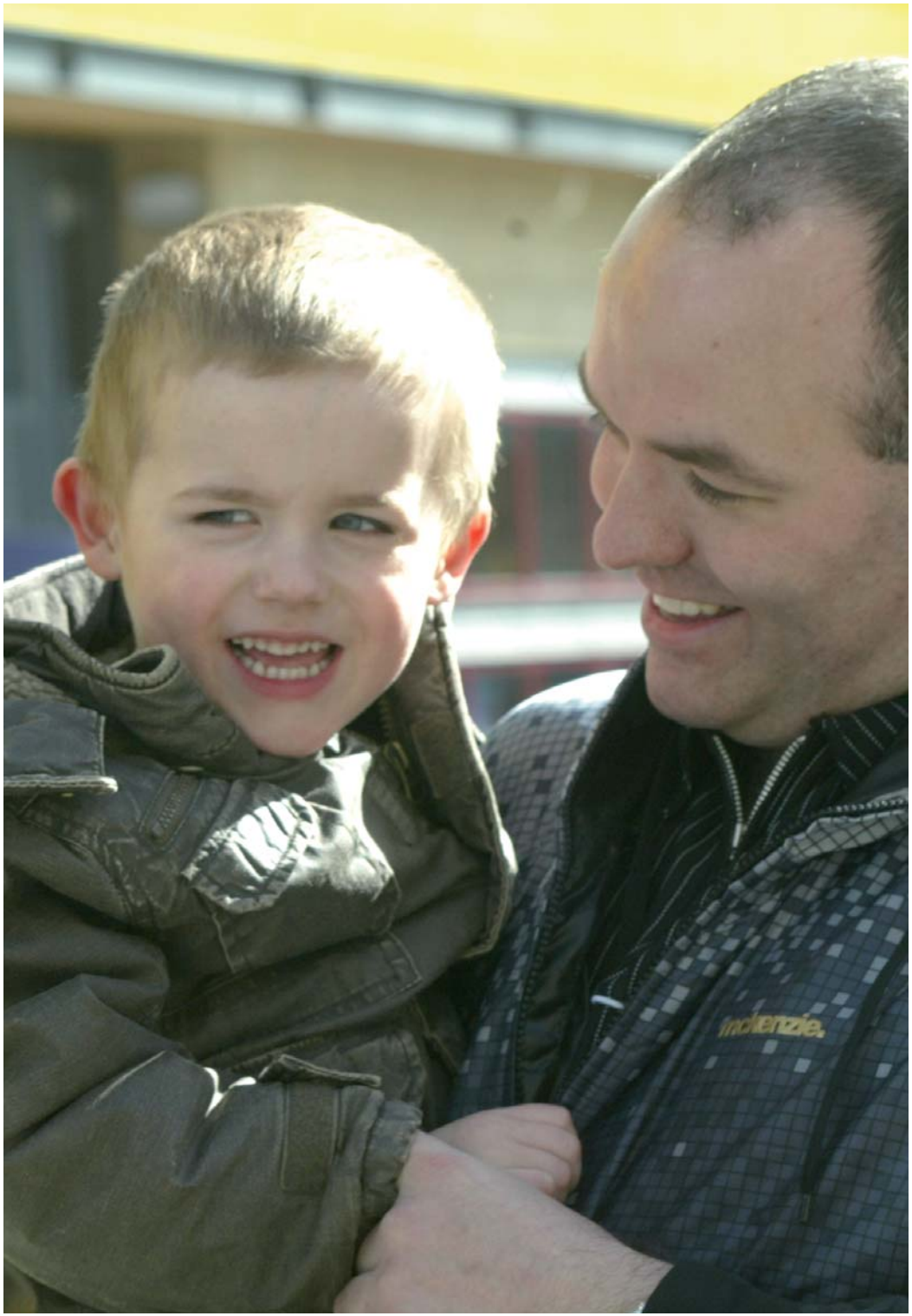
THE DIAGNOSIS AND MANAGEMENT OF DUCHENNE MUSCULAR DYSTROPHY

A guide for families



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Contents

1 Introduction

This guide for families summarises the results of an international consensus on the medical care of Duchenne muscular dystrophy (DMD). This effort was supported by the US Centers for Disease Control and Prevention (CDC), in collaboration with patient advocacy groups and the TREAT-NMD network. The main document is published in Lancet Neurology.

REFERENCES FOR THE MAIN DOCUMENT:

Bushby K, et al. The Diagnosis and Management of Duchenne Muscular Dystrophy, part 1: diagnosis, and pharmacological and psychosocial management, Lancet Neurology 2010, 9(1) 77-93.

Bushby K, et al. The Diagnosis and Management of Duchenne Muscular Dystrophy, part 2: implementation of multidisciplinary care, Lancet Neurology 2010, 9(2) 177-189.

The main document can be downloaded free from http://www.treat-nmd.eu/diagnosis-and-management-of-DMD/

The recommendations are based on an extensive study by 84 international experts in DMD diagnosis and care chosen to represent a broad range of specialties. They independently "rated" methods of care used in the management of DMD to say how "**necessary**", "**appropriate**" or "**inappropriate**" each one was at different stages of the course of DMD. In total they considered more than 70,000 different scenarios. This allowed them to establish guidelines that the majority agreed represented the "**best practice**" for DMD care.

The experts stressed that the best management of DMD requires a **multidisciplinary approach**, with the input of specialists in many different areas, and that there must be a doctor or medical professional that coordinates these efforts. Because everybody is different, the person with DMD and his family should be **actively engaged with a medical professional** who will coordinate and individualise clinical care.

This document will provide you with basic information to allow you to participate effectively in this process. The doctor or medical professional who coordinates care must be aware of all potential issues in DMD and must have access to the interventions that are the foundations for proper care and to input from different specialties. The emphasis of interventions will change over time. This guide takes you through the different topics or domains of DMD care (Figure 1). Not all of these specialists will be needed at all stages of the condition, but it is important that they are accessible if necessary and that the person coordinating care has support in all these areas.

2 How to use this document

This guide contains a lot of information. You can use it in two different ways. Some families prefer to concentrate on the stage of DMD that their child has reached. Others like to understand as much as possible about every aspect of DMD from the start.

In this section and in Figure 1 you can see how experts think about the different stages of DMD and how the care needs of someone with DMD changes over that time. Then if you want to go directly to the parts of the document which are relevant to you now, you should be able to find them easily. At the end of the document, there are two sections that might be important to have as an easy reference: things to remember if surgery is planned and things to consider in an emergency situation. We have colour-coded the different sections to match with the colours in the figure. There is a glossary at the end of the booklet to help you to understand any difficult terms that you find here or that your care team might use.

"This is a guide to the 'medical' aspects of DMD, but always bear in mind that the medical side isn't everything. The idea is that by minimising medical problems, your son can get on with his life and you can get on with being a family. It's good to remember that most Duchenne boys are happy kids and most families do very well after the initial shock of the diagnosis."

Elizabeth Vroom,
United Parent Projects Muscular Dystrophy

TAKING DMD STEP BY STEP (Figure 1)
DMD is a condition that changes as time goes by. Doctors and others often recognise key "stages" in the progression of the condition. They use these key stages to guide their recommendations about care, though these stages can be a rather artificial distinction. Still, it can be useful to use the stages to identify the kind of interventions that are recommended at any particular time and what you should expect of your care team at that time.

Presymptomatic

Most boys with DMD are not diagnosed during the **PRESYMPTOMATIC** stage unless there is a family history of the condition or unless blood tests are done for other reasons. Symptoms of delayed walking or delayed speech are present, but are typically subtle and are often unnoticed or unrecognised at this stage.

Early ambulatory

In the **EARLY AMBULATORY (WALKING)** stage, boys will be showing what are typically regarded as the "classical" signs of DMD – a Gowers' manoeuvre (which means that they need to support themselves with hands on thighs as they get up from the floor), waddling type walking (gait) and walking on their toes. They can still climb stairs, but typically bring the second foot up to join the first rather than going foot over foot.

These two early stages are the time when the diagnostic process is likely to be underway (Section 3).

DIAGNOSIS: Specific tests will be recommended in order to identify the change in the DNA or genetic mutation that caused DMD. Input from specialists may be needed in order to interpret these tests and to discuss how the results may impact your son and other family members.

LEARNING AND BEHAVIOUR: Boys who have DMD have a higher chance of having problems in these areas. Some are due to the effect DMD has on the brain, others to physical limitations. Some medications such as steroids also play a role. Family support is essential, and input from specialists may be needed to address specific issues of learning and behaviour (Section 10).

PHYSIOTHERAPY: An introduction to the physiotherapy team (Section 5) at this early stage will mean that exercise regimes can be introduced gradually to keep muscles supple and prevent or minimise tightness at the joints. The physiotherapy team can also advise on appropriate exercise for school in order to support participation.

STEROIDS: It is a good time to find out about options such as steroids (Section 4) which will be planned for when the boy's gaining of motor skills starts to level out or "plateau". In planning for the use of steroids, it is important to check that all immunisations are complete and to find out if any risk factors for the side effects of steroids can be anticipated and minimised. Guidance on weight control for example might be required.

HEART AND BREATHING MUSCLES: Typically, problems with the heart and breathing muscles are not likely to be present at this stage, but surveillance should be built into the regular follow-up clinic visits to establish the baseline (what is 'normal' for your son). Cardiac monitoring is recommended at diagnosis and then every two years up to age 10. After the age of 10, monitoring should be more frequent. It is also important that the boy has pneumococcal and influenza vaccinations (Section 7).

Late ambulatory

In the **LATE AMBULATORY** stage, walking becomes increasingly difficult and there are more problems with climbing stairs and getting up from the floor.

LEARNING AND BEHAVIOUR: Continued support from professionals will be necessary to help with any learning and behaviour issues, and specific help may be needed to address coping strategies for dealing with the loss of ability to walk (Section 10).

PHYSIOTHERAPY: Rehabilitation input will continue to focus on range of movement and independence (Section 5). If joint tightness becomes too much of a problem for physiotherapy interventions, assessment and input from orthopaedic specialists may be necessary. It is important to make sure that there are appropriate wheelchairs with supportive seating to promote continued independence and comfort.

STEROIDS: Ongoing management of steroid treatment is important at this stage, with attention to the specific regime and dose used (Section 4) as well as attention to side effects.

Twice yearly assessments to monitor strength and function are important. Continued attention to weight control needs to take into account any tendency to be under or over weight with appropriate intervention if there is a problem (Section 9).

HEART AND BREATHING MUSCLES: From a respiratory and cardiac point of view (Sections 7 and 8 respectively), the low risk of obvious problems remains but ongoing assessment of the heart and breathing muscles is necessary. Echocardiogram and other types of tests should be done annually from the age of 10. The doctor will recommend interventions if there are any changes observed on the echocardiogram.

Early non-ambulatory

In the **EARLY NON-AMBULATORY** phase the boy needs to use a wheelchair. To start with he may be able to wheel the chair himself and typically his posture is still good (Section 5).

LEARNING AND BEHAVIOUR: Despite the backdrop of the condition progressing, an increasing emphasis on independence is necessary to encourage normal participation in school and fun activities through adolescence.

