Montrose Access

DUCHENNE MUSCULAR DYSTROPHY

a team approach to management

DUCHENNE MUSCULAR DYSTROPHY

a team approach to management

Montrose Access

Acknowledgements

MontroseAccess gratefully acknowledges a grant from the Commonwealth Learning Special Needs Program to produce this educational package and to NSO, the Heritage Building Society, and the Ian Potter Foundation for their assistance in updating the material in this second edition that comprises a manual and accompanying DVD.

This project aims to provide an understanding of Duchenne muscular dystrophy (DMD) and current research as well as outline the multidisciplinary approach to management and methods of providing support for children, their families and schools. This material has been written for general practitioners and therapists in rural and remote areas who may not see many children with Duchenne muscular dystrophy and for any health professionals, teachers and advisory visiting teachers who are involved in the care of boys with DMD. Finally, it is hoped that the material will assist the families of boys with DMD.

We acknowledge the researchers and authors of the following chapters:

Project leader and coordinator **Introduction, diagnosis, research, clinical features, medical management, physiotherapy management, essential requirements for seating**	Helen Posselt
Shark bait kids program	Sue Nicklin
Occupational therapy management	Jan Bates
	Alice Zuber
	Mathilde Backhouse
	Lyndal Mc Phail
Speech, language and cognitive skills	Pam Bignold
	Jo Wilkinson
	Renee French
	Marion Langdon
Recreation	David Stafford and the Recreation department, Montrose Access
Social and emotional issues	Marea Vanzetti
	Geraldine Burgess
Dietetics	Zoe Davidson, Department of Nutrition & Dietetics, Monash University
Endocrinology	Dr Tony Huynh, Paediatric Endocrinologist, Department of Paediatric & Child Health, Canberra

We extend special thanks to:

- families living with DMD, especially those involved with this project
- the Parent Project for Muscular Dystrophy and, in particular, Mrs Pat Furlong for her invaluable assistance and support over many years
- MontroseAccess management, therapy, support, administration and clerical staff for their assistance
- Education Queensland for support and permission to reprint some photographs.

Video production	Videocam Productions
Design and layout	Goldi
Editing	New Word Order
Illustrations	See-Saw Illustration
Photography	Lyle Radford

ISBN 978-0-9585865-5-9

Contents

Introduction to DMD

Duchenne muscular dystrophy is the most prevalent and disabling of the inherited neuromuscular disorders. It affects one in 3500 live-born males and is found equally in all world populations as well as in many animal species. The milder Becker muscular dystrophy affects one in 20000.[1]

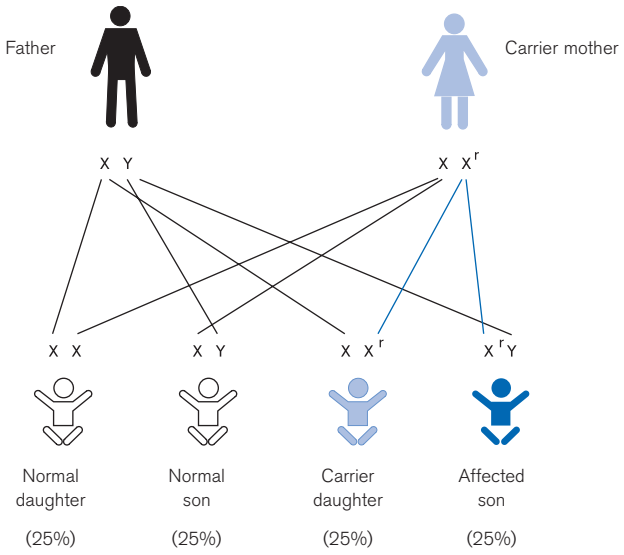
Although present from conception and progressive from the earliest stages of development, the clinical signs do not become evident until between two and five years of age. Initially, weakness is evident in the proximal muscles, especially those of the pelvic girdle, and later the shoulder girdle. Weakness subsequently progresses to the distal muscles.

Smooth and cardiac muscle are also affected and there is some involvement in the brain, particularly in the neocortex, cerebellum and hippocampus.

In the natural course of the condition, boys lose the ability to walk at around 10 years of age and die of respiratory insufficiency or as a result of cardiomyopathy by their late teens to mid-20s. Advances in medical care have extended life span, often well into the third decade and sometimes longer. The use of corticosteroid medication, improved cardiac surveillance and medication, as well as access to nighttime ventilation, daytime support for declining respiration and the cough-assist machine have all contributed. Better care, management and access to education and technology have all improved quality of life immeasurably.

Mode of inheritance

When inherited, the condition follows a typical X-linked recessive pattern, i.e. a son has a 50 per cent chance of inheriting the condition from a carrier mother. A daughter also has a 50 per cent chance of inheriting carrier status.



Sporadic events

There is an extremely high mutation rate in the gene responsible for Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD). One in 10,000 eggs and sperm are thought to carry a new mutation.

Approximately one in three cases is the result of sporadic event (i.e. not inherited). In isolated cases, there is a 33 per cent chance that the mother is not a carrier.

Gonadal mosaicism can also occur in mothers thought not to be carriers. In this case, the mother carries the mutation in some of the ova in her ovaries but it is not found in her somatic tissues. There is then a 20 per cent chance that future children could inherit the defective gene.

Reference

1. Brown SC, Lucy JE. Dystrophin, Gene Protein and Cell Biology, Cambridge University Press; 1997.

Chapter one:
Genetics

The gene responsible for DMD was cloned in 1986 and the protein that it encodes (dystrophin) was identified in 1987:

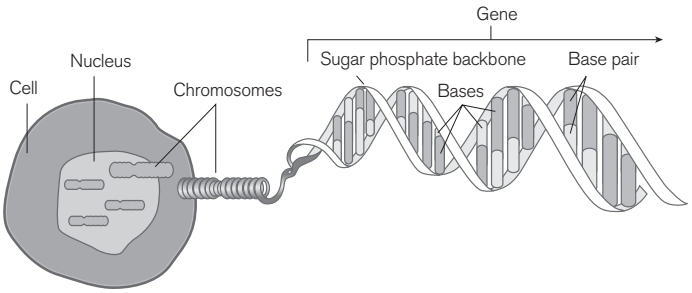
- it is the largest gene yet identified, comprising more than 2.5 million base pairs of genomic material
- it is 10 times bigger than the next biggest gene so far identified in any animal species
- its size makes it a prime target for random mutations and explains the high incidence of the condition worldwide, despite the increasing availability of prenatal diagnostic services for known carriers of the condition and the options available to parents when a diagnosis is confirmed in the foetus.

WHAT IS A GENE?

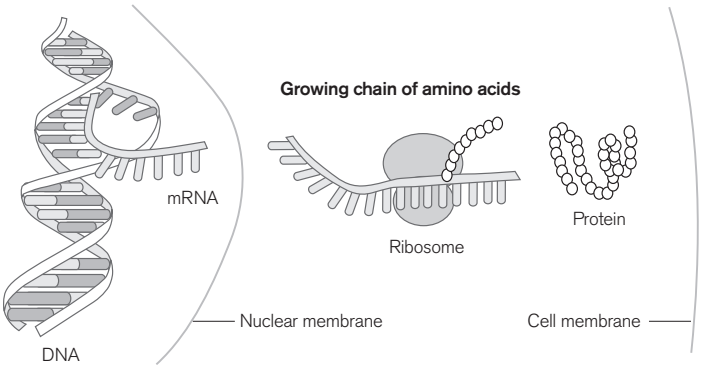
Deoxyribonucleic acid (DNA) is comprised of nucleotides. There are four main types of nucleotides found in DNA – cytosine, guanine, thymine and adenine. These are ordered in a specific sequence known as genes, to provide information required to make proteins. The coding sequences within a gene are known as exons, while interspersed between the exons are sections that do not contribute to making a protein. These are known as introns.

When ribonucleic acid (RNA) is transcribed from the gene, the information from the exons is spliced together exactly, in groups of three nucleotides called codons. The information in the codons determines which amino acids are linked together to make proteins.

Chromosomes, DNA and genes



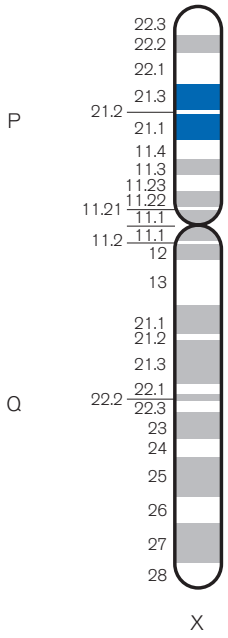
DNA transcription and translation



Redrawn from American Society of Clinical Oncologists
Reprinted with permission. Copyright 2001 ASCO genetic slides

Gene location

The gene responsible for DMD is located on the short arm of the X chromosome, position 21.



This gene is known to code for the protein dystrophin, which is absent in the muscle biopsies of patients with DMD and present to some extent (though abnormal) in BMD. These two conditions are known as the dystrophinopathies because the protein dystrophin is deficient or defective.

Mutations of the gene

Mutations of the gene can be:

- deletions of one or more exons are the most common, and account for 72 per cent of the DMD mutations and 85 per cent of BMD. Deletions are most often found clustered around two hot spots rather than randomly distributed throughout the large gene. These hot spots are from exon 44 to 52 and from exon 2 to 13.
- duplications account for 7 per cent of DMD and BMD.
- point mutations account for 20 per cent and are very small deletions or insertions of one or a few genetic letters.
- nonsense mutations are point mutations that exchange one nucleotide where the change replaces an amino acid with a stop codon, preventing further production of dystrophin. Other point mutations include missense mutations, which are insertions or deletions of one or several nucleotides. For detection and confirmation of a nonsense mutation, patients usually need to first have multiplex ligation-dependent probe amplification (MLPA) screening in order to exclude exon deletions and duplications, followed by sequencing of all exons of the dystrophin gene if none are identified. [2]
- 1 per cent of mutations are accounted for by very rare disruptions of splice sites or rearrangements of large parts of the gene structure.

Clinical severity in the Xp21 dystrophinopathies

The clinical picture ranges from the severe presentation in DMD to the milder phenotype (presentation) in BMD at the other end of the spectrum. Cases of intermediate severity also occur:

- less than 3 per cent of normal dystrophin results in a diagnosis of DMD
- between 5 and 15 per cent of dystrophin results in an intermediate form
- more than 20 per cent dystrophin results in a diagnosis of mild to moderate BMD[3]
- more than 30 per cent dystrophin would also result in a diagnosis of BMD and there have been many reported cases with a very mild clinical presentation.

The presence in muscle fibres of even a small amount of dystrophin confers some functional advantage. [4]

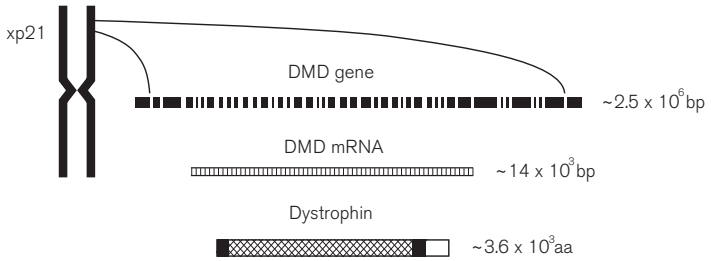
The ability to produce dystrophin, and therefore the severity of the condition, does not correlate with the size of the deletion in the gene. Rather, it depends on whether the reading frame is altered (see box).

THE READING FRAME HYPOTHESIS

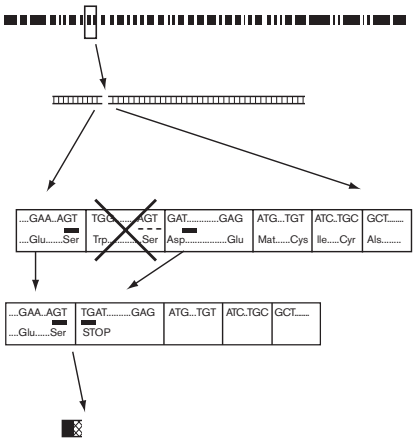
Genetic material is made up of nucleotides, sequenced in triplets called codons, which 'code' for (or are the recipe for) specific amino acids, the building blocks of all proteins. Special codons signal the start of a protein sequence while others (stop codons) signal the end. When a mutation has occurred, nucleotides are added (duplication), deleted (deletion) or replaced by another nucleotide (point mutation) from the sequence.

The clinical severity will depend on the number of added or deleted nucleotides. If the number is divisible by three, then the codons subsequent to the deletion or duplication will be unaffected and the resulting proteins will be similar to the original but with only some amino acids missing. Therefore, they will function partially. The result is the less severe clinical presentation in BMD.

If the number deleted or added is not divisible by three, then the 'reading frame' is shifted and an almost unrecognisable protein will be produced that will not function–resulting in DMD. Sometimes the mutation produces a premature stop codon, which results in a truncated protein with greatly reduced or no function. The more severe clinical presentation of DMD will then result. [2,3]

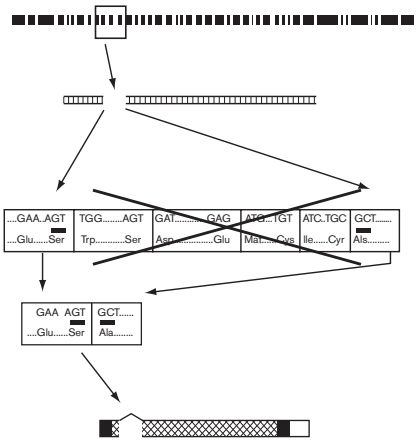


Deletion resulting in severe DMD



Severely truncated dystrophin
Rapidly degraded by cell

Deletion resulting in mild DMD



Internally deleted, semi-functional dystrophin
Allowed to persist by cell

Hoffman & Kunkel (1989)

References

1. Dubowitz V. The Muscular Dystrophies. Postgraduate Medical Journal 1992;68:500-506.
2. Brown SC, and Lucy JE. Dystrophin, Gene Protein and Cell Biology, Cambridge University Press; 1997.
3. Lalic T, Vossen RH, Coffa J, et al. Deletion and duplication screening in the DMD gene using MLPA. Eur J Hum Genet 2005;13: 1231–34
4. Blake DJ, Kröger S. The neurobiology of Duchenne muscular dystrophy: learning lessons from muscle? Trends Neurosci. 2000;Mar;23(3):92

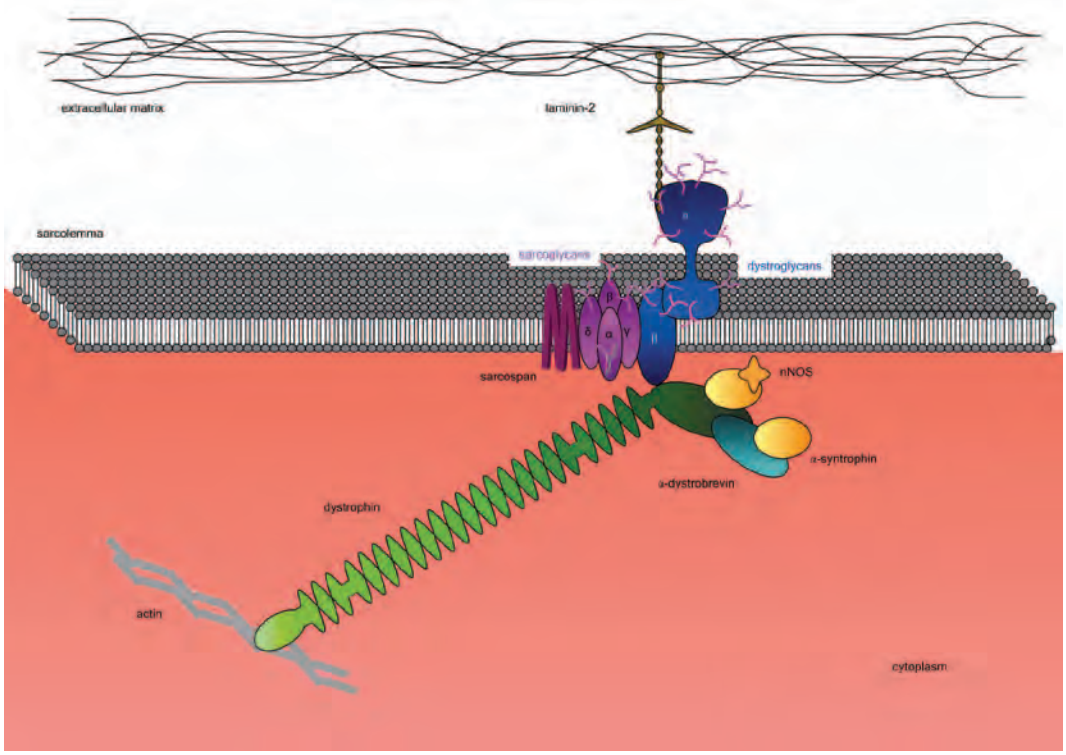
Other References

Emery A. and F. Muntoni. Duchenne Muscular Dystrophy, Oxford University Press, Oxford; 2004.
Emery A. E. H. Duchenne Muscular Dystrophy, Oxford University Press, Oxford; 1993.
Emery A. E. H. Muscular Dystrophy: The Facts, Oxford University Press, Oxford; 1994.
Dubowitz V. A Colour Atlas of Muscle Disorders in Childhood, Wolfe Medical Publications Ltd, England; 1989.
Dubowitz V. The Muscular Dystrophies. Postgraduate Medical Journal 1992;68: 500-506.
Dubowitz V. Muscle disorders in childhood;1995.
Dubowitz V. Forty years of Neuromuscular Disease, A Historical Perspective. Imperial College of Science, Technology and Medicine, Hammersmith Campus, London, UK. New Perspectives in Paediatric Neuromuscular Disorders Conference, Sydney; 1998.

Chapter 2:

The role of dystrophin

Dystrophin is a large rod-like cytoskeletal protein, which is located on the inner surface of the plasma membrane of muscle fibres. It is associated with other proteins known as the dystrophin-associated glycoprotein complex, which is made up of intracellular, trans-membrane and extracellular proteins. This complex is thought to attach the inner cytoskeleton (actin) to the extracellular matrix, which coats the muscle fibres. Within each muscle cell, the bundles of actin and myosin proteins move back and forth in a plane, effecting a muscle contraction. The dystrophin-associated complex and dystrophin form the bridge and act as a shock absorber between these contractile fibres and the cell membrane, permitting smooth movement while remaining firmly anchored. The complex has also been likened to internal scaffolding, which maintains the integrity of the membrane during the stresses imposed on it during contractions.



Reprinted with the permission of Dr Annemieke Aartasma-Rus, Department of Human Genetics, Leiden University Medical Center, the Netherlands.

When dystrophin is absent, there is also a drastic reduction in all of the dystrophin-associated proteins and a subsequent disruption of the linkage between the inner cytoskeleton and the extracellular matrix, causing instability and tears to occur in the membrane.

Dystrophin molecules have four distinct domains.[4]

- Aminoterminal is associated with the F-actin binding cytoskeleton at two distinct locations. If it is missing yet other domains remain intact, dystrophin can still bind F-actin through a third domain located in the central rod, and a severe Becker's muscular dystrophy results.
- Central rod domain is less critical for dystrophin. Deletions in this area that do not completely disrupt the reading frame, permitting a partially functioning dystrophin to be produced which would result in a mild Becker muscular dystrophy. Deletions that disrupt all F-actin binding domains (in aminoterminal and central rod) will cause Duchenne muscular dystrophy. The rod also contains a domain that binds to the nNOS protein, but this is not crucial for dystrophin function.

- Cysteine-rich penultimate domain is a binding region for dystroglycan and through that the glyco-protein complex through the plasma membrane and the extracellular matrix. This domain is crucial for protein function. Mutations in this region will always cause Duchenne muscular dystrophin, even if they do not disrupt the reading frame.
- Carboxyl terminus appears to be less important, though it does contain the binding site for syntrophin protein. When this domain does not function, patients can develop either Duchenne or Becker muscular dystrophy, depending on the location of the mutation (the further down the end, the more chance Becker muscular dystrophy will be the result). [4]

The role of dystrophin is not clearly understood; however, the major role of dystrophin is thought to be the maintenance of muscle membrane structure. In the absence of dystrophin, muscle cells are susceptible to damage induced by the strong torsional forces during muscle contractions. Calcium is thought to flood into the cells through tears made in the plasma membrane. Uncoordinated mini-contractions then occur, causing further damage to the membrane and the death of the cell. Intracellular enzymes leak out into the blood serum, giving rise to elevated serum creatinine phosphokinase (CPK) levels.[17]

Another contributing factor could be the relative absence of nitric oxide synthase (nNOS) in dystrophic muscle. nNOS, found attached at two sites to the dystrophin molecule (syntrophin and in the rod domain) in normal skeletal muscle, is an enzyme that produces nitric oxide (NO). NO is an important cellular signaling molecule, having a vital role in many biological processes. It is the intercellular signal that controls airway tone and peristalsis. Nitric oxide plays a role in vascular tone (hence blood pressure) and permits an increase in blood flow during exercise. In dystrophin-deficient muscle, blood vessels do not dilate, creating a deficit in blood flow and supply of oxygen and nutrients to the muscle. The waste products of metabolism are also not easily removed. An injury, similar to an ischaemic injury, results. Other studies[9] are looking at oxidative stress and the protective role NO could play. NO is also thought to play a major role in the mechanism that acts as a switch for activating muscle precursor cells (satellite cells) to begin the repair process after injury. Inhibiting NO for 30 minutes before injury to the muscle stops satellite cells from being released.[10]

Dystrophin and the dystrophin-associated glycoprotein complex are also thought to play a signalling role. They could possibly be responsible for relaying messages from the inner cytoplasm to the extracellular matrix.[9]

Pathology

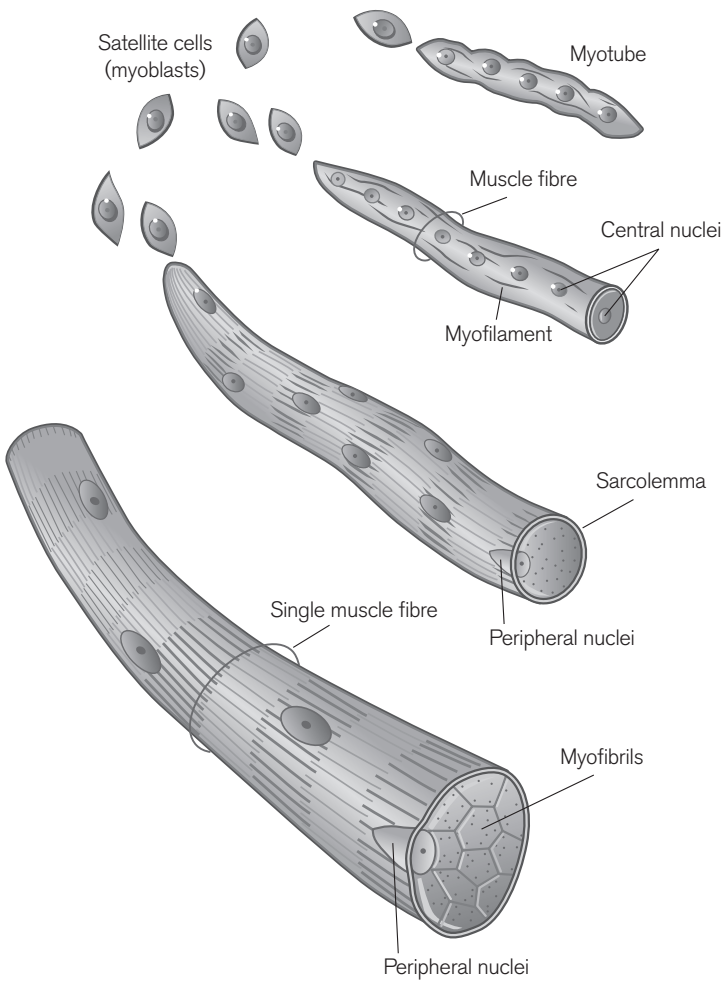
Membrane fragility and subsequent muscle fibre damage induces a chain of events that further disables the muscle. Protease activation occurs and the immune system is activated, bringing an influx of cytotoxic T-lymphocytes, which are thought to destroy more dystrophic muscle through an autoimmune response. The onset of pathology is also characterised by the invasion of large numbers of macrophages and neutrophils, which generate and release free radicals. These are known to cause damage and death of cells.[11] Dystrophic muscle is far more sensitive than normal muscle to attack by the free radicals.[9]

Aggressive proliferation of fibroblasts between the damaged fibres plays a large part in the loss of function. Fibroblasts produce collagen in an attempt to bind the regenerating muscle cells. This is a normal occurrence in the wound-repair mechanism, but successive cycles of

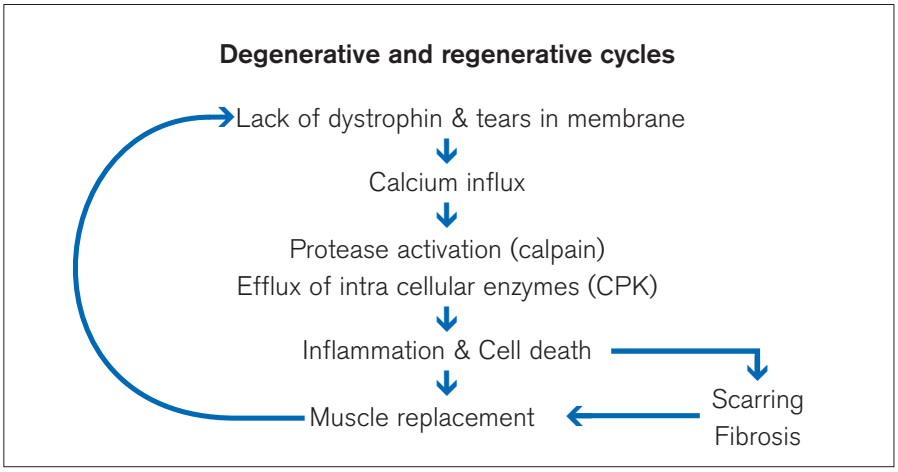
degeneration and regeneration result in a collagen build-up and the proliferation of scar tissue. The build-up of scar tissue not only interferes with the blood supply to the muscle but also the ability of remaining fibres to contract, both of which contribute to failed regeneration. Fat as well as scar tissue then replaces muscle.

Degeneration and regeneration

Initially, the body is able to repair damaged muscle fibres by activating satellite cells. These are precursor muscle cells, which have remained as single cells between the plasma membrane and the basal lamina. When activated they divide and multiply, then fuse together to form myotubes and repair the damaged fibres. However, in dystrophin-deficient myofibres the membrane damage is continually occurring and the cycle of degeneration and regeneration is repeated until satellite cells have exhausted their ability to replicate.



Development of skeletal muscles



The role of dystrophin in the brain

In recent years, there has been increasing understanding of the role dystrophin plays in the central nervous system.

The areas of greatest abundance of dystrophin in the brain are the cerebellum, the neocortex and the hippocampus[1, 2, 3] which have been confirmed by examinations of Northern blot, Western blot and immunochemistry. Other evidence is gained from positron emission tomography (PET) scans, which have revealed hypometabolism in the cerebellum of the DMD subjects studied, and a varying amount of reduced activity in the cerebral hemispheres in these boys. [12]

Dystrophin has been found to be present in the post-synaptic density (PSD) of one normal brain and absent in the same area in a brain from a child with DMD.[13] The PSD is a disc-shaped proteinaceous structure beneath the post-synaptic membrane in chemical synapses. It is thought to play a critical role in synaptic function by stabilising the synaptic structure, anchoring the postsynaptic receptors and transducing signals.[13]

It has long been known that dystrophin exists in many isoforms; these are expressed in different tissues.

There are three full-length isoforms and several that are shorter. The 'brain isoform' of dystrophin was first discussed in the early 1990s.[12] It has been hypothesised that the variation in cognitive functioning of boys with DMD could be partially attributed to the different locations of the mutations on the very large gene and therefore the involvement of these isoforms and the expression of dystrophin in the areas that they control.

The full-length dystrophin DP427 exists in three sub-types: PP427m, DP427c and DP247p. Two of these are expressed in the central nervous system and, with two of the shorter isoforms DP140 and DP71, have proven significance in cognitive function.

The full-length isoform DP427b is found predominantly in neurones of the cerebral cortex and parts of the hippocampus.

The full-length isoform DP427P drives nearly all of the expression of dystrophin in the cerebellum. In recent years there has been increasing interest in the role played by the

cerebellum in processing sensory information, short-term memory, attention, impulse control, emotion, higher cognition and the ability to schedule and plan tasks.[15] The hypotheses connecting cerebellar dysfunction with dyslexia, ADHD and autism all stress links to a deficit in the coordination of sensory data. There is a higher incidence of ADHD, autism and obsessive-compulsive disorder in boys with Duchenne than in the general population.[14]

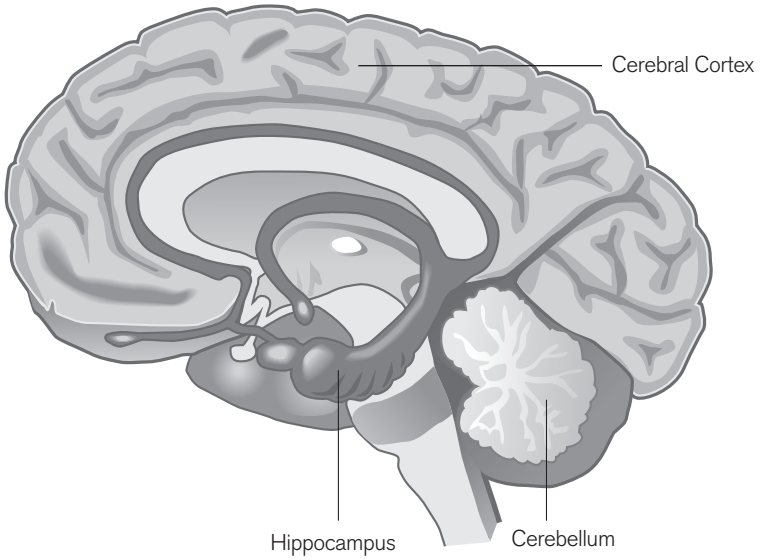
Intellectual impairment appears to be more frequent in those with distal deletions of the gene (including exons 45 through to 54 and upstream of exon 62).

Of the two shorter dystrophin isoforms, DP 140 (promoter upstream of exon 45) shows significant association with mental retardation and its product is found throughout the nervous system cortex, cerebellum, hippocampus, brainstem and spinal cord.

The influence of the other short isoform DP 71 also appears to be significant. The promoter for this isoform is between exon 62 and 63.[16]

Although there is a statistically significant difference between patients with proximal (versus distal) deletions, there are many exceptions documented.

While one-third of the boys with Duchenne have a non-progressive intellectual impairment, others show varying degrees of the recognised cognitive profile attributed to this condition. In others, some cognitive function remains relatively unaffected, with average to above average scores.



Areas of the brain where dystrophin is present

References

1. Dubowitz V. Muscle disorders in childhood, 2nd ed. London; Philadelphia: Saunders; 1995
2. Dubowitz V. The Muscular Dystrophies." Postgraduate Medical Journal 1992;68: 500-506
3. Dubowitz V. Forty years of Neuromuscular Disease, A Historical Perspective. Imperial College of Science, Technology and Medicine, Hammersmith Campus, London, UK. New Perspectives in Paediatric Neuromuscular Disorders Conference, Sydney. 1998
4. Brown SC. and Lucy JE. Dystrophin, Gene Protein and Cell Biology, Cambridge University Press. 1997
5. Lalic T, Vossen RH, Coffa J, et al. Deletion and duplication screening in the DMD gene using MLPA. *Eur J Hum Ge*net 2005;13: 1231–34
6. Entries in the Leiden Duchenne muscular dystrophy mutation database: An overview of mutation types and paradoxical cases that confirm the reading-frame rule - Aartsma-Rus - 2006 - Muscle & Nerve - Wiley Online Library
7. Anthony P. Monacoc, Corlee J. Bertelson, Sabina Liechti-Gallati, Hans Moser and Louis M. Kunkel An explanation for the phenotypic
8. Anderson JL. Satellite cell Activation and Muscle Repair. Proceedings of Duchenne in Millennium Parent Project Conference, Pittsburgh PA. 2000.
9. Hoffman EP Dystrophin gene protein and Cell Biology Proceedings of Parent project conference, University of Pittsburg PA. 1997.
10. Rando T. Regulation of oxidative damage as potential therapeutic approach. Proceedings of Parent Project Conference, University College of Los Angeles. 1999.
11. Anderson J. Satellite cell activation and muscle cell repair, Proceedings of PPMD conference 2000 Pittsburg. 2000
12. Spencer M. Clinical trials for increasing muscle mass. Proceedings of Parent Project Conference Research Treatment Hope, University of California, Los Angeles. 1999
13. Bresolin N, Castelli, E et al. Cognitive Impairment in Duchenne Muscular Dystrophy. Neuromuscular Disorders 1994;4(4): 359-369.
14. Kim TW, Wu K, et al. Deficiency of brain synaptic Dystrophin in human Duchenne Muscular Dystrophy. Annals of Neurology 1995;38(3): 446-44
15. Henderiksen JG, Vles JS. Neuropsychiatric disorders in males with duchenne muscular dystrophy: frequency rate of attention-deficit hyperactivity disorder (ADHD), autism spectrum disorder, and obsessive-compulsive disorder. J Child Neurol 2008;23(5): 477-81.
16. Bower JM, Parsons LM Rethinking The Lesser Brain. Scientific American 2003; 2003.
17. Blake DJ, Kröger S. The neurobiology of Duchenne muscular dystrophy: learning lessons from muscle? Trends Neurosci. 2000;Mar;23(3):92
18. Hoffman EP. Dystrophin Gene product and cell biology; Proceedings of Parent Project conference University of Pittsburg; 1997.
19. Hoffman EP, Brown Jr. RH, Kunkel LM. Dystrophin: the protein product of the Duchenne muscular dystrophy locus. Cell 1987;51(6):919–928.
20. Nicholson LV, Johnson MA, Bushby KM, Gardner-Medwin D, Curtis A, Ginjaar IB. Integrated study of 100 patients with Xp21 linked muscular dystrophy using clinical, genetic, immunochemical, and histopathological data. Part 1. Trends across the clinical groups. J Med Genet 1993;0(9):728–36.
21. Nicholson LV, Johnson MA, Bushby KM, Gardner-Medwin D, Curtis A, Ginjaar IB. Integrated study of 100 patients with Xp21 linked musculardystrophy using clinical, genetic, immunochemical, and histopathological data. Part 2. Correlations within individual patients. J Med Genet 1993; 30(9): 737– 44.
22. Nicholson LV, Johnson MA, Bushby KM, Gardner-Medwin D, Curtis A, Ginjaar IB. Integrated study of 100 patients with Xp21 linked muscular dystrophy using clinical, genetic, immunochemical, and histopathological data. Part 3. Differential diagnosis and prognosis. J Med Genet; 1993.
23. Muntoni F, Torelli S, Ferlini A. Dystrophin and mutations: one gene, several proteins, multiple phenotypes. Lancet Neurol 2003;2: 731–40.11.
24. Bushby, KM, Goodship JA, Nicholson LV, et al. Variability in clinical, genetic and protein abnormalities in manifesting carriers of Duchenne and Becker muscular dystrophy. Neuromuscul Disord 1993; 3(1): 57–64.
25. Kunkel LM, Monaco AP, Hoffman E, Koenig M, Feener C, Bertelson C. Molecular studies of progressive muscular dystrophy (Duchenne). Enzyme 1987;38(1-4):72–75
26. Emery AEH. (The Facts). 2nd edition Oxford university press; 1993
27. Gorospe JR, Hoffman EP. Duchenne muscular dystrophy. Curr Opin Rheumatol 1992;4(6):794–800
28. Matsumura K, Ohlendieck K, Ionasescu VV, et al. The role of the dystrophin-glycoprotein complex in the molecular pathogenesis of muscular dystrophies. Neuromuscul Disord 1993; 3(5-6): 533–535

Chapter Three:
Diagnosis

Early clinical features

A diagnosis of Duchenne muscular dystrophy may be considered at a very young age in the presence of some of the following early clinical features:

- mobility problems:
  - delayed motor milestones
  - gait abnormalities
  - difficulty running
  - difficulty climbing
  - frequent falls
  - perceived as clumsy or lazy
  - toe-walking
  - Gower's manoeuvre (see accompanying DVD, Duchenne Muscular Dystrophy: A Clinical Profile)
- hypertrophy of some muscle groups especially gastrocnemius but also deltoids, quadriceps and temporalis. Macroglossia of the tongue is present in some boys.
- cognitive or language delays may be the first presenting symptom and may overshadow the physical symptoms. Many are alerted to a possible diagnosis of DMD because of a delay in language acquisition, word-finding difficulties, influent speech or other signs of a deficit in auditory working memory and especially if these symptoms co-exist with other developmental delays especially gross motor difficulties.[1]
- behaviour problems are more common than in the general population, with autistic features evident in a small minority. Others have attention deficit difficulties and problems with obsessive and/or compulsive behavior or anxiety.
- liver function tests. An increase in transaminases (aspartate aminotransferase and alanine aminotransferase) would suggest further investigations for DMD before undertaking a liver biopsy.[1]

It is becoming common medical practice to test CPK levels in all boys presenting with developmental delay in motor and language areas.

Family history

An X-linked family history of muscular dystrophy when there are clinical signs of Duchenne is often sufficient for diagnosis. However molecular diagnostic tests are essential for detection of female carriers, prenatal diagnosis, genetic counselling and access to mutation-specific therapies.

Diagnostic tests

Blood tests

Blood tests are used to ascertain levels of creatine phosphokinase (CPK) in the blood serum. Raised CPK levels are an invariant finding in DMD. Normal levels of CPK in males are 30 to 200 U/litre, while females are 30 to 140 U/litre. In DMD, these may be elevated 10 to 100 times. Seventy percent of DMD carriers have raised CPK levels, however normal levels do not preclude the carrier status.

Molecular diagnostic tests

Molecular genetic testing is now considered essential in order to determine the exact mutation to permit genetic counselling, prenatal diagnosis and to assess whether emerging therapies, which are mutation-specific, are appropriate for the child.

The most commonly available DNA-based test is Multiplex polymerase chain reaction (MutiplexPCR), which directly analyses 18 of the 79 exons in the dystrophin gene from a small peripheral blood sample. This method is thought to pick up 98 per cent of deletions from the two 'hot spots' (proximal exons from 2 to 20 and distal 'hot spot' 44 to 55) where most mutations are located. PCR does not always identify the boundaries of the mutation, which are needed for some of the mutation-specific therapies that are undergoing, or about to undergo, clinical trials.

Positive results of a mutation by PCR will confirm the diagnosis of dystrophinopathy (either DMD or BMD). However, the absence of a deletion in either of these spots does not exclude DMD or BMD: 40 per cent of Duchenne cases and 20 per cent of Becker have another form of mutation (duplication or point mutation) or a deletion in another part of this large gene. A definitive diagnosis, with full characterisation of the mutation and its exact boundaries, will require further investigation by one of the other methods offered now in many laboratories:

- multiplex ligation-dependent probe amplification (MLPA) has been developed for deletion and duplication analysis of the whole gene for both probands and carriers.[1] This method is becoming more widely available and is currently the choice of Queensland Health.
- single condition amplification/internal primer (SCAIP) is another method favoured by some laboratories as it will detect deletions and duplications and provide sequence data.[4]
- Multiplex amplifiable probe hybridisation (MAPH) will detect both deletions and duplications of all 79 exons.[5]

- a muscle biopsy-based diagnostic approach was developed and optimised to increase the mutation detection frequency to nearly 100 per cent.[6] This method utilises protein- and RNA-based analyses in combination with direct cDNA sequencing and is favoured in some centres.
- if deletion or duplication testing is negative, then full dystrophin gene sequencing should be performed by one of the methods in order to look for small deletions, insertions or point mutations.[4]

Prognosis of the severity of the condition can be made with 90 per cent accuracy—with reference to the reading frame—if the boundaries of the deletion are known (see The reading frame hypothesis, page 8).

Muscle biopsy

When a genetic diagnosis is available, it is no longer considered necessary to perform a muscle biopsy. However, when the diagnosis is not clear it can be invaluable in not only assisting with the differential diagnosis but also to determine the degree of the dystrophinopathy (whether a Duchenne or Becker MD).

Microscopic examination of frozen dystrophic muscle tissue cut across the longitudinal axis reveals:
- variation in fibre size
- presence of dead fibres and groups of necrotic cells
- regenerating fibres
- centrally placed nuclei
- proliferation of adipose and connective tissue
- the presence or absence of other proteins from the dystrophin-associated glycoprotein complex.

Dystrophin protein analysis

Immunochemistry and immunoblotting for dystrophin on tissue from a muscle biopsy not only provides accurate diagnosis of dystrophinopathy but also can reliably differentiate between Duchenne and Becker.

If dystrophin is absent or only present in very small amounts (3 per cent or less), a diagnosis of Duchenne will be made. When dystrophin is present but reduced and is of abnormal molecular weight, Becker muscular dystrophy is the probable diagnosis.

If dystrophin of normal molecular weight but reduced quantities is found, the primary deficiency could be in one of the associated trans-membrane glycoproteins.[8]

Prenatal testing

Prenatal testing can be carried out by DNA analysis of cells collected through:

- chorionic villus sampling between 11 and 12 weeks gestation. The rate of foetal loss from this procedure is under 1 per cent
- amniocentesis, which can be performed at between 14 and 16 weeks. There is a very small risk of inducing miscarriage (0.05 per cent)
- foetal muscle biopsy of a small subcutaneous muscle sample taken at 18 to 24 weeks. This sample is processed to detect the presence of dystrophin.[7]

References

1. Bushby Finkel R, Birnkranr D, Case L, Clemens P, Cripe L, Kaul A, KinnetK, McDonald K, Pandya P, Poysky P, Shapiro F, Tomeszco J, Conatantin C. For care consideration working group 2010. Diagnosis and management of Duchenne muscular dystrophy Part 1: diagnosis, and pharmacological and psychosocial management Lancet 2010;Jan.
2. Gatta V, Scarciolla O, Gaspari AR, Palka C, De Angelis MV, Di Muzio A, Guanciali-Franchi P, Calabrese G, Uncini A, Stuppia L. Identification of deletions and duplications of the DMD gene in affected males and carrier females by multiple ligation probe amplification (MLPA). Hum Genet 2005;Jun;117(1):92-8. Epub 2005 Apr 20.
3. Lalic T, Vossen RH, Coffa J, et al. Deletion and duplication screening in the DMD gene using MLPA. Eur J Hum Genet 2005;13: 1231–34
4. Flanigan KM, von Niederhausern A, Dunn DM, Alder J, Mendell JR, Weiss RB. Rapid direct sequence analysis of the dystrophin gene. Am J HumGenet 2003;72: 931–39.
5. Dent KM, Dunn DM, von Niederhausern AC, et al. Improved molecular diagnosis of dystrophinopathies in an unselected clinical cohort. Am JMed Genet A 2005;134A:295–9
6. Deburgrave, N, Daoud, F, Llense, S, Barbot, JC, Recan, D, Peccate, C, Burghes, AH, Beroud, C, Garcia L, Kaplan JC, Chelly J, Leturcq F. Protein- and mRNA-based phenotype-genotype correlations in DMD/BMD with point mutations and molecular basis for BMD with nonsense and frameshift mutations in the DMD gene. Hum Mutat. 2007.
7. Hoffman EP, and Wang J. Duchenne-Becker Muscular Dystrophy and the Non - Dystrophic Myotonias: Paradigms for Loss of Function and Change of Function of Gene Products. Archives of Neurology 1993;50:1227-1235.
8. Hoffman EP Proceeding of PPMD conference university of Pittsburg.1998.

Chapter 4:
Clinical Features

The condition affects males almost exclusively. Rare female cases occur when there is only one active X chromosome (with a mutation at Xp21) or when there has been an X autosomal translocation, which disrupts the other dystrophin gene in the second X chromosome. Females who have a dystrophin gene mutation on one of their X chromosomes are 'carriers' of the condition, most of whom are asymptomatic.

MANIFESTING CARRIERS

Approximately 10 per cent of all carriers are 'manifesting' carriers, which is the result of random X inactivation. In the early stage of the life of each cell one of the X chromosomes preferentially switches off. If a greater percentage of the 'good' X chromosomes are turned off in muscle tissue, then a large number of the muscle fibres will not produce dystrophin. The carrier can then show varying amounts of proximal muscle weakness, fatigue, hypertrophy of some muscle groups and occasional cardiomyopathy.

Occasionally, the 'good' X is preferentially switched off in most cells during the process of random X inactivation leading to a severely diminished production of dystrophin and a very severe presentation of weakness in the manifesting carrier, occasionally this resembles the male phenotype. However, unlike the symmetrical distribution of weakness in boys who have DMD, the weakness in manifesting carriers can be quite asymmetrical because of the random nature of the X inactivation.

Early problems in DMD include:

- delayed motor milestones
- mobility problems (e.g. abnormal gait, frequent falls, toe-walking and difficulty with climbing stairs)
- delayed speech acquisition.

Behavioral problems may be the first clinical symptoms.

Musculoskeletal function

Hypertrophy of some muscles is common, especially gastrocnemius, deltoids, quadriceps, extensors of the forearm and the temporalis. A small number of boys have macroglossia severe enough to produce a fleshy protruding tongue. The primary problems are the progressive symmetrical muscle weakness and gait anomalies.

Relentless progression of muscle weakness is the primary disability in Duchenne muscular dystrophy. The muscles of the pelvic girdle are affected first (abdominals, abductors and extensors of the hip). Shoulder girdle weakness also occurs, but it is evident a little later. Weakness then progresses distally, affecting quadriceps (hamstrings are relatively strong) and the anterior compartment of the lower leg while those in the posterior compartment, gastrocnemius, soleus and tibialis posterior remain stronger than anterior tibial muscles. Neck flexors often show weakness from a very early age.[1, 2, 3, 4, 5, 6]

In the normal course of the condition, ambulation ceases at around 10 years of age. However, steroid treatment has been shown to prolong the child's ability to walk for some years. Upper limb weakness follows soon after lower limb weakness. Fine motor muscles are relatively spared for some time.

The specific pattern and progression of muscle weakness produces the easily identified gait and stance of boys with DMD. Early weakness in the abdominals and hip extensors leads to the adoption of a wide-based lordotic stance. This stance shifts the line of gravity so that it passes behind the fulcrum of the hips and in front of the knees, which, in combination with the widened base of support, improves the boy's stability in standing and walking (see accompanying DVD, *A Clinical Profile*).

Hip abductor weakness leads to a loss of pelvic stability in the swing phase of gait. The boy will walk with either a bilateral Trendelenburg gait (with the pelvis dipping down on the side of the non-supporting leg) or the more common 'waddling gait' where there is lateral side flexion of the trunk over the supporting leg with backward, as well as lateral, lean.

Toe-walking may be a necessary biomechanical adaptation to the altered alignment. It is also thought to occur because of the relative imbalance in strength between the dorsi and plantar flexors of the ankle. Forceful contractions of the triceps surae (in the role of ancillary knee extensors) also help to propel the body forward and assist failing quadriceps to brace the knees (see accompanying DVD, *A Clinical Profile*).


Gower's sign

In the early ambulatory stage, many boys compensate for a lack in range of dorsiflexion by pronating the feet with subsequent external rotation at the hips. (Pronation involves plantar flexion at the talo-crural joint, eversion at the subtalar joint and abduction at the mid-tarsal joint.)

Gower's manoeuvre is used by boys who have weak anti-gravity muscles to enable them to rise from the floor to standing. It is considered a diagnostic sign of DMD and is well illustrated in the A Clinical Profile DVD.

Decline in muscle function

Most skeletal muscle is made up of a mixture of the three types of muscle fibres: fibre type 1, fibre type 2A and fibre type 2B.

Type 1 or slow-twitch muscles respond more slowly and have less force but they have a higher resistance to fatigue. Type 1 fibres are found in higher proportions in the postural muscles and are retained longer in dystrophic muscle.

