DUCHEENNE MUSCULAR DYSTROPHY
SCOTTISH
MULTI DISCIPLINARY CARE PATHWAY

Scottish Muscle Network
This care pathway has been developed to support the multi-disciplinary care of boys and young men with Duchenne Muscular Dystrophy in Scotland. The care pathway has been developed from the model of the DMD Scottish Physiotherapy Profile (Journal of the Association of Paediatric Chartered Physiotherapists, 2007) and we are very grateful to the physiotherapy network for all the work they have done in laying the foundations and template design for the multi-disciplinary care pathway.

This care pathway is intended to be used as a guideline in the management of DMD, in a format which is easy to use, by all members of the multi-disciplinary team, in a clinical setting. It is not intended to be prescriptive of management in specialist areas, but rather as an overview of interventions at key stages in the progress of this condition. Further guidance on the multi-disciplinary management of DMD is now available, and this care pathway is best read in conjunction with this- "international TREAT NMD care standards for management of DMD"

The pathway has been written with support from the following working and advisory groups, to whom we are very grateful.

Scottish Neuromuscular Physiotherapy Group
Scottish Muscle Network Paediatric Subgroup
Scottish Paediatric Cardiologists
Scottish Paediatric Respiratory Interest Group
Scottish National Spinal Deformity Services
Scottish Adult Home Ventilation Services
Scottish Muscle Network Transitional Care Subgroup
Children’s Hospice Association Scotland
Scottish Paediatric Endocrinologists

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STAGES

In considering the care pathway, assessments and interventions it is helpful to consider stages of DMD.

Stage 1 Early/pre-symptomatic
This may occur when diagnosis has been made incidentally, for example diagnostic screening for boys presenting with developmental delay or in families with a positive family history. Being given a new diagnosis like this is devastating for families, sensitive counselling and support at this time can influence the family’s ability to manage in the future.

Stage 2 Early ambulant
In this stage boys may present with motor difficulties for example toe walking, or difficulty with activities such as running, hopping and stairs. Later boys may have difficulty rising from the floor (Gower’s manoeuvre). Interventions at this stage are aimed at maintaining mobility and preventing secondary complications.

Stage 3 Late Ambulant/going off feet
Mobility is increasingly difficult. Boys have difficulty managing stairs and walking distances. They may be prone to falls and have difficulty getting back up on their feet without assistance. Long bone fractures can occur, and may lead to loss of mobility. Loss of ambulation can be predicted and anticipatory planning is needed.

Stage 4 Early non-ambulant
In this stage boys are able to sit independently but have lost the ability to walk. In addition to wheelchair provision to aid independent mobility, good postural management is required with particular attention to the increased risk of scoliosis.

Stage 5 Late non-ambulant
In this stage boys have difficulty maintaining posture, and upper limb function is affected, impacting on other activities of daily living. Ability to feed and possible difficulties with oro-motor function can lead to nutritional problems. Respiratory and cardiac functions are more likely to be impaired, and require interventions. Adolescence and transition to adult services, further education and employment require careful planning and co-ordination.

Stage 6 Palliative Care/ End of Life
Young men with DMD do face living with a life-limiting condition. Planning for end of life care should be in place when it becomes appropriate.
<table>
<thead>
<tr>
<th>Difficulties Presenting</th>
<th>Diagnosis/ Paediatrician</th>
<th>After diagnosis</th>
<th>Genetics</th>
<th>Physiotherapy</th>
<th>Orthotics</th>
<th>Equipment/ OT</th>
<th>Social Services</th>
<th>Education</th>
</tr>
</thead>
<tbody>
<tr>
<td>Speech / Global developmental Delay</td>
<td>Referral to Paediatric Neurologist and/or Community Paediatrician for assessment and investigation</td>
<td>Information should be given both in verbal and in written form</td>
<td>Information about genetic basis for diagnosis</td>
<td>Encourage activities as able, consider referral to wheelchair services for reinforced buggy/light wheelchair if mobility impaired</td>
<td>For foot variations such as pes planus (flat feet) it may be useful to refer to an orthotist for insoles / inlays</td>
<td>Equipment provision not usually required at this stage</td>
<td>Information about DLA - Mobility component from age 3</td>
<td>Referral to local pre-school inter agency planning group</td>
</tr>
<tr>
<td>Muscle weakness (particularly proximal) presenting with gross motor difficulties eg waddling gait, running, hopping, stairs</td>
<td>Investigations include: CK Genetic testing for mutation analysis Consideration of muscle biopsy if genetics are negative</td>
<td>Family tree and risk of carrier status determined</td>
<td>Avoid muscle strengthening/ and eccentric exercise (it is advisable to avoid strengthening exercises and eccentric muscle work as this can cause further damage to the muscle cell membrane).</td>
<td>Testing for carrier status and counselling of results, including cardiomyopathy screening of carriers</td>
<td>Night splints may be appropriate if loss of range of dorsiflexion is noted</td>
<td>Consider access issues with physiotherapy eg home and nursery/school</td>
<td>Consider accessibility issues (OT and PT)</td>
<td>Referral to pre-school home teaching services/educational psychology if cognitive developmental concerns</td>
</tr>
<tr>
<td>toe walking (tight TAs)</td>
<td>Referral to Genetic clinic Occupational therapist SALT if language delay</td>
<td>Information about support groups</td>
<td>Testing for carrier status and counselling of results, including cardiomyopathy screening of carriers</td>
<td>Other family members at risk identified and offered counselling</td>
<td>Other family members at risk identified and offered counselling</td>
<td>Referral to social work OT if housing issues</td>
<td>For nursery/school information about diagnosis would be helpful</td>
<td>Transport to school may be helpful</td>
</tr>
<tr>
<td>Boys will rise from the floor via a prone position using their hands to “walk” up their body. (Gower’s manoeuvre)</td>
<td>Referral to Genetic clinic Occupational therapist SALT if language delay</td>
<td>Information about other local support groups</td>
<td>Risk for siblings discussed and offered future genetic counselling when appropriate</td>
<td>Introduce and advise on stretches for tight muscle groups and joints</td>
<td>Introduce and advise on stretches for tight muscle groups and joints</td>
<td>Referral to/information given about social work services for family assessment if need for additional support services</td>
<td>Additional support may be required in educational environment</td>
<td>Additional support may be required in educational environment</td>
</tr>
</tbody>
</table>

*Information about CSP (co-ordinated support plan) to family

| Information about DLA - Mobility component from age 3 | Blue badge for parking |
| Free nappies from age 4 if continence issues (Health visitor) | Referral to social work OT if housing issues |

