# 2017

# Guidance for Paediatric Physiotherapists Managing Neuromuscular Disorders





Written and compiled by the APCP Neuromuscular Committee

# Contents

Introduction	2
Section 1 - Neuromuscular Disorders (NMD)	3
Section 2 - Duchenne Muscular Dystrophy (DMD) and Becker Muscular Dystrophy (BMD)	7
Section 3 - Spinal Muscular Atrophy (SMA)	13
Section 4 - Limb Girdle Muscular Dystrophy (LGMD)	20
Section 5 - Myopathies	22
Section 6 - Congenital Muscular Dystrophy	25
Section 7 - Congenital Myasthenic Syndrome (CMS) and Myasthenia Gravis (MG)	29
Section 8 - Charcot Marie Tooth (CMT)	34
Section 9 - Congenital Myotonic Dystrophy	37
Section 10 - Respiratory Management	40
Section 11 - Orthotic Management	46
Section 12 - Knee Ankle Foot Orthoses (KAFOs)	50
Appendix 2 - Specialist Neuromuscular Centres in the UK	52
Appendix 2 – Borg Fatigue Scale	53
References	54
Authors	58

#### Guidance for Paediatric Physiotherapists Managing Neuromuscular Disorders

#### Introduction

This is a consensus document providing guidance on good practice management of infants, children and young adults with neuromuscular disorders (NMD). The intention of this document is to create a resource for learning about physiotherapy assessment and management in NMD.

The advice given in this document is based on best evidence available and professional experience by the authors working in neuromuscular care.

This document was created in collaboration with members of the Association of Paediatric Chartered Physiotherapists Neuromuscular Committee with the help of additional key specialists.

NMD are genetic conditions affecting the motor unit, either the muscles directly or the neuromuscular junction, the anterior horn cells or the peripheral nerves. They are quite distinct from other neurological conditions and so require specific assessment and management.

Management for children with neuromuscular (NM) conditions is constantly changing, with new developments in medication and therapeutic intervention. It is therefore important that this document is read in this context and the reader should seek the most current information from a number of sources.

The document is divided into 12 sections. The first section looks at the general assessment and management of NMD and is followed by eight sections, each focusing on specific NM conditions. At the end of the document there are sections on Respiratory Management, Orthotic Management, and Knee Ankle Foot Orthoses (KAFOs).

The document is further subdivided into two modules, 'Foundation' and 'Advanced'. Both modules are relevant to physiotherapists, with the Foundation Module being aimed at therapists who are relatively inexperienced in the management of NM conditions or who may only have one or two NM children on their caseload. Those involved in Specialist NM Centres may benefit from the Advanced Module. The Advanced Module may also be a useful tool for less experienced therapists, to see what adjuncts to therapy are available from tertiary care.

#### NOTE:

A list of the Specialist NM Centres in the UK at the end of this document and on the Muscular Dytrophy UK website.

# Neuromuscular Disorders (NMD)

#### Introduction

Children may come to you with a "muscle condition" but may not have a diagnosis as yet or, even following genetic testing, may not have been given a full description of their condition. New genes which cause NMD are being found on a regular basis so undiagnosed children may at some stage receive a diagnosis. For a foundation to ALL conditions this section can be a useful reference point even if you know the child's diagnosis and can be used in conjunction with the more specific advice presented in later sections.

Increased tone is not a feature of almost all NMD, there are some **very rare** dystroglycanopathies, including Muscle Eye Brain disease (MEB) that may have a tonal component linked to brain anomalies. It is therefore not appropriate to directly apply treatment techniques developed for cerebral palsy. NMD often need a specific set of assessments and skills, which are outlined in this document. Medicine is changing rapidly and the rarest NMD may have less than 20 children in the country – or world with that diagnosis. There are Specialist NM Centres around the country where help is on hand. Appendix A: List of Specialist NM Centres in UK.

Children with Duchene Muscular Dystrophy (DMD) and Spinal Muscular Atrophy (SMA) will usually be seen six monthly by a Specialist NM Centre – other children may be seen yearly. Community physiotherapists are key to successful management. You must not wait until a child's next neuromuscular centre appointment if they are increasingly struggling with walking, losing ambulation or have rapid deterioration of their posture – contact the Specialist NM Centre.

Assessment	Foundation Module	Advanced Module
Assessment Development / mobility	<ul> <li>Foundation Module</li> <li>All of your current skills are relevant. Assessments for NMD should include:</li> <li>General attainment of developmental milestones - use any suitable scale, such as Alberta Infant Motor Scale (AIMS) (Darrah, Piper, &amp; Watt, 1998) for under 18 months, modules within gross motor function measure (GMFM) (Russell &amp; Hart, n.d.)</li> <li>Assessment of current ambulatory status, note ability indoors as well as outdoors</li> <li>Wheelchair use - especially occasional use, as aspects of NM weakness (e.g. pain and weakness (can impact on</li> </ul>	Advanced Module Children may attend a diagnostic clinic where a full assessment is conducted. Assessments may include an appropriate measure of development or motor performance scale. Continuity of the scale used is important and needs to be age and ability level appropriate. Young weak infants can be assessed on scales, such as the <u>Children's Hospital of</u> <u>Philadelphia - Infant Test of Neuromuscular</u> <u>Disorders (CHOP-INTEND</u> (A. M. Glanzman et al., 2010) Young ambulant children can be assessed using the <u>North Star Ambulatory</u>
	<ul> <li>pain and weakness (can impact on mobility)</li> <li>Gait, transfers and ability on stairs</li> <li>Note symptoms of fatigue. For example the Fatigue Severity Scale.</li> <li>Simple timed tests can be useful, including:</li> <li>Rise from floor time (supine to standing upright) record to the nearest 1/10<sup>th</sup> of a second</li> <li>10 metre run / walk</li> <li>See reference for norms (Pereira, Ribeiro, &amp; Araújo, 2016).</li> </ul>	Assessment (NSAA) or other appropriate scales (Scott et al., 2012). For non-ambulant children. a simple measure of arm function can be conducted, such as the <u>Brooke Upper Extremity Scale</u> (Lord, Portwood, Lieberman, Fowler, & Berck, 1987) or its modified version, Performance of Upper Limb Module (Anna Mayhew et al., 2013). The 6MWT can be applied but perhaps outside the context of a clinical trial does not add value or information to your clinical management (Mcdonald et al., 2013).
Function / ADL	Functional assessment and use of equipment may of course be conducted by or in conjunction with an occupational therapist. Assessment may include	The <u>Activlim</u> maybe a suitable for both ambulant and non-ambulant children. It has a few child specific questions and is a good example of a Patient Reported Outcome

	handwriting, which can be influenced by fatigue.	Measure (PROM) (Batcho et al., 2016). The Egen Klassifikation Scale is also a good PROM / objective measure for non- ambulant children.(Steffensen, Hyde, Lyager, & Mattsson, 2001)
Posture / spine	<ul> <li>Assessment should include:</li> <li>Observation and palpation of spine</li> <li>Pelvic obliquity</li> <li>Leg length</li> <li>Foot posture</li> <li>Any asymmetry</li> <li>Muscle wasting and hypertrophy*</li> <li>Spinal mobility / rigidity</li> <li>Winging of scapulae both at rest and when the arms are raised through full abduction</li> <li>Posture in wheelchair</li> <li>Logging these on a simple body diagram or chart can be very useful.</li> <li>*Specific patterns of muscle wasting and hypertrophy can be very disease specific</li> </ul>	
Range of movement	Knowledge of normal joint range of movement (ROM) is essential. For general assessment a physical review and discussion with the family will help you identify any key joints to assess. These can be the joints you focus on in subsequent assessments. A focus on ROM or muscle groups should include: ankle dorsiflexion, knee extension, ITB, elbows, forearms and fingers maybe appropriate. ROM of the neck can also be important to assess. Joint laxity can contribute to mobility issues particularly in the very young child. Note any existing orthotics.	Patterns of contractures may assist in establishing a diagnosis. In some children, e.g. MDC1a, a full body review is needed for completeness.
Muscle strength / weakness	Knowledge of MRC grades is important, as is knowledge of antigravity versus sub- gravity power. For example the ability to perform knee extension against gravity is key to walking ability (grade 3 or above) but once this is lost (grade 3 and less) knee control may be compromised depending on other weakness and orthotic management may benefit the child. Standardised testing positions are important and grades are important but are not	MRC grading techniques and record keeping are key to assisting in diagnosing children and for management. The grading used does vary but we commonly use the following system Physiotherapists should be familiar with conducting a full manual muscle testing procedure and these patterns can often assist with diagnosis. Myometry (measuring muscle strength with a device to measure force) may be used in the clinical setting especially grip strength

	covered in this section currently.	but is most often conducted as part of a clinical trial.
Pain / fatigue	An assessment of pain and its location, duration, intensity, and aggravating / easing factors can be important. Certain conditions may be painful or include hypersensitivity. Children with NM weakness are often prone to musculoskeletal (MSK) damage and MSK techniques may be beneficial. Record medication prescribed. Note symptoms of fatigue e.g. <u>Fatigue</u> <u>Severity Scale</u> .	Visual analogue scale can be useful for individuals with significant pain issues. For more information on these issues refer to the following references (de Groot et al., 2013) (Kalkman, Schillings, Zwarts, van Engelen, & Bleijenberg, 2007).
Respiratory	It is important to assess for symptoms of hypoventilation and establish history of chest infections. This history should include frequency of chest infections, antibiotic use and any unplanned hospital admissions. These children may have respiratory equipment and an individualised respiratory care plan in place. For more details see Section 10 - Respiratory Management.	Advanced respiratory assessment may be undertaken by a physiologist, or specialist respiratory physiotherapist, and may include forced vital capacity (FVC) in sitting and in lying, especially if the child is undiagnosed and a cardiac component is excluded. For more details see Section 10 – Respiratory Management (Advanced Module).
Management	Foundation Module & Advanced Module	
Wanagement	Foundation module	& Advanced Module
Development / mobility Posture / spine	In a child with a NMD the key is maintaining furecognising that new development milestones comes a point where your role is to manage de Keeping children active using appropriate type maintaining 'good' muscle. Low impact and s increase stamina, not power – are generally be hydrotherapy). Sessions should be kept short children but the Muscular Dystrophy UK Exercent it is important to think about activity in the whet and Boccia are good examples of accessible ambulation is deteriorating. Fun programmes with stretchy exercise bands non-ambulatory individuals. A sitting assessment will help establish best in Specialist seating / standing frames can all be	Advanced Module unction for as long as possible and a may be achieved in the early years but there lecline and maximise function. es of aerobic activity / exercise is key to ymmetrical exercise building – aimed to beneficial (e.g. cycling, swimming, and built up slowly. No guidelines exist for <u>cise Advice for Adults</u> may be helpful. eelchair dependent child. <u>Wheelchair Football</u> activity for those in wheelchairs or where as tied to the wheelchair can be devised for management. enefit these children.
Development / mobility Posture / spine	In a child with a NMD the key is maintaining furecognising that new development milestones comes a point where your role is to manage development children active using appropriate type maintaining 'good' muscle. Low impact and so increase stamina, not power – are generally be hydrotherapy). Sessions should be kept short children but the Muscular Dystrophy UK Exerce It is important to think about activity in the where and Boccia are good examples of accessible ambulation is deteriorating. Fun programmes with stretchy exercise bands non-ambulatory individuals. A sitting assessment will help establish best in Specialist seating / standing frames can all be Spinal braces may be prescribed to assist in for curvature progression. Postural support at night can be benefical, but includes difficulty with auto regulation of temp	Advanced Module unction for as long as possible and a may be achieved in the early years but there lecline and maximise function. es of aerobic activity / exercise is key to ymmetrical exercise building – aimed to beneficial (e.g. cycling, swimming, and built up slowly. No guidelines exist for cise Advice for Adults may be helpful. eelchair dependent child. Wheelchair Football activity for those in wheelchairs or where as tied to the wheelchair can be devised for management. enefit these children. functional sitting, but will not prevent spinal t difficult to tolerate especially if the condition erature.

	Note hypermobility may also need to be managed. Some joints can change from hypermobile to contracture overtime so assessment should be on-going. Orthotics may be prescribed by the Specialist NM Centre. Insoles may be a first step to improve posture.	
	Certain conditions can be greatly assisted by orthotics. Closely working with an orthotist will enable you to learn more about their impact in this group.	
	In conditions with specific patterns of weakness orthotics can be very effective, yet too much correction can limit mobility more.	
Muscle strength / weakness	Strength training may be suitable but the key advice is to keep resistance / weights very light and increase repetitions as stamina improves.	
	is recommended to avoid eccentric muscle loading (e.g. trampoling). in an undiagnosed child (such as trampolining)	
Pain / fatigue	Pain relief may be prescribed by the local paediatrician. In some cases a referral to a pain clinic may be indicated.	
	Massage, warm baths, gentle heat can be beneficial.	
	Advice on pacing can be beneficial.	
Respiratory	See Section 10 - Respiratory Management	

Neuromuscular Disease Center

Muscular Dystrophy UK Factsheets

Rehabilitation Measures Database

# Duchenne muscular dystrophy, Becker muscular dystrophy (Dystrophinopathies) Not Limb Girdle Muscular Dystrophy – see separate section.

#### Introduction

Duchenne Muscular Dystrophy (DMD) is a result of a genetic mutation leading to inability to produce dystrophin and is one of the most common NMD. Becker (BMD) is genetically the same but a milder form where some dystrophin is present in the muscle enabling stronger motor function for longer. It is thought that dystrophin has a role in protecting the muscle membrane during repeated contraction and relaxation of the muscle. In the absence of dystrophin, muscle fibres break down and are replaced by fibrous and / or fatty tissue, causing the muscle to weaken.

It is exceptionally rare for girls to have DMD, although not unheard of. But girls and mums in particular can be "manifesting carriers" i.e. they can have calf hypertrophy, which is often asymmetrical, some weakness, difficulty getting off the floor and managing stairs. In severe cases, girls are like boys and can lose ambulation.