Type 2 or fast-twitch muscles fire quicker and with more force and are geared for hard, short bursts of activity. They are found in abundance in the muscles of the legs, pelvis, shoulders and arms. They can be further differentiated into Type 2A and Type 2B.

Fast-twitch muscle fibres, and especially fibre type 2B, appear to be affected first in DMD. This could be because of their low resistance to fatigue. They have fewer mitochondria and therefore energy stores, and fewer small blood capillaries, so oxygen and nutrient supply may not be as readily available. They also have low myoglobin content and anaerobic respiration.

The relative composition of fibre types of various muscles will depend on the workload of that muscle and can in part explain why some muscle groups remain stronger than others.

The extraocular muscles around the eyes are known to be spared in DMD and they could provide a better understanding to the process of muscle degradation.

TYPES OF EXERCISE

There is sufficient evidence from many sources to suggest that eccentric exercise causes more damage to the muscle membrane. It is also known that the ability of the muscle to repair itself is eventually exhausted, therefore it is likely that those muscles subjected to repetitive high-load eccentric work will be affected earlier and more severely than others.

The early weakening extensor muscles often work eccentrically in standing and walking. The extensor muscles affected early in the condition include the gluteals (maximus). When walking, they contract concentrically to extend the hips or eccentrically to control the rate or amount of flexion at this joint. Other gluteal muscles (medius and minimus) work to keep the pelvis level when one foot is lifted. The quadriceps straighten the knee in the early swing phase of walking (working concentrically) and then gradually control the bending of the supporting leg as the other leg swings through in walking (eccentric muscle action).

The dorsiflexors lift the foot up so that the initial contact with the ground is with the heel when walking (a concentric muscle contraction) and then control the release of the foot to the floor after heel strike, which is a strong eccentric activity.

The flexor groups of muscle at the hip and knee joints are relatively strong, as are the plantar flexors (the calf) of the ankle. The hip flexors are the only muscles that work only concentrically during walking. When the regeneration of damaged muscle slows and eventually fails, muscle weakness becomes more evident and walking patterns change to accommodate the alteration in the balance of strength around the joints of the lower limb.

Progression

The pattern and progression of weakness follows a fairly stereotypical pathway. Weakness in the hip extensors occurs first and leads to a forward-tilting pelvis and lordosis. Weakening abductors fail to hold the pelvis level during walking, producing either a waddling or bilateral Trendelenburg gait. Failing strength in the quadriceps at the front of the thigh can lead to toe-walking because boys use forceful contractions of the gastrocnemei (in their role of ancillary knee extensors) to help brace the knees. The uneven balance of strength between the stronger calf and the muscles of the anterior compartment of the lower leg contributes to loss of range in the ankle, the loss of heel strike, the development of contractures in the ankle, and varying degrees of toe-walking.

Secondary problems in Duchenne Muscular Dystrophy

Contractures

Contractures develop because of unequal loss of muscle power in opposing muscle groups. They can also be the result of postures and gait patterns that are adopted because of the progressive weakness. Although muscle weakness is the primary disability in DMD, contractures can be the most disabling in the later stages. Contractures are not always symmetrical, as many boys adopt an asymmetric pose consistently favoring one side (as shown in the DVD).

The likely contractures in the lower limb are:

- equinus in the ankle initially, followed by equinovarus deformity especially when weight-bearing has ceased. The 'propping' leg in an asymmetrical stance will develop a more severe equinovarus deformity as well as an increase in knee and hip flexion contractures
- flexion contractures of knees
- hip flexion contractures
- iliotibial band tightness.

Asymmetry in contractures fosters the development of scoliosis.

In the upper limbs, contractures are more likely to develop after ambulation is lost and arms are no longer dependent:

- flexion contractures of the elbows
- limitation of full supination of the forearm
- flexion contractures of the wrist, metacarpophalangeal and proximal interphalangeal joints (long finger flexors).

Spinal deviations, scoliosis, etc.

The ambulatory boy has a lordotic spine when standing but a reversal of the normal lumbar lordosis when sitting. The development of scoliosis was thought to be almost universal in boys with DMD within two years of full-time wheelchair use (approximately 90 per cent). Some curves progressed very rapidly as much as 0.3 to 4.5 degrees per month.[9] With greater acceptance of steroid medication worldwide, the incidence of spinal curvatures has been greatly reduced.[8] Better attention to seating and postural support has also reduced the occurrence of spinal deviations.

Attempts have been made to identify those patients who have a poor prognosis for rapid collapse of the spine and thereby ensure the best timing for surgery.

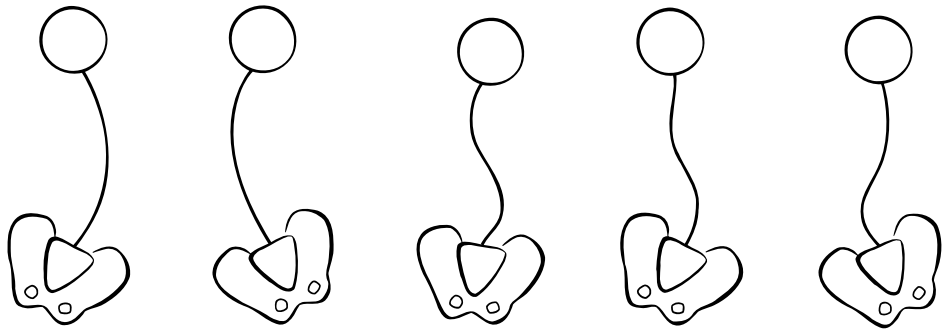
Gibson and Wilkins (1975) identified two pathways:

- stable pathway—when the rate of progression of the curve is slow and never compromises comfort, respiration or appearance. They hypothesised that retaining the extension of the spine protected it from scoliosis by locking the posterior facets. However, other studies have not supported this hypothesis.

- unstable pathway—leading to rapid spinal collapse, extreme discomfort, an inability to maintain 'hands-free' sitting balance, further restriction in vital capacity and abdominal discomfort.

Oda et al. (1993) defined three distinct pathways that could be useful in determining the need and optimal timing of spinal instrumentation:

- type 1 relates to the 'unstable pathway' of Gibson and Wilkins, in this there is an unremitting progression of scoliosis often in the presence of kyphosis (this is often referred to as a 'collapsing spine'). The curve reaches 30 degrees at between 9 and 15 years and progresses rapidly thereafter. Frontal plane curves can present as single curves in the lumbar spine, extending to include the thoraco-lumbar spine or presenting as double major curves. Pelvic obliquity is an almost invariant feature, with the iliac crest higher on the side of the concavity. Pelvic obliquity is always more severe when there is only a single curve.

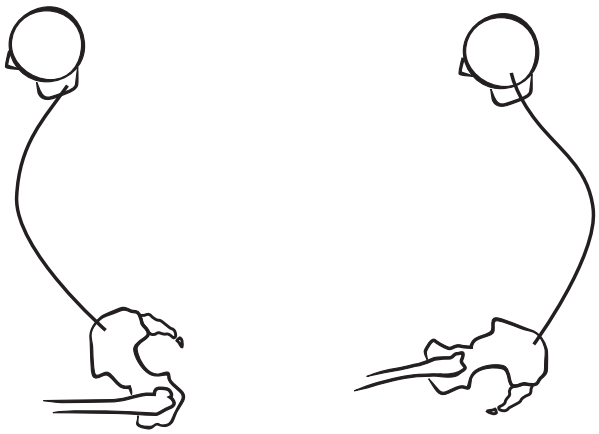


Thoraco-lumbar scoliosis —convex to the right | Thoraco-lumbar scoliosis —convex to the left | Double major —left thoracic, right lumbar | Right lumbar | Left lumbar

Possible deviations in the frontal plane.

- in type 2, the deviation initially occurs in the sagittal plane with the development of kyphosis. The spine then becomes lordotic (before 15 years) and finally hyperlordotic. This is accompanied by severe anterior tilting of the pelvis and a flattened thorax with a diminution in the anterior/posterior diameter, severely compromising respiration. Some type 2 patients follow a stable path, but in others frontal plane deviation and rapid collapse accompany the sagittal plane deformity. Boys with fixed hyperlordotic spines get contractures of the neck extensors, thus making surgical correction and supportive seating difficult (see seating section on page 66).
- type 3 are the stable curves, where there is less deformity (usually not progressing beyond 30 degrees), the curve does not progress with age and there is very little pelvic obliquity. This group has greater forced vital capacity than types 1 and 2 (in excess of 2 litres at 15) and needs to be distinguished from the other two groups, as surgery is not indicated (according to researchers Oda et al. (1993)).

Another deviation seen occasionally, though not documented in the above research, is severe thoracolumbar kyphosis. This severely compromises comfortable sitting in the wheelchair, respiratory excursion and digestion. If allowed to continue, this posture inevitably leads to contractures in the neck extensors.



Hyperlordosis with anterior tilted pelvis

Thoracicolumbar kyphosis with posterior pelvic tilt

Respiratory function

Dystrophic changes in the muscles of respiration are the major cause of the decline in respiratory function. The lungs are essentially normal and normal gaseous exchange will occur as long as the intake of air reaches the alveoli. The progressive weakness of intercostal muscles and the diaphragm restricts the ability to breathe deeply while the loss of strength in abdominal muscles and diaphragm inhibits effective coughing.

Chronic alveolar hypotension occurs when intake is restricted, resulting in severe carbon dioxide retention. Significant blood gas abnormalities occur first at night with chronic nocturnal hypoventilation occurring initially in the rapid eye movement phase of sleep, and then gradually extending throughout most sleep patterns. The blood gas alterations eventually extend to 24 hours a day.

Vital capacity (VC) in boys with DMD plateaus between 10 and 15 years (compared with 19 years in the general population) at about 1800mL on average.[13] VC is then lost at a rate of 200 to 250mL each year, with the rate of loss tapering off below 400mL.

The magnitude of the plateau is thought to indicate the severity of the condition and be a predictor of life expectancy. John Bach (2004) states: 'The long-term suppression of deep breaths or chronic hypoventilation leads to chronic microatelectasis and permanent loss of lung and chest-wall elasticity with decreased static pulmonary compliance'.

Weakness in expiratory muscles results in difficulty clearing secretions from lungs especially during upper respiratory tract infections, this can lead to mucous plugging, atelectasis pulmonary infiltrates and scarring. Boys with DMD are at greater risk of developing pneumonia because of the retention of excess secretions, which provides a rich medium for the growth of bacteria.[14, 15, 16]

Respiratory insufficiency used to be the most common cause of death in patients with DMD. However, since nocturnal ventilation and non-invasive daytime ventilation have become readily available as well as the increasing availability and use of the cough-assist machine, life expectancy has increased dramatically.

Cardiomyopathy

Cardiomyopathy is a common feature in DMD. It is caused by a deficiency of dystrophin in cardiomyocytes leading to a loss of cardiac muscle cells under the stress of constant contractions. The baso-lateral free wall of the left ventricle is most affected with muscle tissue becoming gradually thinner, floppy and unable to function properly (dilated cardiomyopathy). Respiratory muscle weakness leads to low blood-oxygen, which places an additional strain on the heart. Alterations in the input from the autonomic nervous system are evident. Conduction abnormalities are common. Cardiac arrhythmia with a sympathetic predominance resulting in sinus tachycardia is often present.

Although the heart muscle shows signs of the lack of dystrophin from conception, cardiac function usually remains normal until 10 years of age. By age 15, 26 per cent will have problems and approximately 45 per cent by 18 years of age.[17]

Some genetic deletions predispose a boy to have more cardiac problems. Jeffries (2005)31 identified deletion exons 31 to 42 were statistically significant for more cardiac involvement and, to a lesser extent, exons 10 to 12 and 18 to 30 (though not statistically significant). Deletions in

exons 51 and 52 appear to be protective and, although not significant, it would appear that deletions around 53 and 54 and 68 to 71 are less likely to involve cardiac involvement.

As progressive skeletal muscle weakness develops, very few demands are placed on the heart and boys with DMD often remain relatively free of overt cardiac symptoms. If present, they can include:

- weight loss
- cough
- nausea and vomiting
- increased fatigue.[33]

The development of dilated cardiomyopathy usually precedes the development of symptoms by years and surveillance from diagnosis is essential. An initial baseline evaluation at diagnosis is recommended, followed by cardiac screening every two years until the age of 10 and then annual screening.

Boys being treated with steroids have some protection from cardiac decline.[32] Boys on steroid medication need to be evaluated regularly for weight gain and hypertension. Systemic arterial hypertension should be treated and steroid dose may require adjustment.

In BMD, there is a higher incidence of overt heart disease, presumably because of the greater demands placed on the dystrophic heart musculature and the longer life expectancy.[33]

Cognitive function

Thirty per cent of boys who have DMD show a non-progressive intellectual impairment. Cotton[20] conducted an extensive meta-analysis of all cognitive research and concluded that although the mean IQ of boys with Duchenne, 80, falls in the low average range, they showed considerable heterogeneity with full-scale IQ ranging from 14 to 134, with scores following a normal distribution curve.

Analysis of scores from many boys (n=1224) has indicated that they perform significantly better on the performance tasks of the Weschler Intelligence Scale for Children—Revised (WISC-R) than they do on the verbal subtests. The analysis showed that this discrepancy between the verbal and performance scores is greater in the younger boys (up to nine years of age) with an unexpected improvement in the verbal component as they reached 14 years of age and beyond. There was no change in the performance sub-tests or full IQ score and, therefore, it is independent of the increasing functional impairment.

Current research suggests that many boys with DMD have a similar cognitive profile and share the same spectrum of neuropsychological strengths and deficits, regardless of their intelligence. [36, 37, 38, 39, 40, 41]

Strengths

The strengths of the cognitive profile are:

- good vocabulary and naming skills
- good ability to categorise
- good ability for rote learning, both visually and verbally
- excellent memory for detail, especially visual detail
- good auditory discrimination
- good ability to speak fluently
- good general knowledge.

Weaknesses

Many of the learning difficulties found in boys with Duchenne are associated with language and its acquisition. They are often evident before diagnosis and present as a delay in learning to speak. Approximately 40 per cent of boys without a known history of Duchenne have language delay as the first identifiable symptom.

Researchers working in the cognitive field describe learning and behavioral problems differently but all identify three main areas, which may cause problems for some boys:

- verbal or language-based intelligence – learning and memory, especially short-term and working memory
- attention-focusing and regulation – 18.7 per cent, compared with 7.5 per cent in general population[27]
- difficulties with emotional interactions and a higher incidence of neuropsychiatric problems e.g. attention deficit hyperactivity disorder, autistic spectrum disorder and obsessive compulsive disorder.[27]

These weaknesses are discussed below.

Verbal intelligence and language-based learning

Phonological processing

Many researchers have identified the difficulties boys with DMD have mastering the phonological code and acquiring automaticity of learning:

- Dr Mark Mehler in Wahl (1997) believes that the delay in the acquisition of language and continuing difficulties in verbal skills is attributable to the difficulty in breaking down language into the smallest units of sound, the phonemes. Storing and retrieving phonemes is difficult for boys with DMD and acquiring automatic processing is difficult. The ability to use the phonological code is essential for processing the spoken language and subsequently for remembering and retrieving it, as well as learning to read.
- Billard et al. (1998) likened the problem to that experienced by children with dysphonetic dyslexia because of the poor use of graphophonological conversion rules. This is well demonstrated by the greater difficulty experienced by boys with DMD in reading 'non-words'.
- Dorman et al. (1988) reported that DMD children relied heavily on their visual abilities to compensate for poor auditory-based word-attack skills.

Any impairments in acquisition of phonological knowledge disrupts phonological processing and reading:[17, 26]

Hendriksen JG and Vles JS (2006)28 found a 40 per cent higher risk of problems learning to read in boys with DMD which did not correspond to their IQs. This compared to a 5 per cent risk in the general population. They attribute this to the known difficulty boys with DMD have in decoding letters into sounds and phonemes.

Veronica Hinton[35, 36, 38, 39, 40] described the major difficulty as 'limited verbal span', as evidenced in problems processing and retaining sequentially presented information. Cyrulnik SE & Hinton 200744 hypothesised that this automaticity of decoding skills is attributable in part to the lack of dystrophin in the cerebellum. In their hypothesis, the difficulties with verbally mediated

intelligence arise, in part, as a result of the lack of dystrophin in the Purkinje cells of the cerebro-cerebellar pathways of the cerebellum. It is thought that the cerebro-cerebellar loops could be responsible for skilled mental performance similar to skilled motor performance—a long-attributed function of the cerebellum.

Attention focusing and verbal working memory

Many boys are reported to have attention deficit disorders (with or without hyperactivity) because of an inability to focus on tasks or maintain attention for long – 18.7 per cent of boys with DMD compared with 7.5 per cent of general population.[25, 45] Conversely, there is often great difficulty 'letting go' attention with prolonged perseveration on tasks, objects or activities. This inability to modulate attention makes a true assessment of intellectual capabilities difficult. The frontal lobes of the cerebral cortex (known to express dystrophin in the general population) are thought to regulate attention and assist higher executive functioning such as planning and organisation. Hinton,[36] in part, attributes inattentiveness to having a limited verbal span and is often evident in:

- inability or difficulty in following instructions which consist of several parts
- inability or difficulty in recalling a story (boys often remember the parts well, but are unable to retell the plot from beginning to end)
- problems with sentence repetition
- recall of digits (digit span subtests of WIS are consistently depressed).[38, 46]

Several investigators have argued that the cerebellum plays a major role in the coordination of cognitive function and especially in automatisation of all skilled learning. There is a growing consensus that the cerebellum is part of a network mediating verbal working memory and processing of all phonological material, although it does not appear to be involved in spatial working memory.

The hippocampus is also one of the sites in the brain where dystrophin is located and it is here that short-term memories are processed into long-term memories.[29]

Commonly identified deficits in verbal working memory can result in, or coexist with, many other weaknesses such as impulse control, planning, organisation, emotional regulation, mental flexibility, independent task initiation, abstract concept formation and self-monitoring. All of these cognitive abilities rely on the same or similar brain structures and pathways.

Emotional interaction

Some boys have difficulty with emotional interactions, which Mehler[29] describes as a lack of 'connectedness', and that the boy may appear immature. Understanding complex social situations is sometimes difficult and can strain peer relationships. School staff and parents mention a higher incidence of autistic-type behaviours even in those not classified on the autistic spectrum continuum. Many parents report that their sons have difficulty problem-solving, and dislike games where the rules are likely to change. Many boys resent and have difficulty accepting change in any form, even when it is to their advantage.

The role of the cerebellum, once thought to be responsible only for learning and fine-tuning skilled motor movement and coordination, is now thought to be involved in coordination of cognitive function and especially in automatisation of all skilled learning information processing, all verbal cognitive tasks and social connectedness. There is also an increasing interest in the role that cerebellar dysfunction plays in the development of autistic spectrum disorders, ADHD and sensory processing anomalies.

Psychological or behavioral problems may be present

Hendriksen J & Vles JS 2008[27] reported on their questionnaire-based survey of 351 males with DMD in the Netherlands and the United States. They found a higher prevalence of the following neuropsychiatric disorders:

- Attention-Deficit Hyperactivity Disorder (ADHD) 11.7 per cent (more than twice as high as in the general population)
- Autism spectrum disorders (ASD) 3.1 per cent (1 per cent in the general population)
- Obsessive-compulsive disorders 4.8 per cent (twice as high as in the general population).

Their survey also reported a co-morbidity of all three neuropsychiatric disorders in 11 of the 351 boys surveyed, with ADHD and OCD being the most common co-morbidity. The authors recommend further research to discover if boys with an intellectual impairment or with specific deletions may be those at greater risk of neuropsychiatric disorders.

It is important to remember that this is group information only; there is just as much variability in cognitive abilities within the DMD population as there is in the general population.

Smooth muscle involvement

Little research has been done on the role of dystrophin in smooth muscle to date. Further research could shed light on the differences noted in the urinary tract and circulatory systems, particularly with reference to the increased blood loss during surgery in males with DMD. Smooth muscle is found in the walls of all hollow organs of the body except the heart, helping to regulate the flow of blood in arteries, the progress of food through the intestines and the elimination of urine. The flow of air through the lungs is also influenced by smooth muscle. There is known to be a lack of dystrophin in smooth muscle, which could affect function in these systems.

Urinary dysfunction is an occasional problem. M MacLeod, R Kelly, SA Robb and M Borzyskowski[42] examined bladder dysfunction in 87 boys and young men with DMD. They found urinary problems occurred in some young, ambulant boys as well as wheelchair-dependent patients. Day and night-time incontinence, nocturia, urinary frequency, urgency and stress incontinence were found in the boys at a mean age of 10.3 years (range 3–25 years). Urinary hesitancy affected an older group with a mean age of 16.7 years (range 11–22 years).

Of the group, 27 patients had symptoms severe enough to require follow-up. While the pathology is not fully understood and difficult to explain, McLeod et al. concluded that myopathic involvement of the detrusor would be expected to reduce the strength of contractions and cause a large capacity, flaccid bladder and that weakness of striated muscle would be expected to result in stress incontinence. McLeod et al. have drawn attention to the presence of dystrophin isoforms in the brain, which could be implicated in impaired bladder control. Smooth muscle involvement was not questioned in this paper. In this study, 10 patients were treated with oxybutinin and all responded well to treatment, reporting a reduction in frequency, urgency and daytime incontinence. Nocturnal enuresis ceased. They all reported an improvement in quality of life.

Others[48] conclude that urinary incontinence in DMD is most often due to upper motor neuron

dysfunction and not due to a severe myopathy of the detrusor or external sphincter. The most likely causes of the UMN abnormalities were attributed to severe scoliosis or a complication of spinal fusion surgery.

In the gastrointestinal tract, constipation is a common feature, this could be the result of sluggish peristalsis, but, as it is a common problem for many people confined to wheelchairs, it may not result from lack of dystrophin in smooth muscle. Other boys report overactive bowels, with leakage not attributable to overflow around impacted faeces.

More research into the role of dystrophin in smooth muscle could help explain these commonly reported symptoms.

Endocrine issues, including growth and delayed puberty, are discussed on page 42.

Obesity

Obesity can be a problem for boys in wheelchairs as their daily dietary intake often far exceeds their energy requirements. Two distinct body types are seen consistently: boys who appear to gain weight very easily, irrespective of cortico-steroid medication and those who remain thin whatever their dietary intake. Steroid medication, now considered the gold standard of intervention, also contributes significantly to uncontrolled weight gain. Dietietic advice is recommended before medication commences, as success in maintaining a healthy weight is best achieved when the whole family adopts a healthy eating regime. (See page 48 on dietetics).

References

1. McDonald CM, Abresch RT, Carter GT, et al. Profiles of neuromuscular diseases. Duchenne muscular dystrophy. Am J Phys Med Rehabil 1995;74(5 Suppl): S70–92.
2. Siegel IM. Pathomechanics of stance in Duchenne muscular dystrophy. Arch Phys Med Rehabil 1972; 53(9): 403–406.
3. Siegel IM, Weiss LA. Postural substitution in Duchenne's muscular dystrophy. Jama 1982; 247(5):
4. Beenakker EA, Maurits NM, Fock JM, Brouwer OF, van der Hoeven JH. Functional ability and muscle force in healthy children and ambulant Duchenne muscular dystrophy patients. Eur J Paediatr Neurol 2005; 9(6): 387–93.
5. Johnson EW, Walter J, Zeiter. Lecture: pathokinesiology of Duchenne muscular dystrophy: implications for management. Arch Phys Med Rehabil. 1977; 58(1): 4–7.
6. Sutherland DH, Olshen R, Cooper L, et al. The pathomechanics of gait in Duchenne muscular dystrophy. Dev Med Child Neurol 1981; 23(1): 3.
7. Alman BA, Raza SN, Biggar WD. Steroid treatment and the development of scoliosis in males with Duchenne muscular dystrophy. J Bone Joint. Surg [Am]. 2004
8. Smith AD, Koreska J & Moseley CF. Hospital for Sick Children, Toronto, Ontario, Canada. The Journal of Bone and Joint Surgery. 1989:71(7):1066-1074
9. Gibson A, Wilkins KE. The management of spinal deformities in Duchenne Muscular Dystrophy. Clinical Orthopaedics. 1975
10. Oda, T. S., N., K. Yonenobu, et al. . Longitudinal study of spinal deformity in Duchenne Muscular Dystrophy. Journal of Paediatric Orthopaedics.1993;13:478-488.
11. Bach, J. R. (2004). Management of patients with neuromuscular disease. Philadelphia, Hanley & Belfus.
12. Bach, J. R. Pulmonary Rehabilitation Considerations for Duchenne Muscular Dystrophy: The Prolongation of Life by Respiratory Muscle Airs. Critical Reviews in Physical and Rehabilitation Medicine.1992;3(3): 239-269.
13. Finder, J. Respiratory care issues – Proceedings of Duchenne Parent Project Conference, Cincinnatti 2005. 2000
14. Finder, J. Giving a face to Duchenne MD - Understanding the Disease. Giving a face to Duchenne MD. H. Posselt, Parent Project Muscular Dystrophy. 2004
15. Finder, J, Birnkrant D, et al. Respiratory Care of the Patient with Duchenne Muscular Dystrophy. American Thoracic Society Consensus Statement. American Journal of Respiratory and Critical Care Medicine 2004;170:456-465
16. Larry Markham Proceedings of PPMD 2009 conference Atlanta
17. Adams AM, Gathercole SE. Limitations in working memory: implications for language development. International Journal of Language and Communication Disorders. 2000;35(1):95–116.
18. Dorman C, Hurley AD, D'Avignon J. Language and learning disorders of older boys with Duchenne muscular dystrophy.

Developmental Medicine and Child Neurology. 1988;30:316–327. [PubMed]

19. Cotton, S, Crowe SF, et al. Neuropsychological Profile of Duchenne Muscular Dystrophy. Child Neuropsychology. 1998:4(2): 110-117.
20. Cotton S, Voudouris NJ, et al. Intelligence and Duchenne muscular dystrophy: full-scale, verbal, and performance intelligence quotients. Developmental Medicine and Child Neurology 2001;43(7): 497-501.
21. Cotton, SM, Voudouris NJ, et al. Association between intellectual functioning and age in children and young adults with Duchenne muscular dystrophy: further results for a meta-analysis. Developmental Medicine and Child Neurology 2005;47(4): 257-65.
22. Billiard, C, Gillet P, et al. Reading Ability and Processing in Duchenne Muscular Dystrophy and Spinal Muscular Atrophy. Developmental Medicine and Child Neurology 1998;40:12-20.
23. Bresolin, N, Castelli E, et al. Cognitive Impairment in Duchenne Muscular Dystrophy." Neuromuscular Disorders 1994;4(4): 359-369.
24. Dorman C, Desnoyers Hurley A, & D'Avignon J. Language and learning disorders in boys with Duchenne muscular dystrophy Developmental medicine and child neurology 1988;30;316-327
25. Henderiksen JG, Poysky JT, et al. Psychosocial Adjustment in males with Duchenne muscular dystrophy: psychometric properties and clinical utility of a parent-report questionnaire. J Pediatr Psychol 2009;34(1): 69-78.
26. Henderiksen, JG and Vles JS. Are males with Duchenne Muscular Dystrophy at risk of reading disabilities? Pediatr Neurology 2006;34(4): 296-300.
27. Henderiksen, JG, Vles JS. Neuropsychiatric disorders in males with Duchenne muscular dystrophy: frequency rate of attention-deficit hyperactivity disorder (ADHD), autism spectrum disorder, and obsessive-compulsive disorder. J Child Neurol 2008;23(5):477-81.
28. Hendriksen JG, Vles JS. Are males with Duchenne muscular dystrophy at risk for reading disabilities? Pediatric Neurology 2006;34(4): 296-300.
29. Wahl M. The brain in Duchenne muscular dystrophy: are learning disabilities part of the picture? Quest 4 (1) Muscular dystrophy association publication. 1997
30. Wicksell RK, Kihlgren M, Melin L, Eeg-Olofsson O. Specific cognitive deficits are common in children with Duchenne muscular dystrophy. Dev Med Child Neurol. 2004;Mar;46(3):154-9.
31. Jefferies MD JL, Eidem BW, Belmont PhD JW, Craigen WJ, Ware SM, Fernbach SD, Neish SR, O'Brian Smith E, Towbin JA. Genetic Predictors and Remodeling of Dilated Cardiomyopathy in Muscular Dystrophy Circulation. 2005;112:2799-2804 Pediatr Cardiol. 2005;Nov-Dec;26(6):768-71.
32. Cripe L. Giving a face to Duchenne. DVD 2003.
33. Markham LW, Spicer RL, Khoury PR, Wong BL, Mathews KD, Cripe LH. Steroid Therapy and Cardiac Function in Duchenne. 2005.
34. Muscular Dystrophy. Pediatr Cardiol 2005.
35. Hoffman EP, Kunkel LM.Improved diagnosis of Becker muscular dystrophy by dystrophin testing 'Neurology' 1989;39:1011
36. Markham L. Proceedings of PPMD conference Atlanta GA, 2009.
37. Hinton V Cognitive skills in boys with DMD and BMD. Proceedings of focus on Duchenne, Duchenne Parent Project Conference, Pittsburgh PA. 1998.
38. Hinton V, De Vivo D, et al. Investigation of Poor Academic Achievement in Children with Duchenne Muscular Dystrophy. Learning Disabilities Research and Practice 2004;19(3):146-154.
39. Hinton V, De Vivo D, et al. Poor verbal working memory across intellectual level in boys with Duchenne dystrophy. Neurology 2000;54(11): 2127-32.
40. Hinton V, De Vivo D, et al. Selective deficits in verbal working memory associated with a known genetic etiology: the neuropsychological profile of Duchenne muscular dystrophy. J Int Neuropsychol Soc 2001;7(1):45-54.
41. Hinton V, Goldstein E. Duchenne Muscular Dystrophy. Neurogenetic Developmental Disorders: Variation of Manifestation in Childhood. M. M. Mazzocco and J. L. Ross. Cambridge, MIT Press: 2007;105-131.
42. Hinton V, Nereo N, et al. Selective deficits in Verbal working memory associated with a known genetic etiology: The neuropsychological profile of Duchenne muscular dystrophy. Journal of the International Neuropsychological Society. 2001;7:45-54.
43. Hinton VJ, De Vivo DC, et al. Investigation of poor academic achievement in children with Duchenne muscular dystrophy. Learning disabilities research and practice 2004;19(3):146-154.
44. Cyrulink SE, Hinton VJ. Duchenne muscular dystrophy A cerebellar disorder? Neuroscience and Behavioural reviews. 2007.
45. Mehler MF. Brain dystrophin, neurogenetics and mental retardation. Brain Res Rev. 2000;32:277–307.
46. Wicksell, RK, Kihlgren M, Melin L, Eeg-Olofsson O. Specific cognitive deficits are common in children with Duchenne muscular dystrophy. Developmental Medicine and Child Neurology 2004;46,154–159.
47. MacLeod M, Kelly R, Robb SA, Borzyskowski M. Bladder dysfunction in Duchenne muscular dystrophy. Arch Dis Child 2003;88:347–349
48. Caress JB, KMJ, Bauer SB, Shefner JM. Urinary dysfunction in Duchenne muscular dystrophy. Muscle & Nerve 1996(7):819-22.

Chapter 5:
Medical management

Major references:

1. Bushby K, Finkel R, Birnkrant DJ, et al. Diagnosis and management of Duchenne muscular dystrophy, part 2: diagnosis, and pharmacological and psychosocial management. Lancet Neurol 2010;9:77–93.
2. Giving a Face to Duchenne'. Parent Project muscular dystrophy. Two DVD set: Disc two 'steroids'

Medical management aims to slow the rate of decline and lessen the impact of secondary complications.

A paediatric neurologist or paediatrician often assumes the role of overseeing the management of boys with Duchenne muscular dystrophy in Australia. In other countries, a rehabilitation consultant or physiatrist may take on the role of case coordinator, but it can be equally well managed by a geneticist or any specialist who develops a special interest in the condition.

As well as the overseeing primary care physician, the child's medical team will include general practitioner, geneticist, respiratory physician, cardiologist, orthopaedic surgeon, gastroenterologist and endocrinologist as well as the services of a full allied health team including a dietician and neuropsychologist. The primary care physician will monitor all aspects of the child's development and condition to provide the anticipatory and preventative management and ensure referral to different sub-specialties at the appropriate time. The need for services of the sub-specialties will change as the condition progresses.

Steroid therapy

Cortico-steroids are the only proven effective medication to date. They are now considered the gold standard treatment for ambulant boys with DMD against which all other interventions are measured Many physicians recommend continuing treatment after ambulation is lost to preserve core and upper limb muscle strength as well as preserving heart and respiratory function. Both prednisone and its derivative, deflazacort, have been shown in randomised control trials to alter the natural course of DMD.[1, 2, 3, 4, 5, 6, 7, 8, 9, 10]

Major benefits of steroid therapy include:

- preservation of muscle strength and motor function – with prolonged ambulation to mid-teens on average
- improvement in, and prolonged stabilisation of, pulmonary function
- preservation of cardiac function
- delay in, or prevention of, need for scoliosis surgery
- retention of upper limb strength.

By preserving motor function for a longer time, there are significant benefits not only for the young man with DMD but also for the carers. Extension in the ability to perform daily tasks independently, by maintaining upper limb strength as well as extending the ability to roll over in bed, maintains independence for the young man and greatly reduces the burden of care on the parents both during the day and night.

Possible side effects include:

- excessive weight gain
- osteoporosis
- slowing of growth
- delayed puberty

- behaviour changes
- cataracts
- cushingoid facies
- gastritis
- hypertension
- glucose intolerance
- possible suppression of the immune system.

Boys who have pre-existing weight or behavior problems appear to be at greater risk of an exacerbation of these undesirable side effects when taking steroid medication.

Families with children on steroids are provided with information on all aspects of steroid medication including emergency care considerations that become necessary should the child become ill, sustain a fracture, and need surgery or a general anaesthetic. It is recommended that boys wear a Medic Alert bracelet.

It is also recommended that a health care provider with the appropriate expertise undertake the management of the child on long-term steroid medication so that monitoring and dose adjustments may be made as needed.

Initial dose – prednisone or prednisolone

The recommended initial dose is 0.75mg per kg per day. Some physicians choose to begin on a smaller dose of 0.5mg per kg per day. Alternatively, use 0.9 mg per kg per day of deflazacort if it is available.

Deflazacort should be considered first if pre-existing weight or behavior problems are present. Deflazacort is reported to produce fewer side effects, particularly with weight gain and behavior[6, 7, 8] however, it is not available in many countries. Deflazacort is also thought to produce more asymptomatic cataracts and possibly more growth retardation than prednisone. These claims are currently being investigated.

Physicians monitor response to the medication carefully, particularly in the first six months, and adjust the dose if the side effects are unmanageable or intolerable.

The American Academy of Neurology recommends that if side effects persist, to either offer the alternate medication, deflazacort, or to reduce the dose of prednisone slowly to as little as 0.3mg per kg per day. If this fails to produce the desired effect, alternative suggestions for controlling side effects are to consider one of the pulsed dose regimes:
- 10 days on, 10 days off
- alternate days
- weekend dose.

These alternative strategies are outlined in Bushby K, Finkel R, Birnkrant DJ, et al. Diagnosis and management of Duchenne muscular dystrophy, part 2: diagnosis, and pharmacological and psychosocial management. Lancet Neurol 2010; 9: 77–93.

It is well recognised that the daily dose given in the morning provides most benefit.

If no regime is effective in managing the unacceptable side effects, boys are weaned off the medication gradually.

Increasing the dose as the child grows

The dose may be increased incrementally as the child grows, to a maximum dose of 30mg per day for prednisone or prednisolone, or 36mg per day for deflazacort.

The dose may remain unaltered for a time if there has been no functional decline. When the child on a sub-target dose shows deterioration, then the dose is lifted in line with recommendations.

It is now recommended that steroid medication be continued even after loss of ambulation to preserve upper limb function, respiratory and cardiac function, and to protect the spine.

When to begin steroids

The general consensus is that steroids should be offered when there has been a plateau in development of physical skills and after the immunisation schedule (including chicken pox vaccine) has been completed. It appears that when prednisone or prednisolone or deflazacourt is administered relatively early in the disease process, there is more viable muscle to benefit from the medication. The plateau often occurs at around 4-6 years of age. Most physicians are not comfortable prescribing steroids too early because of the growth retardation and other side effects.

How do steroids work?

The reason for improvement in strength and function gained from steroid therapy is not clear, though it is thought to be associated with decreased numbers of dendritic cells and fibroblasts and increased numbers of satellite cells, in the dystrophic skeletal muscle and therefore could:

- reduce the inflammatory response
- possibly strengthen the plasma membrane
- improve the regeneration of damaged cells by activating satellite cells.

The considerable number and serious nature of the side effects associated with steroids have contributed to the cautious approach adopted by many physicians in the past, but, as steroid therapy has become accepted best practice in most countries around the world, it is now considered the 'gold standard' of treatment against which all other interventions are evaluated. Side effects are monitored and managed.

Cardiac evaluation and intervention

As a result of improved respiratory care in boys and young men with Duchenne over the past 20 years, dilated cardiomyopathy has become as significant as respiratory decline and equal to respiratory events as a major cause of death. Improved cardiac surveillance and care has also made a difference to both longevity and quality of life. In the normal presentation of the condition, assessments are made at diagnosis and every second year thereafter until the age of 10, and every year after the age of 10. Any overt cardiac symptoms should receive immediate attention and any abnormalities in left ventricular function would necessitate increased surveillance (six-monthly) as well as earlier consideration of therapy.[42]

Cardiac assessment

It is known that cardiac disease develops long before symptoms are present. A baseline assessment by a cardiologist is recommended at the time of diagnosis or at least by six years

of age and would include an:

- electrocardiogram
- echocardiogram
- cardiac MRI scans (offered in some centres).

In addition:

- boys on steroid medication need regular evaluation of weight and blood pressure
- full cardiac evaluation is needed before any surgical procedure is considered
- cardiac monitoring is required during surgery
- nutritional status and profile of diet are reviewed to assist weight management.

Pharmacological therapy

- ACE inhibitors (angiotensin-converting enzyme inhibitors). In some centres ACE inhibitors are recommended prophylactically from the age of nine.[39, 40]
- beta-blockers are often added earlier than previously recommended. John Bourke 2010, in Towards a Brighter Future Conference, reports beta-blockers should start two weeks after ACE inhibitors begin).[42]
- cardiac arrhythmias should be investigated, with periodic Holter monitoring.
- diueretics are given when fluid retention is a problem.

Orthopaedic surgery for contractures

The role of orthopaedic surgery, as well as the extent of the surgical procedures, varies widely depending on the current beliefs of the child's orthopaedic surgeon and his team. There are three main approaches:

- a conservative or minimalist approach. This is the approach adopted in Queensland at the time of writing. Intervention is highly individualised and usually limited to the relief of symptoms, and only after consultation with the child, his family and the therapy team e.g. lengthening of tendo-achilles tendons in the ambulatory child who has good quadriceps strength may be considered (but not if quads strength is poor). A non-ambulatory child may request surgery in order to be able to wear shoes and rest his feet flat on the footplates of his wheelchair. If the contractures are severe, intervention will not be recommended and alternative solutions will be offered.
- a rehabilitative approach. Tendon-lengthening operations (hips, knees and ankles) are performed at the end of the late ambulatory phase, with the aim of prolonging walking in calipers or HKFOs (hip-knee-ankle-foot orthoses.) It is claimed that this procedure will enable walking to continue for up to three years beyond the average. Some centres wait until the child has stopped walking before embarking on this multilevel surgery. This approach is practised mainly in the United Kingdom, where prolongation of walking in calipers has distinct advantages for those residing in houses which are not wheelchair accessible. The extension in walking time afforded by steroid medication is, however, changing the number of boys requiring surgery and calipers. Serial casting of ankles has been effective in restoring range prior to rehabilitation in calipers and is preferable to tendon-lengthening operations wherever possible.[13]
- extensive prophylactic tendon releases. Tendons around the hip, knee and ankle are released or lengthened at around five years of age, and preferably before the decline in

muscle strength has begun. This protocol is widely practiced in France and Germany.[12, 14] The proponents believe that developing contractures contributes to a further imbalance in the muscles around the joints. The surgery is claimed to extend brace-free walking time to between one and three years. These procedures are also said to negate the necessity for daily passive stretches. Release of contractures does not, however, address the primary problems, which are progressive muscle weakness and the development of compensatory postures and gait, which result because of this pattern of weakness. Before embarking on surgery, consideration should be given in each case to the fact that:

– contractures of hips and knees rarely occur while the boys are still walking
– contractures are a useful and sometimes necessary adaptive response to the unequal loss of power in opposing muscle groups
– function can be lost when the biomechanics are altered e.g. tight iliotibial bands are a result of the wide-based stance adopted to help stabilise the joint and widen the base of support. Lengthening tendo-achilles tendons can alter a precariously balanced stance and result in loss of walking.

Since the 1960s, extensive orthopaedic measures and aggressive rehabilitation have been directed toward prolonging the ability to walk in the belief that assisted walking:

- delays the onset of scoliosis
- maintains independent mobility
- delays the decline in respiratory function
- assists families with manual handling and transfers.

Many changes have occurred in the last 50 years, these have resulted in a re-evaluation of previously recommended protocols. These include:

- an increasing understanding of the pathophysiology and the role dystrophin plays in maintaining muscle membrane structure
- the acceptance of steroid medication as a first line of treatment worldwide has changed the natural course of DMD
- the role of exercise in DMD has been questioned and re-evaluated
- most boys with DMD have been integrated into mainstream schools, and walking in long leg calipers is often considered inappropriate and not tolerated well by boys in the normal school setting
- when scoliosis occurs (albeit less frequently since steroids) spines are surgically 'rodded' or stabilised much earlier in order to intervene at the optimum time when respiratory and cardiac function is good
- provision of power chairs by the government (in Queensland, known as the Mobility Aids Subsidy Scheme) has ensured the restoration of lost mobility. Most boys welcome the provision of their chairs and are reluctant to continue braced walking. Calipers are heavy, progress is laboured and the braces can in no way substitute for weakened proximal muscles. Power chairs enable the child to keep up with his peers and to access all parts of the school ground and his neighborhood
- over-exertion could also be contraindicated if cardiomyopathy is present
- emphasis is now placed on giving the boy more control in the individual choices that affect his life.

Management of the spine

Boys with DMD who are not treated with glucocorticoids are thought to have a 90 per cent chance of developing a scoliosis within two years of loss of ambulation. Glucocorticoids are thought to maintain the strength in the para-spinal and abdominal muscles sufficiently well to maintain postural alignment in all planes, especially through the adolescent years of growth.

Osteoporosis in the spine presents a risk of vertebral fractures and the risk of these occurring increases with glucocorticoid treatment.

Surgical procedures

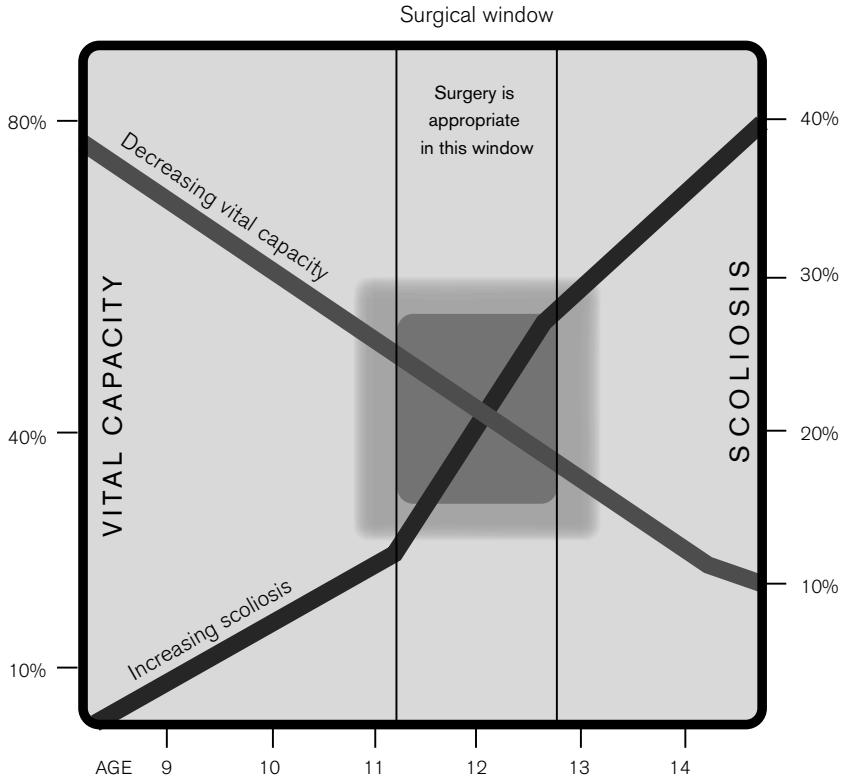
Management by spinal instrumentation and arthrodesis has become a very important part of the management of DMD for those who do develop a curvature.

A posterior procedure is always used, as anterior instrumentation is not appropriate for patients with decreased pulmonary function.[15]

Early intervention for a myopathic scoliosis is recommended as, once established, progression is inevitable and the rate of collapse is often quite rapid. As respiratory function is also declining, careful monitoring is necessary as it is advisable to perform instrumentation when the curve is minimal (20 to 30 degrees), still flexible and there is little or no pelvic obliquity. This can be as early as 12 to 13 years of age if there is evidence of rapid progression.

Many surgeons and anaesthetists prefer not to operate when the forced vital capacity is less than 30 per cent of that predicted for height and age, however successful stabilisations are often performed when respiratory function is considerably less, owing to the increasing expertise of anaesthetists.

Dr John Bach[24] reports that if the respiratory plateau is low (less than 1.7 litres) then the rate



of respiratory decline is greater than normal, scoliosis development is more rapid and there is an increased pulmonary risk if surgery is delayed. If, on the other hand, the respiratory plateau is high (more than 2.5 litres) then the rate of decline is slower and surgeons can afford to wait before determining the need for surgery.[17]

If surgery is delayed until the curve reaches 40 degrees (the standard indication for surgical intervention in idiopathic scoliosis), there is not only the increased pulmonary risk but also the curve may not be flexible and pelvic obliquity may already be present.

Pre-operative assessment

The pre-operative assessment consists of:

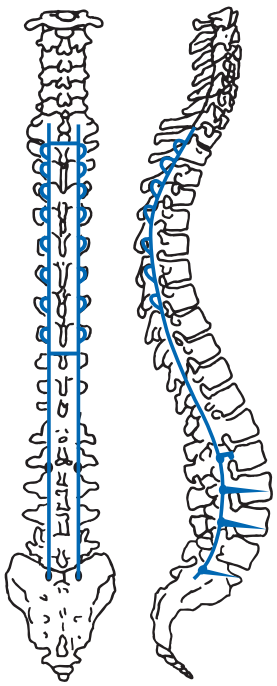
- lung function tests
- blood gas analysis
- cardiac assessment (including echocardiography ECG and Holter monitor)
- assessment by anaesthetist.

Spinal instrumentation

Instrumentation has improved considerably since Harrington designed his rods in 1962. Luque rods with their sublaminar wires allowed shaping to the curvature of the spine and, when combined with improved techniques to fuse the vertebrae, have provided a better alternative for the DMD population, especially as post-operative bracing is seldom required. Many subsequent modifications to Luque's instrumentation have produced further refinements. Orthopaedic surgeons make their choice of instrumentation from the many types now available. Systems commonly used in Queensland include:

- Luque
- TSRH (The Scottish Rite Hospital)
- Isola.

Autograft bone grafts are used more often than donor grafts.



Spinal Instrumentation (rodding)

The rods usually extend from the second, third, fourth or fifth thoracic vertebrae to either the fifth lumbar vertebra or to the sacrum or pelvis. Whether to fuse the sacrum or pelvis or not is still controversial.

Fusion into the pelvis is claimed to provide more stable seating. A neuromuscular scoliosis almost invariably includes pelvic obliquity, which is likely to increase over time if fusion stops at L5 level.[16]

However others, including Dr Walter Greene[16] believe that fusion to the sacrum can cause more problems as perfect symmetry of the pelvis must be achieved at the time of surgery. Because it is impossible for the boys to make even small postural adjustments, comfortable seating may be difficult to achieve. Surgery to include the pelvis takes considerably longer with an increase in blood loss being stated as a reason for care and consideration being given.

