Scottish Muscle Network

Duchenne Muscular Dystrophy (DMD)

Scottish Physiotherapy management profile

NOTE
This guideline is not intended to be construed or to serve as a standard of care. Standards of care are determined based on all clinical data available for an individual case and are subject to change as scientific knowledge and technology advance and patterns of care evolve. Adherence to guideline recommendations will not ensure a successful outcome in every case, nor should they be construed as including all proper methods of care or excluding other acceptable methods of care aimed at the same results. The ultimate judgement must be made by the appropriate healthcare professional(s) responsible for clinical decisions regarding a particular clinical procedure or treatment plan. This judgement should only be arrived at following discussion of the options with the patient, covering the diagnostic and treatment choices available. It is advised, however, that significant departures from the national guideline or any local guidelines derived from it should be fully documented in the patient's case notes at the time the relevant decision is taken.

Contents

Introduction

Background: Duchenne Muscular Dystrophy (DMD) is often described in the literature as a relentlessly progressive muscle wasting disorder that affects mainly boys. It affects 3:10000 live male births across the world and without intervention, young men will rarely live beyond 19 years. There are some instances where females may show similar symptoms and disease progression but are affected to a lesser degree. These females are known as manifesting carriers.

In recent years medical advances have not only progressed in diagnosis but also in treatment and management. Life expectancy has improved, and many can expect to live into their 20's and 30's. In some countries such as Denmark and Holland, where management of this condition has been treated as a specialist area for many years, reaching the fifties is not unusual. In Scotland we also have a growing cohort of men with DMD in their forties and fifties.

Needs analysis: This booklet has been written to assist physiotherapists, to offer effective intervention at the different stages of Duchenne Muscular Dystrophy. Many physiotherapists have expressed a desire for a care pathway or indeed a clinical guideline, as keeping abreast of the changes in management and policy can be difficult. This is particularly relevant where a therapist is newly qualified or has a generic caseload with many different conditions to take account of. This booklet aims to highlight current practice in this field and direct the reader to useful resources where appropriate.

Methodology: This booklet has been written with the collaboration of experienced therapists from around Scotland and is intended for the Scottish physiotherapy service. It reflects current care offered and delivered in Scotland. An extensive literature review was undertaken and in the absence of a very strong or strong evidence base (relating to levels 1 and 2 of Sackett's levels of evidence), expert opinion has been sought using group consensus from experts in the field.

Aim

- this aim of this booklet is to assist healthcare professionals make clinical judgements and provide information to help with the decision-making process. Appendices one to six will offer assessment protocols and an explanation of current practice.
- this booklet is due to be reviewed in March 2027. Please ensure that you are using the most up to date booklet.

Conclusion: Your views on this booklet would be gratefully appreciated. If you have any queries regarding any of the material contained in this booklet, please contact: Marina Di Marco, Consultant Neuromuscular Physiotherapist, Department of Clinical Genetics, West of Scotland Genetic Services, Department of Laboratory Medicine, Queen Elizabeth University Hospital, Glasgow G51 4TF or e-mail: marina.dimarco@ggc.scot.nhs.uk

If you have a specific query and wish to talk to a physiotherapist in your area, please refer to the local services available on the SMN website (www.nn.nhs.scot/smn/).

STAGE I – PRE – DIAGNOSIS / EARLY STAGE								
Description	Assessment	Physiotherapy Intervention	Respiratory Intervention	Orthopaedic Intervention	Orthotic Management	Equipment Provision	Specialist Services	Education
Speech / Cognitive Delay Toe walking Waddling gait / gait abnormalities Developmental delay Tight gastrocnemius complex Enlarged calf circumference Gower's manoeuvre – (Boys will rise from the floor via a prone position using their hands to "walk" up their body). Muscle weakness (particularly proximal). Hypotonia Hypermobility	Physiotherapy assessment (Prior to a definitive diagnosis the physiotherapist should use a developmental assessment and may continue with this in the early stages if the child is unable to follow a more specialised assessment) It is recommended that the child is re-assessed every six months due to the variability in which change occurs throughout growth	Referral to Paediatric Neurologist and/or Community Paediatrician Always copy G.P into referrals. Encourage normal activities as able (Appendix 9) Avoid eccentric exercise (In conditions where there is instability of the muscle cell membrane, it is advisable to avoid strengthening exercises and eccentric muscle work as this can cause further damage the muscle cell). Introduce and advise on stretches for tight muscle groups and joints (Appendix 9)	Not usually required at this stage	Not usually required at this stage	For foot variations such as pes cavus or pes planus it may be useful to refer to an orthotist for insoles / inlays. Night splints may be appropriate if loss of range of dorsiflexion is noted (Appendix 9)	Not usually required at this stage	Refer to Care Advisor / Family Support Advocacy Officer with consent (Appendix 10) Parents may find a copy of the DMD care card (patient information document) helpful at this stage. This can be downloaded from the Scottish Muscle Network website (appendix 10) Family counselling may be required at any stage of this condition	Advice for P.E. teachers on fatigue issues and muscle weakness (Appendix 9) Classroom assistant may be required. Taxi to/from school may be helpful Referral to OT and Access Officer. Consider access in school Social Services can advise on benefits

