Caring for children with muscular dystrophy

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This booklet was initiated and largely written by the late Ilse Langenhoven of the Gauteng Branch of the Muscular Dystrophy Foundation of SA. Ilse was born on 2 February 1966 and passed away on 4 June 2006. Ilse was an inspiration to everyone who crossed her path and knew her.

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CONTENTS

INTRODUCTION 1

WHAT IS MUSCULAR DYSTROPHY? 2
- Whom does MD affect? 2
- What causes MD? 2
- What are the symptoms? 3
- What treatment is available? 4
- What are the different types of MD? 5

THE CAREGIVER'S TASK 5

THE CHILD'S PHYSICAL NEEDS 6
- Diagnostic assessment 6
- How much to tell a child 6
- Ongoing medical assessments 6
- Encouraging physical development 8
- Ways of managing, not curing, the disorder 8
- The danger of bed rest 9
- The use of assistive devices 9
- Diet 9
- The danger of respiratory infections 10

THE CHILD'S NEED FOR INDEPENDENCE 10

THE CHILD'S EDUCATIONAL NEEDS 10
- An important question — why go to school? 10
- Planning for the child's changing needs 11

THE CHILD'S SOCIAL NEEDS 11
- Other children 11
- Play, leisure and recreation 12
- Other family members 12

CARER SUPPORT 13

REFERENCES 14

RESOURCES 15

INTRODUCTION

The aim of this booklet is to tell you about the kind of help you can get, and the professionals you will come into contact with, when you have a child with muscular dystrophy (MD). These professionals — such as social workers, your GP, a practice nurse or health visitor, or teachers and specialist helpers at your child's school — should be able to give you further assistance or different kinds of support as your child grows up. The diagnosis of a muscular dystrophy in all children and adults should be made by a specialist in the field — this is usually a paediatric neurologist or an adult neurologist. Ongoing care can be coordinated through the professionals listed above.

Muscular dystrophy (pronounced "dis-tro-fee") is a lesser-known disability that has a huge impact on people's lives. The picture of a wheelchair user who can still stand up or walk seems to confuse many people because of the popular belief that a wheelchair user is "wheelchair-bound" or "confined to a wheelchair".

The affected person usually seeks help first from general practitioners (doctors) and community nurses, but even these professionals may lack specialised knowledge, probably because so many different conditions fall under the term "muscular dystrophy". Also, general practitioners may come across only a few cases of MD in their entire career!

The term "muscular dystrophy" describes a disorder that causes progressive wasting and weakness of the muscles. Neuromuscular disorders affect not only the muscle but also the nervous system. Muscular dystrophy covers a wide range of neuromuscular disorders with similar symptoms to those dealt with in this booklet. The booklet tries to provide useful information on MD in general as well as specific information on Duchenne MD in particular (DMD), as it is the most common form of neuromuscular disease in young male children and has a profound effect on them and their families.

Do remember that there is help at hand. Contact the Muscular Dystrophy Foundation of South Africa (MDF), which is able to provide information on the condition and can help to put you in touch with other families whose children have the same condition.

WHAT IS MUSCULAR DYSTROPHY?

Muscular dystrophy is a relatively rare genetic disorder that affects muscles throughout the body. There are more than 20 forms of muscular dystrophy. Most of them lead to reduced muscle strength, but the various types affect different muscles in the body and degenerate differently. The most common form of muscular dystrophy is Duchenne MD, a less severe form of which is Becker muscular dystrophy.

Whom does MD affect?

Men, women and children of all ages and races can be affected by MD. The extent of muscular dystrophy in South Africa is unknown as there are no comprehensive epidemiological studies. Duchenne MD affects boys only.

What causes MD?

These disorders are usually inherited. The defective gene responsible for the weakening of the muscles can be passed on from one generation to the next. The genes typically code for a particular protein important for the muscle to work — loss, reduction or absence of these proteins results in impaired muscle function and breakdown. MD can also occur in families that have no prior history of the condition.

What are the symptoms?

Symptoms may be present from birth or develop in early childhood or later in life. Progressive muscle weakness is the main symptom, leading to secondary effects such as fatigue, increasingly limited physical activity, impaired balance and frequent falls.

Routine activities like climbing stairs, running or standing may cause legs to feel tired; and getting up from a chair may require substantial use of the arms. The level of strength may vary from one day to another.

Another symptom may be lordosis, or *curvature of the spine*, with the bend towards the front (the opposite of hunchback). Scoliosis is a form of spinal curvature that involves a sideways bend, which may require surgery to straighten. Severe scoliosis can restrict breathing and use of the upper limbs.

