Q1. What is muscular dystrophy?

A. Muscular dystrophy is the name applied to a group of diseases that are, for the most part, genetically determined and that cause gradual wasting of muscle with accompanying weakness.

Q2. Are these conditions contagious?

A. No.

Q3. What is the common kind of muscular dystrophy, the type that affects young boys?

A. This condition is called Duchenne muscular dystrophy (DMD). Symptoms usually appear between the ages of two and six. It was first described by Duchenne, a French physician, in 1861. More information is available in our Duchenne muscular dystrophy factsheet. [http://www.mda.org.au/Disorders/Dystrophies/DMD-BMD.asp]

Q4. What are the first clinical signs of DMD?

A. The first signs of weakness usually are delayed onset of walking, difficulty in performing a standing jump, a waddling gait and difficulty in rising from the floor.

Q5. What are some of the later clinical signs of weakness in DMD?

A. Later signs of weakness include difficulty in rising from a chair, difficulty climbing stairs and a wide gait with difficulty in balance.

Q6. How is muscular dystrophy transmitted?

A. Muscular dystrophy is a genetic condition that, in most cases, is inherited from a parent (or parents). Humans have about 20,000 genes which contain the instructions to make and maintain our bodies, and errors in certain genes cause muscular dystrophy. We inherit two copies of each gene – one from each parent. For some conditions having an error in one gene is enough to cause muscular dystrophy whereas for others both genes need to have an error.

For example, in most types of limb-girdle muscular dystrophy, both parents carry a gene with an error, although neither has the disease. Children of these parents have a one in four chance of inheriting two copies of the affected gene (one from each parent) and developing the condition. This type of muscular dystrophy can be transmitted to either a male or female child. The technical term for this inheritance pattern is "autosomal recessive".
In facioscapulohumeral dystrophy (FSHD) and myotonic dystrophy, one faulty gene is enough to cause the condition. One parent has to have the disease in order to transmit it (autosomal dominant inheritance pattern). This type can also be inherited by a male or female. Sometimes the parent might be so mildly affected by the condition that they don’t know they have it until much later in life.

In Duchenne and Becker muscular dystrophies, the affected gene is carried by the mother and the disease is transmitted to her son (recessive X-linked inheritance pattern). Each male child born to such a mother has a 50 percent chance of inheriting the disease, and each female a 50 percent chance of becoming a carrier.

Sometimes muscular dystrophy isn’t inherited from the parents, instead the genetic mistake arises in the affected child themselves and then it is known as a ‘spontaneous mutation’. Up to a third of DMD and FSHD cases are the result of spontaneous mutations.

Q7. Is muscular dystrophy always hereditary?
A. No. In approximately one-third of DMD cases the genetic fault is not inherited but arises in the affected boy himself and then it is known as a ‘spontaneous’ or ‘sporadic’ mutation. It is also estimated that 10 to 30 percent of people with FSHD cases have spontaneous mutations.

Q8. What are the chances of a mother without a family history of DMD having a son with the disease?
A. In the population at large, the risk of DMD in a male child is about 1 in 3,200 live male births. The risk of a sporadic (non-hereditary) case is approximately 1 in 12,000.

Q9. Can tests be done to find out if the mother or sister of a boy with DMD is a carrier of the condition?
A. Yes DNA testing can be done. This is easiest if it is known specifically what DNA mutation the family member with DMD has.

Sometimes levels of creatine kinase (CK) in the blood can give an indication of carrier status. CK is normally found in healthy muscles, but if the muscles are diseased or damaged (e.g. in Duchenne and Becker muscular dystrophy) large amounts of CK also leak into the blood, and can be measured. Usually, three blood samples are taken from a person who might be a carrier of Duchenne or Becker muscular dystrophy, over a period of several weeks and the CK level is analysed and averaged to give a result. CK estimation is usually not reliable enough to use as a test in its own right, and is generally carried out in conjunction with other tests.

Q10. If a known carrier decides to have her own children, is there some way of determining whether or not she is carrying a child with DMD?
A. Different types of prenatal tests can be carried out after about 10 weeks of pregnancy. Chorion villus sampling (CVS) can be done at 10-12 weeks, and amniocentesis at about 14-
16 weeks. Women/couples need to consider carefully which test to have and to discuss this with their genetic counsellor - ideally before becoming pregnant.