STAGE II – YOUNG MOBILE								
Description	Assessment	Physiotherapy Intervention	Respiratory Intervention	Orthopaedic Intervention	Orthotic Management	Equipment Provision	Specialist Services	Education
Progressive muscle weakness Hypertrophy of muscle e.g. gastrocnemius / deltoid. Risk of contractures and tightness at end of range particularly ankles, hips, and wrists. Difficulty with jump / run / hop and frequent falls. Difficulty with stairs and rising from chairs. Increased lumbar lordosis and Trendelenburg style gait Muscle cramps and pain after activity. This can be particularly troublesome in bed at night	North Star Ambulatory Assessment (modified version for centres not signed up to data collection appendix 3) Undertake regular monitoring and assess every 6-12 months.	Liaise with the specialist neuromuscular physiotherapist in your area. Encourage activities e.g. swimming as able. Avoid muscle fatigue. Avoid strengthening /resisted exercise especially eccentric muscle work. Continuation of stretches appropriate to stage of progression (refer to MDC booklet on physiotherapy) Liaise with Lead Consultant re. Steroid therapy (appendix 1)	Encourage cardiovascular fitness. Baseline lung function tests. Ensure immunisations are up to date	Not usually required at this stage but do monitor ankles as the gastroc complex can become tight and if appropriate serial casting may be helpful.	Night orthoses for gastrocnemius complex when ankle range of movement is compromised. Consider ultraflex dynamic contracture control devices if child has good walking ability. These are generally worn 2hrs per day and may increase range of movement. Discuss further with specialist physiotherapist and / or Orthotist. Continuation with inlays / insoles if appropriate	Aim to get out of buggy by age five years and into a suitable lightweight manual wheelchair if required for longer distances as fatigue / falling may be problematic. Refer to MD UK wheelchair and seating guidelines. Specialised seating and equipment in school may be beneficial - refer to MD UK booklet on inclusive education	Provide the family with contacts for support / advice and information (appendix 10). Genetic counselling may be helpful for parents. If on steroid therapy, it is important that boys are referred for dietary advice. Pain management Cardiology, Bone Health, Dietetics Care Adviser Social services O.T. (Housing and adaptations) Clinical Psychologist Sleep Scotland if problems with sleeping (appendix 10) Introduce to appropriate sport activities, refer to sports co-ordinator. (appendix 10)	Raise awareness of future challenges re. access to curriculum and accessibility of classrooms and facilities. Additional support for learning strategies may be required – refer to Action Duchenne Learning and behaviour toolkit which can be downloaded from their web site (appendix 10)

STAGE III – GOING OFF FEET								
Description	Assessment	Physiotherapy Intervention	Respiratory Intervention	Orthopaedic Intervention	Orthotic Management	Equipment Provision	Specialist Services	Education
Poor core stability. Increased effort and time to rise from chair / floor Increasing frequency of falls Requires a wide base of support Difficulty standing with heels down Difficulty standing still for >3s Tires easily with physical activity Increased upper limb weakness Asymmetry noted in standing and sitting	North Star Ambulatory Assessment (NSAA) Appendix . Moving and handling risk assessment as per local policy (both at home and school)	Continuation of stretches / passive movements and exercises as abl. Hydrotherapy Standing frame as able (no consensus as to how long or how often – individual assessment as necessary. Be aware of children who complain of back pain especially with increased lumbar lordosis, as vertebral fractures are not uncommon in boys with DMD) Heel wedges may be appropriate to assist standing if dorsiflexion is compromised	Games to promote effective in – expiration Wind instruments Blowing bubbles Incentive spirometer to promote good expiratory technique Inspiratory muscle training using maximal resistance not recommended due to the fact it requires resistance training Monitor spirometry (appendix 5)	Spinal monitoring – discuss referral to Scottish National Paediatric Spine Deformity Service (Edinburgh) with Lead Consultant if asymmetry noted or young person starts to use wheelchair daily. Ensure regular spinal monitoring as scoliosis can develop within a few weeks Continue to monitor lower limb contractures	Walking AFO's not usually recommended as this can cause a deterioration in balance Continue with night AFO's if child is compliant and AFO's recommended when sitting in wheelchair for long periods of time. If wearing daytime AFO's we tend to stop using the nighttime AFO's (Appendix 9)	Manual wheelchair with supportive cushion and backrest. Referral for powered tilt in space wheelchair. It may be appropriate to consider a powered chair with advanced functions including a lap tray (see MDC booklet on wheelchairs and seating). Alternative funding may be required. (appendix 10) Familiarisation with hoisting techniques Profile bed (with lateral tilt if possible, to facilitate more changes in position Bathing Equipment	Motability (adapted vehicle should be considered at this stage). Ensure young person always uses a headrest for transport (refer to MD UK booklet on wheelchair provision and special seating) Introduction of cardiac monitoring recommended at this stage	Moving and handling Postural support within the classroom and increased assistance may be required for toileting Assess for appropriate table height

STAGE IV – EARLY POWER CHAIR USER								
Description	Assessment	Physiotherapy Intervention	Respiratory Intervention	Orthopaedic Intervention	Orthotic Management	Equipment Provision	Specialist Services	Education
Increased risk of scoliosis No static standing balance Sitting balance compromised Some ability to assist in standing transfers Some ability to self-propel manual wheelchair but be aware of fatigue issues and repetitive strain on shoulder joints. Difficulty raising arms above head	NSAA (appendix 3) EK2 Scale (appendix 4)	Continue monitoring upper limb function (PUL 2.0 assessment) 24 hour postural management (appendix 9) Active assisted exercise / passive movements and stretches Particular attention to wheelchair and moving and handling both pre and post spinal fusion (Appendix 8) Pre and post-surgical intervention if orthopaedic surgery undertaken.	Chest clearance techniques (appendix 7) Introduce lung volume recruitment techniques i.e. breath stacking and ambu bag techniques Assisted cough Teach parents / carers chest clearing techniques (Better introduced when patient does not have a chest infection) Regular monitoring of pulmonary function tests. Early use of antibiotics if respiratory infection suspected	Regular review Tenotomies may improve sitting posture if ankle deformity is problematic Surgical gastroc complex lengthening procedures are generally percutaneous and, in most cases, require a general anaesthetic. This is usually followed by two weeks in plaster See ankle management leaflet on the SMN website) Bilateral ankle foot orthoses after removal of plasters are required. Spinal surgery may be appropriate at this stage. For children on steroid therapy scoliosis surgery is less common	AFO's recommended when sitting in wheelchair Consideration to spinal orthoses for function and comfort particularly if person is not a suitable candidate for spinal surgery (if scoliosis is pronounced or person is overweight then surgery may not be appropriate)	Introduction to postural management for sleep if young person is unable to change position in bed Consider 4 –section lateral tilt profiling bed) Standing frame should be discontinued if young person complains of pain and / or contractures at hip / knees are problematic Hoist (assess for appropriate sling with head support, if necessary, in collaboration with occupational therapist) Height adjustable tables may be helpful Consideration of mobile arm supports	Dietician Speech and language therapy assessment for chewing and swallowing may be indicated Be aware of changes in eating pattern and monitor weight. Some children may feel that eating takes too long and fatigue issues emerge. Feeding ability may be compromised and child does not want to ask for help at school Introduction of care agency to assist with areas of personal care Respite services / befrienders	Secondary school access assessment Educational psychologist / Family counselling may be useful at any stage Coordinated support plan if appropriate

