**Adult North Star Network (ANSN): Consensus document for therapists working with adults with Duchenne Muscular Dystrophy (DMD – Therapy guidelines supplementary document**

**Physiotherapy**

***Respiratory management***

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| **Assessment** | **Community therapy services** | **Specialist Neuromuscular service** |
| **History of chest infections** | History of chest infections over the last 12 months; frequency, duration, treatment required (including medical, PT, drug), requirement for A&E attendance or hospital admission | Knowledge of microbiology, obtaining sputum samples.  Future planning and management plan for infections and secretion management. |
| **Physiotherapy routine** | Awareness of the routine PT programme (or previous one) what the individual should be doing, who is helping them with this, techniques and equipment used.  Knowledge of the local specialist service and how to refer/ensure the young adult is being seen regularly. | Specialist assessment to establish and modifying management programme.  Training for the individual and care givers on the PT routine, management techniques, and escalation plan or what to do when unwell.  This will include use of non-invasive ventilation and airway clearance techniques – likely to include Mechanical Insufflation Exsufflation (MI:E; cough assist), Lung Volume Recruitment (LVR) bags, breath stacking and manually assisted cough [10,74].  A key member of the Multi-Disciplinary Team (MDT), liaison to ensure management is optimised, particularly around secretion clearance and Non-Invasive Ventilation (NIV). |
| **Drug** **history** | Working knowledge of the following medications/adjuncts, how often and when should they be used:   * Mucolytics-DNAse/Saline * Bronchodilators * Nebulised antibiotics * Saline nebulisers * Steroids/Salbutamol * Anti-reflux medication * Hyoscine/glycopyrrolate * Management of secretions (suction oral and tracheal) * Known allergies | Specialist knowledge of respiratory medication including specific indications and possible side effects. |
| **Social history** | The following factors will need to be considered in the management of the individual:   * Spouse/partner, family, care givers (who is involved to support) both formally and informally * Package of care/level of care and how this is funded * Access to respite or a hospice, where and how frequently * Housing situation * Work/education situation * Smoking history and alcohol intake | Specialist review with care givers on management plan and level of care required and being provided.  Specialist liaison and training of management techniques with care givers, care providers, hospice staff.  Advocating on behalf of the individual for specialist equipment which may be to Trust management/CCGs/CHC teams (particularly around need for MI:E). |
| **Respiratory Investigations** | Awareness of routine investigations and where to seek help in interpreting these as required:   * Lung function tests * Sleep study * Chest X ray * ABGs/CBGs/TOSCA * Cough strength/Peak Cough Flow (PCF) * Sputum sample | Specialist interpretation of these investigations and adjustment of secretion management or ventilation plan accordingly.  Carrying out these investigations if relevant training/competency has been achieved.  In addition to FVC and FEV1, to complete MIP/MEP/SNIP where appropriate [75,1].  Particular attention on repeated respiratory tests if requiring surgery (pre and post). Liaison with anaesthetic/surgical teams if NIV is required to be established during/post procedure or liaison with PT team for secretion clearance post procedure.  When a young adult with DMD is still ambulant respiratory issues are unlikely but it is important to monitor lung function: Forced Vital Capacity (FVC) measure and Peak Cough Flow (PCF) as a minimum [75,1]. |
| **Other considerations** | Awareness of the following:   * Individuals’ understanding and knowledge of DMD * Relationship with GP, district nurses, community staff * Swallowing problems/risk of OPD * Communication difficulties * Feeding difficulties (NG/NJ/PEG fed and routine) * Monitoring of weight * Gastro-esophageal reflux * Escalation of care * Advanced care planning and wishes for future (what has been discussed and by whom) | Recognising the symptoms of inadequate ventilation, poor cough, episodes of deterioration.  Identifying early signs of nocturnal hypoventilation (progressing to daytime eventually):   * Early morning headaches * Fatigue * Daytime sleepiness * Reduced appetite * Weight loss |

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| **Objective Assessment** | **Community therapy services** | **Specialist Neuromuscular service** |
|  | **Subjective Assessment:** It is essential to establish a clear history of normal respiratory function, recent problems, regular routine and management plan | |
| **Observations** | Working knowledge of basic respiratory assessments and be able to escalate care as required  Understanding of normal values of:   * Respiratory rate * HR * SpO2 * Temperature   Be able to identify increased work of breathing (can be very subtle signs):   * Not feeling well * Fatigue and lethargy * Altered breathing pattern * Excessive use of accessory muscles * Fixing with arms to use accessory muscles * Unable to lie flat * Tracheal/intercostal/subcostal recession | Detailed knowledge and understanding of specific issues related to DMD:  Chronic hypoventilation:   * Sleep disturbance * Waking from sleep * Early morning headaches * Waking catching breath * Difficulty finishing sentences * Poor concentration   Chronic respiratory failure can be very subtle (and may not be accompanied with respiratory distress).  Identify Glossopharyngeal Breathing and other self-insufflation compensatory mechanisms. |
| **Auscultation** | Auscultation (if available) and chest wall palpation for assessment of:   * Chest wall crackles * Audible secretions * Changes to chest wall expansion (may be very limited normally) * Any asymmetry | Advanced auscultation skills in conjunction with other objective assessment. |
| **Secretion** **Clearance** | Assessment of the components of cough and strength of cough with PCF measurement if available  Can the individual clear secretions adequately?  What strategies / techniques / equipment is being used and who is assisting?  How frequently are they experiencing secretions and what is normal for them (colour / amount / thickness)?  Is suction required (oral / nasal / tracheal)? | Specialist assessment at each review on all these aspects.  Interpretation of lung function tests, specialist assessment of ability to clear secretions, including PCF.  PCF value exceeding 270 – 300L/min is required when a patient is well, in order to maintain PCF > 160L/min and effectively clear secretions when unwell [76].  To be able to reason and utilise appropriate secretion clearance technique e.g. LVR or MI:E [6] and to liaise with SLTs and analyse MIC-VC difference if patient has adequate bulbar function for techniques to be beneficial.  Ongoing review and modifying of techniques to maximise adequate secretion clearance. |
| **Spine / Posture** | Ensure chest shape and spinal posture is observed regularly.  Has the individual had spinal surgery previously?  Understanding and knowledge of the impact 24-hour posture can have on respiratory function. | Specialist liaison with MDT and spinal specialists as required.  Liaison with local wheelchair services around adaptions to accommodate changing respiratory needs (carrying equipment / mounting equipment). |
| **Ventilation** | Establish the individual’s dependency on ventilation how many hours are they using it?  Who is helping the individual with ventilation (including cleaning and changing of consumables)?  Awareness of any change in routine if the individual is unwell and be able to contact respiratory team if concerned. | Advanced specialist knowledge on the different types of ventilation options available.  Detailed assessment for the need for ventilatory support, establish on ventilator support; review of ventilatory settings and highlight concerns to MDT.  Use remote monitoring as available, to identify concerns and intervene accordingly optimising management. |