Q11. If you are a carrier of a type of muscular dystrophy, is there a way to prevent it being passed onto future children?

A. Yes, preimplantation genetic diagnosis (PGD) is available for some conditions. This involves using IVF to fertilise eggs outside the body and then testing the resulting embryos for the genetic mutation. Unaffected embryos are then chosen to be placed back in the woman’s womb. Another option is prenatal diagnosis – testing the foetus once the woman is pregnant. The couple then has the option whether or not to continue the pregnancy if the foetus is affected. Alternatively some couples may choose to use egg or sperm donors, or adopt a child.

Q12. Is muscular dystrophy anyone's fault?

A. No. Muscular dystrophies are genetic diseases. Forms of muscular dystrophy can be passed on from generation to generation, or they can occur spontaneously in a single individual as the result of a mutation of a particular gene. In any case, they are not anyone's fault.

Q13. What medical professionals should I enlist to help me manage my condition?

A. Muscular dystrophy can affect you in many different ways so a multi-disciplinary team of professionals is required to optimally manage the symptoms of the condition. A neurologist usually heads the team and team members may include a cardiologist, respiratory specialist, physiotherapist, occupational therapist, speech therapist, specialist nurses, social worker, psychologist/counsellor, dietician, geneticist/genetic counsellor, gastroenterologist and orthopaedic surgeon. Depending on your condition and what stage you’re at, you may only need some of these, and some will only be seen very occasionally. However, some professionals, such as a physiotherapist may need to be seen on a regular basis. Sometimes a neuromuscular coordinator is available to help you manage appointments with all of these people and can help you see them in one visit to the hospital.

Q14. Is there a late-occurring type of childhood muscular dystrophy?

A. Yes. Becker muscular dystrophy is a milder form of DMD with onset between the ages of 5 and 25 and relatively slow progression. There are also many other types of muscular dystrophy with onset later in childhood or in adulthood.

Q15. Is muscular dystrophy strictly a disease of children?

A. No muscular dystrophy and related neuromuscular conditions affects people of all ages. For example, limb-girdle dystrophy usually has its onset in late adolescence, and facioscapulohumeral and myotonic types usually start in adulthood. These forms of muscular dystrophy are slower in their progression and less disabling than the childhood forms.

Q16. What are some of the early signs of adult muscular dystrophy?
A. A weak smile and inability to pucker the lips or whistle occur early in facioscapulohumeral dystrophy. Subtle weakness in the shoulders and hips occurs early in limb-girdle dystrophy. Weakness of the feet and hands and difficulty in relaxing the grip are early signs of myotonic dystrophy.

Q17. What is the incidence of muscular dystrophy and related neuromuscular conditions?

A. The incidence of muscular dystrophy and related neuromuscular conditions is estimated to be 1 per 1,000 head of population. Based on current figures, it is estimated that there would be approximately 30,000 people in Australia who have some form of neuromuscular disease. DMD and spinal muscular atrophy (SMA) are the most common types affecting children estimated to affect 1 in 3,500 live male births and 1 in 6000 live births respectively. Myotonic dystrophy is probably the most common type affecting adults - 1 in 8000 people. Facioscapulohumeral muscular dystrophy (FSHD) is also relatively common affecting 1 in 14,000.

Q18. In what part of the world is muscular dystrophy most common?

A. In general there is no area of the world where muscular dystrophy is more prevalent than anywhere else. There are however some extremely rare types of muscular dystrophy that are more common in some areas, for example, Welander distal myopathy is more common in Sweden and Finland.

Q19. What percentage of body weight is made up of muscle tissue? How much muscle tissue has to be lost before function is affected?

A. Muscle makes up 40 percent of the total body weight. Approximately 33 percent of muscle mass has to be lost for function to be impaired in large postural muscles.

Q20. What muscles are affected in muscular dystrophy?

A. Different muscles are affected in different forms of muscular dystrophy. Although any of the 434 voluntary muscles can be affected, muscles around the spine and that of the limb-girdles (shoulders and hips) are involved most frequently and, when involved, are affected earliest and most profoundly.

Q21. Can DMD be diagnosed at birth?