STAGE V – LONG TERM POWERED WHEELCHAIR USER								
Description	Assessment	Physiotherapy Intervention	Respiratory Intervention	Orthopaedic Intervention	Orthotic Management	Equipment Provision	Specialist Services	Education
Unable to assist with transfers Deteriorating head control Powered wheelchair for independent mobility Increased risk of scoliosis if no spinal fusion Less effective cough Poor circulation to extremities Oedematous ankles Gastrostomy may be indicated Fear of falling asleep	E.K2. Scale Epworth sleepiness scale (appendix 6) Quality of Life Hand dynamometry Upper limb Assessment (PUL)	Passive movements and stretches with particular attention to hand function 24 hr Postural management Encourage regular changes in position perhaps with an advanced powered wheelchair (lie to sit functions) Current recommendations are to tilt for at least ten minutes every hour - appropriate to rest neck muscles Monitor pressure areas and skin folds particularly if not had a spinal fusion Posterior aspect of ankle joints can also be problematic with regard to pressure	Non-invasive ventilation Teach lung volume recruitment techniques (appendix 7) Sleep assessment may be required to monitor nocturnal blood gases (of particular importance during REM sleep) Pulse oximetry	Monitoring of hip joints – some older boys complain of hip pain, particularly those with a scoliosis – be aware of hip subluxation	As previous stage Consideration of wrist splints for resting	Wheelchair provision with adequate degree of tilt (refer to MDC booklet on wheelchairs and seating) Ensure adequate head support (particularly during feeding and travelling) Environmental controls – consider access to games controllers and computers Consider mobile arm supports	Pain Team Palliative team if not already known to patient Transition support worker if available Continence care adviser for those who find toileting difficult Uribags (discreet bottles) are available on prescription from Fittleworth Tel:0800 783 7148) Tissue viability nurse if pressure care is problematic Young disabled school leaver's teams or physical disability teams for onward referral Consider Independent Living via Social Services	Access issues for further education Including fire evacuation strategies Benefits may change when young person is aged 16 years or over Refer to welfare rights officer / social services for up-to-date information on benefits.

STAGE VI – Palliative Phase								
Description	Assessment	Physiotherapy Intervention	Respiratory Intervention	Orthopaedic Intervention	Orthotic Management	Equipment Provision	Specialist Services	Education
Totally dependent for all care Functional muscle power may be limited to finger movements. Difficulty chewing and swallowing / loss of appetite and weight loss Frequent chest infections Multiple contractures Day time ventilatory assistance Difficulty sitting upright in chair – spending long periods of time in bed Pain	Pain and comfort Respiratory effort Circulation Quality of life	Passive movements and regular positioning to relieve pain and pressure Complementary therapies	Cough augmentation and chest clearing techniques if tolerated Ventilation used more frequently at this stage Regular auscultation and monitoring	Not usually appropriate	Generally not tolerated at this stage Positioning with pillows or sleep system for comfort Occasionally a collar is required	Specialised pressure reducing mattress may be required Lateral tilt bed to aid pressure redistribution and facilitate ease of handling.	Family counselling Dietician (augmented feeding) District nurse Tissue viability nurse Respite services Hospice Home support team	Home tuition if appropriate

[illegible] - Steroid Therapy and DMD

In recent years, the prescription of steroid therapy has become the gold standard in the treatment and management of ambulant boys with DMD. The most prescribed steroids are prednisolone and deflazacort. The steroid prescribed will depend upon the Consultant and the dosage prescribed is directly related to the young person's weight. Once steroid therapy has commenced the young person will be weighed regularly and specific health checks such as Vitamin D levels, urine analysis, blood pressure and visual acuity will be carried out to ensure the maintenance of an optimum dosage and to monitor for any side–effects.

Steroid therapy is most effective if commenced when the child has reached their developmental potential usually between the ages of 3 and 6 years. Due to the immuno-suppressant effect of steroids, the child should be up to date with all of their injections.

The exact way the steroids work is not yet fully understood however, it is felt that steroids have an effect on the inflammatory process which occurs in the muscles in DMD. Children who are treated with steroids have been shown to have an increase in muscle strength and functional ability. Some children can hop, run, and ride bikes, tasks they would have been unable to perform without steroid therapy. The frequency of the steroid regime does vary and at the moment there is no clear consensus as to which regime should be used although trials to ascertain steroid prescription are underway (FOR-DMD). Daily steroids have been shown to produce a more successful increase in muscle strength however, side effects are more likely. Young people who have pulsed steroid therapy i.e. 10 days on and 10 days off are felt to have time to recover on the days where steroids are not taken and, in these cases, side effects may be less problematic.

Children on steroids generally maintain ambulation longer than those who do not take steroids. In some cases, walking can be carried on into the mid-teens. These children are also at less risk of scoliosis and some young people also display maintenance of their respiratory function rather than the gradual deterioration of lung function and breathing ability synonymous with the natural progression of this condition.

Side effects commonly associated with prolonged use of steroids are reduced bone mineral density with an increased risk of fractures (particularly vertebral fractures), weight gain and behavioural problems. Ongoing monitoring for children on steroid therapy is important and approximately 1/3rd of children are not suitable for this type of treatment. Glasgow is currently hosting research into bone health in steroid treated boys with DMD and for more information please visit the Scottish Muscle Network website.

Steroids are generally stopped when the young person becomes wheelchair dependent however continuation with steroids is now being undertaken in some centres for respiratory benefits. At the moment, it is not recommended that steroids are commenced when the young person is wheelchair dependent as coming off them can be associated with sudden respiratory failure. Do not stop steroids suddenly as this can be problematic for the patient, weaning is recommended as advised by the lead Medical Consultant www.enmc.org/

More information and leaflets on vertebral fracture, adrenal suppression and puberty can be found on the Scottish Muscle Network website.