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| **Management** | **Community therapy services** | **Specialist Neuromuscular service** |
| **Airway Clearance techniques** | Working knowledge of the following techniques:   * Active Cycle of Breathing Techniques (ACBT) with huff / cough * Manual assisted cough * Positioning | Specialist knowledge of adjuncts for airway clearance and providing these or liaising with other services for provision [5,75,77,78]. These maybe assisted inspiration, expiration or a combination of both for cough augmentation, or peripheral airway techniques [11]:   * LVR bag * Air stacking technique +/- one way valve * Mechanical I:E device * Intermittent Positive Pressure Breathing (IPPB) * Manual hyperinflation * Inspiratory positive airway pressure (IPAP) or Volume mode on NIV can be adjusted to improve tidal volumes during treatment * Manually Assisted Cough * Percussion, vibrations, HFCWO, HFCWC and IPV * Suction if appropriate oral, oral-pharyngeal/ nasal/tracheal as indicated.   Regular evaluation of techniques, effectiveness and modifying management programme. |
| **Ventilation** | Awareness of inadequate ventilation, when an individual’s respiratory care is not optimised (subjective, objective signs).  Sharing concerns promptly with specialist services.  Awareness that oxygen should not be used routinely. In an acute respiratory crisis supplementary oxygen can be used but carefully controlled and monitored, best done in acute care. | Specialist knowledge of management plan to include:   * Ventilator type, use, component parts * Need for second ventilator * Mode of ventilation * Interfaces including mouthpiece ventilation (when indicated and how to establish) * Tracheostomy care * Use of humidifier or not * Battery options * Routine of consumables (supply and delivery)   Key role in setting up and establishing a routine on non-invasive ventilation (NIV) alongside MDT [1].  Key specialist role in the adjustment of the settings / achieving compliance / management plan as required (including proactive planning for when unwell and use of second machine profile if available for times when unwell/for secretion clearance).  This will include regular reviews and at times of acute infection (including weaning) both with NIV and invasive ventilation.  Liaison with MDT (in particular SLT colleagues) regarding use of speaking valves with tracheostomy use. |
| **Other equipment** | Awareness of other equipment / techniques that may be being used:   * Nebuliser * Airway clearance techniques * Suction | Liaising with community services to ensure access to other equipment as required and providing supporting information for these. |
| **Education and Training** | Support the implementation of care plan as part of wider management, support regular review of care plan as care givers / individuals care system changes over time | Specific competency training in specialist equipment / adjuncts completed with the individual and care givers.  Liaison (with wider MDT) on issues around home antibiotics / vaccinations. |

**Rehabilitation physiotherapy**

*Contracture management*

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| **Ambulant DMD** | **Non-ambulant DMD** | **Recommendations** |
| Ambulant individuals require dorsi-flexion ROM for heel strike and weight bearing in stance phaseof gait as well as plantar flexion for effective toe off and propulsion. Eccentric dorsi-flexion is also required for climbing stairs. | Wheelchair users need to maintain ankle dorsiflexion so that feet can be positioned well onto footplates for comfort, also for transfers if still able. Weight bearing through plantar grade foot position maintains a stable pelvis which aids spinal posture and upper limb movement.  Foot positioning in neutral is recommended to avoid hip and knee pain and contractures, caused by external rotation at these joints. To maintain adequate length of Tendo Achilles netural foot position should be facilitated.  If foot is dorsiflexed, check posterior ankle creases for pressure and for feet that are tending toward supination, callous formation at the base of the 5th metatarsal is often common and uncomfortable. | Static and dynamic stretches  Resting splints  Dynamic splints  Avoid central footplates on wheelchair. Bilateral footplates to promote normal lower body alignment.  Standing frames / standing electric wheelchairs can assist weight bearing if the individual is losing ambulation. |

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| **Ambulant DMD** | **Non-ambulant DMD** | **Recommendations** |
| Important to maintain good muscle length to preserve mobility. | At the knee proximal gastrocnemius and distal hamstrings can become contracted particularly in a permanent wheelchair user; due to the 90° knee bend position.  Maintaining knee extension through stretching is important to prevent hamstring contractures that can cause pain and difficulty with positioning in bed, sleep position and seating. | Static and dynamic stretches.  Standing frame / standing electric wheelchairs to encourage weight bearing and maintain muscle length if the individual has enough muscle power to use equipment.  Ambulant individuals may be using Knee Ankle Foot Orthoses (KAFOs) with locked knee joints, to help maintain mobility and joint position sense. |

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| **Ambulant DMD** | **Non-ambulant DMD** | **Recommendations** |
| Important to maintain good muscle length to preserve ambulation and bed / chair mobility. | ITBcan become contracted causing the hips to fall into an externally rotated position (splayed hips) increasing the chance of hip dislocation.  It can also make positioning in a wheelchair difficult if the hips become too abducted and externally rotated. | Static and dynamic stretches.  Standing frame / electric standing wheelchair to encourage weight bearing and maintain muscle length if the individual has enough muscle power to use equipment.  Using pillows or supportive sleep systems to help support while in bed. These can help maintain neutral alignment at the hips and stop the hips abducting out (frog leg position).  Using lateral supports / hip guides in the wheelchair along with good foot position in sitting, can assist with maintaining ITB length. |