Information about CSP (co-ordinated support plan) to family
<table>
<thead>
<tr>
<th>Description of difficulties</th>
<th>Paediatric</th>
<th>Respiratory</th>
<th>Cardiovascular</th>
<th>Physiotherapy</th>
<th>Orthotic/Orthopaedic</th>
<th>Equipment Provision</th>
<th>Support Services</th>
<th>Education</th>
</tr>
</thead>
</table>
| Motor skills affected by progressive muscle weakness eg difficulty with jump / run / hop and frequent falls. Difficulty with stairs and rising from chairs. Changes to gait with lumbar lordosis and waddling/trendelenberg gait. | Discuss role and prescribing regimes of steroids in prolonging ambulation (refer to North star consensus review doc)  
*Steroid Parent information leaflet given  
Issue steroid care card and information leaflet in event of intercurrent illness  
If starting steroids:  
*Check vit D calcium and bone studies if starting steroids. Consider dexa scan baseline and annual/bi-annual screening according to current guidelines consider vit D3 supplement 400-800 IU daily  
Check varicella titres and offer immunisation if negative  
Assess TB risk and offer BCG if appropriate  
Assess growth and BMI.  
*Healthy diet information given  
Recommend family attend local orthoptist annually for cataract screening  
*6 monthly paediatric review for side-effects (BP, growth, behaviour, urinalysis for glucose | Ensure immunisations are up to date,  
Annual influenza vaccination recommended  
Consider pneumococcal vaccination if not already given  
Baseline lung function tests (Annual FVC/FEV1) | *Information leaflet re: cardiac assessment given to families  
ECG and echocardiography biannual after age 6- screening for cardiomyopathy in ambulant boys with DMD  
Blood pressure monitoring at paediatric clinic | Liaise with Lead Consultant and NM physiotherapist re steroid therapy and 6 monthly North Star Ambulatory Assessment (modified version for centres not signed up NS project)  
Encourage activities e.g. swimming as able  
Avoid strengthening exercise especially eccentric muscle work  
Continuation of stretches appropriate to stage of progression (refer to Scottish Physiotherapy profile for specific physiotherapy relating to stage of the condition) | Night orthoses for gastrocnemius when ankle range of movement is compromised  
Continuation with inlays / insoles if appropriate  
Walking AFO’s not usually recommended as this can cause a deterioration in standing/ walking ability  
Night orthoses may be beneficial  
Refer to MDC wheelchair and seating guidelines  
Specialised seating and equipment in school may be beneficial  
Refer to MDC booklet  
Orthotic/Orthopaedic consultant  
Night orthoses | Around age 5 years if mobility/fatigue/ falling is problematic  
Encourage use of lightweight manual wheelchair rather than buggy  
*refer to MDC wheelchair and seating guidelines  
Walking AFO’s recommended as this can cause a deterioration in standing/ walking ability  
Night orthoses may be beneficial  
Refer to MDC wheelchair and seating guidelines | *MDC & Family care advisor/ Action Duchenne/ Information on SMN family events & patient group  
Social services  
Social work O.T. (Housing and adaptations)  
Local sleep counselling or *Sleep Scotland if problems with sleeping through the night  
Support through school, clinical psychology or CAMHS if concern about behaviour or mental health | Raise awareness of associated developmental and learning difficulties, may be global or specific, increased risk of SLD including ASD and ADHD | Increased risk of mental health and emotional difficulties  
May need additional support for learning – *Action Duchenne Learning and behaviour toolkit which can be downloaded  
Consider access to curriculum and accessibility of classrooms and facilities |

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## Stage 3 – Going off feet

<table>
<thead>
<tr>
<th>Difficulties Description</th>
<th>Paediatrician</th>
<th>Respiratory</th>
<th>Cardiac</th>
<th>Physiotherapy</th>
<th>Orthotic/orthopaedic bone health</th>
<th>Equipment and postural management</th>
<th>Support Services</th>
<th>Education</th>
</tr>
</thead>
<tbody>
<tr>
<td>Increased effort and time to rise from chair/floor. May be unable to rise without support.</td>
<td>Paediatrician maintains overview of all medical care</td>
<td>Monitor FVC annually while above 50% then 6monthly</td>
<td>Information leaflet re: cardiac assessment given to families*</td>
<td>Continuation of stretches/passive movements and exercises as able</td>
<td>Night AFO’s if child is still compliant</td>
<td>Manual wheelchair with supportive cushion and backrest.</td>
<td>Social services support at home/respite care as above</td>
<td>As above</td>
</tr>
<tr>
<td>Increasing frequency of falls</td>
<td>Discussion about treatment plan for continuation of steroids if prescribed. Monitoring for side-effects as above</td>
<td>Ensure immunisations up to date</td>
<td>ECG and echo biannual screening for cardiomyopathy From age 6 whilst ambulant</td>
<td>Hydrotherapy if available</td>
<td>Consider daytime AFOs if sitting in wheelchair for long periods of time</td>
<td>Referral for powered wheelchair. It may be appropriate to consider a powered chair with advanced functions (see MDC booklet on wheelchairs and seating).</td>
<td>Motability (adapted vehicle should be considered at this stage).</td>
<td>Moving and handling review as per local policy (both at home and school)</td>
</tr>
<tr>
<td>Requires a wide base of support</td>
<td>Discuss need for spinal and respiratory assessment on loss of ambulation</td>
<td>Encourage annual influenza vaccination and pneumococcal vaccination if not already</td>
<td>6 monthly North Star Ambulatory Assessment (NSAA)</td>
<td>6 monthly North Star Ambulatory Assessment (NSAA)</td>
<td>Referral to local orthopaedic surgeon if contractures present may need future surgery</td>
<td>Introduce standing frame if appropriate</td>
<td>Blue Badge for parking</td>
<td>Raise awareness of need for hoisting</td>
</tr>
<tr>
<td>Difficulty standing with heels down</td>
<td>Discussion of emotional and mental status.</td>
<td>Blood pressure monitoring at paediatric clinic if still on steroids</td>
<td>Continue to monitor lower limb contractures as surgery may be indicated once non-ambulant</td>
<td>Risk of long bone fracture increased, often associated with loss of continuing ambulation.</td>
<td>Risk of long bone fracture increased, often associated with loss of continuing ambulation.</td>
<td>Maintain mobility/standing ability within a standing frame/wheelchair if appropriate.</td>
<td>Safety risk of stairs, housing needs to be accessible and adapted for wheelchair</td>
<td>Postural support within the classroom and increased assistance may be required for toileting</td>
</tr>
<tr>
<td>Difficulty standing still for &gt;3s</td>
<td>Discuss transitional services and introduce yellow folder.</td>
<td>Games to promote effective in – expiration eg wind instruments, Blowing bubbles, Incentive spirometer</td>
<td>Inspiratory muscle training not recommended as it requires resistance</td>
<td>Maintain mobility/standing ability within a standing frame/wheelchair if appropriate.</td>
<td>Referral/discussion re: dxa scan in event of low impact fracture, and bisphosphonates if low bone mineral density present</td>
<td>Familiarisation with hoisting techniques</td>
<td>Wheelchair accessible transport to school</td>
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<tr>
<td>Tires easily with physical activity</td>
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<tr>
<td>Increased upper limb weakness</td>
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<td>Asymmetry noted in standing and sitting</td>
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End of Life Care

DMD is a progressive disease and life expectancy is limited. It is difficult to pinpoint when someone is reaching end stages of their life, but at some point everyone will.

With this in mind, the opportunity needs to be taken to talk about end of life care, to plan ahead about the things that are important to the young person and their loved ones, before and around the time of death. A review of the young person’s wishes, priorities and needs should be undertaken with their active participation, and that of their family and loved ones, at this time.

Whilst practitioners working within children’s and adult’s palliative care services will assist with end of life and bereavement care, a number of tools are available e.g. the Anticipatory Care Planning tool (Living and Dying Well). This can help any staff working with the young person at this sensitive time to engage and involve them in reviewing their wishes and those of their family and loved ones, and to draw up an end of life plan.

Use of the Together for Short Lives Core and Transitional Care Pathways can help ensure good care planning and managing of end of life needs.

District Nurses and GPs are familiar with the Gold Standards Framework, and out of hours palliative care summary systems that assist these services to support and provide effective end of life care at home if this is the young person’s wishes.
1. MDC booklet - Introductory guide for families with a child newly diagnosed with Duchenne Muscular Dystrophy
2. MDC website- www.musculardystrophyuk.org
3. Action Duchenne website - www.actionduchenne.org
4. Scottish Muscle network website- www.smn.scot.nhs.uk
5. Scottish Muscle network- “some useful things to know about DMD” leaflet available on website
6. Scottish Muscle network- DMD care card leaflet available on website
8. Steroids and duchenne muscular dystrophy- FAQs- available on MDC website (appendix 2)
9. Healthy eating for children with NM conditions- available on MDC website (appendix 3)
10. North star project - monitoring for side-effects of steroids (appendix 1)
11. Scottish Muscle Network - guideline for bone management in DMD (appendix 1)
12. Wheelchair provision for children and adults with muscular dystrophy and other neuromuscular disorders www.musculardystrophyuk.org
13. Sleep Scotland- www.sleepscotland.org
14. Learning and Behaviour Toolkit- action Duchenne website www.actionduchenne.org
15. Physiotherapy management for boys with DMD- available on website www.musculardystrophyuk.org
16. Scottish Muscle network leaflet “some useful things to know about transition” available on SMN website www.smn.scot.nhs.uk
17. TREAT-NMD DMD Care Standards www.treat-nmd.eu/care/dmd/dmd-care
Appendix 1

THE USE OF STEROIDS IN DUCHENNE MUSULAR DYSTROPHY

CONSENSUS BEST PRACTICE 2005

Recommendations
These recommendations are based on review of the existing literature and clinical experience of the members of the North Star Network for paediatric neuromuscular disease management.