An early symptom of DMD is *enlarged calf muscles* due to the damaged or missing dystrophin gene, which results in a fatty enlargement of the tissues.

Joint and tendon restriction ("contracture") is another symptom that can progressively affect the ankles, hips, knees and finally upper limbs. Exercises can be done to stretch the joints and improve elasticity. Young men with DMD commonly have foot problems owing to contractures that pull their feet into unnatural positions and affect their ability to walk.

Intellectual disabilities may occur in a small number of males with DMD. These are usually minor learning disabilities, which do not deteriorate with time. Language and communication skills may be limited, but manual and visual skills are often good. Creativity is another typical strength.

Children with DMD may experience *respiratory difficulties* after the age of 10, owing to a weakening of the diaphragm. Impaired

breathing can lead to headaches, drowsiness and inattentiveness. Also, the muscles that are used in coughing grow weaker, which can lead to bronchial infections and pneumonia. Therefore, as respiratory muscles become weaker, it is important to clear the airways of secretions.

What treatment is available?

Although there is no cure yet, the right care can ensure a good quality of life and increase the young person's chances of being productive and fulfilled.

Physical therapy is very important for all kinds of muscular dystrophy and may include active movement, passive stretching and hydrotherapy. Adaptive seating and equipment can also help to reduce physical deterioration.

Braces and standing frames may be helpful in walking and standing. Braces are typically used on the knee, ankle and foot and can reduce contractures. They are sometimes worn at night to keep the limbs and feet in a normal position. Standing and walking for an extended period every day is beneficial.

When a *wheelchair* becomes necessary, it should fit the requirements of the child and the caregiver. Wheelchairs give the child greater mobility and stability. Other useful assistive devices are mechanical lifts, shower chairs, and beds that aid the changing of position.

Medication may be used to slow muscle loss, improve muscle strength and functioning, and prolong the ability to walk. More is said about this in the section on medical assessments later in this guide.

What are the different types of MD?

The outlook for people with MD varies depending on the type and severity of the disorder. In mild cases the disorder may progress slowly, and the person may have a normal lifespan. In more severe cases, the progression of muscle weakness, functional disability and loss of mobility is more marked.

The most well-known and aggressive type is Duchenne MD, in which the disorder progresses rapidly and life expectancy is curtailed. Other types, such as Facioscapulohumeral MD and Limb-Girdle MD, usually progress gradually but this depends on individual factors.

THE CAREGIVER'S TASK

Many disabled children depend heavily upon family members to meet their basic care and treatment needs, most of which do not diminish as the child grows older:

- Eating and drinking — these may include specialist food requirements depending on individual needs
- Using the toilet
- Bathing
- Communication — includes speech difficulties and the ability to express moods and needs
- Mobility — some children cannot walk independently or sit up without assistance
- Maintenance of medical regimes — for DMD this would be steroids and cardiac prophylaxis
- Home exercise programme (physiotherapy)
- Meetings with consultants, health visitors, physiotherapists, occupational therapists, speech therapists, clinical psychologists, educational psychologists and a host of other professional workers — these meetings can use up a lot of time and energy for both carer and child
- Planning of school or education placement

THE CHILD'S PHYSICAL NEEDS

Diagnostic assessment

To diagnose your child's disability or condition, diagnostic assessment should ideally be performed through a specialist unit that manages muscular dystrophy on a regular basis. The referral may be made by your general practitioner (GP) or paediatrician, who should continue to be involved and follow the recommendations from the specialist unit.

How much to tell a child

It is certainly difficult to know how much to tell a child with a progressive (and in some cases fatal) disorder. Once such children recognise the pattern their life is taking, it is certainly important for parents, teachers, doctors, counsellors and perhaps other advisors to talk to them privately about it, and show that they recognise the nature of the problem and are willing to talk frankly about how to deal with it. These children should never be confronted with information they have not asked for — they should feel confident in their carers and be given the opportunity to talk openly with them about their concerns regarding the information given, and they should have access to a psychologist for regular talks as they grow older.

Ongoing medical assessments

To monitor your child's health, ongoing medical assessments are made either directly by the specialist unit (usually run by a paediatric neurologist) or by your GP, hospital consultant and school nursing staff following directions from the specialist unit — usually at least every six months.

You may need the services of:

- a child development centre or unit — to assess your child's developmental needs and organise therapy or services if appropriate
- a physiotherapist
- an occupational therapist
- a speech therapist
- a dietician
- a health visitor
- a community dentist or dental nurse
- a continence adviser (if bladder and bowel control are weak)

You may also need:

- nappies or incontinence aids
- a buggy or a wheelchair (ideally electric) which has been carefully measured and adjusted to fit your child correctly

Medication may be prescribed in some types of muscular dystrophy.