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| **Fingers and wrists** | **Recommendations** |
| The long wrist and finger flexors become particularly tight. | Stretches should be done daily to maintain ROM of fingers and open up palms of hand.    Palm protector splints, finger spacers and resting splints can all be encouraged overnight or alternating over hands throughout the day to help maintain hand position, provide prolonged stretch and manage hand hygiene. |
| **Elbow** | **Recommendations** |
| Elbow stretches to maintain arm length are required so the patient can reach wheelchair controls and to help with continuing independence with feeding. | Resting splints and serial casting can be considered.    Elbow contracture control devices (ECCD) can help maintain elbow range |

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| **Neck and upper thoracic** | | **Recommendations** | |
| Neck position and thoracic extension should be maintained as able, to help preserve an upright posture. | | * Thoracic stretches can be done in supine without a pillow by raising arms above shoulder level. * Specialist orthotics can be made to measure, to help with head and neck support as well as different head supports on wheelchairs should be considered. * Lateral trunk supports and moulded seating for wheelchair to be considered * Neck stretches in the bed are useful to help preserve neck rotation. * Tilt in space wheelchairs help counteract a forward flexion position, however care must be taken of respiratory compromise in this position. | |
| **Assessment** | | |
| **Posture / spine** | **In standing** check for:   * calf hypertrophy * significant lumbar lordosis   (both common in this group)   * axial weakness can be significant * Spinal posture must be assessed, particularly as weakness progresses and especially when ambulation is lost. * Note shoulder and hip symmetry and any scapular winging.   **In sitting and lying**:   * assess the spine * check the position of the legs (common to adopt a ‘Frog-legged’ posture – hip external rotation, abduction and flexion with knee flexion)   In sitting and standing observe for evidence of scoliosis and assess ability to correct scoliosis passively. | |
| **Management** | | |
| Posture must be managed throughout a 24-hour period. This requires close working with OT colleagues and specialist wheelchair services [1,3].  Education of the young adult, families and care givers around 24-hour postural management, particularly optimising chest wall compliance, spinal alignment and upper limb function is important. | | |
| **Posture / spine** | **When in bed:** Use of sleep systems to encourage symmetry may be beneficial but are not always well tolerated.  Adults should be referred for an orthopaedic assessment if a scoliosis is apparent. Those adults who lost ambulation before reaching puberty are most likely to require spinal bracing and surgical stabilisation as interventions for scoliosis. However, with the advances in medical management, posture management with sleep systems / wheelchairs and individuals with DMD staying ambulant for longer, surgical and bracing interventions are used less frequently.  If the spine has been surgically corrected it may still be beneficial to support the legs to maintain alignment at the hips and knees. This can be done by pillows or positioning rolls. An E shaped roll or cushion is particularly helpful in reducing the frog-legged posture.  **When sitting:** This is usually managed in the wheelchair so liaison with wheelchair services is important.  It is important to be aware of the incidence of vertebral fractures which could cause pain as a symptom but could also be asymptomatic.  Postural management at the trunk in terms of therapeutic seating can be considered. However, be mindful of maintaining functional ability, along with truncal support. | |

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| **Wheelchair considerations** | |
| **Assessment** | It is helpful to start by assessing pelvic position.  A seat support assisting the proximal lower limb to rest in a neutral position with feet directly below the knees can help avoid abduction and external rotation at the hips.  A position with abducted and externally rotated femurs will induce a more everted ankle position and so it is important to commence positioning from when the user first starts using a wheelchair.  Some young adults may be very thin, and others may be obese, and some others may lose or gain considerable weight as their condition progresses. Regular reviews to assess changing needs is required. |
| **Arm Function** | With progressive deterioration, arm function may continue to worsen and additional provision for equipment like mobile arm supports may improve arm function.  Individuals may have difficulty using standard joystick controls and modifications of the joystick and interface should be considered.  Progression in trunk weakness could have an impact on arm and hand control and adequate trunk support may be necessary to facilitate hand function.  A few millimetres of change in position of controls can render an individual unable to use the joystick – and when assessing individuals this needs to be high on the therapists’ assessment. |
| **Pain and Fatigue** | As the condition progresses, increased fatigue is a common feature. The user (particularly in the later stages of DMD or following spinal surgery) may need to rest in the wheelchair during the day. |
| **Posture** | When considering posture, it is best practice to aim for a symmetrical position however, if contractures are present then this may not be possible. We know that function is of the utmost importance to maintain independence and sometimes the most functional position is not the most anatomically symmetrical position. In these cases, the position may have to be altered to maximise functionality when undertaking a specific task and having the option to return to a more symmetrical position when resting can be a good compromise.  In many instances the user could be spending up to 15 hours a day in a wheelchair, so comfort, access, function and pressure redistribution will be important issues to address. Individuals could be at high risk of developing pressure areas so proactive management ofpressure is essential.  Individuals may have tightness, contractures and fixed deformities which may require supportive bespoke seating.  A centrally located foot support can bring the feet together and with poor strength in the muscles around the pelvic girdle, this can encourage abduction and external rotation at the hips. Careful assessment of optimum foot support is important. |
| **Head and trunk support** | With progressive deterioration of head and/or trunk muscles, further postural control is a consideration. All options of support and pressure redistribution such as tilt in space, headrest with height adjustment, forward /backward /sideways adjustment should be considered. Head support used with tilt-in-space, enables the user to tilt back with the head well supported and using the tilt function without head support is not recommended.  Some individuals may have had spinal surgery to correct scoliosis. This could have an impact on positioning and arm function. If spinal surgery was not performed, then scoliosis maybe present and additional truncal support or moulded seating should be considered. |
| **Powered tilt-in-space** / **recline** | Having the provision of powered tilt-in-space wheelchair with the addition of powered recline, would enable the user to change position independently and helps avoid the need to go to bed during the day.  The tilt-in-space facility would also assist with changing position thus helping with pain management. It is important to consider powered elevating leg rests (if no fixed contractures), to assist with change in position and pressure.  The recline function is also helpful to open the hip angle and can make toileting and adjustment of clothing after toileting much easier. It is also useful if the user has a gastrostomy or requires manual chest clearing techniques. For some users, having a powered tilt and recline function enables them to lie in a reclined position which is helpful for doctors’ appointments, investigative procedures and dental procedures without the need to transfer out of the chair.  Individuals may possibly have oedema/swelling of the feet and ankles and this can also be supported with electric adjustable leg and foot supports. |
| **Riser function** | As powered mobility should be introduced when young men are still walking short distances, the riser function prolongs independence by raising seat height for transfers.  To maximise functional independence and socialisation with family, peers and society; the riser function enables young men to engage at the same height.  A riser function will enable access to bench tops and work surfaces at all heights, alongside peers throughout college, university and employment. |
| **Assisted standing** | Assisted standing if safe and achievable, even when the adult is unable to walk, can help maintain bone density, prevent/delay the development of contractures and assist with kidney, bladder and gastro-intestinal function. |
| **Environment access** | As the condition progresses, it is important to maintain independence within the wheelchair. This may require the use of environmental control systems. The goal always must be to maximise upper limb and hand function.  Increased weakness in upper limb/hand function may make it difficult to use standard joystick controls. It is important to adapt the joystick control with appropriate mounting and function of joystick. Light touch or sensitive joystick must be considered if indicated. All available alternatives must be assessed and explored with the goal being to provide the user with maximum possible independence.  When setting up the system, ensure the user does not have to lean towards or away from the control to obtain optimum leverage.  Different or repositioned switches may be required to change speed or function and maintain functional skills.  Trays or support for upper limbs if required are considered to increase upper limb functional ability.  Trays also offer support for those with subluxed / dislocated shoulder joints and will support the weight of the arm; relieving stress from the weak shoulder girdle area.  Different activities may require different positions and adequate access to desks, tables and work surfaces should still be attained. Electric seat elevation function should be considered to optimise participation. |