The decision to start glucocorticoid treatment should be made by a physician experienced in management of DMD, after a full discussion with the affected child and the parents. The arrangements for long term follow up should be made at a clinic / centre with medical and physiotherapy facilities for monitoring of disease activity and glucocorticoid therapy are available.

Which Glucocorticoid?
Prednisolone: We consider Prednisolone to be the glucocorticoid of choice in view of our familiarity and experience in the UK studies and clinical practice, its easy availability and low cost, and the high incidence of cataracts reported with the use of deflazacort. Prednesol 5mg water-soluble tablets may have the benefit of ease of administration and less gastrointestinal side effects.
Deflazacort: Deflazacort appears to have less weight gain side effects. Randomized controlled trial to compare deflazacort and prednisolone for their adverse effects is in process. Deflazacort may be a consideration for prednisolone treated boys who have excessive weight gain which is unresponsive to dietary / exercise / dose adjustments.

What dose?
The starting dose is as follows:
Prednisolone 0.75 mg/kg/day, (rounded off to the nearest 2.5 mg e.g. 10, 12.5, 15, 17.5 or 20 mg per day), to a maximum of 40 mg/day.

In glucocorticoid equivalence, 1 mg prednisolone = 1.2 mg deflazacort
0.75 mg prednisolone = 0.9 mg deflazacort

Which regime?
The preceding text explains why there is no overwhelming consensus on which to base didactic recommendations on continuous (daily) prednisolone vs an intermittent (repetitive cycles of 10 days on, 10 days off prednisolone) regime.

In view of these facts a reasonable option is to explain our current understanding of the pros and cons of these two treatments to each family and allow them to make an informed consent decision on whether to opt for the daily regime or the intermittent regime. An information sheet highlighting the two regimes is available
Contraindications to the use of prednisolone in DMD
Hypertension, diabetes mellitus, gastric / peptic ulceration are relative contraindications.

Use of Prednisolone in DMD
The following recommendations for prednisolone therapy have been customised for specific age (and functional) groups of boys with DMD because of the variability in the therapeutic response and the side effects in these groups. A randomised trial of glucocorticoids in young vs older DMD patients has not been published but analysis of subgroup of patients studied by Taylor suggests a more significant response in the younger patients (Dubowitz 1997). This was further highlighted by the recent report by Dubowitz et al (2002) and other unpublished data from the group of Angelini. On the other hand, the long term cumulative side effects of steroids initiated at an early age in DMD have not been systematically assessed in large studies. The age specific recommendations attempt to get the best benefit vs risk ratio at the various disease stages.

"Presymptomatic" phase (age range 0 - 3 years)
This group comprises of boys who are diagnosed early on the basis of family history or following incidental discovery of high CK or ALT/AST in the course of investigating other medical problems. There may be a theoretical advantage in starting glucocorticoid treatment early, prior to major muscle cell necrosis and replacement by fibrosis. With the evidence available however, we are reluctant to expose these young boys to the long term side effects of steroid treatment at a relatively asymptomatic stage and therefore do not recommend routine use of prednisolone in this group.

Early ambulant phase (4 - 7 years)
The option of glucocorticoid therapy should be offered to all families in this age group being followed up at centres with expertise and facilities for long term monitoring of efficacy of treatment and side effects. If the possible side effect profile is acceptable to the parents / child and physician, treatment may be initiated in the context of a nationwide program with regular audit to enable improvements in clinical practice.

Late ambulant phase (7 years till loss of walking)
These are boys who often have lost the ability to rise from the floor and their walking is deteriorating gradually or rapidly, with impending loss of ambulation. Glucocorticoids should also be offered at this stage, provided the adverse effect
profile is acceptable to the family and the physician. It has to be appreciated that the benefit of starting glucocorticoids, from the mobility perspective, at this age might be limited. The decision regarding treatment entails a full discussion with the family and the need for monitoring facilities as above.

**Constant wheel chair dependent phase**
There is little data, in this group, to support the initiation of glucocorticoid therapy, and at this stage, the adverse effects may out-weight potential benefits. In view of the reported long term improvement of respiratory function (Biggar 2002, Alman 2004), pilot studies to demonstrate the effect of glucocorticoids on respiratory muscles are being considered

**When to discontinue glucocorticoid treatment.**
- The functional and clinical benefit is almost always obvious within 6 months treatment. If there is no improvement / stabilisation over the first 6 months of treatment, glucocorticoid therapy may be discontinued.
- The presence of side effects unacceptable to the child / parents / physician
- Loss of ambulation. In this situation, the risks and side effects of ongoing treatment may outweigh the potential benefits. However, in view of the possible benefit to arm and respiratory function, this policy needs to be kept under review and under discussion with families.

3. **Protocol of monitoring - "Good practice"**

**Baseline assessment**
Baseline assessment should include the following
Full physiotherapy assessment
urine dipstix testing
full blood count (FBC),
random glucose,
electrolytes urea, creatinine
25OH Vit D levels
varicella antibodies,
bone mineral density assessment by Dual X-ray Absorptiometery (DXA) scan is desirable (See further under "strategy for prevention of osteoporosis").

**Dietary advice.** Referral to the dietician at initiation of prednisolone should be made. Emphasis on plans to prevent excessive weight gain and maintenance of adequate Vit D and Calcium intake is recommended.

**Screening for Tuberculosis:** Family history of tuberculosis (Tb) should be sought from all at the baseline assessment. If there is history of active Tb in a family member, this would be a contra-indication for steroid use, till the affected member has been fully treated, the boy has received the appropriate screening prophylaxis and infectious diseases specialist's review and advice. Boys from communities with high incidence of Tb (eg recent immigrants from developing world countries) should be offered Mantoux testing and BCG as indicated, with

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parental consent prior to joining the trial; such children should not be started on prednisolone until 4 months following the BCG vaccination.

**Varicella vaccination** should be offered to boys who are susceptible as evidenced by varicella antibody negative status. This will prevent routine future need for Zoster Immunoglobulin (ZIG) and acyclovir on exposure of these boys to chickenpox.

If the boy is coming up to school starting age, consideration may be given to expedite the **MMR pre-school booster** ahead of starting the glucocorticosteroid. MMR pre-school booster can be given from 3.5 years of age onwards.

**Progression of disease activity / response to treatment**
Regular assessments should be done at 3 months and then six monthly or more frequently if indicated in a particular patient.

Physiotherapy assessment (according to North Star assessment protocol)
- MRC score
- Timed Gowers’ manoeuvre (starting from the cross legged sitting position)
- 10 metres walking times
- Motor ability score
- Measurement of contractures
- Myometry of 4 muscle groups (elbow flexion; grip; knee flexion; knee extension)

Forced vital capacity (FVC), as soon as the boys are able to co-operate with the technique.