PHYSIOTHERAPY: Attention to tightness in the upper limbs (shoulders, elbows, wrist and fingers) becomes very important, as does the need for supporting equipment to help keep your son standing. Spinal curvature (scoliosis) is seen much less often with the widespread use of steroids, but monitoring for this is still very important following the loss of ambulation. In some cases scoliosis progresses quite rapidly, often over a period of months (Section 6). Orthopaedic input may also be needed to deal with problems with foot posture which can cause pain or discomfort and limit the choice of footwear.

STEROIDS: Maintenance of steroid treatment continues to be an important part of management in this phase (Section 4) whether started previously and continued into this phase or started at this stage.

HEART AND BREATHING MUSCLES: Monitoring of cardiac function at yearly intervals is still essential and any deterioration should be treated promptly (Section 8). Respiratory function is likely to begin to decline after loss of independent walking and a staged series of interventions to help with breathing and to aid with coughing needs to be introduced (Section 7).

Late non-ambulatory

In the **LATE NON-AMBULATORY** phase, upper limb function and maintenance of good posture is increasingly difficult and complications are more likely.

PHYSIOTHERAPY: It is important to discuss with the physiotherapist what types of equipment will best support independence and participation. Other adaptations may be needed to help with activities such as eating, drinking, toileting and transferring to and turning in bed.

STEROIDS: Decisions related to steroid regimens, nutrition and weight management are reviewed and discussed with the care team.

HEART AND BREATHING MUSCLES: Twice yearly monitoring of heart and lung function is recommended and often more intensive investigations and interventions may need to be implemented.

Many young men with DMD live fulfilling adult lives. It is important to proactively plan for a life as a supported but independent adult with all of the opportunities and challenges that this entails.

The following sections deal in turn with the ten different domains of care for DMD as described in Figure 1.

Figure 1

The different areas of care required at each stage of DMD

Stage 1: PRESYMPTOMATIC	Stage 2: EARLY AMBULATORY	Stage 3: LATE AMBULATORY	Stage 4: EARLY NON-AMBULATORY	Stage 5: LATE NON-AMBULATORY	
May be diagnosed at this stage if CK found to be elevated by chance or if positive family history May show developmental delay but no gait disturbance	Gowers' manoeuvre Waddling gait May be toe-walking Can climb stairs	Increasingly laboured gait Losing ability to climb stairs and rise from floor	May be able to self-propel for some time Able to maintain posture May develop scoliosis	Upper limb function and postural maintenance is increasingly limited	
Requires diagnostic workup and genetic counselling		Likely to be diagnosed by this stage unless delayed for other reasons (e.g. concomitant pathology)			DIAGNOSIS
Anticipatory planning for future developments Ensure immunisation schedule complete	Ongoing assessment to ensure course of disease is as expected in conjunction with interpretation of diagnostic testing At least six-monthly assessment of function, strength and range of movement to define phase of disease and determine need for intervention with steroids, ongoing management of steroid regime and side-effect management				NEUROMUSCULAR MANAGEMENT
Education and support Preventative measures to maintain muscle extensibility/minimise contracture Encouragement of appropriate exercise/activity Support of function & participation Provision of adaptive devices, as appropriate		Previous measures continued Provision of appropriate wheelchair and seating, and aids and adaptations to allow maximal independence in daily activities, function and participation			REHABILITATION MANAGEMENT
Orthopaedic surgery rarely necessary		Consideration of surgical options for Achilles tendon contractures in certain situations	Monitoring for scoliosis: Intervention with posterior spinal fusion in defined situations Possible intervention for foot position for wheelchair positioning		ORTHOPAEDIC MANAGEMENT
Normal respiratory function Ensure usual immunisation schedule including 23-valent pneumococcal and influenza vaccines	Low risk of respiratory problems Monitor progress		Increasing risk of resp. impairment Trigger respiratory assessments	Increasing risk of resp. impairment Trigger respiratory investigations and interventions	PULMONARY MANAGEMENT
Echocardiogram at diagnosis or by 6 years	Maximum 24 months between investigations until age 10 years, annually thereafter	Assessment same as in the younger group Increasing risk of cardiac problems with age; requires intervention even if asymptomatic Use of standard heart failure interventions with deterioration of function			CARDIAC MANAGEMENT
Monitoring for normal weight gain for age Nutritional assessment for over/underweight				Attention to possible dysphagia	GASTROINTESTINAL MANAGEMENT
Family support, early assessment/ intervention for development, learning and behaviour	Assessment/intervention for learning, behaviour and coping Promote independence and social development			Transition planning to adult services	PSYCHOSOCIAL MANAGEMENT

Figure 2 - Gowers' Manoeuvre

3 Diagnosis

Care at diagnosis

The specific cause of a medical disorder is called the diagnosis. It is very important to establish the exact diagnosis when DMD is suspected by your doctor. The aim of care at this time should be to provide an accurate diagnosis as quickly as possible. With prompt diagnosis, everyone in the family can be informed about the general course of DMD, provided with genetic counselling and told of treatment options. Appropriate care can be put in place and ongoing support and education can be provided to the family. Ideally, diagnosis should be performed by a doctor who is a neuromuscular specialist and who can assess the child clinically and can initiate and interpret investigations properly. Family follow-up and support following diagnosis will often be supplemented by support from genetic counsellors.

IMPORTANT FACTS TO REMEMBER:

1. Getting a diagnosis is an important step so that you and your doctor can make plans for your son's care.
2. Doctors cannot diagnose DMD using only a CK test. If your son has elevated CK levels, your doctor will need to confirm the diagnosis using genetic testing.
3. YOU ARE NOT ALONE. Reach out to your doctor to answer any questions you may have and seek consultation from a genetic counsellor.
4. This is also a time when contact with a support group or advocacy organisation can be of particular help. You can find lists of contacts at **www.treat-nmd.eu/dmdpatientorganisations**

When to suspect DMD

The first suspicions are usually raised by one of the following three signs (even when there is no history of DMD in the family):

PROBLEMS WITH MUSCLE FUNCTION: It is often a family member who notices something is wrong. Boys who have DMD walk later than other boys their own age. They have enlarged calf muscles and have trouble running, jumping or climbing stairs. They fall easily and may have a tendency to walk on their toes. They may also have a speech delay. One of the classic signs of DMD is what is known as the "Gowers'" manoeuvre or sign, where the boy has to use his hands and arms to "walk" up his body in order to push himself to an upright position. This is due to weakness in the hips and thigh muscles (see Figure 2).

HIGH LEVELS OF THE MUSCLE PROTEIN CREATINE KINASE (CK) in a blood test. The finding of a high CK level should prompt an urgent referral to a neuromuscular specialist for confirmation of the diagnosis. High levels of CK are seen in people with other kinds of muscle conditions and a high CK alone is not enough to confirm DMD.

HIGH LEVELS OF THE "LIVER ENZYMES" AST AND ALT in a blood test. High levels of these enzymes in the blood are often associated with liver disease, but muscular dystrophies can also cause this elevation. Unexpectedly high levels of these enzymes without another cause should raise the suspicion that the CK will be high as well and so a diagnosis of muscular dystrophy might be suspected. A liver biopsy is not recommended.

DELAYED SPEECH DEVELOPMENT: Children with DMD often also have some delay in their speech development and sometimes that is the symptom that is first noticed (see Section 10).

Confirming the diagnosis of DMD

DMD is a genetic disease – it is caused by a mutation or change in the DNA for a gene called the dystrophin or DMD gene. The diagnosis has to be confirmed by genetic testing usually on a blood sample, but other tests are also sometimes performed as well. Some background about genetic testing is provided in Box 1.

The tests

1) GENETIC TESTING

Genetic testing is always necessary even if DMD is first confirmed by muscle biopsy. Different types of genetic tests are able to provide specific and more detailed information about the change in the DNA or mutation. Having genetic confirmation of the diagnosis is important for several reasons. It will help to determine if the boy may be eligible for a number of mutation-specific clinical trials and will help the family with decisions related to prenatal diagnosis and future pregnancies.

Once the exact mutation or change in the DNA in the dystrophin gene is known, mothers should be offered the opportunity for genetic testing to check whether they are carriers or not. This information will be important for other female family members on the mother's side (sisters, daughters, aunts, cousins) to understand if they may be carriers as well.

Genetic testing and a referral to a genetic counsellor will help the family understand the results of the testing and the potential impact on other family members (see Box 1).

2) MUSCLE BIOPSY ANALYSIS

Your doctor may recommend a muscle biopsy (taking a small sample of muscle for analysis). The genetic mutation in DMD means the body cannot produce the protein dystrophin, or doesn't produce enough of it. Tests on the muscle biopsy can provide information on the amount of dystrophin present in the muscle cells (see Figure 3).

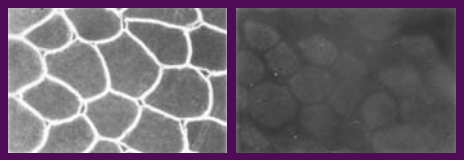
If confirmation of the diagnosis has already been achieved by genetic testing, a muscle biopsy may not be required. However, at some centres, the diagnosis of DMD may be made by muscle biopsy analysis. Genetic testing after a positive biopsy diagnosis of DMD is still essential to determine the specific change in the DNA or genetic mutation causing DMD.

There are two types of tests normally performed on a muscle biopsy. They are immunocytochemistry and immunoblotting for dystrophin. These tests are done to determine the presence or absence of dystrophin and can help to distinguish DMD from a milder form of the condition.

Figure 3

Muscle biopsy; **Left:** normal muscle showing dystrophin round the fibres, **Right:** dystrophic muscle with absent dystrophin.

NORMAL DMD



3) OTHER TESTS

In the past, the tests known as electromyography (EMG) and nerve conduction studies (needle tests) have been a traditional part of the assessment of a child with a suspected neuromuscular disorder. The experts agree that these tests are NOT appropriate or necessary for the evaluation of DMD.

Box 1

Why genetic confirmation is important

GENETIC COUNSELLING AND CARRIER TESTING:

- Sometimes the genetic mutation causing DMD arises by chance in the boy. This is considered a spontaneous mutation. In other cases, it has been passed on by the boy's mother.
- If the mother has the mutation, she is called a "carrier", and she can pass the genetic mutation on to her other children. The boys she passes it on to will be affected by DMD, while the girls will be carriers themselves. If the mother is tested and is found to have the mutation, she can make informed decisions about future pregnancies, and her female relatives (sisters, aunts, daughters) can also be tested to see if they are also at risk of having a boy with DMD.
- Even when a woman is not a carrier, there is a small risk to future pregnancies because the mutation may occur in her ova or egg cells. This is called "germ line mosaicism".
- A carrier also has a small risk of developing a weak heart or even leg weakness later in life. Knowing carrier status helps to identify this risk so the woman can get proper advice.
- You should have access to a genetic counsellor who can explain all this to you in more detail.

ELIGIBILITY FOR CLINICAL TRIALS: There are a number of clinical trials underway in DMD that are targeted to certain types of mutations. Genetic testing is important to understand whether your boy is eligible to participate in these trials. To help doctors find the boys who might be eligible, you should register in a patient registry.

The most important question you need to have answered is whether the genetic testing performed was up to currently accepted standards, allowing the exact mutation to be defined. If it was not, then further testing might be required. You should discuss this with your doctor. The exact mutation is also needed to register with one of the DMD registries. You can find details of the kinds of tests that might be done and how effective they are at detecting the absolute detail of the mutation in the main document.