**Exercise therapy**

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|  | **Recommendations** |
| **Contraindications and exercises to avoid** | End stage cardiomyopathy  Specific hydrotherapy contraindications:   * implications on cardiac output * increased resistance to respiratory function   Exercise using heavy weights or excessive resistance with the aim of muscle hypertrophy  Exercise with eccentric loading |
| **Precautions to consider when prescribing exercise** | Compromised cardiorespiratory function  Implanted cardiac defibrillators or pacemakers  Recent surgery  Recent injury  Marked osteoporosis  Excessive pain and fatigue |
| **Symptoms of overuse in DMD [3]** | Excessive fatigue (leading to temporary loss of function)  Muscle aching and/or pain lasting for between 24-48 hours  Myoglobinuria  Elevated creatine kinase levels above base line (if readings or testing procedure available). |
| **Recommended pre-exercise assessments and goal setting:** | Confirmation of stable cardiac status  Respiratory assessment  ROM of upper and lower limbs  Strength assessment of upper and lower limbs  Subjective and objective assessment of pain and fatigue  Goal setting:   * What does the individual want from exercise? * What interests them with regards to activities? |

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|  | **Recommendations** |
| **Frequency (number of times per week)** | Increasing daily activity from a sedentary baseline would be recommended with a **‘**little and often’ approach. As with all exercise recommendations in DMD there is a paucity of evidence. Frequency would be dependent on physical ability, however one to two times per week could prove beneficial. |
| **Intensity** | An individual’s response to exercise, should be individually assessed and carefully monitored. Individuals experiencing any of the overuse symptoms should modify the exercise undertaken or reduce the intensity of exercise. Peak heart rate is not a good assessment tool, DMD patients frequently have baseline tachycardia and are often treated with beta blockers.  Perceived exertion scales can be used with the caveat that the individual requires a familiarisation with the scale before it can be safely used.  During, immediately after and for 24-48 hours post exercise there should be no loss of function, muscle pain [3], or excessive fatigue. |
| **Type** | The recommendations, although evidence is sparse, are that low intensity aerobic exercise is proven to be beneficial and more importantly, safe [3,15,79,80].  Types of low intensity aerobic exercise would include (but not limited to):   * powered arm or leg pedals * active assisted upper and lower limbs ROM exercise (including stretching) * use of specific gym ball exercise with truncal rotations * hydrotherapy or aquatic therapy (with contraindications ruled out) * very light seated activity such as playing a Wii game or typing [81] |
| **Time** | The amount of time an adult with DMD exercises or engages in increased activity is dependent on the individuals’ ability and availability of support.  As stated previously ‘little and often’ is more than likely going to prove the most beneficial and safest recommendation in terms of time spent exercising. |