A. The clinical signs of muscular dystrophy (weakness and wasting) are usually not apparent until the child is at least several years of age. However, an elevated blood level of the muscle enzyme creatine kinase (CK) is detectable at birth, long before clinical weakness is evident.

Q22. How can a young girl have DMD?

A. DMD occurs in approximately 1 in 3500 male births, and in about 1 in 50,000,000 female births. DMD is extremely rare in females because they have two X chromosomes and usually the unaffected dystrophin gene on the second chromosome can compensate for the faulty one. Note: males have an X and a Y chromosome so they don’t have a second X chromosome to compensate.
A girl could have DMD if for some reason the X chromosome that carries the healthy copy of the dystrophin gene is switched off. This prevents it being used to make dystrophin in the muscles. This is called ‘X-linked inactivation’. Depending on the degree of this X-linked inactivation, their progression could be the same as a boy with DMD, but usually their condition is more mild.

Alternatively, if a young man with DMD had a child with a woman who was a carrier of DMD, it is possible that they could have a daughter with DMD. This is very rare since men with DMD usually have poor health by the time they could have a child and it would be unlikely that their partner would also be a carrier.

Female carriers of DMD can have mild symptoms of DMD that progress very slowly – these women are called manifesting carriers.

Q23. Is muscular dystrophy a painful disease?

A. Pain is not usually a primary symptom of muscular dystrophy but sometimes the muscle weakness can put strain on joints or cause poor posture which causes pain. Surveys have shown that pain is particularly a problem for people with FSHD but the cause and treatment of this pain is poorly understood.

Q24. What are the signs that a person with muscular dystrophy needs to use a ventilator to help with their breathing?

Signs of weakening respiratory muscles are headaches, difficulty sleeping at night, excess sleepiness during the day, poor concentration, and chest infections. Portable, effective ventilation devices are available which can greatly improve quality of life. These are usually only needed at night.

It is helpful if breathing problems are detected and treated early; so patients with MD will be offered regular 'lung function tests' once they start to have significant muscle weakness. These are simple tests which can monitor the strength of the breathing muscles and the level of oxygen in the blood.

Q25. Is arthritis a part of muscular dystrophy?

A. No, although patients with muscle disease can also have arthritis.

Q26. Is there any treatment for muscular dystrophy?

A. No treatment has yet been found to correct the underlying cause or to stop the progression of the disease, but several promising treatments are now being tested in clinical trial. Corticosteroid medication can delay the onset of some of the symptoms of DMD by several years. Supportive and symptomatic aids are available; and care by a team of medical professionals can improve comfort, mobility, and even life expectancy.

Q27. Can muscular dystrophy be diagnosed by X-ray?

A. No, although certain changes (e.g. thinning of bones and increased soft tissue shadows of calves) occur secondarily to the disease.
Q28. Is the heart affected in muscular dystrophy?

A. The heart is a muscle, and heart disease can be a complication many forms of muscular dystrophy. It is common in DMD, and may be seen in limb-girdle dystrophy. It also frequently occurs in myotonic dystrophy.

Q29. Do people with muscular dystrophy have intellectual disabilities?

A. The incidence of intellectual disability is no greater in the muscular dystrophy population than in the total population. However some learning and behavioural difficulties can be present in a minority of boys with DMD. With proper help and support these difficulties can be managed. Some severe cases of myotonic dystrophy can involve some level of intellectual disability.

Q30. What causes deformity in muscular dystrophy?

A. Muscle weakness can cause muscles to shorten (contracture) and imbalance which can result in deformities of the foot in particular. This deformity can be prevented or delayed with the use of special footwear, surgery and with the support of a physiotherapist. Muscle weakness around the spine can also cause scoliosis (curvature of the spine). Surgery is often done to correct this.

Q31. Do people with neuromuscular conditions get tired more easily?

A. Yes, fatigue is often associated with muscular dystrophy and other related conditions. The cause is not fully understood but often stronger muscles are used to compensate for weaker ones which results in them tiring quickly. With some types of muscular dystrophy the heart is weakened which also contributes to fatigue. In myotonic dystrophy the brain is often affected resulting in fatigue and daytime sleepiness. People with myasthenia gravis are particularly susceptible to muscle fatigue due to the way the condition affects the transmission of signals from the nerves to the muscles.