Positive effects of spinal fusion

Spinal fusion:

- prevents further development of curves in all planes
- permits independent hands-free sitting
- enhances sitting comfort and tolerance
- minimises or eliminates pelvic obliquity
- improves body image
- limits the added mechanical restriction imposed on respiration by the collapsing spine
- permits the use of a pneumobelt (a non-invasive respiratory device worn around the waist that provides alternating pressure to assist expiration)
- provides easy access for other respiratory assistive devices such as the Cough Assist, accompanied by manual assistive coughing techniques.

Some research has claimed there is a plateau in respiratory decline after stabilisation for up to three years[17,18], while other research[17,19] states that respiratory decline continues at the expected rate.

Surgery cannot promise to improve or even stabilise respiration, as it can have no effect on the inevitable decline in power of the muscles of respiration.

Non-surgical techniques for controlling scoliosis

Spinal orthoses such as a thoraco-lumbar-sacral orthosis have not been successful in controlling spinal collapse. They are not well tolerated by boys with DMD as they are unable to move away from painful pressure points and are extremely uncomfortable in hot climates. The brace imposes further restriction on respiration, but of most concern is the possibility that wearing an orthosis may delay surgery so that the optimum timing is missed. An orthosis may be worn for a short time after some spinal stabilisation procedures, especially if surgery is performed late when there is a greater risk of complications (e.g. rods pulling away and non-union). In some case, when cardiac complications preclude spinal surgery, an orthosis can be prescribed to offer some support and delay decline.

Modifications to wheelchair seating are an essential part of therapy management and should be provided on every chair. While postural modifications do nothing to lessen the inevitable decline in paraspinal muscle strength, they are able to support the spine in a good position until spinal 'rodding' is performed and in many cases, if seating measures are followed, the development of spinal deviations can be averted entirely. (See Physiotherapy Management for

assessment and prescription of wheelchairs.) Custom-moulded seating or seating modifications on their own are not effective in controlling a rapidly progressing scoliosis and are not viable alternatives to surgical correction.

Future research could examine posture-modified seating in conjunction with electronic tilt-in-space mechanisms as even a small degree of tilt in the seat module minimises the effect of the weight of gravity on the spine, as the line of gravity passes in front of the spine instead of directly through the erect spine. It is essential that the boy is able to control the degree of tilt independently and that thoracic supports and headrests are provided.

Respiratory monitoring and management

Refer to consensus statement: Respiratory Care of the Patient with Duchenne Muscular Dystrophy; Am J Crit Care Med. 2004;170:456-465.

Early stage

In the early stages, normal respiration is thought to occur. An assessment of respiratory function is recommended at the time of diagnosis or early in the course of the condition to obtain baseline readings. Assessments are made at least annually thereafter to monitor forced vital capacity and forced expiratory volume at one second. Routine immunisations as well as annual influenza vaccine and pneumoccal vaccine every five years are recommended. Airways clearance techniques and respiratory therapy are introduced with a physiotherapist or respiratory therapist in preparation for later stages.

Prophylactic care and recommendations for keeping airways clear and free of infections include:

- respiratory therapy (deep and 'sigh' breaths—incentive spirometry techniques, not training)
- swimming and aquatic physiotherapy
- pneumovax vaccine (every five years) to prevent pneumoccocal pneumonia
- influenza vaccine (in autumn every year)
- treat infection aggressively (physiotherapy and antibiotics)
- treat asthma (mucolytics, decongestants, bronchodilators)
- good diet
- drink plenty of fluids (to keep mucous secretions thin)
- avoid irritants and allergens (smoke, dust mites, pollens, moulds, animal dander)
- avoid cough suppressants
- avoid obesity, which limits respiratory excursion.

Late ambulatory years

Ventilation may be adequate but cough may become ineffective. As forced vital capacity drops, regular assessments are needed:

- twice-yearly spirometry will monitor changes and alert the clinician to the time when declining respiratory function requires intervention.
- respiratory infections need to be treated aggressively with antibiotics and therapy.
- incentive spirometry or other methods of achieving maximum insufflation exercises are introduced. These exercises are not considered respiratory muscle training but simply a method to ensure that air does reach the bases of the lung at least once or twice a day and to

help retain chest-wall compliance. Lung and chest-wall compliance can be improved by ensuring these deep insufflations, especially as boys with Duchenne are rarely able to exercise enough to become puffed or require deep breathing. Failure to take deep breaths over a long period does lead to micro-atelectasis and loss of lung and chest-wall elasticity.[24]

- Strategies to improve maximum airway capacity and clearance are taught early in the condition to ensure that they are available when difficulty coughing up retained secretion is experienced. Peak cough flow recordings of under 270L minute or when maximum expiratory pressure is less than 60cm H2O indicate that extra assistance will be needed to clear secretions.34

Techniques to increase inspiratory capacity include:[26, 34, 51, 52, 53, 54]

- glosso-pharangeal breathing (gulping air boluses into the lungs from mouth)
- breath stacking (taking a series of breaths without exhaling)
- use of self-inflating bag and mask such as Ambu or resuscitator bag
- intermittent positive pressure device.

After increasing lung volume by any of these methods, exhalation is improved by:

- manual assisted coughing (operator forcefully pushes on abdomen or chest wall in time with the patient's own cough effort)
- cough-assist machines, which are now widely used in many parts of the world and in Australia. The Duchenne Foundation has been instrumental in not only raising the profile of this device but in supplying machines to many centres caring for boys with DMD around Australia.

Non-ambulatory years

Ventilation may begin to decline in the night. When the forced vital capacity falls below 60 per cent of that predicted for height and age, sleep-disordered breathing can be anticipated. Polysomnography at a sleep clinic is recommended. If it is unavailable, overnight pulse oximetry and transcutaneous tracking of retention of CO2 can be taken at the bedside. [20] At 40 per cent predicted, pulmonary function tests are done at least four times a year, and twice-yearly referral is made to a sleep studies unit for polysomnography. Non-invasive nocturnal ventilation via nasal or facemask may be offered to address symptoms of hypoventilation and disordered sleep. Bi-level positive airways pressure is offered. This intervention has been successful in treating sleep-disordered breathing and nocturnal hypoventilation and has improved both quality of life and survival. [30]

Clinical symptoms of chronic nocturnal hypoventilation include:

- fatigue
- sleep disturbances
- nightmares or night terrors
- morning headaches
- poor concentration
- confusion, disorientation, irritability, depression or anxiety
- poor appetite and weight loss
- weakened or softened voice
- unproductive cough.

Shortness of breath was thought to occur rarely in boys with DMD because of their sedentary lifestyle. However, with longer life expectancy, many men who are on non-invasive nocturnal ventilation are experiencing shortness of breath during the day as evidenced by a reducing

ability to speak without multiple breath intakes. This leads to the fourth stage in respiratory function of boys and young men with DMD.

Inadequate daytime and night-time ventilation

Some young men progress to a state of constant hypoventilation, necessitating 24-hour support.[51, 52, 53, 54] Mechanical volume ventilation is the preferred mode of non-invasive ventilation and can be delivered via a mouthpiece, a nosepiece, or, occasionally, by use of facemask. The mouthpiece interface is mounted on a gooseneck device so that it is easily accessible. Non-invasive daytime ventilation is recommended over invasive ventilation via tracheotomies.[23] However, tracheotomies are offered as a choice in many countries and, though not often recommended, some young men prefer them (Rahbeck 2010, Towards a Brighter Future Conference, Sydney). The advantages of volume ventilators over pressure ventilators are that they can also be used successfully for breath stacking to assist mobilisation of retained mucous secretions.[24]

Oxygen should not be administered to patients with Duchenne muscular dystrophy as it depresses the respiratory drive, resulting in a build-up in blood carbon dioxide.[21]

Anaesthesia

Major reference: Birnkrant DJ, Panitch HB, Benditt JO, et al. American College of Chest Physicians consensus statement on the respiratory and related management of patients with Duchenne muscular dystrophy undergoing anesthesia or sedation. Chest. 2007;132 (6):1977 –1986

A wide variety of anaesthesic agents have been used successfully in patients with DMD. However, there is ample evidence to demonstrate that the use of succinylcholine and vapour anaesthetics is contraindicated (Morris 1997). Complications have been reported including:

- rigidity
- rhabdomyolysis
- myoglobinuria
- malignant hypothermia
- arrhythmias
- cardiac arrest.

It is now recommended that total intravenous anaesthetics (TIVA) using newer, shorter-acting anaesthetic agents be used exclusively due to the risk of malignant hyperthermia-like reactions and rhabdomyolisis, which have occurred when patients with DMD have been exposed to vapour anaesthetics. Depolarising muscle relaxants are contraindicated.[49, 50]

Anaesthesia-related complications have been reported in boys who have not previously been diagnosed with DMD. A full history of motor milestones, including the ability to run properly, should be obtained and evaluated before excluding a DMD diagnosis in any preschool boy who may require anaesthesia.[49] (See Appendix 7 for a discussion of the risks of anaesthesia in children with muscular dystrophy.)

Endocrine issues

The increasing recognition and acknowledgement of the endocrine features of DMD in more recent times have contributed to a considerable expansion of both laboratory and clinical research in this important area. Endocrine aspects of DMD result from a combination of inherent characteristics of the condition and iatrogenic factors—in particular, glucocorticoids. Growth, pubertal progression, nutrition and weight gain, bone health and glucocorticoid-related adrenal suppression are all important issues in DMD and should be addressed early and on a regular basis by a paediatric endocrinologist within a multidisciplinary team.

Growth

Normal childhood growth is influenced by genetic potential, a predictable pattern of hormonal secretion, nutritional status and various other factors that ensure normal homeostasis. Boys with DMD have short stature. Studies of normal growth patterns in glucocorticoid-naive children with DMD show:

- normal birth weight and length
- slow growth with a fall in percentiles in the first years of life followed by normal growth, with a velocity appropriate for age
- average mid-parental height
- concordant skeletal maturity as estimated by bone age
- a final height which is typically 1 SDS below the population.[1, 2]

This attenuated growth may reflect the importance of mechanical stimulation by muscle to not only bone-mineral density but longitudinal bone growth.[3] Dystrophin isoforms are not present in the pituitary gland and hypothalamic-pituitary dysfunction is not an inherent characteristic of DMD. Glucocorticoid therapy contributes to a substantial reduction in the already attenuated growth potential of children with DMD. Glucocorticoids have been shown in in-vivo and in-vitro studies to cause growth failure by various mechanisms in a number of organs including the hypothalamus, pituitary gland, growth plate and liver.[4]

Regular auxological measurements and the exclusion of treatable causes of growth failure are important elements in managing children with DMD. The assessment of growth hormone secretion using conventional dynamic stimulation studies, with all of their intrinsic limitations, is further complicated by the pervasive effects of glucocorticoids on the hypothalamic-pituitary axis. This makes interpretation of stimulation tests more difficult. Growth hormone treatment, used in some international centres, improves growth[4, 5] and body composition[5], but there is currently no published evidence to delineate its effects, positive or negative, on muscle function. Side effects, such as the worsening of scoliosis, are important considerations of growth hormone therapy.

Puberty

Although there are no published studies specifically addressing the pattern of pubertal progression in children with DMD, pubertal delay is a consistent observation seen by clinicians in most centres. The precise mechanism of hypothalamic-pituitary-gonadal dysfunction is incompletely understood but glucocorticoid influences are thought to be an important contribution. Primary gonadal failure is not a characteristic feature of DMD. Delayed puberty in boys with DMD, like in other children, can have significant psychological consequences and

adversely affect the development of relationships with their peers. Regular assessment of pubertal progression should be conducted within a multidisciplinary team approach. Pubertal induction with testosterone, at a time appropriate for the child and parent, should be considered.

Nutrition, weight gain and insulin resistance

Nutritional assessment and dietetic support are essential components of the multidisciplinary team's approach to the boy with DMD. This should be initiated early to address potential weight gain due to the significant alterations in appetite, metabolism and body composition caused by the early initiation of glucocorticoids.[6] The pattern of weight gain is relatively unremarkable before the onset of significant muscle weakness and initiation of glucocorticoids.

Weight gain in mid to late childhood from a combination of reduced physical activity and glucocorticoids is characteristic and management can be very challenging if not anticipated and addressed early. Malnutrition is a relatively common feature of the later stages of the condition due to feeding difficulties secondary to progressive muscle weakness and gastrointestinal dysfunction. Assessment of micronutrient and electrolyte status, such as calcium and vitamin D, is also essential.

Significant weight gain can result in the development and progression of metabolic complications such as hyperlipidaemia, insulin resistance, glucose intolerance and overt diabetes. The rates of these complications have not been comprehensively studied and are therefore relatively unknown. The focus should be on anticipatory guidance on weight management before the development of these complications. In some international centres, metformin, in conjunction with dietetic support, has been used with some promising results5. There have been no formal clinical trials assessing the efficacy of metformin in the treatment of insulin resistance in children with DMD.

Bone health

The risk factors for poor bone health in children with DMD include malnutitrition, hormonal disturbances, glucocorticoids and decreased physical activity. Dietary deficiencies in calcium and vitamin D deficiency/insufficiency secondary to inadequate sunlight exposure is not uncommon in children with DMD[7] and the disturbances in calciotropic hormones are exacerbated by glucocorticoid therapy. Weight-bearing is a fundamental requirement for physiological bone remodelling in the developing skeleton.[8]

Glucocorticoids have a profound effect on the developing skeleton, both directly and indirectly, and lead to glucocorticoid-induced osteoporosis (GIO) and fractures (especially at sites rich in cancellous bone, such as the vertebral bodies).[9] Bone-mineral density, calcium and vitamin D levels have been shown to be lower in children with DMD when compared to age-matched controls. This difference is accentuated in glucocorticoid-treated children.[7]

Long-bone fracture prevalence in children with DMD has been estimated to be around 20-25 per cent[10], while vertebral fractures rates in those treated with glucocorticoids is approximately 32 per cent.[11] The site of fractures is dependent on the degree of independent mobility, the use of orthoses and treatment with glucocorticoids. Fractures—especially vertebral—can be asymptomatic and may contribute to the accelerated loss of ambulation.

Children with DMD should be evaluated for bone health prior to commencement of glucocorticoids and monitored at regular intervals by a paediatric endocrinologist. Calcium and vitamin D status should be optimised. A dietetic assessment is recommended. There is increasing evidence for a correlation between BMD measured via DEXA and future fracture in children with conditions such as DMD.[12] A baseline DEXA should be performed and can be repeated on a yearly basis.

Non-pharmacological interventions to optimise muscle strength and reduce contractures may be useful for promoting bone health. Bisphosphonates are anti-resorptive compounds that have been used extensively in adults with osteoporosis but less commonly in children. A number of studies—predominantly observational— have shown that bisphosphonates are effective in improving BMD and mobility in children with osteogenesis imperfecta, a primary metabolic bone disease. The evidence for reductions in fracture rates is less conclusive.[13] Bisphosphonates have also been used extensively in the management of secondary osteoporosis, including GIO, in adults but less frequently in children. Improvements in spinal and femoral neck BMD have not translated to a significant difference in fracture rates.[14]

Many experts have recommended that the use of bisphosphonates be limited to those children with recurrent extremity fractures, symptomatic vertebral collapse and reduced bone mass.[15] More studies are required to clarify the appropriate indications for bisphosphonate therapy and the optimal agent, the formulation (oral versus intravenous), dose and duration of use in paediatric patients with osteoporosis and increased risks for fractures. Side effects of bisphosphonates, such as hypocalcaemia and flu-like symptoms, are common in children. Osteonecrosis of the jaw has not been reported in the paediatric population.

Bisphosphonates are deposited and remain in the skeleton for years. The long-term effects on the growing skeleton have not been clearly defined. The use of bisphosphonates in children should be conducted under the supervision of a paediatric endocrinologist and progressive assessment of BMD and monitoring for fractures are essential.

Adrenal insufficiency secondary to chronic glucocorticoid therapy

Glucocorticoids have been shown to be effective in prolonging ambulation in children with DMD[16]. Large doses (up to 0.75mg per kg per day, prednisone) are used from a very young age (as young as three years) and can be continued at variable doses until loss of ambulation during adolescence or longer. Another complicating factor is the variable glucocorticoid regimes used in different centres including daily, second daily and pulse therapy (commonly 10 days on/10 days off). The doses of glucocorticoids utilised in DMD are supra-physiological[17] and, therefore, adrenal suppression is a significant risk when glucocorticoids are ceased—during periods of weaning and during non-glucocorticoid days in on-off regimes.

During periods of stress in these situations, adrenal suppression should be assumed and stress doses of glucocorticoids should be utilised. Chronic glucocorticoids should never be ceased abruptly but weaned in a rational manner. Consideration should be given to formal assessment of the hypothalamic-pituitary-adrenal axis once glucocorticoids have been ceased.[18]

References

1. Angelini C. The role of corticosteroids in muscular dystrophy: a critical appraisal. Muscle Nerve 2007; 36: 424–35.5
2. Manzur A, Kuntzer T, et al. Glucocorticoid corticosteroids for Duchenne muscular dystrophy (Cochrane Review). The Cochrane Library 2005;(2),,7
3. Bushby K, Muntoni F, Urtizberea A, Hughes R, Griggs R. Report on the 124th ENMC International Workshop. Treatment of Duchenne musculardystrophy; defining the gold standards of management in the use of corticosteroids. 2-4 April 2004, Naarden, The Netherlands. Neuromuscul Disord 2004;14(8-9): 526–34.
4. Biggar WD, Harris VA, Eliasoph L, Alman B. Long-term benefits of deflazacort treatment for boys with Duchenne muscular dystrophy in theirsecond decade. Neuromuscul Disord 2006;16: 249–55.
5. Houde S, Filiatrault M, Fournier A, et al. Deflazacort use in Duchenne muscular dystrophy: an 8-year follow-up. Pediatr Neurol 2008;38(3):200–06.
6. Bushby K, Griggs R, MSG/ENMC FOR DMD Trial Study Group. 145th ENMC International Workshop: planning for an International Trial of Steroid Dosage Regimes in DMD (FOR DMD), 22-24th October 2006, Naarden, the Netherlands. Neuromuscul Disord 2007;17(5): 423–28.
7. Biggar WD, Politano L,Harris VA, et al. Deflazacort in Duchenne muscular dystrophy: a comparison of two different protocols. Neuromuscul Disord. 2004;14(8-9): 476–82.
8. Biggar WD, Gingras M, Fehlings DL, Harris VA, Steele CA. Deflazacort treatment of Duchenne muscular dystrophy. J Pediatr 2001;138:45–50.
9. Bonifati MD, Ruzza G, Bonometto P, et al. A multicenter, double-blind, randomized trial of deflazacort versus prednisone in Duchenne muscular dystrophy. Muscle Nerve 2000;23(9):1344–47.
10. Griggs RC, Moxley RT 3rd, Mendell JR, et al. Prednisone in Duchenne dystrophy. A randomized, controlled Clinical Investigation of Duchenne Dystrophy Group. Arch Neurol 1991; 48(4): 383–88.
11. Kerr T, Lin J-P, Gresty M, Morley T, Robb S. Spinal stability is improved by inducing a lumbar lordosis in boys with Duchenne Muscular Dystrophy: a pilot study. Gait & Posture. 2008;28(1):108-12.
12. Rideau Y, Duport G, Delaubier A, Guillou C, Renardel-Irani A, Bach JR. Early treatment to preserve quality of locomotion for children with Duchenne muscular dystrophy. Semin Neurol 1995;15: 9–17.
13. Kinali M, Main M, Mercuri E, Muntoni F. Evolution of abnormal postures in Duchenne 14muscular dystrophy. Ann Indian Academy Neur2007;10:44-54.
14. Forst J, Forst R. Lower limb surgery in Duchenne muscular dystrophy. Neuromuscul Disord 1999; 9: 176–81.
15. Smith AD, Koreska,JP& Mosley CF(1989)"Progression of scoliosis in Duchenne Muscular Dystrophy The journal of joint and bone surgery, 71-A(7):1066-1077.
16. Robinson R.Scoliosis surgery setting the record straight: Quest 4(1) Muscular dystrophy Association publication; 1997.
17. Rideau Y, Glorion B, Delaubier A, Tarle A & Bach J.The Treatment of Scoliosis in Duchenne muscular dystrophy Muscle & Nerve 1984;7;281-286.
18. Galasko CSB, Delaney C & Morris P. Spinal stabilization in Duchenne muscular dystrophy. The journal of bone and joint surgery 1992;74-B;210-214
19. Granata C, Merlinil L, Cervellati S, Ballestrazzi A, Gianni S, Corbascio M, Lari S.Long Term Results of spinal surgery in Duchenne muscular dystrophy' Neuromuscular disorders 1996;6(1):61-68
20. Mellies U, Ragette R, Schwake C, et al. Daytime predictors of sleep disordered breathing in children and adolescents with neuromuscular disorders. Neuromuscul Disord 2003;13:123–128.
21. Bach JR. Pulmonary Rehabilitation Considerations for Duchenne Muscular Dystrophy: The Prolongation of Life by Respiratory Muscle Airs. Critical Reviews in Physical and Rehabilitation Medicine 1992;3(3): 239-269
22. Bach JR. Mechanical Insufflation Exsufflation: Comparison of Peak Expiratory Flows with Manually Assisted and Unassisted Coughing Techniques. Chest 1993;104(5):1553-1562.
23. Bach JR. Update and Perspective on Non-invasive Respiratory Muscle Aids - Part 2: The Expiratory Aids." Chest 1994;105(5): 1538-1543.
24. Bach JR. Management of patients with neuromuscular disease. Philadelphia, Hanley & Belfus; 1994.
25. Bach JR, Alba AS. Management of chronic alveolar hypoventilation by nasal ventilation. Chest 1990;97: 52-57.
26. Bach JR, Bianchi C, et al. Lung inflation by glossopharyngeal breathing and air stacking in Duchenne muscular dystrophy. Am J Phys Med Rehabil 2007;86:295-300.
27. Bach JR, Ishikawa Y, et al. Prevention of Pulmonary Morbidity for Patients with Duchenne Muscular Dystrophy. Chest 1997; 112(4): 1024-1028.
28. Simonds AK. Recent advances in respiratory care for neuromuscular disease. Chest 2006;130:1879–1886.
29. Chatwin M, Ross E,Hart N, Nickol A H, Polkey MI, Simmonds AK Cough. Augmentation with mechanical insufflation/exsufflation in patients with neuromuscular weakness. European Respir J 2003;21:502-508
30. Eagle M, Baudoin SV, Chandler C, Giddings DR, Bullock R, Bushby K. Survival in Duchenne muscular dystrophy; improvements in life expectancy since 1967 and the impact of home nocturnal ventilation. Neuromuscul Disord 2002;12:926–929.
31. Simmonds AK. Nocturnal Ventilation in neuromuscular disease – when and how? Monaldi Arch Chest Dis 2002;57;5-6 273-276
32. Ward S, Chatwin M, Heather S, et al. Randomised controlled trial of non-invasive ventilation (NIV) for nocturnal hypoventilation in neuromuscular and chest wall disease patients with daytime normocapnia. Thorax 2005:60; 1019-1024
33. Toussaint M, Steens M, Soudon P. Lung function accurately predicts hypercapnia in patients with Duchenne muscular dystrophy. Chest 2007;131: 368–375.

34. Toussaint M, Boitano LJ, GathotV, Steens M, Soudan.Limits of effective Cough augmentation in Patients with Neuromuscular disease Respiratory care 2009;4(3)
35. Boitano L, Dobrozi J, Hilsen M, Johnson J, Jordan T, Stryker T, Benditt J. University of Washington Resource Manual For Non- invasive Mechanical Ventilation
36. McNally, E. New approaches in the therapy of cardiomyopathy in muscular dystrophy. Annu. Rev. Med 2007;58:75–88..
37. Nigro G, Comi LI, Politano L, Nigro V. Dilated cardiomyopathy of muscular dystrophy: A multifaceted approach to management. Semin Neurol 1995;15(1):90–92.
38. Saito T, Matsumura T, Miyai I, Nozaki S, Shinno S. Carvedilol effectiveness for left ventricular-insufficient patients with Duchenne muscular dystrophy PEDIATRICS 2005;116(6):1569-1573 (doi:10.1542/peds.2005-2448)
39. Duboc D, Meune C, Pierre B, et al. Perindopril preventive treatment on mortality in Duchenne muscular dystrophy: 10 years' follow-up. Am Heart J 2007;154(3):596–602.
40. Duboc D, Meune C, Lerebours G, Devaux JY, Vaksmann G, Bécane HM. Effect of perindopril on the onset and progression of left ventricular dysfunction in Duchenne muscular dystrophy. J Am Coll Cardiol 2005;45(6): 855–57.
41. Meune C, Duboc D. How should physicians manage patients with Duchenne muscular dystrophy when experts' recommendations are not unanimous? Dev Med Child Neurol 2006; 48(10): 863–64.
42. Bourke JP. Cardiac monitoring and treatment for children and adolescents with neuromuscular disorders. Dev Med Child Neurol 2006; 48(3):dystrophy. Rinsho Shinkeigaku 2001;41(10):691–94.
43. Hendriksen JG, Poysky JT, Schrans DG, Schouten EG, Aldenkamp AP, Vles JS. Psychosocial Adjustment in Males with Duchenne MuscularDystrophy: Psychometric Properties and Clinical Utility of a Parent-report Questionnaire. J Pediatr Psychol 2009;34(1): 69-78.
44. Hinton VJ, De Vivo DC, Nereo NE, Goldstein E, Stern Y. Selective deficits in verbal working memory associated with a known genetic etiology:the neuropsychological profile of Duchenne muscular dystrophy. J Int Neuropsychol Soc 2001;7(1): 45–54.
45. Cyrulnik SE, Fee RJ, De Vivo DC, Goldstein E, Hinton VJ. Delayed developmental language milestones in children with Duchenne's muscular dystrophy. J Pediatr 2007;150(5):474–78.
46. Cotton SM, Voudouris NJ, Greenwood KM. Association between intellectual functioning and age in children and young adults with Duchenne muscular dystrophy: further results from a meta-analysis. Dev Med Child Neurol 2005;47(4):257–65.
47. Hendriksen JG, Vles JS. Are males with Duchenne muscular dystrophy at risk for reading disabilities? Pediatr Neurol 2006;34(4):296–300.
48. Hendriksen JG, Vles JS. Neuropsychiatric disorders in males with Duchenne muscular dystrophy: frequency rate of attention-deficit hyperactivity disorder (ADHD), autism spectrum disorder, and obsessive-compulsive disorder. J Child Neurol 2008;23(5):
49. Morris P. Duchenne muscular dystrophy: a challenge for the anaesthetist. Paediatr Anaesth. 1997;7 (1):1 –4[
50. Birkrant DJ. New challenges in the management of prolonged survivors of pediatric neuromuscular diseases: a pulmonologist's
51. Bach J.Mechanical Insufflation and Exsufflation 'Comparisonof Peak Expiratory flows with manually Assisted and Un assisted coughing Techniques Chest 1993:104(5)
52. Bach JR, Ishikawa Y, Kim H. Prevention of pulmonary morbidity for patients with Duchenne muscular dystrophy. Chest. 1997;112 (4):1024–1028
53. Bach JR. Update and Perspective on non invasive Respiratory muscle aids. Chest 1994;105:1538-1544
54. Boitano LJ. Equipment options for cough Augmentation,Ventilation,and non invasive Interfaces in Neuromuscular Respiratory Management Pediatrics 2009;123;S226-S230

Endocrine references

1. Schoenau E. Journal of Musculoskeletal and Neuronal Interactions 2005;5(3):232-238
2. Allan DB et al. Journal of Clinical Endocrinology and Metabolism 1998;83(8): 2824-2829
3. Unpublished data – Cincinnati Children's Hospital.
4. Davidson Z & Truby H. Journal of Human Nutrition Dietetics 2009;22:383–393
5. Bianchi ML et al. Osteoporosis International 2003;14:761-767
6. Bailey DA. Osteoporosis International 2000;11:S2-S3.
7. Canalis E, et al. Osteoporosis International 2007;18: 1319-1328Mcdonald DGM et al. Developmental Medicine & Child Neurology 2002;44:695-698
8. King WM, et al. Neurology 2007;68:1607-1613
9. Henderson RC et al. Journal of Bone and Mineral Research. 2009 Oct 12. Epub
10. Phillipi CA, et al. Cochrane Database Systematic Reviews 2008;4:CD005088
11. Homik J, et al. Cochrane database of Systematic Reviews 1999;1:CD001347
12. Bachrach LK & Ward LM. J Clin Endocrinol Metab 2009;94:400–409
13. Manzur AY, et al. Cochrane Database Systematic Reviews. 2008;Jan 23(1):CD003725
14. Walter LM, Achermann JC, & Fluck CE. The Adrenal Cortex and its Disorders. In Sperling MA (ed), Pediatric Endocrinology 2008. Philadelphia: Saunders Elsevier.
15. Chrousos GP, et al. Neuroimmodulation 2009;16: 272-283.
16. Biggar, D. The use of Deflazacort and Alendronate in treatment with DMD. Proceedings of Parent Project Conference UCLA, Los Angeles; 1999.
17. Biggar, D. Deflazacord and Osteoporosis Treatment in DMD. Proceedings of Duchenne in the Millennium Conference, Pittsburgh PA; 2000.
18. Biggar, W.D, Bachrach LK, et al. Bone health in Duchenne muscular dystrophy. Neuromuscular Disorders 2005:15(1): 80-85.
19. Biggar, WD, Gingras M, et al. Deflazacort treatment in Duchenne muscular dystrophy." J Pediatr 2001:138: 45-50.

ALLIED HEALTH MANAGEMENT

The International Classification of Functioning, Disability and Health (also known as **ICF)** is a classification of the health components of functioning and disability. Reviewing DMD in this framework can assist even the novice allied health practitioner to understand the condition and how it affects function and also the environmental and personal factors that might hamper the boy and young man's participation in his community.

Body structure	Body function (impairment)	Activity limitation (function)	Participation restriction
Skeletal muscle	↓ Functional muscle mass ↑ Skeletal muscle fibrosis ↓ Strength ↓ Endurance ↑ Fatigue	↓ Mobility (walking, running, wheeling) ↓ Upper extremity tasks (reaching, throwing) ↓ Fine motor tasks (writing, typing, object manipulation) ↓ Self care and ADLs ↓ Communication ↓ Ability to undertake tasks ↓ Learning and applying knowledge ↓ Psychosocial adjustment	↓ Educational opportunities ↓ Employment opportunities ↓ Community integration ↓ Socialisation ↓ Family functioning ↓ Recreation ↓↓ ↓ Quality of life
Body composition	↑ Body fat and obesity ↓ Lean tissue		
Bone and joint	↑ Joint contractures ↑ Spine deformity ↑ Osteoporosis ↑ Fractures ↑ Pain		
Lungs	↑ Restrictive lung disease ↓ Pulmonary function ↓ Cough/pulmonary toilet		
Heart	↑ Cardiomyopathy ↑ Conduction defects ↓ Cardiopulmonary capacity		
GI & Nutrition	↑ Dysphagia ↑ Constipation ↑ Cachexia (late onset)		
CNS	↓ Mental functions ↓ Intellectual capacity		

Chapter 6:

Dietary issues in DMD – monitoring and management

The body-composition changes seen in DMD are unique, and hence nutritional management is complex. Boys with DMD can move between the spectrums of over- to under-nutrition within their shortened lifespan. Delayed growth, short stature, muscle wasting and increased fat mass are characteristics of DMD and impact on nutritional status and energy requirements. The early introduction of steroids has altered the natural history of the disease but can exacerbate weight gain in a population already susceptible to obesity. Prior to commencing steroids, anticipatory guidance for weight management should be provided. Malnutrition is a feature of end-stage disease requiring a multidisciplinary approach such as texture modification and supplemental feeding. Micronutrient requirements are yet to be determined but due to corticosteroid treatment, vitamin D and calcium should be supplemented. Some evidence exists to supplement with creatine monohydrate to improve muscle strength.

There is limited high-quality evidence to guide the nutritional management of boys with DMD. Currently, the majority of evidence is based on expert opinion and clinical expertise.

KEY PRACTISE POINTS FOR NUTRITIONAL MANAGEMENT IN DMD

Nutrition requirements:

- monitor weight to guide energy prescription
- ensure adequate intake of micronutrients as per dietary reference values
- supplement vitamin D (1000 IU daily) and calcium (750mg daily), especially in those receiving steroid therapy
- monitor serum 25-hydroxyvitamin D.

Nutrition assessment and monitoring:

- short stature is a characteristic of boys with DMD
- measure height and weight six-monthly and plot on standard growth charts
- upper-arm length, tibial length or knee height can be measured in the advanced stage of disease
- body composition is characterised by a decreased lean body mass and increased intramuscular fat mass
- BMI as a screen for obesity is not accurate in boys with DMD
- various tools can be used to measure body composition with DXA and MRI being accurate, appropriate and non-invasive measurement instruments.

Management:

- prevent excess weight gain by providing anticipatory guidance around energy balance prior to commencement of steroid therapy
- advise reduction in energy intake with caution. Follow-up with regular measurement of fat-free mass to monitor progress where available
- dietary texture modifications may be required to accommodate eating difficulties
- support patients and families in decisions regarding enteral feeding and/or gastrostomy tube placement
- a multidisciplinary team should be involved in the management of feeding difficulties.

Nutriceuticals:

- there is a limited evidence base to support the use of nutriceuticals in boys with DMD
- short and medium-term creatine monohydrate treatment improves muscle strength in people with NMD. (Cochrane)

Chapter 7:

Physiotherapy management

The primary aims of physiotherapy intervention are to:

- maintain functional ability as long as possible
- minimise development of contractures and deformity
- anticipate and minimise other secondary complications of a physical nature, including respiratory decline and development of spinal deviations
- prescribe and provide equipment and aids
- assist the boys to enjoy as fulfilling a lifestyle as possible by giving advice on activities that can be enjoyed throughout life.

Other physiotherapy involvement includes:

- providing information and education on the condition for the boy, his family and the staff at his school
- advocating on behalf of the child, parent and school as needed
- planning with other team members a wide range of adapted programs and recreational options which facilitate inclusion at school and the wider community.

Boys should have an opportunity to enjoy as wide an experience of life as possible and are encouraged to develop interests in the early years that can be continued when mobility is lost.

The main stages of physiotherapy management are:

1. Early stage, including the pre-symptomatic stage from the time of diagnosis, if it is made early, and through the years when the child is acquiring new skills and may have few gross motor symptoms
2. Ambulatory stage, from the stage where symptoms are evident until the time when walking begins to become more laboured and difficult
3. Late ambulatory stage, when walking is possible but is slow and laboured. At this stage,

spontaneous falls are becoming more frequent and rising to standing is difficult. Walking ceases on average at 10 years of age without steroid treatment but this transition stage can be prolonged from between two and five years and occasionally longer with medication.

4. Non-ambulatory stages begin with the full-time use of a wheelchair.

The physiotherapist's role

The physiotherapist's role throughout these stages may include:

- active exercise recommendations—monitoring and supervising
- aquatic physiotherapy
- stretches: instruction, monitoring and supervising
- respiratory therapy and assessment—periodic testing of respiratory function, instruction in breathing and coughing techniques, treatment during acute phases and instruction in use of breathing devices
- scoliosis—monitoring and seating interventions
- orthoses—assessment of need, casting and manufacture, or referral to an orthotist
- wheelchair and seating—assessment, prescription and application to the relevant authority for supply of chair, as well as the assessment of need for modifications
- equipment—assessment and provision of other aids (with other team members)
- recreation—advisory role on suitable activities.

ACTIVE EXERCISE

The role of active exercises and, in particular, strengthening regimes for boys with DMD has been, and still is, a controversial topic.[1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13] Many physiotherapist believe that strengthening programs should only prescribed when there is a reasonable chance of improving function or at least slowing the decline in muscle function.

Many therapists now believe that strong strengthening exercises should not be a part of therapy programs for boys with DMD as it may hasten the degeneration in muscle. Gathering evidence-based data in order to recommend exercise levels has been problematic. It would be difficult to perform randomised control studies, as there is a large heterogeneity in the clinical presentation of boys with DMD even in those with the same mutation. Even boys from the same family can present very differently. The clinician's first aim is to do no harm.

Knowledge of the role dystrophin plays in the maintenance of cell membrane integrity, especially during muscle contractions, has reinforced the belief that strong exercise is detrimental. Regeneration of muscle can only continue while there is a supply of satellite (stem) cells to effect the repair. As the supply of satellite cells is being depleted, muscle cells are replaced by fat and connective tissue. It is hypothesised that strenuous exercise can hasten the cycles of degeneration and regeneration. Knowledge gained from experiments with animal models of Duchenne mouse model (mdx) and the golden retriever dog (GRMD) has also increased understanding of the exacerbation of muscle cell degradation caused by exercise and, particularly, by eccentric contractions.

Some active exercise is, however, essential for maintaining health and wellbeing and to prevent disuse atrophy. Physiotherapists are well placed to recommend activities that provide suitable active exercise but will not cause damage or undue fatigue. These activities in the early years can best be provided in a play or recreation-based form. Water-based exercise and non-weight-bearing exercise become increasingly important as the condition progresses.

As eccentric exercise is known to damage muscle fibres more than concentric activity, attempts should be made to minimise it where possible. Eccentric muscle contractions are, however, occurring in all 'anti-gravity' muscles to keep the body upright against the force of gravity, so it should be addressed by minimising long arduous walking, stairs, slopes and squatting activities wherever possible. Some endurance work may be considered in the swimming pool as the muscles are working concentrically and the load on them is small with the elimination of gravity.

Recommendations for exercise:

- recreational activities appropriate for age (as opposed to strengthening regimes)
- beware of overdoing it—no pushing or encouraging the boy with DMD to do more than he is willing to do
- permit the child to self-monitor (allow him to say when he is tired)
- balance activity with restful occupations
- encourage concentric low-load exercise (water-based) instead of eccentric high-load activities
- minimise, where possible, eccentric activity (stairs, slopes, distances)
- provide transport when long distances are involved to save energy (manual wheelchairs, buggies, scooters, segways etc.).

Active exercise

Early physiotherapy intervention may be necessary in some cases to ensure that developmental milestones are reached. A small percentage of boys have global delays and benefit from a developmental skills program from an early-intervention team.

In the early stage, participation in normal developmentally appropriate activities should provide enough active exercise. This is especially so in Australia where most children are involved in many outdoor activities and simply keeping up with peers provides sufficient exercise.

Stretches

Stretching those muscle groups that are known to tighten and eventually contract is a major part of physiotherapy management and is an essential part of any program throughout all stages of the condition. Parents, carers or teacher's aides will need careful and repeated instruction to ensure that the stretches are performed correctly. Regular monitoring will allow modifications to be made to their execution when necessary. Stretches should be introduced early (as soon as the diagnosis is confirmed).

If possible, parents and others involved in this routine are encouraged to make this time a special time for the child, when they can share the events of the day and emotional ties can be strengthened.

Suggestions from parents include:

- make it a set routine e.g. always after a bath
- include the boy in the process (count or sing songs together; ask him to help e.g. 'help me bring these toes up towards your nose' when doing a tendo-achilles (TA) stretch)
- play favourite CDs, DVDs or stories
- massage the muscles to be stretched beforehand
- precede stretches with a warm-up activity such as a bike ride or warm bath.

See the accompanying DVD, *An Overview of Physiotherapy Management*, and Appendix 5 for some examples of stretches for those muscle groups that are likely to tighten.

Important points to remember when performing stretches:

- Position the child to ensure that he is well supported and comfortable
- Stabilise the joints that are not being moved
- The intensity of the stretch should be submaximal and should never cause pain, however the stretch should be felt. Applying a little extra pressure at the end of the range where some resistance is felt is sufficient. The 'discomfort' felt could be likened to that experienced when performing a good sports stretch. If the child resists strongly, another method must be employed
- The duration that the stretch should be maintained remains a debatable point. The longer the stretch can be held, the better. A prolonged stretch permits the muscle spindle to habituate to the new length and allows the lengthening reaction to occur. However, most parents find it difficult to hold a sustained manual stretch for more than 30 seconds. Three stretches of 30 seconds duration should suffice
- An individual program should be made for each child, this will vary depending on the type of stretch, the circumstances of the child and the person who is performing the stretches
- Overstretching is to be avoided as the muscle responds by contracting further. The stretch is managed better if it begins gently and increases slowly to the maximum intensity (without pain)
- If the boy actively resists, a lengthening contraction occurs which may cause further damage to the muscle fibres
- The person doing the stretches should be taught proper use of body mechanics to make the process easier for them.

Active self-stretches, both static and dynamic, can be most effective for the ambulatory child. Some suggestions can be found in the accompanying DVD, An Overview of Physiotherapy Management, and in Appendix 5.

Respiratory therapy

Refer to: Respiratory Care of the Patient with Duchenne Muscular Dystrophy. Am J Crit Care Med 2004;170:456-465.

Respiratory problems arise as weakening of the muscles of respiration reduces the ability to inhale air and to exhale air forcefully. Coughing to expel mucus from the lungs becomes difficult as muscle power declines, and retained mucus not only reduces the gas exchange in the alveolar sacs but also provides a fertile medium for infection. Arterial oxygen saturation (SaO_2) is a good guide to adequate ventilation, as well as an awake end-tidal carbon dioxide ($ETCO_2$) of >50.

Spirometry assessment

At diagnosis, a baseline recording of respiratory function is taken (although many boys under the age of seven cannot coordinate well enough to perform the usual spirometry tests). Annual recordings are made while the readings are within the normal range (75 per cent of that predicted for age and height). It is recommended that sitting forced-vital-capacity assessment is made at least in six-monthly intervals as volume declines.

Boys taking steroid medication often record average to above average results throughout the ambulatory years.

In the early stage, the child is instructed in deep breathing techniques to ensure adequate ventilation of the lungs and that he has the ability to cough effectively. Techniques used include:

- 'sigh' breaths and open glottis 'huffing'—effective cough techniques
- respiratory toys and bubble blowing, bubble 'pep'
- respiratory work in swimming sessions to encourage deep breathing.

In the late ambulatory stage, boys are naturally less active and less likely to achieve full expansion of lungs. Many boys on steroid medication still register excellent results.

Spirometry assessment every six months is recommended and instruction in techniques to improve chest-wall compliance, prevent atelectasis and assist cough augmentation is introduced when insufficiency becomes apparent. Teaching these techniques before they become necessary is recommended. Indications that airway-clearance measures need to be introduced are a peak cough flow of <270L/min and/or a maximum expiratory pressure of less than 60cm H_2O.

Techniques in the late ambulatory stage may include:

- traditional physiotherapy techniques such as postural drainage and manually assisted coughing
- some (but not all boys) with DMD learn to perform glossopharyngeal or 'frog' breathing. It involves using the muscles of the throat to gulp boluses of air into the lungs and by closing the glottis after each gulp, recruiting extra volumes of air [27]
- breath stacking is an easier process to master: A series of deep breaths is taken, closing the glottis after each breath without exhaling. In this way, the maximum insufflation capacity is increased. In the early stages this exercise is performed by the young man unassisted[27]
- hyperinflation therapy using an Ambu or resuscitator bag is introduced and practised toward the end of the late ambulatory stage to ensure good lung inflation once or twice a day. All methods of volume recruitment can then be followed by manually assisted coughing techniques to assist mucus clearance[21]
- instruction in the use of the cough-assist machine is begun. For the novice therapist a guide can be found at http://www.respironics.com/UserGuides/UserGuideCoughAssist.pdf on the Respironics website.

Physiotherapy assessment in the non-ambulatory stages:

- spirometry is performed at least every six months, including sitting forced vital capacity when hypoventilation is suspected (when FVC is <50 per cent predicted for height, weight and age)
- peak cough flow is assessed regularly and especially when respiratory tract infections occur (PCF <270 L/min indicates expiratory muscle weakness and risk of respiratory failure during respiratory tract infections)

- maximum inspiratory and expiratory pressures. To achieve adequate airways clearance, the boy with DMD needs to be able to have sufficient inspiratory muscle strength to generate a maximal inspiratory capacity (MIC) of more than one litre or 60-80 per cent of the total lung capacity[30] and strong enough expiratory muscles to enable a peak cough expiratory flow to expel retained mucous secretions from the airways (>270L/min is considered adequate)
- yearly or twice-yearly polysomnography (overnight assessment at sleep studies unit) is recommended and available in many larger cities. If not available, overnight pulse oximetry, plus transcutaneous end-tidal capnography (>50) can identify nocturnal hypoventilation
- end-tidal capnography is recommended at any time FVC is <50 per cent and boy has a respiratory infection.

Techniques used to assist airways clearance techniques include:

- traditional physiotherapy techniques as tolerated and in absence of scoliosis or cardiac insufficiency
- manual hyperinflation therapy (resuscitator or Ambu bag)
- cough augmentation combining manual hyperinflation and manually assisted cough manoeuvres (patient coughs in conjunction with therapist assisting with abdominal thrust)
- cough-assist machine.

> "Effective airway clearance is critical for patients with DMD to prevent atelectasis and pneumonia. Ineffective airway clearance can hasten the onset of respiratory failure and death, whereas early intervention to improve airway clearance can prevent hospitalisation and reduce the incidence of pneumonia."

American Thoracic Society, Consensus Statement for DMD

Scoliosis monitoring

Early detection of the onset of scoliosis, and careful monitoring thereafter, is a very important part of management. Ninety per cent of the boys not on steroid medication will develop a scoliosis within two years of going into the wheelchair and approximately 60 per cent of these will progress rapidly (a collapsing neuromuscular scoliosis). Careful monitoring of sitting position is essential and photographic records can assist objective assessment between X-rays. The rate of progression can be between two and four degrees per month.

The physiotherapist may be the only professional able to make regular appraisals and recommend an early referral to an orthopaedic surgeon. The surgeon will determine the need and the optimum time for surgery in conjunction with the respiratory physician, cardiologist and anaesthetist. The parents and the child are involved in the decision-making process. Over many years, many boys have agreed to have surgical 'rodding' but as the use of corticosteroid medication has become more accepted and widespread, the need for surgery has been declining.

Aquatic physiotherapy

Hydrotherapy is an integral part of physiotherapy management throughout life because it is the safest form of exercise.

The primary aim of this form of therapy for boys with DMD is to teach them to swim independently as soon as possible.

Water-based therapy also aims to:

- promote independence and confidence in the water
- provide a venue for good active exercise which is low load and where muscles work concentrically
- ensure water safety throughout the stages of DMD
- foster integrated activity with family and friends at all stages of the condition
- permit active movement when it is no longer possible on land
- provide excellent respiratory work
- above all, provide freedom of movement and enjoyment.

Training in swimming techniques and stroke correction may not be appropriate. Adaptations and modifications to strokes are often needed. Many boys adopt a hybrid stroke using breaststroke arms and a flutter kick, often assisted by use of flippers or fins. Strokes requiring heavy use of shoulder girdle muscles are to be avoided.

Orthoses

Polypropylene ankle foot orthoses (AFOs) night splints[15,16] are provided prophylactically from a very early age. They are set at the maximum comfortable range of dorsiflexion. Custom-made splints with a padded lining, at least over the bony prominences, are more likely to be accepted. There are many different styles of AFO and individual boys may accept some more readily than others. As well as the usual lined polypropylene splints, other varieties include:

- AFOs that are covered in a soft, padded material to reduce the 'clunking' noise made when they make contact as the child turns at night
- articulated splints that permit some momentary stretching into plantar flexion but which recoil to the preset desired dorsiflexed position. These 'ultra-flex' AFOs can also be used for some hours during the day if not accepted as night splints and are therefore may be a useful alternative for those boys who resist wearing night splints
- AFOs with non-slip soles to permit short walks to the toilet if needed. It is recommended that older boys whose walking is compromised use a non-spill urinal bottle at night instead of trying to walk in AFOs
- SMOs (or supra-malleolar orthoses) are sometimes worn to prevent pronation
- An orthotic with ramped-up fore foot is sometimes successful in giving continuous stretch to the calf as the child walks. It also addresses pronation by supporting the subtalar joint in a neutral position

Once ambulation has ceased, it is recommended that boys wear AFOs during the day to prevent the relentless pull into an equino-varus position, which occurs almost inevitably when the pull of relatively strong tibialis posterior muscle is no longer controlled by weight-bearing.