**Occupational therapy**

*The Person Environment Occupation Model [82]*

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| **PERSON** | | |
| **Problem Area Affecting Participation** | **Assessment** | **Interventions/Outcome to Maximise Participation** |
| Poor fine motor skills and ROM.  Non-existent or limited upper limb function – related to muscle weakness, affecting ability to make active movements [84]. | * Observations (contracture, colour, hygiene, position, use of hands during discussion). * Assess ROM, pain, contractures. * How they use their upper limb now? * What compensations do they use? * Equipment used? * Use of splints (functional and management) – current/previous. * Functional upper limb assessmentse.g. PULM [32], to measure change in function and as an outcome measure. | * Develop compensatory strategies. * Refer red flags to MDT team. * Equipment - arm supports, assistive technology [83]. * Positioning – Strategies to maintain strength and ROM to promote participation [30]. * Education about condition and how it affects their upper limbs and function. |
| Emotional and Psychosocial Functioning:  Link between feelings, thoughts and behaviours for an individual who has a progressive, life-limiting condition, which can have a negative impact on capacity to participate in meaningful activities [31]. | * Emotional and psychosocial impact of disease progression, constant changes, series of losses of ability and skills. * Support required to plan for future needs related to disease progression e.g. wheelchair, respiratory, cardiac, medication. * Adjustment - grief and loss relating to function/participation. * Body image, self-esteem, resilience, mood, frustrations. * Quality of and impact on relationships with others. | * Referral to appropriate professionals e.g. Psychology, Neuromuscular Care Advisor. * Facilitation of support groups/ education groups. |
| Cognitive Functioning:  Ability to acquire information and learn new skills or adapt to support participation given the incidence of additional learning needs in some individuals with DMD [85]. | * Attention/Awareness. * Response to cues – visual, auditory, motor. * Diagnosis of conditions that affect cognitive functioning? * What do they want to achieve? * What are the cognitive barriers to reach this goal? * Task and Environment Analysis – of identified activity to determine areas of difficulty/barriers for participation. * Cognitive assessment * Canadian Occupational Performance Measure (COPM) [33]. | * Changes to physical/social environment/task to match cognitive abilities to support participation. * Facilitating understanding of the affects that the lack of dystrophin has on the brain especially executive functioning skills. * Referral to disability officers in university/college. * Referral for assistive technology assessment. |
| **ENVIRONMENT** [86,87] | | |
| **Problem Area Affecting Participation** | **Assessment** | **Interventions/Outcome to Maximise Participation** |
| Environmental barriers/ facilitators to participation:   * Physical – Natural and man-made surroundings * Social – Relationships and social groupings * Cultural – ethnicity and routine practices of particular social groups * Institutional – Policies, services and economic issues. | * Identification of appropriate environmental adaptations. * Ability to access community. * Availability of support networks and relationships. * What are their perceptions around disability and dependency and the influence that has on participation. * Availability of finances, transport, carers. * Task and Environment Analysis – to determine areas of difficulty/barriers for participation. * COPM. | * Referral and signposting to relevant agencies: Local Authority and Community Care services - benefits advice, re-housing advice, accessible transport, support groups. * Referral for assessment for Continuing Health Care/ * Personal Health Budget– may be eligible for Personal Assistant to support access to environment. * Environmental adaptations. * Education: increase awareness about the environmental barriers/facilitators. |
| **OCCUPATION** [28] | | |
| **Problem Area Affecting Participation** | **Assessment** | **Interventions/Outcome to Maximise Participation** |
| **Self-Care**  Washing and bathing:   * Decreased function affects participation * May need to use a ventilator during bathing.   Toileting:   * Reduced hand function for cleaning self. * Use of urine bottle affected by positioning and impact of steroids on body mass and development.   Grooming:   * Reduced strength impacts on shaving, brushing teeth and styling hair.   Dressing:   * Ability to participate is reduced due to muscle weakness.   Eating and Drinking:   * Fatigue affects ability to feed.   Sleep:   * Will need repositioning during night due to joint pain/stiffness and immobility. * Decreased physical exertion will alter sleep patterns. * Impact of awake carer present overnight on sleep/partner/ family. * Impact of lack of sleep on mental health and well-being.   Expression of Intimacy and Emotions:   * Individuals should be empowered to discuss their sexual health and well-being. * Physical barriers to demonstrating emotion. | * Physical support required, time taken, carer’s involvement and equipment used. * Moving and Handling - equipment and who is using it, accessibility, transfers, techniques, environment * Bath vs shower – pain, positioning, carer safety, transfers. * Toileting - equipment, transfers, moving and handling, postural support, slings, wash/dry toilet. continence, pads, bottles, any problems with skin integrity * Environmental issues - Access to sink/tap/hair dryer and mirror. * Empowerment to choose products and voice preferences. * Sensory issues e.g. creases in socks under orthoses – risk of pressure sores. * Poor circulation – potential impact on outfit choices. Cold feet need to be considered when in/out of bed. * Ability to use cutlery; to lift food or cup to mouth; to cut up food; any equipment used; problems with chewing and swallowing? * Weight and nutrition status. * Knowledge of specialist equipment e.g. arm supports/neater eaters. * Environmental considerations e.g. where do they eat? With partners/family or alone in bed? * Consider portion size if fatigue an issue/ re-heating meal if cold due to length of time taking to eat. * Quality of sleep, triggers for disrupted sleep, respiratory issues, equipment, pain, turning required, type of bed and mattress, sleep system, sleep routine/hygiene practices? * Turning beds – may have impact on carers. * Anxiety can reduce quality of sleep (does the person have unanswered questions related to prognosis or future deterioration? Who can they have these conversations with?) * Respiratory impairment will impact on quality of sleep – *look at respiratory section for more details.* * The importance of physical intimacy can be underestimated and lost when an individual has regular physical contact for care needs. * Effect of continence or medication issues, self-view, parental/family attitude. | * Moving and handling advice. * Specialist Equipment assessment and advice, positioning, pacing, adapting (Access sling may not be appropriate due to poor core strength and balance). * Encouragement to make healthy eating choices due to side effects of steroids. * Ensure carers are educated in safe and social interactive feeding. * Referral to group individual therapy/counselling to explore issues relating to sexuality or anxiety related to prognosis. |
| **Problem Area Affecting Participation** | **Assessment** | **Interventions/Outcome to Maximise Participation** |
| **Productivity**  Ability to participate in meaningful activities that contribute to wider society | * Is the environment accessible? * What are their aspirations and the value and meaning of work/education? * What is the skill set or previous experience? * What support would the individual need to meet entry level requirements for academic courses? * What compensations/adaptations are utilised (equipment, carers, reasonable adjustments)? * Impact on carers/family. * The person’s knowledge and utilisation of the Equality Act 2010. * Access to outdoors – pubs, restaurants, theatre, sporting events clubs etc. * Explore role as a parent * Explore barriers to parenting   Occupational Performance Assessments:   * Activity analysis * COPM * Completion of vocational rehab assessment | Referral and signposting to relevant area of expertise and access to specialist Vocational Rehabilitation support, e.g. Access to Work, Specialist Employment Services, Disability Support Services, Educational and Health Care Plan assessment.  Sign posting to and utilising volunteering and paid employment opportunities available through third sector organisations  Young person who is not ready or able to participate fully in adult activities, families might need guidance on Guardianship.  Problem solve parenting difficulty  Referral to appropriate charities |
| **Problem Area Affecting Participation** | **Assessment** | **Interventions/Outcome to Maximise Participation** |
| **Leisure** | * Consider personal relationships, peer groups * Hobbies and what barriers are impacting participation; physical and social environment; financial * How does the individual spend their time? * Do they want to increase leisure activities? * Completion of interest checklist | Referral and sign posting to local services  Interventions should be focused on strengths, interests and be meaningful  Compensatory skills/adaptation of chosen leisure activity |