NB - Take note if mum is a manifesting carrier, ask her about difficulties, as this can affect what she is able to do for her child, and more emphasis will have to be placed on management at nursery or school.

Assessment	Foundation Module	Advanced Module
Development / mobility	Children with DMD are often not diagnosed until three or four years of age, or even later in some cases. Children may have presented earlier and been identified as having generalised developmental delay / clumsiness / laziness and even 'low tone'. This developmental delay may be predominantly gross motor, but often includes fine motor speech and cognitive delays. Some gross motor milestones such as hopping and jumping may also never be achieved without management. On examination it is common to see calf hypertrophy which may be associated with a tiptoe gait. The classic sign associated with DMD is the Gower's manoeuvre - where a boy stands up from lying on the floor, he may roll onto all fours, adopt a wide stance and use his hands to push up on his legs to achieve an upright position as illustrated below This manoeuvre is predominantly due to weakness in the hip extensors. Be aware that very young children may well roll prone prior to standing up. Other conditions may present with this classic sign including LGMD and many children with benign hypermobility. Diagnosis is made genetically and boys are commonly started on oral steroids from around the age of 5 or six. This intervention often results in the boy gaining some motor	<ul> <li>Specialist NM Centres use several standard assessments to monitor progression and guide management in DMD.</li> <li>Young children under the age of 3 can be assessed using Bayley Scales of Infant Development (BSID III) which have been validated for under 3's with DMD (Connolly et al., 2014).</li> <li>For older children, until they use ambulation, the most commonly used scale of motor performance is the <u>North Star</u> Ambulatory Assessment (NSAA). This is well validated.</li> <li>NSAA contains two timed tests:</li> <li>Rise from floor (RFF): Time to rise from supine to an upright standing position to the nearest 1/10<sup>th</sup> second.</li> <li>Time 10 metre walk / run: Time to "go as fast as you can" over 10 metres to the nearest 1/10<sup>th</sup> second.</li> <li>Details of the predictive value of the RFF test can be found in (Mazzone et al., 2016).</li> </ul>

	<ul> <li>skills such as running, jumping and even hopping. From this point boys usually enter a plateau phase for months or even years. However weakness is progressive and loss of ambulation generally occurs whilst they are under paediatric services. With steroids this loss of ambulation generally, though not always, occurs at a later age, 12 years old plus.</li> <li>The weakest boys may lose the ability to walk as young as 7 years.</li> <li>BMD is a milder phenotype and boys remain ambulant for longer and often walk much later into adult life. However it is important to monitor mobility, a timed rise from floor and 10m run/walk can be a simple way of tracking progression.</li> </ul>	
Function / ADL	Observe how they rise from the floor as they often employ a Gower's manoeuvre. Observe carrying out stairs and a step as in NSAA. Timed RFF is useful information to assess disease progression. In the latter stages of ambulation assessing bed mobility is important to advise on transfers and equipment provision.	North Star Ambulatory Assessment (NSAA) and the Performance of Upper Limb module (PUL 2.0) are recognised and validated measures for DMD (A Mayhew, Cano, Scott, & Eagle, 2011) (A. G. Mayhew et al., 2013) (Anna Mayhew et al., 2013) (Pane et al., 2014). For BMD the NSAA is often not difficult enough. Items which test higher levels of function may be needed such a squat or rise through half kneeling.
Posture / spine	When assessing posture in standing, check for calf hypertrophy and significant lumbar lordosis, which are common in this group and can assist in establishing a diagnosis. Spinal posture must be assessed, particularly as weakness progresses and especially when ambulation is lost. Note shoulder and hip symmetry and any scapular winging. Axial weakness can be significant in this group and may act as a diagnostic clue.	In sitting and standing observe for evidence of scoliosis and assess ability to correct scoliosis passively.
Range of movement	Loss of ankle range commonly occurs. Restricted dorsiflexion may appear to be more dynamic (up on toes whilst walking) or observed as passive loss of range. Other commonly affected joint ranges are hip extension (often evident as a lack of extension when walking), tight ITB's and as ambulation becomes more difficult or is lost, range may be lost in knee extension. Even whilst the boy or young man is walking the upper limbs may also be affected particularly long finger flexors and supination.	Specialist NM Centres need to monitor joint range with a particular focus on the ranges detailed here.

Strength / weakness		Manual Muscle Test (MMT) can be conducted around diagnosis and to monitor progression.
		Myometry may be used to gain a more objective values and to identify asymmetry.
Pain and fatigue	DMD: Pain is not usually a problem in this population in the ambulatory phase. However pain in older boys, whether ambulant or not, must be taken seriously as prolonged used of corticosteroids result in reduced bone density and fractures are not uncommon.	
	Back pain if persistent, especially in non- ambulant children may require an x-ray.	
	A fall or knock may also require radiological review as a precaution. Many boys may not have severe pain associated with their fractures so even mild pain should be taken seriously.	
	BMD: Pain can be a significant issue in this group. Painful cramps can be a problem in this population, so it is important to ask specifically about this.	
Respiratory	Respiratory issues are not generally a problem in ambulant DMD boys. However as the disease progresses respiratory muscles are affected.	Forced Vital Capacity (FVC) is usually monitored from soon after diagnosis to get the boys used to this test. Some will never achieve a satisfactory test.
	In the non-ambulant phase it is important to assess for symptoms of hypoventilation and establish history of chest infections including frequency of chest infections, antibiotic use and any unplanned hospital	As boys get older a measure of their ability to cough effectively via Peak Cough Flow (PCF) (using a mask to achieve a good seal) is included in respiratory assessments.
	admissions. These children may have respiratory equipment and an individualised respiratory care plan in place.	A falling FVC may prompt an overnight pulse oximetry trace to be done to check for symptoms of hypoventilation especially if the boy or family has mentioned poor sleep, nausea or headaches in the morning).
	Respiratory Management.	For more details see Section 10 – Respiratory Management (Advanced Module).
Management	Foundation Module & Advanced Module	
Mobility	Ambulatory Phase:	
	Maintaining ambulation is the primary goal an PE and activity, especially low impact exercise	d boys should be encouraged to participate in e such as swimming.
	Boys may not be able to keep up with their pe to short periods and paced appropriately.	ers and participation may need to be limited
	Active trampolining, which involves significant	impact and eccentric muscle activity, is

	strongly discouraged. Overuse activity can lead to muscle damage resulting in myoglubinuria (Coca-Cola coloured urine).
	The use of scooters is also discouraged, as it is encourages leg and hip power asymmetry. Children should try bicycles or tricycles or low gear tricycles.
	NB – low-geared tricycles are VERY expensive and may need charity funding.
	As boys become weaker you could consider the use of KAFOs (see Orthotics)
	Progression to non-ambulatory phase:
	Managing a progressive disease such as this requires sensitivity and boys and families sometimes need to be given "permission to stop walking".
	Ensure early referral for wheelchairs and offer appropriate advice. As steroids keep this boys and young men 'stronger for longer' manual chairs with assisted power chairs or even sit-on scooters maybe appropriate. Since steroids often significantly limit growth be aware that not all wheelchairs that are wide enough fit leg length appropriately
	Provision of an electric wheelchair often gives them back independence and allows them to use their energy to do other things. Participation in sport is still important and continuing swimming, wheelchair football or Boccia can be encouraged.
	It is important to advise and assist in obtaining appropriate mobility equipment at the relevant time, such as manual wheelchair to help with school trips etc. when a boy's ability to walk for long distances ay be reduced.
	Provision of a standing frame is important to help maintain lower limb soft tissue length, joint range and can also provide psychological and physiological benefits.
Function / ADL	See above for advice on participation.
	Early referral for home and school adaptations i.e. ramps, lifts, wet rooms, bathroom equipment is imperative. It is not unusual for major work to be required to future proof any adaptations to incorporate tracking hoists etc.
	Sleeping and appropriate mattress and bed provision are also important and should be reviewed with the appropriate support from additional professionals.
	As arm function is lost feeding may become difficult. Mobile arm supports may be beneficial.
Posture / spine	Non-ambulatory phase:
	Use of sleep systems to encourage symmetry may be beneficial but are not always well tolerated.
	Boys should be referred for an orthopaedic assessment if a scoliosis is apparent. Spinal bracing and surgical stabilisation are not used a frequently in DMD now, due to the advances in medical management and the DMD boys are staying ambulant for longer. Boys who lose ambulation before reaching puberty are most likely to require intervention.
Range of movement	Ambulatory phase:
	The first strategy is usually daily stretches for tight joints. However do not give a programme of stretches to families if joints are not tight.
	Dorsiflexion of the ankle is the main joint that needs attention and teach stretches for hips, knees and ITB if necessary if weakness leads to contractures.
	In younger children these stretches are usually taught to families and carers and are passive in nature. However active or self-directed stretches can be shown and are important for older boys.
	Remember a loss of range is not always due to a lack of physiotherapy as growth or a sudden increase in strength as steroids are started can lead to a further loss of range.

	If stretches are not sufficient to maintain a reasonable joint range (10 degrees ankle DF) orthotics or serial casts can be used.
	Ambulatory boys:
	Night-time AFO's are commonly prescribed for ambulatory boys. However daytime AFO's are NOT beneficial.
	Insoles can be useful and some centres use contracture control devices (CCD) for ankles in this group.
	Non-ambulatory:
	Daytime AFO's can be important to maintain good foot position but are sometimes poorly tolerated and poorly understood by the boys. As fixed plantar flexion progresses they tend towards inversion, which can be painful and can make it difficult to find footwear that fits. Prevention is preferable than the need for surgery in the future.
	Surgery may be considered earlier rather than later particularly in boys with splint compliance issues – to ensure that deteriorating cardiac and respiratory function do not prevent surgery – it may be better to suggest that surgery occurs at a specific neuromuscular service.
Exercise / Activity (Strength)	Age appropriate exercise programmes and activity advice should be given and PE encouraged.
	Avoid eccentric muscle activities (e.g. trampolines) and asymmetrical activities (e.g. use of scooters).
	The <u>Muscular Dystrophy UK Education Guidelines on Physical Education</u> provides useful information on inclusion in PE at school
Pain and fatigue	BMD: Management of pain / cramp in this group is important and boys can benefit from warm baths, massage and stretches. Some boys find Lycra shorts or leggings beneficial to reduce cramping. Refer to the medical team if you think medication to control pain is
	required.
	required. Some families benefit from advice on managing activity levels. It is important to find a balance, as painful muscles are a sign of overuse. Schools need to understand the need to NOT over exercise and allow rests, whilst at the same time not preventing any participation in activities.
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wheelchair often gives them back independence and allows them to use their energy to doother things. However participation in sport is still important and continuing swimming, wheelchair football or Botcha are great activities to encourage.
Ensure early referral for wheelchairs and offer appropriate advice. As steroids keep this boys and young men 'stronger for longer' manual chairs with assisted power maybe appropriate, or even sit-on scooters. Since steroids often significantly limit growth be aware that not all wheelchairs that are wide enough fit leg length appropriately.

**Diagnosis and Management of DMD** 

#### Spinal Muscular Atrophy (SMA)

#### Introduction

Spinal Muscular Atrophy (SMA) is an autosomal recessive neuromuscular (NM) disease caused by homozygous mutations of the survival motor neuron (SMN1) gene. The estimated incidence is 1 in 6,000 to 1 in 10,000 live births. The carrier frequency is 1 in 40 to 60. The SMN1 gene encodes for the survival motor neuron protein, which maintains the health of alpha motor neurons in the spinal cord. Lack of this protein causes degeneration of the alpha motor neurons resulting in progressive, generalised muscle weakness and atrophy.

Muscle weakness affects both sides but most children have some asymmetry. Weakness is greater proximally than distally, and generally greater in the legs than the arms. Diaphragm, facial muscles and cognition are spared although bulbar and swallowing problems are frequent in the weaker children. Breathing is affected in SMA I and the weaker SMA II children, although stronger children with type II or type III can get chest infections.

The disease is classified by severity (SMA I, SMA II, SMA III, SMA IV) based on age of onset and maximum motor function achieved, but there is a spectrum within each classification. Severity is generally, but not exactly, related to the loss of the number of copies of the SMN2 gene, a gene almost identical to the SMN1 gene but producing only around 10% of the survival motor neuron protein. Although SMA is classified according to maximum function achieved (Munsat & Davies, 1992) it is one disorder with a very large spectrum of functional ability, from the very weakest, unable to hold their own head, to the mildest who have relatively mild difficulties with stairs, running and getting off the floor.

**SMA I** is the most severe form of SMA. It is the leading genetic cause of infant death in under 2 year olds. Symptoms appear within the first few months of life, sometimes before birth. Children are never able to sit unaided and the weakest rarely survive their second birthday. The spared diaphragm, combined with weakened intercostal muscles, results in paradoxical breathing. The involvement of bulbar motor neurons often causes tongue fasciculation and poor suck and swallow with increasing swallowing and feeding difficulty over time. Pulmonary disease is the major cause of morbidity and mortality in SMA I and SMA II and swallowing dysfunction and reflux are important contributors to pulmonary morbidity (Wang et al., 2007). There is increasing evidence that some patients with severe SMA I may have heart defects and involvement of the autonomic system that may be responsible for arrhythmia and sudden death.

SMA I is sub-categorised into:

- SMA Ia the severe neonatal variant with joint contractures and a paucity of movement present at birth has a poor prognosis, often needing ventilatory support as a neonate
- SMA lb is the typical SMA I patient having poor head control and difficulty handling oral secretions upon or shortly after presentation and has an intermediate prognosis
- SMA Ic is the minority of type I patients who achieve head control or who can sit with support and have the best prognosis (Bertini et al., 2005). Some SMA Ic will survive into childhood and even transition. They should be considered as a weak SMA II in terms of assessment and management.

**SMA II** symptoms appear between the ages of 6 and 18 months. Children achieve sitting, though may lose the ability over time, but are never able to walk unaided. Fine tremors of the hands and fingers may be seen and at rare times can affect fine motor function. Joint contractures and kyphoscoliosis can occur in all children in the first years of life although there is often a mixture of joint contractures and hypermobility, increasing the functional difficulties. Weak swallowing can be present but is not common; weakness of the masticatory muscles can affect the ability to chew. There is a very broad spectrum of severity ranging from weak children who are *just* able to sit unsupported and are more prone to respiratory signs and early scoliosis to relatively stronger children who have much stronger trunk, limb and respiratory muscles. Patients at the weak end of the spectrum may develop respiratory failure requiring mechanical ventilation.