Long-leg braces are offered in many parts of the world when independent ambulation is no longer possible. Standing in supportive devices or standing wheel chairs is an alternative.

Other roles

- Wheelchair assessment and modification is discussed in 'Essential Requirements for Seating' (see page 66)

- Equipment—Standing frames and other useful equipment is discussed in 'Occupational Therapy Management' (page 77)
- Recreation—an overview of suggested recreation can be found in 'Recreation and Leisure' (page 114).

Early years

In the years from diagnosis until the time the boy reaches the plateau in the development of his gross motor skills, it is important for him to be exposed to a wide range of enjoyable activities that will enhance his physical, social and emotional development.

Before he reaches this plateau in strength and skill acquisition, he will enjoy acquiring gross motor skills, such as bike riding, swinging and ball handling, and being involved in sensory motor programs at his preschool. All of these activities will provide good opportunities for learning as well as assist the development of balance and coordination and aid sensory integration. Adaptations to activities will need to be made as he loses strength or tires. Care must be taken never to encourage a boy with Duchenne to do more than he can comfortably perform.

It is at the end of this time, when a plateau has been reached in the acquisition of motor abilities, that steroid medication is most likely to be introduced.

Physiotherapy intervention in the early stages and throughout the mid-ambulatory stage may be summarised under the following:

- provide early intervention to help the child achieve developmental milestones, as needed
- encourage normal developmentally appropriate activities that do not cause undue fatigue. Advise on appropriate school gross motor activities
- provide a program of stretches for those muscle groups that are tightening (tendo-achilles, hamstrings and iliotibial band). These may include active self-stretches, passive self-stretches and manual stretches
- provide orthoses to be worn at night (AFOs) and daytime in shoe orthotics as needed
- conduct baseline assessment of respiratory function if possible, as young boys are often not able to perform the test
- provide instruction in deep breathing, 'sigh' breaths, open glottis huffing and use of respiratory toys to encourage deep inspirations. Give instruction in effective coughing
- provide aquatic physiotherapy, including 'learn to swim' classes and respiratory work in the pool
- supply mobility equipment as needed for long-distance travel e.g. a buggy or stroller if he is a preschool child, a small manual wheelchair (more age appropriate for an older child) or motorised scooters, which are becoming very popular and have the advantage of permitting independent mobility
- supply other equipment, such as a prone wedge for 'tummy time' to stretch hip flexors
- support and advocate for the child, family and school.

Late ambulatory stage

This stage is often the most trying for the child, who is facing the reality of his progressive loss of function. His parents also struggle to hold on to their son's physical abilities and attempt to delay or deny the inevitable decline. The boy may already be using a power scooter, however the timely provision of the power chair (for part-time use) can smooth the progression to the next stage. Boys are usually eager to accept the wheelchair, while parents often grieve about this obvious sign of deterioration in their son's condition.

Physiotherapy programs would include the following interventions:

1. *Stretches:*
   - provide regular daily passive stretches for tendo-achilles, hamstrings, hip flexor and iliotibial band. Self-stretches may still also be performed on the standing board (tendo-achilles), Nada chair (hamstrings) or prone lying on a wedge for the hip flexors
   - perform regular assessment of range in the upper limb muscle groups e.g. elbow flexors, forearm pronators, and the wrist and long finger flexors. Stretches are provided as needed. (See the DVD, An Overview of Physiotherapy Management, and Appendix 5.)
2. *Orthoses:*
   - encourage the use of night splints (AFOs) to maintain the range of dorsiflexion. Always recommend conventional polypropylene splint first and, if unsuccessful, Ultraflex hinged orthotics worn for several hours during the day may be accepted. These splints are expensive but do have the ability to change the degree of dorsiflexion as needed and provide a prolonged stretch. The hinges can be re-used for subsequent orthoses.
3. *Respiratory function:*
   - monitor at least twice a year
   - instruct in using the Ambu bag and cough assist.
4. *Aquatic physiotherapy:*
   - continue and increase aquatic physiotherapy as walking becomes more difficult. Ease of movement in water can be maintained long after activity on land has become too tiring
5. *Equipment provision:*
   - assess the need for equipment, prescribe as needed and apply to the relevant authority for financial assistance
   - provide power chairs toward the end of the late ambulatory stage. The chairs are used on a part-time basis at first, for long-distance travel (school excursions, lengthy shopping or walking trips with family or friends). At school, the wheelchair will ensure the child's safety in the playground, as it is at this time that spontaneous falls become more frequent. Chairs are often kept at school during the week and are used only during the lunch hour or when long distances are to be travelled. Boys enjoy the ability to access all parts of the school grounds with ease, especially as children at this age often spend their breaks on the sports oval. A power chair permits him to be a part of the action and means he can be truly included in as many of his friends' activities as possible.

- for seating assessment and guidelines for providing seating modifications, see 'Essential Requirements for Seating' (page 66) and review the seating suggestions on the DVD An Overview of Physiotherapy Management.
- apply for a manual wheelchair as well as a power chair as is sometimes more appropriate for family and school outings because it is easy to transport in an ordinary car. The Mobility Aids Subsidy Scheme (Queensland) will provide a second-hand chair from stock for all children who have a power chair.
- provide either a standing frame, easy stand, tilt table or supine stander to be used for regular standing time either at school or at home. A standing program provides a prolonged stretch to calves, hamstrings and hip flexors. Some boys really enjoy this activity while others do not tolerate it at all.
- consider provision of a prone wedge for the boy to use while lying in front of the television or playing with toys such as Lego. Sandbags placed on either side of the hips (to anchor him in the correct position) and another on the bottom will provide a stretch for the hip flexors. Weights placed over the ankles will keep legs straight and stretch the hamstrings. (See the DVD)
- consider long sitting in the Nada chair or sitting supported against a wall with weights on knees as another alternative.

6. *Advisory role*

   As this is a difficult time for the child, his family and the school staff, the therapist and other team members may be needed more often to advise on issues regarding:
   - the wheelchair and how often it should be used
   - adaptations to classroom furniture
   - access issues in the school environment
   - inclusion in physical education programs
   - management at school camps and excursions
   - information on the condition and preparation for the next stage.

   It may be very important to communicate to well-meaning staff at schools where there are children with a wide range of physical disabilities that this condition requires very different management to the other physically disabling conditions. Striving to reach maximum physical potential is cruel and can be physically damaging. It is essential that team members raise awareness of the condition and the relentless decline in functional abilities.

   In the later stages of the condition, it is important for therapists to remember that efforts should be directed toward offering choices and involving the boys in all decisions which affect their lives, rather than striving for independence.

SHARK BAIT KIDS PROGRAM

Shark Bait Kids (SBK) is an exciting and innovative program devised and run by a MontroseAccess physiotherapist and combines the skills and knowledge of all team members. It has been enormously successful and well received by both the children and parents. It is offered to boys in the late ambulatory stage of their condition.

SBK is a modified scuba diving program for children with Duchenne muscular dystrophy. The 10-week hydrotherapy program gives young clients with DMD the unique opportunity to experience the wonders of the underwater world freely in a tightly controlled therapeutic environment. The children are taught scuba diving skills in a pool environment, with a camp at the end including a dive in the artificial reef at SeaWorld.

The program is also designed to help with written language skills, assignment planning and computer literacy. The children write blogs about their adventures and share their tales with their friends through the website and Facebook.

As their condition deteriorates, children with DMD are unable to participate in organised sport. Particularly, they miss out on the unique social, cultural and health benefits that Saturday morning sport offers to children, their mums and dads and their families. Shark Bait Kids is run on a Saturday morning to provide a replacement for other organised sport. This provides not just the children with peer support but also a safe, non-threatening environment for the parents to get together and support each other.

Although still in its infancy, this program is expanding every year. Shark Bait Kids will provide this unique experience to other metropolitan and regional families throughout Queensland.

Non-ambulatory stage

The provision of power chairs helps boys regain lost mobility and independence at home, in the community and at school. Modifications are essential to ensure that a good posture is maintained as the paraspinal and trunk muscles become weaker (see pages 69 for recommended modifications).

Physiotherapists may assist the occupational therapist to give advice on home issues such as comfort in bed and techniques to facilitate turning at night.

The physiotherapy program at this stage will include the following interventions:

Stretches:

- continue a manual stretch program as before to minimise the development of contractures in hips, knees and ankles. Without an aggressive standing program there will be loss of range in the hips, knees and ankles, however these contractures can be minimised with a regular stretch program. The aim is to retain sufficient range to ensure comfort in bed, ease of dressing and a good position for the feet on the wheelchair footplates. A separate stretch for tibialis posterior is usually necessary. Stretching the hip flexors may not be possible in prone and an alternative, side-lying position may be more comfortable. Alternatively, the stretch can be performed in the Thomas position (see the DVD *An Overview of Physiotherapy Management* and).
- pay particular attention to the upper limbs, as contractures are now more likely to occur. Maintenance of range in wrist and hands is essential for fine motor function, which is usually retained for some time. Contractures in the upper limbs are likely to occur in the elbow flexors, forearm pronators, wrist flexors and the long finger flexors. Avoidance of contractures, particularly in the long finger flexors, ensures access to a wide range of technology and ease of controlling the power chair. General mobilising of the shoulder is recommended.
- include standing (if tolerated) in the easy stand, standing frame, tilt table or supine stander in the daily regime at school or at home. A long stretching time is preferable and negotiating times in conjunction with favourite TV programs or computer time may improve acceptance of the program. Regular daily standing does retain range but must be balanced with other needs. Standing wheelchairs are available and fulfill this role easily. Unfortunately, the expense of these chairs makes them out of reach for most Australian families and they are not provided by any of the funding bodies. Some boys accept routine standing in the standing wheelchair well while others do not, which makes the decision to make this big financial outlay all the more difficult.
- provide other time out of the wheelchair with prone lying on a wedge (with the addition of weights or sandbags to maintain the position and to provide stretch) to give a sustained stretch to the hip and knee flexors. This is tolerated well by some boys but not in those who have hip contractures or reduced respiratory volumes.
- provide orthotics to help minimise the pull into the equino-varus position of the feet and to help keep the feet flat on the footplates. Boys at this stage are encouraged to wear their splints during the day. Some boys will also wear night splints.

Spinal monitoring:

- monitor the spine to detect the onset of scoliosis. Minor changes in skin fold depth and symmetry can alert the clinician to beginning curves. A reversal of the normal lumbar curves requires monitoring and attention. Careful attention to sitting posture by everyone will detect small changes. Regularly compare physical examination with photographic records. Referral to an orthopaedic surgeon for baseline radiological assessment is usually made. Regular reappraisal by the radiologist and orthopaedic surgeon will then be made.

Respiratory function monitoring:

- monitor respiratory function regularly. Respiratory volumes start to decline as the muscles of respiration weaken. It is particularly important to identify reduced capacity and the likelihood of sleep-disordered breathing. It is also important when a boy is developing scoliosis and spinal instrumentation is being considered, as many orthopaedic surgeons and anaesthetists prefer not to operate when the recorded forced vital capacity (FVC) falls below 30 per cent of the predicted value for age and height.

RESPIRATORY FUNCTION ACTIONS

When recorded FVC falls below normal (75 per cent of that predicted), respiratory function is tested more regularly (three to four times per year). The therapist will train the boy in use of Ambu (resuscitator) bag as well as teaching glossopharangeal breathing, breath stacking and familiarisation with the use of the cough-assist machine.

When the recorded volumes fall below 60 per cent of that predicted for his age and height, there is a possibility that he may have sleep-disordered breathing. A referral to a sleep studies clinic for evaluation will be made. In Queensland, all boys are monitored at the pediatric sleep study unit at the Mater Children's Hospital well before this time. At a later stage, they are assessed at either the Princess Alexandra Hospital or the Prince Charles Respiratory unit or the sleep labs in regional hospitals. Mechanical ventilators, which deliver air at a preset pressures, such as bileval positive airways pressure (BiPAP and VPAP) machines, are offered when nocturnal hypoventilation occurs.

The Cough Assist machine is very effective in expelling retained mucus from the lungs. It imitates a strong cough by pushing a preset volume of air into the lungs and then rapidly withdrawing the same volume, with the accumulated secretions. The Duchenne Foundation has been instrumental in securing many of these machines to centres around Australia as well as for MontroseAccess.

NON-INVASIVE DAYTIME VENTILATION

In early 2010, the first non-invasive daytime ventilators were supplied to three young men. It is foreseen that this non-invasive ventilatory assistance from a volume ventilator via mouth or nose piece will become popular in keeping with worldwide trends. Ventilation via tracheotomy is still favoured in some countries, such as Denmark, however, in other countries noninvasive ventilation is recommended and preferred by the young men.

BRACED AMBULATION

Braced ambulation is not offered through MontroseAccess for several reasons:

- it has not been well tolerated in the past the increased acceptance and use of steroids has extended the time of functional walking
- Australian funding bodies tend to provide only one expensive item for mobility and a power chair would always be the item of choice over long leg calipers
- the Australian lifestyle and style of housing obviates the necessity for retaining the ability to stand and mobilise short distances indoors. However, it is a well-recognised advantage in some countries where boys spend more time indoors in houses not built for wheelchair access
- it was considered inappropriate in the normal school setting, as it does not promote the aim of inclusion in the normal Australian school environment, which involves a greater percentage of outdoor time and the need to travel long distances in the playground.
- in the past, boys were encouraged to maintain ambulation in calipers as it was thought to delay the onset of scoliosis past the most vulnerable time of adolescent growth spurt. The benefits for the spine are no longer as important, as boys taking steroids are often walking independently for much longer. In those who have not taken steroids, the trend toward performing early spinal instrumentation continues. It is now considered more advisable to consider the surgery at a time when respiratory function is still at least 30 per cent of that predicted for height and age, and when the curve is around 30 degrees. This often occurs at around 12 to 13 years of age (see the diagram on page 37). In recent years, the improvement in anaesthetic techniques has made surgery possible with much lower respiratory reserves
- mobilising or standing in calipers does minimise the development of contractures most effectively, however other methods such as standers, standing wheelchair or supine stander can be a preferred option
- when given the choice, very few boys choose to ambulate in long-leg braces.

Aquatic physiotherapy:

- continued to provide ease of movement and some respiratory work, but, most of all, it provides social and recreational opportunities. As muscle is replaced by fat and connective tissue, the body becomes less dense and some parts float more easily. As strength is lost, it may be necessary to provide a neck collar or other flotation devices to help maintain the head above water. Forward recovery is always difficult because of weakness in abdominals and neck flexors.

Pre and post-surgery physiotherapy is conducted as required:

Surgery for tendo-achilles lengthening, with or without tibialis posterior transfer to the dorsolateral aspect of the foot, is still the preferred choice for some surgeons and for the boys when contractures have occurred:

- to maintain flat feet on the wheelchair footplates
- to enable shoes to be worn
- for cosmetic reasons
- because polypropylene AFOs are too hot in the summer in some areas of Queensland.

After tendo-achilles lengthening, the physiotherapist will:
- provide post-operative respiratory exercise
- provide AFOs (which will need to be worn for a designated time after surgery)
- assist with pain and oedema relief if needed.

Please note: Pain after this surgery can be quite considerable when fixed contractures are present. Attempts to correct the position can sometimes result in multiple fractures of osteoporotic bones. Both parents and boys should be well informed before making the decision to embark on a late correction. In recent years, very few boys have agreed to tendo-achilles lengthening surgery.

Alternative choices should be discussed. (See seating recommendations on page 72.)

AFTER SCOLIOSIS SURGERY

General post-operative management after scoliosis surgery is documented in Spinal Fusion Management – A Physiotherapy Perspective (an initiative of the Paediatric Special Interest Group of the Australian Physiotherapy Association). Adaptations to the recommended protocols are needed to take into account the level of dependence of most boys with DMD.

Important post-surgery actions include:
- instruct in post-operative respiratory exercise
- avoid rotation (log rolling only) and forward flexion
- avoid distraction forces until fusion is complete (a normal two-person safety lift is not recommended)
- transfer via hoist, slide board or lifting slings only, or a cradle lift if the child is small
- ensure hoist slings are supportive, shaped and rigid or boned to prevent flexion. The sling should provide good support for the head. A removable polypropylene backboard in the normal sling can provide the easiest solution for the required post-operative period
- avoid prone lying and standing in a standing frame
- reassess wheelchair needs e.g. tilt in space and pressure-relieving cushion
- adjust the wheelchair to accommodate gain in height (raise height of back rest and armrests, etc.)
- assess changes in functional ability with an occupational therapist, especially hand to mouth for self-feeding which may have been lost with the fixation of spine. A simple resolution may be raised trays or tables to enable elbow flexion in the frontal rather than sagittal plane with gravity eliminated.

Regular manual handling techniques may be resumed once fusion is complete (on the advice of the orthopaedic surgeon).

Provide information and guidance:
- with other members of the multidisciplinary team to assist the boys to live fulfilling lives with particular emphasis on individual choices, educational and employment opportunities, wheelchair sports and other recreational pursuits.

References

1. Eagle M. Report on the muscular dystrophy campaign workshop: exercise in neuromuscular diseases Newcastle, January 2002. Neuromuscul; 2002.
2. Vignos PJ, Jr, Watkins MP. The effect of exercise in muscular dystrophy. Jama 1966;197(11):843–48.
3. Scott OM, Hyde SA, Goddard C, Jones R, Dubowitz V. Effect of exercise in Duchenne muscular dystrophy. Physiotherapy 1981; 67(6):174–76.
4. De Lateur BJ, Giaconi RM. Effect on maximal strength of submaximal exercise in Duchenne muscular dystrophy. Am J Phys Med 1979;58(1):
5. Fowler WM, Jr. Importance of overwork weakness. Muscle Nerve 1984;7(6):496–99.
6. Armstrong RB, Warren GL, Warren JA. Mechanisms of exercise-induced muscle fibre injury. Sports Med 1991;12(3):184–207.
7. Fowler WM, Jr, Taylor M, Rehabilitation management of muscular dystrophy and related disorders: I. The role of exercise. Arch Phys Med Rehabil 1982;63(7):319–21.
8. Fowler WM, Jr. Role of physical activity and exercise training in neuromuscular diseases. Am J Phys Med Rehabil 2002; 81(11 Suppl):S187–95.
9. McDonald CM. Physical activity, health impairments, and disability in neuromuscular disease. Am J Phys Med Rehabil 2002; 81(11 Suppl):S108–20.
10. Sockolov R, Irwin B, Dressendorfer RH, Bernauer EM. Exercise performance in 6-to-11-year-old boys with Duchenne muscular dystrophy. Arch Phys Med Rehabil 1977;58(5):195–201.
11. Petrof BJ. The molecular basis of activity-induced muscle injury in Duchenne muscular dystrophy. Mol Cell Biochem 1998;179(1-2):111–23.
12. Ansved T. Muscular dystrophies: influence of physical conditioning on the disease evolution. Curr Opin Clin Nutr Metab Care 2003;6(4):435–7
13. Grange RW, Call JAw. Recommendations to define exercise prescription for Duchenne muscular dystrophy. Ex. and Sports Sci, Rev. 2007;35(1):12-17. Review.
14. Allen DG. Eccentric muscle damage: mechanisms of early reduction of force. Acta Physiol Scand 2001;171(3):311–319.Disord 2002;12(10):975–83.
15. Hyde SA, Fllytrup I, Glent S, et al. A randomized comparative study of two methods for controlling Tendo Achilles contracture in Duchenne muscular dystrophy. Neuromuscul Disord 2000;10(4-5):257–63.
16. Scott OM, Hyde SA, Goddard C, Dubowitz V. Prevention of deformity in Duchenne muscular dystrophy. A prospective study of passive stretching and splintage. Physiotherapy 1981;67(6): 177–80.
17. McDonald CM. Limb contractures in progressive neuromuscular disease and the role of stretching, orthotics, and surgery. Phys Med Rehabil Clin N Am 1998;9(1):187–211.
18. Bakker JP, De Groot IJ, De Jong BA, Van Tol-De Jager MA, Lankhorst GJ. Prescription pattern for orthoses in The Netherlands: use and experience in the ambulatory phase of Duchenne muscular dystrophy. Disabil Rehabil 1997;19(8): 318–25.
19. Bakker JP, de Groot IJ, Beckerman H, de Jong BA, Lankhorst GJ. The effects of knee-ankle-foot orthoses in the treatment of Duchenne muscular dystrophy: review of the literature. Clin Rehabil 2000;14(4):343–59.
20. Finder JD, Birnkrant D, Carl J, et al. Respiratory care of the patient with Duchenne muscular dystrophy: An official ATS consensus statement.m J Respir Crit Care Med 2004;170:456–65.
21. Boitano JL. Management of airway clearance in neuromuscular disease. Respir Care 2006;51(8);913–22.
22.
23. Ward S, Chatwin M, Heather S, Simonds AK. Randomised controlled trial of non-invasive ventilation (NIV) for nocturnal hypoventilation in neuromuscular and chest wall disease patients with daytime normocapnia. Thorax 2005;60:1019–24.
24. Bach JR, Alba AS. Management of chronic alveolar hypoventilation by nasal ventilation. Chest 1990;97:52–57.
25. Mellies U, Ragette R, Dohna Schwake C, Boehm H, Voit T, Teschler H. Long-term noninvasive ventilation in children and adolescents with neuromuscular disorders. Eur Respir J 2003;22:631–36.
26. Simonds AK, Muntoni F, Heather S, Fielding S. Impact of nasal ventilation on survival in hypercapnic Duchenne muscular dystrophy. Thorax 1998;53:949–52.
27. Piastra M, Antonelli M, Caresta E, Chiaretti A, Polidori G, Conti G. Noninvasive ventilation in childhood acute neuromuscular respiratory failure. Respiration 2006;73:791–98.
28. Bach JR, Bianchi C, Vidigal-Lopes M, Turi S, Felisari G. Lung inflation by glossopharyngeal breathing and air stacking in Duchenne muscular dystrophy. Am J Phys Med Rehabil 2007;86:295–300.
29. Bach JR, Kang SW. Disorders of ventilation: weakness, stiffness and mobilization. Chest 2000;117:301–03.
30. Tzeng AC, Bach JR. Prevention of pulmonary morbidity for patients with neuromuscular disease. Chest 2000;118:1390–96.
31. Miske LJ, Hickey EM, Kolb SM, Weiner DJ, Panitch HB. Use of the mechanical in-exsufflator in pediatric patients with neuromuscular disease and impaired cough. Chest 2004; 125: 1406–12.

32. Mellies U, Ragette R, Dohna Schwake C, Boehm H, Voit T,Teschler H. Daytime predictors of sleep disordered breathing in children and adolescents with neuromusculardisorders. Neuromuscul Disord 2003;13:123–128.1

33. Simonds AK. Home ventilation. Eur Respir J 2003; 22:Suppl. 47, 38S–46S.

34. Eagle M, Baudoin SV, Chandler C, Giddings DR, Bullock R, Bushby K. Survival in Duchenne muscular dystrophy:improvements in life expectancy since 1967 and the impact of home nocturnal ventilation. Neuromuscul Disord 2002; 12:926–929.

35. Baydur A, Layne E, Aral H, et al. Long term non-invasive ventilation in the community for patients with musculoskeletaldisorders: 46 years experience and review. Thorax 2000;55:4–11.

36. Philipps MF, Quinlivan RCM, Edwards RHT, Calverley PMA. Changes in spirometry over time as a prognostic marker in patients with Duchenne muscular dystrophy. Am J Respir Crit Care Med 2001;164:2191–2194.

37. Bach JR. Management of neuromuscular ventilatory failure by 24 hour noninvasive intermittent positive pressure ventilation. Eur Respir Rev 1993;3:284–291.

38. Toussaint M, Steens M, Wasteels G, et al. Diurnal ventilation via mouthpiece: survival in end-stage Duchenne patients. Eur Respir J 2006;28:549–55.

39. Toussaint M, Chatwin M, Soudon P. Mechanical ventilation in Duchenne patients with chronic respiratory insufficiency: clinical implications of 20 years published experience. Chron Respir Dis 2007;4:167–77.

40. M Toussaint, P Soudon, W Kinnear. Effect of non-invasive ventilation on respiratory muscle loading and endurance in patients with Duchenne muscular dystrophy Thorax 2008;63;430-434;

41. Chatwin M, Ross E, Hart N, Nickol AH, Polkey MI, Simonds AK. Cough augmentation with mechanical insufflation/exsufflation in patients with neuromuscular weakness. Eur Respir J 2003;21:502–08.

42. Bach J. Mechanical Insufflation and Exsufflation 'Comparisonof Peak Expiratory flows with manually Assisted and Un assisted coughing Techniques. Chest 1993;104(5)

43. Bach JR, Ishikawa Y, Kim H. Prevention of pulmonary morbidity for patients with Duchenne muscular dystrophy. Chest. 1997;112(4):1024 –1028

44. Bach JR. Update and Perspective on non invasive Respiratory muscle aids. Chest 1994;105:1538-1544

45. Boitano LJ. Equipment options for cough Augmentation, Ventilation, and non invasive Interfaces in Neuromuscular Respiratory Management. Pediatrics 2009;123;S226-S230

Chapter 8:

Essential requirements for seating: assessment and wheelchair modifications

It is very important to make regular assessments of DMD boys' postures in their chair and adjust the seating as required. Frequent adaptations and modifications may be necessary: progressive loss of strength in the paraspinal muscles results in the inability to maintain an upright position against gravity. Efforts should be directed toward maintaining the best possible sitting position by providing postural adaptations and modifications as, or preferably before, they are needed. Other considerations may include adaptations to accommodate contractures and modifications to maximise declining upper limb strength and prolong functional ability.

The importance of the social and emotional wellbeing of a child with DMD cannot be emphasised too strongly. It is imperative that he is not isolated by his mobility problems and that he is given every opportunity to be involved in as many of the activities enjoyed by his peers as possible. It is for these reasons that provision of powered mobility (either a scooter or chair) is recommended earlier than in the past and to coincide with the time when walking becomes difficult and more spontaneous falls are occurring. The scooter or power chair will also afford some protection from excessive falls in the playground and busy traffic areas and save energy for the activities of his choice rather than expending the effort simply getting there.

The early stage

When providing the first chair for an ambulatory boy, the main consideration is to provide a comfortable means of transport for the child to cover longer distances and to avoid fatigue. Therapists will take time to determine the special needs of each family and discuss with them the relative attributes of different wheelchairs, buggies, scooters and other means of mobility.

Some boys learn to pedal a tricycle or bicycle with or without trainer wheels, which is a good alternative means of transport and is highly recommended for shorter distances. Other families prefer to provide a motorised option as soon as feasible so it can be put to limited use to enable their son to participate fully in family activities.

An infant's stroller or buggy may be appropriate for a very young child.

Advantages:
- it is perceived as more acceptable and 'normal' for a young child
- it is much easier to transport
- parents who have not come to terms with the disability accept a stroller more readily.

Disadvantages:
- it offers very little room for growth
- the peer group is not as accepting after a time as the stroller is perceived as 'babyish'
- mobility is totally dependent
- most strollers do not provide good supportive seating.

A larger buggy is a good alternative for some families for a short time. Some prefer to have both the wheelchair and buggy available so they can choose which is more appropriate for each outing.

When prescribing the first wheelchair, the therapist may not have the luxury of obtaining the perfect chair, as it may be one provided from stock or be on loan while waiting for provision of a power chair. However, features to consider should include:
- firm seat
- hips and knees should be well positioned with particular emphasis on pelvic alignment with even weight distribution on both ischial tuberosities and a neutral, or even a small anterior, tilt to the pelvis to help restore the normal lumbar curve
- full-length, height adjustable armrests to provide support for the arms at the height of the bent elbow. This will prevent the boy leaning down for support and, if extended, they will provide a means of preventing the legs rolling out
- feet flat on foot plates – make sure the plates are set at the correct height
- seatbelt
- brakes
- lightweight and folding (for ease of transport)
- push handles at the right height for carer comfort.

Self-propulsion of the manual wheelchair should never be recommended as a source of physical exercise for boys with DMD as the energy demands are high and strength in the muscles of the upper limb girdle begins to decline at around the time the chair is needed. In times past, a child was given a manual chair first, progressing to a power chair when they could no longer self-propel. It is now recommended that a power chair be provided early, with a manual chair provided as a back-up chair.

Scooters and other powered mobility tools are considered at any time in the ambulatory stage to assist with mobility and especially when walking is becoming tiring and falls are occurring more frequently.

1. There are many power scooters on the market, some of which require very little or no adaptation to seat height and depth to suit a young boy. Other scooters may need adaptations to provide a good fit and ensure good posture and support for the feet

2. Segway scooters are a stand-on, foldable and easily transportable scooter. They are becoming popular overseas
3. Zappy scooters are also stand-on scooters but have an optional seat to rest on when tired.

Advantages

- they are often more acceptable to the user in the ambulatory stage
- some scooters have a swinging seat to make transfers easier
- they can be disassembled to go into a car
- a seat elevator, if fitted, is very useful to rise to standing when the muscles in the buttocks and thighs are becoming weaker
- three-wheeled vehicles are more maneuverable than four wheels and boys do not have the balance problems of the elderly.

Disadvantages

- they are not as suitable for a progressive condition and are only recommended for boys who are walking at least part-time
- very few postural needs can be provided
- the electronics are basic – speed and gears only, no 'add-ons'
- they are not as suitable for inhouse use
- they are not as stable
- boys cannot travel in vans or taxis when seated in their scooters.

Monitoring closely for changes in sitting posture will enable intervention at the optimum time to prevent known problems.

Assessment at all stages will include:

- **the pelvis:** Both pelvic tilt (sagittal plane) and obliquity (frontal plane) and rotation (transverse plane) are assessed. A posterior pelvic tilt is a common feature accompanying the loss of the normal lumbar curve. An anterior tilting pelvis is also found in some boys who have progressed from the usual early kyphotic sitting posture to lordosis and later to a hyperlordotic posture. Regular reassessment and provision of seating modifications can prevent these postures becoming fixed postural deformities.

  Pelvic obliquity is rarely seen while the child is still walking unless ankle contractures are asymmetrical. Provision of a firm, narrow seat in the chair should be sufficient in the earlier stage to provide him with a stable level base of support.

  Provision of height-adjustable armrests is essential to help eliminate leaning which affects pelvic obliquity.

- **the spine:** Assess for deviation in the frontal plane (an incipient scoliosis is thought to be present in approximately 20 per cent of ambulatory boys and is often the result of uneven contractures in the ankle). In the sagittal plane, a reversal of the normal lumbar lordosis is extremely common when the child is seated (though lordotic when walking). A slumped kyphotic posture is common. Rotation in the transverse plane often accompanies a progressive curve
- **hips and knees:** Iliotibial bands (ITB) are assessed. ITBs are often tight, resulting in widely abducted and externally rotated legs. When seated, this splayed-leg position favours the development of equino-varus positioning and later contractures of the feet
- **hypertrophied calves** need to be accommodated in the later stages but are rarely a problem in a manual wheelchair where the seat is thin.

Late ambulatory stage

In the late ambulatory stage, physiotherapists will assess:

- pelvis (tilt and obliquity)
- spinal alignment in sagittal and frontal plane
- range of movement in hips, knees and ankles
- tightness in iliotibial bands
- size of calves.

The first power chair is provided toward the end of this stage. It is recommended that consideration be given to providing all of the following modification from the beginning to ensure good postural support from the outset, as well as preventing refusal of necessary modifications later on. Many boys with Duchenne do not readily accept change, others resist any modifications that they see as evidence of decline in their function.

- **the seat** should be firm but comfortable and help provide the best orientation of the pelvis. It should be narrow and provide a snug fit, to discourage sideways leaning. Ideally, thighs and buttocks will take 65 per cent of the body weight so it is important that the full length of the thigh is well supported. Approximately one to two fingers' space is needed between the front of the seat and the bend at the back of the knee. It is important that footplates are not too high as this will result in the thigh not being fully supported and more weight being taken through the ischial tuberosities which may cause discomfort and pressure problems. Most commercially available chairs come with room for growth in all dimensions (width, seat depth, back height and leg length). Many boys gain or lose weight during the expected life of a chair. One method of providing for weight gain and/or subsequent weight loss is to supply inset armrests mounted on horizontal brackets, allowing the width of the chair to be expanded or minimised to retain the desired fit. A narrow seat, which is a snug fit, is essential to prevent leaning down to the side for support. Pelvic blocks at the back lateral margins of the seat or incorporated in the cushion can help maintain a level pelvis. The front margin of the seat may need to be bevelled backward and downward to accommodate enlarged calves.
- **the backrest** should provide support for the normal curves and extend to the spine of the scapula. It absorbs 14 per cent of the body weight. If a harness is provided, a higher backrest is needed to prevent the straps digging into the shoulders. Keepers will prevent the straps slipping off the shoulders. A slightly reclined or tilted backrest (10 to 20 degrees) takes the weight of gravity off the erect spine and therefore provides some protection from rapid collapse. Currently, the MontroseAccess seating clinic is providing an I-shaped or indented backrest for mounting inset lateral supports. (See accompanying DVD, *An Overview of Physiotherapy Management Seating*.)
- **lateral supports** (also known as thoracic or scoliosis pads) key the child into the best sitting position and help prevent sideways leaning. Ninety per cent of boys (who have not had corticosteroid medication) will develop a scoliosis within two years of full-time wheelchair use. While thoracic supports cannot prevent a scoliosis developing on their own, they do discourage any sideways leaning and, in combination with other postural recommendations, such as a slightly reclined backrest angle and electronic tilt in space, they do assist in maintaining a good postural alignment. Currently, the MontroseAccess seating clinic is using snug curved laterals such as those supplied by AEL in combination with an I-shaped backrest to firmly support the chest wall.

- **a lumbar support** in the backrest will encourage a neutral or slightly anterior pelvic tilt (which is the desirable orientation of the pelvis) and thereby maintain the normal lumbar curve. One of the first signs of spinal collapse is the reversal of the normal lumbar curve. Newer chairs come with height-adjustable lumbar support on a firm backrest. Other chairs come with other means of providing a 'sacral push'. A separate lumbar-support cushion is often provided and attached to the backrest with velcro
- **a headrest** is supplied from the beginning as boys with DMD almost invariably have weakness in the neck flexors and abdominal muscles. This weakness makes it difficult for the child to bring his head forward when displaced backward. This backwards thrust often occurs when accelerating suddenly or when travelling up steep inclines. Without a headrest, boys adopt a protective, forward-leaning position, 'hanging' on the neck extensor muscles, in time these contract (in the same way that contractures in the feet occur). Eventually, the boys are unable to rest back against their backrests because the contractures in the neck extensors makes it impossible for them to bring their chin to their chest, which effectively results in a loss of the lower third of their field of vision. Headrests also allow the child to relax and recline when tired and they are essential when using the 'tilt in space' or recline mechanisms. Providing the headrest early will ensure that it is accepted. If offered later, boys inevitably see it as a sign of deterioration in their condition and reject it. Headrests are required by the Department of Transport if the child travels seated in his power chair in vans and taxis. Ideally, headrests should be adjustable in three planes to secure the most comfortable position. They should be shaped to provide some lateral support.
- **armrest height** must be adjustable to provide correct support for the forearm at the level of the bent elbow. Persistent leaning down for support will encourage the onset of scoliosis. If the armrests are too high, shoulders will be hunched and internally rotated. It is anticipated that 2 per cent of body weight is taken through the forearms when they are positioned correctly. Armrests that are adjustable inward so the width of the chair can be varied are also recommended as they can provide a narrow seat while allowing room for growth. Armrests that are removable make sideways transfers and toileting easier. When the boy needs to urinate, one armrest can be removed and the leg on that side moved out to the side. A urinal bottle can then be put in place without the need to move the child. Boys sometimes find tilting or reclining the chair assists this manoeuvre.
- **adductor pads:** Tight iliotibial bands lead to widely abducted and externally rotated thighs when seated**.** This position encourages the pattern of equinovarus contracture so common in boys with DMD**.** Some correction into adduction can be maintained by providing armrests, which extend the full length of the thigh and incorporate an adductor pad to hold the leg in the desired position. Alternatively, pads may be mounted on the footplate hangers. Another method involves raising the sides of the cushion to hold the thighs in place or separate pads slipped in between the thighs and the wheelchair arms (or kept in place with velcro). Some pressure-relieving cushions have adductor wedges as optional extras. Each method has advantages and disadvantages. Ease of toileting (when using a urinal bottle) must be considered. The easy one-step, removable armrest with adductor pad is often preferable to the need to remove two or more parts of the chair.
- **footplates:** 19 per cent of body weight is taken through the feet. To distribute this weight evenly, the feet should be positioned as flat as possible on the footplates. All boys who are no longer walking are encouraged to wear AFOs (orthotics) during the day while sitting in their wheelchairs to help prevent contractures in the ankles. A good position is then maintained and the feet can be well supported on the footplates. If feet are contracted and surgery is not a

consideration, then adjustable-angle footplates can provide more support for the feet. Pressure-relieving pads on the footplates can improve comfort (such as Roho air-filled pockets or gel pads). When feet are severely contracted, all the weight will be borne on a very small area (usually at the base of the fifth metatarsal) and pressure points are inevitable. Accommodation of severe contracture may necessitate specially constructed footplates that incorporate a softly padded well or cushioning made of micro polystyrene pellets to distribute and absorb the weight. The footplate-to-hanger angle can be adjusted for best range and maximum comfort. Adjustable-angle footplates can accommodate some degree of equinus. Swing-away footplates make transferring in and out of the chair easier for the boy who is using the chair part-time.

- **control box and position of the joystick**: The control box should be mounted on the armrest of the dominant hand so that the control knob lies directly under the hand at its normal resting position. This way, no reaching is required. Many therapists advocate the regular alternation of controllers from one side to the other in the belief that this will prevent leaning to one side, which, in turn, encourages scoliosis. Other suggestions include mounting the control box centrally or having it mounted on a swing-away arm, which is also mounted centrally. If careful monitoring of the seating issues mentioned above is carried out, all of which discourage leaning for support, these measures are not necessary and the control knob is best positioned on the armrest of the dominant hand directly under the hand. These points that minimise the risk of a scoliosis are:
  – position the joystick so that it is directly under the dominant hand
  – ensure the armrests are adjusted to elbow height
  – seat should be a snug fit, as seats that are too wide encourage leaning down for support (e.g. insert arm rests on horizontal brackets to permit outward adjustment as the boy grows)
  – provide thoracic supports to 'key' the child into an upright position
  – provide electronic tilt-in-space.
- **a tilt-in-space mechanism** is a highly desirable feature on all chairs for boys with DMD. The Mobility Aids Subsidy Scheme (MASS) in Queensland now funds this device and is prepared to include it on the first chair. All boys are encouraged to use it frequently to both protect their spines and relieve areas of pressure. A boy who has the tilt-in-space chair mechanism fitted on his chair will also be able to alter his own position in space at will. Tilt-in-space differs from the recline option found on most chairs as the whole seat module is moved into the tipped position and the angle at the hip is maintained. When chairs are reclined, only the backrest is moved, which opens the hip angle and can induce shearing and discomfort in those who have restricted range in hips.

  Fatigue: Older boys and adults often appreciate a daytime rest and find a full tilt (perhaps combined with a little recline) provides a good alternative to bed rest.

  Respiration: A tilted position can permit greater chest expansion, as well as facilitate the use of respiratory therapy and equipment such as the Ambu bag and the Cough-Assist machine.

  Two modes of tilting operation are available:

  Gas-operated mechanism: These are less expensive but are not recommended, as the boy is reliant on others to perform the tilt and he does not have the ability to move in and out of a tilted position independently. Boys are understandably reluctant to be tilted by any other person, for fear of being abandoned or stuck in one position.

  Electronically operated mechanism: This is the preferred option and it is recommended that it be provided as soon as the boy is using his chair full-time or preferably on the first chair.

- **backrest recline:** most chairs come with a reclining facility, which enables a boy to rest back in his chair. The angle at the hips is opened up, which is not a problem in the early years, but this position is uncomfortable for those boys who have significant hip-flexion contractures and shearing forces can occur. Tilting the whole seat module is then a better option.
- **elevating leg rests:** These are an optional extra and can be manually or electronically operated. They may provide some comfort when reclining and can assist in the management of lower-limb oedema. However, they are only useful if knee-flexion contractures have not yet occurred or are minimal.
- **seat belts** are essential requirements as many fractures of osteoporotic bones occur as a result of falls from wheelchairs. Traditionally, they are set at a 45-degree angle at the junction of the back and seat.

Non-ambulatory stage

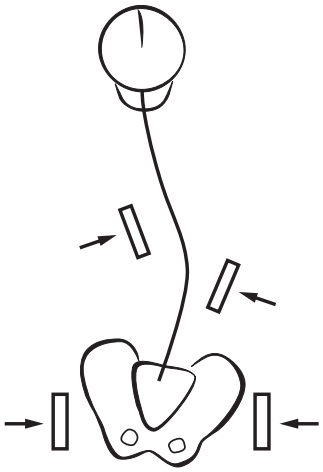
This stage will arrive gradually and with fewer problems for the child who has been a part-time user of a power chair. Therapists will need to monitor the boy's growth and any change in spinal alignment to ensure the optimum sitting position is maintained.

Regular appraisal will again include assessment of:
- pelvis – tilt and obliquity and rotation
- spinal alignment in sagittal, frontal and transverse planes
- range of movement in hips, knees and ankles
- tightness in iliotibial bands
- size of calves.

Particular consideration should be given to the seating requirements needed for:
- a scoliosis which is developing
- an existing scoliosis
- a hyperlordotic (excessively swayed back)
- fixed pelvic obliquity.

If a scoliosis is present or developing, three (sometimes four) points of control are used:
- one thoracic support at the apex of the curve
- another higher support on the opposite side to the apex of the curve
- one hip block on the same side as the high thoracic support, i.e. on the opposite side to the apex of the curve
- another hip block on the other side is optional but provides better control and prevents pelvic drift.

Specialist seating clinic advice is often invaluable once a fixed spinal deviation is evident.



Application of system of forces to support the trunk and spine and maintain neutral pelvis in the case where the pelvic obliquity is flexible.

A good chair will have provision for growth in all dimensions.

Likely alterations to the chair as the child grows include:
- adjusting armrest height as the child gets taller
- altering the depth of the chair's seat board and cushion as the thighs grow in length (the thighs should always be well supported)
- widening the armrests as the boy grows wider (on chairs that have horizontal adjustment) and expanding the chair's width, if needed
- providing a wider or a channeled top to the armrest for more support as upper-extremity weakness becomes apparent
- consider providing a lip on the outer edge to prevent the arms dropping off and a ledge at the back of the armrest to maintain the arm position when tilting
- lengthening the footplate hangers as lower legs grow – inadequate length will mean more weight through ischial tuberosities.

Other adaptations to the wheelchair are likely as the condition progresses, and other accessories may be provided. These include:

- **footplates**: Boys are encouraged to wear AFOs (orthotics) during the day while sitting in their chairs. A good position is then maintained and feet are well supported on the footplates. However, if the feet are contracted and surgery is not a consideration, adjustable, angled footplates can provide support for the feet over a larger area than what is available on normally positioned plates. When feet are severely contracted, all weight will be borne on a very small area and pressure points are inevitable. Accommodating contractures so that some weight can be more evenly distributed may necessitate a specially constructed footplate incorporating a softly padded well or an overlay of pressure-relieving material of either a gel pad or air such as provided by Roho.

- **elevating leg rests** are an optional extra on most chairs and can provide some comfort when reclining and also assist with lower limb oedema management. They are either manually or electronically operated.

- **harnesses** are invaluable for some boys in order to support the trunk against the backrest. A Bodypoint, Daher or similar four-point harness is possibly the most effective, but other forms of anterior chest support (such as a wide, soft chest strap by Bodypoint) can be successful. If a harness is to be used it should have:
  – large, clear neck opening
  – non-adjustable bottom straps (to prevent the harness riding up)
  – main area of support over the sternum (to permit free movement of the diaphragm)
  – place-guides or keepers on the backrest to prevent straps slipping off the shoulders
  – higher backrest so those straps do not dig into shoulders.

  It is recommended that harnesses always be worn when traveling in vans or taxis while seated in power chairs.

- **pressure-relieving cushions**: Cushions are rarely needed in the early stages of full-time wheelchair use, as boys are usually able to make small postural adjustments and rarely complain of discomfort that could be attributed to pressure. In the early days of wheelchair use, they are also able to wriggle forward in their chairs to allow independent use of the urinal bottle. A gel overlay cushion (e.g. Pilot or Akton Gel cushion) is a useful interim

cushion as it is comfortable and prevents shearing forces while still permitting the boy to move himself around in his chair. These gel cushions are also useful for long-distance car travel and make them more comfortable for boys with Duchenne. In the later stages, pressure-relieving cushions become very important means of maintaining not only comfort but stability and postural alignment as well. (See below for more about cushions.)

There have been no randomised control studies to evaluate the effectiveness of this seating regime, however anecdotal evidence and considered expert opinion would support the hypothesis that using a tilt in conjunction with lateral supports, headrest and a slightly reclined back rest, greatly improve postural alignment and reduce the incidence of spinal deviations.

Accessories

The following accessories are available on most wheelchairs and have obvious advantages for the school-aged child and young adult:

- a clear perspex or polycarbonate tray is a very useful accessory, not only as a work surface but also for upper-limb support and as a means of transporting needs and equipment
- drop-down or swing-away bracket for the control box and joystick enables the chair to roll under normal size desks and tables
- mid-wheel drive chairs are more manoeuvrable than rear-wheel drive chairs, however they are possibly not as suitable for the active young man doing a lot of outdoor cross-country driving as the driving position in the middle of the chair can increase the likelihood of becoming stuck where the terrain is uneven
- wider armrests are useful as upper-limb strength declines
- enviromental control units are available on better chairs.

(For more information, see the Occupational Therapy section on page 77.)

Cushions

Cushions come in four main configurations:

1. air cushions
2. foam and gel combinations
3. foam combinations
4. gel cushions.

All have distinct advantages and disadvantages depending on the particular needs of the child.

Air cushions

Advantages:

- provides the best pressure relief
- able to contour to any bony prominences
- best shock-absorbing qualities
- easy to clean.

Disadvantages:
- unable to secure the correct location of the pelvis
- unstable (rolls)
- difficult to maintain (punctures)
- easy to either over or under-inflate.

Many therapists like this cushion because of its undoubtedly superior pressure-relieving ability. However, extreme caution is recommended when providing a cushion for a boy who has a scoliosis, who is likely to develop a scoliosis, or who has had spinal surgery that did not extend to include the pelvis. The first principal of good seating is to locate the pelvis firmly, as it is the foundation of good positioning, supports the spine and provides alignment for the trunk. Air cushions are not as able to provide a secure stable base for the pelvis, especially for those with declining postural stability.

Roho can be the cushion of choice if:
- there is no or minimal scoliosis (stable spine)
- there is no pelvic obliquity present or likely to occur, e.g. in a spine fused to the sacrum
- the boy is heavier and wedged firmly in his seat
- other cushions have been tried and persistent pain or skin breakdown has not been relieved.

Foam and gel combinations

Examples of these cushions are Invacare Flo-tech Solution Xtra, Jay 3, Jay 2, Jay 2 Deep Contour, Otto Back Cloud, Invacare Infinity and Action Xact.

The base of these cushions is foam, which provides the stability, while the overlay is a gel or fluid-filled pad, which provides the pressure-relieving attribute.

Advantages:
- stable support
- accommodates mild to moderate bony prominences
- easy to maintain
- accessories and add-ons in some of these cushions (e.g. Flo-tech Xtra, Jay 3, Jay 2 Contour and Deep Contour) can secure the pelvis and provide correct alignment in both the sagittal and frontal planes, thereby accommodating or correcting pelvic obliquity and tilt. Add-ons can also minimise abduction of the thighs.