**Speech and language therapy**

**Assessment of oropharyngeal dysphagia (OPD)**

|  |  |  |
| --- | --- | --- |
| **Subjective Assessment** | | |
| **Self-evaluation** | | |
| **Benefits and considerations** | The unique benefits of self-evaluation to identify OPD are:   * practicality and cost [88]. * can be completed at home without direct access to a health-care professional. * promote independence and self-advocacy, consistent with DMD specialist guidelines and government strategy [1,3,89]. * regular, repeated patient-centred monitoring from home is possible * promotes a proactive approach to OPD self-management.   However, accurate self-evaluation relies on intact cognition, sensation, and self-awareness [90,91]. Intellectual impairment is present in approximately one-third of patients [70] and may impact ability to detect changes in their condition.  On this basis consideration should also be given to proxy-reporting and or adjunctive measures that yield quantitative information regarding the likelihood of presence or absence of OPD. | |
| **Tools** | There are currently no existing validated self-evaluation tools for diagnosis of OPD in DMD.  However, the tools below may be considered:   * Sydney Swallow Questionnaire (SSQ) [92]. The SSQ is proven to have validity in capturing changes in OPD symptoms overtime and has undergone early validation as a diagnostic tool in DMD [62]. A score of 224.5/1700 is suggested as a diagnostic cut-off. * Munich Dysphagia Test – Parkinson’s Disease (MDT-PD) [93]. An as yet, unpublished synthesis of literature has identified the MDT-PD as containing the greatest number of symptom specific items of OPD in DMD. This tool has not been validated for use in DMD.   With both questionnaires their downfalls should be borne in mind should they be integrated into clinical practice.  Additional questions may be asked to supplement symptoms not represented within the questionnaire as part of screening. | |
| **Screening** | | |
| **MDT clinical team screening** | There is consensus in existing literature supporting the existence of the following symptoms specific to swallowing changes in DMD:   * Difficulties in chewing * Difficulty clearing the mouth or food residues * Slowed eating or prolonged mealtimes * Drinking fluids to help push food down * Hard or effortful swallowing * Difficulties with food more so than drink * Feeling of an inability to swallow or initiating a swallow * Coughing, choking or throat clearing * Feeling of food catching in the throat * Needing to swallow more than once * Feeling unable to get food to the back of the mouth   Symptom-specific questions should be incorporated into clinical screening interviews by a suitably qualified healthcare professional which may include the patients’ neurologist, GP, clinical nurse specialist or therapist.  Questions and clinical history suggestive of weight loss and/or recent history of chest infections should also be incorporated. | |
| **Adjunctive screening tools** | Additional screening tools should be considered for patients undergoing more comprehensive clinical assessment.  There are currently no validated screening tools in this population however tools such as the timed test of chewing could be considered a relevant tool to capture early signs of chewing difficulty and oral dysphagia. | |
| **Clinical Assessment** | | |
| **Assessment of OPD** | In the presence of signs or symptoms of OPD, patients should be referred for assessment by a SLT, ideally with specialism or access to specialty advice in neuromuscular disease +/- respiratory care.  Assessment should aim to include:   * A comprehensive case history * Liaison with MDT specialisms to gather information on patient mobility, functional independence, spirometry and ventilation status as appropriate * Clinical bedside assessment including bulbar assessment and assessment of peak cough flow   The proforma in *Appendix A* is offered as a tool to ensure thorough information gathering and tailored assessments for patients with DMD. This tool has been developed by a SLT specialised in neuromuscular diseases. Whilst the tool has not yet undergone validation, it has been used clinically.  The ability of the clinical bedside assessment to identify early or sub-clinical dysphagia in DMD is neither agreed nor understood [48,49,60].  Until validated assessment tools are developed, instrumental assessment via VFSS or fibreoptic endoscopic evaluation of swallowing (FEES) is considered gold-standard for assessment and management of OPD in this population.  Benefits of instrumental assessments include:   * opportunities to tailor management programmes * support understanding of the dysphagia as a possible contributor to underlying respiratory infections * assess the impact of interventions on swallowing * provide an opportunity to monitor the dysphagia longitudinally.   **Limitations of instrumental assessment:**  Equipment limitations:   * lack of portability and accessibility * high cost * radiation exposure (VFS) and * discomfort (FEES), * regional variability of availability of access to instrumental assessment   Individual limitations:   * poor visualisation secondary to altered posture [47,48] * inability to sit upright due to scoliosis [61] * anxiety [48] * inability to complete protocols due to pre-initiated dietary adaptations [19]   With this in mind, the flow-chart listed in Figure 2 of the main document is offered as a support-tool to help non-specialist clinicians streamline referrals for instrumental assessment. | |
| **Interdisciplinary Interactions** | | |
| OPD should not be assessed and managed on a unidisciplinary basis. The need to consider ‘the whole person’ is particularly paramount in DMD given the multi-system nature of the disease.  For comprehensive and specialist management of OPD, working relationships should be built and maintained with respiratory physicians and PTs, neuromuscular dietitians, OTs, gastroenterologists, ear nose and throat consultants, ophthalmologists and anaesthetists.  In assessing OPD, the following should also be considered:   * Respiratory status * Cough function * Gastroenterological impairment including reflux disease * Ophthalmology * Nutrition and hydration * Posture and mobility | | |
| **Respiratory status** | | Patients with low lung capacity reduce the time of expiratory apnoea after swallowing. Reduced expiratory apnoea triggers early inspiration which, in turn, increases the risk for aspiration and O2 desaturation [67]. This risk is further increased in the presence of pharyngeal residue [61].  In patients with normal SpO2 but who are complaining about dyspnoea and respiratory fatigue during the daytime [94], interventions to normalise blood gases/treat these symptoms may be relevant. Small scale research currently suggests that invasive ventilation via tracheostomy may improve aspects of swallowing [95] and mouthpiece ventilation rather than non-invasive positive-pressure ventilation during mealtimes a preferred and safer method for swallowing [96]. |
| **Cough function** | | The ability of the patient to reflexively and voluntarily clear their airway should be considered as part of the OPD assessment.  Measurement of cough peak flow should be integrated into assessment to guide the clinician in risk assessing consequences of dysphagia. Those with a cough peak flow under 270l/min should be referred to a respiratory PT for chest clearance and/or cough augmentation. |
| **Gastroenterological function** | | Whilst the prevalence of gastroenterological function in DMD is unknown, disturbed gastrointestinal dysfunction such as reflux disease, dysmotility, hypomobility and delayed gastric emptying is increasingly reported [97].  Reflux is frequent in non-ambulatory individuals; the main cause is the long term sitting position that induces a permanent pressure gradient between the abdomen content and the thoracic one, combined with the progressive degradation of the diaphragmatic muscular tone.  Early satiety, progressive lack of appetite and decreased intake of food may be suggestive of primary oesophageal issues and warrant further investigation by a gastroenterologist.  Careful exploration of symptoms in consultation with a gastroenterologist is therefore warranted to refine assessment and treatment. Oesophageal manometry and 24pH monitoring may be amongst their choice for assessment. |
| **Nutrition and hydration** | | Low weight /weight loss could be considered as a possible sign of undiagnosed OPD but could also contribute to a worsening profile of OPD.  Careful attention should be paid to changes in meal-time regimes such as lengthened meal-times, food avoidance and adaptations which may provide vital clues to emerging OPD. |
| **Posture and mobility** | | Scoliosis may also contribute to OPD profile. OPD should be assessed in the context of the wider mealtime +/- occupational therapists and physiotherapists, with consideration for:   * Posture * Positioning for meals * Level of assistance required * Upright vs flat posture   Postural adjustments have been reported as strategies to support management of OPD [98]. |
| **OPD Assessments in the acute hospital** | | |
| Whilst the emphasis of OPD assessment is on proactive, preventative identification and management, there is always the possibility that OPD maybe present /apparent only after emergency hospital admission.  Given the potential for decompensation of the swallow at times of acute illness, attention should be paid to the cause of hospital admission and likelihood of any reversible issues, including alterations in swallow ability. | | |