All of the national patient registries for DMD across the world are listed at: www.treat-nmd.eu/patientregistries.

Please see this website for more details.

4 Neuromuscular management - maintaining strength and function

What assessments should be done and why

Your son should have regular checkups with a specialist doctor who has the expertise to monitor how things are going and to understand if there is anything unusual that might need additional evaluation. This is important in order to make decisions about new treatments at the most appropriate time and to anticipate and prevent problems to the maximum extent possible. It is recommended that your son sees the doctor every 6 months and the specialist physiotherapist and/or occupational therapist about every 4 months if possible.

Tests used in different clinics to follow individuals with DMD may vary. The most important thing is that there is regular review so that interventions can be properly monitored. This regular assessment should include tests that help show how the condition is progressing, including:

STRENGTH: Strength may be measured in a number of different ways to see if the force that can be generated at specific joints is changing.

RANGE OF JOINT MOTION: This is done to monitor if contractures or joint tightening is developing and to help to guide what stretches or interventions will be most helpful.

TIMED TESTS: Many clinics routinely time activities such as the time to get up off the floor, time to walk a certain distance, and time to climb several steps. This gives important information on how the condition is changing and how it is responding to treatment.

MOTOR FUNCTION SCALES: There are a large number of different scales, but your clinic should routinely use the same one to monitor the condition in a systematic way. Different scales may be needed at different times.

ACTIVITIES OF DAILY LIVING: This allows the team to tell if some additional help might be needed to assist independence.

IMPORTANT FACTS TO REMEMBER:

1. Because your son does not have dystrophin, his muscles will gradually get weaker.
2. Some types of exercise and getting tired can make muscle damage worse.
3. The doctor understands the progression of muscle weakness and can help your son get ready for the next step.
4. It is important for the doctor to know how your son's muscles are working so that they can start the right therapy as early as possible.

Drug treatments for muscle symptoms

There is a lot of research happening at the moment in the area of new drugs for DMD. In this document the experts only gave recommendations where there is already sufficient evidence for a treatment. These recommendations will change in future when new evidence (such as the results of clinical trials) becomes available. The guidelines will be reviewed as new results become available.

Although it is expected that in future a wider range of treatment options will be available, at the present time, the only drug treatment for the musculoskeletal symptoms of DMD that the experts agreed there was sufficient evidence to be able to recommend is steroid treatment. Steroids are discussed in detail in this section. Drug treatments for other specific symptoms of the disease, such as heart problems, are discussed later.

Steroid treatment – a step by step guide

Steroids are used in many other medical conditions and there is a lot of experience in their use worldwide. There is no doubt they can benefit many boys with DMD but this benefit needs to be balanced with proactive management of possible side effects. Use of steroids is very important in DMD and should be discussed with all families early.

THE BASICS

- **Steroids (also called glucocorticoids or corticosteroids) are the only drugs known to slow the decline in muscle strength and motor function in DMD.** The goal of steroid use is to help the child walk independently for longer to allow enhanced participation and to later minimise breathing, heart and orthopaedic problems. They can also reduce the risk of scoliosis (curvature of the spine).
- **Prevention and management of steroid side effects needs to be proactive and anticipatory. Interventions should be put in place EARLY in an effort to prevent problems and to make sure they do not become severe.** Side effects associated with steroid use vary and are listed in Table 1.

STARTING AND STOPPING STEROIDS

- **The optimal time for starting steroid treatment is when motor function is in a "plateau phase"** – when the boy's motor skills have stopped improving, but have not yet started to get worse. This is normally sometime between the age of 4-6 years. It is not recommended to start steroids in children who are still gaining motor skills, especially if they are under 2 years of age.
- The **recommended national vaccination schedule should be complete before steroid treatment is started**, and varicella (chicken pox) immunity should be established.
- **Starting steroid treatment in boys/young men who are no longer walking independently is a matter for individual decision and needs to be discussed with the doctor, taking into consideration the effect of pre-existing risk factors.** In boys who used steroids when they were walking, many experts recommend continuation of medication after loss of ambulation. The goal in the non-ambulatory person is to preserve upper limb strength, slow the progression of scoliosis, and delay the decline of respiratory and cardiac function.

IMPORTANT FACTS TO REMEMBER:

1. Steroids are the only medicines known to help slow down muscle weakness.
2. Always tell doctors and other healthcare providers that your son is taking steroids. It is especially important if he is having surgery or has an infection or injury because steroids can suppress the immune system.
3. Your son should never stop taking steroids suddenly.
4. Your son should have regular visits with a doctor who is skilled in managing steroids. The doctor will explain possible side effects and tell you if your son is at risk of developing them.

The different steroid regimes

One of the potentially confusing things in DMD care is that different doctors and different clinics often prescribe different regimens of steroids, which means you will find information about different drugs and different regimes. These guidelines have tried to establish a clear route to use steroids effectively and safely based on regular assessments of function and side effects (see Box 2).

- **Prednisone (prednisolone) and deflazacort** are the two types of steroids that are mainly used in DMD. They are **believed to work similarly**. Neither one is clearly better. Planned trials of these drugs are important and should help us understand them better in future.
- The choice of which steroid to use depends upon availability in a particular country, the cost to the family, the way the drug is taken, and the perceived side effects. Prednisone has the advantage of being inexpensive and is available in both tablet and liquid formulation. Deflazacort may be preferred to prednisone for some individuals because there may be a slightly lower risk of weight gain.
- Starting with daily use of a steroid was preferred by the experts to the alternative regimes. Data from ongoing and future studies may modify this recommendation.

Box 2

Doses for starting and maintaining steroids

- **The recommended starting dose of prednisone is 0.75 mg/kg/day and that of deflazacort is 0.9 mg/kg/day**, given in the morning. Some children experience short-lived behavioural side effects (hyperactivity, mood swings) for a few hours after the medication is given. For these children, administration of the medication in the afternoon may alleviate some of these difficulties.
- **For ambulatory individuals, the dosage is commonly increased as the child grows until he reaches approximately 40 kg in weight.** The maximum dose of prednisone is usually capped at approximately 30 mg/day, and that of deflazacort at 36 mg/day.
- **Non-ambulatory teenagers** maintained on long-term steroid therapy are usually above 40 kg in weight and **the prednisone dosage per kg is often allowed to drift down to the 0.3 to 0.6 mg/kg/day range**. While this dosage is less than the approximate 30 mg cap, it demonstrates substantial benefit.
- Deciding on the maintenance dose of steroids is a balance between growth, how good the response to steroids is and the burden of side effects. So this decision needs to be reviewed at every clinic visit based on the result of the tests done and whether or not side effects are a problem that can't be managed or tolerated.
- **In boys on a relatively low dosage of steroids (less than the starting dose per kg body weight) who start to show functional decline, it is necessary to consider a "functional rescue" adjustment.** The dosage of steroids is increased to the target and the individual is then re-evaluated for any benefit in approximately two to three months.
- **There is no consensus on the optimal steroid dosage if initiated in the non-ambulatory individual.** Nor is it known how effective steroid treatment is in preventing scoliosis or in stabilising cardiac or respiratory function in this setting. This issue warrants further study.

Box 3

Management of steroid medication

- **A dose reduction of approximately ¼ to ⅓ is suggested if intolerable or non-manageable side effects occur**, with a reassessment by phone or clinical visit in one month to assess control of side effects.
- If a daily dosing schedule results in **unmanageable and/or intolerable side effects that do not improve when the dose is reduced**, then it is appropriate to **change to an alternative regime**.
- **Steroid therapy should not be abandoned even if side effects are NOT manageable and/or tolerable until at least one dosage reduction and change to an alternative regime have been pursued.** This recommendation holds for both ambulatory and non-ambulatory individuals.
- **Should adjustments to the steroid dosing and/or schedule regimens prove ineffective** in making any significant side effects sufficiently manageable and tolerable, then it is necessary to **discontinue steroid therapy**. These decisions need to be made individually in partnership with the child and family. **Steroids should never be stopped suddenly.**

Steroid management and side effects
(Boxes 3 and 4 and Table 1)

Attentive management of steroid-related side effects is crucial once a boy has started on long-term steroid therapy. While steroid therapy is currently the mainstay of medication therapy for DMD, it should not be undertaken casually by the doctor or family, and should be undertaken only by doctors with appropriate expertise.

Box 4

Other drugs and dietary supplements

The experts considered a range of other drugs and supplements that are known to be used in some cases for DMD treatment. They reviewed published data on these substances to see if there was enough evidence for their safety and efficacy to be able to make recommendations.

The experts concluded the following:

- The use of **oxandrolone**, an anabolic steroid, is not recommended.
- Safety in the use of **Botox** has not been studied for the treatment or prevention of contractures in individuals with DMD and is not recommended.
- There was no support for the systematic use of creatine. A randomised controlled trial of **creatine** in DMD did not show a clear benefit. If a person is taking creatine and has evidence of kidney problems, it is necessary to discontinue this supplement.
- **No recommendations** can be made at this time about other supplements or other drugs that are sometimes used in DMD treatment, including co-enzyme Q10, carnitine, amino acids (glutamine, arginine), anti-inflammatories/anti-oxidants (fish oil, vitamin E, green tea extract, pentoxifylline), and others including herbal or botanical extracts. The experts concluded that there was not enough evidence in the published literature.
- The experts agreed that this is an area where additional research is needed. Active involvement of families in activities that develop further knowledge, such as patient registries and clinical trials, was encouraged.

Steroids are the only drugs that the experts have agreed can be recommended. Though some of the drugs mentioned in Box 4 are quite widely used, there is just not enough evidence to say whether these other supplements really work or not. It is important to discuss all medication with your doctor before you think about adding or stopping medication.

Table 1 (overleaf) summarises the main side effects of steroids that should be monitored and useful interventions. Factors to take into account in maintaining or increasing dose are response to therapy, weight and growth, and whether side effects are present and manageable.