**SMA III** Symptoms usually appear after 18 months of age. They are classified into SMA IIIa – onset of symptoms before the age of 5, and SMA IIIb, whose symptoms appear later. The latter are invariably milder and more likely to walk into adulthood, though may still lose ambulation in later adulthood. Children typically experience reduced walking ability over time. Those with SMA IIIa may never achieve hopping, jumping or running and as with many other NM children, can struggle with stairs and getting off the floor. Patients who lose ambulation before puberty often develop scoliosis and other medical problems related to poor mobility such as obesity and osteoporosis.

Other types of SMA exist e.g. Distal Spinal Muscular Atrophy (DSMA) and SMA with Respiratory Distress (SMARDI). SMARDI shares some clinical features with SMA I but is caused by a different gene. SMARDI has diaphragmatic involvement leading to respiratory failure, with relative sparing of the intercostals and muscle weakness is greater distally than proximally.

Assessment	Foundation Module	Advanced Module
Assessment	Children with SMA are almost always bright and engaging. They delight in stimulation and play and progress quickly in cognitive and communication skills despite their motor limitations. Assessment will vary depending on the stage and severity of the disease. While assessing development, mobility, function and range of movement it is important to review respiratory status and monitor bulbar function. There are several assessments in use for SMA I and depending on your caseload and experience, you may want to learn some of these - see advanced section.	Functional scales should be recorded in conjunction with assessment of joint range, spinal posture and for SMA III, muscle power. Testing muscle power in SMA II, may be less helpful (though can be used for the upper limbs). It can be useful for the ambulant child as it enables prediction of loss of skills.
Respiratory and Bulbar	In SMA I and weaker SMA II assess chest and breathing and sleep pattern. Bulbar assessm child gets on with eating, if they ever choke or swallowing. Some may only eat soft food – yo taken to feed or eat meals is also a useful indi- interventionist approach with ventilation and s section). Oxygen saturation may be assessed oximetry. Recent advances in therapeutic strategies will management. For SMA I, SMA II and SMA III: monitor freque and ability to clear secretions as well as cough respiratory function using FVC and a PCF from	d spine shape, respiratory rate, work of ent would include asking parents how the cough on their food or drink or have difficulty oghurt and ice cream being favourites. Time icator. iative but some parents may choose a more cometimes tracheostomy (see management d in clinic settings and/or home overnight l increase the likelihood of more intensive ency of chest infections, hospital admissions h strength. Tertiary centres monitor m age 3-4 years
Mobility	Non-ambulant SMA I and SMA II children- assess floor and bed mobility and transitions and the strategies used – relating them to power and contractures. Children with SMA II with good head control: Assess for suitability of KAFOs from normal walking age, if appropriate having tried standing in leg gaiters first. Stronger children may be able to take steps/walk in gaiters/KAFOs. Most will need to use a posture control walker – but a timed 10m walk test can be performed in the most able. Assess arm power for the suitability of a manual wheelchair (lightweight provision is now more widely available on the NHS) or dynamic stander. <u>Ambulant:</u> For children with SMA III assess for need for KAFOs for prolonging ambulation once losing or lost ambulation i.e. Able to walk	Children's Hospital of Philadelphia - Infant Test of Neuromuscular Disorders (CHOP INTEND) (Allan M. Glanzman et al., 2011) (A. M. Glanzman et al., 2010) is the most common one specific to SMA I. Baby scales such as the Alberta Infant Motor Scale (AIMS) can also be used. The Expanded Hammersmith Function Motor Function Scale Expanded Hammersmith Functional Motor Scale for SMA II and SMA III (HFMSE)

	less than 10 m or having frequent falls with risk of fractures. Try standing in leg gaiters as part of assessment/suitability process for KAFOs Timed tests such as the 10m walk/ run test and the four stair climb/descend test can be used to give an objective measure of speed and function.	<ul> <li>(A M Glanzman et al., 2011) or Revised Hammersmith Scale (RHS) for SMA II and SMA III are the most common and can help in making management decisions.</li> <li>Revised Upper Limb Module (RULM) is specifically designed for SMA.</li> <li>The Gross motor function measure can also be used in SMA but is very long (Nelson, Owens, Hynan, Iannaccone, &amp; AmSMART Group, 2006)</li> <li>SMA III - Endurance may be assessed using 6 minute walk test but is unusual in the clinical setting.</li> </ul>
Function and ADL	For SMA I and IIa assessment in supported sitting and in supine is key to providing good seating and postural control systems. It is important to ensure that any provision does not restrict the limited movement the child can achieve. Remember to take into account respiratory function and fatigue. Some SMA I babies will not cope with being upright or even partially reclined. Head and trunk control is poor in the weakest children and many type I have plagiocephaly. Neck and head posture need good assessment to ensure the correct supports are put in place. SMA II seating: Assess neck and trunk control in sitting. Take in to consideration muscle fatigue which will affect position achieved and how long it is maintained/tolerated. Tremor may affect fine motor function, which may worsen initially if prescribed oral salbutamol. Timed Up and Go (TUG) may be used as a measure of ability in ambulant patients with SMA (Dunaway et al., 2014).	The Egen Klassifikation Scale (Steffensen et al., 2001) (Steffensen BF, Mayhew A, 2008) can be useful in older non-ambulant individuals as a global indicator of function
Posture / spine	Older children with SMA I and weaker children with SMA II and many of those with SMA III who lose ambulation before puberty, will develop a scoliosis. Scoliosis can begin in early years with 'growing rod' spinal surgery required before spinal maturity (approximately age 11) is reached. Definitive fusion may be performed at this stage. The spine therefore needs careful and regular monitoring. N.B growth spurts and puberty precipitate, often rapidly, increasing scoliosis.	It is important to observe the spinal posture clearly to see if scoliosis is evident or curvature is progressing This can be done in sitting (and standing where possible – even in KAFOs) where both active and passive correction are noted. Check the sacral dimples are level. Check shoulders and neck levels. Check spine with child in forward flexion in both sitting and standing (if standing is possible) to assess for asymmetry of ribs, and scapulae +/- rotation. Pelvic obliquity is strongly associated with

		hip subluxation or dislocation.
Range of movement	Contractures are very common in SMA II. In the lower limbs, the knees are most affected followed by the hips and ankles (Fujak, Kopschina, Gras, Forst, & Forst, 2011). The shoulders are the most severely affected in the upper limbs followed by the elbows and wrists (Fujak, Kopschina, Gras, Forst, & Forst, 2010).	May need to perform a full ROM assessment which includes neck range and jaw opening (which can get tighter in older children with SMA).
Strength	Measurement of muscle power in SMA may be part of routine neuromuscular assessment but it is not essential for those working in the community. Changes in power are usually matched by a change in function and are particularly noticeable as children grow. Exercise cannot damage SMA muscle but should be encouraged and paced. What is most important is that lack of activity / exercise can lead to disuse atrophy in addition to their NM atrophy (McDonald, 2002).	Muscle strength can be quantified using myometry (Kroksmark, Beckung, & Tulinius, 2001).
Pain and fatigue	<ul> <li>Pain and fatigue can be an issue in SMA (Lager &amp; Kroksmark, 2015). Back pain is seen in children with posture problems mostly in standing – independently or in KAFOs and can be due to dislocated hips, or sprains, etc. Fatigue can be significant and can cause muscle aches. Simple pain and fatigue scales can be used to monitor change.</li> <li>Hip dislocation needs referral to an orthopaedic surgeon or the local NM team.</li> <li>Occasionally, hypersensitivity of hands, feet and skin may result in a dislike of being handled.</li> </ul>	If fatigue is an issue consider using fatigue measurement scales such as the <u>Fatigue</u> <u>Severity Scale</u> as this can assist with managing this on a day-to-day level and monitoring change. Hip relocation surgery is not usually recommended in SMA due to the likelihood of recurrence and post-operative complications (Sporer, Smith, S.M., & B.G., 2003) (Zenios, Sampath, Cole, Khan, & Galasko, 2005)
Management	Foundation Module a	nd Advanced Module
Respiratory	<ul> <li>Patients and their carers may require teaching of lung volume recruitment and airway clearance techniques, manually assisted coughing and/or provision of cough assist machine.</li> <li>Respiratory care may include: non-invasive ventilation (NIV), secretion management and suction. Ventilation via tracheostomy is unusual in this country although more common in other European and countries worldwide.</li> <li>Advanced Care Plans should be discussed early.</li> <li>Home suction equipment may also be needed.</li> <li>Home oxygen may also be prescribed for comfort in the later palliative stage in SMA I.</li> </ul>	
Mobility	Non-ambulant SMA II – Early power chair provision for mobility and development. Children are often able	

	to use a power wheelchair from as young as 18 months or less (Jones, McEwen, & Hansen, 2003). Some wheelchair services do not prescribe power chairs to under 5's but there is no justification for this and families benefit from therapist's support in helping them source a suitable wheelchair. Some charities may provide powered mobility for very young children (see resources).
	Floor scooter for floor mobility and play.
	Standing frame from normal standing age. Mobile (dynamic) standing frame if the child has enough strength to propel. May need to use in conjunction with leg gaiters and AFO's.
	Lightweight ischial weight-bearing knee-ankle-foot orthoses (KAFO) or reciprocal gait orthoses (RGO) may be considered for standing or assisted ambulation. Not suitable if poor neck/trunk strength (seek advice from specialist physiotherapist).
	<u>Ambulant</u> - – Balance difficulties and falls are not uncommon in SMA III as well as difficulties with distance, changes in terrain and climbing stairs.
	Wheelchair provision is often needed e.g. lightweight, self-propelling or attendant, for fatigue/energy preservation. Some young people may prefer a mobility scooter.
	Advice on suitability of wheelchair features such as powered add-ons, standing or rising facility, elevating leg rests, recline, etc.
	Several types of power assist wheels can be purchased to assist self-propelled mobility and are more portable than a powered chair for travel and independence.
	Keep footwear lightweight with firm heel support.
	Consider orthotics to aid foot position and ambulation eg heel cups, supra malleolar foot orthosis (SMAFO).
	SMA I and weak SMA II: Neck support may be needed for travelling in the car, when sick, or due to fatigue.
	Fatigue management: Strategies must be put in place to reduce fatigue. This is particularly important in SMA III where fatigue can result in falls and possible fractures.
	See section 11a for prescribing KAFO's which may prolong ambulation or support loss of ambulation
	Assessment for appropriate mobility aids, adaptive equipment, assistive technology, and environmental access to allow patients to maintain independence and mobility and to conserve energy.
Strength (and weakness)	Evidence suggests that exercise can help to build muscle in SMA. Exercise as play sport and activity should be encouraged in all types of SMA.
	In the weakest children (SMA I) play can be easiest in side lying. Taking the weight of the limb, arm or leg, can aid active movement.
	For all types, supporting the limb, such as when in the bath or hydrotherapy, can allow the child to move more easily. Hydrotherapy may not be appropriate in weakest children due to pressure of water on body when fully immersed.
	Fatigue is problem in SMA. Activities / exercise should focus on "little and often". Daily and targeted exercise is important and should be fun.
	Specific muscle groups can be targeted with use of Thera-band or light weights. It is best to avoid attempting to strengthen hip flexors, knee flexors, foot plantar flexors, forearm pronators and elbow flexors for all types. Symmetrical exercise and activity is preferred – swimming, cycling, martial arts and horse riding. Strengthening combined with aerobic exercise is more likely to be more effective than strength training alone (Cup et al., 2007)
	and strength training appears to be safe (Lewelt et al., 2015).
	Swimming/hydrotherapy is encouraged (Cunha, Oliveira, Labronici, & Gabbai, 1996) (Salem & Gropack, 2010). Adapted trike or bike may be used. Horse riding may be

	beneficial (Lemke, Rothwell, Newcomb, & Swoboda, 2014).
Contracture management	Contractures are common in SMA and prevention is the optimum strategy for management. Children can have a mixture of hypermobility and tightness. While it is not necessary to stretch hypermobile joints, it is important to keep reviewing as they can begin to tighten up.
	The most commonly contracted joints are ankles, hips and knees, elbows and supination. The ITBs can get tight when the child persistently "frog" sits and/or their legs roll out in sitting. Wheelchairs and seating with abduction wedges – not pommels - will help to prevent this and hip asymmetry, which frequently develops.
	It is important that stretches are done gently as the ankles are easily sprained. Active assisted stretches are most effective.
	Splints and orthotics are important to maintain a good position, particularly of the feet and night splints should be considered for all children. Splints for the weakest children can prevent what little movement they have so should be kept for sleep – if tolerated.
	Stretches can be done through positioning: standing, side lying, using a therapy ball and in hydrotherapy.
	Serial casting, CCDs, standing in KAFOs, all help to reduce the speed of development of contractures. Asymmetry of lower limb contractures can impact on the pelvis and spine. This is true of all types of SMA.
Range of movement	SMA I - Use of neck rolls or plagiocephaly pillows to prevent/manage plagiocephaly.
	Passive movements incorporated into songs and games.
	Facilitation of any active movement through careful positioning of child and toys for movement with gravity assisting. Use of an A-frame for attaching toys or mobile arm supports may be helpful.
	Symmetrical positioning and regular changes of position. Generally it is best to avoid prone because of restriction of diaphragmatic excursion. Prone position may be tolerated if the child is ventilated but requires careful assessment of vital signs, oxygen saturation and signs of distress.
	<b>SMA II and weaker SMA IIIs</b> - contracture prevention and management with regular changes of position, use of standing frame or KAFO's, stretching programme, orthoses, active exercise/activities, serial casting. Advice about extra vigilance around time of puberty or growth spurts.
Spine and posture	Non-ambulant children with SMA will get spinal asymmetry. Many weaker children with SMA have asymmetry of the hips. Consider spinal bracing to aid function and seating in SMA IIs and potentially in treated / stronger SMA Is.
	Weaker children with SMA III who lose ambulation before puberty and some ambulant children will develop scoliosis. Spinal posture therefore needs to be reviewed in all children in SMA. Good posture in sitting in school and in wheelchairs as it is for long periods, is very important.
	Fractures and injury can also increase asymmetry and the child will tend to lean away from the side that has been hurt. Upper limb fractures can also cause this.
Orthotics	The use of orthotics in SMA is an important part of management and it is helpful if your orthotist has experience of working with this group of children. Guidance from your regional neuromuscular service is helpful in ensuring the correct orthoses are used at the

correct time.
Spinal jackets are helpful to aid sitting, improve symmetry and sitting posture and reduce the speed of progression of a spinal curve. As the children are diaphragmatic breathers, the TLSO's should always be made with an abdominal hole.
Putting a jacket on should be done in lying and the jacket pulled down to the pelvis. If too loose it will ride up under the arms. When bringing the child up to sit, the knees should be bent up to stop the jacket slipping.
Some children prefer the jacket to be loosened to eat. To fasten it again, the child will ideally be lying.
Feet in SMA II have a tendency to evert and splints can maintain a better midline posture and prevent TA tightness. The splints can be worn at night or during the day (except in very weak children) but day splints should be cut to mid-foot to make it easier to wear shoes.