Disadvantages:
- some are heavier
- there is a bottoming-out risk in Jay 2, particularly if an incorrect prescription has been made. Measurements need to be from greater trochanter to greater trochanter, rather than providing a cushion to fit the chair. Jay 3 measurements are from ASIS to ASIS and easier for the novice prescriber.
- no resilience in some fluids, which can move completely out of the area
- in extremely hot conditions, the fluid can become viscous and less effective
- airflow is limited
- needs careful prescription. The prescriber of the Jay 2 cushion range needs to be trained as incorrect prescription will not fulfill any of the seating aims. The Flo-tech cushion and Jay 3 are more forgiving and do not need to be so carefully prescribed.

These cushions are arguably the best for boys with DMD. They are comfortable and have the capacity to make postural corrections or accommodations with the use of the add-ons such as hip guides, pelvic obliquity pads, adductor pads, hip and thigh aligners and extra fluid supplements or overfill gel pads, if more pressure relief is needed. The Flo-tech also comes with lateral and front wedges to assist lateral stability and tilt respectively. Boys with DMD very rarely have serious pressure problems that necessitate the highest pressure-relieving qualities of the air cushion, but almost invariably need support to control the position of the pelvis.

Foam combinations

Combinations of foam are thought by some to be the most effective for those boys who simply need a comfortable firm base. Examples include Flo-tech Contour and Flo-tech Lite, Invacare Ultimate Base, T-Foam and Jay Basic.

Advantages:

- stable
- lightweight
- good shock absorption
- adequate airflow
- low maintenance
- no risk of bottoming-out.

Disadvantages:

- postural adjustments are difficult
- hotter sitting surface
- pressure-relief is not as good as previous types of cushion
- needs protection from incontinence and spills.

Gel cushions

Gel cushions, as mentioned, are excellent interim cushions as they provide comfort and protection from shearing forces. These cushions come in three different thicknesses: 12mm (the 'commuter'), 16mm thickness ('the centurion') and 25mm-thick gel pad (the 'pilot'). The 25mm version is recommended for people who are sitting for long periods.

One disadvantage of gel cushions is that they are heavy. The Akton Grid gel cushion is a flexible, lightweight overlay, which provides very good pressure relief and protection from shearing forces and can be transferred between chairs easily, if this is required.

Chapter 9:
Occupational therapy management

As part of a multi-disciplinary team, occupational therapists aim to assist the child, carers, and family and school staff in managing DMD clients. When working toward the overall aim of improving quality of life, occupational therapists may adopt the Person-Environment-Occupation Model as a frame of reference. This is due to the dynamic and changing nature of the boys' lives.

To facilitate occupational performance, therapists aim to increase the congruence of person-environment-occupation as interlocking elements that are ever-changing.

Occupational therapists endeavor to work toward maximal participation of the child in home, school and community life. This may include assessment of developmental delays and learning difficulties, prescription of aids and equipment, activity adaptations, modifications in the home and school, as well as linking with community facilities and services.

Ambulatory stage – early and mid

Preschool

Play and development assessment

Play is the child's primary occupation and is important in the development of language, social, cognitive and fine motor skills. It is important to ensure the play environment is conducive to these developmental skills.

To determine the child's level of ability and if intervention is required, you may complete a developmental assessment in conjunction with a play assessment.

Some suggestions for assessments may include:
- Winnie Dunn's checklist[2]
- Hawaii Early Learning Profile (HELP)[3]
- Sensory Profile (W Dunn)[4]– sensory processing deficits can impact on a child's attention span and quality of play
- check for age-appropriate fine motor skills
- visual perceptual skills: WRAVMA[5] or Beery VMI[6]

If cognitive delays are evident, a referral to a psychologist may also be recommended.

Functional and self-care assessment

Begin by taking a parental history, and observe the child in a home, kindergarten or prep environment.

Toileting

Check for night-time incontinence and toilet training.

School readiness skills

Assess independence in self-care tasks (eating, drinking, toileting, dressing), social/peer interaction, organisational/planning skills and fatigue levels.

Intervention

Play and development:

- facilitate development of drawing and pre-writing skills
- provide activity ideas to facilitate the development of fine motor skills, scissor skills and age-appropriate grasp patterns
- position for play – recommend appropriate height tables and chairs to maximise function, as well as a footrest for support
- teach and model play skills with the goal of playing interactively with peers
- create specific treatment plans to address any delays in visual perception, cognition or general task-planning behaviours.

Self-care and daily routines:

- provide therapy sessions to address specific problems in self-care tasks
- develop independence in self-care tasks prior to school age e.g. putting on shoes and socks, washing and drying hands, feeding self
- encourage avoidance of excessively tiring routines
- liaise with parents and teachers to set up a routine for toilet training if required.

Links with other services:

- refer to educational support staff e.g. Advisory Visiting Teachers for Physical Impairment who can assist parents to choose an appropriate school, support transition to primary school and provide continuous support throughout the school years
- refer to inclusion-support agencies for teacher-aide assistance or equipment needs in long-daycare facilities.

Equipment

Play and development:

- Stirex-type scissors for children unable to grasp regular scissors
- spring-loaded scissors for children with reduced strength or delay in developing scissoring skills
- commercially available thicker pencils or pencil grips, crayons and built-up paintbrushes for children with immature grasp patterns or reduced tripod grasp strength
- softer lead pencils and felt pens that produce less friction and do not require as much pressure/effort
- slope board for drawing/writing.

Self-care:

- prescribe a toilet step with rails to allow the child to step up, turn safely and sit back onto the toilet
- provide a reducer ring insert for the toilet
- provide wet wipes to assist with bottom-wiping.

Early school years

Assessment

School:

- review the school set-up i.e. ergonomics of chair and desk, work area, access to implements/equipment, and undertake an environmental scan for any areas that can be avoided, such as stairs
- handwriting: quality and speed
- visual perceptual skills
- cognition, learning, social skills and behaviour
- toileting
- fatigue issues.

Home:

- toileting
- dressing skills
- bathing, showering, grooming
- routines
- goal-setting with family e.g. Canadian Occupational Performance Measure[7]

Intervention

School:

- information: Provide information on DMD and an overview of the expected pathway of the condition.
- assessments: Relay results from sensory processing, living skills and any other assessments so that an appropriate individual education plan can be developed. For example, many boys with DMD are reported by parents to be over-responsive to auditory, touch or oral sensory input. Problems maintaining or shifting attention appropriately and difficulty with changed routines are also often reported.
- learning: Boys with DMD are often described as visual learners. They have strengths in the visual perceptual domain while many have difficulties in verbal learning and verbal working memory. Visual prompts, visual diaries and photographic sequences are often successful strategies used.

- typing: Begin boys on a typing program to improve keyboard skills in preparation for computer use in the future.
- ergonomics: Ensure work areas such as desk and chair are the correct height.
- toileting: Many boys experience urinary frequency and/or urgency, which could be related to smooth muscle involvement. This may necessitate establishing routines or setting of regular toileting times. Regular hydration is encouraged in the early years to establish good habits in preparation for the later years when adequate fluid intake is needed to prevent constipation and for respiratory health.
- energy conservation: Suggestions for adaptations to the environment to minimise those activities that cause fatigue e.g. repeated standing up and sitting down on the floor, heavy loads, walking long distances, etc.
- rest breaks: For students who are having difficulty managing full school days, it is recommended to schedule rest breaks so the boys can have some time out. This will assist with behaviour management as outbursts are often more prominent when the boys are fatigued.

Home:

Dressing

- sitting will decrease energy expenditure and overcome any standing balance problems
- elastic-waisted or larger pants will be easier to pull on and off
- shoes with velcro fasteners are easier to fasten.

Home modifications

- begin early discussion and planning for major adaptation at home and at school, such as modifications to the bathroom, future outside and inside access for wheelchair use, and the bedroom.

Transport:

- if parents are ready to discuss future vehicle modification requirements, early provision of general information can assist families to make cost-effective choices.

Behaviour:

Behavioural problems are well documented in a small percentage of boys with DMD[9] Parents are encouraged to develop their child's strengths employing the following strategies:

- maintain the same expectations for behaviour for all the children in the family
- establish and maintain family routines, however be sure to provide warning time for changes of tasks and activities
- modify household tasks (e.g. chores) so that the child with DMD can participate
- implement sensory strategies to minimise the impact of sensory processing difficulties on behaviour.

Equipment

School and home:

- provide an angled writing surface such as a slope board
- provide supportive seating for the classroom
- assess whether the toilet may need an over-toilet frame, grab rails or a toilet step with rails
- provide bath and shower aids in the short-term to deal with increasing balance and stability difficulties. These can include a bath board, static plastic shower chair or stool, grab rails, non-slip matting, long-handled washer, soap-on-a-rope, etc.

Late ambulatory stage

Assessment

- identify parental concerns using a goal-setting tool such as the Canadian Occupational Performance Model[7]
- provide an environmental scan of the home with a view to modification. Circulation space, doorway and hallway widths need to be checked
- provide an environmental scan of the school in conjunction with the Advisory Visiting Teacher. Note stairs, steps, grates, slopes and surfaces, plus access to toilets, tuckshop, etc.
- check the student's ability to access all programs in liaison with the physiotherapist e.g. music, physical education, swimming, cooking and library
- make bathing, toileting, grooming observations and checklist
- check posture and seating at desk
- provide a computer assessment: access, keyboarding skills, mouse skills
- assess social skills, peer interaction and self-esteem with social worker and speech pathologist
- review strategies for learning with school staff
- assess transport to school, as the student may have difficulty in accessing a bus without assistance.

Intervention

Planning for access at home

- base recommendations on relevant Australian Standards, such as AS1428.1–2009
- apply for assistance for planning and/or building of major and minor home modifications through Home and Community Care Program (HACC) in each Australian state. They can advise on the suitability of installing a lift in highset homes or discuss incorporating a ceiling hoist into the design
- apply for purpose-built housing if using public housing or renting
- ensure continuous level access from the street or garage into the house and to all relevant rooms. Covered access from the garage to the house is preferred
- ensure the garage height is sufficient for a van and allow for flat loading space at the rear or side
- ensure floor surfaces are suitable for power wheelchair and for hoist.

Bathroom planning

- providing an en-suite adjoining the child's bedroom is ideal
- hob-free shower recess will accommodate a shower chair and allow room for an attendant
- long extension shower hose allows easier use
- toilet should be positioned within the bathroom to allow room for up to a bariatric-sized mobile over-toilet chair
- bathroom heater, exhaust fan and light combination provides warmth in winter
- if modification or relocation is not possible, other solutions are sought e.g. remove shower doors and replace with a floor-length curtain, then install a raised aluminium insert floor with drainage holes. A ramp bridges the hob into the shower recess. Custom-made models are available.

Bedroom planning

- close proximity to parents' room is preferred for turning the boy at night
- space should be allocated in the child's bedroom for a power wheelchair and mobile hoist, both of which require overnight charging

- provide a king single or, for larger boys, a double electric bed, positioned in bedroom to provide access on both sides for the carer
- consider the location of the TV and other entertainment in relation to the boy's bed.

Planning for access at school

- assist the Advisory Visiting Teacher recommending modifications required for access from the carpark into and around the school e.g. disabled parking bay, lift, ramps, threshold ramps, covered walkway, play areas, etc.
- ensure the toilet area is accessible with sufficient space to accommodate an electric hoist, a mobile over-toilet shower chair and a change bench
- provide change facilities at the swimming pool that are appropriate for female teacher aides to assist the male student. Pool hoists may be mobile or fixed and need to be suited to the design of the pool
- establish a parking place for the scooter or power wheelchair either inside or just outside the classroom
- place the boy's desk at the end of a row near the doorway to assist with safe movement into and out of the classroom
- store books within reach e.g. in a set of elevated trays alongside the boy's desk
- provide access to lunch box, hat and school bag.

Dressing

- boys may be fussy about clothing and/or textures, so offer assistance if tasks become too frustrating or time consuming
- choose garments that are easy to manage and free from buttons e.g. elastic-waisted shorts, loose-fitting t-shirts and pants
- encourage boys to sit when dressing.

Toileting

- some boys who are in the late ambulatory phase of their condition can experience urinary frequency and urgency. Accidents and bed-wetting are not uncommon and often resolve. A medical review is recommended should these problems interfere with daily life
- ensure adequate hydration, as there is a common tendency for boys to restrict fluid intake to avoid going to the toilet. This can result in health problems including urinary tract infections, constipation and thickened mucous secretions in the lungs.

  Strategies to avoid these problems include:
  - inform parents and teachers that urinary frequency is common with this condition and boys may need to attend the toilet more often
  - encourage boys to commence using the school's disabled toilet as the increased toilet height makes both sitting and rising easier
  - regular toileting times can prevent the embarrassing need for the child to ask for assistance. A buddy system may be an option
  - A collapsible urinal bottle with a latex pouch, e.g. a Uribag, is an option for long-distance car travel

Peer interaction and self-esteem

- maintain self-esteem and growth in personal maturity by encouraging participation in appropriate chores, e.g. fetching and carrying using the scooter or wheelchair
- employ strategies so the boy can continue to play at home with his siblings, e.g. provide a sturdy seat just inside the sandpit into which the child can swivel his legs

- provide a scooter or wheelchair to enable the boy to engage with his peers at lunchtime
- appealing indoor activities, such as Wii or table games, promote friendships
- referral to an inclusion-support agency for aide assistance to enable the boy's involvement in before and after school care or vacation care if parents are working.

Learning and school

- collaborate with school staff to develop an Individual Education Plan
- inform teachers of the current implications of the condition, e.g. effects of fatigue on school and homework, emotional issues from being unable to keep up with peers and how to manage falls
- suggest alternatives, keeping in mind that no boy wants to be different from his peers, e.g. review homework load, reduce written demands, instigate a buddy system, foster good organisation of belongings (desk pencil caddy, colour-coded books), timetable swimming classes with consideration to fatigue levels, etc.
- liaise with school regarding teacher aide time for learning support, keyboarding instruction, physical education, dressing after swimming class, etc.
- explore learning support software to enhance literacy and learning
- provide early preparation for the child's participation in camps and excursions in conjunction with the Advisory Visiting Teacher, e.g. transport to and from the venue, accessibility of accommodation, suitability of terrain for scooter use, program plans and teacher aide hours.

Managing falls

As the condition progresses, more frequent, spontaneous falls will occur. Rising from the floor to standing and regaining a balanced position becomes increasingly difficult. Some of the suggested techniques to help the boy regain standing are:

- props: Place a chair or other sturdy furniture close by so it can be used as a prop. Encourage the boy to move into four-point kneeling before leaning on the prop to climb into standing. Some physical support can be provided at the hips
- the through-arm safety lift could be used in an emergency situation, if the boy is able to offer some assistance, or if he is of small stature. One person takes the standard through-arm, cross-over position of the upper body while another attendant supports the child under his bent knees. They then lift in unison. This lift is appropriate as it provides protection for the boy's weakened shoulder joints (see the DVD for demonstration)
- the **Here to There Chair** divides the load between two or more attendants and as well as being a safer option for those performing the lift, it is comfortable for the child
- use of the hoist is recommended for most transfers. Education Queensland's guidelines for manual handling should be followed

Transport

- the School Transport Assistance for Students with Disabilities Program may provide taxi transport for the child to school. The school makes the application. Limited financial reimbursement of travel expenses is also available for parents who drive their child to a non-government school
- occupational therapists can help families apply for a Disability Parking Permit and for half-price taxi vouchers through the Taxi Subsidy Scheme. Both are administered by the Department of Transport in Queensland.

Equipment:

Seating

- use a swivel, gas-lift office chair. Check the safety of wheels on the floor surface
- a chair with sliders enables the student to push back to stand up
- a higher seat with a footrest at school or a foam wedge in a favourite lounge chair at home helps the student retain independent standing.

Bath and shower

- to facilitate stepping in and out of the bath, use a clamp-on bath rail or a bathboard
- electric bath lifters are a short-term solution
- provide a static, height-adjustable shower chair.

Toilet aids

- provide a raised toilet seat or over-toilet chair
- provide a footstool for under the boy's feet
- provide grab rails to provide steadying balance, e.g. for holding while a carer assists with cleansing.

Bed

- a mattress of medium firmness combined with a lightweight blanket helps to retain independent rolling in bed
- an electric bed may help the boy to retain his independence by assisting him in rising to stand.

Transfer aids

- these include sliding boards and transfer belts.

Transport

- a portable step can allow the student to step up into the school bus in the short-term.

Non-ambulatory stage

Assessment:

- reassess home and school access for independent mobility in a power wheelchair (see late ambulatory stage)
- monitor upper limb range of motion, especially flexors of elbow, wrist and fingers
- observe functional upper limb skills such as eating, drinking and handwriting
- determine equipment needs for personal care including showering and toileting
- introduce adapted clothing for dressing
- trial transfer equipment
- monitor sleeping needs, including comfort in bed and frequency of requests to be turned
- assess the boy's needs for assistive technology by observing keyboarding, mouse skills, etc.
- provide a school assessment with the Advisory Visiting Teacher regarding:
  - assistance for participation in practical subjects, and for note-taking, eating lunch, etc.
  - arrangements for toileting including regular or emergency bowel movements
  - school equipment needs
- assess leisure interests with a recreational officer
- advise on transport requirements for a boy seated in his electric wheelchair.

Intervention

Dressing

- use an adjustable-height bed or change table to ensure carer comfort
- loose shorts without underwear (or boxer shorts) simplify the use of the urinary bottle
- adapted clothing includes velcro-sided pants, trackpants with a disguised front opening flap, pleats on the back of t-shirts, ponchos that fold like jumpers when worn and backless pants
- as an alternative to purchasing modified pants, a dressmaker can modify existing school pants e.g. replace the side seam with velcro, zippers or buttons for easy access.

Eating and drinking

- a raised eating platform made from perspex allows the arms to be supported on an elevated plane, reducing the distance to reach the hand to the mouth
- a height-adjustable, over-bed table suitable for wheelchair access may assist
- dynamic pressure-driven lever arm supports may increase independence
- recommend a change in presentation or type of food e.g. pre-cut, bite-sized meat and vegetables and finger foods
- trial lightweight, extended cutlery
- encourage regular fluid intake by storing a drink bottle in a wheelchair cup-holder
- backpack drink systems can be useful but only if the boy has strong suction
- bendable straws, long straws, valved (one-way) straws and straw holders are other drinking options.

Toileting

Constipation is common once walking has ceased. The following simple management strategies should be trialled with the aim of developing healthy bowel routines:

- providing a fibre-rich diet
- drinking sufficient fluids
- providing suitable equipment, e.g. footstool and back support
- ensuring optimum positioning, e.g. leaning forward for bowel movements
- providing personal carers who ensure privacy and dignity.

If the above are not successful, natural stool softeners e.g. pear or prune juice could be trialled in the short-term or consult the boy's doctor regarding medication.

For more detailed information, see *Managing Toileting Issues for People with DMD: Some Practical Guidelines.*[9]

Hand function

- despite regular stretching, contractures of the long finger flexors, elbow flexors and pronators of the forearm may become apparent
- monitor tightening of the finger flexors. Resting pan splints may be of benefit.

Transfers

- train families, carers and teacher aides in using the hoist

demonstrate the use of slide sheets and correct manual handling techniques.

Learning and school

- as shoulder weakness progresses, adaptations or aide assistance may be required for practical classes such as art, manual arts and science, and for scribing, eating and toileting
- as handwriting becomes less efficient and more tiring for the boy, keyboarding is recommended
- email homework or notes to home, and provide photocopied notes

- print lesson notes from interactive whiteboards
- a second set of textbooks should be kept in appropriate classrooms
- fatigue is common in high school, so reducing the number of subjects provides 'spares'
- students with associated learning difficulties may benefit from alternative curriculum programs
- offer some alternate activity choices during lunchtimes with peers, such as Wii
- a power wheelchair facilitates participation in physical education and camps, however assistance, adaptations and equipment will be required for activities requiring transfers, arm strength and body involvement
- some extra-curricular activities will not require significant adaptation e.g. chess, choir or computer games. Others are specifically designed for children in wheelchairs e.g. wheelchair sports.

Computers

- laptops or netbooks should be trialled before ordering to ensure the keyboards are the correct size and layout for the boy to use
- USB sticks allow easy transfer of information from school to home
- scanning software enables documents to be scanned in (such as pages of books or worksheets) and the text is read aloud and highlighted e.g. *Texthelp Read and Write Gold*
- textbooks may have a computer-based version
- mouse alternatives include a trackball or mini trackball, an external touchpad, a joystick, eye gaze or infrared technology. Switches can be added to a switch-adapted mouse or to a switch interface to enable access to various mouse functions. Some advanced wheelchair controllers can also be programmed to control a computer mouse
- predictive text software can reduce the number of keystrokes and assist with literacy
- Windows has an on-screen keyboard as an accessibility option. It allows people to type using a mouse instead of a keyboard. It can be set to row or column scanning and activated by the mouse or through an external switch. This is time-consuming but provides independence
- text to speech software reads words on the screen out loud. It is available within the Windows program or online in a limited capacity
- voice-activated software converts speech to text. Voice commands can be used to edit and move within a document, to switch between programs and to access the internet. Good articulation is required and perseverance in training is necessary. Noisy classrooms are unsuitable for this technology.

Mobile phones

- some thumb or finger control is usually maintained. Using the speaker overcomes the difficulty in bringing the phone to the ear. Technology is available to allow mobile phone access and dialling of numbers through a switch option or through voice activation.

Environmental control units (ECUs)

- these devices allow independent control of any electronic devices in their immediate environment e.g. TVs, DVD players, air conditioners, lamps, light switches or electric garage doors. Some wheelchair controllers can be programmed as ECUs. Other ECUs can be accessed via a touchscreen, scanning switch control or by eye gaze. Expense must be weighed against benefits.

Gaming controllers can be adjusted for a lighter touch control.

Leisure and hobbies

- collaborate with the recreation officer and other team members to provide suitable leisure pursuits
- link the boy and his family with relevant community services, clubs and facilities for leisure and socialisation. Provide advice on transport and access issues.

Emotional support

- some strategies to assist insomnia include: night lights, personal pets, intercom system, siblings to share a room, relaxation activities prior to bed including visualisation, muscle relaxation, music and emotional coaching.[10]

Equipment:

Shower

- an attendant-propelled mobile over-toilet shower chair is now ordered. Boys are not encouraged to self-propel because of high-energy demands on weakening shoulder muscles. Usually supplied are a front-opening padded seat, a contoured head support, flip-back armrests, height-adjustable swing-away footplates, a pelvic strap (for safety), pan and pan racks (allowing for the potential of bowel urgency later)
- if accessible facilities are not available, e.g. when travelling, an option for showering is a portable shower tray, i.e. a vinyl tray with durable plastic sides that a mobile shower chair can be wheeled over. A hose can be connected to a drainage point.

Toileting aids

- a discrete, collapsible urinal bottle with a latex pouch and lid allows toileting while seated in the wheelchair, e.g. a Uribag
- an alternative is a non-spill urinal bottle with a one-way valve
- the task is made easier if the arm of the wheelchair is first removed and the boy's leg is abducted
- the Australian Government's Continence Aids Payment Scheme helps eligible families to meet some of the cost of their incontinence products, e.g. nappies, absorbent bedding protectors or urinal sheaths for night time.

Hoists

Once boys are unable to weight-bear, lifting aids become essential:

- a compact mobile hoist is suited to the home environment. Recommendations are a high-lifting range to clear a power wheelchair or bed, a wide leg spread, a pivot frame (for ease of positioning following surgery for scoliosis) and a sling with head support. Mesh slings dry quickly. Occupational therapists apply to the Medical Aids Subsidy Scheme in Queensland
- ceiling hoists are less intrusive, take up no storage space and are easy to use, however, they do not currently attract Queensland Government funding. They can either be portable, i.e. interchanged with other track locations in the house, or they can be fixed
- a hoist will be required at school for transfer to the over-toilet chair and to the change bench for undressing and dressing for swimming
- a pool hoist sling will require head support.

Beds

- an electric bed provides the boy with some independence to reposition himself at night. The bed can be adjusted to a good ergonomic height for carers
- the electronically operated backrest assists with sitting up as the abdominal muscles lose strength
- a king single-sized bed is often preferred as it allows additional space for ease of turning
- bed should have a hi-lo function, sufficient clearance under the bed for use of a hoist, an adjustable back rest, a knee break and a reverse Trendelenburg position
- funding for an electric bed may be available through organisations such as Muscular Dystrophy Association of Queensland
- electronically operated turning beds and turning mattresses are available and are designed to offer gentle repositioning throughout the night. They may suit some boys
- if an electric bed is not available, bed raisers provide clearance under the bed for use of the hoist.

Overlays and mattress replacement systems

- as the condition progresses, the boy gradually loses the ability to roll over or reposition himself independently in bed and may require frequent turning at night. At this stage, the young man also has the potential to gain or lose weight, which may have an impact on his level of comfort
- it is hoped that selecting an appropriate overlay or mattress replacement system will reduce the number of requests to parents for repositioning throughout the night and so minimise parental sleep deprivation
- overlays or mattress-replacement systems matching the level of pressure risk (high, medium or low) need to be trialled over a couple of weeks. The following are commonly used: static inflatable air-cell system, alternating air, gel, foam or a combination, e.g. foam and air
- trials of various types are essential as mattress comfort varies from one individual to the next
- the mattress chosen should be suited to use on an electric bed
- funding for pressure-relieving overlays or mattress replacement systems is available through the Queensland Health Medical Aids Subsidy Scheme.

Slide sheets

Slide sheets used by carers to simplify the task of turning adolescents in bed include:

- Smart Move slide sheets, which are made from an ultra-low-friction fabric
- WendyLett 2-way (sideways) or WendyLett 4-way (sideways, up/down) slide sheets have a central low-friction panel and are designed to remain in the bed
- A TopSheet can be placed over the WendyLett sheets, then attached to the sling bar of the hoist for turning heavier young men to ease manual-handling demands.

Mobility and transfer equipment

- small ramps such as the Personal Decpac can be carried on the back of a power wheelchair
- if an unsafe footpath or gutter is located on a regular route for the boy, apply to the local council for modifications
- beach wheelchairs can be hired throughout Australia (the wheels remove for easy transport). Refer to the Hire category on the Independent Living Centre or LifeTec websites in Australia
- information on specific transfer and evacuation equipment is also available from LifeTec or Independent Living Centres in Australia. Ensure that postural and head support is available when considering options. Boys with advanced DMD generally have osteoporosis so avoid jarring or bumping in all transfers.

School equipment

- wheelchair-accessible desk and/or a wheelchair tray
- hoist and sling with head support (see Hoists), a change bench and an attendant-propelled mobile over-toilet chair
- pool hoist with a sling with head support.

Vehicles

- financial assistance is not readily available in Queensland for families to purchase a modified vehicle
- some vehicles are not suitable to modify so families are advised to first consult with vehicle-modification specialists. Final approval of the modification must be sought from the Queensland Department of Transport
- Standards Australia sets standards for conversions, modifications and restraints
- minimum door-opening heights apply, plus the child requires sufficient headroom as he grows. It is advisable to ensure the boy will have good visibility from his position seated in the wheelchair.
- Options include equipping the vehicle with a dropped floor, which can allow ramped access, or to fit an automatic wheelchair loader which raises/lowers the boy into the vehicle while he is seated in his wheelchair. Choose either a rigid platform or a split platform for enhanced rear vision. Ensure the platform will be large enough for a solidly built young man's power wheelchair with a long wheelbase and extended footplates
- the wheelchair must be clamped to the floor of the vehicle by means of a wheelchair restraint system. Several types are available, including a vehicle conversion where a steel plate fitted to the base of the wheelchair docks with a base plate on the vehicle floor
- it is recommended that the boy wears a harness when travelling in conjunction with the required seatbelt
- second-hand vehicles that are already modified are an alternative choice
- LifeTec or Independent Living Centres in each state can provide further information.

Planning for adulthood

Planning for life after school is essential. Young adults with DMD may engage in tertiary study, paid employment, voluntary work and/or active recreation.

***Early planning*:**

- early planning is needed to address physical limitations, access issues and carer assistance. The process should commence at around 14 years of age
- it is important to assist with a holistic, collaborative plan, involving a wide range of stakeholders and support people. This may include parents, extended family, close friends, school case manager or class teacher, and therapists.

***Continuous planning*:**
- regular meetings with stakeholders ensure action plans are carried out and adjusted as the young person grows, alters his hopes and dreams, and comes to terms with changes in his physical abilities.

***Senior transition officers*:**
Where possible, a gradual transition to post-school activities is encouraged during the last six months of schooling. Specialist teachers who work as senior transition officers are available in many Queensland schools to assist with:

- planning for post-school options
- making applications for funding
- making referrals to appropriate agencies such as Centrelink and vocational support agencies.

Services to assist transition into adulthood:
- as supports from formal schooling reduce upon completion of secondary education, assistance from outside the family is helpful and age-appropriate for young adults
- it is important at this stage that daily routines and levels of activity and socialisation are maintained
- MontroseAccess has a lifestyle and leisure program that assists young adults to plan for their future by identifying and achieving goals. The program is aimed at assisting the transition into adulthood by providing opportunity for developing life skills, community access and social independence
- the program also monitors quality of life using the Personal Wellbeing Index (PWI-A) for adults over 18 years
- young adults with disabilities undertaking vocational training, looking for work, etc., and are unable to use public transport may be eligible for the Mobility Allowance. Application is made through Centrelink.

***Sexuality*:**
- young people with DMD have identical hopes and dreams as their able-bodied peers, including a desire for intimacy and sexual relations. Adolescents should be provided with opportunities to discuss these issues in private with support people of their choice
- some family planning and sexual health clinics may provide guidance. In addition, some consulting psychologists who specialise in working with young adults with a disability may be of assistance
- it is recommended that family culture and beliefs be considered to facilitate access to appropriate services
- limited expectations for people's lives should not be presumed, as a few young men with DMD have married and had children.

Occupational therapists, together with the other team members, aim to enable boys with DMD to continue to live a full and meaningful life, participating maximally at home, school and in the community.

References

1. Dunbar SB. Occupational Therapy Models for Intervention with Children and Families. Thorofare, NJ: SLACK; 2007.
2. Dunn W. Best Practice Occupational Therapy: In Community Service With Children and Families. Slack Incorporated; 2000.
3. Hawaii Early Learning Profile (HELP) VORT, 1995-1999 Palo Alto, CA.
4. Dunn W. Sensory Profile. The Psychological Corporation, San Antonio; 1999.
5. Adams W, & Sheslow D. Wide Range Assessment of Visual Motor Abilities (WRAVMA). Wide Range Inc; 2003.
6. Beery KE, Beery NA. Beery VMI (5th ed) NCS Pearson. Minneapolis; 2004.
7. Law M, Baptiste S, Carswell A, McColl MA, Polatajko H, Pollock N. The Canadian Occupational Performance Measure: A Research and Clinical Literature Review. Canadian journal of occupational therapy 2004;71(4), 210-22.
8. Hinton VJ, et al. Social Behavior Problems in Boys with Duchenne Muscular Dystrophy. Journal of Developmental and Behavioral Pediatrics 2006;27(6): p. 470-476.
9. Backhouse M, Managing Toileting Issues in People with Duchenne Muscular Dystrophy: Some Practical Guidelines. Montrose Access. Brisbane; 2005.
10. Hendriksen J. Towards a Brighter Future Conference. Sydney; 2010.
11. Forest M, Lusthaus E. Making Action Plans (MAPs) Pearpoint. Toronto
12. Pearpoint J,O'Brien J, Forest M. Planning Alternative Tomorrows with Hope (PATH) Toronto; 1991.
13. Home Modification Booklet: Duchenne Foundation website: http://www.parentproject.org.au/files/XCMOMCTU3E/Home per cent20Modifications per cent20Booklet per cent202004.pdf
14. International Wellbeing Group (2006). Personal Wellbeing Index. Melbourne: Australian Centre on Quality of Life, Deakin University. http://www.deakin.edu.au/research/acqol/instruments/wellbeing_index.htm.

Chapter 10:

Speech, language and cognition

The communication and learning difficulties experienced by many boys with DMD are often overshadowed by their medical condition and prognosis. Unlike their physical condition, their cognitive and communication skills do not deteriorate over time. Furthermore, with increased life expectancy and vocational opportunities, sound cognitive and communication skills allow boys with DMD to better access their environment and participate in valued community roles.

Researchers and anecdotal clinical reports indicate that boys with DMD have a greater incidence of cognitive and communication deficits than their non-affected peers. Boys with DMD also perform significantly more poorly on verbal IQ subtests than on performance subtests, however the exact nature of this deficit is not well defined. Both clinical observation and research indicate that boys with this profile demonstrate a broad spectrum of abilities. However, where cognitive deficits are significant, the DMD profile is usually more pronounced.

Language skills

Boys with DMD are often delayed in acquiring language milestones (first words, sentence length and complexity) in the same way that they are delayed in reaching their motor milestones. Early intervention is critical in this population as these delays are often related to impairments in cognitive skills.

As boys with DMD progress through school, more specific language characteristics become evident. Sentence generation in written and verbal tasks, complex language comprehension, verb and function-word deficits, and understanding and expression of abstract concepts are

frequently areas of weakness. In contrast, vocabulary, single-word expression and semantics are often areas of strength, which facilitate success in the classroom.

Phonological awareness (sound awareness) skills

Research has suggested that boys with DMD may have difficulties with processing and analysing sequences of speech sounds (phonemes). These phonological processing skills are necessary to learn to read and spell – important skills for later quality of life and vocational opportunities. Despite initial difficulties, some boys acquire these skills at a later age than their peers. For others, they continue to be areas of weakness. Alternative methods of accessing written communication may need to be considered for these boys.

Verbal memory skills

Studies have suggested that boys with DMD have significant difficulties attending to and processing complex verbal information. Of particular difficulty are tasks that require manipulation of verbal information e.g. summarising, inference and reverse digit span. It has been noted that boys often have a surprising strength in recalling specific words or concepts. However, more global comprehension and recall are generally weak.

Social communication skills

Many boys with DMD have difficulties initiating conversations, interpreting social situations, expressing emotions, communicating in novel situations and transitioning through activities in the classroom. These characteristics can impact on their ability to relate to, and interact with, their peers.

Home and school support

Throughout their lifetime, boys with DMD will benefit from a variety of interventions from both educators and therapists. Below are details of key issues that may arise throughout childhood and suggestions for assessment and intervention. We recognise many professionals have alternative assessments and intervention programs that they prefer. These ideas are recommended as a guide only.

Implications for the early years (0-5 years)

There are number of possible speech and language difficulties that can present in the early years. These include:

1. ***Delayed language skills***
   i) 0-3 years
   ii) 3-5 years
2. ***Auditory processing difficulties – 3-5 years***
3. ***Speech sound delays – 0-5 years***
4. ***Fluency disorders.***

1. Delayed language skills

i) 0-3 years:

It is important to assess receptive and expressive language early to obtain a baseline measurement of the boy's communication development relative to his same-age peers, as well as to identify areas that may require intervention. These skills can be assessed using a variety of checklists.

Assessment

Rossetti Infant-Toddler Language Scale, Receptive-Expressive Emergent Language Scale (2nd Edition, REEL) and *Macarthur Bates Communicative Development Inventories*. Other, more formal, assessments include the *Pre-School Language Scale (4th Edition, PLS-4)* and *Preschool Language Assessment Instrument (2nd Edition, PLAI)*.

Therapist intervention

- recommend Hanen's *It Takes Two To Talk* or *Target Word* as parent-based intervention programs focusing on developing interaction and early communication skills in children with language delays
- provide individual therapy sessions to stimulate language development
- provide parent education on language stimulation options.

Strategies for home and childcare

- develop early communication skills by encouraging imitation, choice-making, sound play, turn-taking and joint attention
- use short, simple and repeated instructions
- label new objects
- provide appropriate language models
- sing simple songs and say rhymes (e.g. Round And Round The Garden). Encourage the child to fill in the missing word at the end of the line
- encourage active listening skills
- encourage vocalisation and verbalisation to communicate, rather than gestures
- encourage early labelling of emotions.

ii) 3-5 years:

It is important to continue to monitor language skills throughout the preschool years in order to develop the boy's areas of weakness and to begin to encourage skills for school readiness.

Assessment

Commonly used assessments include: *Pre-School Language Scale (4th Edition, PLS-4)*, *Clinical Evaluation of Language Fundamentals – Preschool (2nd Edition, CELF-P2)*, *Renfrew Language Scales* and *Expressive & Receptive One Word Picture Vocabulary Tests (EOWPVT* and *ROWPVT)*.

Therapist intervention

- Hanen's *It Takes Two To Talk*, a parent-based intervention program focusing on developing interaction and early communication skills in children with language delays
- individual therapy sessions to support further language development
- parent education on language-development ideas.

Strategies for home, childcare and pre-prep programs

- refer to the above strategies for home and childcare for children aged 0-3 years
- encourage the development of spoken language by talking about 'here and now' events
- take turns talking and listening
- copy and expand out what the child says
  e.g. Child: "Car."
  Parent: "Yes, Mummy's car."
- model grammatically correct sentences
- frequently repeat new words
- read and talk about storybooks
- regularly read aloud
- use short, simple and repeated instructions
- use visual timetables to support comprehension and transition between activities
- offer opportunities for social interactions with peers.

2. Auditory processing difficulties – 3-5 years:

Auditory processing skills are essential to develop early language and literacy skills. These skills can be developed from a very early age and therefore are a good target for early intervention.

Assessment:

The following assessments can be used to identify delayed auditory processing skills: *Preschool and Primary Inventory of Phonological Awareness (PIPA)*, *Test of Auditory Processing Skills (TAPS)*.

Therapist intervention

- training active listening skills.

Strategies for home, childcare and pre-prep programs

- listen to music. Clap, march and dance to the beat
- model the correct pronunciation of mispronounced words by repeating the word and emphasising the correct sound
- encourage active listening: look at the speaker, keep your body still, think about the message
- limit background noise and distractions when communicating with the child
- model rhymes and songs for child to imitate.

3. Speech sound delays – 0-5 years:

Speech sound difficulties are present in all populations of young children, including boys with DMD. It is therefore appropriate to monitor these skills in the early years.

Assessment

The following assessments can be used: *Goldman Fristoe Test of Articulation*, *Articulation Survey*, *Diagnostic Evaluation Articulation and Phonology (DEAP).*

Therapist intervention

- articulation and phonological therapy.

Strategies for home, childcare and pre-prep programs

- model the correct pronunciation of mispronounced words by repeating the word and emphasising the correct sound.

4. Fluency disorders:

Some anecdotal reports suggest a greater incidence of fluency disorders in boys with DMD.

Assessment

Calculation of percentage of syllables stuttered.

Therapist intervention

- Lidcombe Program – early fluency program.

Strategies for home, childcare and pre-prep programs

- encourage the boy to start words and sentences gently with a slow, smooth voice
- limit questions
- limit talking when fatigued
- give the boy time to respond.

Implications for the early school years

There are a number of possible language and literacy difficulties that can present from Prep to Year 3. Depending upon the nature and severity of these difficulties, boys may require additional learning support in their educational setting.

These difficulties may include:

1. phonological awareness difficulties
2. expressive and receptive language delays
3. auditory processing difficulties.

1. Phonological awareness difficulties

Phonological awareness skills underpin early literacy development. Without appropriate support, boys with DMD may experience significant difficulties learning to read and spell.

Assessment

The following assessments can be used to identify delayed phonological awareness skills: *Preschool and Primary Inventory of Phonological Awareness (PIPA)*, *Test of Auditory Processing Skills (TAPS)*, and *Sutherland Phonological Awareness Test – Revised (SPAT-R)*.

Therapist intervention

- encourage Early Literacy Foundations (ELF) support program
- encourage sound-awareness skills
- develop the boy's ability to process and analyse sequences of speech sounds, e.g. sound-letter correspondence, sound blending and segmenting words
- develop programs for school and support their implementation in class.

Strategies for home and school

- establish the use of multimodal learning to reduce working memory load e.g. spelling word pictures and letter tiles
- clap out syllables in words
- talk about how sounds are made
- listen to and make up rhymes
- identify words that begin with the same sound
- show the letter that goes with each sound.

2. Expressive and receptive language delays

During the first years of schooling, boys with DMD may find the classroom environment challenging as they experience a variety of new language demands for the first time, such as interacting with a range of peers, following more complex directions and listening in a distracting environment.

Language delays may make it difficult for boys with DMD to participate successfully in learning experiences.

Assessment

Assessment regularly used for this age group includes *Clinical Evaluation of Language Fundamentals–Preschool* or *4th Edition (CELF-P2, CELF-4)*, *Renfrew Language Scales* and *Expressive & Receptive One Word Picture Vocabulary Tests (EOWPVT* and *ROWPVT)*.

Therapist intervention

- individual therapy programs
- school programs.

Strategies and activities for home and school

- present information in short, simple sentences
- repeat information frequently
- check if instructions have been understood
- use visual information such as pictures and gestures to accompany words, particularly in instructional situations
- explain new words and demonstrate the meanings of concepts
- read and talk about storybooks, and encourage the child to tell you the story in the correct order: "What happened first?", "What happened next?", "What happened last?"
- play games and explain the rules. Discuss situations the child may meet and how he might react. Encourage discussions about feelings
- encourage role-plays, e.g. with puppets, to act out stories and situations
- play barrier games
- play games such as Simon Says to develop listening skills and the ability to follow instructions.

3. Auditory processing difficulties

Auditory processing skills are essential for developing early language and literacy skills. These skills can be developed from a very early age and therefore are a good target for early intervention.

Assessment

The following assessments can be used to identify delayed auditory processing skills: *Preschool and Primary Inventory of Phonological Awareness (PIPA)* and *Test of Auditory Processing Skills (TAPS)*.

Therapist intervention

- train active listening skills
- do temporal patterning training, teaching children to analyse and imitate rhythms and beats, and discriminating different words in sequences e.g. "tick, tick, tick", "tick, tock, tick"
- train listening with background noise
- do localisation training, e.g. identifying where a sound is coming from.

Strategies for home and school programs

- listen to music – clap, march, dance to the beat
- model the correct pronunciation of mispronounced words by repeating the word and emphasising the correct sound
- encourage active listening – look at the speaker, keep body still, think about message
- limit background noise and distractions when communicating with the child
- model rhymes and songs for the child to imitate
- use appropriate classroom seating, e.g. facing the front of room, near the front and mid-centre of the classroom. Research suggests that immediately front and to the sides is also poor for children with auditory processing difficulty.

Implications for later primary school

Boys with DMD may present with difficulties in the following areas in the later primary years:

1. literacy and reading comprehension
2. verbal working memory and comprehension
3. narrative.

1. Literacy and reading comprehension

Boys with DMD may have difficulty associating letters and sounds and therefore may tend to use a whole word or visual approach in their acquisition of reading. They may have few strategies to use when attempting to read and spell unfamiliar words.

Assessment

Sutherland Phonological Awareness Test (SPAT-R), *Queensland University Inventory of Literacy (QUIL)*, *Neale Analysis of Reading Ability (3rd Edition)*, *South Australian Spelling Test*.

Therapist intervention

- individual, tailored therapy programs
- group programs, e.g. Phonological Awareness for Literacy (PAL)
- reading comprehension, e.g. *Visualising and Verbalising* program
- liaison with educational staff.

Strategies for school

- use multimodal learning to reduce working memory load e.g. spelling word pictures and letter tiles
- undertake over-learning of phonic skills
- explicit teaching of spelling rules
- use paired reading to improve sight-word vocabulary
- use reading comprehension activities e.g. keyword identification.

2. Verbal working memory and comprehension

Boys with DMD can present with difficulties attending to, remembering and understanding sequentially presented material, such as complex verbal instructions or stories.

Assessment

Clinical Evaluation of Language Fundamentals (4th Edition, CELF-4) and *Test of Problem Solving – 3rd Edition (TOPS-3)*.

Therapist intervention

- undertake auditory memory programs, e.g. Memory Magic
- visualise, chunk, categorise and rehearse programs
- train active listening skills.

Strategies for school

- present information in short, simple sentences
- repeat information frequently
- check if instructions have been understood
- use visual information such as pictures and gestures to accompany words, particularly in instructional situations
- play barrier games
- encourage child to repeat back instructions or information
- present information in chunks
- present information in context
- encourage self-talk to recall information, work through steps sequentially and support comprehension
- encourage memory strategies such as acronyms, diaries and post-its.

3. Narrative

Boys with DMD can present with difficulties producing verbal and written narratives.

Assessment

Parent and teacher report, language sample, informal narrative assessments, *Clinical Evaluation of Language Fundamentals (4th Edition, CELF-4)*.

Therapist intervention

- teach narrative structures
- use the Visualising and Verbalising program
- identify the most important information in paragraphs
- use 'who', 'when', 'where' and 'why' questioning strategies.

Strategies for school

- use narrative structures
- brainstorm ideas
- encourage role-plays, e.g. with puppets, to act out stories and situations
- illustrate stories to develop content
- present material to the class.

Implications for high school years

As boys with DMD transition into high school, the following difficulties may present:

1. literacy and reading comprehension
2. verbal working memory and comprehension
3. narrative and report writing.

1. Literacy and reading comprehension

Boys with DMD may continue to have difficulties with reading and spelling as they progress through school. As their academic load increases, they face increasing numbers of new or unfamiliar words. Intervention generally focuses on improving functional literacy outcomes, rather than literacy fundamentals.

Assessment:

The following assessments are used in this client population: Parent and teacher report, *Queensland University Inventory for Literacy (QUIL)*, *Neale Analysis of Reading Ability (3rd Edition).*

Therapist intervention:

- *Univ*ersity of Queensland Phonological Awareness Training for High Schools (PATHS) program
- *Visualising and Verbalising* program
- reading comprehension activities
- predictive text software, voice recognition software, text to voice software
- liaison with educational staff.

Strategies for school:

- provide a scribe for writing tasks
- provide access to predictive text and other software options
- provide access to audio books.

2. Verbal working memory and comprehension

Teenagers with DMD may continue to experience difficulties attending to, remembering and understanding material presented verbally. This can include complex verbal instructions, stories or subject content.

Assessment:

Clinical Evaluation of Language Fundamentals (4th Edition, CELF-4) and *Test of Problem Solving 2 – Adolescent (TOPS)*.

Therapist intervention:

- use auditory memory programs e.g. Memory Magic
- visualise, chunk, categorise and rehearse programs
- train active listening skills.

Strategies for school:

- use visual information, such as content summaries, pictures and gestures, to accompany lectures, particularly in instructional situations
- present information in short, simple sentences
- repeat information frequently
- check if instructions have been understood

- encourage repeating back of instructions or information
- present information in chunks
- present information in context
- encourage self-talk to recall information, work through steps sequentially and support comprehension
- encourage use of memory strategies such as acronyms, diaries and post-its
- develop note-taking and summarising skills
- highlight key words or concepts.

3. Narrative and report writing

Teenagers with DMD may continue to experience difficulties producing verbal and written narrative, and may experience difficulties planning, organising and completing assignment and report writing tasks.

Assessment:

Common assessments completed with this population include: Parent and teacher report, informal language sample, informal narrative assessments and *Clinical Evaluation of Language Fundamentals (4th Edition, CELF-4)*.

Therapist intervention:

- individualised programs to develop brainstorming and planning skills
- *Visu*alisi*ng and Verbalising* program.

Strategies for school:

- teach or provide clear scaffolding for assignment tasks
- encourage brainstorming of ideas
- teach assignment structures
- provide assignment-planning support.

Mealtime issues

There are a variety of mealtime issues that are observed in the DMD population. In younger boys with DMD, sensory feeding issues are often present. As the condition progresses, reduced oral motor skills lead to mealtimes becoming more and more challenging.

1. Infants and school age

Young boys with DMD can present with a variety of sensory-based mealtime issues. They often also present with other sensory issues, e.g. they don't like touching wet or soft textures. Anecdotal therapy reports suggest that these sensory difficulties can influence mealtimes, leading to strong food preferences or aversions. These children may be described as fussy eaters who choose only to eat certain food types or textures.

Assessment

Assessments should include food and mealtime profile (including food types, textures and tastes preferred), the Sensory Profile and informal mealtime observations.