Q32. Is muscular dystrophy a progressive disease, or are there times when it stops for a while and starts again?

A. The weakening process in muscular dystrophy is a continuous one, although there may be times when the processes of normal growth and development seem to overtake the disease and the patient appears to have gained strength because of this.

Q33. Does muscular dystrophy affect hearing, eyesight, or speech?

A. In general muscular dystrophy does not damage eyesight. However, cataracts sometimes develop in myotonic dystrophy and damage to the retina of the eye is occasionally associated with FSHD, and in infantile onset FSHD this may progress to an eye condition called ‘Coat’s disease’ which can cause significant loss of sight if not treated. There are also some rare conditions where the muscles that move the eye are involved - such as oculopharangeal muscular dystrophy (OPMD). Hearing is usually unaffected except in infantile onset facioscapulohumeral dystrophy (FSHD). Speech may be impaired in FSHD where there is weakness of those facial muscles controlling speech.
Q34. Are the muscles of the hands affected in muscular dystrophy?

A. It depends on the type of muscular dystrophy. Some types such as distal myopathy and myotonic dystrophy do affect the hands. In other types such as Duchenne muscular dystrophy the large muscles of the legs and pelvis are affected first and the hands are much less severely affected.

Q35. If there is no known cure for muscular dystrophy, why is it important that diagnosis be made early?

A. For three main reasons. Firstly, so that links can be made with a neuromuscular specialist ready for if and when a treatment becomes available. Secondly, so that medical, psychological and social needs can be anticipated, planned for and addressed quickly as they arise. Early intervention is key to successful treatment of some of the symptoms and complications of the conditions. Third, so that parents and female relatives can receive genetic counselling concerning risk in future pregnancies.

Q36. Of what use are blood muscle enzyme determinations in the diagnosis of muscular dystrophy?

A. An elevated muscle enzyme (e.g. CPK, adolase, L.D.H.) indicates that muscle cells have been damaged. Typical elevations are found in various muscle diseases, including the muscular dystrophies. Genetic testing is generally required to confirm the diagnosis though.

Q37. What is electromyogram (EMG)?

A. An electromyogram is an examination performed by inserting very fine needle electrodes into selected muscles and recording and interpreting the electrical patterns that they produce at rest and during muscular contraction. Typical patterns may be diagnostic of various diseases of muscle or nerve.

Q38. What is a muscle biopsy?

A. A muscle biopsy is a minor procedure in which a small piece of muscle is removed under general or local anaesthesia, sliced very thinly, treated with a variety of stains, and examined under a microscope for changes that are typical of various diseases of muscle or nerve.

Q39. Do patients with muscular dystrophy sometimes feel stronger on some days and weaker on others?

A. Yes, like other people, muscular dystrophy patients have good and bad days.

Q40. Is muscular dystrophy primarily a disease of muscle, or are the nerves also affected?

A. Muscular dystrophy is primarily a disease of muscle but some other related neuromuscular conditions such as Charcot-Marie-Tooth disease affect the peripheral nerves; others such as amyotrophic lateral sclerosis (ALS) and spinal muscular atrophy (SMA) affect the motor neurons. Myasthenia gravis affects the structures at the junction between the nerves and the muscle.
Q41. Is the bowel or urinary bladder involved in muscular dystrophy?

A. Constipation sometimes occurs in people with reduced mobility. Otherwise the bowels are rarely affected in diseases of muscle. Similarly, function of the urinary bladder is normal.

Q42. Is there any correlation between the age of onset of muscular dystrophy and its progression?

A. As a general rule it can be said that the earlier clinical symptoms appear, the more rapidly the disease progresses. There are always exceptions to the rule though and some of the congenital muscular dystrophies are known to progress very slowly.

Q43. Do acupuncture, electrical stimulation, psychic-healing, detoxifying the body through colonic irrigation and cleansing through fasting, massage, hydrotherapy, or hot and cold alternating baths or showers help in the treatment of muscular dystrophy?

A. Generally such measures do not help the treatment of muscular dystrophy; however, many individuals have improved their level of comfort, mobility and dexterity through massage, hydrotherapy and physiotherapy.