#### Resources

Spinal Muscular Atrophy Support UK Muscular Dystrophy UK - Spinal Muscular Atrophy Newlife- Foundation for Disabled Children Designability - Wizzybug MERU - Bugzi

# Limb Girdle Muscular Dystrophy (LGMD)

#### Introduction

Limb girdle muscular dystrophy (LGMD) describes a large group of conditions, which mainly affect the shoulder and pelvic girdle muscles, hence the name. Hip and thigh muscles are affected first but foot drop and hand weakness is not uncommon. Symptoms vary between the different types, as does the rate at which the disorder progresses. LGMD may also affect the heart and breathing muscles but this usually manifests later in life.

Diagnosis can be made at any age. Some children with milder forms of the condition will never become seriously affected. Others may struggle to lift their arms above their heads or may lose the ability to walk. Falls may be an issue in this group and children may find walking long distances, climbing stairs and getting up off the floor difficult.

Roughly 25% of people with LGMD are not given a precise genetic diagnosis but even with one, progression is hard to predict. About 1,400 people in the UK are affected by various forms of LGMD. Classification is alphanumeric, given on mode of inheritance (1=dominant and 2=recessive) and a letter for the order of discovery of the type. They usually have a common name too, e.g. LGMD2b is known as dysferlinopathy.

Assessment	Foundation Module	Advanced Module
Development / mobility	There are many patterns of muscle weakness in the LGMD however symptoms are often first noted in the proximal regions – shoulders and pelvis. Patients are usually ambulant in childhood and only lose ambulation later in life, if at all. LGMD's are not known to be asymmetrical in presentation but children may show differences between left and right. Assessment using timed tests can be useful - see below. First symptoms may appear as difficulty keeping up with peers in sport, falls, difficulty on stairs and over long distances. In facioscapulohumeral dystrophy (FSHD), facial weakness, hearing loss and increasing lordosis can be presenting features. Girls tend to be more severely affected than boys. In the most severe form of FSHD, the children rarely achieve ambulation.	Now over 30 sub-types identified. For details of patterns of weakness refer to (Guglieri et al., 2008) and (Murphy & Straub, 2015). LGMD2A (calpain) is most common and presents in first - fourth decades. LGMD1C (caveolin) is rare, about 1-2% but does present in childhood.
Function	<ul> <li>Modules within the Gross Motor Function measure (GMFM-88) for higher function children may be a useful starting point and simple timed tests such as <ul> <li>10m run</li> <li>time to climb a standard set of stairs</li> <li>timed rise from supine</li> </ul> </li> <li>will always be useful for comparative purposes. Rise from floor may reveal a 'Gower's manoeuvre' similar to DMD due to the proximal weakness.</li> </ul>	For ambulant children a good starting point is the NSAA however it may not pose enough of a challenge to high functioning individuals. For ambulant and non-ambulant children the <u>Activlim</u> may be a suitable Patient Reported Outcome Measures (PROM). (Batcho et al., 2016).

Posture / spine Range of movement	<ul> <li>Key points of assessment are:</li> <li>Visualisation and palpation of spine</li> <li>Pelvic obliquity</li> <li>Particular attention to foot posture</li> <li>Muscle wasting and hypertrophy*</li> <li>Spinal mobility / rigidity</li> <li>Winging of scapulae both at rest and when the arms are raised through full abduction.</li> <li>Contractures can be an issue in the group but are not common. Ankles are more</li> </ul>	Same as Foundation Module. *Specific patterns of muscle wasting and hypertrophy can be very specific to different forms of LGMD. See <u>LGMD Syndromes</u> for more detail.
Muscle strength / weakness	A thorough Manual Muscle Testing (MMT) assessment is very important in this group and can help guide functional support.	MMT in this group often assists in diagnosis and patterns are very disease specific. For example inability to stand on tiptoes, weak plantar flexors is a common first symptom in dysferlinopathy.
Pain and fatigue	Fatigue may be a presenting factor whereas pain is more unusual.	Fatigue Severity Scale. Visual Analogue Scale can be useful for individuals with significant pain issues.
Respiratory	It is important to monitor chest infections and note any symptoms of hypoventilation although respiratory issues are more commonly seen later in life.	Perform forced vital capacity (FVC) in sitting and in lying especially if the child is undiagnosed and cardiac involvement could be an issue. Knowledge of FVC in lying also may inform the potential need for night time ventilatory support. Several LGMD have cardiac involvement. Respiratory issues most likely in LGMD21 (FKRP).
Management	Foundation Module a	nd Advanced Module
Development and mobility	A thorough knowledge of levels of mobility will help inform your management. Practising tasks can be beneficial not because you can fix affected muscle fibres but you can make the most of good muscle fibres. NB - For children with some LGMD's where hamstrings are notably weak while they are ambulant, KAFOs are very difficult to walk in when the children lose ambulation and their use very frustrating.	
Range of Movement	Stretches and orthotics can be applied on a need to do basis rather than asking families to carry out unnecessary tasks. Orthotics may be prescribed by a tertiary centre. See specific conditions for details. Customised insoles are often a first step to improve posture.	
Muscle strength / weakness	Strength training may be suitable but the key advice is to keep resistance / weights very light and increase repetitions as stamina improves. It is recommended to avoid eccentric muscle loading (such as trampolining).	
Pain and fatigue	Advice on pacing is beneficial in this population <u>Dystrophy UK Education Guidelines on PE</u> ca	on, as they can fatigue quickly. <u>Muscular</u> In be useful for advice to schools.

#### Myopathies

#### Introduction

Myopathies are diseases of skeletal muscle that are not caused by nerve disorders. These diseases cause the skeletal or voluntary muscles to become weak or wasted.

The muscle architecture shows changes on biopsy that characterise the different types.

As with all NMDs, they can vary significantly in severity and presentation; but many show symptoms from birth or in infancy. Those children presenting at birth can have severe neonatal complications although some babies and children gain improvement in muscle strength and over time and with supportive therapies make progress with their motor skills and function. Many progress to standing and walking though there is a wide spectrum and others who achieve ambulation lose it over time.

Specific diagnosis is essential in giving accurate information about progression of the disorder and likely complications associated with them. There are different groups and sub groups of inherited myopathy, some forms having several different genetically distinct types that can give rise to different presentation and pattern of improvement or deterioration.

Children with congenital myopathy have normal intellectual and learning ability.

In general, children with congenital myopathies change only very slowly over time, with any rapid loss of ability and increasing asymmetry of posture being through growth spurts and around puberty.

The four main subgroups are:

- Nemaline myopathy rod body disease
- Centronuclear and myotubular myopathy (MTM1)
- Central core
- Mini core and multi minicore

One infantile form of Centronuclear MTM 1 myopathy is a severe x-linked disorder presenting with profound weakness and respiratory problems. Independent mobility is not achieved and there is a high risk of morbidity. There are milder forms.

N.B. Bethlem Myopathy is in fact not a myopathy but is a form of Congenital Muscular Dystrophy – see Congenital Muscular Dystrophy.

Assessment	Foundation Module	Advanced Module
Functional assessment	Hypotonia and generalised muscle weakness are features in myopathies. Nemaline myopathy and myotubular myopathy may present neonatally. In young babies assess levels of spontaneous activity and voluntary movement. Monitor achievement of developmental milestones. Most children with central core and multi- mini core myopathies achieve ambulation. In nemaline myopathy function is related to underlying genetic diagnosis. Many do eventually walk. Those with milder forms of Centronuclear myopathy often walk until teenage years. Therefore the assessment chosen will depend on age and severity of the disorder.	Infant assessments such the <u>CHOP-</u> <u>INTEND</u> can be useful to monitor progress in the weakest babies. For older children who are non-ambulant an SMA style assessment of function can be useful. For the ambulant children, ambulant scales such as the <u>NSAA</u> can also be used.
Respiratory	Bulbar weakness and ineffective cough means these babies/children are vulnerable to chest infections and respiratory	FVC and PCF should be performed. Monitor signs of nocturnal hypoventilation such as morning headaches, nausea, a lack

	compromise. Therefore it is important to monitor the frequency of chest infections, hospital admissions and assess a child's ability to clear secretions.	of feeling rested, etc.) and refer if appropriate especially if associated with poor respiratory function.
	is a risk of morbidity from feeding and respiratory problems however this risk reduces with age, but monitoring remains important.	
	Central core and mini core disease are frequently less severe and while a small number of children may not achieve independent ambulation, respiratory problems are usually later manifestations.	
Function/ADL	Assess the strength of the trunk as appropriate trunk supports may be required to enable / maximise upper limb function in weak children.	
	Writing may be tiring due to shoulder and upper limb weakness and may need assessment at school.	
Posture and spine	Scoliosis is a common feature of myopathies and regular monitoring is needed to ensure symmetrical seating, posture and gait.	
ROM	Check for evidence of muscle tightness and contractures or excessive range/laxity. Formal measurement should be recorded if contractures noted as these may well change over time depending on growth and level of weakness.	Assessment of hips in the neonatal period is important. Incidences of hip incongruity may require orthopaedic opinion. Talipes may be present and need assessment and management.
	Central core patients require assessment	
	and monitoring of their hip incongruity.	
Strength	and monitoring of their hip incongruity. Weakness occurs in a more proximal presentation and is mostly symmetrical but this does vary across myopathy type. Seek specialist support about different types of myopathy for more specific information.	
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	Myotubular myopathy.
Mobility	It is important to advise families that mobility may well be delayed and therefore provide advice and activities to promote motor development.
	Assess for walking aid and when considering which type of walker is best try and avoid heavy walkers that the child will be unable to propel or is required to lift as they are unlikely to have the strength for this.
	Consider early referral for wheelchair for less ambulant or those with limited ambulation for longer distances / outdoor mobility. Electrically powered wheelchairs should be considered from around two years of age for the more severely affected. Lightweight self propel wheelchairs should be sought where possible. These can be provided through the NHS or charity funding.
	KAFO's may be appropriate in some children. It can be useful to try a young child in gaiters first to determine suitability. Liaise with specialist centres for this.
Function/ADL	Considerations for suitable footwear: avoid heavy and bulky shoes / boots that weaker child will not be able to manage.
	Congenital Talipes Equinovarus (CTEV) management – Ponseti if applicable.
Range of movement	Stretches should be taught and stretching programme provided for carers where loss of range is noted, particularly in young children.
	Splints / gaiters may be appropriate to help maintain range, but should not inhibit functional mobility.
Posture	Advise parents on positioning and postural support to prevent asymmetry. In the neonatal period, positioning must take into account respiratory function as well as head and neck posture to prevent any cranial asymmetry and hip positioning.
	Children will need on-going appropriate seating and postural management.
	Non-ambulant children may benefit from upright standing frames. However they should be used only if a symmetrical posture is achieved and the child is enjoying standing.
	Consider standing frames where a child can have degree of independence and control over activity (dynamic standers).
Spine	Bracing may be needed to assist spinal posture, and provide a stable base to improve arm function.
Exercise and activity (strength)	Be mindful of respiratory insufficiency (physiological effects of submersion for example when using hydrotherapy).
	Symmetrical and fun activities are encouraged and most beneficial. Consider low-geared trikes.
	Be mindful of high energy activities as they may not appropriate for weaker children due to the risk of injury. For example treadmills and trampolines.
Pain and Fatigue	Pain is not a feature of myopathy as such, but fatigue often is. Give family advice on pacing, use of buggy / wheelchair when going longer distances for more ambulant children.
	Feeding difficulties and not getting enough nutrition may affect fatigue levels

#### Congenital Muscular Dystrophy (CMD)

#### Introduction

Congenital Muscular Dystrophies (CMD) are progressive conditions, although many deteriorate only very slowly. Learning difficulties may be present but intellectual ability does not deteriorate over time. As with all disorders, the spectrum of disease even within a diagnostic group can be wide. In CMD it is no different and in most cases it is relevant that the earlier the appearance of symptoms and difficulties, the more severe the eventual outcome.

The disorders that come under the umbrella of CMD can be generally divided into two main clinically different types; those such as Merosin Deficient MDC 1a, Bethlem/Ullrich, and Rigid Spine Muscular Dystrophy, where the contractures are often more debilitating than the underlying weakness, and the disorders which fall into the dystroglycan sub-group such as Muscle-Eye-Brain disease (MEB) where learning difficulties and upper motor signs can have a significant effect on function.

In the former group, the contractures can be present from birth, or develop quite rapidly with growth and therefore early diagnosis and management are important.

Neck and spine rigidity and jaw limitation are features of MDC 1a, and unusually, shoulder tightness, particularly contractures of deltoid, occur along with the more frequent limb contractures. These can have a marked effect on function. While it is important to try and maintain joint range, over time contractures *will* increase due to the specific underlying pathology.