Table 1

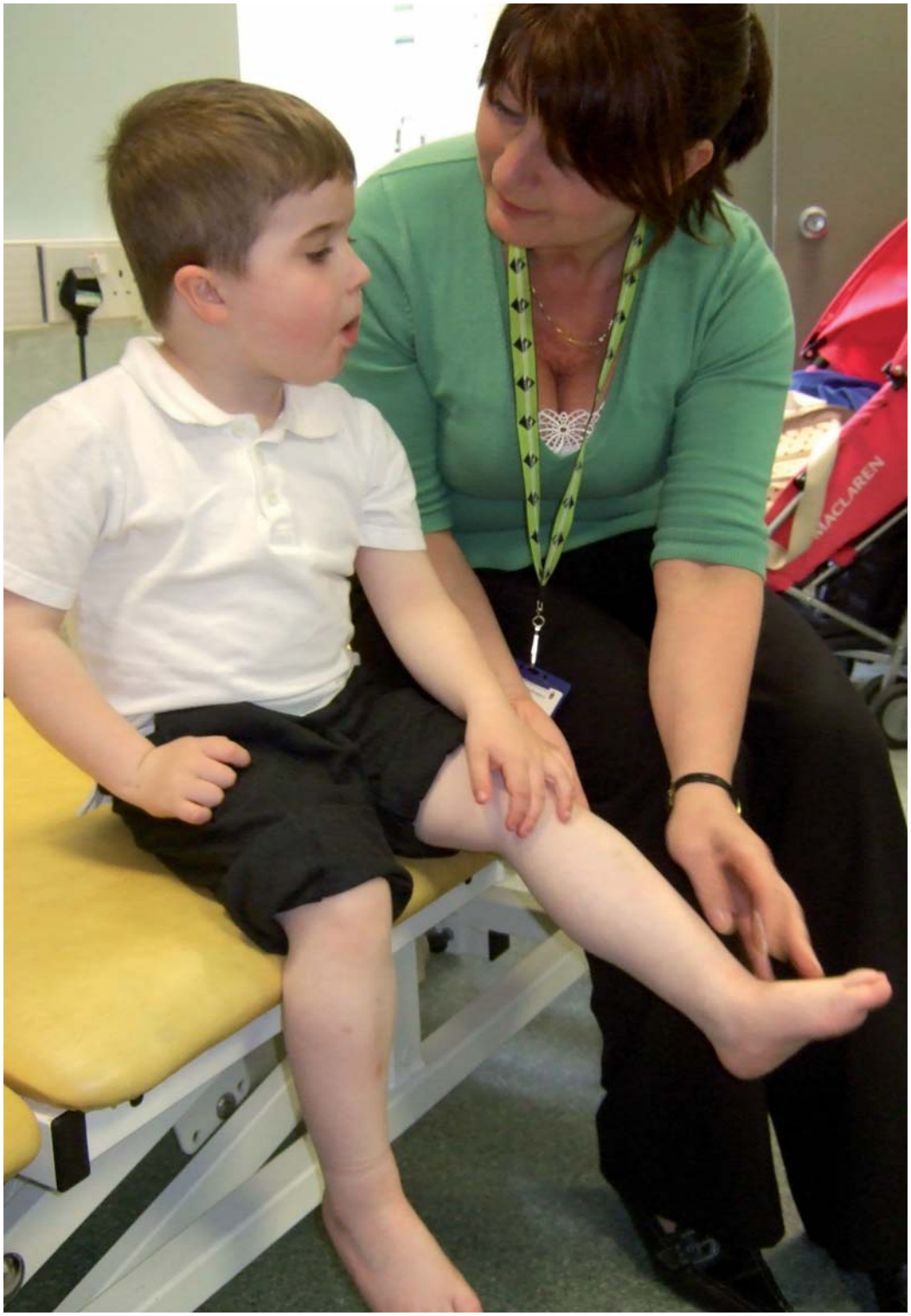
STEROID SIDE EFFECTS: RECOMMENDED MONITORING AND INTERVENTION

Some of the more common long-term side effects of high-dose steroid administration in growing children are listed here. It is important to note that different people will have very different responses to steroids. The key to successful steroid management is to be aware of the potential side effects and work to prevent them or reduce them where possible. Reduction in steroid dose is necessary if side effects are unmanageable or intolerable. If this is unsuccessful, then further reduction or a change to another dosing regimen is necessary before abandoning treatment altogether.

STEROID SIDE EFFECT	COMMENT AND RECOMMENDED MONITORING	POINTS FOR YOU TO THINK ABOUT AND TO DISCUSS WITH YOUR DOCTOR
General and cosmetic Weight gain Obesity	Dietary advice needs to be provided to all families before starting a steroid regimen. They should be warned that steroids increase appetite.	It is important that the whole family eat sensibly in order to prevent excess weight gain. Look for advice for the entire family regarding diet and nutrition.
Cushingoid features ("moon face")	Fullness in the face and cheeks becomes more noticeable over time.	Careful monitoring of diet and restricting sugar and salt intake will help with weight gain and may minimise Cushingoid features.
Excessive growth of hair on the body (hirsutism)	Clinical examination.	This is not usually severe enough to warrant a change in medication.
Acne, Tinea, Warts	More noticeable in teenagers.	Use specific treatments (topical prescription) and do not rush to change the steroid regimen unless there is emotional distress.
Growth retardation	Monitor height at least every 6 months as part of general care (height tends to be small in DMD even without steroid treatment.)	Ask if your son is concerned about his short stature. If so, you should discuss with your doctor if he needs an endocrine check up.
Delayed puberty	Monitor development. Identify any family history of delayed sexual maturation.	Encourage discussion about puberty. Ask your son if he has concerns about any delay. Discuss with your doctor about getting an endocrine evaluation if you or your son are concerned.
Adverse behavioural changes (There is a lot more information about behaviour in Section 10 of these recommendations)	Identify any baseline mood, temperament, and ADHD issues. Be aware that these often temporarily worsen in the initial six weeks on steroid therapy.	Consider if baseline issues should be treated prior to starting steroid therapy, e.g. ADHD counselling or prescription. It may help to change the timing of steroid medication to later in the day – discuss this with your doctor, who may also consider a behavioural health referral.
Immune / adrenal suppression	Be aware of risk of serious infection and the need to promptly address minor infections. Inform all medical personnel that the child is on steroids, and carry steroid alert card. Ensure that the steroid is not stopped abruptly. It is very important that someone on chronic steroids does not miss their dose for more than 24 hours at the most, especially if they are also unwell.	Obtain chicken pox immunisation prior to starting steroid therapy; if not done seek medical advice if in contact with chicken pox. If there is a regional problem with TB, there may need to be specific surveillance. Discuss with your doctor how you would cope if there was a break in taking steroids, for example substituting prednisone equivalent if deflazacort is temporarily unavailable, or how you might need IV coverage during illness or fasting. Discuss use of intravenous (IV) "stress dose" methylprednisolone coverage for surgery or major illness. Give IV coverage if fasting.

STEROID SIDE EFFECT	COMMENT AND RECOMMENDED MONITORING	POINTS FOR YOU TO THINK ABOUT AND TO DISCUSS WITH YOUR DOCTOR
Hypertension	Monitor blood pressure (BP) at each clinic visit	If BP is elevated, reducing salt intake and weight reduction can be useful first steps. If ineffective, your doctor will need to consider possible ACE or beta-blocker medication.
Glucose intolerance	Test urine for glucose with dipstick test at clinic visits. Enquire about increased passage of urine or increased thirst.	Blood tests may be needed if urine tests are positive.
Gastritis/ gastroesophageal reflux	Look out for reflux symptoms (heartburn).	Avoid non-steroidal anti inflammatory drugs (NSAIDs) - such as aspirin, ibuprofen, naproxen. Drugs and antacid can be used if symptoms occur.
Peptic ulcer disease	Report symptoms of stomach pain as this can be a sign of damage to the lining of the stomach. Stool can be checked for blood if anaemic or suggestive history.	Avoid NSAIDs (aspirin, ibuprofen, naproxen). Drugs and antacid can be used if symptomatic. Seek gastrointestinal consultation.
Cataracts	Annual eye exam.	Consider switching from deflazacort to prednisone if cataracts evolve that affect vision. Seek ophthalmology consultation. Cataracts will only need to be treated if they interfere with vision.

STEROID SIDE EFFECT	COMMENT AND RECOMMENDED MONITORING	POINTS FOR YOU TO THINK ABOUT AND TO DISCUSS WITH YOUR DOCTOR
Bone demineralisation and increased fracture risk	Take careful fracture history. Yearly DEXA to monitor bone density. Yearly vitamin D blood level (ideally late winter in seasonal climates) and supplement with vitamin D3 if levels are low. Dietician assesses calcium and vitamin D intake.	Vitamin D supplements may be needed depending on level in blood. Recheck vitamin D level again after 3 months on therapy. Weight-bearing activities can be helpful. Make sure that calcium intake is good in the diet and if not supplements may be needed.
Myoglobinuria (Urine looks coca-cola coloured because it contains breakdown products of muscle proteins. This needs to be tested for in a hospital lab.)	Enquire about abnormal colouration of urine after exercise – urine testing.	Avoid vigorous exercise and eccentric exercises, such as running downhill or trampolining. Good fluid intake is important. Kidney investigations are needed if it carries on.



5 Rehabilitation management - physiotherapy and occupational therapy

People with DMD need access to different types of rehabilitation management throughout their lives. Much of this will be delivered by physiotherapists and occupational therapists, but other people may also need to help, including rehabilitation specialists, orthotists, providers of wheelchairs and other seating. Orthopaedic surgeons may also be involved.

Management of muscle extensibility and joint contractures is a key part of rehabilitation management.

The goal of stretching is to preserve function and maintain comfort. The program of stretching will be monitored by the physiotherapist but needs to become part of the family's daily routine.

There are many factors in DMD that contribute to the tendency for joints to get tight or "contracted". These include the muscle becoming less elastic due to limited use and positioning or because the muscles around a joint are out of balance (one stronger than another). Maintaining good range of movement and symmetry at different joints is important. This helps to maintain the best possible function, prevent the development of fixed deformities, and prevent pressure problems with the skin.

Box 5

Management of muscle extensibility and joint contractures

- The key contact for management of joint contractures is your physiotherapist. Ideally input from a local physiotherapist will be backed up by a specialist physiotherapist about every 4 months. Stretching should be performed at least 4-6 times each week and should become part of the daily routine.
- Effective stretching to counteract development of contractures may require different techniques which your physiotherapist will show you, including stretching, splinting and standing devices.
- Regular stretching at the ankle, knee, and hip is important. Later on, regular stretching for the arms becomes necessary, especially the fingers, wrist, elbow and shoulder. Additional areas that require stretching may be identified on individual examination.
- Night splints (ankle-foot orthoses or AFOs) can be used to help control contractures in the ankle. These need to be custom-made and not provided 'off the shelf'. After the loss of ambulation, daytime splints may be preferred, but daytime splints are not recommended for boys who are still walking.
- Long leg splints (knee-ankle-foot orthoses or KAFOs) may be useful around the stage when walking is becoming very difficult or impossible. KAFOs can be useful to help control joint tightness and to prolong ambulation and delay the onset of scoliosis.
- Standing programs (in a standing frame or power chair with stander) are recommended after walking becomes impossible.
- Resting hand splints are appropriate for individuals with tight long finger flexors.
- Surgery can be offered in some situations in an effort to prolong the period of walking. However, this approach must be strictly individualised. More information about the different options is available in the main document.

Wheelchairs, seating and other equipment

- **During the early ambulatory stage, a scooter, stroller or wheelchair may be used for long distances to conserve strength.** When your son starts using a wheelchair for longer periods, it becomes more important that posture is carefully looked at, and customisation of the chair is usually necessary.
- As difficulty with walking increases, it is recommended that a power wheelchair is provided sooner rather than later. Ideally, the initial power wheelchair should be adapted and customised to optimise comfort, posture and symmetry. Some experts also recommend a power standing feature if available.

- With time, arm strength becomes more of an issue. Physiotherapists and occupational therapists will be helpful in recommending assistive devices to help maintain independence. It is best to think proactively about the kind of equipment that will best support independence and participation and plan ahead to provide it in as timely a manner as possible.
- Additional adaptations in the late ambulatory and non-ambulatory stages may be needed to help with getting upstairs and transferring, eating and drinking, turning in bed and bathing.