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| --- | --- |
| **Treatment and Management of OPD in DMD** | |
| **Self-management:** | * Dietary adaptation is common [48,50,57,61] * Move to small frequent calorie-rich diet with fluids washes [63]. * Given the issues with pharyngeal clearance, thickening of fluids is not recommended in routine practice. |
| **Interface with NIV:** | At a late stage of disease progression, the extension of nocturnal NIV into the daytime reduces dyspnoea, restores respiratory muscle endurance and improves eating [94,99].  Improvements have been seen with breath-swallow coordination for patients drinking whilst on NIV [63], however until frontline devices are developed to allow users to halt the ventilator inspiratory phase for swallowing it is down to the individual to find a way to regulate the ventilator with their swallowing which can be challenging for some but has been witnessed in small numbers. |
| **Airway clearance:** | The effectiveness of instrumental and non-instrumental non-invasive tracheal clearance techniques should be evaluated in conjunction with the respiratory physiotherapist.  When aspiration is evident with clinical sequalae, the choice of the fastest and most effective technique must be made:   * manual chest compression and air-stacking in combination or * MI:E |
| **Enteral feeding:** | * First line management would routinely be oral supplementation with dietetic overview * Invasive options such as gastrostomy are second line [69]. In many, gastrostomy is often used as a complimentary adjunct to oral intake rather than alternative.   Points to consider:   * Early information and decision-making re gastrostomy to support advanced decision making is advised [100] * Early involvement of cardiac, respiratory and anaesthetic care teams to address changing risks that would be associated with gastrostomy procedure. * Recent advent of bedside gastrostomy placement helps to reduce risk of mortality and intra and post-surgical complications [54]. |
| **Treatment of underlying impairment:** | There is limited data addressing the benefits of exercise therapies for management. The limited evidence available suggests that exercise therapies improve chewing or swallowing skill rather than strengthen underlying muscles [100]. |

**A tool to ensure thorough information gathering of dysphagia signs and symptoms in DMD.**

|  |  |
| --- | --- |
| **Patient**  **perception of swallowing difficulties**  Description of signs and symptoms, compensations |  |
| **Chest history:**  🞟 Last 12 months in detail  🞟 Antibiotic use  🞟 Hospital admission  🞟 Chronic chest diagnosis  🞟 Spirometry (FVC/FEV1)  Required ventilatory support   * + Invasive   + Non-invasive (mouthpiece/nasal mask/full face mask)   + Required hours of use   Need for cough augmentation |  |
| **Weight history**  🞟 Last known weight  🞟 Current weight  🞟 Explanations for weight change (gastro, mood, appetite, mobility, food preparation, swallow) |  |
| **A ‘usual’ day of meals**  🞟 Compare to 12 months ago  🞟 Explore food avoidance/adaptation |  |
| **Baseline observations**  🞟 Including other comments/relevant additional information |  |

**A tool for bedside dysphagia assessment in DMD**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Oro-motor assessment** | | | | | |
| **Face** | Brow furrow (assess loss of movement in eyebrow raise) | Normal | Mild loss of movement | Moderate loss of movement | Severe loss of movement/No movement |
| Eye close (assess eye closure for sleeping) | Normal | Mild gap\* (>1mm) | Moderate gap\*  (1-3mm) | Severe gap\* (>3mm) |
| Eye squeeze (assess eye closure with effort) | Normal | Mild loss of squeeze | Moderate loss of squeeze | Severe loss of squeeze/no squeeze |
| Lip seal against resistance (assessment ability to seal air in cheeks with pressure) ●● | Normal (no air loss) | Mild (transient or short burst of air loss) | Moderate (prolonged air loss) | Severe (inability to seal any air in cheeks) |
| **Bite** | Masseter activation (assessor place three middle fingers over masseter muscle) (select up to two responses) | Normal (provokes finger movement) | Reduced palpable activation (does not provoke finger movement) | No/minimal palpable activation | Asymmetrical activation (describe here): |
| **Jaw** | Jaw opening (assess ability to open mouth fully without resistance) | Normal (full range of opening) | Some restriction but doesn’t impact function | Notable restriction impacting function | Marked restriction heavily impacting function |
| Jaw laterality (assess ability move lower jaw left and right) | Normal (full ROM) | Some restriction | Minimal or no movement |  |
| **Palate●●** | Elevation | Normal | Some (elevates but does not meet pharyngeal wall) | None | Asymmetrical elevation (describe here): |
| Speech nasality | Normal | Mild (hints of nasality or nasal escape) | Moderate (nasality persistently evident throughout) | Severe (notable hypernasality with blending of nasal-non-nasal phonemes) |
| Soft palate function | No collapse of cheeks | Easy collapse under pressure |  |  |
| **Tongue●●** | Protrusion (select up to two responses) | Normal | Some deviation | Marked deviation | Fasciculation’s or wasting |
| Against resistance (push tongue into cheek & against assessors thumb) | Normal (full resistance) | Mild (unable to sustain resistance/easy collapse) | Moderate (can achieve tongue into cheek but easy to push tongue back) | Severe (barely able to achieve tongue into cheek) |
| IOPI score | 51-70 | 35-50 | 20-34 | <20 |
| Wasting | None | Subtle | Evident | Marked |
| **Voice** | Pitch | Normal | Lacks some inflection | No inflection |  |
| Volume | Normal | Lacks some change in volume or stress | No changes in volume and/or stress |  |
| Quality●● | Normal | Not normal | Describe: | |
| **Cough** | Peak flow | >500l/min | 350-500l/min | 270-350l/min | <270l/min\*\* |
| Glottic closure (assess via voice, cough and breath hold) ●● | Good glottic closure | Evidence of impaired glottic closure in voice, cough or breath hold. | Not demonstrating any ability to close glottis. |  |