**Monitoring of possible side effects at each assessment – (Table 3)**
- Weight plotted on Child Growth Foundation Chart
- Standing height
- BP
- Urine dipstix testing for glucose
- Cushingoid features
- Mood / behavioural / personality / GI / skin changes
- Screening for cataracts with red reflex
- Record any bone fractures
- Record any intercurrent infections

**Specialised assessments to include**
Ophthalmologic review for cataracts is not recommended as routine, as the huge majority of steroid induced cataracts have been asymptomatic. Red reflex should be screened for on each assessment. Visual acuity should be tested annually, and this can be done by the opticians)

**Echocardiogram** to be performed, as clinically indicated, before any surgery and at time of loss of ambulation. Please note the ENMC workshop recommendations
<table>
<thead>
<tr>
<th>Adverse event group</th>
<th>Measure</th>
<th>Prophylactic measures</th>
<th>Events as criterion for dose reduction</th>
<th>Events as criterion for drug withdrawal</th>
<th>Specific comments</th>
</tr>
</thead>
<tbody>
<tr>
<td>Behaviour changes</td>
<td>Parental reporting</td>
<td>Advice on behaviour modification</td>
<td>Behaviour changes disrupting family/ school life</td>
<td>Severe behaviour changes disrupting family/ school life</td>
<td></td>
</tr>
<tr>
<td>Weight</td>
<td>Weight for age/ height/ BMI</td>
<td>Dietary advice</td>
<td>25% or 3 centile increase from baseline</td>
<td>Weight gain unacceptable to child/ family despite dietetic input/ dose reduction</td>
<td></td>
</tr>
<tr>
<td>Height</td>
<td>Standing height or arm span in non ambulatory</td>
<td></td>
<td>Failure to gain height that is unacceptable to child/ family</td>
<td>Failure to gain height that is unacceptable to child/ family despite dose reduction</td>
<td></td>
</tr>
<tr>
<td>Bone Density</td>
<td>DEXA annually if available, recording of fracture history</td>
<td>Dietary advice about Ca &amp; Vit D, sunshine, exercise</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Glucose tolerance</td>
<td>Glycosuria</td>
<td>Dietary advice</td>
<td>Fasting blood sugar &gt;110 &lt;126mg/dl after dietary modification or blood glucose two hours after meal &gt;140&lt;200mg/dl</td>
<td>Diabetes mellitus as defined as fasting blood sugar &gt;126 mg/dl or blood glucose 2 hours after a meal &lt;200mg/dl</td>
<td>Blood glucose to be checked if glycosuria present on urinalysis</td>
</tr>
<tr>
<td>Blood pressure</td>
<td>Blood pressure compared to age norms,</td>
<td>Advice about dietary sodium intake</td>
<td>Consistent increase in systolic blood pressure 15mmHg over the 97th centile or diastolic blood pressure of 10mmHg over 97th centile for age after sodium restriction</td>
<td>Confirmed hypertension as defined as an increase in systolic blood pressure of 15-30mmHg over the 97th centile or diastolic blood pressure increased 10-30mmHg over 97th centile for height</td>
<td></td>
</tr>
<tr>
<td>Immune/ adrenal suppression</td>
<td>History of infection.</td>
<td>Ensure varicella zoster immunity. Advise on steroid cover for surgery/ injury</td>
<td>Unusually high frequency of infection/ unusual organisms- seek guidance from immunology expert.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Gastrointestinal symptoms</td>
<td>Parental reporting</td>
<td>Advise to avoid NSAIDs</td>
<td>Persistent GI symptoms despite treatment</td>
<td>Abdominal pain/ peptic ulceration- treat with gaviscon, zantac</td>
<td></td>
</tr>
<tr>
<td>Cataract</td>
<td>Optician's check yearly for visual acuity, cataracts and intraocular pressure</td>
<td></td>
<td></td>
<td>Cataracts- if symptomatic, surgery. Increased IOP- follow ophthalmological advice</td>
<td></td>
</tr>
<tr>
<td>Skin changes</td>
<td>Atrophy, easy bruising, fragility, striae, cutaneous/ oral infections</td>
<td></td>
<td></td>
<td>Treat infections as indicated</td>
<td></td>
</tr>
</tbody>
</table>

**Table 3. Managing steroid side-effects / adverse events**
of echocardiogram at diagnosis, before any surgery, every two years till 10 years of age and then annually (Bushby et al 2003)

Management of side effects – Please refer to tables 3

**Strategy for prevention of osteoporosis**

This issue was discussed in detail in an MDC sponsored workshop (Quinlivan 2005).

The key aspects recognized were as follows:

- Three published studies have demonstrated that in boys with DMD, the bone mineral density as measured by DEXA scanning, is lower than normal for age (Larson 2000, Aparicio 2002, Bianchi 2003).
- The high fracture incidence in DMD, ranging from 20 - 40 % is well recognised. In-patients not treated with glucocorticoids these fractures are almost exclusively in the limbs, and spare the vertebrae.
- Nutritional aspects for prevention of osteopenia / osteoporosis are important. One study (Bianchi 2003) documented low 25 OH Vit D levels in boys with DMD. Checking for this at diagnosis and treatment if indicated is recommended
- With long term glucocorticoid use in long non-randomised studies, a high incidence of vertebral fractures has been reported (Bothwell 2003)
- Long term steroid use, with cumulative doses of glucocorticoid, inevitably leads to trabecular bone loss, osteoporosis, and consequent vertebral fractures. The effect on long bones is not as marked.
- DEXA scan measured bone density scores have been used in adults for WHO definition of osteoporosis, but these criteria cannot be applied in children.
- The pitfalls of DEXA scanning, and the need to interpret DEXA bone density Z scores in context of child's age, height, weight (Fewtrell 2003) emphasized.
- "Given the difficulties discussed, a child may most appropriately act as his or her own control, with serial (DEXA) scans to monitor progress" (Fewtrell 2003)
- At present, in the paediatric age group, the precise correlation / threshold (if any) of Bone density Z scores and risk of vertebral fractures is not known. DEXA scan, therefore, in an individual boy with DMD not predict the vertebral fracture. Serial DEXA scans in the individual patient can identify trends of change, and may be used to monitor the effect of specific treatment.
- In patients on Glucocorticoid treatment, there is no evidence base to demonstrate prevention of osteoporosis with the routine use of Ca or Vit D. (This will obviously not be applicable to the individual patient who is Ca or Vit D deficient)
- There is not enough paediatric data to support routine clinical prophylactic use of bisphosphonates in boys with DMD on glucocorticoids.
- Glucocorticoid therapy induced osteoporotic vertebral fractures can be treated effectively with IV pamidronate treatment, and the anticipated duration of treatment is usually 1 year
- Dr Quinlivan is the paediatric lead for submitting a grant proposal for a study of bone density and effects of bisphosphonate treatment in DMD.
- The clinical role of DEXA scanning in DMD and it's glucocorticoid treatment is still being clarified. Acquisition of data and further studies in this area may allow us to better delineate the clinical and prognostic value of DEXA scans over the coming years.
- DEXA scans of children should be interpreted by or in collaboration with centres with a clinical team with a specific interest and expertise in bone densitometry in children, especially if the scan results are to be used to start or change treatment.

The North Star Network will review the osteoporosis assessment and prevention policy in future with the further availability of data.

Approved July 2015
Review July 2018
As of now, the following are recommended

- Ca and Vit D intake dietary assessment at diagnosis and initiation of Glucocorticoids
- 25OH Vit D levels check at diagnosis / initiation of glucocorticoids
- DEXA scan is desirable at starting glucocorticoids
- In DMD boys on glucocorticoid treatment, lateral XR of thoraco lumbar spine and DEXA scan will be indicated for
  - back pain
  - spinal deformity
  - loss of height
- Serial DEXA scans may be indicated to monitor patients with vertebral fractures on bisphosphonate treatment.
- Please ensure that the DXA scan software uses age appropriate normative bone density data from paediatric population to define normal ranges and to calculate z scores, and that any abnormal results are discussed with the regional "Bone metabolism expert"

Management of Osteoporosis / Fractures
Single long bone fracture is not considered an indication for DEXA scan or discontinuation of glucocorticoids.
Recurrent long bone fractures or a single vertebral fracture will need to be discussed with the "bone metabolism specialist" / endocrinologist. Effective treatment with IV palmidronate is available. The decision to continue/discontinue prednisolone depends on the quantum of benefit to that child, and his and the parental wishes.