Box 6

Pain management

It is important to ask boys/young men with DMD if pain is a problem so that it can be addressed and treated properly. Unfortunately, very little is currently known about pain in DMD. More research is needed. If your son is in pain you need to talk to your doctor and explain to them that this is a problem.

- **For effective pain management, it is important to determine why there is pain** so the doctors can provide appropriate interventions.
- As a lot of pain results from problems with posture and difficulty getting comfortable, interventions include **provision of appropriate and individualised orthoses (braces), seating, bedding, and mobility as well as standard drug treatment approaches** (e.g. muscle relaxants, anti-inflammatory medications). **Interactions** with other medications (e.g. steroids and non-steroidal anti-inflammatory drugs [NSAIDs]) and associated side effects, especially those that might affect cardiac or respiratory function, should be considered.
- Rarely, **orthopaedic intervention** might be indicated for pain that cannot be managed in any other way but that might respond to surgery. **Back pain**, especially in people using steroids, means the doctors should **check carefully for vertebral fractures, which respond well to bisphosphonate treatment**.

6 Orthopaedic management - help with bone and joint problems

People with DMD who are not treated with steroids have a 90% chance of developing progressive scoliosis (a sideways curvature of the spine that gets worse as time goes on). Daily steroid treatment has been shown to reduce the risk of scoliosis or at least delay its onset.

Proactive management of the risk of scoliosis requires:

Surveillance

- **Spinal care should include monitoring for scoliosis. This is done by clinical observation throughout the ambulatory phase** and with a spinal X-ray only if scoliosis is observed. In the non-ambulatory phase, clinical assessment for scoliosis is essential at each clinic visit.
- **Spinal radiography (X-ray) should be done as a baseline assessment around the time of becoming wheelchair-dependent.** Special X-rays getting two views of the full spine are needed. Follow up X-rays should be done at least once per year if there is a problem. Gaps of greater than one year between X-rays have the risk of missing a worsening of scoliosis. After growth has stopped X-rays are only needed if there is any change clinically.

Prophylaxis (preventive measures)

- **Attention to posture at all times: prevention of asymmetrical contractures in boys who are still walking, proper seating system in the wheelchair** giving support of spinal and pelvic symmetry and spinal extension. Spinal bracing is not appropriate to try and delay surgery but may be used if surgery cannot be done or is not the chosen option.

IMPORTANT FACTS TO REMEMBER:

1. Boys and young men with DMD have weak bones, especially if they are taking steroids.
2. It is important for your son to have the right amount of calcium and vitamin D to help keep his bones strong.
3. The doctor should watch your son's spine closely after he stops walking, especially while he is still growing, as scoliosis can change quickly.
4. Key to success of spinal surgery, if it is needed, is the identification of an experienced surgeon and proper attention to the breathing muscles and heart.
5. If your son has back pain he should see the doctor.

Treatment

- **Surgery with posterior spinal fusion** is indicated when the degree of the curve (known as the Cobb angle) is greater than 20° in boys who have not yet stopped growing and who are not taking steroids. The aim of surgery is to preserve the best possible posture for comfort and function. When boys are taking steroids, there is less risk of deterioration and the decision to proceed to surgery can be left until the Cobb angle is greater than 40°.
- It is important to discuss what type of operation is needed with your surgeon and express any concerns you may have.

Bone health management

- Bone health is important in both the ambulatory and non-ambulatory phases of DMD. Boys with DMD at all ages have weak bones, especially if they are taking steroids. They have a lower bone mineral density and are at increased risk of fractures (broken bones) compared to the general population.

Long bone fracture management

- A broken leg can be a significant threat to the continued ability to walk. This is why treatment with surgery should be considered to allow the boy with DMD to get back up on his feet as soon as possible. If a fracture does occur, make sure that your physiotherapist is notified.
- **If a boy who is still walking breaks his leg, internal fixation (that is surgery to stabilise the break as quickly as possible)** is needed to resume walking and to have the greatest possible chance to maintain ambulation.
- In boys who are no longer walking, a broken leg can be safely treated by **splinting or casting, taking into account the functional position of the limb and possible development of contractures.**

Bone health in general

- Steroid treatment is known to add to the risk of low bone density and is also associated with the risk of fractures of the spinal vertebrae. Fractures of the vertebrae are not usually seen in non-steroid treated boys. Bone density may need to be assessed with blood tests, bone scans and other X-rays (see Box 7). This is an area where further research is needed to establish the parameters for best practice.

Box 7

Bone health management

UNDERLYING FACTORS FOR POOR BONE HEALTH ARE:

- Decreased mobility
- Muscle weakness
- Steroid therapy

POSSIBLE INTERVENTIONS:

- Vitamin D – needed if there is a real deficiency, supplement should be considered in children.
- Calcium – intake is best in the diet, but supplementation should be considered if diet is not adequate with advice from a dietician.
- Bisphosphonates – IV bisphosphates recommended for vertebral fractures.

7 Pulmonary management - looking after the breathing muscles

Usually boys do not have trouble breathing or coughing while they are still walking. Because the breathing muscles become affected, as boys with DMD get older they are at risk of chest infections, often due to an ineffective cough. Later on they develop problems with their breathing when sleeping. When they are older, they may require help with breathing during the day as well. As this is a staged progression of problems, a planned and proactive approach to respiratory care is possible based around appropriate surveillance, prophylaxis and interventions. The team must include a doctor and therapist with skill in looking after the delivery of non-invasive ventilation and associated techniques for increasing the amount of air that can enter the lungs (lung volume recruitment), and manual and mechanically assisted cough.

IMPORTANT FACTS TO REMEMBER:

1. Keep a copy of your son's latest breathing tests to show any doctor who takes care of him.
2. Your son should never be given inhaled anaesthesia or the drug succinylcholine.
3. Your son's lung function should be checked before surgery. It is good to pick up on silent problems so that they can be treated promptly.
4. Your son will need help with coughing and antibiotics if he has a chest infection.
5. Symptoms of hypoventilation and weak cough should be monitored for and be reported to the medical caregivers so that therapy can be initiated.
6. If your son's oxygen level drops when he is ill or injured, the doctor must be very careful giving him oxygen because this can cause a situation where his own urge to breathe is decreased.

Surveillance

- While a boy with DMD is still walking, minimal assessment of pulmonary function (such as measurement of forced vital capacity [FVC] at least annually) allows the child to become familiar with the equipment and the team to assess the maximum respiratory function achieved.
- The **main emphasis of pulmonary assessment is after the loss of independent walking**, and should include FVC measurement and peak cough flow. Other measures may also be useful, including studies of oxygen levels during sleep, and should be introduced as time goes by. Assessment frequency will depend on the stage of the condition, but at a minimum FVC measurement should be done **at least every 6 months**.

It is very important to look out for the kinds of signs that suggest your son may be having trouble breathing as he gets older. If you think you are seeing any of these you need to report them to your doctor. Contact your doctor if your son:

- is experiencing prolonged illness with apparently minor upper respiratory infections. For example, recovery from common colds is slow, with colds progressing to chest congestion and bronchitis, often requiring antibiotic therapy;
- is more tired than usual;
- is short of breath, acts as if he cannot catch his breath or has difficulty finishing sentences;
- has headaches all the time or in the morning;
- is often sleepy for no reason;
- has trouble sleeping, wakes up a lot, has trouble waking up or has nightmares;
- wakes up trying to catch his breath or says he can feel his heart pounding;
- has trouble paying attention.

Prevention of problems

- **Immunisation** with pneumonia vaccine is indicated for persons two years of age and older and may need to be repeated according to local policy. Annual immunisation with **influenza** vaccine is indicated. Both can be given to individuals treated with steroids, though the immune response to vaccination may be diminished in those individuals. **Up-to-date, detailed information on immunisation indications, contraindications, and schedules can be obtained from various national sources** – see the "resources" section at the end of this document. **It is essential to keep up to date with vaccination policies as they can change regularly according to new threats**, such as the emergence of H1N1 flu in 2009.
- If chest infection occurs, then in addition to use of manually and mechanically assisted cough, **antibiotics should be considered**.

Interventions (this requires special expertise)

- **Interventions** are dependent on disease phase. First of all, it may be helpful to use ways to increase the amount of air that can enter the lungs through deep breathing **(lung inflation techniques). As DMD progresses, coughing will become less effective**, and ways to improve this can be very helpful, **such as with manual and assisted cough techniques**. With time, support will be needed **initially for breathing overnight and then later during the daytime (non-invasive nocturnal / daytime ventilatory support) as symptoms listed under the surveillance section develop. Support of breathing through the use of non-invasive ventilation is a very important way to maintain health. Ventilation may also be delivered via a surgically placed tube in the neck (tracheostomy tube)** depending on local practice (this is known as invasive ventilatory support). All these interventions can help to keep people healthy and avoid acute illnesses.
- Particular attention to the breathing is required around the time of planned surgery (see Section 11 regarding **respiratory considerations for surgery**).

Box 8

IMPORTANT - CAUTION

- **In the later stages of DMD supplemental oxygen therapy should be used with caution.**
- While oxygen therapy can apparently improve low oxygen levels, using oxygen will mask the underlying cause, such as a collapsed lung or poor breathing.
- Oxygen therapy may reduce the drive to breathe and lead to carbon dioxide retention.
- Manual and mechanically assisted cough and non-invasive ventilatory support are likely to be necessary. **Using oxygen therapy instead is not recommended and may be dangerous**.
- If oxygen is given, and it may sometimes have to be, then there needs to be very careful monitoring of the gases in the blood and/or help with the breathing should be given at the same time.

8 Cardiac management - looking after the heart

The aim of cardiac management in DMD is early detection and treatment of the deterioration of heart muscle function (usually cardiomyopathy - involvement of heart muscle, or rhythm problems leading, for example, to palpitations) that commonly accompanies the overall progression of the disease. As this often happens silently (that is without the development of significant symptoms) it needs to be looked out for so it can be treated promptly. The key factors to consider in cardiac management are surveillance and proactive management. You need to be sure that there is a cardiologist involved with the care team.

IMPORTANT FACTS TO REMEMBER:

1. Your son's heart should be checked regularly starting from the time he is diagnosed.
2. In DMD the heart may be already damaged before symptoms appear.
3. This means that your son may need to start heart medication even if he does not have symptoms of heart problems.
4. It is good to pick up silent problems so that they can be treated promptly.
5. Keep a copy of your son's latest heart tests to show any other doctor who may see your son.

Surveillance

- **Baseline evaluation** of cardiac function should be performed **at the confirmation of the diagnosis or at latest by the age of six years**. Minimum evaluation should include an **electrocardiogram (ECG)** and **echocardiogram**.
- **Evaluation of cardiac function should occur at least once every two years until the age of ten. Yearly complete cardiac evaluations should begin at approximately ten years of age or at the onset of cardiac signs and symptoms**, if earlier. If non-invasive cardiac tests show abnormalities, increased surveillance is required, at least every six months, and drug treatment should be initiated.

Treatment

- **Angiotensin converting enzyme (ACE) inhibitors should be considered as first-line therapy**. Other medicines such as beta-blockers and diuretics are also appropriate and should follow published guidelines for the management of heart failure. There is some evidence from clinical trials to support the **prophylactic treatment of cardiomyopathy with ACE inhibitors prior to any signs of abnormal functioning**. Further studies are awaited to allow firm recommendations in this regard.