Appendix Two - Bladder Function and Continence Care

Whilst bladder function is not felt to be directly affected by the deterioration in muscle cells associated with dystrophinopathies, boys with DMD often experience periods where bladder control is compromised.

In young people with DMD who are starting to go off their feet, standing balance can be affected and in fact, standing still for more than a second or two can be challenging. This can cause boys to “have an accident” when using a urinal / toilet in the standing position. If this happens often, sitting down is possibly a better option in these circumstances and it is important to make sure that the toilet at school / home has handrails to assist the young person back into the standing position. The optimal toilet height and position of handrails can be assessed by the Occupational Therapist (OT).

If the young person has been used to toileting independently, loss of hand strength can mean going to the toilet is more difficult. Undoing buttons and zips takes time and for many children, going to the toilet is put off until the last minute. Suddenly there is urgency, and this can make undoing clothes more difficult when time is of the essence. In these circumstances, it is important that school staff can recognise when this is happening and perhaps gently remind the young person to go to the toilet prior to interval or lunchtime. If the young person is wetting repeatedly, he may feel embarrassed, and this issue requires a degree of sensitivity.

In some instances, school staff may not be fully aware of the reasons behind the wetting instances and occasionally it is suggested that the young person wears a pad to school. This is not recommended as wearing pads encourages the young person to “let go” whenever they wish, and this quickly infiltrates to evening and weekend behaviour becoming the norm for that child/young person. Trying to re-establish a toileting pattern when this has commenced can be more difficult and prevention is a better option.

If the child/young person does begin to wet more frequently, a toileting programme can be commenced. It is normal to go to the toilet every two hours and it is important to give the child/young person time to fully empty their bladder. If help is required with clothing, there are various options readily available to make things a bit easier for the young person to remain independent when undoing clothing and an OT can advise.

Young people can maintain independent toileting when in a wheelchair with the use of a bottle and if assisted, they can shuffle forwards and use the bottle without difficulty, but a carer should be on hand to take the bottle off the young person if required. When full, the bottle may be too heavy for the young person to hold and empty. Maintaining good continence care can also be problematic if the young person is embarrassed due to the fact he requires assistance to position the bottle. Often, due to the sitting position and pubertal delay, boys have a small penis and locating the penis in the bottle can be more difficult. Some boys have found a bottle with an extended neck or a female bottle with the wider aperture is often easier to use. An Occupational Therapist or Continence Care Adviser can usually advise but often it comes down to trial and error. When trying to establish regular toileting, some

families have found helpful advice via their local Enuresis Clinic and if the young person is over the age of five years, a referral can be made via the healthcare professional.

Some literature describes young men with DMD as having a neurogenic bladder. Neurogenic bladder dysfunction and symptoms vary depending upon the cause. It is thought in DMD that young people become used to holding onto urine as they prefer to wait until they get home to go to the toilet. Often this is because their personal assistant at school / college may not be confident in helping them toilet efficiently and for some, toileting takes too much time out of their busy day. Other young people report that if they are moved within the chair, it takes a long time to re-position them and their caregivers at home can do this quickly and with much less fuss. Holding on to the urine causes the bladder to become overstretched. If the bladder is regularly overstretched, the stretch receptors in the bladder wall take longer to be stimulated. When bladder distension occurs regularly, the bladder becomes large and can accommodate much higher volumes of urine. When the stretch receptors are finally stimulated, the bladder will be overly full and often urgency is experienced by the young person who may not be able to wait long enough to be positioned for a bottle.

In cases like this, working with the young person and their family to establish a good routine is essential. There are various options on the market such as a sheath, a condom like glove that fits over the penis snugly enabling the young person to urinate more frequently. The urine drains into a catheter bag that can be attached to the leg hidden under the trousers. For some, a short-term indwelling catheter may be used to prevent the bladder becoming distended. This in turn encourages more frequent emptying of the bladder and a return to normal sensitivity of bladder filling. Caregivers and personal assistants may require more in-depth training in moving and handling and this is often best approached in collaboration with the family who understand the moving and handling needs of the young person best.

Some young people will refrain from fluids through the day to enable them to get through the day without going to the toilet. Dehydration leads to tiredness and fatigue and can cause issues with kidney stones. If the urine becomes concentrated, crystal formation of minerals (and chemicals) within the urine is often experienced and these stones vary in size. Establishing a good toileting regime is better than risking kidney stones, which are often painful when passed.

Myoglobinuria
Myoglobin is a protein found in heart and skeletal muscles. When a muscle is exercised, it requires oxygen. With continued activity, more oxygen is required, and myoglobin provides extra oxygen for the muscle to maintain this level of activity for a longer period. When muscle is damaged, the myoglobin is released into the bloodstream and excreted via the kidneys into the urine. In large amounts, myoglobin can cause damage to the kidneys and episodes of myoglobinuria should be investigated. The child will often tell parents that his urine is red or looks like Coca-Cola.

Myoglobinuria is a recognised complication of steroid treated boys with DMD. It is felt that boys who are participating in steroid therapy may be more active than those who are not on a steroid prescription. It is thought that the increased activity places the dystrophin-deficient muscles under greater mechanical stress, predisposing to further muscle fibre damage and consequent myoglobinuria.

Appendix Three - North Star Assessment Protocol

This protocol has been developed via a lengthy process of review and consensus by the Physiotherapy Assessment and Evaluation Group of the North Star Clinical Network for Paediatric Neuromuscular Disease Management (NSCN). One of the key aims of this national, multidisciplinary project has been to standardise assessment techniques for ambulant children with DMD. Twenty specialist paediatric neuromuscular centres from across the UK have participated. The charity Muscular Dystrophy UK has substantially funded and supported the activity of the NSCN.

For a copy of the NSAA and its manual please go to:
Home - Pod NMD (pod-nmd.org)

This is a web site hosted by TREAT-NMD and contains lots if education, information and assessments for children and adults with muscle conditions.