Therapist intervention:

- introduce food desensitisation program to introduce challenging food types
- change mealtime routines
- support parents to choose progressively more challenging foods.

Strategies for home and school:

- undertake a gradual exposure to a range of food options, e.g. food play, trying plate
- teachers and therapists need to be aware of food preferences to accommodate children at mealtimes and to stretch the child's tolerance of food options

2. Late teens and young adults

As their condition progresses, mealtimes become more challenging for boys with DMD. As their muscles continue to deteriorate, their oral motor skills begin to decline. They generally begin to lose strength and mobility when chewing and manipulating food in their mouths and begin limiting both food options and food intake as muscle weakness and fatigue increase. They may also be at risk of choking and/or aspiration during mealtimes due to weakened or slow triggering of swallowing function, reduced oral skills and difficulties coughing to clear food in the airway or pharynx.

Assessment:

Assessments should include a clinical evaluation of oral motor and swallowing skills, cervical auscultation and referral for a modified barium swallow if required.

Therapist intervention:

- refer for a modified barium swallow if required
- recommend food textures and fluid modification, e.g. soft diet, easy chew, thickened fluids
- establish mealtime strategies to ensure swallowing safety, e.g. moistening food, effortful swallow, assisted airway clearance
- provide recommendations to optimise food choices
- refer to dietician to optimise kilojoule intake
- liaise with medical staff to provide information about the risks of oral feeding and the appropriateness of percutaneous endoscopic gastrostomy (PEG)
- provide information to the young man and his family about feeding options and support their decision-making.

Strategies for home and school:

- modify food and fluid at mealtimes
- supervise mealtimes
- implement mealtime strategies.

Chapter 11:

Social and emotional issues

Families with children with disabilities and families with children without disabilities are more alike than different, and every family at differing times has particular needs.

Current philosophies and trends in service provision for people with disabilities are the basis for this discussion of how best to assist families with the social and emotional issues of caring for their children with Duchenne muscular dystrophy.

Principles underpinning state and federal legislation in this area include the rights of disabled people to respect for their human worth and dignity, to realise their individual capacities for physical, social, emotional and intellectual development, and to full participation in community life.

For children, others state that the family is the best place for a child to be raised and supported, that families must be fully involved in decisions about their children, that services must be focused on meeting the needs of the child and family, and that children have a right to a safe, secure and nurturing environment.

The progressive nature of Duchenne Muscular Dystrophy means that the families' needs do change over time and there are certain critical periods when stress is likely to be greater.

These are:

- diagnosis
- early childhood
- adolescence
- end of life.

Grief

Grief has been defined as the psychological and emotional process set in motion as a result of a loss of – or failure to realise – expectations in a situation where there has been a degree of emotional involvement. Families of children diagnosed with Duchenne muscular dystrophy face many losses: their 'normal' child, their hopes and expectations for his future, their lifestyle and, ultimately, the child himself.

One way of understanding their emotional responses is to use *The Stages Theory of Grieving*, which was originally identified by people working with terminally ill patients and their families. The generally accepted stages and common emotional and behavioural responses are:

Shock
stunned
numb
unable to focus
crying
sighing

Denial
disbelief – 'this can't be happening', 'it's not true'
refusal to believe test results
searching for proof of misdiagnosis and second opinions

Bargaining
engaging in activity in the hope of a change in the situation
searching for alternative treatments

Anger
blaming others, e.g. doctors
outbursts of rage

Guilt
blaming oneself
belief that they are being punished

Depression
hopelessness
embracing not only the present but the future
overwhelming sadness
crying
apathy
loss of energy and motivation
suicidal thoughts

Acceptance
adapting to the reality of the loss and the changes that are necessary to live with loss in a constructive way and get a more realistic understanding of the child's limitations and potential.

These stages are considered a positive, natural process that assists people to gradually come to terms with a loss that would be too traumatic to cope with immediately. Not all parents experience grief in the same way, certainly not in any particular order. Any or all of the reactions may recur throughout the child's life and this is often referred to as chronic grief.

Diagnosis

The experience:

Most families are utterly devastated by the diagnosis. Up to this point, they have had a reasonably normal, healthy child.

For some, the symptoms that have prompted them to seek medical advice have prepared them a little for what is to come but others are completely overwhelmed by the news.

Shock and denial, which are very common around the time of diagnosis, are protective reactions, which insulate the grieving person from the full horror of the news. The expression of strong emotions such as anger and guilt is important in the healing process.

The issues:

How the news is given

Whether parents react to the diagnosis with an emotional outburst of despair and crying or the numbed silence of shock, most of them will have a vivid recollection of the occasion in emotional terms. They may not remember much of *what* they were told but they do remember *how* they were told.

It is therefore extremely important for health professionals at this stage to show sensitivity, empathy and a caring attitude, even though they also find the situation personally stressful.

Information

In their shocked state, parents' ability to absorb and understand all of the information given to them may be impaired and health professionals need to be patient and prepared to go over information more than once if necessary. They also need to individualise the amount of information and how it is presented, as some parents will want much more as a way of gaining some control (knowledge) in a situation where they have little control. Others will want only basic information for a start, but will be more open as they adjust to the news.

Time

Many parents have said that they felt rushed through this process and that it would have been nice to have more time with the professional to absorb information, ask questions, etc. If time is limited in a busy clinic, it would be ideal if parents could be referred to a social worker who could offer them support at this difficult time.

Genetic counselling

The diagnosis of DMD has a significant impact on a couple's plans and expectations for their whole family, including existing children who may be carriers and future pregnancies. Early referral to a genetic counselling service is important to assist parents to make informed decisions in this area.

‘What do we tell our son?’

This will depend on his age at diagnosis.

Experience has shown that children can be given information that is commensurate with their level of understanding and relevant to the present. For example, a preschooler may have his condition explained as having weak muscles so he can’t run as fast as others or falls over more. It is not necessary to tell him he will need a wheelchair when he is older or that he is going to die. He will not understand these abstract concepts and he may become unnecessarily anxious. He will probably pick up on his parents’ anxiety anyway. He needs their love and the security of their care for him more than he needs the whole story.

If he does ask more specific questions, parents can be encouraged to give simple answers that are relevant to the context.

Relationships under stress

Differences between family members’ grief processes may result in increased stress on family relationships. For example, when one partner has come to a degree of acceptance of the diagnosis while the other is in denial, there may be arguments about treatment plans. One partner may express anger in unacceptable ways, resulting in domestic violence. These difficulties may be evident quite early after diagnosis and may result in sudden family breakdown.

Health professionals should be aware of these possibilities and refer families for supportive counselling early, rather than wait for obvious signs when it may be too late.

Grandparents and others

Parents need considerable support from extended family and friends at this time. However, these people may be experiencing their own grief reactions and may themselves require support as well as being unable to fully meet the needs of the child’s parents.

This is another reason for referring families early to a supportive counsellor, who can take some of the responsibility from the extended family so they don’t burn out and are still around and able to help later when they are needed more.

Financial assistance

Parents of boys with DMD may be eligible for a carer allowance to help with the costs of caring for them. This allowance is free of income or assets tests and is not taxable. Parents should be encouraged to apply even if the child is very young. They may also be eligible for a carer payment.

Contact the local Centrelink office.

How to help:

- remember that people are individual and unique
- remember that this is a new experience for them
- don’t judge or assume
- be patient
- be prepared to give information more than once
- don’t say, “You should…”, instead say, “You can…”
- accept reactions as natural. Crying is OK.

Early childhood

The experience:

There remains some doubt as to whether parents of children with disabilities ever reach a stage of acceptance in their grieving. For those whose children have DMD, these early years can bring joy tinged with pain as they watch them develop skills and achieve milestones while, at the same time, they see evidence of increasing weakness. While they can be encouraged to see the positive aspects of their child's development, they continue to need support as they may feel acutely the irony in view of the future prognosis.

The issues:

Alternative treatments

Some parents continue to doubt the diagnosis and prognosis in the light of their son's development or they may seek alternative treatments or regimes, which, at this stage, appear to have positive results in halting or slowing the progress of the disease.

Health professionals need to understand that seeking alternative treatments is a common response to the powerlessness many parents feel. However, parents may be vulnerable to exploitation, both emotionally and financially, and the provision of adequate and accurate information continues to be necessary in order for them to make informed decisions about treatment.

More family stress

Ongoing grief and other stresses may continue to affect relationships. Individual support to parents remains important but parent-support groups can also provide many opportunities to express feelings and share experiences and ideas. Unfortunately, these are not always available in all areas. MontroseAccess currently offers groups and also has a program of weekend retreats for parents.

Siblings

The increased attention given to the disabled child may lead to difficulties for siblings. Feelings of guilt are common and can be related to ambiguous feelings toward the disabled child (envy of the attention he receives and sympathy for his condition) or to the fact that they have escaped the condition. They may also fear 'catching' the disease and may have extra responsibility placed on them to care for the disabled child. There are some programs available for siblings of children with disabilities through MontroseAccess and Queensland Council of Carers, as well as some publications.

The child

As the child himself grows more aware of the implications of his condition he, too, will experience grief for his lost abilities and experiences and may express his anger and frustration in his behaviour.

Behaviour management

Parents may experience difficulties in managing their disabled child's behaviour. This may be linked to their own continuing grief and difficulties in establishing consistent routines and

management strategies. However, it is important to remember that many boys with DMD have significant communication and learning difficulties and that they, too, may be expressing emotional reactions to their loss. All of these factors should be taken into consideration before recommending any form of intervention in this area.

It is particularly important that parents are not made to feel inadequate.

School

School is a normal part of life for all children. It is the place where they make friends as well as learn, where they can experience achievements in many areas apart from the physical, all of which contribute to their self-esteem and emotional wellbeing. Although many boys go on to further education and/or vocational training and employment, school should not just be seen as a preparation for life.

The process of enrolling the child in preschool and school can be difficult emotionally as families are faced with choosing a placement with appropriate facilities to accommodate the child's increasing disability. Education Queensland has advisory visiting teachers for children with disabilities in most areas of the state. They can assist parents with information about schools with appropriate facilities and can make arrangements for support for the children at school.

Education Queensland has a system of verification, which is the process of confirming that a student's identified impairment, and the associated activity limitations and participation restrictions, will require significant education adjustments. The system also verifies that they meet criteria for one or more of the six Education Adjustment Program (EAP) disability categories, including physical impairment, learning disabilities or intellectual impairments. Verification can proceed for students when they are of prep-eligible age or older and enrolled in a state education facility. Verification in the EAP disability category of Physical Impairment can also occur for eligible students attending non-state schools.

Once verified, the student is eligible for EAP. This is a process for identifying and responding to the educational needs of students with disabilities. Adjustments are made for students with disabilities to enable them to access the curriculum, achieve curriculum outcomes and participate in school life.

Intellectual impairment, if identified at this stage, may be an added blow that sets the whole grieving process in motion again.

Throughout the child's school life, there will be many meetings as the child's condition deteriorates and his needs change. Again, while the benefits of continuing good communication between school and families cannot be denied, it is well to remember that parents may experience stress on these occasions. Some parents have appreciated having someone accompany them to these meetings for support. This could be a family member or member of the support team.

'What do we tell the children?'

Parents will vary in their willingness to allow their child's classmates to be told about his condition. Where they agree to some information being given, parents and teachers can devise a plan for this together. Therapists working with the children can also assist in this process.

A useful resource for teachers is *Positive Peer Relationships–Including a Student with Duchenne Muscular Dystrophy* (Rocky Bay Inc., PO Box 53, Mosman Park WA 6912).

Wheelchair

A particularly difficult time for parents is when the child is no longer able to walk and has to rely on a wheelchair for mobility. Therapists working with the child have a good idea when this is likely to happen and will be preparing the child, his parents and teachers in advance. This is also necessary as it can take some time for the wheelchair to come after it has been ordered.

Many parents try to put off this moment for as long as possible, denying that it is necessary and again looking for alternatives. They may need gentle persuasion to accept even the ordering of the wheelchair. The child himself will often be relieved, however, as, leading up to this stage, he will have found it increasingly tiring getting around, with frequent falls and some discomfort.

Paradoxically, the arrival of a power wheelchair and relief for the child often coincides with a period of aggressive behaviour. The child up to this point has been losing mobility and becoming increasingly dependent on others for everyday needs. Compliance and politeness are necessary for survival. The power wheelchair represents a return to some independence and control as well as an instrument for expressing anger and resentment. Ramming the wheelchair into people and furniture is not uncommon, as well as increased verbal aggression and non-compliant behaviour.

Management of such behaviour needs to be firm, especially if it is dangerous, but sensitive to the fact that children at this age are beginning to develop more adult concepts about life, death and disability and becoming more aware of the reality of their condition. They, therefore, feel the pain of their loss more acutely than previously, especially as they see their friends becoming more independent.

Boys at this stage may require more direct individual supportive counselling, or their parents and carers can be assisted to understand their responses and to provide appropriate support.

Other assistance

- Queensland Transport provides assistance through its Taxi Subsidy Scheme and Disability Parking Scheme
- Queensland Housing can provide specially designed rental housing for families with a disabled child.

Adolescence

The experience:

This is a time of turmoil for many young people, with or without disabilities. For boys with DMD, the usual issues of adolescence are compounded by their increasing disability and dependence.

The issues:

High school

Careful planning, well in advance, can facilitate the transition from primary to high school. Education staff, especially advisory visiting teachers, need to identify issues such as access, curriculum, equipment and support needs, and to arrange in-servicing of teachers and other staff about the condition and the student's individual needs. The therapy team working with the student can often help with this.

Independence

This is the time when most adolescents are exerting their independence and is the very time that boys with DMD are becoming more dependent on others for their physical needs. Parents, carers, teachers and health professionals can help by encouraging them to be more involved in decisions about their lives.

Surgery

Parents often feel ambivalent about spinal surgery, which may be recommended for their sons around this age. Adequate information about the benefits and risks of the procedures, as well as the opportunity to discuss their concerns, is essential for them to feel comfortable about their decisions.

It can be helpful for them to be put in touch with other families whose son has had surgery but care should be taken that the contact does not have a negative impact. Helping professionals should provide continuing support for their decision and avoid judging, whatever the outcome.

Anxiety

Many boys experience increasing anxiety, which can become a significant problem in adolescence. This anxiety is often related to their physical management and they express considerable fear of any change in routines or carers. Boys may object to anyone but their parents caring for them so that even if families are able to access help in this area, the parents are unable to take advantage of it.

This can also affect the boys' social relationships and recreational and vocational opportunities and can increase social isolation for them and their families. Boys at this age may benefit from individual counselling to address these issues.

Intellectual impairment or autistic tendencies compound the issue for some boys and support for parents to find strategies to manage the difficulties is more appropriate.

Where respite care or personal care assistance is planned for families, care must be taken in selecting and training personnel, and patience and persistence is needed to gradually introduce changes.

Anxiety about the future becomes more of an issue as boys become increasingly aware of their deterioration. Open communication within families from the early stages will help to minimise this anxiety. Families require continuing support in this area and some boys will benefit from individual counselling. Some boys will turn to others for help, most likely people with whom they have established a trusting relationship, such as a carer, teacher or teacher's aide. These people can help by listening supportively, answering general questions as simply as possible, and encouraging them to talk to their parents or other helping people.

While educational inclusion policies have improved the lives of boys with DMD, the downside can be the isolation a boy may feel when he is the only student in his school with a disability, let alone with DMD. Getting to know other boys with DMD can be helpful and they will often share concerns that they would not share with their families. MontroseAccess organises camps for boys of different ages through the year and there are others held interstate. Access to a local wheelchair-sporting group is a good way to establish relationships, too.

Boys may also like to communicate with each other via the internet. LiveWire, run by the Starlight Children's Foundation of Australia, provides a safe online community for teens living with serious illness or disability. Facebook and texting are becoming popular means of communicating, as are list servers on the Internet such as DMD Pioneers.

Physical care

The physical demands of caring for a boy with DMD, of course, increase as his condition deteriorates and can leave his family exhausted, adding to emotional stress. Practical assistance with equipment and advice on managing these tasks is discussed in previous sections.

Some services are available to assist families with the physical aspects of care, such as bathing, dressing and exercise programs. These vary from region to region. Contacting Disability Services Queensland in the local area can assist families to obtain information.

Some form of respite, either in a residential setting away from home or in the home (while the carer goes out or away) can provide carers with a break from these demands.

Post-school options

Vocational opportunities for boys with DMD are limited by their increasing disability. Some boys, after completing their secondary schooling, go on to participate in tertiary or further education and obtain employment suited to their abilities.

They require considerable support to achieve these goals but the outcomes for these young men in terms of their self-esteem and independence are well worth the effort.

For others, it is important for them to have some meaningful and enjoyable activities that will enhance their life after school.

The Disability Services Queensland website at http://www.communities.qld.gov.au/disability provides details of how to apply for assistance.

Financial assistance

When boys turn 16, they may be eligible for a Disability Support Pension, Mobility Allowance and Pensioner Education Supplement. Parents may qualify for Carer Payment or Carer Allowance also. Contact the local Centrelink Office.

End of life

Despite the relatively predictable course of the condition, many factors combine to influence the final outcome for individual boys. For some, there is a long and gradual decline where the type of assistance required will not change very much but the amount will increase as they become more dependent on their carers for everyday tasks. Government funding of such assistance still does not meet the demand and many families struggle through this period, often suffering health problems of their own through injury and stress. Any assistance available needs to be investigated, whether through local government-funded agencies or community organisations, such as church groups.

Generally, the boys and their families need to be supported to make the most of opportunities in life and to be involved in normal family, educational, vocational, recreational and community activities.

Emotional issues at this time may include anxiety about impending death and the ability to cope.

Helping professionals need to acknowledge a family's anxiety and assure them of their continuing support or help them to establish a relationship with someone who can do this. Even when families avoid discussion of this issue, regular contact from a caring person has been found to be very reassuring.

Families require special consideration when their son dies, whether suddenly and unexpectedly as a result of a secondary illness or after a long decline. They will grieve again for this final loss just as other bereaved families do and will vary in their ways of expression of this grief. Helping professionals who have developed a caring relationship with the family will be aware of these needs and will be able to offer appropriate support.

Families may express ambivalent feelings such as relief and sadness, anger at the difficulties faced by their son and joy at the happy memories. These feelings should be acknowledged but not judged and helpers should avoid making statements like "it's for the best" or "he's better off". Affirmation of the value of the boy's life and the family's care for him during his life can be expressed through attendance at the funeral and some contact afterwards.

Families have sometimes felt 'abandoned' after their son's death when they lose contact with people (professionals) who have been intimately involved in their lives. Relatives and friends may also not be as available once the initial mourning period is over and it is well documented that continuing support for grieving families can help in their adjustment to their loss.

Siblings

The siblings of a boy with Duchenne need special mention as their relationship is often the longest relationship in the lives of their brother. It is therefore not surprising that growing up in a family with a sibling with a life-limiting condition is sometimes referred to as a challenging experience.

It is yet another concern for parents who are already suffering from the grief of a child's diagnosis with DMD and the stress associated with organising appropriate medical and therapy treatment. In fact, it has been shown that while their childhood experiences may be different from their peers, siblings growing up in a family with a sibling with a life-limiting condition often demonstrate real strengths in later life. They can be more mature, responsible, tolerant and altruistic, and they appreciate the importance of family. However, it is true that siblings have their own special needs and stresses.

Siblings experience a similar grief to their parents at the longer-term prognosis of their brother's condition. They sometimes experience "survivor" guilt as they increasingly realise they will be able to achieve what their sibling may not. At the same time they may feel jealous of the special attention their sibling receives from their parents, extended family and even doctors and therapists. They may find different behavioural expectations within the family hard to understand and accept. They may feel embarrassed by unwelcome attention to their family in public. They are often confused by their contradictory emotions but may feel unwilling to bother their parents whom they know are already stressed.

It is possible to manage these concerns in positive ways, and parents need to be reminded that all family relationships – with or without medical issues – have their good and not-so-good times.

Some suggestions include:

1. siblings need age-appropriate information about their brother's condition and language, which they can use to answer questions from friends and even members of the public
2. they should be included in discussions and plans for their brother's treatment and future as his future will affect theirs
3. feelings shouldn't be completely hidden by parents from their children. By letting children see that their parents can manage the tough times, they can learn to do the same
4. some individual time with each sibling is very important – even if this means taking them out of school for a few hours. This lets them know they are important to their parents, too
5. siblings should be encouraged to develop their own skills and interests. This is good for their sense of identity and confidence. They don't always want to be known as their brother's sibling
6. parents need to be careful about how much they expect siblings to be involved in personal care. Children often like to help but what type of help and how often this is expected should be sensitively discussed with them, especially as they grow older. Nobody likes to be taken for granted
7. all sibling relationships can be turbulent at times. It doesn't mean they don't care for each other. The child with DMD and their siblings should be allowed the benefit of sorting out some things between themselves
8. as parents' time may become limited, some thought could be given as to whether a relative or close friend could become a supportive mentor to siblings
9. siblings should be encouraged to attend sibling activities, which may be organised by service providers or parent groups. Contact with other siblings lets them know they are not the only ones and that what they are experiencing is normal for families with their challenges
10. in summary, siblings know their parents love and care for them. It sometimes happens that they need some explicit reassurance from them.

References and reading

In creating the content for this chapter, we have drawn heavily on the following publications and acknowledge these authors' contribution to our content.

1. Erby LH, Rushton C, Geller G." My Son is Still Walking" Stages of Receptivity to Discussions of Advance Care Planning Among Parents of Sons with Duchenne Muscular Dystrophy. Seminars in Pediatric Neurology 2006;13(2) 132-140.
2. Kenneson A, Bobo JK. The effect of caregiving in families with Duchenne/Becker muscular dystrophy. Health and Social Care in the Community 2010;18(5) 520-528.
3. Miller NB. Nobody's Perfect – Living and Growing with Children Who have Special Needs, Paul H Brookes Publishing, Baltimore; 1994.
4. Roos S. Chronic Sorrow: A Living Loss, Routledge, NY; 2002.
5. Thompson CE. Raising A Child with A Neuromuscular Disorder, Oxford University Press, USA; 1999
6. Read J, Kinali M, Muntoni F, Garralda ME. Psychosocial adjustment in siblings of young people with Duchenne muscular dystrophy. Journal of the European Paediatirc Neurology Society 2009;14,340-348
7. Strohm K. Siblings: Brothers and Sisters of Children with Special Needs, Wakefield Press, South Australia; 2002.

Chapter 12:

Recreation and leisure

The main aim of recreation and leisure is to provide opportunities that result in the participant experiencing 'flow'. Flow is a pleasurable experience that results when a person's skills and abilities are matched with a level of challenge in activities. This promotes success in an activity that, in turn, enhances a person's self-esteem.

Recreation workers are able to be involved with children throughout all the life stages. The main purposes during the early stages is to establish social networks, encourage physical activity that will aid later stages and create a pattern of leisure that can be maintained throughout the later life stages. The later stages involve maintaining leisure involvement through the adaptation of equipment and activities.

Early stage: preschool

During this stage, physical activity is encouraged to develop the child's balance, strength, coordination and awareness of body parts. Encouraging children to explore new experiences through sensory activities enhances the senses of touch, hearing and seeing.

Social networks are important for both the child and family as they provide an avenue for support and encourage physical interactions between children.

Activities:

- play with shaped sponges and colourful toys during bathtimes
- outside, aquatic activities can include using small pools or garden hose
- use play equipment, especially in public areas
- play games, e.g. identify this object/sound and role-playing
- try obstacle courses that encourage simple movements

- try to complete all activities with a warm-down element, e.g. lie down and pretend to be a lazy lizard
- try to discuss the child's feelings in regard to the activity and themselves.

NB: Adult supervision is recommended during these activities.

Early stage: early school years

Maintaining and developing social networks for both the child and family is important, as this will help enhance interpersonal skills and provide another form of support network outside the immediate family.

The type of activities should try to meet the interests of the child while encouraging activities that will allow the child to discover what they can and can't do. As this may result in some forms of stress, developing good relaxation techniques is encouraged.

During the early school years, preparing for later life stages is important. Identifying accessible and inclusive activities that can be continued through the other life stages is important. Adaptations to equipment and activities may be required during this stage.

If the boys are encouraged to stay engaged in a variety of social activities in earlier stages, they are more likely to stay socially active later.

Activities include:

- social groups and clubs, including cubs/brownies and remote control clubs
- those that facilitate the leisure goals of the child, with some adaptations as needed, such as using ramps for bowling, and being involved in ways other than being a competitor, such as umpiring
- activities to build coordination and movement such as chase and tag, catch, breathing and stretching exercises, aquatic exercises
- reading visualisations to help the child sleep, develop concentration and awaken creativity, e.g. *The Inner Garden* by Maureen Garth.

NB: During this stage children may begin to experience fatigue during activities, so planning for sufficient breaks and pacing activities is important.

Late ambulatory stage

This life stage is a period when many changes occur. Children may experience changes in their body and feelings. These changes may require adaptations and considerations to be made to leisure activities. Children may also begin to develop a need to be independent and challenge authority figures.

Being part of peer groups and having individual friendships is important to feeling socially accepted. Therefore, identifying accessible and inclusive leisure activities and venues that precipitate social interactions is encouraged.

Activities during this life stage should be age-appropriate, provide opportunities for children to practise social skills, encourage peer involvement and develop new skills and refine old skills.

Activities:

- continue activities that can be continued into the next stages, e.g. breathing and stretching exercises, aquatic exercises, social interaction and outings with friends, electric wheelchair sports, computer-related activities and remote control clubs
- attend camps that provide opportunities to mix with peers, discuss issues related to living with a disability, allow for the child to be responsible for their own possessions and provide activities that challenge their abilities
- join some disability-specific activities or groups, e.g. Sporting Wheelies.

Adaptations may be required to activities and equipment, e.g. may require bumper bar around footplates for wheelchair sports, or reducing the time in a game so that the fatigue effect is lessened.

Non-ambulatory stage

During this stage, muscle power diminishes although the hand and finger muscles remain relatively strong. This allows for the operation of an electric wheelchair by an individual. This level of functioning affects the type of leisure involvement a person has.

During this stage, the leisure activities introduced earlier with this stage in mind can be continued. Other passive-type leisure activities can take effect during this stage.

When the boys are very fatigued, they tend to restrict their outings and activities considerably (especially in the colder months). Young men who have non-invasive daytime ventilation are finding they are able to participate in activities outside the home more freely.

It is important to try to maintain all interpersonal relationships developed over the person's life and continue them through this stage.

Activities:

- maintaining involvement in activities begun in earlier stages, e.g. breathing and stretching exercises, aquatic exercises, social interaction and outings with friends, electric wheelchair sports, computer-related activities and remote control clubs
- attending camps that provide opportunities to mix with peers and discuss issues related to living with a disability allows the child to be responsible for their own possessions and provide activities that challenge their abilities
- maintaining involvement with disability-specific activities or groups, e.g. Sporting Wheelies.

Further adaptations may be required to activities and equipment, e.g. the boy may require a chest harness for electric wheelchair sports.

Chapter 13:
Research

In the recent years much research has been conducted worldwide on the many animal models of Duchenne (mdx mice and GRMD dogs are used most often). Successful trials in animals have paved the way for clinical trials to begin in boys.

To date, Phase 1 clinical trials have been completed in a number of molecular genetic therapies, some of which have progressed to Phase 2A and 2B trials.

The short overview of the main candidates has been summarised below from *Research Approaches to Duchenne Muscular Dystrophy* by Guenter Scheurbrandt, PhD for Treat NMD, PPMD and Action Duchenne. The full text can be found on the internet at www.duchenne-information.eu

A review of current clinical trials can be found on http://clinicaltrialsfeeds.org/clinical-trials/show/NCT01182324

Exon skipping

Exon skipping is a molecular therapeutic technique that involves removing extra exon or exons adjacent to the existing mutation of the gene in order to restore the reading frame, permit the recipe to be read and the production of sufficient functioning dystrophin to occur. It aims restore at least 30 per cent of dystrophin to muscle and thereby convert Duchenne into the milder Becker MD. It is not a cure but hopes to reduce the symptoms significantly and make a difference to longevity and quality of life of boys and young men with Duchenne.

Exon skipping is achieved using short pieces of manufactured genetic material called antisense oligonucleotides (AOs) which have specific chemical sequences to complement and attach to the desired exon/exons exactly. Because they are exon-specific, they will need individualised chemistry for all of the different mutations.

AOs are being manufactured in many laboratories around the world. Some labs claim to have AOs to match most of the 79 exons in the dystrophin gene, but a few genetic mistakes will not be suitable for exon skipping because of their critical position in relation to binding sites with other proteins.

Clinical trials are progressing with proof of principle trials successfully accomplished. Two different chemistries have now moved on to further trials. Both the AONs in trial are directed at skipping exon 51, which would confer an advantage to the greatest number of the mutations (approximately 13 per cent) on the databases worldwide. Preparations are being made for skipping other mutations once trials of exon 51 skipping are proven successful. Exon 44 trials are planned.

In the Netherlands, PRO-051 is being developed by GlaxoSmithKline PLC under license from Leiden University Medical Center and Prosensa Therapeutics BV. This oligonucleotide sequence induces skipping of exon 51 of the dystrophin gene by binding to a sequence within the dystrophin pre-mRNA and masking the exon inclusion signals that are used for splicing.

Removal of exon 51 from an exon 45 to 50, 47 to 50, 48 to 50, 49 to 50, 50, 52 or 52 to 63 deleted transcript allows restoration of the open reading frame and synthesis of an internally truncated, semi-functional dystrophin protein. Observable dystrophin expression has been most encouraging in the early trials. (At the time of publication, a phase I/IIa trial to evaluate subcutaneous delivery of PRO-051 had been completed, although full results were yet to be published.)

In the United Kingdom, clinical trials are also underway by the MDEX consortium and AVI Bio Pharma Inc under the direction of Prof Francesco Muntoni. The AON being trialled is the Morpholino AVI-4658. Promising levels of protein expression have been expressed and further trials are underway.[1] Antisense oligonucleotides inserted into U7snRNA/AAV vectors, has been shown to successfully restore expression of the dystrophin gene in cellular and animal models of DMD. The advantage of AAV based U7snRNA vectors is that they show more sustained expression than naked antisense oligonucleotides. Researchers in Dame Kay Davies lab (MRC functional Genomics Unit) are currently testing vectors that can skip multiple exons to cover many of the regions of the protein mutated in DMD patients.[2]

Challenges include:

- developing appropriate delivery mechanisms
- enhancing bio-availability and uptake
- discerning the effects of long-term treatment
- progressing to delivery of AON applicable to a range of other mutations.

(S. Wilton, personal communication *Rome 2011*)

Transfer of the dystrophin gene

This method aims to address the primary genetic and biochemical defect by delivering a high level of therapeutic dystrophin gene to every affected muscle without inducing an immune response. Vectors for transporting the corrected dystrophin gene being developed are both viral and non-viral vectors. Viral vectors are engineered (gutted of their own DNA) so they can carry the required DNA but cannot replicate themselves within the host body.

Viruses commonly used in gene therapy trials in the mdx mouse and dogs are the adeno-associated viruses (AAVs). These viruses are the smallest and arguably the safest viral vectors used for gene therapy to date as they do not provoke an immune response in the host and can be targeted to specific tissues, including non-dividing cells. They are, however, only able to carry 5000 base pairs with exons 18-59 and 70-79 deleted. The micro gene, which is only a third of the length of the full-length dystrophin, may be able to produce sufficient dystrophin levels to reduce the dystrophinopathy to Becker MD. Clinical trials of biostropin (Asklepius Bio Pharm) are being conducted by Jerry Mendell in Columbus, Ohio. An unexpected immune response in some of the trial participants is being investigated.[3]

Gene transfer with plasmids is also being studied and trialled as vectors for a stripped-down mini-dystrophin gene at the Functional Genetics Unit in Oxford. Some researchers are investigating delivery of plasmids under pressure for better uptake.[4] Plasmids are extra-chromosomal circular DNA molecules in bacteria that are used as vectors for cloned segments of DNA.

There are four major problems preventing successful gene transfer to date in DMD:

- the full-length dystrophin gene is so large that most viruses cannot transport it and it remains to be seen whether the mini and micro genes can confer enough functional improvement with their reduced dystrophin production.
- muscle tissue, especially mature muscle, is not readily attacked by commonly used viruses in gene therapy. As muscle represents 30 per cent of the body, the size of the target is a major problem. Viral vectors invade immature muscle cells (myoblasts, satellite, and stem cells) more readily.

- a systemic mode of delivery for the viral vector carrying the corrected gene is thought to be essential. Injection-based delivery has been shown to be extremely localised, without sufficient spread into neighbouring tissue. The unique structure of muscle fibres with their three layers of connective tissue is a contributory factor to reducing spread into neighbouring fibres.
- the immune system attacks the many viral vectors before they reach the target cells. It also thought that the immune system may attack those cells that have been invaded by the virus and, possibly, the introduced dystrophin which is not recognised as 'self'.

Myogenic cell transfer – the delivery of precursor muscle cells – in therapeutic numbers has, to date, been unsuccessful. However, researchers in Canada[5,6] are continuing this line of research on its own and in combination with myostatin inhibition.

The benefits of this approach and the reason for continuing this line of research are:
- the new dystrophin would be normal length
- the benefit would be long lasting
- it could be combined with pharmacological treatments
- it could benefit older boys and young men.

As with the other research strategies, there are many hurdles associated with myogenic cell transfer including a need for a systemic mode of delivery. At present, 100 injections per centimetre of muscle are required.

Stem cell research

The development of pluripotent human stem cells, which are capable of giving rise to most tissues lines, has given the tantalising promise of many new therapies including a possible cure for DMD. Pluripotent stem cells undergo further specialisation into multipotent stem cells that are committed to cells, which have a particular function, e.g. skin, bone, muscle or blood cells. Pluripotent stem cells have been harvested from the inner cell mass of human embryos made for in-vitro fertilisation purposes and from the foetal tissue of terminated pregnancies. Both methods have provoked enormous debate on ethical grounds over the years and this has led to the development of alternative techniques without the ethical dilemmas.

Donor cells that are procurable from living sources are being sought, thereby avoiding the ethical issues associated with the stem cells from foetal or embryonic tissue.

Results from many research projects into muscular dystrophy has increased understanding of the plasticity which exists in many adult stem cells, permitting muscle cells to be produced from different sources including blood, fat, bone marrow, skin and other tissues.

For effective muscle regeneration to occur using muscle stem cells, the cell lines should all:
- be easy to isolate from their tissue
- be able to multiply effectively in the laboratory
- be able to carry a viral vector transporting the healthy dystrophin gene sequences
- be able to be systemically delivered into the bloodstream
- be able to successfully migrate into muscle
- give rise to sufficient functioning muscle cells, complete with satellite cells within the dystrophic muscle tissue
- produce no side-effects, specifically the risk of producing cancer.

Two cell types have been identified that appear to fill the requirement:

Mesangioblasts, found outside the walls of small blood vessels within muscle, have been shown to restore muscle structure and function to a significant extent when injected into the bloodstream. Mesangioblasts can cross from the bloodstream into muscle tissue, diffusing throughout the body and integrating into existing muscle. This makes them ideal candidates to carry viral vectors with the corrected genetic material into the muscle. Some success in animal models has been achieved.

Pericytes located inside the capillaries in muscle have been targeted for trials at The Stem Cell Institute of the San Rafffaele hospital in Milan.[8, 9, 10] Dr Cossu's team progressed from the mouse model to treating four dystrophic dogs with pericytes carrying a microdystrophin gene. They also treated six dogs with pericytes from healthy dogs. Better results were obtained with the cells from healthy dogs (heterologous) than the corrected autologous cells. Further studies are being conducted, looking at transfer of a larger dystrophin molecule within the cells. After further animal trials, it is envisioned that human safety trials may be contemplated.

Genetically exon skipped cells: Research continues on stem cells carrying lentoviruses, which carry in their genes antisense oligonucleotides for skipping exon 51. Experimentation will continue in the mouse model.

Up-regulation of utrophin

Utrophin is an intra-cellular protein which is very similar to dystrophin in both its structure and function. The utrophin gene, spanning 75 exons, is located on chromosome 6 and its product, utrophin itself, is found mainly at the neuromuscular junction after birth. In foetal life it is more widespread, occurring with dystrophin on the inner side of the muscle membrane.

It is hypothesised that if up-regulation of utrophin were achieved, there would be amelioration of the muscle degradation and symptoms in boys with Duchenne. The initial work on utrophin carried out by Professor Dame Kaye Davies at Oxford University[6,11] is now being continued and expanded at biotech company Summit Corporation PLC under the direction of John Tinsley. A number of compounds have been identified by high-throughput screening as possible candidates to up-regulate utrophin. Testing on mice and zebra fish is continuing.

Myostatin inhibition

Myostatin is a naturally occurring protein that inhibits muscle growth. A mutation in the myostatin gene has been responsible for double-muscled cattle in Belgium and one child with myostatin inhibition has also been studied[12, 13.] Myostatin blockade has been shown in treatment of mdx mice to significantly increase muscle mass and to increase the tetanic force in muscles. However, treated mice showed diminished endurance on the treadmill. It has been hypothesised that a lack of myostatin results in a lack of oxidative fibres and a conversion of fibre types towards fast glycolitic fibres.[12]

In summary, it would appear that myostatin inhibition makes normal muscle bigger but they may not be stronger. It makes mdx muscle both bigger and stronger but the mice do appear to lack endurance and therefore cannot run as far.

Idebenone

Idebenone is a synthetic analogue of co-enzyme Q10, a powerful antioxidant. DMD is known to weaken and disrupt the muscle-cell membranes but it also thought to affect the mitochondria within the cytoplasm of the cells. Mitochondria are the cellular energy stores, and CoQ10 and idebenone are thought to facilitate cellular energy as well as being powerful antioxidants.[14] Santhera Pharmaceuticals, in collaboration with the University of Leuven, has been conducting randomised, double-blind, controlled studies to determine the effect of idebenone on heart and respiratory function. Promising results were recorded on functional cardiac test and an improved respiratory peak flow. Santhera is continuing the development of this drug.

Pharmacological research

Inducing a read-through of premature stop codons is being considered by Ataluren (PTC 124).

Between 13 and 15 per cent of boys with DMD have a nonsense mutation, which is a single point change resulting in the introduction of a premature stop codon into the dystrophin mRNA. This effectively shuts down the production of dystrophin beyond this point. Gentamycin, a common aminoglycoside antibiotic, had been found to allow cells to ignore an abnormal signal and to continue to make the protein in muscular dystrophy mice (mdx mice). Clinical trials (early 2000) tested its efficacy in a small group of children, however the doses required to be effective were likely to cause liver damage and hearing loss.[15]

Pharmaceutical company PTC Therapeutics developed a drug by automated screening of many small molecule compounds to determine their ability to achieve this same read-through of the nonsense mutations. The successful compound, initially called PTC124 and now called Ataluren, had the required potential. Extensive lab testing proved its effectiveness in cultured muscle cells and later the mdx mice trials showed production of full-length dystrophin.

Phase 1 clinical trials were conducted to assess safety and Phase 2A trials were conducted from December 2005 until May 2007 on 38 boys with DMD. Promising results led to the multi-centre, long-term, randomised controlled Phase 2B trials, enrolling boys who were over five and still walking. They were randomised into three groups: low dose, higher dose and placebo. The treatment continued for 48 weeks. At the end of 2009, open-label trials continued in all of the international centres that had been involved with the Phase 2B trials. All boys received the real drug Ataluren.

The initial results released in February 2010 did not uphold the highly anticipated promise. When reviewing the chosen endpoint result for improvement in function – the six-minute walk test – there was no statistically significant improvement in function for either dose as compared to the control group, although the lower dose did appear to have more effect. A review of all data is continuing, particularly looking at the results of the low-dose ataluren.

Sildenafil

Researchers at Kennedy Kreiger hospital in Baltimore are looking at sidenafil (revatio) and the possible benefits to cardiac function. Dystrophin is normally localised to the muscle cell membrane where it interacts with the dystrophin-associated glycoprotein complex including

neuronal nitric oxide synthase (nNOS). DMD gene mutations not only lead to the loss of dystrophin but also to mislocalisation, and reduced activity of, neuronal nitric oxide synthase. Researchers aim to prove the drug can lead to favorable cardiac remodelling and improved vascular tone.

The current (2010) study will be a phase 2, randomised, double-blind, placebo-controlled, single-centre study for six months, followed by open-label period of six months in which all enrolled subjects receive revatio (a PDE5 inhibitor). A single dose of revatio (20mg three times daily) will be tested based on the safety and efficacy of that dose for treatment of pulmonary hypertension.

The primary endpoint will be the change in cardiac left ventricular end-systolic volume (LVESV) as determined by cardiac MRI after six months of revatio, compared to baseline. A 10 per cent change in LVESV will be considered significant.

Aminophylline is another anti-inflammatory drug and is already in clinical use to treat asthma. A non-selective phosphodiesterase (PDE) inhibitor, it blocks the same biochemical pathway as sildenafil citrate and preliminary studies are under way. Other anti-inflammatory drugs are being considered.

Immune system

Investigation continues into ways to regulate the large contribution made by the immune system in the death of dystrophic muscle. Researchers aim to provide therapeutic intervention, which will control the added destructive effects on muscle by the cytotoxic T-lymphocytes, the helper T-lymphocytes and macrophages. It is possible that pharmacological control of specific immune cells could slow the rate of necrosis in dystrophic muscle.

Anti-inflammatory agents are being examined to discern whether any of the approved drugs could act to prevent the gradual build-up of scar tissue within muscle that is a normally occurring part of the normal wound-repair mechanism. Studies in mice have shown that some cytokines – molecules that promote inflammation and the development of fibrosis – accelerate the progression of the disease.

As muscle cells die and are constantly being replaced, the wound-repair mechanism causes an influx of fibroblasts to stabilise weak tissue and aid the healing. However, this gradual build-up of scar tissue in muscle also contributes to further loss of muscle function.

Research aiming to prevent this scarring in muscle, without inhibiting the production of collagen in other parts of the body where it is needed to maintain tissue strength, continues in the hope of preventing and reversing the damage. Some of the drugs of interest include Galectin-1, Remicade and Enbrel, all being used to combat arthritis and Anti-asialo GM1 – an antibody used in Parkinson's disease.

References

1. Aartsma-Rus A, Fokkema IF, Verschuuren J, Ginjaar I, Deutokom JCT, Van Ommen GJ, den Dunnen JT. Human Mutation 2009; 30:293-299.
2. Arechavala-Gomeza V, Graham IR, Popplewell LJ, et al & Muntoni F. (2007) 'Comparative Analysis of Antisense Oligonucleotide Sequences for Targeted Skipping of Exon 51 During Dystrophin Pre-mRNA Splicing in Human Muscle'. Human Gene Therapy, 2007:18. pp 798-810)
3. Goyenvalle A, Babbs A van Ommen G, Garcia L, and Davies KE. Enhanced exon-skipping induced by U7 snRNA carrying a splicing silencer sequence: Promising tool for DMD therapy. Mol Ther. 2009;7(7):1234-40
4. http://quest.mda.org/news/caution-immune-response-seen-dmd-gene-therapy
5. SebestyénM1, Budker VG2 Budker T2, . Subbotin VM1, Zhang2 G, Monahan SD, Lewis DL1, Wong SC1, Hagstrom JE1, Wolff JA. "Mechanism of plasmid delivery by hydrodynamic tail vein injection. I. Hepatocyte uptake of various molecules. The Journal of Gene Medicine 2006;8(7):852–873,
6. Nowak KJ, Davies KE. Duchenne muscular dystrophy and dystrophin: pathogenesis and opportunities for treatment. EMBO reports 2004;5(9):872–876
7. Fakhfakh R, Michaud A, Tremblay JP. Blocking the Myostatin Signal With a Dominant Negative Receptor Improves the Success of Human Myoblast Transplantation in Dystrophic Mice. Molecular Therapy 2011;19(1):204–210.
8. Cossu G, Sampaolesi M. New therapies for Duchenne muscular dystrophy: challenges, prospects and clinical trials. Trends Mol Med. 2007;13(12):520-6.
9. Cossu, G. and Tajbakhsh, S. Oriented Cell Divisions and Muscle Satellite Cell Heterogeneity. Cell 2007;129, 859-861.
10. Dellavalle, A, Sampaolesi, M, Tonlorenzi, R, Tagliafico, E, Sacchetti, B, Perani, L„ Innocenzi, B, Galvez, BG, Messina, G, Morosetti, R, Li, S, Belicchi, M, Peretti, G, Chamberlain, JS, Wright, WE, Torrente, Y, Ferrari, S, Bianco, P and Cossu, G.. Pericytes of human skeletal muscle are myogenic precursors distinct from satellite cells. Nature Cell Biol. 2007;9:255-67.
11. Miura P, Jasmin BJ. Utrophin upregulation for treating Duchenne or Becker muscular dystrophy: how close are we? Trends Mol Med 2006;Mar;12(3):122-9.
12. Wagner, Kathryn R. Muscle regeneration through myostatin inhibition. Current Opinion in Rheumatology: 2005;17(6):720-72
13. Amthora H, Ottoc A, Vulina A, Rochata A, Dumonceauxa J, Garciaa L, Mouisela E, Hourdéa C, Machariad R, Friedrichsb M, Relaixa R, Zammite PS, Matsakasc A, Patelc K, Partridgef T. Muscle hypertrophy driven by myostatin blockade does not require stem/precursor-cell activity. PNAS 2009;106(18):7479-7484
14. Buyse GM, Van der Mieren G, Erb M, D'hooge J, Herijgers P, Verbeken E, Jara A, Van Den Bergh A, Mertens L, Courdier-Fruh I, Barzaghi P, Meier T. Long-term blinded placebo-controlled study of SNT-MC17/idebenone in the dystrophin deficient mdx mouse: cardiac protection and improved exercise performance. Eur Heart J. January 2009;30(1):116–124.
15. Welch EM, Barton ER, Zhuo J. PTC targets genetic disorders caused by nonsense mutations. Nature 2007;447; 87-91)

Useful organisations and contacts for support

Montrose*Access*

(formerly Queensland Society for Crippled Children)
www.montrose.org.au

Corinda office
54 Consort Street
Corinda Qld 4075
Phone 07 3379 9200
Fax 07 3717 1146
Email: information@montroseaccess.org.au

Gold Coast office
Cnr Byth and Allied Drive
Arundel Qld 4214
Phone 07 5509 9300
Fax 07 5571 5879

Sunshine Coast office
Level 2, 20 Innovation Parkway
Birtinya Qld 4575
Phone 07 5439 1300
Fax 07 5437 8372

Strathpine office
3 Jockers St
Strathpine Qld 4500
Phone 07 3881 7900
Fax 07 3889 8979

MontroseAccess provides therapy, social work, recreation and respite services to children aged 0–18 years with physical disabilities and their families. In addition, respite is available for young adults.

Since 1995, Montrose has run twice-monthly neuromuscular clinics on site at Corinda. Paediatric neurologists from Royal Children's Hospital and Mater Children's Hospital attend and the full allied health team and orthotist see the children. MontroseAccess also runs a monthly wheelchair clinic.

MontroseAccess runs many programs focusing on all stages, from early childhood through to 'lifestyle and leisure' for young adults. Referrals can be made by families, medical and health professionals, teachers or other caregivers.

Areas covered by Montrose services include the Brisbane metropolitan area, Gold and Sunshine Coasts, Darling Downs, Burnett and Wide Bay and some areas in central, western and northern Queensland.

Duchenne Foundation (DF)

www.duchennefoundation.org.au

Formerly Parent Project Australia Inc, DF was incorporated in 2003 with the aim of improving the treatment, quality of life and long-term outlook for persons affected by DMD and BMD through research, education and advocacy.

DF is a member of the international network of United Parent Projects (www.uppmd.org). Parent Projects worldwide are committed to networking to find the best treatments and research avenues, lobbying and fundraising, gathering information and compiling consensus documents and multimedia educational materials.

DF has been the lead organisation lobbying to establish the Duchenne registry in Australia. DF has co-convened three conferences, in Brisbane, Toowoomba and Sydney, bringing together people living with neuromuscular conditions, researchers and clinicians from all over the world, as well as educators parents and all those involved in care and management of INMDs.

