr>
<tr>
<td>normal strength</td>
<td>decreased/ absent DTRs</td>
</tr>
<tr>
<td>normal/ increased DTRs</td>
<td>decrease/ proximal</td>
</tr>
<tr>
<td>+/-seizures</td>
<td>fasciculations</td>
</tr>
<tr>
<td>+/-dysmorphic features</td>
<td>often described as alert</td>
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**Aetiology:**

| Central hypotonia (most common) | Hypoxic-ischaemic encephalopathy
- Intracranial haemorrhage
- Cerebral malformations
- Chromosomal abnormalities (e.g. Trisomy 21, Prader-Willi syndrome)
- Congenital infections (TORCH)
- Acquired infections/sepsis
- Peroxisomal disorders
- Drug effects (e.g. Benzodiazepines) |
| Spinal cord | Birth trauma (esp. breech delivery) |
| Anterior horn cell | Spinal Muscular Atrophy |
| Neuromuscular junction | Myasthenia gravis (transient or congenital) |
| Muscle | Muscular dystrophies (inc. congenital myotonic dystrophy)
- Congenital myopathies |
| Peripheral nerves | Hereditary motor and sensory neuropathies |
| Metabolic myopathies | Acid maltase deficiency,Carnitine deficiency
- Cytochrome-c-oxidase deficiency |
Investigations:
Order in which investigations are performed may change depending on the history and clinical picture in each individual infant.

- Serum electrolytes, including calcium and phosphate, serum alkaline phosphatase, blood gas, thyroid function
- Creatinine kinase
- Chromosomal analysis (trisomy), testing for Prader-Willi syndrome (15q11–13)
- Metabolic screen (please refer to metabolic guideline)
- Neurology opinion
- Medical genetics opinion
- Ophthalmology opinion
- Brain imaging (CT/MRI)
- Muscle biopsy (histology, immunohistochemistry, electron microscopy, respiratory chain enzyme analysis)
- Genetic testing (SMA, myotonic dystrophy)
- EMG

Muscle biopsy:
Should be considered only after discussion with the neurology team, pathology and biochemistry.
Careful patient selection as increased anaesthetic risk (post-operative respiratory failure, malignant hyperthermia, rhabdomyolysis).
Discuss biochemical investigations (ie mitochondrial enzymology) in advance with Lydia Llewellyn (head scientist for neurohistopathology, ext 42019).
Consultant Pathologist needs to be available at time of biopsy (contact Jim Neal on ext 44273).
Consent needs to be obtained by paediatric surgical team.
Inform medical biochemistry laboratory (ext 43560) of day, time and location for biopsy.

On the day of the biopsy:
- Confirm the time with theatre, medical biochemistry lab and the consultant pathologist.
- Ensure consent form signed.
- Request form with all relevant investigations required (eg histopathology, mitochondrial) to be attached to the notes.
- SHO or SpR to go to theatre to collect muscle biopsy specimen in sterile universal container and take it straight to lab (1st floor, B-block). Specimens for mitochondrial enzyme analysis need to be frozen immediately (lab will do) and will be forwarded to relevant labs that may be outside UHW.

Overall principles of management:
Physiotherapy: aimed at preventing contractures. Occupational therapy: appliances, improvement of posture and function, facilitating activities of daily living.
Evaluation and treatment of associated cardiac dysfunction.
Respiratory support: assessment of requirement for invasive or non-invasive ventilation and/or tracheostomy.
Feeding: nasogastric feeding, caloric supplementation, gastrostomy. Management of gastroesophageal reflux: medical or fundoplication.
Orthopaedic intervention: prevention and correction of scoliosis and joint contractures.
Encouragement of overall development and stimulation of learning.
Prevention (influenza and pneumococcal vaccination) and prompt treatment of respiratory infections.
Genetics team: pre-natal diagnosis and counselling for future pregnancies.
Ethical considerations: appropriateness of CPR.

References:
Evaluation of the floppy infant, P Sender, J Parr, S Jayawant, Current Paediatrics 2003:13; 345-349
Congenital hypotonia Neubauer D Paro-Panjan J child Neurol 2004
Genetic Evaluation of the floppy infant Prasad AN Sem Fet and Neo Med 2011

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