4. **Data collection and maintenance of database**
It is important to systematically collect sequential data on disease progression and the effects and side effects of prednisolone treatment and maintain a database of all affected boys at the local neuromuscular centres. This is essential to facilitate audit and research and improve clinical practice. The North Star network project co-ordinator will be instrumental in this regard.
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Committee on Safety of Medicines, Medicines Control Agency (1998). Calcort (Deflazacort): advertising has been withdrawn. Curr Prob Pharmacovigilance, 24:10


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FASEB J. 2005 Feb 25; [Epub ahead of print]


Mendell JR, Moxley RT, Griggs RC. et al (1989). Randomized controlled trial of prednisolone in Duchenne's muscular dystrophy. NEJM 320:1592-1597,

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Review July 2018
Steroids and Duchenne Muscular Dystrophy (DMD)

This leaflet has been produced to help you think about whether your child with Duchenne muscular dystrophy should have steroids or not. It includes some of the main questions people have asked when in this situation and also gives you some further information to look up. It is not intended to replace discussion with the muscle clinic team, so feel free to ask them any questions you may have after reading this.

**Why are steroids used in DMD?**
It has been known for a number of years now that steroids have an effect on muscle strength in DMD. If they are used in boys who are still walking, they *may* have an effect on stabilising or even improving muscle strength for a period of time. Not all boys respond to steroids and the way that steroids have this effect on slowing the dystrophic process is not known.

**What steroids are used?**
The main steroid that is used is called prednisolone (prednisone in the USA). Deflazacort is also used in some countries. These are not “anabolic steroids” which is what athletes use illegally to build up muscle- these do not have an effect in DMD.

**What are the possible beneficial effects?**
The studies which are now being reported on steroids are showing that overall boys who are treated with steroids walk for longer than those who are not. This effect varies from child to child but there are some studies coming out which are showing that some boys may carry on walking for years longer than they otherwise would. These children also seem to develop other complications of the condition less frequently too – for example they may develop breathing difficulties later than they might have and have fewer problems with their spines.

**What are the possible risks?**
The down side of steroid treatment, and the reason that people are still very cautious about using them, is that they may have side effects. Steroids have many side effects, but the chances of getting these vary from person to person, and on the dosage of the regime used. The most common side effects reported in the studies of using steroids in DMD in the short term are weight gain and mood changes. Weight gain seems to be most of a problem just at the time that the boys start taking the steroids, so it is a really good idea to keep a close eye on food intake at that time to avoid running into problems. If you would like further information on this, please ask the muscle team or your local dietician in the longer term (after many years of treatment) there may be growth suppression, the development of cataracts and thinning of the bones. The risk of these long-term effects cannot be measured. You should discuss these fully with your doctor. There are whole lists of other possible side effects, which include raised blood pressure, diabetes, thinning of the skin and poor wound healing and increased susceptibility to infection. A rare side effect of taking steroids in DMD is stomach irritation. It is important not to take non steroidal anti inflammatory tablets or medicines like neurofen or aspirin while you are on steroids. If your son develops tummy pain, or there is any sign of bleeding, you should contact your GP. This may not mean that they have to stop taking the steroids but it is important that this is noted. It is also very important that steroids are not
stopped suddenly but tapered off as the body becomes used to their effect and needs time to adjust if they are withdrawn.

**How do the benefits and risks balance out?**

This is a difficult question. The reason that steroids are often used is because the studies which are coming out now are showing some significant benefits. In some studies some boys with DMD are still walking at the ages of 14 or 15. But there is no doubt that there can be important side effects, and the worst of these include significant growth delay and weak bones. The muscle team aim to minimise the risk of side effects by checking for them when you go to clinic. If significant side effects were picked up then the dose of the steroids could be altered, or tapered off completely. This may also be done if it seemed like they were not having a positive effect. It is important to realise that if the decision to use steroids is made this is not an ‘all or nothing’ thing, but that the regime would be carefully worked out on an individual basis and changed if required. It is possible to change your mind at any time, though you must realise that steroids cannot be stopped suddenly.

**What dose would be used and how often?**

There are two alternatives. All of the studies that have shown a useful benefit of steroids have given them on a daily basis. However, it is possible that using steroids intermittently (i.e. not all the time) could be effective while reducing the risk of side effects. If you were particularly worried about the side effects this might be a good alternative. However, it has not been proven in controlled studies that this is as effective as using steroids every day.

**What happens if we decide to use steroids?**

If the decision to use steroids is made, then the muscle team will need to be keeping a close eye on your son and organise more tests than would usually be done. It will be very important that you attend these appointments. Your son will need to have some blood and urine tests to check everything is OK to use the steroids, and to check that he is immune to chicken pox (if not he will need to be immunised against it as chicken pox can be very serious in children who are on steroids). He will also need a scan of his bones and an eye check (these will need to be repeated at intervals while he is on the steroids), and a good assessment of how he is getting on at the moment.

Once the blood test results are back, and providing they are all OK, your GP will be asked to prescribe the steroids and you should be seen by your muscle team approximately three months after. At this appointment the team will check for any side effects and see if there is any difference in your son’s physical performance and discuss with you if the dose seems right. He will then be seen as usual at six monthly intervals, though the GP will be asked to check his blood pressure and his urine for sugar in between appointments as well.

At every appointment, you can discuss the dose etc and what you want the plans for the future to be. It is possible that if your son responds well to the steroids that he will need to be on them for a long time. New information is becoming available every year about the long term use of steroids in DMD, and this should be fed back to you at the clinic.

**What happens if we decide not to use steroids?**
Your son will continue to be followed-up in the usual way and the muscle team will discuss with you all other options, as they become relevant. If you change your mind and want to think about steroids again in the future, you just need to mention this at one of your routine appointments.

Where can I find out more?
There are lots of sites on the web that discuss the use of steroids in DMD. Here is one example, but you will be able to find more. Not all of them are completely up to date and it is important to realise that some of the most recent studies have not been formally published yet.

http://www.parentprojectmd.org/treatment/supplements.html

If the information in this fact sheet has been of use then you may like to consider becoming a member of the Muscular Dystrophy Campaign. Membership is free and is open to anybody over 16. By becoming a member you will receive Target MD magazine, which contains news and information on conditions, and you can also receive research and condition up-dates as soon as they are available. To become a member please call 020 7803 4800 or freephone 0800 652 6352 or visit our website at www.muscular-dystrophy.org.

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Healthy eating for children with neuromuscular conditions

Children with neuromuscular conditions may have a tendency to put on weight due to:
- reduced ability to walk around (so less energy is burned off), or
- an increase in appetite (which may be a side effect of some medications e.g. steroids)

This factsheet looks at how to eat a healthy, well-balanced diet.

Why is it important to not be too overweight?
Too much weight will:
- increase the burden on already weakened muscles
- increase risk of surgical procedures
- make it difficult for carers to assist children to move
- reduce the ability to walk
- cause a strain on respiratory muscles
- lead to difficulty in wearing spinal jackets (if needed for treatment for scoliosis)

Prevention is better than cure
It is very important to have enough nutrition to help children grow in height. However, too much nutrition - excess calories - will be stored as fat. This invariably results in children becoming overweight.

Up until the end of the adolescent growth spurt all children grow in height and require adequate nutrition to grow. However, it is important that the diet is balanced and takes into account the amount of energy a child needs and uses.