Bethlem myopathy (BM) and Ullrich Congenital Muscular Dystrophy (UCMD), are genetically differing severities of the same condition (collagen VI disorders). This is a spectrum with Ulrich at the more severe end and Bethlem the milder. An overlap of more severe Bethlem/milder Ullrich in the mid-range.

In the more severe Ullrich, the children may never walk or lose ambulation before puberty; respiratory problems can be an early feature. Diaphragm involvement can cause difficulties breathing when lying flat.

Children with rigid spine may remain ambulant into adulthood but have progressive rigidity and weakness of the neck and spine making neck ache and back ache a particular problem. Scoliosis may be less significant and rarely need treating. Respiratory compromise increases with age.

Non-ambulant children, in particular those with marked contractures and/or scoliosis, may frequently wake in the night to be repositioned due to discomfort. It is not unusual for parents to say that 8 times is a "normal" night. Some may be using NIV, but this does not prevent them needing repositioning.

Assessment	Foundation Module	Advanced Module
Mobility	Children with CMD differ in their ability to walk but it would be unusual for a child over the age of 3 to begin walking independently.	
	Non-ambulant assess floor mobility.	
	<b>Ambulant</b> children with Intermediate, Bethlem/Ullrich, milder MEB, and more severe rigid spine can all lose ambulation.	
	Assess gait in shoes and barefoot.	
Function and ADL	Ambulant: consider NSAA	
	<b>Non-ambulant</b> : particularly smaller children, SMA scales are helpful.	
	For older non-ambulant who have marked upper limb weakness and/or contractures, the EK scale is helpful.	
	Hand function or upper limb assessments will need to consider the reduced range of	

For an overview see Sparks et al. (1993).

	movement.	
	ADL, developmental scales and QOL scales can all be used depending on the setting and relevance to the child, family and situation.	
Posture Spine and neck	Non-ambulant children assess in sitting and lying.	
	Neck ROM,	
	Spinal mobility and rigidity	
	Thoracic rotation,	
	Hip and pelvis alignment	
	Ambulant children assess in sitting and standing particular note being taken of differences between the two. This may lead to further assessment of leg lengths, or other parameters.	
Strength /Power		Can make use of MRC grading scales.
		Contractures may restrict ROM so measure power through the available range; e.g. elbow flexion contracture of 25° but power in the available range is 3+.
ROM	All joints should be assessed at screen	
	including neck and spine, jaw and shoulders.	
	Then use targeted follow up ROM assessments.	
	Measurement using goniometry for elbows, knees, hips and ankles may be helpful.	
Pain and fatigue	Pain: spine and neck pain often postural.	
	Hip pain and pain from contractures particularly when the child is moved or stretched.	
	Check type, frequency, intensity, aggravating and relieving factors, duration and management.	
	Fatigue: am/pm, length of school day consider respiratory factors.	
Management	Foundation Module	Advanced Module
Mobility	Consider gaiters and / or standing frame to pr	omote standing.
	Powered wheelchairs should be considered fr and for nursery/pre-school for the more sever Intermediate, Bethlem/Ullrich, milder MEB, an ambulation.	om the age of two for children with MDC1a e Ullrich. Ambulant children with d more severe rigid spine can all lose
	A powered wheelchair will be necessary as an	m power is rarely adequate to self-propel.
	Consider KAFOs for promoting ambulation or	supporting loss of ambulation.

Function and ADL	All children will need for help in function, ADL and mobility.	
	Early wheelchair mobility for development of non-ambulant children is important but those who can still walk, stand or mobilise in orthotics should continue to do so.	
Exercise and activity	Encourage daily exercise/activity daily through play for all.	
	Symmetrical activity is best so scooters used for long distances should be avoided.	
	Swimming, horse riding, cycling, martial arts can be beneficial.	
	Rugby, trampolines, weights, more advanced gymnastics, ballet are generally discouraged.	
	Hydrotherapy is useful for the least mobile and those losing ambulation.	
Posture and spine	Regular reviews of the spine, neck and pelvic alignment are needed and key during periods of rapid growth (ideally as often as every three months).	
	Evaluation of posture in the home and school situation with changes made where necessary to seating, positioning and/or orthoses.	
	Collars should not be used except for travelling if needed and in times of severe pain or fatigue. Prolonged use can further weaken neck muscles making head control harder.	
Joint range	Stretches should be active assisted and child is asked to "help". Be aware of over stretching as it can cause pain and reduce co-operation. In the older child, self-stretches are encouraged and should be reviewed and supervised as required.	
	N.B. Children with Ullrich have increased dorsiflexion and reduced plantarflexion, so TA stretches are un-necessary.	
Orthotics	The use of orthotics is widespread and varied in this group.	
	<b>Spinal support</b> : TLSO (jackets, braces see Orthotics section) are used to enable some of the younger / weakest children to achieve independent sitting:	
	Improve symmetry of sitting,	
	Increase sitting tolerance	
	Reduce the fatigue of sitting.	
	Try to reduce the progression of scoliosis.	
	Improve upper limb function	
	Early intervention with 'growing' spinal rods or magnetic rods may not alleviate the need for a TLSO as spinal surgeons may still advocate their use to protect the instrumentation.	
	A TLSO is not used for kyphosis and in the children with neck rigidity, it may actually restrict their ability to look forwards	
	Gastrostomy and/or respiratory compromise necessitate the use of an abdominal hole in all jackets to make them easier to tolerate for long periods.	
	Lycra suits for spinal posture control has not been researched and no evidence supports their use at this time.	
	The consensus among experienced therapists is that the level of control needed for posture support, improved sitting and reduced rate of scoliosis progression needs at least a semi-rigid orthoses.	
Pain and fatigue	Pain: seating and positions can be a major factor in pain management.	
	Stretches in the bath may be easier for small children.	
	Fatigue: pacing is very important particularly in school at break times and with PE/games sessions.	

For details of the many subgroups refer to the subgroup of Online Gene Table - Neuromuscular Disorders

Cure CMD

Treat-NMD

Muscular Dystrophy UK - CMD

#### Congenital Myasthenic Syndrome (CMS) and Myasthenia Gravis (MG)

#### Introduction

When most people talk about **myasthenia**, they mean **Myasthenia Gravis** (MG), a chronic autoimmune neuromuscular disease characterised by varying degrees of weakness of the skeletal (voluntary) muscles of the body. MG affects children as well as adults and is caused by antibodies, 'immune' proteins that block, alter or destroy the acetylcholine receptors resulting in less effective signalling from nerves to muscles. This is an autoimmune condition like rheumatoid arthritis and can be treated with steroids, immunosuppressive drugs and thymectomy (surgical removal of the thymus gland).

**Congenital Myasthenic Syndrome** (CMS) on the other hand is the collective term for a group of inherited disorders of neuromuscular transmission - the mechanism by which messages from the nerves are sent across to muscles to make them work. This causes weakness in the muscles (myasthenia), which fatigue easily when they are required to work. CMS does not affect the 'automatic' muscles such as the heart, gut, bladder, etc. They do not respond to steroids and the other treatments suitable for myasthenia gravis, which should be avoided in CMS patients. In Congenital Myasthenic Syndromes, the muscle weakness typically begins in early childhood but can also appear in adolescence or adulthood. Facial muscles, including muscles that control the eyelids, eyes, limb muscles, and the muscles used for chewing and swallowing are the most commonly affected.

Management of these conditions is a combination of medication and guidance on pacing physical activities. Most children with myasthenia will be ambulant and live active healthy lives.

Assessment and management of this group is very often led by the specialist centre rather than involving community staff. However an understanding of these conditions and assisting with fatigue management locally and particularly in schools is important.

Assessment	Foundation Module	Advanced Module
Respiratory and bulbar function	Children with all types of myasthenia can have difficulties with their breathing, speaking and bulbar function. On the whole deterioration in these components is evident after prolonged activity and with fatigue. In certain sub types of CMS – e.g. RAPSYN & EPSILON respiratory function can be severely compromised in early childhood and often improves to near normal after the first 2 years of life. Assessments for respiratory function may include forced vital capacity (FVC). It is important to ask the child/ parent about swallowing, choking episode, chewing difficulties. Noting that changes may indicate a need to review management.	It is important to recognise the specific sub- types of CMS and their different presentations. Early detection can be of significant importance when supporting medical management in infancy – e.g. respiratory function severely compromised in infants with RAPSYN. Regular sleep studies are important in recognising where the provision of non- invasive ventilation is indicated. (Hull, 2012) Specific speaking assessments are used to ascertain any slurred or nasal speech. The "Me-bee"/count 1-50 assessments are often used in clinic.
Mobility	<ul> <li>In babies and young children a detailed histor is important to highlight any risk factors of con It is important to ascertain their current function fluctuations (e.g. within the last 4 weeks). Stree all affect a child's function and mobility.</li> <li>An assessment should include details of: <ul> <li>Current level of ambulation – including intrable to walk before fatigue</li> <li>Wheelchair use – when and how often is has adequate support to reduce fatigue</li> </ul> </li> </ul>	y of early year's development of motor skills atracture / weakness. onal baseline, including any recent ess, temperature and hormonal changes can doors, outdoors and the distance the child is it required. It is important to ensure the child

	• Stairs – how do they ascend/descend, support required, fluctuations throughout the	
	<ul> <li>Gait analysis – look for muscle weakness both proximally and distally, plus trunk and neck position. Note any changes in gait pattern with prolonged walking, e.g. 6MWT</li> </ul>	
	It is important to note that several factors can impact on the performance observed on day of assessment, including recent levels of activity, hormonal changes, timing and changes to medication, fatigue and stressors; such as exams.	
Function and ADL	Children with myasthenia may fatigue quickly and it is therefore important to keep the number of assessments performed to a minimum, ensuring they are standardized and relevant to each individual.	Timed Tests can be very useful in evaluating the effectiveness of therapy intervention and medication. Repeated tests should highlight any muscle fatigability. These may include but are not limited to: Repeated myometry Timed rise from floor 10 meter run / walk 6MWT Gait analysis Sit to Stand in a minute
		• Static arm raise/ leg raise Care must be taken to time these assessments with medication as this can bias the results.
		There are currently no widely available disease specific assessments appropriate to use in children with myasthenia.
Posture / spine	Visualization and palpation of spine is important in both sitting and standing as some children with myasthenia are susceptible to developing scoliosis, especially those who have limited ambulation.	
	These children will need assessment and provision of an appropriate wheelchair to support independence.	
	The wheelchair needs assessing regularly to ensure patients have a good spinal posture and adequate support. Taking special care of head posture to accommodate possible altered vision and neck weakness.	
	Children often have visual difficulties due to restricted eye movements, diplopia (double vision) or ptosis (eye lid closure) therefore positioning in the classroom needs to be optimal. Neck weakness and muscle fatigue can be caused by continual looking down and up (e.g. from the whiteboard at school). An Occupational Therapist could help to improve this with aids such as sloping boards and advice on seating.	
	Not all children with congenital myasthenia will develop a scoliosis. DOK7 and COLQ are the most common subsets that may develop scoliosis, due to limb girdle weakness.	
Range of movement	Knowledge of normal joint range in children is important. When assessing a child with CMS, the therapist should note range of movement into hip extension, knee extension and ankle dorsiflexion. This is especially important in the weaker infant or a child that sits for prolonged periods of time, e.g. wheelchair user.	
	Evaluate difference between passive ROM (PROM) and active ROM (AROM) to highlight any muscle weakness.	

	Always assess for the need of orthotics and check fitting, use and compliance with existing orthotics.		
	Joint contractures can develop in adolescents with CMS, due to prolonged immobility.		
	Contractures can develop in this population of children due to reduced mobility and		
	prolonged static postures, secondary to fatigue.		
Strength	<ul> <li>Fatigable muscle weakness is a key feature of myasthenia should be assessed separate from muscle strength. Look at repeated movement or prolonged static postures to help assess muscle fatigue.</li> <li>Repeated movement: e.g. sit to stand in one minute, look for fatigue during the minute.</li> <li>Straight Leg raise in supine, at 45 degrees for as long as possible, up to 100 seconds can assess fatigue.</li> </ul>	MRC grading all major muscle groups should be carried out in the assessment. It is important to note antigravity versus sub- gravity muscle power. Repeated myometry assessment for fatigability: e.g. hand grip, ten maximal repetitions with a five second hold. Evaluate fatigue by comparing the difference between the average first and last three scores.	
Pain and fatigue	Fatigue is a key feature of myasthenia. It is important however to note the difference between general fatigue and muscle fatigue. Patient reported fatigue with the absence of muscle weakness should be treated with caution and may be a feature of depression or chronic fatigue. <u>Fatigue Severity Scale</u> or Borg Scale (see Appendix 2).		
	<ul> <li>Fatigue in children with myasthenia can fluctuate though out the day, on a day to dan a dan affect all muscle groups – including respiratory muscles. Often muscles of face and mouth can fatigue causing droopy eyelids, slurred speech and difficulties and swallowing.</li> <li>Pain is not a key feature of myasthenia, but can be associated with increased patier activity levels, muscle imbalance or injury. Exercise induced myalgia is occasional reported in children with myasthenia. Location, duration, intensity, aggravating and factors are all important to assess when patients report pain. The use of a pain diabe of benefit.</li> </ul>		
	Knowledge of medication regime is an importa patients with myasthenia, as time from / until n function.	ant factor to consider when assessing next medication can have a direct impact in	
	<b>Vision:</b> assessment of Diplopia (double vision Remembering both of these can affect other to the school setting.	n) and Ptosis (droopy eyelids) is important. ests and recommendations, especially within	
Other	<b>Medication:</b> a detailed history of medication should be taken, including timing of medication (to establish if fatigue / weakness seen on examination is because the next dose of medication is due) and any recent changes to regime.		
	A child's medication regime is often altered to reflect changes in a child's function and can be adjusted at times of growth, puberty, illness, etc.		
	Certain medications work fast on reversing the effects of myasthenia and others take longer to establish in the system, for example:		
	<ul> <li>Pyridostigmine – works within 30 minutes fatigue may be seen towards the timing or</li> </ul>	s and wears off after 4 hours, symptoms of of the next dose.	
	<ul> <li>Salbutamol – takes several weeks to buil does may not be evident in showing fatig</li> </ul>	d up in the patients system and a missed ue.	