FVC	Absolute value	%age pred for height	Comments
Test 1			
Test 2			
Test3			

Joint range	R	L	Comments
Elbow extension			
Hip extension			
Knee extension			
Ankle dorsiflexion			
ITB tightness			
Wrist and Fingers			
Supination			

Management of joint range (link to assessment of joint range)

Stretches	summary of advice given & to who, include frequency and reps
Orthotics	e.g. FO's**,** AFO's, KAFO's
Usage	e.g. night splints + estimated wear time and compliance
General advice given	e.g. hydro/swimming
Surgery (If any – specify)	
Comments	e.g. any problems with any of the above, include compliance where possible

Equipment

Mobility Equipment	
Wheelchair/buggy	Y/N
Model	
Wheelchair cushion	Y/N
Model	
Wheelchair services	
Contact	
Comments	

Gait analysis: (brief description, include any aids used, frequency of falls)

General activity levels:
Include 'extra-curricular' activities such as swimming, cycling horse-riding

Parent/carer perception of general health & well being
Improvement/deterioration/no change
Comments:

Patient perception of general health & well-being:
Improvement/deterioration/no change
Comments:

Spinal Posture

Sitting Draw, describe	**Standing** Draw, describe
Cobb angle (if known)	**Rate of progression** (if known)
Correctable Y/N	
Comments (Include any action, spinal jacket – type and wear time, surgery)	

Appendix Four: EK2 Scale (wheelchair dependent)

For the EK2 Scale and manual:
EK2_engelsk.pdf (rcfm.dk)

Stage	Aim	Objective	Outcome measure
Young ambulant (Diagnosis to approximately 10 years)	To maintain and promote good inspiratory capacity	Encourage wind instruments, blowing bubbles, singing etc	Regular peak flow to monitor respiratory muscle fitness. Peak cough flow is a good way of measuring expiratory muscle function.
	To maintain and promote respiratory fitness	Encourage young people to maintain an active lifestyle within their capabilities	
	To teach an awareness of breathing control	An incentive spirometer can be used to teach breathing control awareness.	
Wheelchair dependent (Approx 11-16 years)	To maintain chest compliance	Increase persons awareness of improving lung volume recruitment such as breath stacking/ and /or glosopharyngeal breathing. Teach parents /carers how to increase inspiratory measures using an ambu bag	Regular assisted inspirations (aim for daily and this can be increased when young person has a chest infection)) Referral to Respiratory Consultant for formal monitoring
	To prevent chest infections	People with DMD are encouraged to have their flu and pneumococcal jabs where appropriate particularly if VC <50% of predicted value	Referral to Orthopaedic Consultant for spinal assessment

Stage	Aim	Objective	Outcome measure
	To introduce effective measures for clearing the chest during infections	Respiratory assessment by physiotherapist to assess best method of clearing the chest If peak cough flow is less than 280l/min teach cough augmentation techniques such as manual splinting of the diaphragm and thoracic holds. This is only an indication, and some people require secretion mobilisation techniques with a PCF>280l/min as it can be greatly reduced during episodes of fatigue or infection Use of cough assist machine (mechanical in-exsufflator) if appropriate Early delivery of antibiotics during a chest infection	Many patients with respiratory muscle weakness benefit from a prescription for antibiotics to be kept at home. Advise them to have a low threshold for commencing antibiotics when they develop a respiratory tract infection
	To prepare for non-invasive ventilation (NIV)	Practice regular lung volume recruitment techniques with use of ambu bag	
	To monitor scoliosis	Refer to Royal Hospital for Children and Young People, Edinburgh for monitoring of scoliosis	

Stage	Aim	Objective	Outcome measure
Wheelchair Dependent on Non-Invasive Ventilation (NIV) Indications for ventilation Poor FVC Frequent chest infections Poor Sleep Hygiene Early morning well being Poor Appetite Weight loss (see Epworth Sleepiness scale)	To prevent/manage chest infections	If cough is ineffective at clearing secretions, further techniques to increase inspiration might be helpful such as an ambu bag or cough assist machine. The cough assist machine can be used in conjunction with manually assisted cough techniques. It can also be used to clear secretions in the absence of a chest infection and some patients like to use it prophylactically If aspiration or weight loss is evident, refer to a speech and language therapist and / or dietician	Liaise with respiratory physiotherapist and breathing support /nursing staff.
	To maintain chest compliance	As above	
	To manage transition process	Liaise with adult physiotherapy team	

Symptoms of respiratory failure	**Symptoms of nocturnal hypoventilation**	**Symptoms of Bulbar Dysfunction**	**Signs of respiratory muscle weakness**
Shortness of breath Orthopnoea (Dyspnoea when lying flat) Recurrent chest infections Lethargy Weight loss	Frequent nocturnal wakening Excessive daytime sleepiness Reduced concentration Un-refreshing sleep Fatigue Early morning headache	Nasal regurgitation Choking or coughing episodes at mealtimes Weak cough Recurrent chest infections	Weak sniff or cough Abdominal paradox Recruitment of accessory muscles at rest Increased rate of respiration Reduced chest expansion Cyanosis Papilloedema (severe hypoventilation)

Appendix Six - Epworth sleepiness scale

SITUATION	CHANCE OF DOZING
Sitting and reading	_____
Watching TV	_____
Sitting inactive in a public place (e.g. a theatre or a meeting)	_____
As a passenger in a car for an hour without a break	_____
Lying down to rest in the afternoon when circumstances permit	_____
Sitting and talking to someone	_____
Sitting quietly after a lunch without alcohol	_____
In a car, while stopped for a few minutes in traffic	_____

0 = no chance of dozing
1 = slight chance of dozing
2 = moderate chance of dozing
3 = high chance of dozing

If score is between 6 and 8 there are some concerns with sleep hygiene. Scores of 9 or above are considered significant and patient should be referred to his respiratory specialist.