If a child has become overweight, but they are still growing in height, they do not need to lose weight as such, they just need to keep their weight stable. However, this still means that a change in the diet is necessary.

Generally, it is easier to keep weight stable rather than lose it i.e. prevention is better than cure. Keeping the weight stable can be done in two ways: eating a balanced diet and where possible, using up energy by exercising, but the latter is often difficult in children who are less active than their peers.

Can dieting harm the muscles?
Research has shown that sensible dieting does not reduce muscle bulk or function. Reducing intake of the foods which are high in calories (such as fat and sugar) is a safe and effective way of losing excess fat.
Should I exercise?
Exercise is a good way of using up stored energy and will help to keep weight off in the long term. It is important to be as active as possible whatever your mobility. Again, prevention is better than cure.

What is my ideal weight?
A child’s ideal weight is determined by their height and is therefore different for everyone. If a child’s actual weight tracks along the same centile as their height centile they are correct weight for their height.

What is a balanced diet?
Food and drink must provide energy from a variety of sources. These are:

- **Fruit and vegetables** - these are a rich source of minerals, fibre and vitamins. Try to eat five portions of fruit and/or vegetables a day.

- **Protein-rich foods** - for example, meat, fish, eggs, lentils or beans. Foods rich in protein help to build and repair tissues. Most people in the UK eat enough protein. If more is eaten than the body needs the extra calories will be converted to fat.

- **Starchy food** - for example, bread, cereal, pasta, rice and potatoes. These foods are a good source of energy. The wholegrain varieties are best to choose as they contain more fibre and are therefore more filling. Try to include a good portion at each meal.

- **Milk & dairy foods** - these are good sources of calcium and they help to keep growing bones strong. However, they can be very high in calories unless you choose the low fat variety. If you avoid dairy products then you should discuss this with your dietitian.

- **Fat & fatty foods** - these should be eaten in very small amounts as they are very high in calories. It is easy to eat many more calories than you intend to when eating fatty foods.

- **Sugar & sugary foods** - these should be eaten in very small amounts as they are high in calories and don’t give any other valuable nutrients.

Healthy eating for the whole family
Try to:
- Eat regularly and don’t skip meals otherwise you may end up eating a lot more at your next meal.
- Cut down on the amount of high fat foods e.g. crisps, chocolates, biscuits, pastries, pies, sausages and cakes. Choose low fat varieties, e.g. ‘Lite’ mayonnaise, but eat them sparingly as even these can be high in calories.
- Avoid all fried foods. Instead try grilling, boiling or steaming foods instead.
- Eat plenty of fruit and vegetables: aim for five portions of fruit and vegetables each day.
Although we encourage children to eat fruit it is important **not** to drink too much fruit juice - it is high in fruit sugar but doesn’t contain the fibre that whole fruit does.

- Cut down on foods and drinks high in sugar e.g. sweets, chocolates, sugary drinks, pastries.
- Don’t add sugar to drinks, cereals or in cooking. Try sugar-free or diet drinks instead.
- Try not to eat in between meals, but if you are hungry eat fruit.
- Don’t eat in front of the TV or eat snacks out of the bag, it’s much easier to overeat that way.
- Instead of treating yourself with food, try buying a magazine or new book, or a CD.
- Agree on one or two “treat” foods a week for you and the family.
- Cook more vegetables than you would normally prepare. Put the vegetables on your plate first, so that they take up half the space, followed by starchy foods, such as potatoes, followed lastly by fish or meat. This will help you to eat the right amount of each food group.

**Cooking tips**

**Meat & fish**

- Trim off all visible fat and skin before cooking.
- Do not fry food but instead, try grilling, poaching, microwaving or baking.
- If you use oil, limit it to one teaspoon per person. Measure out the oil on a spoon as opposed to pouring from the bottle.
- If you are making a gravy, use gravy powder or juice from roast meat which has had the fat skimmed off.
- Cook the meat or chicken a drip tray to allow the fat and juices to drain away.

**Potatoes**

- It’s best to have boiled, mashed or jacket potatoes, and try not to add butter or margarine - use skimmed milk instead.
- Have chips occasionally and when you do make sure they are large and/or low fat oven chips instead of making your own in the chip pan.
- Try dry roasting by putting peeled, chopped and parboiled potatoes in a very hot oven without any oil, or instead spray the potatoes with a very small amount of oil spray.

**Healthy lunch ideas**

Include a variety of foods from the different food groups as that will help make the lunchbox healthier. Try to include one or more servings of fruit as they are nutritious, filling and full of vitamins and minerals. Dilute fruit juices with water.

**Lunchbox idea 1**

- Pasta twirls with cherry tomatoes, or pasta with tuna, sweetcorn and peppers
- Fruit bun or scone
- Banana or apple

**Lunchbox idea 2**

- Rice salad with ham, chicken, mushroom and peas/chick peas
- Dried fruit e.g. apple rings, apricots, raisins
- Breadsticks, Twiglets or crackers
• Fromage frais or low fat yoghurt

**Lunchbox idea 3**
• Cous cous with chick peas, raisins, or chopped peppers
• Cherry tomatoes & cucumber
• A slice of malt loaf

**Lunchbox idea 4**
• Potato salad with tinned salmon, cucumber and spring onions
• Muffin or wafer biscuit
• Piece of fruit

**Sandwich ideas**
• Choose lean meat e.g. chicken, turkey, lean ham or beef.
• Try to use tuna fish that's tinned in brine or water, not oil.
• Garnish your sandwiches to make them appetizing and tasty. Use spinach, grated carrot, tomatoes, peppers, cucumbers, pickles, salad, piccalilli, grapes, mustard, low calorie mayonnaise or dressing.
• Make sandwiches with wholegrain bread as they contain lots of fibre and are therefore more filling than white bread.
• Use a low fat spread in your sandwiches and spread it very thinly.

**Healthy sandwich fillings**
• Grated cheese and carrot
• Ham and pineapple
• Tuna and sweetcorn
• Banana
• Ham and cheese
• Salmon and cucumber
• Chicken and salad

If you want to add mayonnaise use only one or two teaspoons and always get the lower fat varieties.

**Healthy fast food ideas**
Fast food doesn't have to be unhealthy. Here are some nutritional ideas:
• Baguettes, pitta bread, and granary, rye or pumpemickel bread filled with salad, cold meat, egg, tuna (in brine or water) make good meals. Use low calorie mayonnaise – but only a teaspoon. Pickles, piccalilli and relish help to add flavour without too many calories.
• Jacket potatoes with either baked beans, cottage cheese, low fat cheese, tuna or salmon (but don’t add butter or margarine).
• Baked beans, sardines, or a poached or boiled egg on toast.
• Pasta in tomato sauce with (or without) tuna or salmon (in brine) and sweetcorn. Spice it up with two or three slices of chopped salami and chilli sauce. Avoid creamy pasta sauces as they are high in calories and fat.
• Bean salad, brown rice, or pasta salad, with low calorie dressing.
• Vegetable and bean or lentil soup. There are many ready-made ones, both fresh and tinned. Again, try to avoid the creamy ones.
• Always choose tinned fish (e.g. salmon or tuna) in brine not oil.

The importance of calcium
When you are active, your muscles "pull" on your bones. This pulling action helps to keep the bones strong. Children with neuromuscular disease may have bones which are less strong because they have weaker muscles and are less active.

This can make them more prone to bone fractures than other children. In addition, a child is more at risk of fractures if they are on daily steroids. It is important therefore, that as much activity as possible is encouraged in order to help keep bones strong. A diet with adequate calcium and vitamin D is also important for optimal bone strength.

Calcium requirements:
• 4 - 10 years-old: two portions a day
• 11 - 18 years-old: three portions a day

Rich sources of calcium include dairy products. If your child excludes or eats very little dairy products this increases the risk of bone fractures. Please discuss with your dietitian.