Management	Foundation Module and Advanced Module
Respiratory	Respiratory muscles can be weak in children with myasthenia (especially those with RAPSYN) and may require non-invasive ventilation to support breathing, particularly at night time, when unwell or going through a crisis.
	In order to support children and their families, knowledge and application of secretion clearance / maintenance techniques are important. (See respiratory section for further information).
	Children with myasthenia may be at risk of having chewing and swallowing difficulties leading to choking or aspiration. These can be ascertained by taking a detailed history from the patient of parent and timely referrals on to relevant services.
	Assessment and provision of respiratory support can be a key adjunct in a child's therapy.
	Children with myasthenia are susceptible to quick deterioration in their respiratory function and timely intervention is key in best management.
	Swallowing assessments such as the Slurp test should only be carried out in a safe environment in case of aspiration / choking.
Mobility	Children should be provided with suitable mobility equipment to support optimal participation in life. This may include walking aids to support motor skill acquisition when young and timely referral to wheelchair service.
	Patients, families and schools need to be taught about structuring and pacing of activities to avoid fatigue. Each individual should be able to recognize their first sign of fatigue; this is very variable and could be weakness of the legs, slurred speech or droopy eyelids, to name a few.
	In children with myasthenia gravis, early rehab post-acute onset of crisis is important to support return to normal function as soon as possible.
Function and ADL	The physiotherapist will play a key role in educating adults supporting the child about the effects of myasthenia and strategies to help manage fatigue.
	Timely referrals, where necessary, is key in supporting access and independence. Including early referral for home and school adaptations (e.g. wheelchair accessible housing / stair bannisters) and wheelchair services.
	If feeding concerns are present patients will need speech and language therapy input and may need support around altered meals and longer lunch times at school, etc.
	Optimal placement in the classroom environment may reduce the need to continuously focusing on and/or looking up, which is difficult if neck muscles are weak.
	The timing of medication can be a key factor in the management of fatigue and regular medication may need to be taken into school and dispensed.
	The physiotherapist at the tertiary centre will offer support and guidance to the community physiotherapist as required. As myasthenia (especially CMS) is a relatively rare condition, it is not uncommon to adopt a collaborative approach to therapy between community and tertiary centres.
	Advice and education to school can be hugely beneficial in supporting a child's access to the curriculum. Options could include recording sessions, provision of handouts, or an interactive tablet to reduce head and neck movements and assist with visual concerns.
Posture / spine	As with all children who present with NM weakness the management of the spine is essential. This includes advice and education on good postural habits to the family, child and school, on the importance of symmetrical postures, activities and good seating options in spinal care.
	A small number of children with CMS will require spinal surgery and is more common in

	<ul> <li>certain genetic subsets, (COLq and DOK7). Pre-operative planning and post-operative management will need to be considered, ensuring adequate support is available for likely post-op fatigue.</li> <li>Continued monitoring of spine posture and timely referral for spinal assessment / intervention and orthotic provision as required.</li> </ul>
Range of movement	There are a number of management options that could be considered in maintaining joint range of movement, including: <ul> <li>Stretches</li> <li>Orthotics</li> <li>Serial casting</li> <li>Postural management</li> <li>Activity and exercise</li> </ul>
Exercise and activity (Strength)	Children with myasthenia should <i>not avoid</i> exercise. Instead an age appropriate exercise program and activity advice should be given and monitored closely. Specific work around pacing of activities may be carried out at the tertiary centre, where advice is given in tandem with medication management. The child / family will be aware of 'tell signs' of fatigue – ptosis, slurred speech, breathing changes. Activity plans should be adhered to. Keeping an activity diary may be of benefit to identify any times of increased fatigue. Children with myasthenia can increase muscle strength with targeted strengthening programs. Caution should be used with the number of 'sets' and 'reps' a patient can complete before fatigue. Education to school / nursery regarding the condition and appropriate inclusion of PE and games activities within limitations. Although a child with myasthenia may struggle to carry out high levels of aerobic activity (e.g. swimming, running)
Pain	<ul> <li>Muscle cramps and exercise-induced myalgia are often reported in individuals with myasthenia. Advice for cramp management may include:</li> <li>Warm baths</li> <li>Massage</li> <li>Stretch</li> <li>Although it is important to be aware that heat often makes the symptoms of myasthenic fatigue worse and should be used with caution.</li> <li>Referral to a pain clinic may be required in certain cases.</li> </ul>

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 Neuromuscular Disease Center

 National Institute of Neurological Disorders and Stroke

#### Peripheral Neuropathies / Charcot-Marie-Tooth / Hereditary Motor Sensory Neuropathies

#### Introduction

Charcot-Marie-Tooth (CMT) disease the most common inherited neurological disorder that comes under the neuromuscular umbrella. Symptoms often present in childhood and usually demonstrate a slowly progressive course. It is a group of conditions that affects the peripheral nerves and is sometimes known as hereditary motor and sensory neuropathy. There are many different types of CMT, some of which are dominantly inherited and some recessively inherited thus other members of the family may be affected too.

CMT is caused by mutations in the genes that produce the proteins involved in the structure and function of either the peripheral nerve axon (axonal) or myelin sheath (demyelinating). These proteins affect the normal function of the peripheral nerves. In the axonal form, the nerves slowly degenerate from distal to proximal and this results in muscle atrophy and weakness in the extremities. This can also lead to degeneration of the sensory nerves, which can result in a reduced ability to feel temperature and pain. In the demyelinating form sensory involvement is rare and degeneration is often slower with less functional difficulties. The most common types found in children are CMT 1A (demyelinating) and CMT 2 (axonal neuropathy).

As a result of the muscle weakness particularly in the lower legs and feet, these children and young people may develop secondary foot deformities and have difficulties standing, balancing and have altered gait pattern. Most often this will be a pes cavus (+/- hammer toe) type but pes planus may also be seen.

Although this is a genetically inherited condition, symptoms and severity can vary hugely between family members.

Assessment	Foundation Module	Advanced Module
Mobility	Subjective assessment can give an idea of what functional difficulties these children and young people have. Gait analysis both barefoot and in footwear is essential with these patients. Look out for a high stepping gait, foot drop, reduced heel strike and altered balance. Gait analysis should be considered over varying distances and terrain to give a clear indication of the patient's difficulties. It is also important to complete a stair assessment. Time over 10 metre walk test can also be useful. Foot assessment – looking for a cavus or planus foot, hammer toes, claw toes, patterns of callous formation, altered position of calcaneus. Consider also any difficulty in maintaining a static standing posture and static and dynamic standing balance assessment.	<ul> <li>Formal gait assessment tools can be used to assess deterioration in gait parameters. (Pagliano et al., 2011) including:</li> <li>6 minute walk test.</li> <li><u>Foot Posture Index</u></li> <li>CMT paediatric scale (CMT Peds) (Burns et al., 2012)</li> <li>Drawing around feet can also be helpful to monitor change.</li> </ul>
Function and ADL	Assessment of fatigue levels can be useful if this is an issue.	<ul> <li>More advanced measured include:</li> <li>Adapted Paediatric Evaluation of Disability Inventory</li> <li>Grip strength</li> <li>Modified Fatigue Impact Scale</li> </ul>
Posture / spine	Scoliosis may develop due to muscle imbalance, altered gait patterns and altered posture therefore monitor 6 monthly or more frequently during periods of rapid growth.	Monitoring of the spine and referral to the Spinal Surgeons for further assessment if indicated.

Range of movement Sensory Assessment and Pain	Accurate assessment of ROM is important. Range of movement can be limited by muscle weakness or tightness in opposing muscle groups and contractures are more common in finger flexors, wrist flexors and ankle plantarflexors and toe flexors. Ensure that the subtalar and forefoot position are fully assessed. Be aware of a potential sensory component.	Referral on to Orthopaedics/plastics for assessment and possible surgery if foot deformity is causing pain, falls, significant pressure concerns/skin breakdown Joint working with Orthotics may be necessary to develop appropriate orthoses particularly for the feet and ankles. Axonal neuropathies more commonly involve sensory nerves so assessment of proprioception, light and firm touch, pain and ability to distinguish hot and cold may	
		form part of your assessment. Pain may also be an issue. (Ribiere et al., 2012).	
Strength		Muscle strength is measured through manual muscle testing and myometry. Full assessment using the oxford muscle scale is required especially of the distal muscles. Do not forget to assess the smaller muscle groups in the hands and the feet.	
Respiratory		The respiratory system is rarely affected by peripheral neuropathies. However, it is something to be aware of in extremely rare forms.	
Management	Foundation Module and Advanced Module		
Mobility	Consider appropriate orthotic management such as insoles or splints if required. These patients may benefit from ankle supports such as the trilock ankle support for physical activities if they have a significant pes cavus.		
	Contracture controlled devises (CCDs) are ge	nerally ineffective with this patient group.	
	Refer to: (Manila M, Kadian C, Futterlieb E, 2014).		
Function and ADL	These children and young people can have difficulties with fine motor skills, e.g prolonged writing, opening bottles, buttons etc and may benefit from referral to an occupational therapist for an assessment within the school or at home.		
	Patients can have altered proprioception and sensation thus providing advice about careful foot care and hygiene; prevention of pressure sores may be required especially with regards to orthotic management.		
	Fatigue management education is important. Try pacing and spacing, prioritisation, energy conservation.		
Posture / Spine	Advice on appropriate seating, postural mana rehabilitation and monitoring of the spine is es	Advice on appropriate seating, postural management and back care advice, gait rehabilitation and monitoring of the spine is essential.	
ROM	When teaching passive stretches for tight muscles in the young child it is important to be aware awareness that because of the inherited nature of CMT the parent MAY also be affected so may struggle to do the stretches. Orthotics or teaching a support teacher in school to perform stretches may be able to help ameliorate this In CMT, patients can have a degree of joint hypermobility so advice on joint protection and		

	management of this hypermobility may be required. Do not forget that a hypermobile joint may become a tight joint with growth and disease progression (Lencioni et al., 2014).
Strength	Randomised, controlled studies have previously shown the positive effect of resistance training in CMT populations (Chetlin, Gutmann, Tarnopolsky, Ullrich, & Yeater, 2004). Furthermore, exercising should be encouraged, since a sedentary lifestyle and secondary weight gain can cause a deterioration in symptoms in CMT.

Muscular Dystrophy UK - CMT National Institute of Neurological Disorders and Stroke - CMT NHA Choices - CMT Neuromuscular Disease Center CMT UK

# **Congenital Myotonic Dystrophy**

#### Introduction

This is the early childhood form of Myotonic Dystrophy (DM1) with symptoms being present at or shortly after birth.

The mother (or father in 9% of cases) already has the disease though s/he may be unaware of it and the baby has the more severe form.

N.B. Mothers / fathers and even grandparents will be affected but may have been undiagnosed. Some may have social/communication difficulties and need extra support to look after the affected child.

Myotonia - <u>delayed relaxation of a muscle after a strong contraction</u> - is rarely present in young children but develops in adolescence and is often evident in grip release initially.

#### Types of congenital myotonic dystrophy

- Neonatal and Infant
- Childhood-onset (apparent between 1 to 10 years)
- Juvenile (signs before <18 20 years)</li>

At birth the baby may have significant respiratory difficulties requiring support due to severe hypotonia. There may be poor head control and facial and bulbar weakness leading to feeding difficulties and risk of aspiration. This is particularly significant if the baby is premature. Gastrostomy is often required.

Once helped through the neonatal period the child will make progress and the low muscle tone does improve with age and there is increased strength and movement in limbs, though there will be significant developmental delay, which responds to physiotherapy input. Most children will become ambulant by age 5-6 years though may need orthotics and walking aids/wheelchair for distance. Talipes is a common feature; some children require corrective surgery though others respond to stretching/use of splints.

Alongside motor delay these children have a facial myopathy with reduced facial expression and 'tented' or 'carp' mouth affecting communication, speech and lip closure when eating. Squint or visual impairment may also be present.

Learning delay is often significant with poor motivation and concentration delaying motor progress further and excessive fatigue limits exercise tolerance, shorter bursts of activity with rests can work well.

Anaesthetics pose a risk of malignant hyperpyrexia and doctors should be aware of the diagnosis prior to any surgery.

Natural history of overall disease progression in congenital myotonic dystrophy is still to be fully determined.