In order to successfully expel secretions from the lungs the patient requires a good expiratory volume and a forced expiration. In DMD the respiratory muscles and diaphragm are severely compromised by weakness and many patients are unable to successfully increase lung volume and forcefully expire air.
In order to maximise respiratory volume, the patient can use a number of techniques to increase lung volume recruitment:

- Glossopharyngeal breathing
- Breath stacking with assistance of
  - an ambu bag
  - mechanical in-exsufflator (cough assist)
  - ventilator

Glossopharyngeal breathing
This requires good bulbar control, and some patients can develop this technique naturally. It involves 'gulping' air into the lungs and breath stacking. Patients who require daytime ventilation can use this technique to come off their ventilator for bathing and showering etc whereas others use it to add volume to their voice.

Ambu Bag
An ambu bag fitted with a one-way valve is recommended (available from Intersurgical, 0118 9656376 or www.intersurgical.com). These ambu bags are also known as lung volume recruitment ambu bags. (Always mark ambu bag with a notice "Not to be used for resuscitation).

Current recommendations are that it should be used up to four times per day for those who have an ineffective cough. Aim to use it first thing in the morning to clear secretions that may have gathered overnight and again last thing at night. It is also recommended that the ambu bag is used before meals however if bulbar control is poor and patient is likely to aspirate, it may be beneficial to use after eating. Refer to the supplier for infection control / use of filters.

It may also be helpful to use it more often if patient requires to cough or if there is a chest infection, however, avoid overuse as patient can become fatigued. Early use of antibiotics is essential if a chest infection is suspected.

Technique

- clear explanation should be given to the patient
- best done in sitting but can be done in lying or a semi-recumbent position. Head should be supported against a headrest and if in the wheelchair, ensure that the brakes are on and chair is positioned against a wall particularly during assisted cough
- position nose clip if tolerated
- ask patient to take a deep breath in and hold
- immediately place mouthpiece into mouth and ensure there is a good seal

- gently squeeze bag and instruct patient to take a second deep breath
- repeat again if possible. In this way the patient is stacking breath on breath until lungs are full. The patient may adapt more readily if LVR is initiated at the end of normal exhalation. As chest expands, the patient will feel a stretch in his chest
- Exhale or cough as desired.

Encourage the patient to take as much air as possible and maintain eye contact throughout the treatment. Watch for initiation of inspiration so the helper can time the squeeze on the ambu bag. If air leakage is a problem, change interface to a mask if preferred. This technique should not induce dizziness or chest discomfort – in the event of these symptoms occurring, discontinue treatment.

Mechanical in-exsufflator (Cough assist)
As with the ambu bag, the cough assist machine will deliver a positive pressure inhalation but will deliver it throughout the inspiratory cycle. This can be administered via a mask or mouthpiece. The machine very quickly changes to negative pressure and forces expiration. This expiration can be done in conjunction with manually splinting the diaphragm for a more effective cough.
These machines are becoming more popular as assistance from one helper is required but two helpers may be necessary for the ambu bag technique and assisted cough. Also the additional negative pressure during expiration can be enough to clear secretions without the need for a forced assisted cough. Many patients report that the cough assist helps with secretion clearance without the same degree of muscle fatigue as other techniques.
The cough assist machine also has the capacity for automatic or manual timing of the inspiratory, expiratory and pause phase.

Assisted cough
When undertaking an assisted cough, some patients find it more comfortable to dissipate the force required during the upward thrust by using a towel or small cushion across the abdomen.

Appendix Eight - Spinal Fusion

Around 90% of boys with DMD will develop a scoliosis if not on steroid therapy. Even with steroid therapy, scoliosis may still develop but at a later stage due to prolonged ambulation and standing ability but data for this group is sparse due to the lack of historical perspective in this "new" older population. Scoliosis monitoring should commence before the loss of ambulation, to ensure surgical intervention can be offered at the appropriate stage. The Spinal Consultants and Respiratory team will determine if the young person is a suitable candidate for this procedure. The orthopaedic consultant will monitor the Cobb angle and surgery is best undertaken when this angle is between 20^0 and 40^0 although in some instances, the spinal team may operate well before the scoliosis reaches this level. Spinal surgery is a complicated procedure, and families can feel very stressed around this time. Prior to spinal surgery, good preparation is essential. If the young person does not have a tilt in space wheelchair, it is highly recommended that this is in place before hospital admission. Planning is essential as delays in the provision of equipment are not uncommon.

After spinal fusion, the young person may be taller and therefore lateral supports / back support contours in their seating and wheelchair symptoms may not be in the correct position. It is recommended that the young person has a complete re-assessment of their wheelchair / seating provision and if this can be pre-arranged for 2/3 weeks after surgery, then this will avoid delays when waiting for an appointment with local seating services.

The young person may have difficulty with head control as he will be in a different position therefore adequate head support is essential. For the first few weeks the young person may not feel comfortable sitting upright and the tilt and/or recline function in the wheelchair / shower chair will be beneficial. Some young people lose the ability to feed themselves as their 'trick' movements are more difficult and the hand has to lift the food to the mouth through a greater distance against gravity. Loss of this ability can be distressing for the family as well as frustrating for the young person. Increasing tray and table heights can alleviate this situation, however, in some instances the young person may not resume independent feeding.

Hoisting both at school and at home is essential and manual lifts are not recommended particularly in the first year after spinal fusion. Bone grafts can take between nine and twelve months to heal and during this time particular attention should be given to moving, handling and postural management. The hoist sling should support the head and neck and slings with strengthening in the back are generally best although some patients are more comfortable without the strengthening due to their spine and head alignment. It is best to avoid excessive hip flexion beyond 90^0 particularly in the first nine months and the spinal consultant will be able to further advise on this. In some cases, the pelvis is also fused to the spine and excessive hip flexion can increase the strain in the lower back through flexion of the lumbar spine. Each consultant will manage the spine differently and it is important to check the post-operative care with the consultant in charge.

Rotation at the spine should be avoided as this can place undue stress on the healing spine. ‘Log’ rolls are recommended, and a symmetrical sleep posture is desirable. Appropriate sleep support in terms of pillows or pressure redistributing mattresses are generally necessary once the young person is no longer able to turn in bed independently and this may have to be reviewed following surgery. Changes to sleep posture may be best undertaken in incremental stages and consideration to respiratory function and how this may be affected by positioning is important especially in those with very weak respiratory muscles who require non-invasive ventilation. Care and attention to hip joints is also recommended through 24-hour postural management.