A serving of the following foods gives approximately 200 mg calcium:
• One glass of semi-skimmed or skimmed cow's milk or sweetened soya milk
• A matchbox size piece of cheese (low fat varieties e.g. Cheddar, Red Leicester, Double Gloucester)
• 125g carton low-fat yoghurt
• Two 112g carton of cottage cheese
• Two small (60g) cartons low fat fromage frais
• Low fat drinking yoghurt
• Iced low-fat yoghurt
• One tin of sardines
• One tin of pilchards
• Two large low fat cheese triangles
• Three scoops of low fat dairy ice cream
• One small handful of dried figs

The importance of vitamin D
Vitamin D is a fat-soluble vitamin and is found in some fatty foods. However, it can also be made in our own bodies by the action of sunlight on the skin. Most of the vitamin D in our bodies comes from this source.
Vitamin D is vital for the absorption of calcium from our food and to maintain healthy bones. A child may be at risk of vitamin D deficiency if the child is kept indoors or, when outside is covered thoroughly, or has pigmented skin.

Sources of vitamin D include:
- Oily fish (e.g. mackerel, herring, salmon, pilchards, sardines)
- Margarine
- Eggs

Requirements:
- Try to get outside to play once a day for around 20 to 30 minutes in the summer months. If you are unable to get outside during the summer it is worthwhile taking a supplement that includes vitamin D. A multi-vitamin may be recommended if calcium and other vitamins are needed.

Other factsheets that may be useful
- Nutrition and feeding in individuals with neuromuscular conditions
- Gastrostomy
Appendix 4

Scottish Muscle Network - Guideline for the Management of
Bone Mineral Density Problems in
Boys with Duchenne Muscular Dystrophy

Introduction

Duchenne Muscular Dystrophy (DMD) is the most common muscular dystrophy of boys, and is characterised by poor muscle function and progressive loss of ability to walk, generally with patients becoming wheelchair bound by teenage years. Children and young people with poor mobility are at increased risk of osteoporosis and pathological fractures.

There is good evidence that corticosteroids given to ambulant boys with DMD improve muscle function, at least in the short term\(^1\). Most boys with DMD will be started on corticosteroids, either on intermittent pulse therapy or continuous treatment.

Long term use of corticosteroids and prolonged poor mobility are independent risk factors for pathological fracture. Over time as children get older and, possibly, less mobile, bone mineral density (BMD) falls in DMD and this fall may be most marked in those treated with corticosteroids\(^2\).

Bone mineral density can be measured by a Dual Energy X-ray Absorptiometry (DXA) scanner and this service is available in most large hospitals for monitoring osteoporosis in the elderly. However, for children, the results should be interpreted by a clinician or paediatrician with a knowledge of DXA scans in children.

There is evidence in adult practice that prophylactic use of bisphosphonates may be beneficial (3) but insufficient evidence exists for this in children, and is not currently recommended.

This guideline is based on consensus between local and regional paediatricians with interests in neuromuscular disorders and metabolic bone disease.

Aim

The aim of this guideline is to maintain health, mobility and function as much as possible, by promoting bone health, measuring bone mineral density and treating those with fractures.
Before starting steroids

Check plasma calcium, phosphate, PTH (check tube) and alkaline phosphatase (yellow tube) and 25 Hydroxyvitamin D (separate yellow tube, covered to exclude light)
Give healthy diet advice leaflet
Arrange baseline DXA scan

One year after starting therapy

Check plasma calcium and vitamin D
Arrange DXA
Repeat DXA 1-2 yearly thereafter depending on results.

Fractures
In children with DMD who have a fracture, DXA scan should be considered if this has not been done previously- including those with low impact fracture and poor mobility who have not received steroids. Bisphosphonate therapy should be considered in those children who are on steroids, have low impact fractures and especially if they have low BMD. However, ensure that they have a normal Vitamin D status.

Arranging DXA scans
As other sheet
1. Where there is no facility for paediatric DXA locally referrals can be made to RHSC in Glasgow.
2. Where DXA is available locally but no specialist available for reporting scans can be sent to and reported by Dr Faisal Ahmed, Consultant Paediatrician Metabolic Medicine, Royal Hospital for Sick Children, GlasgowG3 8SJ
3. DXA can be arranged and reported locally

References


Appendix 5

Cardiac assessment in Duchenne Muscular Dystrophy

An information leaflet by SMN in Collaboration with Scottish Paediatric Cardiologists

Current guidelines in the management of boys and young men with muscular dystrophy recommend cardiac (heart) assessment. This leaflet has been designed to give parents some explanation why this happens, and what treatments are available if that should become necessary.

Why do you need a referral to the heart (cardiology) clinic?
Muscular dystrophy causes weakness of the muscles of the body from a young age, and the muscles of the heart can also be affected, although this is not usually until boys are older. Specialised heart tests can detect changes in the heart muscle function some time before any symptoms or other signs develop and for this reason, screening for early signs of heart involvement is recommended for boys with Duchenne Muscular Dystrophy.

What are these tests?
Specialised screening tests involve assessing the heart function by a heart scan (echocardiogram) and electrical test (ECG). Neither of these are uncomfortable or painful at all. In future this is likely to be accompanied by blood tests which may be more sensitive at assessing the stretch of the filling chambers of the heart.

How often should these tests be done?
Tests are usually done initially after diagnosis, every two years until around 10 years of age, and every year after that.

What happens if the tests show the heart muscle is not working properly?
If the screening tests show a reduction in heart muscle function, it does not usually cause any symptoms or outward problems for the individual. If there are any symptoms of the heart not working properly, current guidelines recommend treatment with the same sort of drug treatment for the heart that would be given to patients who have reduced heart function for other reasons. These medicines can help the heart to work better, and can slow down any further deterioration, but they can have some side effects. Side effects are not usually serious, and you will be given further information about this if treatment is started.
All these medicines are used in every day life by many adults for the treatment of heart problems or high blood pressure.

Is there any benefit in starting treatment early?
it is not known for sure that these medicines give any benefit if given before any symptoms develop, but there is some research which suggests there might be. The heart specialist (cardiologist) will be able to discuss the pros and cons of starting early treatment with you at the clinic.

What will happen if treatment is started?
Treatment always starts with small doses and increase them after regular blood tests have shown there is no harm to kidney function. Initially one drug is used from a group of drugs called ACE inhibitors. There are many of these and their names end with "..pril " such as captopril, enalapril, lisinopril. Sometimes these do not agree with patients and we use drugs which end in " ...artan" When these drugs are at a suitable dose, we would then add in drugs called beta-blockers.
Will the results be given at the clinic?
The cardiologist will not usually be able to tell you right away if the heart is working normally or if there has been any change. This is because he/she often needs to look at the scans several times after the clinic, and to compare each scan with previous ones. The doctor will, however, write to your GP once he/she has assessed the scans and let him/her know whether things seem unchanged or whether drug treatment should be started. A copy of these letters will be sent to you, and you can have the opportunity for a further appointment if you wish you discuss starting treatment further before it begins.
## Appendix 6