Assessment	Foundation Module	Advanced Module
Neonatal & Infant presentation	Neonatally, babies have respiratory and bulbar weakness that can be severe requiring ventilation and tube feeding.	
	The following signs denote more severe or early onset phenotypes. Usual assessment techniques of observation and handling apply.	
Development	Severe hypotonia, with respiratory compromise and motor delay +/- visual impairment are significant	
	<ul> <li>Activity of lower limbs may be reduced compared to upper limbs</li> </ul>	
	<ul> <li>Head and neck weakness, often inability to protect airway particularly in prone position</li> </ul>	AIMS can be used to assess development in youngest children
	<ul> <li>Facial myopathy - 'carp' mouth, ptosis, atrophy and lack of expression (Harper PS, 2001)</li> </ul>	
	• Dysarthria	

	Plagiocephaly and head preference	
	<ul> <li>Talipes - at or afterbirth, or may develop within first few months – toe</li> </ul>	SALT referral for assessment and guidance on swallowing and feeding
	<ul> <li>Wrist and hand contractures (i.e. arthrogryposis)</li> </ul>	Orthopaedic team referral and orthotics when appropriate
	<ul> <li>Spinal deviations (scoliosis, kyphosis) with sacral sitting a common feature</li> </ul>	
Childhood onset and young child (2 years	Developmental history and motor milestones will be delayed.	May require TA release prior to ambulation.
plus)	Foot posture/stability may be altered.	Scales up to 42 months old) – (Harris, 2008).
Development and	Check.	
Mobility	gait and ability to run	
	frequency of falls	
	walking distance	
	ability on steps/stairs	
	ability to get on/off floor	
Function and ADL	Learning delay and reduced social and communication skills play a significant part in motor delay and function in this group.	Picture Vocabulary Peabody Test (currently considered as an outcome measure for clinical trials in early onset DM1 to assess
	Delayed toilet training is a frequent feature.	cognitive level).
	As part of this condition some children will	
	be diagnosed with autism spectrum disorder	
	or psychiatric disorders (e.g. anxiety) and	
	abnormal visual-spatial abilities that affect	
	Plate, Samuelsson, Tulinius, & Wentz,	
	2008) (DOUNIOL et al., 2012).	
Older child and	Additional features may be:	
.luvenile onset	<ul> <li>poor concentration</li> </ul>	
	low motivation	
10-10 years	fatigue	
	<ul> <li>in second decade child will start</li> </ul>	
	developing symptoms of myotonia -	
	difficulty with grip release particularly.	
Range of movement	Generalised joint laxity and low tone are common features.	
	Check for contractures, prior talipes management is common. Check for	
	tightness in calt complex, and tendency to to toe-walk.	
	Assess spine, thoracic kyphosis frequently develops as the child grows.	
Differential Diagnosis	(Para) Myotonia Congenita is a distinct condit	ion with different features
Respiratory	Note frequency of chest infections.	Respiratory – FVC in sitting and lying - may
	Consider aspiration if infections occurring	be difficult due to learning, compliance and facial myopathy. Overnight oximetry may

	frequently.	be required. - 71% might require respiratory support at start (Campbell, Sherlock, Jacob, & Blayney, 2004).
Management	Foundation Module	Advanced Module
Development	<ul> <li>Children do progress significantly with their development despite severe neonatal difficulties and the usual techniques to promote motor development apply:</li> <li>Use positioning for play and development</li> <li>Avoid prone initially in the weakest children as they are unable to protect their airway even beyond 2 years of age.</li> <li>As development progresses and trunk and upper limb strength improves e.g. rolling develops, prone can be used with care, start with the child positioned prone over the carer's shoulder.</li> <li>Supportive seating/ standing equipment</li> <li>Stretches to improve/maintain ROM in weaker limbs</li> <li>Orthotics –AFOs to maintain ankle ROM and/or provide stability for standing/ walking</li> <li>Activities and exercises to maintain spinal and pelvic mobility</li> <li>Active aerobic play to promote cardio vascular fitness within limits of fatigue</li> <li>Hydrotherapy</li> </ul>	Manage progression of joint contractures Fatigue compensation techniques – graded exercise in evenly spread bouts (not yet tested in paediatrics but giving good results in juvenile and adults). Balance and proprioception exercise program has shown increased in balance confidence in adults – may be useful for children with risks of falls (Hammarén, Kjellby-Wendt, & Lindberg, 2015). Pain management.

#### **Respiratory Management of the child with NMD**

#### Introduction

Respiratory impairment in children with neuromuscular disorders can vary greatly among the different conditions. Acute respiratory failure associated with respiratory infection is the most frequent reason for unplanned hospital admissions, and chronic respiratory failure is a frequent cause of death. With appropriate intervention, the incidence of unplanned hospital admission can be reduced and life expectancy can be improved (Hull, 2012).

Muscular weakness can lead to impairments in cough strength and limitations in achieving good ventilation, most notably at night time during REM sleep. Physiotherapy assessment and suitable intervention is very important in this group of children and seen as a vital component of their continuous care. Vital capacity and peak cough flow should be measured at each routine hospital appointment (Hull, 2014). Airway clearance techniques, such as mechanical insufflation-exsufflation, manually assisted cough and breath stacking should be adopted as appropriate.

Appropriate ventilation support is important in this population and is often monitored through routine polysomnography. This may include the use of non-invasive ventilation or tracheostomy ventilation.

Each child should have a child specific respiratory care plan identified and provided by a physiotherapist in consultation with the MDT. Child specific respiratory care plans are an important part of ensuring good communication between the tertiary centre and community teams and should be reviewed on at least an annual basis.

Assessment	Foundation Module	Advanced Module
Subjective	It is important to establish a clear history of no	ormal respiratory function and exacerbations
History of chest infections	A good indication of a child's respiratory health is to establish how they have been within the last 12 months. This includes noting frequency and duration of any chest infections, if they have required hospital admission, and their antibiotic history – including prophylactic and additional use (Hull, 2012).	Knowledge of the child's recent microbiology & intervention is an important part of planning their future management. This may include obtaining oral, nasal or tracheal secretions.
Home physiotherapy routine	The community therapist should be aware of the child's routine respiratory physiotherapy programme, who devised it and when the programme was last updated. (For example, use of airway clearance techniques and equipment used). It is important to establish who the child's local / specialist respiratory physiotherapy contact is. Understanding a child's compliance to treatment is important in supporting their care. This includes prophylactic management and escalation of care.	Children with Neuromuscular Weakness (NMW) who have an ineffective cough or a history of recurrent respiratory exacerbations, should have an individualised written management plan to deal with respiratory exacerbations (Hull, 2014). The specialist respiratory physiotherapist is responsible for assessing the child and establishing and monitoring the respiratory physiotherapy regime. They should review the child during planned and unplanned hospital admissions and the plan should be reviewed at least annually (Hull, 2012).
Drug History	<ul> <li>The local physiotherapist should have a working knowledge of the following adjuncts to their patients care, and know which medications their patient is using and how often</li> <li>Mucolytics–DNAse/ Saline (HTS)</li> <li>Bronchodilators</li> </ul>	The specialist physiotherapist should have a sound knowledge of respiratory medication, including the indications of use and possible side effects. They should establish effective liaison with the medical team and local respiratory physiotherapist, to ensure the optimal medical management is achieved.

	Nebulised antibiotics	
	Saline Nebulisers (0.9%)	
	Management of oral secretions	
	Steroids / Salbutamol	
	Anti-reflux medication	
	Hyocine / glycopyrrolate (Rutkowski et al., 2015)	
	Known allergies	
Social History	<ul> <li>The following factors can contribute to a child's respiratory health and management. They should be considered when taking a subjective history and implementing a management plan</li> <li>Housing situation</li> <li>Smoking</li> <li>Family – parents / guardians who will support the physiotherapy intervention</li> <li>Respite &amp; home package of care</li> <li>Education environment</li> </ul>	It is important to be aware of the child's home care provider and provide guidance on how training needs can be supported. The specialist physiotherapist may need to liaise with the patients CCG provider, when considering the provision of specialist equipment (e.g. mechanical insufflation- exsufflation). The physiotherapist should establish clear links to help prevent delayed equipment provision where required.
Respiratory Investigations	<ul> <li>The local physiotherapist should have an awareness of the following investigations and know where to seek support with interpreting the results</li> <li>Bronchoscopy</li> <li>Sleep Study</li> <li>CXR</li> <li>Cough Swab, cough plates and sputum samples</li> </ul>	At an advanced level the physiotherapist should be able to interpret the results of these investigations and adjust their treatment / management of the child accordingly. They should demonstrate sound clinical reasoning and implementation of appropriate intervention. They should be able to obtain sputum samples, cough swabs and cough plates if they are required.
Other considerations	<ul> <li>The local physiotherapist should have an awareness of the following and know where to seek support with concerns or regarding interpreting the results of investigations</li> <li>Feeding – type and timings of nasogastric (NG) / nasojejunal (NJ) / percutaneous endoscopic gastrostomy (PEG) or oral</li> <li>Possibility of gastro-oesophageal reflux (GORD)</li> <li>Communication</li> <li>Advanced Care Plan &amp; Escalation Care Plan</li> <li>Bone density</li> </ul>	It is important to understand and recognise the symptoms of inadequate ventilation and the appropriate escalation of care. Symptoms of hypoventilation, both daytime and nocturnal are commonly seen in children with NMW. These symptoms may include • Transient morning headaches (should resolve after 60 minutes) • Morning nausea • Daytime tiredness and lethargy
Objective		
Observations	The local physiotherapist should have a working knowledge of the basic respiratory	Specialist physiotherapists should have a deeper understanding of condition specific

	<ul> <li>assessments and be able to clinically reason whether an escalation of care is required.</li> <li>Knowledge of the normal age specific ranges including</li> <li>Respiratory Rate</li> <li>SpO2 (what is normal for that patient)</li> <li>Heart Rate</li> <li>Children with NMW may become</li> </ul>	<ul> <li>differences in distribution of respiratory muscle weakness (Hull, 2012).</li> <li>For example:</li> <li>Respiratory muscle function in SMA features. Children with SMA present with very weak intercostal muscles and a relatively stronger diaphragm. The diaphragm is the primary muscle used for breathing in children with SMA type I or II. For this reason lying flat helps reduce their work of breathing and</li> </ul>
	<ul> <li>tachycardic before changes in respiratory rate and work of breathing are observed (Schroth, 2009).</li> <li>It is important to be able to recognise the signs of increased work of breathing in a child, including:</li> <li>Nasal flaring</li> <li>Tracheal/ intercostal / subcostal recession</li> <li>Change to pattern of breathing- including paradoxical breathing (see-</li> </ul>	<ul> <li>reduce their work of breathing and caution should be taken when lying prone (especially SMA type I) (Schroth, 2009)</li> <li>Children with Dystropinopathies have fibrotic muscles and can develop stiff rib cages with restricted bucket handle movement</li> <li>Ullrich CMD – do not assess/ manage in supine as breathing is severely compromised in this position</li> <li>Recognising signs of respiratory fatigue is</li> </ul>
	saw breathing pattern, often seen in SMA type 1&2 and SMARD) • Fatigue	an important part of the assessment of a child with NMW. Often children with NMW will compensate when they become unwell, but can rapidly deteriorate if they are not adequately supported in their ventilation.
Auscultation	Chest wall palpation can be a useful assessment tool when the physiotherapist is not skilled in the use of stethoscope auscultation or one is not available. Feeling for chest wall crackles, audible crepitus and changes in chest wall expansion.	Advanced auscultation techniques are an essential adjunct in the respiratory assessment of a child with NMW. The physiotherapist should be skilled in interpretation of auscultation and adjust their treatment / management accordingly.
Secretion clearance	The effectiveness of a child's cough is an essential component of their respiratory health. Simply listening to their cough and noting if they achieve adequate inspiration and the cough is effective in clearing audible secretions can achieve this. If the child is productive of secretions, observe to see if they swallow or spit the secretions out or if they pool at the back of the throat or lower in the respiratory tract. Observations of the secretions produced are also important to note. Including the colour, amount & thickness. It is important to establish the child's normal routine. For example; does the child have suction at home? If so	Children should have a respiratory assessment at each hospital appointment. This includes performing lung function spirometry. The specialist physiotherapist should be able to interpret lung function results, (Bianchi & Baiardi, 2008) including normal values for cough peak flow (CPF) and forced vital capacity (FVC). They should know when to liaise with respiratory consultants and nurse specialists for further investigations if they fail to achieve adequate values. CO2 levels – capillary blood gases can be a useful part of your physiotherapy assessment and could be used if the therapist is appropriately trained and has available equipment.

	<ul> <li>Is it oral or nasopharyngeal suction?</li> <li>How often do they require suction?</li> <li>Who is trained in carrying out this procedure?</li> </ul>	
Spine /Posture	Ensure chest shape and spinal posture is observed without clothing to ensure clear examination. Observe the spinal posture when the child is lying down and when they are sitting up as gravity can have a pronounced effect. Note if the child has any spinal curvature (scoliosis, kyphosis, pescavus or excavatum). Assess the child's thoracic mobility, looking at both bucket handle and pump handle movements. The therapist should establish if the child has had any previous spinal surgery or there are plans to in the future. Awareness of the restrictions spinal jackets (TLSO) can have on a child's respiratory function.	Liaison / referral to spinal specialists should be carried out where appropriate. Established links with the local and specialist orthotics department to ensure effective provision of spinal jackets that do not compromise respiratory function.
Ventilation	Children with NMW may require a combination of day and/or night time ventilation to manage their respiratory fatigue. It is important to establish what is their normal 24-hour pattern and level of dependency on mechanical ventilation. It is likely that a child using night time ventilation only, may need it for periods during the day when unwell (Hull, 2012). Interface – the therapist should know the difference between different ventilation interfaces; including non-invasive ventilation (NIV) e.g. face mask and invasive ventilation, e.g. tracheostomy.	<ul> <li>The specialist NM physiotherapist should have an advanced understanding of the different types of ventilation support available. They should have an awareness of the appropriateness of current ventilation settings and when the settings on the ventilator were last reviewed.</li> <li>The specialist physiotherapist may play a key role in setting up and monitoring ventilation in the child with NMW.</li> <li>They should have a sound knowledge of the child's respiratory management plan and escalation of care if required.</li> <li>This should include</li> <li>Machine type</li> <li>Mode of Ventilation</li> <li>Interface – mask, nasal, full face</li> <li>Tracheostomy – cuffed/un-cuffed plan, type, speaking valves etc.</li> <li>Mask rotation</li> <li>Wet/ dry circuit</li> </ul>
Other equipment:	Awareness of other techniques the child / parent may be using Nebulisers Airway Clearance Techniques Suction	