Physiotherapy such as passive movements to hip joints through their full range should be discussed with the spinal surgery team as should hydrotherapy and sporting activities as some wheelchair activities may have to be postponed until the spine is fully healed. In most circumstances, activities can be resumed after 6 months with supervision, as falls out of the wheelchair must be avoided. In some circumstances, healing may take longer so communicate with the spinal surgeon to ensure safe re-introduction of activity If there are any queries or concerns with specific activities then please discuss with the young person’s spinal consultant. When it is known that a young person has been accepted for spinal surgery, local physiotherapists are advised to contact the Clinical Specialist in Spinal Surgery, Address: Royal Hospital for Children and Young People, 50 Little France Crescent, Edinburgh Bio Quarter, Edinburgh, EH16 4TJ. Switchboard Telephone: 0131 536 1000

Exercise

Muscle weakness is defined as the reduction in strength of one or more muscles and can be subjective or objective in nature. It can also be exhibited as fatigue, exhaustion, or debility Muscle Weakness and Fatigue: Causes and Treatment (patient.info) The Physiotherapist is responsible for providing advice on exercise and activity in the person with Duchenne Muscular Dystrophy (DMD). This should be tailored to the person and stage of their condition. People with DMD should be encouraged to be active in normal, age-appropriate activities and sport within the limit of fatigue. There is a paucity of evidence regarding the effect of exercise in persons with DMD. The numerous benefits of exercise, including social and psychological, are well recognised by specialist clinicians (Eagle, 2002).

Intervention	Recommendations
Aerobic activity	Normal, age-appropriate activity, including play in children, should be encouraged. Also recommend non-weight bearing activities such as swimming, cycling, horse riding. Research into aerobic and anaerobic exercise in children with DMD has not been shown to influence functional ability.
Eccentric exercise	Prescription of eccentric activities are not recommended because of the evidence reported in animal studies. Studies report eccentric exercise leads to increased muscle damage (Eagle, 2002). Examples of eccentric activity to avoid include heavy resisted activities such as weights, unnecessary walking downstairs and running downhill and trampolining/bouncy castles.
School based exercise	Children with DMD should be encouraged to participate in Physical Education lessons and other school sports within limits of their fatigue. Sports and classroom teachers should receive guidance from the child's local Physiotherapist.
Disabled Sports Association	Children and their families may be referred to a local organisation for participation in appropriate sports. This may include horse riding / wheelchair sports / swimming / disabled scouts etc.
Hydrotherapy	Recognised as beneficial for people with DMD and recommend if available. In the absence of the service, families should be encouraged to pursue warm water activities. Due to a paucity of evidence, this has been extrapolated from other neuromuscular disorders.

Parents	It is the responsibility of the Physiotherapist to provide guidance, education, and recommendations regarding appropriate levels of activity. For example, awareness of the dangers of fatigue or periods of inactivity should be discussed.
Over exercise	Therapists should be aware of the risk of over exercise which may result in myoglobinuria. This is particularly relevant in children/young people who are on steroids.
Steroids	The introduction of steroids as standard treatment may be a factor in increased frequency of myoglobinuria, as children are more active. DMD boys are known to be osteoporotic, and this may increase with the long-term use of steroids. Long bone fractures are particularly common when ambulation is becoming more difficult, and this may lead to complete loss of ambulation. Therapists should be aware of the possibility of fractures particularly prior to the loss of ambulation when they are more unstable.

Stretching

For many boys with DMD, contractures are a part of the condition. The disease process causes muscle weakness. The link between muscle imbalance and contractures has not been established however, there is an association between them and long-term positioning and posture. The development of joint contractures and deformity can also be associated with pain. Promotion and maintenance of muscle length and joint range of motion (ROM) is recognised as essential. Stretching is defined as a prolonged passive movement applied to the muscle at the end of range.

Intervention	**Recommendations**
Early implementation of a tailored stretching programme where tightness at end of range is noted.	Assess all four limbs for joint tightness. Early implementation of a stretching programme is an integral part of the physiotherapy management of DMD particularly the gastrocnemius complex. It is recommended that stretches commence when there is any loss of dorsiflexion. N.B. Upper limb contractures can occur in ambulant children i.e. tightness in long finger flexors or supinators. Self-stretches, passive and/or active assisted should be tailored to the individual and be introduced once there is loss of normal range across a joint. The MD UK Physiotherapy guidelines booklet is a useful resource as it has pictures of common stretches.
	The first muscle groups at risk of contracture are recognised as gastroc-soleus complex, hip flexors and iliotibial band (Eagle, 2002). Utilise 24-hour positioning and appropriate equipment in addition to stretches. Standing wedges may be useful to stretch the gastroc/soleus complex.

	Other muscle groups of the lower limb are at risk of developing contractures at a later stage. Monitor muscle length in all groups of the lower limb and implement additional stretches as appropriate.
	Upper limbs are at risk of development of contractures. Generally, this occurs in the non-ambulant stage, but not exclusively. Monitoring of the muscle length of the upper limb is essential from an early age.
	There is no evidence to prescribe the exact frequency of stretches, length of hold etc however, normal muscle responds to 20secs of stretch. In practice, maintaining a stretch of 20sec may be difficult in the person with DMD however, clinicians do agree on a daily programme where possible.
	Symmetry to be promoted in posture, exercises and activities.

Orthoses

Night-time AFOs can be utilised from an early age to provide a sustained stretch of the gastroc/soleus complex, with the aim of maintaining ROM at the ankle. There is evidence to support use of AFO's for night-time. Scott et al., (1981) conclude that early and persistent use of AFO's at night delays tendo-achilles contractures and enhances walking ability. Hyde et al., (2000) concludes AFO's and stretching are more effective in control of a tendo-achilles contracture compared with stretching alone.