Surgery is sometimes used to release contracted muscles. However, regularly performing hamstring and Achilles tendon stretches can avoid invasive interventions. Some children with MD may require a limb to be *splinted* in order to prevent the development of deformities and to optimise function. Progressive scoliosis may require surgical interventions if the curvature is likely to lead to impaired lung function. Maintaining walking into the main pubertal growth spurt tends to reduce the need for this.

However, *anaesthetics*, if administered, can negatively affect heart function, blood circulation, body temperature and breathing, with potentially damaging consequences. A mechanical respirator may be used during anaesthetic procedures, but the muscle-relaxing drugs that are used may slow the recovery of respiratory function afterwards, among other dangers. Therefore careful testing of the patient has to be done before any anaesthetic procedures are

performed. (An article on this topic by Margaret Vroom is listed in the references at the end of this guide. Your doctor should be advised to consult the article before undertaking any anaesthetic procedures on a child with muscular dystrophy.)

Encouraging physical development

Encouraging physical development is an important part of any child's education. The child should take part in physical activities, though some of these may need to be modified.

Children should be encouraged to be as active and mobile as possible, for as long as possible, but should not be over-exerted.

Swimming is an excellent form of exercise for people with MD as it helps maintain muscle strength, joint range of motion, and cardiovascular fitness. In addition, the buoyancy of the water helps support the body and may be used to exercise limbs too weak to lift against gravity. The warmth of a hydrotherapy pool aids in relaxation and soothes sore muscles.

Ways of managing, not curing, the disorder

Flexibility training and passive exercise can be used to manage the disorder (but not to cure it). These may involve the following:

- Range-of-motion and stretching exercises. These prevent the development of contractures (shortening of the tendons and muscles, which results in deformities and loss of function). These passive exercises may also help in relieving painful muscle spasms. Mobilising the shoulders also maintains a good breathing pattern and prevents postural deformities which might affect the respiratory system.
- Moderate-resistance strengthening exercise (but strenuous exercise should be avoided).
- Aerobic exercise when the disability allows.

- Breathing exercises to encourage correct breathing patterns and to improve lung expansion.
- Speech therapy for impaired speech and difficulty swallowing.
- Massage therapy.

The danger of bed rest

Staying as active as possible is important, and therefore bed rest should be avoided as far as possible.

The use of assistive devices

Children with MD may require one or more assistive devices in order to optimise their mobility and help with activities of daily living.

Such devices include:

- walking frames and rollators (rolling walkers with a seat)
- electric wheelchairs (children using wheelchairs should have sufficient trunk muscle strength to be able to sit in a good position with minimal support, otherwise there is a risk of developing truncal deformities such as scoliosis)
- specialised back supports
- buggies

Diet

It is common for children with muscular dystrophy to become overweight. This is a very trying problem for those who are wheelchair users. They tend to put on weight partly because of overeating and partly because of an unavoidable lack of muscular activity to "burn up" the excess intake.

The intake of carbohydrate foods should be restricted, and protein intake should be improved.

The danger of respiratory infections

When the muscles involved in breathing are affected by the disorder, apparently trivial coughs and colds can lead to much more serious respiratory infections. It is therefore important to take them seriously. Accordingly, family doctors are likely to prescribe antibiotics for seemingly slight infections in MD-affected individuals, when this would be unnecessary for most people. Preventative measures such as the yearly influenza vaccination are advised.

If your child's strength of coughing is reduced, you must learn how to help clear secretions from the lungs. Ask your physiotherapist to show you how to do this safely and gently.

THE CHILD'S NEED FOR INDEPENDENCE

Independence in young children comes gradually as they learn to do things without adult help. However, a progressive neuromuscular condition may cause children to lose the ability to do things by themselves. Consequently, their striving for independence will be counterbalanced by their need for well-judged assistance.

THE CHILD'S EDUCATIONAL NEEDS

An important question — why go to school?

A frequent question asked in connection with DMD-affected children is: "If they're going to have a short life, what's the point of going to school and working hard? Surely we need to make their life as enjoyable as possible?" This is a very understandable attitude, but a far more positive question would be: "How do we ensure that these individuals have experiences that add to their quality of life?" This can be achieved by having the same life experience and same expectations as other children. As a result of positive thinking and an independent spirit, some males with DMD are cur-

rently studying at university, and others have jobs. It is essential for children with any type of muscular dystrophy or neuromuscular condition to aim high academically so as to compete in the job market and make the most of skills they can build on.