### Respiratory Chart for DMD

<table>
<thead>
<tr>
<th>Stage</th>
<th>Aim</th>
<th>Objective</th>
<th>Outcome measure</th>
</tr>
</thead>
<tbody>
<tr>
<td>Young ambulant (Diagnosis to approximately 10 years)</td>
<td>To maintain and promote good inspiratory capacity.</td>
<td>Encourage wind instruments, blowing bubbles, singing etc</td>
<td>Annual spirometry from age 5</td>
</tr>
<tr>
<td></td>
<td>To maintain and promote respiratory fitness</td>
<td>Encourage young people to maintain an active lifestyle within their capabilities.</td>
<td></td>
</tr>
<tr>
<td></td>
<td>To teach an awareness of breathing control</td>
<td>An incentive spirometer can be used to teach breathing control awareness</td>
<td></td>
</tr>
<tr>
<td>Wheelchair dependent (Approx 11-16 years)</td>
<td>To maintain chest compliance</td>
<td>Increase persons awareness of improving lung volume recruitment such as breath stacking/ glosopharyngeal breathing. Teach parents /carers how to increase inspiratory measures using an ambu bag.</td>
<td>Regular assisted inspirations (aim for daily and this can be increased when young person has a chest infection)</td>
</tr>
<tr>
<td></td>
<td>To prevent chest infections</td>
<td>People with DMD are encouraged to have their flu and pneumococcal jabs where appropriate particularly if FVC &lt;50% of predicted value</td>
<td>Referral to Respiratory Consultant for formal monitoring</td>
</tr>
</tbody>
</table>
| | To introduce effective measures for clearing the chest during infections | Respiratory assessment by physiotherapist to assess best method of clearing the chest (Positive Expiratory End Pressure / Ambubag) If indicated teach cough augmentation techniques such as manual splinting of the diaphragm and thoracic holds. Use of cough assist machine (mechanical in-exsufflator) if appropriate. Early delivery of antibiotics during a chest infection | Minimum of annual spirometry. Once FVC falls to <50% predicted, sleep investigations should be performed to look for signs of nocturnal hypoventilation. This should consist of overnight oximetry and capnography. More detailed respiratory sleep studies may be considered once FVC is <30%
<p>| | To prepare for Non invasive ventilation (NIV) | Practice regular lung volume recruitment techniques with use of ambu bag | Referral to Orthopaedic Consultant for spinal assessment |
| | To monitor scoliosis | Refer to specialist spinal services for monitoring of any scoliosis | It is advisable that patients with respiratory muscle weakness have a low threshold for commencing antibiotics when they develop a respiratory tract infection. |</p>
<table>
<thead>
<tr>
<th>Wheelchair Dependent on Non Invasive Ventilation (NIV)</th>
<th>To prevent/manage chest infections</th>
<th>If peak cough is less than 160/l/min Assess the need for a mechanical in-exsufflator (MI-E) if other assisted cough techniques are ineffective. The cough assist machine can be used in conjunction with manually assisted cough techniques. The MI-E can also be used to clear secretions in the absence of a chest infection and some like to use it prophylactically If aspiration or weight loss is evident, refer to a speech and language therapist and/or dietician.</th>
<th>Liaise with breathing support/nursing staff. Regular assessment of adequacy of ventilation and early treatment of complications (eg skin breakdown from pressure of face masks).</th>
</tr>
</thead>
<tbody>
<tr>
<td>Indications for ventilation FVC&lt; 1.25l Frequent chest infections Poor Sleep quality Signs of nocturnal hypoventilation on sleep study Poor Appetite Weight loss (see Epworth Sleepiness scale)</td>
<td>To maintain chest compliance</td>
<td>As above</td>
<td></td>
</tr>
<tr>
<td></td>
<td>To manage transition process.</td>
<td>Liaise with adult physiotherapy teams</td>
<td></td>
</tr>
<tr>
<td>Advanced stages NIV and daytime ventilation for short/extended periods of time.</td>
<td>Ensure person and carers are able to clear chest effectively. At this stage, muscle weakness and fatigue are of particular concern in cough augmentation techniques.</td>
<td>As well as all of the above, a suction unit may be necessary.</td>
<td>Regular spirometry Regular contact with breathing support team Regular ventilator checks to monitor respiratory pressures. Ventilator may be used in conjunction with chest clearing techniques. Discuss with respiratory physiotherapist.</td>
</tr>
<tr>
<td>Emergency admission procedure</td>
<td>To manage chest infection To manage end of life care</td>
<td>Preparation of advance directives should be encouraged in DMD. These can be discussed with the respiratory consultant or neurologist.</td>
<td></td>
</tr>
</tbody>
</table>

### Symptoms of respiratory failure

- Shortness of breath
- Orthopnea (Dyspnoea when lying flat)
- Recurrent chest infections
- Lethargy
- Weight loss

### Symptoms of nocturnal hypoventilation

- Frequent nocturnal wakening
- Excessive daytime sleepiness
- Reduced concentration
- Un-refreshing sleep
- Fatigue
- Early morning headache

### Symptoms of Bulbar Dysfunction

- Nasal regurgitation
- Choking or coughing episodes at mealtimes
- Weak cough
- Recurrent chest infections

### Signs of respiratory muscle weakness

- Weak sniff or cough
- Abdominal paradox
- Recruitment of accessory muscles at rest
- Increased rate of respiration
- Reduced chest expansion
- Cyanosis
- Papilloedema (severe hypoventilation)
Appendix 7

Lung Volume Recruitment Techniques
In order to successfully expel secretions from the lungs the patient requires having 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 assist0
  - 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 ambubag is used before meals however if bulbar control is poor and patient is likely to aspirate, it may be beneficial to use after eating.

It may be 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 clients 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. For an international review of respiratory literature refer to [https://eshare.bah.com/sites/CDC_SGCC/default.aspx](https://eshare.bah.com/sites/CDC_SGCC/default.aspx)
Appendix 8

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

Epworth Sleepiness Scale

<table>
<thead>
<tr>
<th>Score</th>
<th>Chance of Dozing</th>
</tr>
</thead>
<tbody>
<tr>
<td>0</td>
<td>No chance of dozing</td>
</tr>
<tr>
<td>1</td>
<td>Slight chance of dozing</td>
</tr>
<tr>
<td>2</td>
<td>Moderate chance of dozing</td>
</tr>
<tr>
<td>3</td>
<td>High chance of dozing</td>
</tr>
</tbody>
</table>

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.
Appendix 9

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 but data for this group is sparse due to the lack of historical perspective in this “new” older population. Monitoring the scoliosis should commence before the loss of ambulation to enable surgical intervention to be offered at the appropriate stage 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° and 40°. 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. Advance 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 may not be at 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 is unable to 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 can be more comfortable without the strengthening due to their spine and head alignment. It is best to avoid excessive hip flexion beyond 90° particularly in the first nine months. In some cases the pelvis may be fused to the spine and excessive hip flexion can increase the strain in the lower back through flexion of the lumbar spine.

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 (hip flexion to 90° is permitted) 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.

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 Clinical Specialist in Spinal Surgery, Edinburgh Sick Children’s Hospital, Sciennes Road, Edinburgh EH9 1LF Tel:0131 536 0000 Bleep:9126 (a patient leaflet can be downloaded from www.gla.ac.uk/muscle).

Approved July 2015
Review July 2018
Appendix 10

References


Approved July 2015
Review July 2018


Muscular Dystrophy Campaign. (2002). Inclusive education for children with muscular dystrophy and other neuromuscular conditions: Guidance for primary and secondary schools. Published by MDC

Muscular Dystrophy Campaign Wheelchair Provision for children and adults with muscular dystrophy and other neuromuscular conditions 2006 [www.musculardystrophyuk.org](http://www.musculardystrophyuk.org)


Approved July 2015
Review July 2018


Appendix 11

Useful Contacts

1. The Scottish Muscle Network - www.smn.scot.nhs.uk
   Manager: Hugh Kennedy – 0141 300 1336
   Network Co-ordinator: Laura Craig – 0141 300 1424

2. Muscular Dystrophy UK - www.musculardystrophyuk.org
   Care Advisors: East: Gillian Mitchell: 0131 536 0973
                  West: Wilma Stewart: 0141 354 9204
                  North: Julie Burslem 01463 706106 (secretary)

3. Action Duchenne – www.actionduchenne.org


5. Sleep Scotland, 8 Hope Park Square, Edinburgh, EH8 9NW Tel: 0131 651 1392. E-mail: sleepscotland@btinternet.com

This guideline is not intended to be construed or to serve as a standard of care. Standards of care are determined on the basis of 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.