- **Abnormalities of heart rhythm should be promptly investigated and treated.** A fast heart rate is a commonly noted harmless feature of DMD, but can also be seen with heart problems. If it develops as a new finding it should be investigated.
- Individuals undergoing **treatment with steroids need additional attention** from the cardiovascular perspective, especially monitoring for **hypertension (high blood pressure)**. Steroid dose may require adjustment or further treatment may need to be added (see Table 1).

9 Gastrointestinal management - nutrition, swallowing and other gastrointestinal issues

Access to the following experts may be needed at different stages: a dietician or nutritionist, a swallowing/speech and language therapist, and a gastroenterologist.

Nutritional management

- **Thinking ahead to maintain good nutritional status to prevent both under nutrition and overweight is essential from diagnosis throughout life.** It is important that weight for age or body mass index for age is kept between the 10th and 85th percentile on national percentile charts (see resources section). Provide **a well-balanced diet with a full range of food types**. Information for the whole family on eating a well-balanced diet can be found from most national sources.
- Boys should be monitored regularly for their weight and height (which can be calculated from arm measurement in non-ambulatory boys). The triggers for referral to an expert dietician/nutritionist are if a boy is overweight or underweight, if he is experiencing unintentional weight loss or gain, or poor weight gain, if major surgery is planned, if he has chronic constipation and/or if he has difficulty swallowing (dysphagia). Referral will also be made at diagnosis and when starting steroids. The diet should also be assessed for calories, protein, fluid, calcium, vitamin D, and other nutrients.
- **It is recommended that people with DMD take a daily multivitamin with vitamin D and minerals.**
- If there is weight loss, it is important to look for **problems with swallowing**. However it is important to note that **complications in other systems, such as cardiac or respiratory systems, may contribute to weight loss. If there is unexpected weight loss, it may be important to check out other areas as well**.

IMPORTANT FACTS TO REMEMBER:

1. Your son's height and weight should be checked at every visit to the doctor.
2. It is important for your son to have a well-balanced diet, especially one that includes the right amount of calcium and vitamin D.
3. Nutritionists and dieticians are important members of your son's healthcare team, who can check your son's diet and help him eat better.
4. Your son should be evaluated if he has signs of swallowing problems.
5. Getting a gastrostomy tube is another option after trying other ways to maintain your son's weight.

Swallowing management

In later stages, weakness of the throat muscles can lead to swallowing problems (dysphagia), further accentuating nutritional issues. This can often come on very gradually, meaning it can be difficult to spot.

- **Clinical and X-ray tests of swallowing are necessary when there are clinical indicators of possible aspiration (getting food in the windpipe) and poor movement of the swallowing muscles (food feels like it is getting stuck in the throat)**. Such indicators include unintentional weight loss of 10% or more, or insufficient weight gain in growing children, prolonged meal times (>30 minutes) or mealtimes accompanied by fatigue, drooling, coughing or choking.
- **Pneumonia caused by fluid going down into the lungs (aspiration pneumonia), unexplained decline in pulmonary function, or fever of unknown origin may be signs of swallowing problems** necessitating assessment.
- In case of swallowing problems, **a Speech Language Therapist (SLT) should be involved to deliver an individualised treatment plan**. The aim is to preserve good swallowing function.
- Gastric tube placement should be offered when efforts to maintain weight and fluid intake by mouth do not help enough. Potential risks and benefits of the procedure should be discussed carefully. A gastrostomy may be placed by endoscopic or open surgery, taking into account anaesthetic considerations and family and personal preference. A feeding tube provided at the right time can relieve a lot of pressure from trying to eat enough. Provided the swallowing muscles are OK, having a feeding tube doesn't mean you can't still eat the food you want to – just that you don't have to rely on mealtimes to get the calories and other nutrients you need so you can enjoy the food more.

Other areas of gastrointestinal management

Constipation and gastroesophageal reflux (which causes heartburn) are the two most common gastrointestinal conditions seen in individuals with DMD. Constipation typically occurs at an older age and after surgery. With increasing survival, other complications are being reported, including gastric and intestinal swelling related to air swallowing due to ventilator use.

- **Laxatives and other medicines can be useful**. It is important that there is enough fluid intake. Increasing fibre may make symptoms worse especially if fluids are not increased.
- **Reflux is typically treated with appropriate drugs**. Acid blockers are commonly prescribed to children on steroid therapy or oral bisphosphonates to avoid complications.
- **Oral care is an important area**, and although this was not included in the published International consensus on the care and management of DMD, TREAT-NMD have developed expert recommendations for oral care that are outlined in Box 9.

Box 9

Oral care recommendations

- Boys with DMD should see a dentist with extended experience and detailed knowledge of the disease, preferably at a centralised or specialist clinic. The dentist's mission should be to strive for high-quality treatment, oral health and wellbeing and to function as a resource for the families and the boy's own dentist in his home community. This dentist should be aware of the specific differences in dental and skeletal development in boys with DMD and collaborate with a well informed and experienced orthodontist.
- Oral and dental care is to be based on prophylactic measures with a view to maintaining good oral and dental hygiene.
- Individually adapted assistive devices and technical aids for oral hygiene are of particular importance when the muscular strength of the boy's hands, arms and neck begins to decrease.

10 Psychosocial management - help with behaviour and learning

People with DMD may have an increased risk of psychosocial difficulties, such as problems with behaviour and learning, and medical care is not complete without support for psychosocial wellbeing. Difficulties in social functioning may be due to specific challenges in particular skills, such as getting on with others, judging social situations, and perspectives, while the consequences of DMD (such as physical limitations) may result in social isolation, social withdrawal, and reduced access to social activities. For many parents, the stress caused by the psychosocial problems of the child and difficulties in getting them recognised and properly treated exceeds the stress associated with the physical aspects of the disease.

IMPORTANT FACTS TO REMEMBER:

1. The psychosocial health of your son and your family is important.
2. Your son may have a higher chance of having psychosocial difficulties.
3. You and your family are at risk of some problems such as depression.
4. The best way to manage psychosocial problems is to identify them early and start treatments.
5. Correct use of language may be a problem, as may continuing difficulties at school. These behaviors are often seen in DMD and can be helped with proper assessment and input.
6. Learning problems in DMD are not progressive and most boys catch up when they receive good help.

If you think your child has worries about his condition, openness and a willingness to answer his questions can go a long way to preventing further problems. Boys with DMD often understand more about their condition than their parents think. It is important to answer questions openly, but be age-appropriate in your answers and just answer what is being asked. This can be very difficult, but the staff at your clinic can offer help and guidance about what has worked for other families, as can patient support groups.

Not everyone with DMD will have psychosocial difficulties, but families should keep an eye out for:

- Weaknesses in language development, comprehension, and short-term memory;
- Learning problems;
- Difficulty with social interactions and/or making friendships (i.e., social immaturity, poor social skills, withdrawal or isolation from peers);
- Anxiety/worry;
- Frequent arguing and temper tantrums;
- There is also increased risk of neurobehavioural and neurodevelopmental disorders, including autism-spectrum disorders, attention-deficit/hyperactivity disorder (ADHD), and obsessive-compulsive disorder (OCD);
- Problems may be encountered with emotional adjustment and depression. Anxiety may also be an issue and can be made worse by deficits in mental flexibility and adaptability (i.e. an overly-rigid thought process);

- This can also result in oppositional/ argumentative behaviour and temper problems;
- In addition, increased rates of depression in parents of children who have DMD underscore the need for assessment and support of the entire family.

The **emphasis in psychosocial management should be strongly on prevention of problems and early intervention**, as this will maximise the potential outcome. In general, the psychosocial problems should be treated with the same effective, evidence-based interventions that are used in the general population. This means it is important to look for help if you think there are problems in this area.

Box 10

Speech and language management – The details:

- There is a well-documented pattern of **speech and language deficits in some children with DMD**, including problems with language development, short-term verbal memory, and phonological processing, as well as impaired IQ and specific learning disorders. These do not affect all children with DMD but should be looked out for and helped if they are present.
- Delay in attainment of early language milestones is common in boys who have DMD compared to children of the same age. The differences in gaining and improving language may be something that can be seen across childhood. It is important that this problem is looked for and treated. **Evaluate and treat for delayed speech and language problems**.
- **Referral to a Speech Language Therapist (SLT) for speech and language evaluation and treatment is** necessary if problems in this area are suspected.
- **Exercises for the muscles involved in speech and help with articulation** are appropriate and necessary for both young boys who have DMD with difficulties in this area and in older individuals who have deteriorating oral muscle strength and/or impaired speech intelligibility.

- For older individuals, **compensatory strategies, voice exercises, and speech amplifications** are appropriate if it becomes difficult to understand the person with DMD due to problems with respiratory support for speech and vocal intensity. Voice Output Communication Aid (VOCA) assessment may be appropriate at all ages if speech output is limited.

Assessments

Although the needs of each child will vary, crucial times to consider assessments include at or near the time of diagnosis (a 6- to 12-month window for some evaluations may be beneficial in order to allow for adjustment following diagnosis), prior to entering school, and following a change in functioning. While not every clinic will have direct access to all of the assessments and interventions listed, these recommendations can serve as a guide to filling gaps in clinical staff and directing referrals, where appropriate.

- Areas of emotional adjustment and coping, development in learning relative to age, speech and language development, the possible presence of autism spectrum disorders, and social support should be assessed. (A social services professional can help access financial resources, develop social support networks, or provide mental health support to the family as needed).
- The psychosocial wellbeing in the individual with DMD, parents, and siblings should be a routine part of care for DMD.

Interventions

Care and support interventions

- **A care coordinator** can be the crucial person here: they can serve as a point of contact for families and become a trusted person. This person needs to have sufficient knowledge and background in neuromuscular disorders to be able to meet routine family information needs.
- **Proactive intervention is essential to help avoid social problems and the social isolation** that can occur in the context of DMD. Examples of useful interventions include increasing awareness and education about DMD in school and with peers, ensuring participation in appropriate sports and camps, provision of service dogs and contact with others via the internet and other activities.

A special individualised education plan should be developed to address potential learning problems and to modify activities that might otherwise prove harmful to the child's muscles (e.g. physical education), reduced energy/fatigue (e.g. walking long distances to/from lunch), safety (e.g. playground activities), and accessibility issues.

- Making sure the school is fully informed about DMD is important. Share with them all the information you have and identify the person at the school who is there to support children with additional needs. A proactive approach is important to make sure that the child with DMD accesses the full range of education he needs to develop good social interactions and prepare for further education and employment. So the school needs to be on side!
- **Promoting independence and involvement in decision making** (in particular, as relates to medical care) is necessary and of significant importance to promote autonomy and independence. This should

be part of a planned transition program from paediatric to adult care.

- **Helping to develop social and learning skills** will make it easier to find a job and be part of normal daily life in adulthood. Boys with DMD benefit from having support to reach their personal goals.
- Access to palliative care services is appropriate to relieve or prevent suffering and to improve quality of life, as needed. In addition to pain management (Box 6), palliative care teams may also be able to provide emotional and spiritual support, assist families in clarifying treatment goals and making difficult medical decisions, facilitate communication between families and medical teams, and address issues related to grief, loss, and bereavement.