Management	Foundation Module	Advanced Module
Airway clearance techniques	Effective airway clearance techniques are an essential component in the physiotherapy management of a child with NMW (Kravitz, 2009). An effective cough is essential for secretion clearance and protection against respiratory infections (Michelle Chatwin & Simonds, 2009).	The advanced therapist should have a sound knowledge base of the following adjuncts to physiotherapy. They should be able to identify the correct adjunct required for the child. They should evaluate and adapt the child's physiotherapy management accordingly.
	<ul> <li>The minimum intervention required to achieve effective secretion clearance, to reduce the risks associated with fatigue.</li> <li>It is important to have a working knowledge of the following respiratory manual techniques and when it would be appropriate to use them.</li> <li>These include</li> <li>Percussions</li> <li>Vibrations</li> <li>Manual assisted cough technique</li> <li>PEP devices</li> <li>Incentive spirometry</li> <li>Positioning</li> <li>Consideration of humidification / nebulisers</li> </ul>	<ul> <li>A sound knowledge of suitable adjuncts to therapy, that can help shorten the length of airway clearance sessions (M. Chatwin, 2009) although this may not help reduce patient fatigue.</li> <li>Adjuncts may include</li> <li>Lung Volume Recruitment (LVR) bag</li> <li>Air stacking technique, +/- one-way valve</li> <li>Mechanical Insufflation-Exsufflation (Cough-Assist device)</li> <li>Intermittent Positive Pressure Breathing (IPPB)</li> <li>Manual Hyperinflation</li> <li>Oscillatory devises i.e. High Frequency Chest Wall Oscillation (HFCWO)</li> <li>Tracheal pressure technique (if trained and used appropriately)</li> <li>IPAP on NIV can be adjusted to improve tidal volumes during treatment</li> <li>Suction – experience in the use of suction, including oral, oral-pharyngeal (OP) and nasal phalangeal (NP) and how to modify technique to ensure optimal secretion clearance</li> <li>Negative Pressure Ventilation (e.g. Hayek Jacket)</li> <li>(Michelle Chatwin &amp; Simonds, 2009) The specialist physiotherapist will have a role in educating others on the use of the above techniques, including patients, parents,</li> </ul>
Posture & Seating	Awareness of how the 24-hour postural management programme impacts on the respiratory system e.g. spinal brace and seating.	Close links and joint assessments where required to ensure suitable seating and postural support is provided. Pre & post-operative respiratory management is important in supporting timely discharge from hospital post spinal surgery.
Education & Training	Support the implementation of a child specific physiotherapy care plan, including basic manual techniques.	Identify, provide and implement written child specific care plans: including on-going training, evaluation and completing child

	Be able to identify when further specialist support is required. Educate the child, families and carers on the importance of the 24-hour postural management programme and trying to optimise chest wall and spinal alignment.	specific competencies. Review the training and competencies annually for each child and each trained parent / carer. This should be carried out sooner if the child's specific respiratory needs change.
Ventilation	Awareness of inadequate ventilation and when to escalate care appropriately. This would include signs of respiratory difficulty, marked air leaks, and information gathered in the subjective assessment.	Effectively ventilation support is an essential component in the management of a child with NMW. The advanced physiotherapist should know how to manage changes to the child's ventilation plan.
	Children with NMW may require oxygen if they have low saturations. However, it is important to be aware that oxygen alone will not adequately support the child with NMW, due to respiratory muscle fatigue.	The use of continuous positive airway pressure (CPAP) is not recommended in children with neuromuscular disorders, as it is unable to treat nocturnal hypoventilation due to respiratory muscle weakness (Rutkowski et al., 2015).
		This may include adjusting pressures, weaning time following an acute infection, mode of ventilation, mask rotation and the adjunct of speaking valves.
Other:	Awareness of how other components of the child's medical care can impact on their respiratory function. Liaison would usually be via the specialist centre	
	Gastrologist - reflux	
	Oral secretions	
	<ul> <li>Ear, Nose &amp; Throat specialist – tonsils / adenoids</li> </ul>	
	<ul> <li>Speech and Language Therapist – feeding</li> </ul>	
	Deteriorating respiratory function	
	Timing and side effects of other medication	
Palliative Care	A sound knowledge of the child's condition specific prognosis and disease progression.	The specialist physiotherapist will be involved in the discussions around ceilings of treatment.
	Appropriate links with specialist centre – especially the neuromuscular physiotherapy team.	
	Knowledge of the details of the child's advance care plan (ACP). If one is in place and which team is responsible for reviewing it.	

Right to Breathe - Muscular Dystrophy UK (2015)

# **Orthotics Management**

Why are orthotics important? Orthotics can play a key role in management of NM conditions however there are a large variety of types and indications. Both static and dynamic devices can be used and careful assessment and ongoing review is essential. This requires working closely with your orthotic provider. Advice for your local orthotist can be sought from national orthotics specialists. Experience suggests custom made paediatric devices are better tolerated, produce better results and in the long term more cost effective. Evidence on their benefit however is limited.

Role of orthotics:

- Providing a stable base for support and movement
- Establishing an efficient gait
- Reducing excessive energy used to move
- Reducing, or eliminating, hip and knee hyperextension
- Increasing a child's ability to function physically and mentally
- Strengthening weak muscles
- Controlling muscular imbalances
- Correcting poor posture
- Reducing the progression of deformity
- Preventing deformity and pain where possible

#### Orthotics and splints most frequently used in neuromuscular disorders

Abbreviation	Orthosis	Indications, treatment notes and precautions
СО	Cervical Orthosis (collar) – can be soft, semi-rigid or rigid	Travelling, head fatigue (for short periods) atlanto-axial instability.
		Commercially available types are suitable.
TLSO	Thoraco-lumbar-sacral orthosis	Rigid or semi-rigid orthoses can be the most effective in managing NM scoliosis.
		To assist in the management of scoliosis or spinal asymmetry.
	Jacket/brace	To support a weak trunk and improve upper limb function.
		To try and assist develop sitting balance.
		Can provide comfort/support in children with reduced trunk control.
		Important to put on correctly and prevent sores or pressure.
		Enough space under the arms.
		Should not gape.
		Should be comfortable enough to be worn for a minimum of 6 hours.
		Needs a 'tummy hole' cut for most children and ALL children with gastrostomy.
		Lycra may be indicated for occasional use in specific cases but not with marked trunk weakness.

LSO	Lumbar support orthosis	For ambulant children with back pain and discomfort
		or those with marked abdominal muscle weakness (e.g. Central Core Myopathy) but not for correction of hyperlordosis.
НКАГО	Hip ankle knee foot orthosis	Rarely used following improvement in standing frame design.
KAFO	Knee ankle foot orthosis	Used for standing and mobility. It can be used for non-ambulant children to achieve some independent walking. In most it is useful for exercise only but some of the stronger non-ambulant children can achieve functional ambulation. For children losing ambulation, it can prolong ambulation and reduce the progression of contractures and deformity. Not all KAFOs look the same; those for DMD having a different knee configuration to those for SMA. For further advice on prescribing and managing children in KAFOs you may need support from your local neuromuscular service. See Section 11a for specific advice.
RGO HGO	Reciprocating gait orthoses Hip guidance orthosis	Effective in a small group of children. Can be cumbersome and difficult to apply. Seek advice on application
КО	Knee Orthosis Gaiters/knee immobilisers	<ul> <li>Very useful as</li> <li>Assessment tool pre KAFO use</li> <li>In small children</li> <li>For improving stability in standing frames</li> <li>For maintaining knee ROM</li> <li>It is important when putting on gaiters that there is no metal bar directly over the front of the knee joint and patella.</li> </ul>
AFO	Ankle foot orthosis – any orthosis that has control over the foot and ankle	There are many different types of AFO as described below. They can be used as a dynamic splint to assist ambulation or as resting/positional splints. There is strong evidence that AFOs are not helpful in ambulant children with DMD, although many children with peripheral neuropathies may need AFOs for walking especially when they have foot drop. Night-time AFOs commonly used in DMD must be bespoke to ensure comfortable fit.

SMAFO	Supra-malleolar AFO	These are splints, which are primarily used to control the unstable foot, especially around the hind-foot. They allow dorsiflexion and plantarflexion and should be flexible in the mid-foot. Padding is often required around the malleoli.
DAFO	Dynamic AFO	Not used in neuromuscular disorders– these are similar to SMAFOs but have a tone reducing footboard.
GRAFO	Ground reaction AFO	Rarely used in this country – these are splints with anterior leg support.
SAFO	Silicon AFO	Expensive but very useful in children with foot-drop. They are however difficult for children with peripheral neuropathies to put them on unaided as they often have hand weakness.
Hinged AFO		Children will need passive dorsiflexion past neutral. They are bulky and parents can find difficulty with getting shoes.
Leaf spring AFO		Useful for children with foot drop or weakness but have passive dorsiflexion past neutral. The offset type is very well tolerated
FO's	Foot Orthotics	
	Insoles	There is little evidence to support the use of insoles in flat feet. However they have a role to play if feet are significantly pronated etc.
	Heel cups	Used to control inversion or eversion of the foot caused by instability at the hind-foot. The heelcup is much deeper around the heel and should be "posted" under the heel to correct the inversion or eversion.
CCD's	Control Contracture Devices	Without good access to serial casting some centre use these regularly. Ankle CCD's are particular helpful where loss of dorsiflexion is an issue. The hinges are expensive so best used in compliant families.
Tri-lock supports		These are elastic or neoprene ankle supports that have three straps to control the ankle and hind-foot. They are flexible and well tolerated when SMAFOs or AFOs are not found to be helpful.
Upper limb orthotics		
Less commonly used but may be effective in specific cases. Suggestions for specific joints are detailed below		

Shoulder	Shrug in FSHD can be useful especially if shoulders are painful. Bilateral shrugs exist. Lycra maybe an option to assist in FSHD as well and can correct asymmetry and provide support.
Elbow	Soft splints may be beneficial. Rarely used.
Wrist and hand	For very weak DMD young men tight long flinger flexors may warrant some intervention.
	Neoprene hand splints may be helpful in young children to aid wrist and hand function.

# Knee Ankle Foot Orthoses (KAFOs)

These can be used to promote standing and 'walking' in non-ambulant children with SMA 2, some congenital myopathies and arthrogryposis or in children that have been ambulant but have reached a stage where they are no longer walking independently. KAFOs are predominantly used in DMD but may be relevant to SMA 3, some other forms of limb girdle dystrophy and congenital myopathy.

KAFOs can enhance mobility, maintain joint range and soft tissue length promote exercise and activity and may have a role in reduce the progression of spinal asymmetry. It is not, however appropriate for all children and careful selection of children will help make the use of KAFOs successful.

The 'walking' in almost all cases is done with the knees locked into extension and can be slow and effortful, and useful only for exercise. Some children with stronger knee extensors are able to manage 'free knee' KAFOs. Some children are able to use the KAFOs effectively for walking for longer distances.

Apart from careful selection of patients, it is important to have an experienced orthotist and the feet must be able to achieve plantargrade. If the feet do not come to a neutral position, serial casting or surgery may be considered.

Percutaneous release of the Achilles tendon does not require the use of POP and standing and walking can be commenced on the first post-operative day if the KAFOs are pre-cast. **However** orthopaedic opinion is likely to vary considerably regarding this management. It is important to note that children who are able to weight bear for transfers, particularly with DMD, even though they cannot walk – will not be able to fully weight bear following surgery and the manual handling implications of this need to be considered.

#### General considerations for KAFOs in Duchenne Muscular Dystrophy

Although some centres will be used to working with orthotists who are familiar with making KAFOs, this will not always be the case. These general considerations may be helpful, but it may be beneficial to seek further advice.

Discuss with child and family in late ambulatory stage, i.e. when boy still able to walk across a room.

- Motivation of child and family and ability to comply with therapy for whatever reason.
- Family, child, school and support teachers, Orthotist and PTs (specialist and community) must all be on board if it is to be successful.
- Presence of LL contractures if more than -15 degrees at TA/ankle boy will struggle to gain independent standing balance and probably won't be able to walk in KAFOs. It is possible to wedge the KAFO heel a little to assist balance. Serial casting should be considered.
- TA release can also work well but it is recommended that you work closely with the orthopaedic surgeon to discuss post-op management and minimise post-op period in cast as good TA length will be achieved but knees will tighten (1-3 weeks may work as opposed to 'standard' orthopaedic 6 weeks in cast).
- If more than -5 degrees extension at knees, again, balance is more difficult though KAFOs can be off-set a little to accommodate this.
- Similarly hip flexion/abduction, contractures < 20 degrees make it difficult for child to balance, boy will hyperlordose to compensate and may experience low back pain.
- Space at home/school to enable therapeutic standing/mobilizing for exercise.

#### Guidance for casting KAFOs

- KAFOs need to be ischial/gluteal fold weight bearing and should be cast with the child in supine with leg supported in the air at 45 degrees to ensure cast goes right up to gluteal fold posteriorly.
- KAFOs can be cast prior to TA release and foot section set to 90 degrees at manufacture.
- It is not necessary to have a full foot / toe section, can trim to metatarsal heads to make it easier to fit shoes.
- KAFO should have lever-type release knee hinges for ease of release.

It is a time consuming process with several (usually 3) orthotic appointments – timely orthotic provision aiming for 6 weeks from cast to final fit. Cast (1 hour), mid fit (45 mins), final fit (45 mins) to allow time for orthotic adjustments and trial stand.

Unless your service is familiar with making KAFOs for children with NM disease it is helpful for the physio to attend all the orthotic appointments.

Once KAFOs are provided, it is important for the physiotherapist to plan physiotherapy rehabilitation sessions - ideally daily or 3 x week for first week in a gym with height adjustable plinth and parallel bars and these sessions may need repeating the next week to help child gain balance, etc.

#### Example KAFO Rehabilitation in DMD

Apply KAFOs in lying at least initially, child in loose shorts, apply pump type shoes / adjustable sandals over KAFO.

Lock knee joints in extension prior to stand.

Assist child from lying to sit to stand using height adjustable plinth and therapist in front and behind on plinth.

Balance work encouraging hip extension, 'stick your tummy out' so child 'sits' onto the KAFO – this may take a couple of sessions to achieve reliably and for the child to be able to maintain securely.

Once standing balance achieved, teach weight transference and hip hitch in standing and progress to walking as able.

Parallel bars work well for rehab at this point.

N.B. Take care on stand to sit, use height adjustable plinth, taking care to assist feet/legs forward a little at a time to slowly lower. Release knee locks only once weight supported on plinth.



Example Pathway from Oxford NM Service

#### Specialist Neuromuscular Services in UK

University Hospitals Bristol NHS Foundation Trust

University Hospitals Southampton NHS Foundation Trust

Oxford University Hospitals NHS Foundation Trust

Evelina London Children's Hospital

Great Ormond Street Hospital, London

The Royal Brompton and Harefield NHS Foundation Trust, London

Cambridge University Hospitals NHS Foundation Trust

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Borg Fatigue Scale



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