Muscular Dystrophy Queensland (MD Qld)

www.mdqld.org.au

191 Hedley Ave
Hendra Qld 4011
Phone 07 3607 1800
Fax 07 3607 1899

MD Qld assists people with various neuromuscular conditions by providing services and supporting research. Family support services can include counselling information, education, loan of equipment such as electric beds and wheelchairs, support to carers, emergency respite care and emotional crisis intervention.

Queensland Clinical Genetics Service

Phone 07 3636 1686

This service, based at the Royal Brisbane and Women's Hospital, provides diagnosis and counselling for people with an inherited condition in the family.

A referral must be made by a doctor.

Other centres for this service are at Mater Hospital (Brisbane), Cairns Base Hospital, Gold Coast Hospital, Queensland Health Molecular Genetics Laboratory, Toowoomba Base Hospital and Townsville General Hospital.

Clinics are also held in Ipswich, Bundaberg, Rockhampton, Mackay, Mt Isa, Sunshine Coast, Logan and Princess Alexandra Hospital (Brisbane).

Carers Queensland

www.carersqld.asn.au

Phone 07 3900 8100
Toll free phone 1800 242 636

The purpose of Carers Queensland is to improve the lives of carers. Services include counselling, advice, advocacy, education and training.

Centrelink

www.centrelink.gov.au

Centrelink provides financial assistance to families who have children with disabilities and to young adults with disabilities.

This assistance includes the Carer Allowance and Carer Payment, Assistance for Isolated Children, Disability Support Pension, Pensioner Education Supplement and Mobility Allowance.

Disability Services Queensland

www.disability.qld.gov.au

DSQ helps people with a disability and their families to access the support and services they need as they move through the different stages of their life.

Services include adult lifestyle support program, post-school services program, supporting families-family support program and local-area coordination.

Disability Information Service

Phone: Brisbane callers: 07 3224 8444
Toll free (outside Brisbane): 1800 177 120
TTY (telephone typewriter): 07 3896 3471
Fax: 3224 8037
Email: disabilityinfo@disability.qld.gov.au

This is a free, statewide information, resource and referral service for people with a disability, family, friends, carers, professionals and community organisations, etc.

The service provides information about all services – both government and community-based – in all areas.

Queensland Health

www.health.qld.gov.au

Queensland Health provides hospitals – inpatient, outpatient and emergency services – as well as community health centres, where services vary but may include domestic assistance, personal care, social support, physiotherapy, community nursing and social work.

Queensland Health's Home and Community Care program provides funds for services that support frail aged and younger disabled people to stay living at home. These services include domiciliary nursing (e.g. Blue Care), home care and respite care.

Contact your local community health centre for further information.

Mobility Aids Subsidy Scheme (MASS)

PO Box 281
Cannon Hill Qld 4170
Phone 07 3136 3524
Fax 07 3136 3525
Wheelchairs, modifications and other mobility equipment are funded through Queensland Health.

The therapist prescribes the equipment, following the guidelines as set out in the procedural manual.

Funding for daily living aids include:
- hoists
- pressure-reduction mattresses
- bath boards
- mobile toilet commodes.

Funding for incontinence aids includes:
- pads and nappies
- catheters
- drainage bags
- bottles and urinals
- gloves
- bed sheets.

Apply to the Mobility Aids Subsidy Scheme for assistance.

Home Assist Secure

Home Assist Secure provides assistance, advice and information on home repairs, maintenance, and minor modifications and security for homeowners. Locate your local provider through the Queensland Department of Housing.

Department of Communities – Housing and Homelessness Services

This Queensland government-owned business builds adaptable housing or modifies existing housing for eligible clients.

Occupational therapists are available for advice on housing solutions.

LifeTec Queensland

www.lifetec.org.au

Brisbane:
Newmarket Rd
Newmarket
Phone 07 3552 9000
Fax 07 3552 9088

Townsville
Domain Central
103 Duckworth St
Garbutt Qld 4814
Phone 07 4759 5600
mail@lifetec.org.au
LifeTec is a non-government, not-for-profit community service. It provides information and advice on equipment and resources for people with disabilities. Anyone can access LifeTec.

Visits can be arranged by appointment, with a fee applying for a professional consultation, and there is a phone-in service for information.

A mobile unit tours around Queensland.

Brisbane South Home Modifications Scheme

Princess Alexandra Hospital
Ipswich Rd
Woolloongabba Qld 4102
Phone 07 3240 2776
Fax 07 3240 2778

This service provides home modifications to people living in private dwellings. Modifications include bathroom, toilet, ramps, stairlifts, etc. A grant of 50 per cent toward major modifications is available.

NICAN

www.nican.com.au

Unit 5
48 Brookes St
Mitchell ACT 2911
Free call 1800 806 769
info@nican.com.au

NICAN provides information on recreation, tourism, sport and the arts for people with disabilities.

For domestic air travel in Australia NICAN administers the Qantas Carer Concession Card which is issued to people with a disability who need high level assistance in-flight. Cardholders receive 10% discount on full cost domestic economy class airfares and 50% discount for nominated carers. See www.nican.com.au

Queensland Transport

www.transport.qld.gov.au

Queensland Transport provides the following support for eligible people:

- students with disabilities—conveyance allowance
- disability parking scheme
- taxi subsidy.

Seating clinics – Rehabilitation Engineering Centre

Royal Brisbane Hospital
Ground floor, Coles Health Services Building
Royal Children's Hospital
Herston Road
Herston Qld 4029
Phone 07 3636 7773
Fax 07 3636 1785

The Rehabilitation Engineering Centre provides services in the following areas:

- assessment and prescription of wheeled mobility
- assessment, design and manufacture of customised seating
- seating interface pressure mapping and evaluation of cushions and mattresses
- powered wheelchair control interface development and driver skills training
- consultation service for customised assistive technology and rehabilitation engineering, including videoconference consultations for remote clients and care providers.

Referrals are accepted from medical officers, allied health professionals, patients and their families.

For general enquires or to receive a referral form, call 07 3636 7773 between 7.30am and 4.30pm, Monday to Friday.

Rehabilitation Technology Service

20 Keane St, Currajong
Townsville Qld 4814
Phone 07 4759 2030
Fax 07 4779 9914

Cootharinga Rehabilitation Technology Service assists people with a disability who have a need for repairs or modifications to wheelchairs or other mobility equipment.

The goal of RTS is to enhance individuals' lives by providing appropriate and timely modifications to equipment, primarily wheelchairs, which have been funded by Medical Aids Subsidy Scheme (MASS), self-funded or obtained from other parties.

This is a unique service.

Mobile seating assessments are carried out by the team at RTS from Cairns to Mackay and regions in between.

Advice on diet and nutrition

Specialist advice on nutrition can be obtained from a qualified dietitian/nutritionist, who can help you to provide a balanced, healthy diet.

Your paediatrician can refer you to a dietitian/nutritionist at the Royal Children's Hospital (phone 07 3636 3777), the Mater Children's Hospital (phone 07 3840 8195) or a dietitian/nutritionist in private practice.

Physiotherapy assessment chart

DUCHENNE MUSCULAR DYSTROPHY ASSESSMENT

Name _____

Date of birth _____

Date of diagnosis_ _

Date							
Clinics attended							
Medication							
Dose							
Surgery							
Equipment							
• AFOS							
• Manual wheelchair							
• Standing frame							
• Power chair							
• Hoist							
• Electric bed							
• BiPAP/VPAP							
• Incentive spirometer							
• Respiratory aids							
• Other							
Sleep pattern							
Sleep studies							
Pain							
Dietetic consult.							

FUNCTIONAL ASSESSMENT CHART

	TIME TO ACCOMPLISH					
Therapist						
Date						
1. Run 10 metres						
2. Walk 10 metres						
• independent—normal gait						
wide base						
Trendelenberg						
waddling						
lumbar lordosis						
heel strike						
plantargrade feet						
on toes						
pronated						
internal rotation						
external rotation						
Stance						
• symmetric						
• asymmetric (R) (L) prop						
3. Climb 8 stairs						
• independent—foot over foot						
• independent—one step at a time						
• hands on knees						
• railing—one hand						
• railing and hand on knee						
• railing—two hands						
4. Walk down stairs						
• independent—controlled						
uncontrolled						
• railing—one hand						
two hands						
• sideways holding rails						
5. Stand on left leg (timed)						
Stand on right leg (timed)						
Hop						
6. Rise from floor from prone position						
• independent through supine						
• independent through prone						
• Gower's sign						
• uses furniture						

Adapted from *"The Clinical Management of Muscles Disease",* Irwin M. Seigel 1977

FUNCTIONAL ASSESSMENT CHART (continued)

	TIME TO ACCOMPLISH					
Therapist						
Date						
7. Rise from chair • independent						
• Gower's sign						
• turn to side, then push up						
• pull up with aid of table						
8. Gets to all fours from prone						
9. Crawls—palms flat						
—palms raised						

10. Roll over from supine to prone (left)						
Roll over from supine to prone (right)						
Roll over from prone to supine (left)						
Roll over from prone to supine (right)						
11. Lifts head supine						
12. Supine to sitting (a) independent						
(b) pulls up on leg or clothing						
(c) pulls up and pushes on elbows						
(d) turns to side and pushes up						
(e) to hands and knees then sits up						

13. Sitting balance • stable						
• unstable						
14. Sitting posture • normal						
• reversed lumbar curve						
• scoliosis						
• convexity to left (L) or right (R)						
• hyperlordotic						
• kypholordotic						

FUNCTIONAL ASSESSMENT CHART (continued)

	TIME TO ACCOMPLISH					
Therapist						
Date						
15. Upper limb function						
• can abduct arms in a full circle until they touch above the head						
• raises arms above the head only by shortening the lever arm or using accessory muscles						
• cannot raise hands above the head, but can raise a 180 ml cup of water to mouth, using both hands, if necessary						
• can raise hands to mouth, but cannot raise a 180 ml cup of water to mouth						
• cannot raise hands to mouth, but can use hands to hold a pen or pick up a coin						
• cannot raise hands to mouth and has no functional use of hands						

16. Height—standing or arm span						
17. Weight						

18. Respiratory function																		
	Predicted						Measured						%					
FVC																		
VC																		
FEV1																		

19. Swimming						
• blows bubbles						
• immerses face in water						
• independent in prone						
• independent in supine						
• floats independently						
• uses floatation devices						
• independent vertically						
• water safe						

LEFT						RANGE OF MOVEMENT						RIGHT
Therapist												
Date												
						Hip • extension						
						• adduction						
						Popliteal angle						
						Rectus femoris length						
						Knee • extension						
						Ankle • dorsiflexion						
						• eversion						
						Shoulder • flexion						
						Elbow • extension						
						Forearm • supination						
						Wrist • extension						
						Long finger flexors						
						Neck flexion						

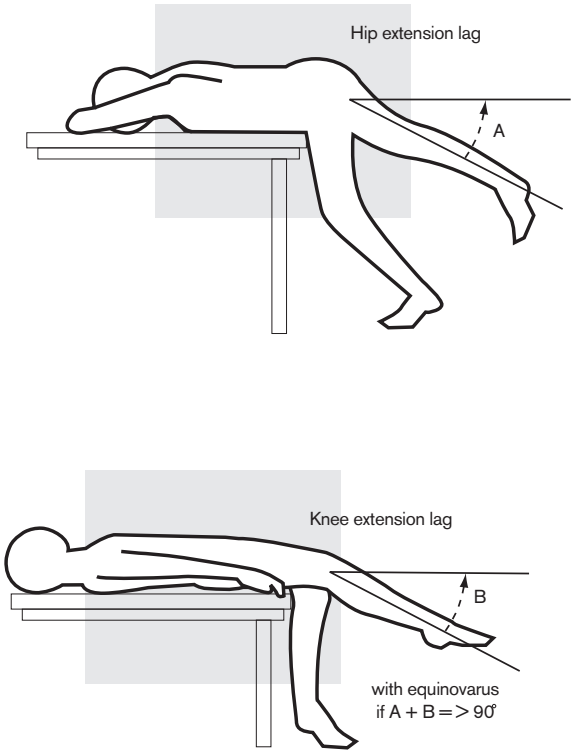
MANUAL MUSCLE TEST

						Shoulder • abduction						
						• flexion						
						• extension						
						• protraction						
						Elbow • flexion						
						• extension						
						Wrist • extension						
						Neck flexors						

MANUAL MUSCLE TEST (continued)

LEFT						RANGE OF MOVEMENT	RIGHT					
						Abdominals						
						Hip • abduction						
						• flexion						
						• extension						
						Ankle • dorsiflexion						
						• eversion						

Assessment of hip and knee extension lag



Ambulation ends
When the sum of knee and hip extension lag reaches 90 degrees, ambulation ends. (M. Siegel, *The Clinical Management of Muscle Diseases*. London, William Heinemann Medical Books, 1977)

Hip						
Knee						
Combined						

EXPLANATION OF TESTING PROCEDURES

Functional Assessment Chart

ACTIVITY—timed (1 through 10) and observed (11)

1. **Run 10 metres**
   Both feet off the ground at one time.

2. **Walk 10 metres**
   Independent—no assistance from mechanical aids.

3. **Climb 8 stairs**
   Stairs measurements:depth—29·5 cm; height 17·5cm; railing height 91·5 cm.
   (a) Independent—foot over foot—no assistance such as pushing on knees, using rail.
   (b) Independent—one step at a time.
   (c) Hands on knees assistance from rail.
   (d) Railing—one hand.
   (e) Railing and hand on knee—child pulls on rail with one hand, pushes on knee with other.
   (f) Railing—two hands, some boys may use two rails.

4. **Walk down stairs**
   (a) Independent—no assistance from rail. May go foot over foot or one step at a time.
   (b) Railing—one hand, often used for safety but not for support.
   (c) Railing—two hands or rail and therapist's hand for safety.

6. **Rise from floor from prone position**
   (a) Independent—no assistance required.
   (b) Gower's sign—can bring himself to his feet but must push on knees to assume erect posture.
   (c) Chair to standing—child pulls himself to feet with aid of chair, then pushes on chair to achieve upright position.
   (d) Chair to sitting, then to standing—child pulls himself to sitting position in the chair, then pushes himself to upright position using chair. Child should be placed on edge of table with thighs fully supported and knees flexed at 90°.

7. **Rise from chair**
   Care must be taken to seat the child in a chair which places his feet flat on the floor and his knees in a 90° flexion. This is particularly important with children as a higher chair would give them a mechanical advantage.
   (a) Independent—rising without pushing on chair or knees, arms folded across chest or extended.
   (b) Gower's sign—includes pressure on knees or pressure on seat of chair or both.
   (c) Turn to side, then push up—child turns sideways in chair to sit on one hip, with feet on floor pushes with arms to 90° hip flexion and pushes off to upright position or climbs up to chair to upright position.
   (d) Pull up with aid of table—child takes support from table with hip flexed while extending knees. Once knees are locked child extends hips by pushing up on table.

10. Roll over from supine to prone contractures may inhibit this act.
    Roll over from prone to supine contractures may inhibit this act.
    Get to all fours position from prone—push up to hands and knees.

12. **Sit up from supine**
    (a) Independent—no aids other than having ankles held down by therapist which is within the range of normal: must achieve sitting balance.
    (b) Pull up—child holds on to leg or clothing to pull himself up.
    (c) Pull and push on elbow—begins to pull up then pushes on at least one elbow.
    (d) Turn to side, then push up—child will roll to side then push up with both arms to achieve sitting balance.
    (e) To hands and knees, then sit up—roll to prone, to hands and knees, then to side or other sitting balance.

13. **Sitting balance**
    (a) Stable.
    (b) Unstable—child sits erect but cannot recover balance if it is lost. Or child is unable to maintain his balance without support of his arms.

DUCHENNE MUSCULAR DYSTROPHY ASSESSMENT

Modified Physiotherapy Assessment (Ambulatory)

Date of Ax ___/___/_____

Name	DOB	URN
Date of Dx	Height	Weight

Steroids	Type	
	Dose	
	Regime	

Respiratory Function

	Date ___/___/___ (most recent)			Date ___/___/___ (prior)			Date ___/___/___ (prior)		
	Predicted	Measured	Percent	Predicted	Measured	Percent	Predicted	Measured	Percent
VC									
FVC									
FEV1									

Timed 10m (Standing start, "as fast as you can go", best of 2 trials) _____

Gait
- ☐ Normal
- ☐ Wide base
- ☐ Waddling / lateral sway
- ☐ Bilateral Trendelenberg

Posture
- ☐ Lumbar lordosis
- ☐ Pronated feet
- ☐ Leg internal rotation
- ☐ Leg external rotation

Foot fall
- ☐ Heel strike
- ☐ Plantar-grade
- ☐ On toes

Timed Supine to Standing _____
- ☐ Independent through supine
- ☐ Independent through prone
- ☐ Uses furniture
- ☐ Gower's sign – 1 hand
- ☐ Gower's sign – 2 hands
- ☐ Gower's sign – 2 hands / laboured

Time to Descend 6 Stairs _____
- ☐ Controlled
- ☐ Uncontrolled
- ☐ Foot over foot
- ☐ One step at a time
- ☐ Railing – 1 hand
- ☐ Railing – 2 hands (sideways)

Time to Ascend 6 Stairs _____
- ☐ Foot over foot
- ☐ One step at a time
- ☐ Hands on knees
- ☐ Railing – 1 hand
- ☐ Railing – 2 hands
- ☐ Railing and hand on knee

Timed One Leg Stance Right _____ Left _____

Able to Jump ☐ Yes ☐ No

Number of Hops Right _____ Left _____

DUCHENNE MUSCULAR DYSTROPHY ASSESSMENT

Modified Physiotherapy Assessment (Non Ambulatory)

Date of Ax ___/___/_____

Name	DOB	URN
Date of Dx	Height	Weight

Steroids	Type	
	Dose	
	Regime	

Respiratory Function

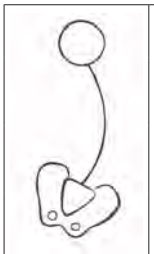
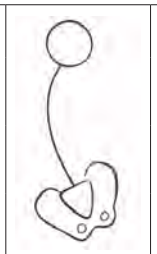
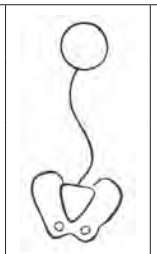
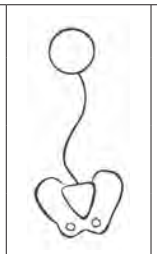
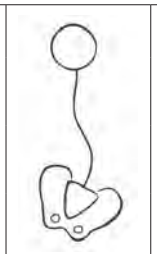
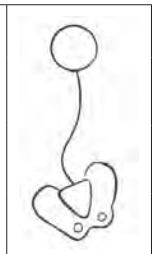
	Date ___/___/___ (most recent)			Date ___/___/___ (prior)			Date ___/___/___ (prior)		
	Predicted	Measured	Percent	Predicted	Measured	Percent	Predicted	Measured	Percent
VC									
FVC									
FEV1									

Upper Limb Function

- ☐ Can abduct arms in full circle until they touch (elbows extended)
- ☐ Can abduct arms above head but shortens the lever arm (elbows flexed)
- ☐ Has antigravity elbow flexion
- ☐ Cannot raise arms above head but can raise a glass of water to mouth using both hands if necessary
- ☐ Cannot raise hands to mouth but can use trick movement
- ☐ Cannot raise hands but can use hands to hold a pen or pick up a coin
- ☐ No antigravity power but has functional fine motor use (eg typing)
- ☐ Has little functional use of hands

Posture

- ☐ Normal
- ☐ Reversal of lumbar curve
- ☐ Lumbar kyphosis
- ☐ Thoracic kyphosis
- ☐ Hyperlordosis
- ☐ Scoliosis ➔➔➔
- ☐ Kyphoscoliosis

					
Thoraco-lumbar scoliosis – convex to the right	Thoraco-lumbar scoliosis – convex to the left	Double Major-Left thoracic, right lumbar	Double Major-Right thoracic, left lumbar	Right Lumbar	Left Lumbar

Power Wheelchair Modifications

- ☐ Headrest
- ☐ Standard Contoured backrest
- ☐ "I" Back
- ☐ Custom backrest
- ☐ Lateral supports x 2
- ☐ Inset armrests
- ☐ Extended armrests (adductor pads)
- ☐ Power tilt-in-space
- ☐ Pressure relieving cushion

  Type _____
- ☐ Harness
- ☐ Elevating legrests (power / manual)
- ☐ Spec switches
- ☐ Other _____
- ☐ Other _____

NORTH STAR PROJECT
Physiotherapy Test Detail

User Manual
Version 1.2

(Updated following Workshop 9th & 10th March 2005)

General Test Issues

1. **'Inclusion criteria'** - Children for the North Star project will have a definitive diagnosis of DMD. Children should be ambulant (minimum of 10m unaided) at time of initial assessment. The target age group to be assessed are primarily 4-8 year olds; however younger children or those who are older and still ambulant may be assessed using this protocol for inclusion in the database.
2. **Who can assess for the North Star database?** – wherever possible senior physiotherapists in permanent positions who have attended the North Star Workshop sessions and who specialise in the assessment and treatment of neuromuscular disorders should assess for the database. Where this is not possible assessments from other therapists will be considered on an individual basis. The project coordinator is happy to offer further input in teaching test method to new staff as needed. All staff involved with assessment should be available for informal reliability 'assessments' which will be done individually and on site.
3. Wherever possible the same **physiotherapist should assess and reassess** the patients. Intra-rater reliability has generally shown to be better than inter-rater for the tests. This also gives continuity to the patient and his family/carers.
4. **Do I have to do all tests?** Wherever possible we are asking centres to undertake all tests. Where resources – in particular therapist time – are an issue we ask that the functional testing (North Star Ambulatory scale and timed tests), joint ROM and respiratory testing are completed.
5. **Order of tests** – this is a debatable point. Ideally all centres would follow the same test order – function, joint range, respiratory tests then muscle test, as fatigue can be an issue with these children. However the practicalities of busy clinic settings make this unfeasible. It was also generally considered more important by the group to maintain a child's interest and avoid fatigue in order to get as 'true' a measure of ability as possible. This realistically means that most therapists will tend to do, e.g., all activities in sitting, then lying etc.
6. **Need for measurement rigour** – please be as exact as possible with measurement techniques.
7. Please do not examine previous results prior to reassessing a patient as this is likely to bias your current assessment.
8. Where gaining and maintaining **compliance** is an issue, therapists are asked to make a value judgment as to whether test results give a true value of the patient's ability. Where it is felt that data is poor this should be clearly noted on the assessment sheet. Distractions should be kept to a minimum wherever possible during testing.
9. **Appropriately clothed** – shorts/t-shirts or tracksuits to allow for testing. This is especially important for assessing joint range of movement and MMT. Shoes should be worn for the timed walk tests, but not for MMT and measurement of joint ROM. They may be worn or not for other tests but please be consistent.

Functional and Timed Tests

This document includes detail of the functional and timed tests for the project.

General Method

- Explain the tests to the child prior to undertaking them. E.g. show them the start and finish lines for the timed walk test, demonstrate an action if needed.
- Timed tests should be done in conjunction with the NSAA.
- The EK scale is for use with non-ambulant children
- Use a stopwatch for timed tests.
- Tests are generally completed from easiest to most difficult
- Where compliance is an issue note this. If so much so that a 'true' test score is not gained, results should not be included in the North Star database. If lack of compliance is due to patient's inability to complete the test, i.e. patient starts well but compliance is an issue with more difficult test components, a value judgment must be made on the part of the therapist as to score.
- Two assessment forms are included for the functional scale testing; one includes the Hammersmith Motor Ability Scale. Those who will be collecting information on both scales can use this form.

1. Timed tests

10 metre timed walk/run test

The patient should wear shoes for this test. If he usually wears orthoses but is capable of walking without them, testing can be done, as he prefers, with or without orthoses. The use of any aids/orthoses should be marked on the assessment sheet.

A 10m measured walkway is marked in the physiotherapy department or hallway. The patient is shown start and finish lines asked to traverse this as ***quickly*** as he safely can. He should not be asked to run – but rather to get there as quickly as he can, the choice is then his. 'Prime' him by saying ***'ready, steady, GO'.*** Time is recorded with a stopwatch from when his first foot crosses the start line to when the second foot crosses the finish line. If wall is touched, note how often.

NB Care needs to be taken to ensure that the patient is safe when completing this test. The assessor can walk nearby to provide 'emergency' help should it be needed, but must not support or provide manual assistance for the patient in any way.

Timed rising from floor

The patient starts in supine with his arms by his side. He is told to stand up as quickly as possible when the command "GO" is given. Time is recorded with a stopwatch from the initiation of movement until the assumption of upright standing. The area should be free from furniture and the patient should not be wearing orthoses or using any aids.

2. North Star Ambulatory Assessment

We have attempted to give clear explanations of the possible methods employed to achieve motor goals, but it is not possible to be exhaustive in the descriptions, particularly of modifications to activity. Whilst DMD children may generally present with recognizable adaptations to activity due to the underlying progressive muscular weakness, they may modify their activity to achieve functional goals in slightly differing ways. Generally, activities are graded in the following manner:

2 – Normal – no modification of activity

1 – Modified method but achieves goal independent of physical assistance from another

0 – Unable to achieve independently

It would be useful to have the method of achieving the goal briefly noted in the comments section whenever activity is modified.

We are asking that you further grade rising from the floor:

Definition of Gowers' manoeuvre:

The child turns towards the floor (generally into a four-point kneeling position) to place hands on the floor to assist rising, walks hands back in towards him then uses arms to 'climb' up legs to achieve upright standing. A wide base of support is often assumed through the phases of rising from the floor.

The following method of 'grading' rise from floor should be used in conjunction with the NSAA – please note in the comments section alongside the scale:

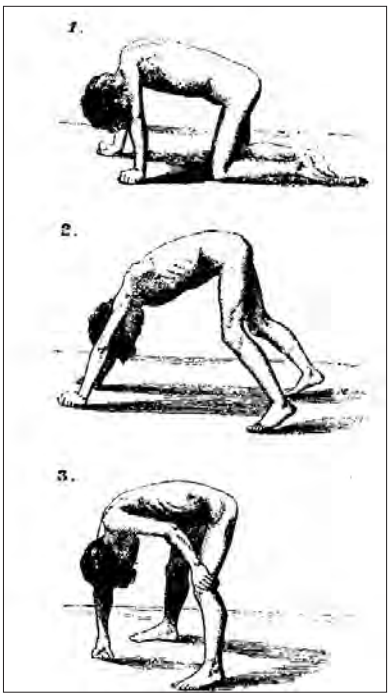


Figure: Gowers' Manoeuvre (from W.R. Gowers' Pseudohypertrophic muscular paralysis, 1879)

2	No evidence of Gowers' manoeuvre
1A	Turns towards the floor and places hand/s on floor to start to rise, does not need to place hands on legs
1B	Turns towards the floor and places hand/s on floor to start to rise, one hand on leg
1C	Turns towards the floor and places hand/s on floor to start to rise, two hands on legs
0D	*HAS* to use furniture
0E	Unble

Stair Climb

As it is not possible to ensure standardization of flights of stairs across the country, we are asking that a box step (approximately 15cm high) is used to assess step climb and descend. A plinth or other immovable object may need to be available to provide support. However, if a flight of steps is available to you please use these, and gain subjective information from child/parents as to how they manage stairs at home. Note this information in the comments section of the data collection sheet.

North Star Ambulatory Assessment

	Activity	2	1	0	Comments
1	**Stand**	Stands upright, still and symmetrically, without compensation (with heels flat and legs in neutral) for minimum count of 3	Stands still but with some degree of compensation (e.g. on toes or with legs abducted or with bottom stuck out) for minimum count of 3	Cannot stand still or independently, needs support (even minimal)	
2	**Walk**	Walks with heel-toe or flat-footed gait pattern	Persistent or habitual toe walker, unable to heel-toe consistently	Loss of independent ambulation – may use KAFO's or walk short distances with assistance	A value judgement needs to be made – if the patient generally toe walks but occasionally gets heels flat, or can on request but doesn't usually, they should score 1
3	**Stand from chair sitting**	Keeping arms folded Starting position 90° hips and knees, feet on floor/ supported on a box step.	With help from thighs or push on chair or prone turn	Unable	Feet should be supported on the floor or on a box step in starting position
4&5	**Stand on one leg R&L**	Able to stand in a relaxed manner (no fixation) for count of 3	Stands but either momentarily or needs a lot of fixation e.g. by knees tightly adducted or other trick	Unable	
6&7	**Climb box step R&L**	Faces step – no support needed	Goes up sideways or needs support	Unable	Support may be provided by the use of a height adjustable plinth, or, if not available a 'neutral' hand from the therapist.
8&9	**Descend box step R&L**	Faces forward, climbs down controlling weight bearing leg. No support needed	Sideways, skips down or needs support	Unable	Support may be provided by the use of a height adjustable plinth, or, if not available a 'neutral' hand from the therapist.
10	**Gets to sitting**	Starts in supine – may use one hand to assist	Self assistance e.g. – pulls on legs or uses head-on-hands or head flexed to floor	Unable	If patient turns into prone or towards the floor to work their way into sitting 1 should be scored
11	**Rise from floor***	From supine – no evidence of Gowers' manoeuvre	Gowers' evident	NEEDS to use external support object e.g. chair OR Unable	This item is scored on the 6-point scale detailed below
12	**Lifts head**	In supine, head must be lifted in mid-line. Chin moves towards chest	Head is lifted but through side flexion or with no neck flexion	Unable	Ask patient to keep arms crossed over chest during the activity to avoid self-assist. Also ask to look at toes to ensure neck is flexed 'lift your head and look at your toes'
13	**Stands on heels**	Both feet at the same time, clearly standing on heels only (acceptable to move a few steps to keep balance) for count of 3	Flexes hip and only raises forefoot	Unable	Watch for eversion. If substantial eversion but forefeet are still lifted – score 1. If only eversion with lateral border of foot still on the ground score 0.
14	**Jump**	Both feet at the same time, clear the ground simultaneously	One foot after the other (skip)	Unable	Instruction to patient 'how high can you jump'. Want height, not forward movement. Small amount of forward movement acceptable
15&16	**Hop R&L**	Clears forefoot and heel off floor	Able bend knee and raise heel, no floor clearance	Unable	Needs obvious floor clearance to score 2
17	**Run (10m)**	Both feet off the ground (no double stance phase during running)	'Duchenne jog'	Walk	'Duchenne jog' - not a true run (there probably IS a double support phase), but more than a walk. Typically characterized by excessive use of arms, trunk rotation, substantial 'waddle'. No real 'push-off'

* see definition above

Ambulatory Assessment and timed functional testing

Patient ID:

Date of assessment:

Assessor name (print):

Assessor name (signature):

	Activity	**Score 2,1,0**	**Time (s) (00.0s)**	**Comments**
1	Stand			
2	Walk (10m)			
3	Sit to stand from chair			
4	Stand on one leg - R			
5	Stand on one leg – L			
6	Climb step - R			
7	Climb step – L			
8	Descend step - R			
9	Descend step – L			
10	Gets to sitting			
11	Rise from floor			
12	Lifts head			
13	Stand on heels			
14	Jump			
15	Hop – R			
16	Hop - L			
17	Run			
	TOTAL (out of 34)			

General comments

Patient ID: **Date:** **Assessed by:** **Print**

	Hammersmith Motor Ability Scale	Score 2,1,0		North Star Ambulatory Assessment	Score 2,1,0	Time (00.0s)	Comments
1	Lifts Head		1	Stand			
2	Supine to prone over R		2	Walk (10m)			
3	Supine to prone over L		3	Sit to stand from chair			
4	Prone to supine over R		4	Stand on one leg - R			
5	Prone to supine over L		5	Stand on one leg – L			
6	Gets to sitting		6	Climb stair - R			
7	Sitting		7	Climb stair – L			
8	Rise from floor		8	Descend stair - R			
9	Stand		9	Descend stair – L			
10	Sit to stand from chair		10	Gets to sitting			
11	Stand on one leg - R		11	Rise from floor			
12	Stand on one leg – L		12	Lifts head			
13	Climb stair - R		13	Stands on heels			
14	Climb stair – L		14	Jump			
15	Descend stair - R		15	Hop – R			
16	Descend stair – L		16	Hop - L			
17	Stand on heels		17	Run			
18	Stand on toes						
19	Hop – R						
20	Hop - L						
	TOTAL HAMA (out of 40)			**TOTAL NSAA** (out of 34)			

General comments

3. The EK SCALE

The scale is comprised of 10 categories relating to the overall physical functional ability of non-ambulant subjects.

Each of the ten categories consists of four items (0-3) and the EK sum is the sum of the scores over categories.

Administration of the scale

The individual and his carer are questioned as to how each task is performed in daily life. The items are scored according to the explanation given and observation of the performance.

Category 10 focuses on respiratory insufficiency and the descriptions of items 0-3 are used for questioning and scoring. For instance: do you need to rest during the day? Do you sleep well at night? How is your appetite?

The administration of the EK scale consists of a question to the individual and his carer. Items are scored according to the explanation given and observation of the performance (to give a score out of 30).

1. How do you drive your wheelchair (items 0-3)? Please show me how you do it (items 0-1).
2. How do you transfer from the wheelchair (items 0-3)? Please show me how you do it (items 0-1).
3. Do you stand up (items 0-3)? How do you stand? Please show me or explain to me how you do it (items 0-2).
4. Do you change position in the wheelchair (items 0-3)? Please show me how much you can lean forwards, to the sides and get back to the upright position (items 0-2).
5. Can you move your fingers, hands and arms against gravity (items 0-3)? Please show me how you do it (items 0-3).
6. How do you feed yourself (items 0-3)? Please show me or explain to me how you do it (items 0-2).
7. How do you turn in bed at night (items 0-3)? Please explain to me how you do it (items 0-1) and how often (items 2-3).
8. What do you do to produce the most effective cough (items 0-3)? Please show and explain to me how you do it (items 0-3).
9. Do you speak loudly and clearly enough to make people understand you at the other end of the classroom (items 0-3)?
10. How is your general health (items 0-3)? This category focuses on symptoms of respiratory insufficiency and the descriptions of the items from 0-3 are used for questioning and scoring e.g. do you need to rest during the day? Do you sleep well at night? How is your appetite?

The EK Scale

1 Ability to use wheelchair

0 Able to use a manual wheelchair on flat ground, 10 metres in less than one minute
1 Able to use a manual wheelchair on flat ground, 10 metres in more than one minute
2 Unable to use a manual wheelchair, requires electric wheelchair
3 Uses electric wheelchair, but occasionally has difficulty in steering

2 Ability to transfer from a wheelchair

0 Able to transfer from a wheelchair without help
1 Able to transfer independently from a wheelchair with use of an aid
2 Needs assistance to transfer with or without additional aids (e.g. sliding board, easiglide)
3 Needs to be lifted with support of head when transferring from wheelchair

3 Ability to stand

0 Able to stand with knees supported, as when using braces
1 Able to stand with knees and hips supported, as when using standing aids
2 Able to stand with full body support
3 Unable to be stood, marked contractures

4 Ability to balance in wheelchair

0 Able to push himself upright from complete forward flexion by pushing up with hands
1 Able to move the upper part of the body more than 30 degrees from the upright position in all directions, but cannot push himself upright from the total forward flexed position
2 Able to move the upper part of the body less than 30° from one side to the other
3 Unable to change position of the upper part of the body, cannot sit without total support of trunk and head

5 Ability to move arms

0 Able to raise arms above head with or without compensatory movements
1 Unable to lift arms above head, but able to raise forearms against gravity i.e. hand to mouth with or without elbow support
2 Unable to lift arms against gravity, but able to use hands against gravity when the forearm is supported
3 Unable to move hands against gravity but able to use fingers

6 Ability to use hands and arms for eating

0 Able to cut meat into pieces and eat with a fork and spoon. Can lift a filled cup (approx 250ml) to the mouth without support at the elbow
1 Eats and drinks with support at the elbow
2 Eats and drinks with elbow support and with reinforcement of the opposite hand +/- feeding aids
3 Has to be fed

7 Ability to turn in bed

0 Able to turn himself with bedclothes
1 Able to turn himself on a couch, but not in bed
2 Unable to turn himself in bed. Has to be turned 3 times or less during the night
3 Unable to turn himself in bed. Has to be turned 4 times or more during the night

8 Ability to cough

0 Able to cough effectively
1 Has difficulty coughing and sometimes needs manual reinforcement. Able to clear throat
2 Always needs help coughing. Only possible to cough in certain positions
3 Unable to cough. Needs suction and/or hyperventilation techniques or IPPB in order to keep airways clear

9 Ability to speak

0 Powerful speech. Able to sing and speak loudly
1 Speaks normally, but cannot raise his voice.
2 Speaks with a quiet voice and needs a breath after 3-5 words
3 Speech is difficult to understand, except to close relatives

10 Physical well-being

0 No complaints, feels good
1 Easily tires. Has difficulty resting in a chair or in bed.
2 Has loss of weight, loss of appetite. Scared of falling asleep at night, sleeps badly
3 Experiences additional symptoms such as: change of mood, stomach ache, palpitations, perspiring

EK Scale for non-ambulant patients

Patient ID:

Date of assessment:

Category	**Score 0-3**	**Comments**
1. Wheelchair use		
2. Transfer from wheelchair		
3. Standing		
4. Balance in wheelchair		
5. Arm movement		
6. Eating		
7. Turning in bed		
8. Cough		
9. Speech		
10. Physical wellbeing		
TOTAL (out of 30)		

General comments:

Assessor name (print):

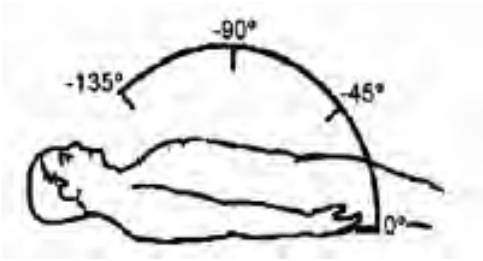
Assessor name (signature):

Joint Range Of Movement

General Issues

1. Wherever possible the same therapist should assess and reassess the patient as intra-rater reliability has been found to be better than inter-rater (Pandya et al, 1985).
2. ROM should be recorded to the nearest 5º.
3. **If possible** 2 people should assess ROM – one to hold the limb, one to position the goniometer. The patient's parents or a carer can give support if needed
4. Use an appropriately sized goniometer
5. Passive range is being measured so the patient needs to relax whilst the therapist ranges the joint.
6. Measurements should be taken with a 'moderate' degree of stretch applied. Variations in manual pressure applied have been identified as one of the key factors in inter-rater variability when passive ROM is being monitored (Gajdosik & Bohannon, 1987).
7. The mean range of 'normal' joint movement, as reported by the American Academy of Orthopaedic Surgeons, is noted for each measure.
8. Normal end-feel of each of the ranges is also noted for information (Norkin & White, 1995). These authors describe these as 'soft – due to soft tissue opposition, firm – due to muscle, capsule or ligamentous tension, or hard – due to bony opposition. Changes in this may be predictors of incipient contractures and may want to be noted as 'comments' in notes.
9. If needed use a towel under the upper arm to gain full range of elbow extension, or under the calf to gain full knee extension. This will also allow for easier alignment of the goniometer.

1. Elbow extension



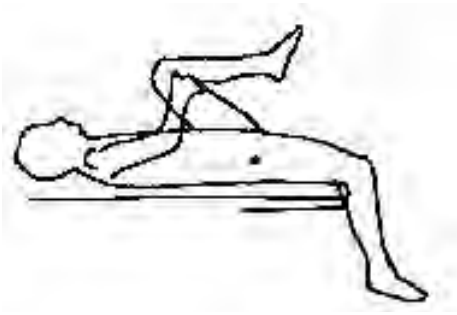
Patient position: supine with arm resting by side, forearm supinated.

Goniometer position: place centre of goniometer at lateral epicondyle. Align stationary arm with greater tubercle, parallel to lateral midline of humerus. "Moving arm" is aligned with radial styloid, along forearm. Measurement is recorded as degrees lacking from neutral.

Normal range: 0-150º of movement in the saggital plane

End feel: bony due to olecranon process of ulna in olecranon fossa of humerus.

2. Hip extension



Patient position: supine with the plinth supporting trunk and thigh, knee flexed over end of plinth. Flex contralateral hip and knee towards chest to fixate lumbar spine and stabilise pelvis (approximately 80º - NOT so much that the buttocks start to lift from the bench). The leg to be examined is lowered whilst pressure on the opposite thigh is maintained. Measurement is taken when resistance is felt or the pelvis becomes unstable. Foot must not rest on the floor. To measure range of extension beyond this, a subjective measure can be gained in side lying, or in prone.

Goniometer position: place axis of goniometer at greater trochanter. Align stationary arm parallel with plinth, along the side of the trunk. Align "moving arm" with lateral epicondyle, parallel with lateral femur. Note range of extension past neutral as +xº's, range lacking from neutral as -xº's.

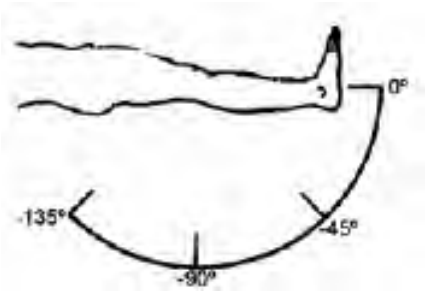
NB external rotation and abduction of leg being examined must be prevented

*Normal range: 1*0º of extension beyond neutral to 120º of flexion.

End feel: usually firm due to tension in the anterior joint capsule, the iliofemoral ligaments and possibly the hip flexor muscles.

3. Knee extension

Patient position: supine on plinth/floor.



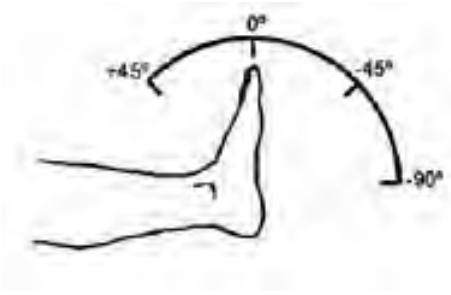
Goniometer position: place axis of goniometer over axis of knee (lateral epicondyle). Align fixed arm with greater trochanter, parallel with the lateral midline of the femur. Align "moving arm" with the lateral malleolus, parallel with the fibula. Measurement is recorded as degrees lacking from neutral.

Normal range: 10° of hyperextension to 135° flexion.

End-feel: firm due to posterior capsule and ligaments.

4. Ankle dorsiflexion

Patient position: Supine. As we are interested in the effects of gastrocnemius shortening on ankle dorsiflexion, this test is undertaken with the knee in full extension. The calcaneum is held in neutral alignment whilst pressure is applied over the mid-section of the foot to dorsiflex the ankle as much as possible, preventing inversion.



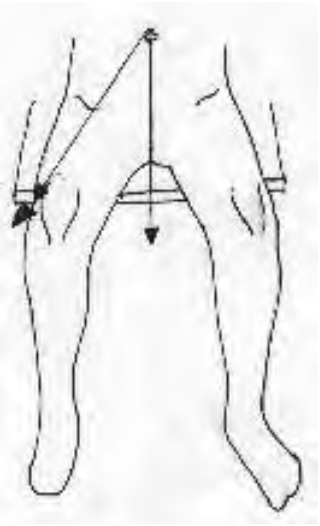
Goniometer position: Axis of the goniometer over the lateral malleolus. Stationary arm aligned with the fibular head, along the lateral aspect of the lower leg. "Moving arm" held parallel to the lateral aspect of the 5th metatarsal, aligned with the posterior third of the foot (this is to ensure that gastrocnemius range is being monitored and not that of the planter structures of the foot). Note range of dorsiflexion past plantergrade as +x°'s, range lacking from plantergrade as -x°'s

Normal range: 20° dorsiflexion to 50° planterflexion.

End feel: firm due to joint capsule, Achilles tendon and ligaments.

5. Iliotibial band

Patient position: supine with plinth supporting trunk. Both hips extended and in neutral rotation, knees should be flexed over side of plinth, feet should be off the floor. ASIS should be level. Note any asymmetry of legs.



Goniometer position: centre of the goniometer placed over the umbilicus. Stationary arm aligned straight down from umbilicus. Align 'moving arm' with lateral border of the patella.

Interpretation: <20° of abduction from midline – no contracture

20°-30° of abduction from midline – moderate contracture

>30° of abduction from midline – severe contracture

References

American Academy of Orthopedic Surgeons (1965). Joint Motion: Method of measuring and recording. London, Churchill Livingston.

Gajdosik, R. & Bohannon, R. (1987). "Clinical measurement of range of motion: review of goniometry emphasizing reliability and validity." Physical Therapy 67(12): 1867-1872.

Kendall FP, McCreary EK, Provenance PG. 1993 'Muscles: Testing and Function' 4th Ed Williams & Wilkins

Norkin, C. & White, D. (1995). Measurement of joint motion: a guide to goniometry. Philadelphia, FA Davis.

Pandya, S., Florence, J. M., King, W. M., Robison, J. D., Oxman, M. & Province, M. A. (1985). "Reliability of goniometric measurements in patients with Duchenne muscular dystrophy." Phys Ther 65(9): 1339-42.

Joint Range of Movement

Patient ID:

Date:

Assessed by:

	ROM		Comments
	R	**L**	
Elbow extension			
Hip extension			
Knee extension			
Ankle dorsiflexion			
ITB tightness			

Management:

Orthotic management	
Maintenance of ROM – include advice re stretches, who performs them, frequency, problems	
Community physiotherapy	

Manual Muscle Testing

General method:

1. Explain or demonstrate movement to the patient.
2. Ask the patient to perform the movement in the antigravity position.
3. If unable to then check patient's understanding, ROM available, weakness.
4. Based on the limiting factor(s) either instruct and demonstrate again, decide if ROM is affecting performance or repeat the test using antigravity position.
5. Resistance is given through full available range.
6. In the case of a contracture limiting ROM, grades are given as stated in table 1, for available range. Range limitation should be noted in the comments section.
7. If uncertain as to what grade should be given, the lower option should be chosen.
8. If general compliance is such a problem that the results do not give, in the assessors considered opinion, a true indicator of the patient's ability, then this should be noted and the results should not be included in the database. If specific muscle groups are a problem e.g. due to pain, please note this and do not include in calculation of the MRC %.
9. Equipment –a height adjustable plinth should be available.
10. Clothing – ideally shorts and t-shirt. If not shorts then tracksuit bottoms. Jeans may restrict movement – especially in very weak children.
11. Watch for compensatory movements being used to produce an action – if uncertain palpate the muscle group which should be prime movers. Be clear with positioning and stabilization.
12. The muscle groups required for the North Star constitute the minimum data set to be assessed. If you need to include further muscle groups to assist in clinical decision making for the individual patient, please do so, but DO NOT include them in the calculation of MRC%.

Table 1 – North Star MMT Grading system

Grade	Grade for %MRC	Definition
0	0	Nothing, no muscle contraction
1	1	"Flicker" – palpable contraction, no movement
2-	2	Part ROM with gravity counterbalanced
2	2	Full ROM with gravity counterbalanced
2+	2	Full ROM with gravity counterbalanced and a little resistance, no antigravity movement
3-	2	Antigravity movement through part most of range
3	3	Full range antigravity movement
3+	3	Full antigravity ROM with some resistance through part of range
4	4	Full antigravity ROM with some resistance through the whole range
4+	4	Full antigravity ROM with normal power in part of range
5	5+	Full ROM with full normal resistance throughout range

The above scale is based upon the modifications made to the original Medical Research Council 6-point scale by staff at the Hammersmith Hospital. Most physiotherapists who regularly muscle chart are familiar with this scale (or the expanded 16-point version) from previous workshops.

A minus grade denotes loss of range. This is why 4- is not graded, as a grade of 4 is, by definition, full range.

The MRC percentage is to be calculated on the 'whole' score, rather than with the + and - grades. The 'whole' grades for each element of the scale are noted in column 2 of the above table.

The test methodology which follows (Table 2) has been taken from a variety of sources – the comprehensive work undertaken by the physiotherapists involved with the CIDD projects in assessment of DMD in the States, and from a number of MMT textbooks. References are given, and further information/clarification should be sought from them as needed.