Q44. Why is it important to correct foot instability in children with DMD?

A. Contracture in weight-bearing joints can interfere with balance and impede walking so correction is very important to maintain mobility.

Q45. Is there anything to help muscular dystrophy patients breathe better?

A. Breathing can be helped through improving posture, through diaphragmatic breathing exercises (which can be taught by a physiotherapist), and through the use of certain non-invasive ventilator equipment prescribed by the doctor.

Q46. Why is it dangerous to keep a child with muscular dystrophy at bed rest for more than a day or so?

A. Children with DMD suffer rapid acceleration of weakness when they are kept in bed for more than a day or so. It is usually unnecessary to keep these children at absolute bed rest during the treatment of the common cold or the ordinary childhood diseases.

Q47. What is postural drainage?

A. A technique to help clear the airways by assuming a variety of positions in which mucus is easily drained from the chest. Postural drainage can be taught by a physiotherapist.

Q48. Why is it important to measure the strength and functional ability of a child with muscular dystrophy at regular intervals?

A. So that subtle changes that indicate progressive weakness can be detected early and appropriate treatment started.
Q49. What are diaphragmatic breathing exercises and intermittent positive pressure breathing (IPPB)?

A. Diaphragmatic breathing exercises train the patient in the use of the diaphragm for breathing. They should be taught to all patients suffering loss of breathing capacity because of chest muscle weakness. IPPB treatment involves the use of portable therapy unit that aids the patient in ventilating the lungs and removing bronchial secretions. Its operation is simple and can be easily mastered in a few minutes time.

Q50. What is Glossopharyngeal Breathing (GPB) (also called Stacking or Frog Breathing)?

A. Frog breathing, or technically speaking, glossopharyngeal breathing (GPB) is a learned skill that can be used as a substitute (voluntary) method of breathing. Frog breathing involves using the tongue and throat muscles in a pumping mechanism to force air into the lungs. It can produce adequate ventilation for either short or long periods of time even when there is a total paralysis of the respiratory muscles.

Q51. Are exercises helpful in the treatment of muscle disease?

A. Stretching exercises are helpful in preventing muscle contracture and subsequent joint deformity. Aerobic exercise is important to keep the muscles and cardiovascular system healthy for as long as possible. However, it is important to do exercise in moderation; too much exercise or the wrong type of exercise can cause additional muscle damage. Swimming or hydrotherapy is a good way to exercise without putting undue stress on muscles. The buoyancy of the water gives great assistance to weak muscles and breath holding underwater allows exercising of the respiratory muscles. This form of exercise can be enjoyed long after walking is too difficult.

Q52. What can be done to help a child with DMD who finds increased difficulty with walking?

A. Surgical release of contracted muscles followed by light plastic bracing can enable some children to continue walking independently for a longer period of time. A manual wheelchair may be required for longer distances though.

Q53. Why do some children with DMD walk with a swayback, wide-based gait?

A. Because of a selective weakening of certain muscle groups, with secondary over pull of others. This can be helped by daily stretching of those muscles that tend to contract.

Q54. What can physiotherapy offer children with muscular dystrophy?

A. The prevention and / or correction of muscle contractures, the effective use of residual strength and increase in the efficiency of functional activities such as standing, walking, and transfer can all be improved by physiotherapy.

Q55. What is the difference between active and passive exercise?
A. Active exercise is exercise performed by the patient. In passive exercise, the patient's body is manipulated in stretching exercises by the therapist or parent.

Q56. Why is it important to keep the heel cords well stretched in a child with muscular dystrophy?

A. In order to maintain alignment support in the body. In a child with DMD, severe contractures of the heel cord can make the differences between walking and not walking.

Q57. Are any special diets or vitamins helpful in the treatment of muscular dystrophy?

A. Patients should be encouraged to eat a well-balanced diet and to avoid becoming overweight. Undernourishment can also become a problem if there are swallowing difficulties. Nutritionists/dieticians are important members of the healthcare team: they can make sure the diet is balanced and help with eating plans.

For boys with Duchenne muscular dystrophy a daily multivitamin with vitamin D and minerals is recommended. This is especially important for those who are taking corticosteroids and in this case, monitoring of vitamin D and calcium levels may also be required.