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| --- | --- | --- | --- | --- | --- |
| **Swallow** | | | | | |
| **Oral**  **/pharyngeal secretions** | Quantity\*\*\* | Normal | Mild (some pooling or reports of pooling in sulci/back of tongue and/or pooled secretions only at night) | Moderate (some drooling & intermittent wet voice at rest) | Severe (constant drooling & persistent wet voice at rest) |
| **On fluid** | Oral spillage | None | Some loss but doesn’t pass beyond border of lips | Loss beyond lips onto chin | Loss beyond chin |
| Range of hyolaryngeal movement● | WNL | No/very little anterior movement | Markedly impaired anterior and superior movement | Bobbing only |
| Abnormalities● | Up to 2 swallows per mouthful | Multiple swallows per mouthful (3 or more) | Reports of fluid sticking | Gurgling/throat-clearing/coughing/altered breathing or other signs aspiration. |
| **On diet** | Chewing | WNL (rapid) | Slow but complete mastication | Slow with suspicion of incomplete mastication before swallowing |  |
| Oral clearance (on diet) | WNL –cleared in one swallow | Slow with repeated swallows | Incomplete spontaneous clearance |  |
| Oral control (on fluids) | Able to hold fluid bolus without issue | Signs of some premature spillage into pharynx pre-swallow | Complete oral spillage into pharynx before swallow. |  |
| Range of hyolaryngeal movement● | WNL | No/very little anterior movement | Markedly impaired anterior and superior movement | Bobbing only |
| Abnormalities● | Up to 2 swallows per mouthful | Multiple swallows per mouthful (3 or more) | Reports of food sticking | Attempts to cough and clear |

\*Ask about dry eye, history of eye infections. Provide basic eye-care advice (e.g. patches/taping/drops) and ensure referral to neuro-ophthalmology.

\*\* Refer to respiratory PT for cough augmentation – provide information re glottic function with this referral (i.e. can the patient audibly close their glottis in speech, cough & breath-hold)

\*\*\* Check secretion tolerance at night. If regularly coughing on secretions when lying flat – sleep propped up or on the side. NB Usual secretion management drugs may be out of bound for certain disease subtypes due to interactions with the heart.

● Use this information to build an impression about patients’ pharyngeal sensation.

●● Use this information to liaise with the respiratory PT re suitability for various cough augmentation devices (significant spasticity and/or flaccidity impacts tolerance and/or ability to access certain cough augmentation approaches)

**List of abbreviations:**

A&E – Accident and Emergency

ABGs – Arterial Blood Gases

ACBT – Active Cycle of Breathing Techniques

ADT – Assistive Devices and Technologies

ANSN – Adult North Star Network

CBGs – Capillary Blood Gases

CCGs – Clinical Commissioning Groups

COPM – Canadian Occupational Performance Measure

CHC – Continuing Health Care

DMD – Duchenne Muscular Dystrophy

ECCD – Elbow Contracture Control Devices

ECG – Electrocardiogram

FEES – Fibreoptic Endoscopic Evaluation of Swallowing

FEV1 – Forced Expiratory Volume in 1 second

FVC – Forced Vital Capacity

HR – Heart Rate

HFCWO – High Frequency Chest Wall Oscillations

HFCWC – High Frequency Chest Wall Compression

IPAP – Inspiratory Positive Airway Pressure

IPPB – Intermittent Positive Pressure Breathing

IPV – Intrapulmonary Percussive Ventilation

ITB – Iliotibial Band

KAFOs – Knee Ankle Foot Orthoses

LVR bag – Lung Volume Recruitment bag

MDT – Multi-Disciplinary Team

MDT-PD – Munich Dysphagia Test – Parkinson’s Disease

MEP – Maximal Expiratory Pressure

MHR – Maximal Heart Rate

MIC-VC – Maximum Insufflation Capacity – Vital Capacity

MI:E – Mechanical Insufflation Exsufflation; cough assist

MIP – Maximal Inspiratory Pressure

NG tube – Nasogastric tube

NIV – Non-Invasive Ventilation

NJ tube – Nasojejunal tube

OPD – Oro-pharyngeal Dysphagia

OT – Occupational Therapy

PCF – Peak Cough Flow

PEG – Percutaneous Endoscopic Gastrostomy

PT – Physiotherapy

PUL – Performance of Upper Limb

ROM – Range of Movement

SNIP – Sniff Nasal Inspiratory Pressure

SpO2 – oxygen saturation

SLT – Speech and Language Therapy/Therapist

SOC – Standards of Care

SSQ – Sydney Swallow Questionnaire

TOSCA – sleep study with transcutaneous CO2 and O2 monitoring

VFSS – Videofluoroscopic Swallow Studies

VO2max – maximal oxygen uptake

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