Psychotherapy and drug interventions

Several well-known techniques exist to help in various areas. These include training for parents in trying to cope with bad behaviour and conflicts, individual or family therapy and behavioural interventions. Applied behaviour analysis may help with certain behaviours related to autism.

Some children and adults may get benefit from the use of prescribed medicines to help with emotional or behavioural problems. These medicines can be used under specialised supervision and monitoring for depression, aggression, OCD or ADHD when these problems have been specifically diagnosed by specialist doctors.

11 Considerations for surgery

There will be a variety of situations, both related to DMD (e.g. muscle biopsy, joint contracture surgery, spinal surgery, or gastrostomy) and unrelated (e.g. acute surgical events), where general anaesthesia may be needed. There are a number of condition-specific issues that need to be taken into account for the planning of safe surgery.

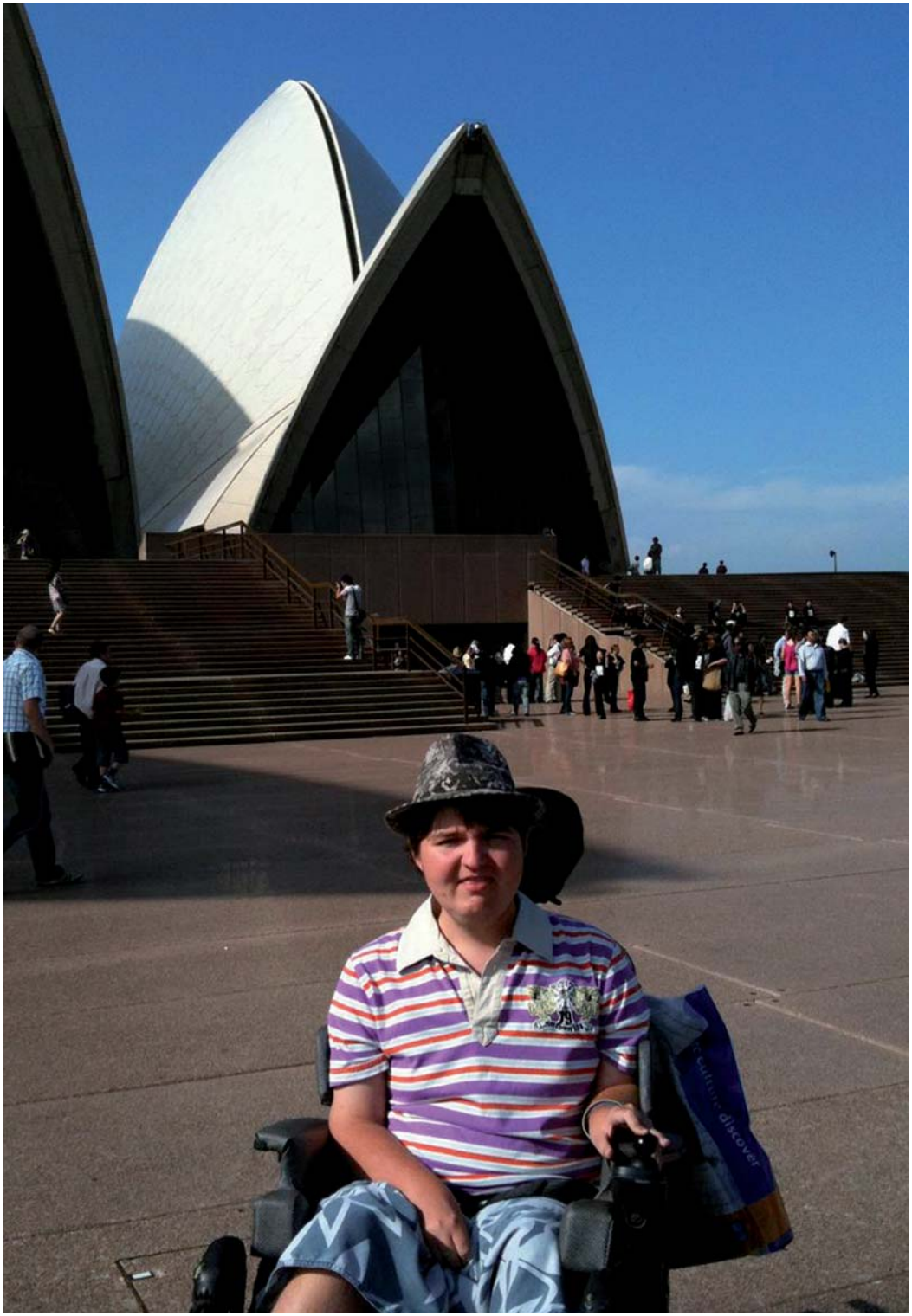
- **Surgery should be done in a hospital where personnel involved in the operation and after care are familiar with DMD and willing to work together to be sure everything goes smoothly.** In addition, consideration needs to be given to "stress steroid" coverage during surgery, for people treated with steroids at home.

IMPORTANT FACTS TO REMEMBER:

1. There are always risks with anaesthesia; however, special considerations in DMD can allow anaesthesia to be given more safely, such as the use of a total intravenous anaesthesia technique and absolute avoidance of the drug succinylcholine.
2. Proper assessments of the heart and lungs are important when planning for surgery.
3. Make sure that all doctors are properly informed about DMD and all interventions (medicines) your son is getting.

Anaesthetic agents and other considerations for safe operative care

- There are always risks with anaesthetics and there are special considerations in DMD to allow anaesthetics to be given safely including using total intravenous anaesthetics and avoiding specific drugs.
- Minimising loss of blood is important especially in major surgery such as spinal fusion. In this situation, the surgeon and anaesthetist may decide to use specific techniques to help with this.
- Full details are available in the main document.



Cardiac considerations

- **An echocardiogram and electrocardiogram should be performed prior to general anaesthesia**. They should also be performed before undergoing conscious sedation or **regional anaesthesia** if the last investigation was more than one year previously, or if there had been an abnormal echocardiogram in the preceding 7-12 months.
- For **local anaesthesia**, an **echocardiogram** should be performed if there had been an abnormal result obtained previously.

Respiratory considerations

- **Even if someone with DMD already has problems with the breathing muscles, certain measures can make surgery safer**, though there will still be an increased risk. A pre-operative assessment of breathing function in a centre familiar with DMD is very important. Pre-operative training in the use of non-invasive ventilation and assisted cough, and specialised interventions may be needed.
- Physiotherapists should always be involved if someone with DMD is having surgery.
- Planning and proactive assessments and management of risk is the key to safe surgery in DMD.

12 Emergency care considerations

If you find yourselves needing to go to the hospital in an emergency situation, there are a range of factors that should be taken into account.

- The **diagnosis of DMD, current medication, presence of any respiratory and cardiac complications and the people who are your key medical input should be made clear** to the admitting unit.
- As many health professionals are not aware of the potential management strategies available for DMD, the **current life expectancy and expected good quality of life should also be explained**.

IMPORTANT FACTS TO REMEMBER:

1. You are very likely to know more about DMD than the doctors in Accident and Emergency.
2. Advise the doctor or healthcare staff if your son is taking steroids.
3. If your son has a broken bone, insist that they speak with your doctor or physiotherapist.
4. If you can, bring copies of your son's most recent test results, such as FVC and LVEF.
5. If your son's oxygen level drops, the doctor must be very careful about giving him oxygen or sedating medication.

Steroids

Chronic steroid use needs to be made clear. Tell the staff **how long your son has been using steroids** and if he has **missed a dose**. It is also important to let the doctors know if your son used steroids in the past.

- Steroids can dampen the stress response so extra steroids may be needed if someone on chronic steroids is unwell.
- Steroids can increase the risk of stomach ulceration.
- Rarely other complications can present acutely.

Broken bones

Boys with DMD are at risk of broken bones and breaking a leg bone can mean that it is difficult to walk again if walking is already very difficult. Let your physiotherapist and the rest of the care team know if there is a fracture so they can talk to the surgeons if necessary.

- **Surgery is often a better option** than a cast for a broken leg if someone is still walking.
- **Input from a physiotherapist is crucial** to make sure that the boy gets back on his feet as soon as possible.
- If the broken bone is one of the vertebrae (backbones) with a lot of pain in the back, input from a bone doctor or endocrinologist is needed to provide the right treatment (see Section 6).

Breathing problems

Try and keep a note of, or remember what the latest tests of breathing **(e.g. forced vital capacity, FVC)** were. This information can be useful for the doctors assessing your son if he does become ill acutely.

The main risks with breathing problems come when FVC and coughing strength have reduced:

- Help with clearing the chest may be needed;
- It may be important to help with coughing;
- Antibiotics may be needed;
- Sometimes it may be necessary to give support with a ventilator;
- Risk of the breathing muscles needing extra support during an infection can be high in those with borderline respiratory function. **Care in the use of opiates and other sedating medication is essential, as is care in the use of oxygen without ventilation due to the risk of rising carbon dioxide in people with compromised breathing muscle strength**;
- If nocturnal ventilation is already used, then **access to the ventilator** is essential during any acute event or intervention. For those who are already ventilated, the team involved with the respiratory care should be involved as soon as possible.

If you have a ventilator (or similar equipment) it is a good idea to bring it with you to the hospital.

Heart function

Try and keep a note of what the latest test results of heart function **(e.g. left ventricular ejection fraction, LVEF)** were, and what, if any, heart medication your son is on and which cardiologist sees him. This will help the emergency doctors decide if it is likely that the problems they are seeing are due to a problem with the heart.

- Awareness of the **risk of heart rhythm problems and cardiomyopathy** is important.

Anaesthetic risks (see Section 11) need to be taken into account at all times if surgery or sedation is needed.

Abbreviations

ACE	angiotensin converting enzyme (ACE inhibitors are used to control cardiac problems and high blood pressure)
ADHD	attention deficit hyperactivity disorder
ADL	activities of daily living
AFOs	ankle-foot orthoses (splints which are used to control tightness at the ankles)
ALT	alanine aminotransferase
AST	aspartate aminotransferase
BP	blood pressure
CDC	Centers for Disease Control and Prevention (the major public health body in the USA)
CK	creatine kinase (an enzyme found at high levels in the blood in DMD and other forms of muscular dystrophy)
DEXA	dual energy X-ray absorptiometry (a test done to look at the strength of the bones). Also referred to as DXA
DMD	Duchenne muscular dystrophy
ECG	electrocardiogram (the main test done to look at heart rhythm)
FVC	forced vital capacity (a test of breathing muscle strength)
GC	glucocorticoid
IV	intravenous (into the vein)
KAFOs	knee-ankle-foot orthoses (long leg splints that can be used especially over the period that walking is becoming impossible and thereafter to help prolong walking)
kg	kilogram
L	litre
LVEF	left ventricular ejection fraction (one of the main tests of heart function)
mg	milligram
nmol	nanomoles
NSAIDs	non-steroidal anti-inflammatory drugs (which are used for pain relief, the most common of which are ibuprofen, diclofenac and naproxen)
OCD	obsessive-compulsive disorder
TA	achilles tendon
TB	tuberculosis
VOCA	voice output communication aid

List of terms that you may come across

A

Aetiology
cause

Anterior spinal fusion
a way to correct scoliosis via an anterior approach

Aspiration pneumonia
pneumonia caused by irritation or bacteria from stomach content entering lungs due to faulty swallowing

Atelectasis
condition in which the lungs are not fully inflated

B

Baseline
the starting point to compare other tests

BiPAP
bi – two way; PAP – positive air pressure. Used to maintain lung expansion

Body Mass Index
relationship between weight and height according to formula weight, in kg, divided by the square of length, in metres

C

Cardiomyopathy
deterioration of heart muscle function - also known as “heart muscle disease”

Cobb angle
measurement of scoliosis angle from spinal X-ray

Contractures
tightness round a joint leading to its becoming fixed in a particular position or having less than full range of motion at that joint

Cushingoid features
term used to describe the round or “moonlike” face that people on steroids may develop. (This may be prominent even if weight gain overall is not an issue and can be hard to control without a change in steroid or in the dosing schedule)

D

Depolarising muscle relaxants
drugs that decrease the muscle tone by acting on muscle receptors involved in depolarisation

DEXA
see abbreviations

Dysphagia
swallowing problems

Dystrophinopathy
the term used to cover all the different conditions caused by faults in the dystrophin gene (Duchenne muscular dystrophy, Becker muscular dystrophy, manifesting carriers of one of these conditions and rare patients who have only heart disease)

E

Eccentric Exercises
exercises such as going downstairs or trampolining that involve lengthening rather than contraction of the muscle

Electrocardiogram (ECG)
methods used to assess the electrical activity of the cardiac muscle. The ECG involves stickers placed on the chest to record heart signals.