Q58. Why do so many people with muscular dystrophy become obese, and how can this be prevented?

A. Obesity occurs because food intake is in excess of the reduced energy demands of people with limited mobility who use a wheelchair. Corticosteroids which are often prescribed to boys with Duchenne muscular dystrophy also contribute to weight gain. People with a chronic medical condition also often overeat to deal with depression or anxiety and parents/friends/family/carers also often feel the need to treat them with unhealthy food. Being overweight can put extra stress on the muscles so a carefully controlled diet with the help of a dietician/nutritionist is important.

Q59. What are some of the aids available to help in the care of a child with muscular dystrophy?

A. A variety of self-help aids are available, including raised chairs and toilet seats, grab bars, bath tub benches, high stools, clothing adaptions (utilising special zippers on trousers, easily manipulated velcro fastenings, etc.) footboards to diminish heel cord contracture and electric, hydraulic, or mechanical lifts to transfer to and from bed and bath.

Q60. Can patients with muscular dystrophy receive routine immunisations and be treated with the usual medications and/or surgery for conditions unrelated to muscular dystrophy?

A. This is best discussed with your specialist but in general children with muscular dystrophy should receive their routine shots, including flu shots. Care must be taken when undergoing surgery to make sure that the anaesthetic drugs used are safe for people with that particular condition and that the patient’s respiratory health is carefully monitored.

Q61. Are all neuromuscular conditions the same?
No, neuromuscular conditions have a wide variety of causes and can affect people in many different ways at any age.

Muscular Dystrophy Australia supports people with any one of more than 60 different neuromuscular conditions. Some neuromuscular conditions are so severe that babies die within the first few years of life, whereas others only affect people in old age and don’t cause severe disability. Most of the conditions have a genetic cause and are inherited within families, the rest are autoimmune conditions or the cause is unknown or poorly understood.

The source of the muscle weakness within the body differs too. For example, muscular dystrophy is primarily a disease of muscle, however, for some other neuromuscular conditions the muscle weakness originates in the nerves that control the muscles, the motor neurons or the neuromuscular junction.

Each neuromuscular condition causes a characteristic pattern of muscle weakness: it could be just the muscles around the eyes or the large muscles in the legs, or just one side of the body. The most severe conditions affect all the muscles of the body including the heart and respiratory muscles. Some conditions such as myotonic dystrophy affect many parts of the body in addition to the muscles.

Q62. I’m interested in wheelchair football, how can I find out if it is for me?

Wheelchair football is played on an indoor basketball court with players using either electric or manual wheelchairs. Players are able to have support on court if needed to push their chair and can use their feet to kick the ball or chair to propel the ball forward. A lot of people with muscular dystrophy enjoy playing so if you’d like to find out about giving it a go contact Football Federation Victoria. [http://www.footballfedvic.com.au/index.php?id=1100]

Q63. Do parents of a child with muscular dystrophy often have emotional problems themselves?

A. Yes. Because of undue demands on their time and strength as well as normal feelings of anger and guilt that may arise, parents of children with muscular dystrophy often find themselves with psychological issues. These can usually be worked out through individual counselling or parent groups.

Q64. Who can help the family with psychological problems (such as feelings of anger and guilt) caused by the disease?

A. Referral to a qualified medical social worker, psychologist, or psychiatrist can be obtained from you doctor, or Muscular Dystrophy Australia can help point you in the right direction.

Q65. What should be expected of the child with DMD?

A. That he live as productively and as normally as possible while realistically recognising the specific limitations imposed by the condition. Honest expectations must be set for these children, as for anyone with a disabling condition. At the same time, one should avoid over-protection, allowing the child to accomplish tasks by his own efforts. Over-protection, rather than helping, leads inevitably to isolation and dependence.
Q66. How does one plan ahead (high school, university, job, etc.) for a child with muscular dystrophy?

A. All children with muscular dystrophy should be able to attend high school and many even attend university. Distance education is one option for tertiary study if travelling to a campus is not practical. Many pursue careers after graduation. Unfortunately, the aspirations and ambitions of disabled people are often limited by prejudice but legislation is place to try to prevent discrimination. Employers and educational institutions are obliged to make reasonable adjustments, which could include: allowing flexible working; part-time working hours; working from home; making adaptations to toilets; redesigning a job description.