Intervention	**Recommendations**
Night splints (AFO's)	There is no evidence for the timing of the intervention but there is a consensus for the introduction of AFO's when ankle power is reduced or loss of range of motion into dorsiflexion is noted. Expert opinion agrees on early introduction and nightly use. Care must be taken if there is lining in the splints and the risk of infection should be considered. Night splints should be extended beyond the metatarsal heads.
Day time AFOs	There is no evidence for use of daytime AFO's in the ambulant child. Solid AFO's prevent the dynamic equinus which is an essential compensation to maintain independent gait. Insoles may assist in providing stance phase control of the foot. Non-ambulant patients should utilise daytime AFO's to maintain good foot posture and promote supported symmetrical sitting in a wheelchair. Day time AFO's should be supplied at the time of loss of ambulation. Trimlines may be behind the metatarsal heads for ease of donning footwear. DAFO's are not often recommended. Consider Northvane (or similar) material for casting AFO's if rigid splints are not tolerated.

KAFOs	KAFO's are not often used although some young people use them for therapeutic walking. Assessment of suitability should take place following discussion with specialist muscle clinic team.
Spinal Bracing	Spinal bracing can be used with young men who do not undertake spinal surgery. Consultation with the Spinal surgery team and Orthotist is essential.
Other splints	Contracture correction devices can be utilised for the upper and lower limbs. Occupational Therapy / Orthotics may be consulted for advice regarding upper limb splints.

24-hour Postural Management

Physiotherapists have a crucial role in the assessment and management of posture in lying, sitting, and standing.

Postural management is defined as “the use of any technique to minimize postural abnormality and enhance function” (Clark et al, 2003). It includes physiotherapy management in the form of stretches, and passive movement as well as within the prescription of equipment, orthoses, medication, and surgery.

24-hour postural management may be best introduced prior to the onset of contractures and deformity. Many therapists undertake postural management at different levels and intensity from diagnosis in the form of night splints, in soles, stretches and advice on seating and posture.

Postural management is best delivered in such a way that it encompasses rest, recreation, and activity. It acknowledges that static postures can be detrimental to the maintenance of function and that a variety of different postures are adopted throughout the day that will include asymmetrical positions. It also acknowledges the fact that prolonged static postures can be destructive, and a variety of different postures may be a useful goal.

Standing
Once standing posture becomes compromised, it may be useful to introduce specific standing support as often as is able.

This may occur when:

- Gastrocnemius / soleus complex becomes tight
- heel contact is lost when stationary
- there is asymmetrical standing posture
- when child is unable to stand still and /or standing balance is compromised
- the child is no longer able to stand independently

Initially, a standing frame may be appropriate for those still able to move independently or with some assistance. Some centres may use tilt tables however moving and handling should be assessed in all situations. For those requiring maximal help, sit to stand wheelchairs or powered sit to stand frames may be more appropriate.

There are many benefits associated with continued standing which include management of contractures, osteoporosis as well as maintaining good spatial awareness and self-esteem however, if comfort is compromised and pain is evident, standing may need to be discontinued. As with all treatments patient partnership and informed consent are essential.

For those children/young people when loss of walking is imminent, an informed decision must be reached whether to continue standing with supportive equipment. It is important to ensure equipment is provided quickly, prior to the onset of contractures.

For those children/young people with hip and knee contractures, there is no consensus as to when it is no longer appropriate to stand. Care should be given to position within the standing frame particularly hyperextension of the lumbar spine.

In some situations, appropriate orthoses may also be worn in the standing position.

Lying
Patients are generally reluctant to accept support in bed until independent changes in position are no longer possible however, by this time contractures may already exist. Lying postures can become problematic when

- the young person is no longer able to change position independently
- the young person requires frequent turning / changes of position through the night resulting in disturbed sleep for both his parents and himself
- the young person experiences pain from pressure or joint pain generally caused by weak muscles not being able to provide adequate support to the joints in certain positions

Patients are best when supported in a symmetrical position. This can be in side lying or supine however, there are pros and cons with each position. RCN recommends supine lying with 30^0 turns to prevent an increase in pressure on the greater trochanter.

Prior to the start of ventilation, side lying may be best for respiratory purposes as airways are more likely to be able to remain patent. The diaphragm is more efficient in side lying however care must be given to ensure that the child/young person spends equal amounts of time on either side. A preference on single side lying results in an asymmetrical thorax and often there can be difficulty when the young person commences supine lying when ventilation is required.

Supine lying enables the young person to ventilate both sides of his chest equally. It is often easier to maintain the head in midline avoiding asymmetrical contractures at the neck and hip joint integrity can be maintained with the use of pillows / positioning equipment.

Pressure re-distribution can be achieved using specific mattresses and if pressure is problematic, specific advice from the tissue viability service may be helpful. Visco elastic foam mattresses can be useful with pressure re-distribution however it should be noted that an increase in temperature can result. Sheets with a two way stretch are advisable to ensure that the sheet does not tighten underneath the young person hence increasing pressure and cotton bedding and nightwear for temperature control is advisable.

In later stages, boys with DMD are unable to undertake heat avoidance strategies by altering the bedding therefore care should be taken to maintain the bedroom at a constant temperature.

Pillows should also contain pressure re-distributing material to ensure that pressure areas do not develop on the back of the head (when supine lying) or ear (in side lying).

Sitting
It is acknowledged that sitting is a posture in which all activities are undertaken whether rest, recreation, or work. Sitting with an anteriorly tilted pelvis (forward sitting) is required for working whereas a posteriorly tilted pelvis is accepted as a position for rest and relaxation. Chairs that tilt and recline (whether wheelchair or static chair) are advantageous as they facilitate independent changes in position throughout the day.

It is important that physiotherapists are aware that hip dislocation and subluxation is common in DMD (Chan et al., 2001). Hip dislocation is secondary to pelvic obliquity which is secondary to scoliosis with muscle weakness as an important factor. Painful subluxation of the hip should be referred to an orthopaedic surgeon for advice.

Appendix Ten - Contact Details

Scottish Muscle Network

Muscular Dystrophy UK

Action Duchenne

Duchenne Family Support Group

Sleep Action

Disability sports co-ordinators for all areas are available via:
https://www.scottishdisabilitysport.com/about-sds/branches/

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