Echocardiogram ("echo")
method used to assess the structure of the heart. The Echo is also known as a "cardiac ultrasound" and gives pictures of the beating heart

Electromyography
a test that measures electrical signals from a muscle and can give a clue to whether a nerve or muscle disorder is present

F

Forced Vital Capacity
the maximum volume of air that can be exhaled after maximum inhalation

G

Gastritis/gastroesophageal reflux
occurs when the muscle joining the oesophagus (swallowing tube from the mouth) to the stomach opens on its own, or does not close properly and stomach contents rise up into the oesophagus. Also called acid reflux or acid regurgitation, because digestive juices, called acids, rise up with the food

Gastrostomy
surgical opening into the stomach, in this case to insert a feeding tube. Sometimes referred to as a PEG

Germline mosaicism
condition in which the cells in the gonads that will develop into germ cells (ova and spermatozoa) are a mixture of two genetically different cell types

Glucose intolerance
defines a pre-diabetic state associated with insulin resistance

Gowers' manoeuvre/sign
a sign of weakness in the muscles round the hips and upper part of the lower legs. It describes the way that someone with weakness in these muscles gets up of the floor, needing to turn onto their front, keep their legs wide apart and using a hand on their thigh to rise. It is commonly seen in DMD but other conditions causing weakness in the same muscle groups also cause a Gowers' manoeuvre

H

Holter
method used for continuous ambulatory 24 hr ECG recording

Hypercapnia
too much carbon dioxide in the blood

Hypertension
high blood pressure

Hypoventilation
reduced breathing efficiency of ventilatory capacity

Hypoxemia
decreased oxygen levels in the blood

I

Immunoblotting
a way to measure the amount of dystrophin in the muscle

Immunocytochemistry
a way to look at the muscle under the microscope and see how much dystrophin is present

K

Knee adductors
the muscles that keep the knees together

Kyphoscoliosis
abnormal curvature of the spine with both sideward (scoliosis) and hunched forward or backward (kyphosis)

M

Malignant Hyperthermia-like reaction
a response to anaesthesia that causes a high temperature and can be life-threatening

Motor Function Scales
tests which are used to assess activities of motor performance in a standardised way

Myoglobinuria
presence of myoglobin in the urine as a sign of breakdown of muscle (urine looks coca-cola coloured because it contains breakdown products of muscle proteins)

O

Osteopenia/osteoporosis
decrease in bone mineral density

Oximetry
the measurement of oxygen in the blood stream using a machine to detect it through the skin

P

Palpitations
awareness of abnormal heartbeats

Pelvic obliquity
describes a condition in which the pelvis is uneven, such as being rotated downward on one side

Prophylaxis
prevention

R

Rhabdomyolysis
breakdown of muscle

S

Scoliosis
curvature of the spine

T

Tanner stage
defines pubertal development based on external primary and secondary sex characteristics, such as the size of the breasts, genitalia and the development of pubic hair

Tenotomy
surgical cutting of a tendon

Thrombolitic events
formation of a clot (thrombus) in a blood vessel that breaks loose and is carried by the blood stream to plug another vessel

Tinea
a fungal skin infection

Tracheostomy
surgical procedure on the neck to open a direct airway through an incision in the trachea (the windpipe)

V

Varus
inward rotation of the foot due to an imbalance of the foot muscles

Volume recruitment
increasing the amount of air taken in by the lungs using a device to help inflate the lungs. Such devices include Ambu bags and in-exsufflators. Ventilators can also be used to increase volume

Videofluoroscopic study
assessment tool to view and determine the nature and extent of an oropharyngeal swallowing problem. A video X-ray is taken as the child swallows the food

Resources for families living in the UK and Ireland

The UK and Ireland version of this guide was produced by TREAT-NMD (**www.treat-nmd.eu**). Please contact **info@treat-nmd.eu** or **0191 241 8605** with any questions. Additional copies can be obtained from TREAT-NMD or any of the patient organisations listed in this section.

PATIENT ORGANISATIONS

UK

ACTION DUCHENNE

Action Duchenne is a national charity set up by Duchenne families in 2001 to promote new research to cure Duchenne. The charity has directly funded a $1.2m research programme with AVI Biopharma and the MDEX consortium to develop exon skipping drugs for Duchenne. Action Duchenne campaigns to implement best practice standards of care for all Duchenne patients.

E: info@actionduchenne.org
W: www.actionduchenne.org
T: 020 8556 9955

DUCHENNE FAMILY SUPPORT GROUP (DFSG)

The DFSG is a national charity run by families for families affected by Duchenne. It provides a positive national support network of parents, their families and professionals. It also fundraises and organises subsidised holidays, events, and workshops.

E: info@dfsg.org.uk
W: www.dfsg.org.uk
T: 0800 121 4518 (free phone)

MUSCULAR DYSTROPHY CAMPAIGN (MDC)

The Muscular Dystrophy Campaign is the leading UK charity focusing on muscle disease and has pioneered the search for treatments and cures for over 50 years. The MDC funds world-class research to find treatments and cures, campaigns to raise awareness and provides free practical advice, emotional support and signposting to local professionals and groups.

The MDC established and supports the North Star database which records the progression of Duchenne by gathering data from patients across the UK, making this an invaluable research and clinical tool.

E: info@muscular-dystrophy.org
W: www.muscular-dystrophy.org
T: 0800 652 6352 (free phone)

IRELAND

DUCHENNE IRELAND

The aims of Duchenne Ireland are to raise awareness of Duchenne muscular dystrophy at local, national and government level, and to raise funds that go directly to the researchers and clinicians we believe have the best chance of developing improved therapies which will benefit this generation. We are also working towards achieving an infrastructure which is on a par with best international practice.

W: www.duchenne.ie

MUSCULAR DYSTROPHY IRELAND (MDI)

MDI provides information, advice and support to people with neuromuscular conditions and their families through a range of support services. MDI's objective is to promote practical empowerment and independent living for people with neuromuscular conditions to fully participate in society and to live the life of their own choosing.

E: info@mdi.ie
W: www.mdi.ie
T: (+353) (0) 1 8721501

OTHER USEFUL INFORMATION

DMD PATIENT REGISTRY

The DMD Registry is a UK and Ireland-based patient registry that collects information about patients who are affected with both Duchenne and Becker muscular dystrophy, and also female carriers. The information collected in the UK registry is fed into the TREAT-NMD global registry for DMD. Registration is voluntary and done by the patients/parents themselves. To register online, visit the website or contact the registry at the address below.

WHY SHOULD I REGISTER?

- Registered patients may be able to participate in clinical trials more easily
- Registered patients are kept informed about the results of current research into new treatments for DMD
- Registries may help raise urgently needed funding for research

W: www.dmdregistry.org
A: DMD Registry, Action Duchenne, Epicentre, 41 West Street, London, E11 4LJ

EDUCATIONAL AND BEHAVIOURAL SUPPORT

Decipha is a comprehensive learning and behaviour programme that can be used by schools, parents and education professionals. It offers training and a structured web based literacy intervention programme. Decipha gives an expert assessment of young people's learning and behaviour needs. Visit the website for more details of the programme and how to become involved.

E: janet@decipha.org
W: www.decipha.org

EMERGENCY CARE PACK

Most people living with DMD will never need to visit an emergency department, however if your son becomes unwell and needs to go to hospital this pack contains vital information that you can pass onto the emergency doctors. Visit the Action Duchenne website to register for your free copy.

W: www.actionduchenne.org/r-nav/443.jsp

IMMUNISATION SCHEDULES

UK: Information provided by the NHS on the current UK immunisation schedules.

W: www.immunisation.nhs.uk

IRELAND: Information provided by the HSE on the current Irish immunisation schedules.

W: www.immunisation.ie

HEALTHY EATING ADVICE

Information provided by the Food Standards Agency on healthy eating for the whole family.

W: www.eatwell.gov.uk

BMI CALCULATOR

A tool provided by the NHS to calculate if the whole family is a healthy weight, providing practical tips and information for maintaining a healthy diet.

W: www.nhs.uk/tools/pages/healthyweightcalculator.aspx

GROWTH CHARTS

CHILDREN AGED 0-4 YEARS: The early years UK growth charts have been produced by the Royal College of Paediatrics and Child Health, and endorsed by the World Health Organisation (WHO).

W: www.rcpch.ac.uk/Research/UK-WHO-Growth-Charts

CHILDREN AGED 5-19 YEARS: WHO have produced growth charts for children aged 5-19 years.

W: www.who.int/growthref/who2007_bmi_for_age/en/index.html

WHEELCHAIR SERVICES

UK: A list of NHS wheelchairs services around the country, allowing easier contact with local services.

W: www.wheelchairmanagers.nhs.uk/services.asp

IRELAND: The Irish Wheelchair Association is a national charity established by a group of wheelchair users to provide quality services to people with limited mobility throughout the country.

W: www.iwa.ie

DIRECTGOV

Provides information on a variety of issues specifically related to any UK government schemes, such as applying for a disabled facilities grant to adapt your house, and support for your child at school.

W: www.direct.gov.uk

Photographs used in this guide were provided by Duchenne Parent Project Netherlands, MDA, PPMD, Parent Project Czech Republic and TREAT-NMD. We would like to thank all the boys and families who allowed their photos to be used.

MDA, PPMD, TREAT-NMD and UPPMD have all been directly involved in the writing and production of this guide.

MDA Helping Jerry's Kids

Parent Project Muscular Dystrophy
LEADING THE FIGHT TO END DUCHENNE

TREAT-NMD Neuromuscular Network

From 2007 to 2011 inclusive, TREAT-NMD is a "network of excellence" supported through Priority 1 (Life Sciences, Genomics and Biotechnology for Health) of the European Union's Sixth Framework Programme.

United Parent Projects Muscular Dystrophy