Planning for the child's changing needs

Children with muscular dystrophy entering school may not seem physically different from other children. However, the pace of deterioration can vary, and adaptations should be made at the school as and when necessary. Decisions should be made about whether to educate children at special or mainstream schools.

Computers, for example, can offer many new opportunities for children with special needs, especially if writing is difficult or slow for them owing to weakness in the arm and hands.

The following types of contact should also be made:

- Contact with the child's physiotherapist or occupational therapist or the MDF
- Contact with a family counsellor
- Contact between parents and school staff for additional support (schools will need to decide who should take on this role, for example the educational psychologist or child guidance worker)

The child's physical management must include a programme of regular exercises to help prevent later problems (especially for boys with DMD). These exercises may need to be carried out daily.

THE CHILD'S SOCIAL NEEDS

Other children

Working towards social integration is probably the most difficult part of the integration process. It is not enough to place a child

in a mainstream school (physical integration), make arrangements for access to the curriculum (academic integration) and then regard the integration as complete.

The earlier that children with a disability can be integrated into school, the earlier other children will accept them as equals. As muscular dystrophy is a progressive condition, it may take some time before other children realise their friend is different in some way. Good adult attitudes and responses are very important in fostering positive attitudes in children. The key is to answer questions simply and honestly but to give only the information that the children want.

Later on, if it becomes clear that the child is losing mobility or needs splints, it may be more of a challenge to answer questions. This will be especially true if parents have not discussed the condition with the child.

Affected children who require more adult support may find it difficult to get involved with other children in the way they would like. There must be a plan whereby children can be as independent as possible, especially at break times.

Play, leisure and recreation

All children need the chance to play.

If you have a child with a disability or health condition, you need to think about the types of play opportunities your child could be interested in.

Other family members

Having a child with a disability affects the whole family, including brothers, sisters, aunts, uncles and grandparents. This often leads to many members of a family needing information and support, not just the parents.

Siblings of disabled children often feel that, because of the additional care needs of their brother or sister, not enough time is spent on their needs. Everyday family activities may be limited, as parents try to juggle the needs of all the children in the family, and this can lead to additional pressures at home. Other friends and relatives could play an important role here by offering some special time for the brothers or sisters, or including them in activities they would otherwise miss out on.

CARER SUPPORT

Those who care for people with muscular dystrophy can benefit from finding a support group with whom to share information and encouragement. Common interests include health and education, care provision, finances, the concerns of other family members, the challenges that come as children with MD mature, and where to obtain resources and other help.

The list of references and resources on the following pages may provide further useful information and inspiration.

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RESOURCES

In addition to the sources in the preceding list of references, you could consult the following:

Factsheets of the Muscular Dystrophy Campaign. http://www.muscular-dystrophy.org/information_resources/factsheets/index.html
At this site there are three main categories of factsheet: "medical issues", "medical conditions" and "daily living issues". Some of the "medical issues" factsheets for people with neuro-muscular conditions are listed below:

- Achilles tendon release
- Anaesthetics
- Carrier detection tests and prenatal diagnosis
- Gastrostomy
- Heart check
- Inheritance and the muscular dystrophies
- Making breathing easier
- Muscle biopsies
- Newly diagnosed?
- Questions about ventilation to ask your consultant
- Risks of dehydration (lack of water)
- Surgical correction of spinal deformity in muscular dystrophy and other neuromuscular disorders
- Ventilation — questions to ask your consultant
- Alternative therapies
- Guidelines for exercise and orthoses in children with neuromuscular disorders
- Steroids and Duchenne muscular dystrophy — some questions and answers
- Nutrition and feeding in individuals with neuromuscular conditions
- Spinal surgery — questions to ask
- Pressure ulcers

Parent Project Muscular Dystrophy. *Giving a face to DMD: Understanding the disease and guidelines for care and management*. Double DVD set: the first DVD gives an overview of Duchenne muscular dystrophy, its cause and how a diagnosis is reached; the second DVD gives more information on the care and management of Duchenne. http://www.parentprojectmd.org/site/PageServer?pagename=nws_dmd_dvd040107. You can order this DVD set from your local branch of the Muscular Dystrophy Foundation.

Siegel, Irwin M. [Undated]. *Everybody's different, nobody's perfect / Elke mens is anders, niemand is volmaak nie*. A booklet of the Muscular Dystrophy Association, USA, reprinted in English and Afrikaans by the Muscular Dystrophy Foundation (MDF) of South Africa. For younger readers.