Choice of muscle groups has been based upon present clinical practice (from the centres which currently assess muscle activity) and a review of the literature relating to reliability of MMT.

Useful texts

Kendall FP, McCreary EK, Provenance PG. 1993 'Muscles: Testing and Function' 4th Ed Williams & Wilkins *KEY TEXT*

Clarkson HM. 2000. 'Musculoskeletal Assessment: joint range of motion and manual muscle strength' 2nd Ed. Lippincott, William & Wilkins, Philadelphia

Daniels & Worthingham. 1980 'Muscle testing: techniques of manual examination' 4th Ed. WB Saunders Company, Philadelphia

Starting position – SITTING		
Muscle group	**Test detail**	**Grade**
Shoulder flexors	Starting position – sitting, arm at side with elbow slightly flexed. Assessor stabilises scapula Action – shoulder flexion, with slightly bent elbow, palm facing dow Resistance – just proximal to elbow	-3-5
Shoulder abductors	Starting position – sitting, arms by side Action – lift arm away from side, until 90° abduction achieved, with elbow flexed and forearm pronated. Resistance – downward pressure just proximal to the elbow. Stabilise so patient does not lean	-3-5
Elbow flexors	Starting position – sitting, with arm in slight flexion at shoulder, forearm supinated, elbow in full extension Action – take hand to shoulder Resistance – just proximal to wrist. Stabilise under elbow, or anterior surface proximal humerus.	-3-5
Elbow flexors (alternate)	Starting position – sitting, shoulder abducted to 90° and passively supported by the assessor (can also be tested in side-lying) Action – take hand to shoulder, horizontal plane Resistance – 2+ only - just proximal to wrist	0-2+
Elbow extensors (alternate)	Starting position – sitting, shoulder flexed to 90°, elbow flexed towards body in the horizontal plane. Assessor supports upper arm (can also be tested in side-lying) Action – straighten elbow in horizontal plane Resistance – 2+ only - just proximal to wrist	0-2+
Hip flexors	Starting position – upright sitting. Stabilises self by holding front of bench. Action – lifts knee towards chest (min 30° from bench) Resistance – anterior distal surface of the thigh NB – thigh should be in neutral rotation, patient must not flex knee to provide support for leg on the bench. 3- graded if leg is lifted just enough for assessor to slide hand under thigh, or if thigh is lifted but not in neutral rotation	-3-5
Knee extensors	Starting position – sitting, patient can 'prop' if needed Action – straighten knee Resistance – just above ankle NB – be aware of the effects of tight hamstrings on knee extension in this position. A 5-10°extension lag in this position is acceptable. Any 'lag' over and above this should be interpreted as an inability to actively move through full range, and grading reduced appropriately.	-3-5
Ankle dorsiflexors	Starting position – sitting, assessor supports leg just above the ankle joint Action – bring foot upwards Resistance – over dorsum of foot NB – dorsiflexion should be achieved without inversion. If can only dorsiflex with inversion – assessor's decision on grading	-3-5

Starting position – SUPINE		
Neck Flexors	Starting position – supine Action – neck flexion until chin touches chest (or whatever is full ROM for pt) Resistance – given at forehead NB – take care with resistance and be prepared to support head – one hand under occiput NOT graded if flexion is achieved through circumduction, or if head is lifted with NO neck flexion	-3-5
Elbow Extensors	Starting position – supine, assessor supports upper arm in 90° of flexion, forearm is in neutral and lies across patient's chest. Action – extend elbow Resistance – given just proximal to wrist	-3-5
Resistance – over dorsum of foot	NB – dorsiflexion should be achieved without inversion. If can only dorsiflex with inversion – assessor's decision on grading	0-2+
Hip abductors (alternate)	Starting position – supine, assessor supports leg – usually under heel Action – abduct hip Resistance – 2+ only – just proximal to knee NB – do not allow extreme hip rotation, hip or knee flexion	0-2+
Hip adductors (alternate)	Starting position – supine, assessor supports leg – usually under heel. Action – adduct hip until contact is made with other leg Resistance – 2+ only – just proximal to knee NB – do not allow extreme hip rotation, hip or knee flexion	0-2+
Starting position – PRONE		
Neck extensors	Starting position – prone, arms by sides Action – neck extension – should be able to lift head until face is directly forward Resistance – given to occiput NB – keep one hand just under chin to offer support if needed	-3-5
Shoulder extensors	Starting position – prone, arm medially rotated and adducted. Assessor stabilises scapula Action – lift arm away from bench Resistance – just proximal to elbow	-3-5
Hip extensors	1. Starting position – prone, with knee flexed to 90° Action – lift knee off bench, whilst keeping knee bent. Assessor stabilises at pelvis. Resistance – over distal thigh If unable, or just able to achieve – 2. Starting position – prone with hips flexed to 90° over plinth Action – lifts thigh away from horizontal Resistance – distal thigh NB – thigh should be in neutral rotation	-3-5 -3
Knee flexors	Starting position – prone Action – flex knee, take heel towards bottom Resistance – just proximal to Achilles tendon	-3-5
Planterflexors	Starting position – prone, knee flexed to 90° Action – point toes towards ceiling Resistance – support calcaneum with slight upward pressure whilst applying downward pressure over the ball of the foot	-3-5

Starting position – SIDE-LYING		
Muscle group	**Test detail**	**Grade**
Hip abductors	Starting position – side-lying, bottom leg slightly flexed for support, can hold front of bench for support. Assessor stabilises pelvis to stop backward rotation Action – maintaining knee extension, and hip in slight extension and lateral rotation, lift leg towards ceiling Resistance – just proximal to knee – adduction and slight flexion	-3-5
Hip Adductors	Starting position – side-lying, lower leg extended at knee and in neutral extension at hip. Assessor stabilises uppermost leg in abduction. Patient can hold edge of bench to stabilise trunk Action – lift leg lower leg towards upper one. Resistance – just proximal to knee	-3-5
Neck Flexors (alternate)	Starting position – side-lying , head supported by assessor if necessary Action – neck flexion until chin touches chest Resistance – 2+ only – given to forehead	0-2+
Neck extensors (alternate)	Starting position – side-lying, head supported if necessary Action – neck extension Resistance – 2+ only – given to occiput	0-2+
Shoulder flexion (alternate)	Starting position – side-lying, assessor supports upper arm Action – shoulder flexion with palm facing backwards Resistance – 2+ only – just proximal to elbow, anterior aspect	0-2+
Shoulder extension (alternate)	Starting position – side-lying, assessor supports arm and stabilises scapula Action – arm extension, keeping palm facing backwards Resistance – 2+ only - just proximal to elbow, posterior aspect	0-2+
Hip flexors (alternate)	Starting position – side lying, uppermost leg supported at thigh and calf by assessor. Action – take knee towards chest Resistance – 2+ only – anterior surface distal thigh	0-2+
Hip extensors (alternate)	Starting position – side lying, assessor supports thigh and calf of uppermost leg. Action – hip extension with knee flexed Resistance – 2+ only just proximal to knee joint, posterior thigh	0-2+
Knee flexors (alternate)	Starting position – side lying, uppermost leg supported at thigh and calf by assessor. Action – bend knee, taking heel towards bottom Resistance – 2+ only - just proximal to ankle, posterior aspect	0-2+
Knee extensors (alternate)	Starting position – side lying, uppermost leg supported at thigh and calf by assessor. Action – straighten knee Resistance – 2+ only – just proximal to anterior ankle	0-2+
Ankle dorsiflexors (alternate)	Starting position – side lying, uppermost leg and foot supported by assessor Action – pull foot towards head, from the ankle only Resistance – 2+ only – over dorsum of foot	0-2+
Ankle planterflexors (alternate)	Starting position – side-lying, uppermost leg supported by assessor Action – point toes Resistance – 2+ only – over ball of foot	0-2+

Manual Muscle Testing

Patient ID:

Assessed by (initial and sign)			
Date of assessment			Comments
Muscle Group	Grade (0-5)		
	R	L	
Neck Flexors			
Neck Extensors			
Shoulder Flex.			
Shoulder Exts			
Shoulder Abds			
Elbow Flexors			
Elbow Ext.			
Hip Flexors			
Hip Extensors			
Hip Abductors			
Hip Adductors			
Knee flexors			
Knee Extensors			
Dorsiflexors			
Planterflexors			
Total score			
MRC%			

MRC% = [Total actual score ÷ (Total number of muscle scored x 5)] X 100
i.e. = [Total actual score ÷ 140 (if all muscle groups are scored)] X100

General comments:

Pulmonary Function Testing

Introduction

To control for equipment variation, the preference is for sites to use the Microlab ML3500 if possible. If not available, ensure that the same pulmonary function machine is used at each visit. The spirometer must be able to calculate both percentage and absolute values.

Spirometry will be performed to measure the maximal volume of air forcibly exhaled from the point of maximal inhalation (forced vital capacity, FVC). The percentage and absolute value of the FVC from the best of three tests in a sitting position should be documented. The best FVC (both % and absolute value) will need to be recorded. As FVC % predicted for height is being calculated, height measurement needs to be accurately recorded and input prior to pulmonary function testing.

Pulmonary Function Testing Technique

The patient should be sitting comfortably in a chair. The assessor explains to the subject what the test technique is. The wording of instructions should be followed closely. The assessor asks the patient to take as big a breath in as possible and then to *blow out through the mouth as hard and as long as they can. The assessor should verbally encourage the subject to keep going for as long as possible.* The test should be repeated three times making sure that the patient has recovered between attempts. If at each attempt there is an improvement in the results further tests can be conducted until the subject has achieved his best result.

The subject can place the mouth piece in his own mouth unless he is unable to reach his mouth with his hands or flexes his head and shoulders to reach the tube. In this case the assessor should place the tube into the subject's mouth.

The tube can be placed in the mouth either before the beginning of inspiration or at the end of inspiration which ever the subject finds most comfortable. Whichever technique is used the same method should be used at each evaluation.

Where a tight seal around the mouthpiece cannot be achieved, the lips may be held by the evaluator or the subject to ensure no air leaks occur. This is not likely to be an issue with young children with Duchenne muscular dystrophy. To ensure consistency, the same technique should be used at each evaluation.

The subject should not be allowed to flex forward excessively during expiration.

Height calculation

Standing: A stadiometer should be used. The patient must stand erect with heels flat on the floor. Where a child can ONLY stand on their toes, then the distance of heel from floor needs to be measured and subtracted from overall height measurement.

Sitting: At present there does not appear to be a consensus on the method of measuring height for children who are unable to stand. This is not an immediate issue for the North Star as we are initially monitoring FVC in ambulant children. Current recommendation is for centres to continue using their present method of estimating height from a seated position, which in general is to equate arm span to height.

Calibration

Calibration of the spirometer should be undertaken on a weekly basis. A 'biological' control is recommended i.e. – the same member of staff performs an FVC test.

Cleaning & disinfection

This should be undertaken in line with manufacturers and local Trust guidelines. Single use disposable mouthpieces are recommended. Filters are also available.

Printer paper

'Archive' printer paper should be used. This is guaranteed non-fade for a 25 year period.

North Star Pulmonary Function Testing

Patient ID:

Date of assessment:

Assessed by (print and sign):

Height (cm):

	Absolute Value	%age predicted for height	Comments
Test 1			
Test 2			
Test 3			

Myometry

General Test Method

The myometer is a very sensitive measuring tool therefore care must be taken to be consistent with test method.

- A 'make' test will be used unilaterally (dominant side) for the following muscle groups:
  Elbow flexors
  Hand grip
  Knee extensors
  Shoulder abductors
  Hip flexors
- For those who are time limited and cannot do all 5 groups the following are recommended – elbow flexors, hand grip, knee extensors.
- Care must be taken to apply the myometer to the same point on the limb at each test session
- The myometer must be applied evenly and perpendicularly to the direction of force from the muscle group being tested.
- Muscle groups are generally tested in their mid-range and in a gravity compensated/ limb supported position
- Stabilisation of both the patient and the myometer is important and should be standardised.
- Wherever possible, the same physiotherapist will reassess the patient.
- The test is explained to the patient in a way that they understand. The intent is to build a maximum isometric hold, so the command is effectively 'HOLD' or 'Keep still/don't let me move you'. The words push or pull should not be used.
- The patient will be encouraged verbally to build to and maintain a maximum hold over a period of approximately 5 seconds, to allow for full physiological recruitment of muscle.
- The patient shall be told/shown the results of each test.
- Following one 'trial' test to allow for learning, the best of 3 tests shall be noted. Test results should be fairly closely grouped – a 10% variation is not unusual, e.g. 30N +/- 3
- Any discomfort will limit the patient's ability to offer maximum resistance. As much as is possible, ensure comfort when applying the myometer. The applicator can be padded to allow for comfort.
- If the therapist does not feel that they have been able to gain compliance from the patient for any reason (e.g. understanding or poor concentration), this should be noted.
- Calibration – should be checked once a month. Weights will be provided.

1. Knee extensors

Patient position – patient sitting with thigh supported and hip and knee at 90°, on plinth, chair or in wheelchair. Feet must be clear of the floor. Femur in neutral rotation. Patient can hold onto front of plinth or chair to stabilise himself.

Stabilisation – if needed, given by therapist over lower third of thigh, just above knee.

Myometer position – anterior surface of tibia, at junction between middle and lower third of tibia.

Therapist position – sitting in front of child, on a chair or on floor

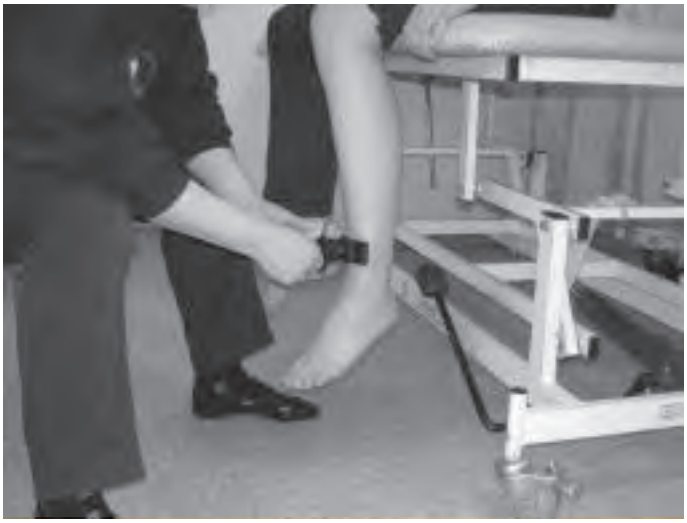


Figure 1: Knee extensors

2. Hip Flexors

Patient position – Supine with hip and knee at 90°. Femur in neutral rotation. Patient is asked to concentrate on keeping knee steady, not to move foot, as this discourages hamstring involvement.

Stabilisation – under knee and calf to support weight of leg and prevent unwanted hip movement.

Myometer position – anterior aspect of lower thigh, just proximal to condyles.

Therapist position – at side of, or kneeling on, plinth. Facing the patient.

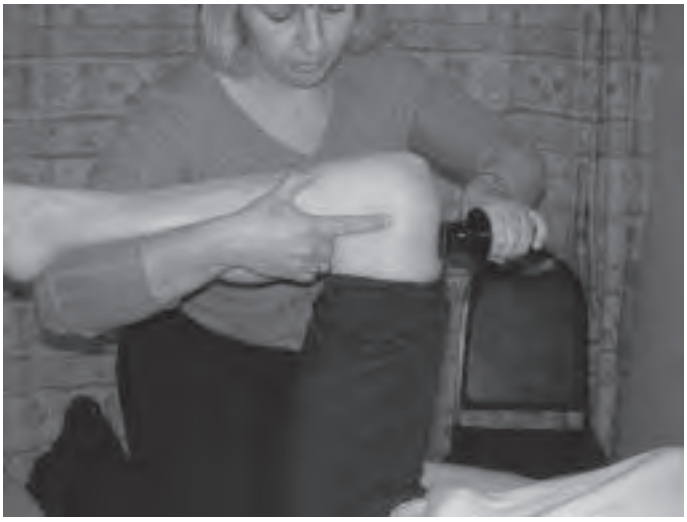


Figure 2a: Hip Flexors - Option 1

Figure 2b: Hip Flexors - Option 2

3. Shoulder abductors

Patient position - supine on plinth. Shoulder abducted to 45°, elbow flexed to 90° with forearm in neutral.

Stabilisation – support at wrist, with arm just off plinth

Myometer position – Lateral aspect of humerus, just proximal to the lateral epicondyle.

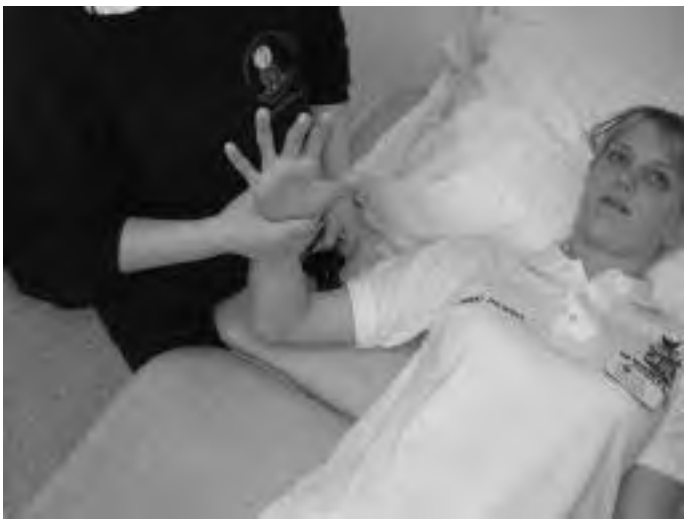


Figure 3: Shoulder abductors

4. Elbow flexors

Patient position – supine, upper arm against side of body. Elbow flexed to 90°, forearm in supination

Stabilisation – given by therapist over lower humerus.

Myometer position – at junction of lower and middle third of forearm.

Therapist position – at side of plinth, facing the patient

Figure 4: Elbow flexors

5. Grip

Patient position – supine, upper arm against side of body. Elbow flexed to 90°, forearm in neutral.

Stabilisation – if needed, over lower end of humerus.

Myometer position – in patient's hand. Fingers need to be around correct part of grip.

Grip can also be tested in a sitting position for patients who are wheelchair users and unwilling to be transferred out of their chairs.

Figure 5: Grip

North Star Myometry Data Collection Sheet

Patient ID

Date:

Assessed by (print & sign):

Dominant side: Right/Left*

***delete as appropriate**

Muscle group	Test Results (N)	Best Score (N)	Comments
Knee extensors			
Hip Flexors			
Elbow flexors			
Shoulder Abductors			
Grip			

Stretches for lower limbs and upper limbs

ACTIVE CALF STRETCHES STANDING POSITION (GASTROCNEMIUS)

Position

- Stand facing wall
- Keep back leg straight
- Heel on floor
- Knee straight
- Toes point to wall

Stretch

- Lean towards wall until stretch is felt in calf of back leg
- Keep bottom in

Hold ☐ seconds

Repeat ☐ times

Special instructions

ACTIVE CALF STRETCHES STANDING POSITION (SOLEUS)

Position

- Stand facing wall with both knees bent and foot to be stretched behind

Stretch

- Lean into the wall, squat down slowly until stretch is felt in lower calf of back leg

Hold ☐ seconds

Repeat ☐ times

Special instructions

PASSIVE SELF-STRETCH FOR TENDO ACHILLES ON STANDING BOARD

Position

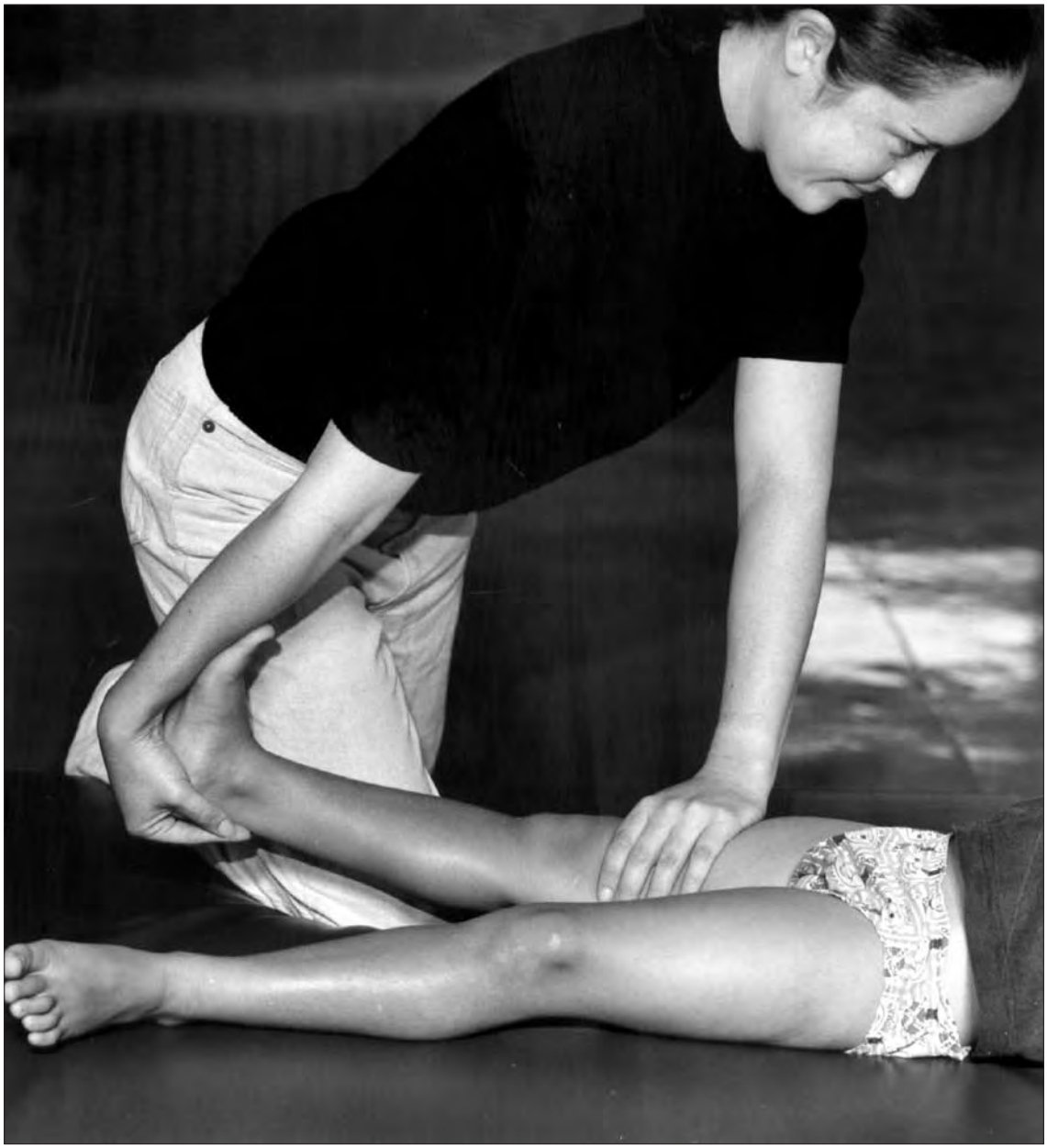
- Position standing board against wall
- Child stands on board resting back against wall
- Feet point straight ahead (or slightly pigeon toed)

Stretch

- Child positions heels as far back on board as possible
- Keep knees straight
- Keep heels down
- Stretch is felt at back of both calves

Duration ☐

Special instructions



MANUAL ACHILLES TENDON STRETCH

Position

- Child lying on back
- Place small rolled towel under the knee of the leg being stretched
- Cup heel in hand
- Rest sole of foot on forearm
- Stabilise above the knee with the other hand

Stretch

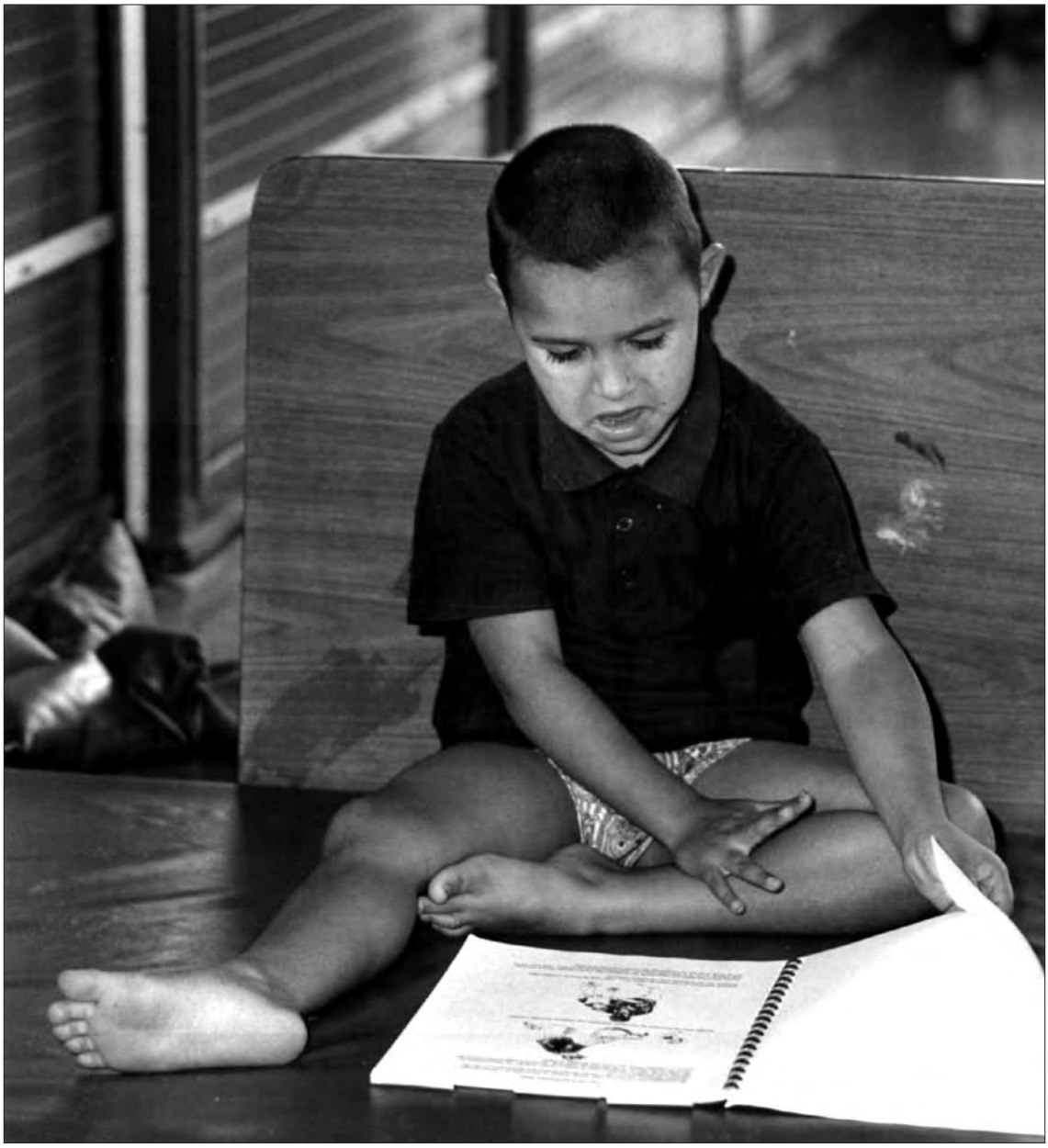
- Pull down firmly on the heel while pushing the ball of the foot up. Keep knee straight
- Stretch is felt in calf

Hold ☐ seconds

Repeat ☐ times

Special instructions

If resistance to stretch is felt, bend the knee, stretch the ankle then straighten the knee while maintaining the ankle stretch. Place support under the knee to prevent hyperextension.



PASSIVE SITTING HAMSTRING STRETCH POSITION

Position

- As in photograph
- Knee should be as straight as possible and leg slightly out to the side
- Lower spine straight
- Sit with hips well back against the wall

Stretch

- Stretch is increased by leaning forwards
- Stretch is felt at back of straight thigh

Duration ☐

Repeat other side

Special instructions

PASSIVE SELF-STRETCH FOR HAMSTRINGS

Position

- Child lies on back in doorway or beside post
- Place leg to be stretched on the wall with knee slightly bent and bottom close to the wall
- Keep other leg straight

Stretch

- Straighten the knee until stretch is felt in back of thigh

Hold ☐ seconds

Repeat ☐ times

Special instructions

MANUAL HAMSTRING STRETCH

Position

- Child lies on back
- Place ankle on shoulder (as in photograph)
- Stabilise the opposite leg with other hand
- Keep knee of moving leg straight with hand

Stretch

- Rock forward using this movement to perform the hamstring stretch
- Stretch is felt at back of upper thigh

Hold ☐ seconds

Repeat ☐ times

Special instructions

HIP FLEXOR STRETCH (PLUS ILIOTIBIAL TRACT)

Position

- Child lies flat on tummy
- Cup bent knee in hand
- Ankle rests on elbow or upper arm
- Place other hand on bottom

Stretch

- Pull knee up and **towards** the other leg while applying downward pressure on the bottom
- Stretch is felt in groin and outside of hip

Duration ☐

Repeat on other side

Special instructions

ILIOTIBIAL TRACT (MANUAL STRETCH IN PRONE)

Position

- Child lies on tummy
- Grasp leg to be stretched at knee
- Stabilise and keep pelvis and trunk flat with knee and hand

Stretch

- Lift leg up
- Pull leg across towards other leg
- Apply pressure on buttocks to keep pelvis flat
- Stretch is felt down outer thigh

Hold ☐ seconds

Repeat ☐ times

Special instructions

ILIOTIBIAL TRACT (MANUAL STRETCH IN SIDE LYING)

Position

- Child lies on side with lower leg bent
- Leg to be stretched uppermost with knee straight
- Stabilise pelvis with hand and knee

Stretch

- Take leg backwards as far as possible
- Apply firm downward pressure at the knee
- Stretch is felt down outer thigh

Hold ☐ seconds

Repeat ☐ times

Special instructions

HIP FLEXOR STRETCH IN SIDE LYING

Position

- Child lies on side with underneath leg bent
- Hold as in photograph
- Lower hand cups knee
- Other hand applies pressure on uppermost buttock
- Stabilise pelvis with knee.

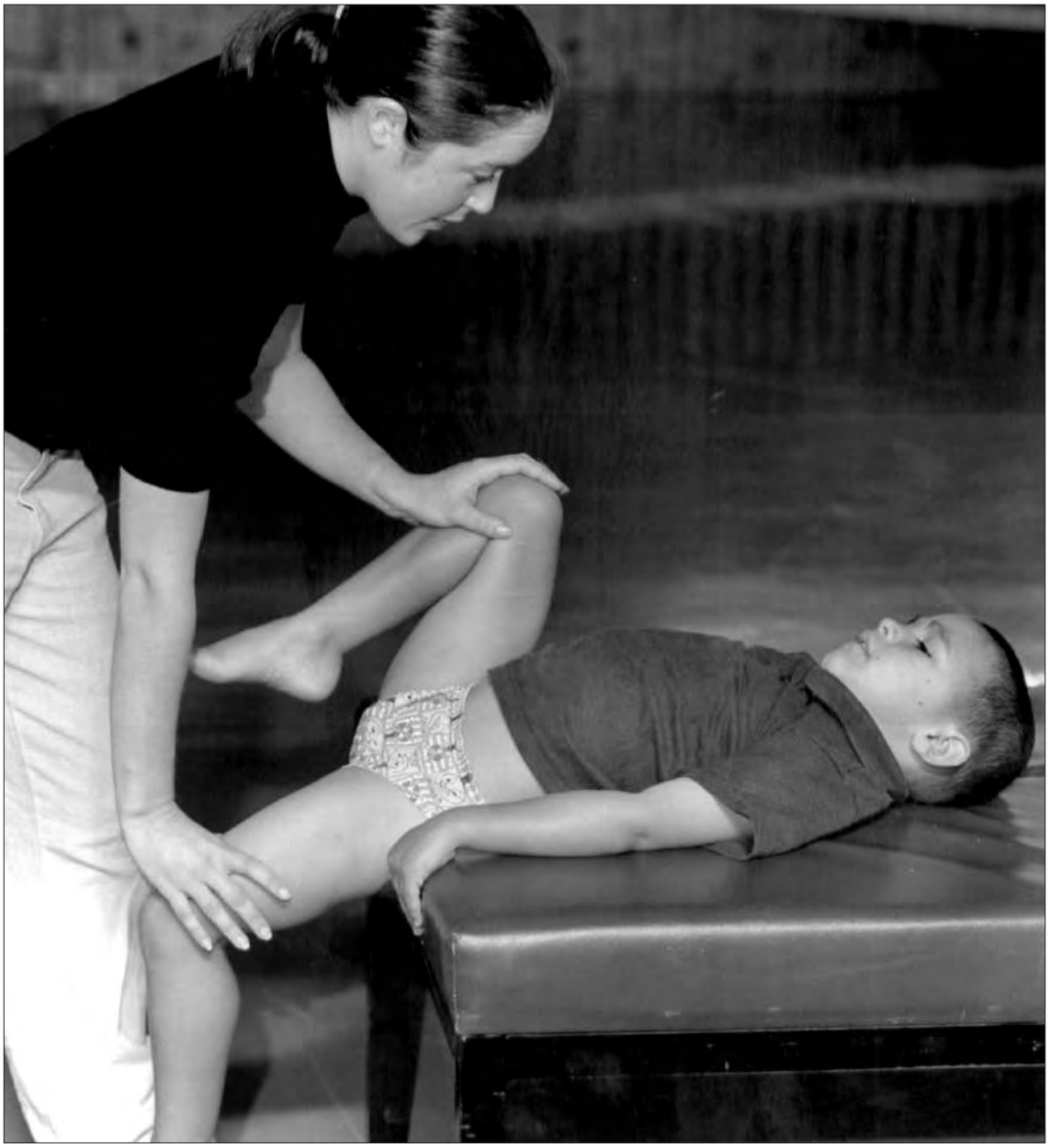
Stretch

- Move top leg backwards until stretch is felt in groin

Hold ☐ seconds

Repeat ☐ times

Special instructions



HIP FLEXORS ON BACK

Position

- Child lies on back
- Stabilise the lumbar spine by holding non-moving leg bent up on chest

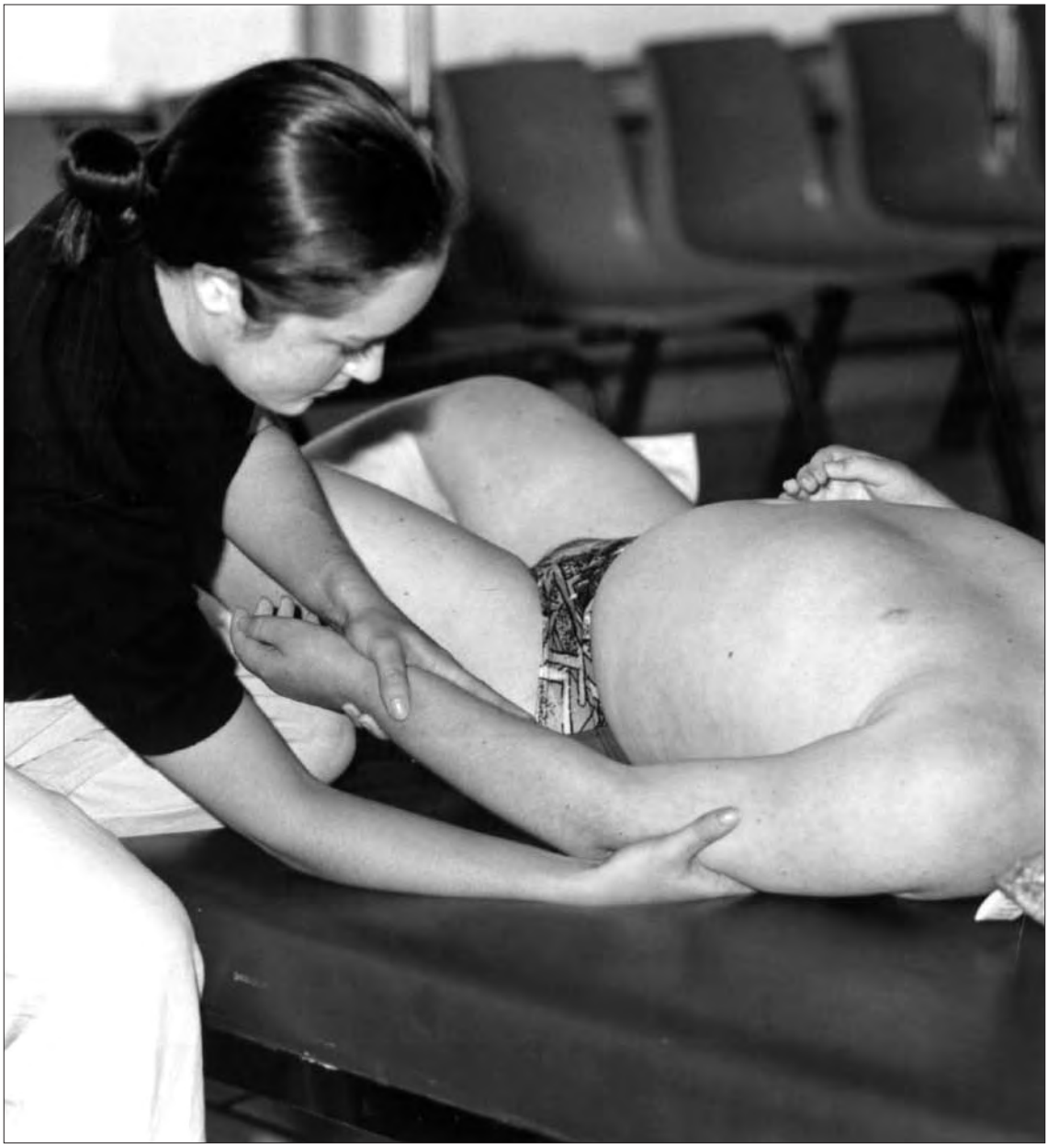
Stretch

- Push down on leg to be stretched while holding other knee in bent position (as in photograph)
- Stretch is felt in groin

Hold ☐ seconds

Repeat ☐ times

Special instructions



ELBOW STRETCH

Position

- Child lies on back or sits on chair
- With palm facing upwards
- One hand supports shoulder joint or upper arm
- Hold above wrist with other hand

Stretch

- Straighten elbow as far as possible, until stretch is felt in front of elbow

Hold ☐ seconds

Repeat ☐ times

Special instructions

FOREARM STRETCH (PRONATORS)

Position

- Hold child's hand as in photograph
- Stabilise the wrist
- Stabilise at the elbow

Stretch

- Slowly turn hand to 'palm up' position until stretch is felt

Hold ☐ seconds

Repeat ☐ times

Special instructions

LONG FINGER FLEXORS

Position

- With elbow as straight as possible
- Support the child's palm, maintaining straight fingers
- Keep thumb out to the side
- Support the wrist

Stretch

- Slowly bend the wrist and hand back until a stretch is felt in the forearm

Hold ☐ seconds

Repeat ☐ times

Special instructions

TIBIALIS POSTERIOR STRETCH

Position

- Boy lies on back
- Operator grasps foot firmly around the head of 1st Metatarsal
- Provide counter pressure on the lateral aspect of heel
- Pull foot into eversion and dorsiflexion

Hold ☐ seconds

Repeat ☐ times

Special instructions

Additional notes

Hospital admission form

Name _____

Address _____

_____

_____

Telephone no. _____

☐ Duchenne Muscular Dystrophy

☐ Becker Muscular Dystrophy

Other _____

_____

_____

_____

A. Physical	I can	I cannot
1. a. Walk unaided		
b. Walk aided		
2. a. Stand unaided		
b. Stand aided		
3. Raise my arms		
4. Move my arms		
5. Move my legs		
6. Straighten my legs		
7. a. Lie on my stomach		
b. Lie on my back with supportive pillows		
c. Lie on my back without supportive pillows		
8. Sit myself up		
9. a. Sit up unsupported		
b. Sit up with support of pillows/wedges		
10. Turn myself in bed		
11. Lift up my head when lying down		
12. a. Dress myself unaided		
b. Dress myself aided		
13. Get in and out of bed myself		
14. Get up and down from a chair myself		
15. Call for assistance (and require a buzzer to hand)		

B. Feeding	I can	I cannot
1. a. Feed myself unaided		
b. Feed myself using aids		
2. Cut up my food		
3. Eat lying down		
4. Take drinks from my side locker		
5. Hold a cup unaided		
6. Drink from an ordinary cup		
7. Drink through a straw		

C. Toilet		
1. a. Sit on a toilet unaided		
b. Sit on a toilet aided		
c. Wipe myself		
2. a. Get up from the toilet unaided		
b. Get up from toilet aided		
3. a. Use a bed pan/bottle unaided		
b. Use a bed pan/bottle aided		
4. a. Wash myself		
b. Wash my hands and face only		
5. Comb my hair		
6. Clean my teeth		
7. Blow my nose		
8. Shave myself		

D. Medication		
1. Swallow tablets		
2. Take liquid medicine		

Risks of anaesthesia in children with muscular dystrophy

By Dr. Margaret Vroom
From *Parent Project Newsletter* 4/97

When a patient with muscular dystrophy is subjected to general anaesthesia, a number of serious problems may arise. As a parent, it might be useful to be informed about these possible anaesthesia-related risks and how these risks can be minimised by careful selection of the administered anaesthetic agents. Therefore, a brief overview of the literature and some of the current ideas and concepts will be discussed.

Anaesthesia-related complications in children with muscular dystrophy can be subdivided into general and specific categories.

General

Muscular dystrophy not only affects the muscles of the extremities, but as the disease progresses, heart and respiratory muscles become involved as well.

Almost all anaesthetics decrease contractility of the heart. Many children with muscular dystrophy have an impaired cardiac function (cardiomyopathy). As a result, difficulties may arise with the circulation in the perioperative period.

Prolonged spontaneous respiration during general anaesthesia may result in inadequate ventilation of the lungs with increased carbon dioxide levels in the blood. This in turn can cause severe heart rhythm disturbances, especially in those patients with an already impaired cardiac function and even more so in combination with certain (volatile) anaesthetics. During the majority of general anaesthetic procedures, however, the respiratory function is controlled by a mechanical respirator via insertion of a tube in the upper airways, which requires the use of drugs resulting in a temporary relaxation of the muscles. The administration of these muscle relaxants also carries specific risks which will be discussed below.

Following general anaesthesia, problems may arise when the patient has to regain his respiratory function. The respiratory drive is diminished as a result of residual drug effects, coughing is impaired and aspiration easily occurs. All of this may result in low levels of oxygen and high levels of carbon dioxide in the blood. Furthermore, certain areas of the lung may collapse (atelectasis), impairing oxygenation of the blood and increasing the risk of respiratory infections.

Therefore, it is important (and for large operations even mandatory) to perform extensive preoperative screening, not only to estimate the perioperative risks, but to provide optimal perioperative monitoring. The screening may include a cardiac ultrasound, an electrocardiogram, pulmonary functions tests as well as blood gases.

Specific

The membranes of muscle fibres and/or the receptors on the muscle cells can alter as a result of the disease, both in function as well as in number. Receptors are specific proteins on a cell membrane which, upon stimulation, modulate the function of the cell.

In patients who have been bedridden for a long period or who are unable to actively use their muscles, the administration of the 'depolarising' muscle relaxant succinylcholine may result in the release of large amounts of the ion potassium from the muscle cell into the bloodstream, since depolarising muscle relaxants cause the muscles to contract briefly before they relax. This sudden increase in potassium concentration in the blood may result in life-threatening heart rhythm disturbances. The 'non-polarising' muscle relaxants (for instance vecuronium, atracurium or

mivacuronium) do not initiate muscle contraction and, therefore, their use does not carry this risk. Nonetheless, the affected muscles may have become more sensitive to non-depolarising muscle relaxants and, therefore, the dose administered has to be carefully chosen.

The brief but profound muscle contraction associated with the use of succinylcholine leads to elevated levels of creatinine kinase (CK) and myoglobin which can be detected in the blood afterwards. The use of succinylcholine and inhalational (volatile) anaesthetics, such as halothane, can result in increased muscle breakdown, called rhabdomyolysis. Muscle breakdown leads to the release of high concentrations of potassium, CK and myoglobin in the circulation. As mentioned previously, high concentrations of potassium in the blood may result in cardiac arrest and the high concentrations of muscle proteins may severely impair the kidney function.

In addition, the use of succinylcholine and inhalational anaesthetics may result in a syndrome called malignant hyperthermia. Malignant hyperthermia is a life-threatening disturbance of the calcium-homeostasis in the muscle cell provoked by certain anaesthetics. Succinylcholine and inhalational anaesthetics are particularly renowned for causing this syndrome. Malignant hyperthermia occurs relatively frequently in patients with muscular dystrophy. Malignant hyperthermia is characterised by an extremely elevated metabolism within the muscle cell. As a result, the temperature of the entire body rises to life-threatening levels, oxygen consumption and carbon dioxide production of the body increase dramatically and waste products are released into the circulation. It is of extreme importance that this syndrome is readily recognised so that the appropriate measures can be taken, including drastic cooling of the body and prompt initiation of therapy with the drug dantroleen. Unfortunately, despite appropriate measures, the course of this syndrome is often fatal. Children with muscular dystrophy should always be considered of being at high risk for the development of malignant hyperthermia and, therefore, the high-risk anaesthetics should be avoided.

Conclusions

General anaesthesia is accompanied by a number of important risks. When general anaesthesia is required in order to undergo a specific procedure, succinylcholine and inhalational anaesthetics need to be AVOIDED.

Painkillers (opioids), the anaesthetic propofol, midazolam and hypnomidate can probably all be used safely. When it is essential to use muscle relaxants, short-acting, non-depolarising muscle relaxants can be used albeit at a reduced dose (one-fourth to one-fifth of the usual dose). Controversy exists as to whether antagonising these muscle relaxants with cholinesterase inhibitors carries additional risks. Therefore, it is of utmost importance to inform your anesthetist as early as possible about the medical history of your child. The necessary preoperative screening procedures (heart and lungs) can be performed and also the appropriate anaesthetics can be selected. In addition, the anesthetist will be enabled to provide optimal perioperative monitoring.

You may want to show this document to the anaesthetist involved in caring for your child during any surgical or emergency procedure.

Useful internet sites

Muscular Dystrophy Western Australian

http://www.mdwa.org.au/

Muscular Dystrophy Tasmania

mdtasmania.org.au

Muscular Dystrophy Queensland

mdqld.org.au

Muscular Dystrophy New South Wales

www.mdnsw.org.au

Muscular Dystrophy South Australia

mdasa.org.au

Muscular Dystrophy Victoria

www.mda.org.au

Duchenne Foundation

www.duchennefoundation.org.au

Muscular Dystrophy USA

www.mdausa.org

Parent Project Muscular Dystrophy

www.parentprojectmd.org

Action Duchenne UK

www.actionduchenne.org

United Parent Project Muscular Dystrophy

www.uppmd.org

Since 1932, when it was established as The Queensland Society for Crippled Children, Montrose*ACCESS* has been providing therapeutic and respite services for children with physical disabilities. Currently, staff of Montrose*ACCESS* offer a holistic, community-based service with members of an interdisciplinary team visiting the children in their homes and schools. Duchenne muscular dystrophy is the largest diagnostic group managed by this organisation.

Over the years, many dedicated staff from Montrose*ACCESS* have treated children with this devastating condition, from their early childhood into adulthood. Thus the practitioners have had the opportunity to make clinical observations, observe patterns and make predictions. In short, a knowledge base has developed. Informally, information has been shared and there have been requests for assistance from other practitioners, especially those located in rural and remote areas.

This package has been developed in response to a perceived need for an understanding of the current research and clinical information about the patterns of the disorder and the treatment techniques. It is hoped that the material in this package will also stimulate interest in and discussion of the issues for the best management for boys with Duchenne muscular dystrophy.


Montrose Access


NSO
Non School Organisations
Commonwealth Special Education


People first
Heritage
Building Society


THE
IAN POTTER
FOUNDATION