It is important for both employers and disabled people to be open and upfront about what works best for them. Talking about challenges and potential ways to alleviate them is the best way to find a creative flexible working environment.

Q67. How should parents respond to a child's inquiries about the disabling and even fatal aspect of his or her disease?

A. Parents should give answers in words the child can understand that are realistic, hopeful, honest, yet as reassuring as possible. It is important to recognise exactly what the child is asking and respond to this specific request for information and not to one's own anxieties.

Q68. How and when does a parent tell other children in the family about a brother or sister who has muscular dystrophy?

A. If lines of communication in the family are kept open many potential problems can be avoided. Brothers and sisters of a child with muscular dystrophy should be told of his or her condition and its implications as soon as they are able to understand. Parents must realise that their unaffected children also have physical and emotional needs that must be met and avoid focusing all their attention on the child with muscular dystrophy.

Q69. Can a patient with muscular dystrophy participate in sports?

A. As long as strength permits. Because the buoyancy of water supports the body, most patients are able to enjoy water activities after weakness prevents participation in more vigorous sports.

Q70. What types of recreational activities are available to a child with muscular dystrophy?

A. Many activities are available including wheelchair sports – such as tennis, soccer and basketball. Boccia is a sport which can be played socially or competitively, and is designed for people with physical disabilities. Some children enjoy swimming, horse riding or indoor rock climbing. Organisations like Disability Sport and Recreation can help you find suitable activities. [www.dsr.org.au]

Q71. Should children with muscular dystrophy attend ordinary school?

A. Yes, most children attend mainstream schools. But of course you should evaluate the options available and decide what is suits your child the best.
Q72. What Social Security and other benefits can a muscular dystrophy patient obtain from the government?

A. Federal and State governments provide a number of benefits by way of services or financial assistance to people affected by a disability or for the primary carer. In some cases there are eligibility criteria that need to be met and you may not be eligible for each and every service.

You may be able to apply for taxi vouchers: http://www.taxi.vic.gov.au/passengers/mptp. Information on benefits and concessions such as companion cards can be found on the MDA website http://www.mda.org.au/Concessions/Index.asp/

If you need help with benefits and concessions contact the MDA (details at the bottom of this page).

Q73. Where can I find reliable information about DMD?

A. The MDA website has a DMD Portal called “MD-Net” that brings together authoritative, evidence based information which can be shared with your health care provider if necessary. For example, you might want the latest advice on spinal surgery or find out about stem cell research. [http://www.mda-net.md/]

Q74. Do children with muscular dystrophy develop pressure sores from sitting for long periods of time in a wheelchair?

A. People with muscular dystrophy who use wheelchairs have fewer problems with pressure sores than those with spinal injuries who have lost physical sensation. This is because they are able to feel the discomfort, and shift themselves or ask a caregiver to shift them. It is recommended that wheelchair users shift their bodies every 15 minutes to avoid sores. For people who can’t shift on their own, caregivers should help them reposition at least once every hour. There is also a wide range of wheelchair cushions available to reduce the risk of pressure sores.

Q75. Is spinal curvature (scoliosis) a frequent complication of muscular dystrophy?

A. Children with DMD usually do not develop spinal curvature until they are using a wheelchair full time. Scoliosis can occur in Becker's dystrophy even while still able to walk, and in the childhood forms of facioscapulohumeral dystrophy, congenital muscular dystrophy and myotonic dystrophy. Surgery may be necessary to correct scoliosis.

Q76. Can patients with muscle disease engage in sexual activity, and is such activity harmful to them?

A. Many patients with muscle disease engage in sexual activity, and is in no way harmful to them.

Q77. What is myotonic dystrophy? Can it be treated?

A. Myotonic dystrophy is a disease that usually affects adults but may have its onset in childhood. It is characterised by progressive weakness and muscle stiffness (myotonia).
Additionally, symptoms of this condition may include heart problems, cataracts, trouble breathing, adverse reactions to anaesthesia, difficulty swallowing, digestive problems, excessive daytime sleepiness and, in children, learning difficulties. No treatment is available for the weakness, but several drugs are effective in relieving the myotonic aspect of this disease. Please see the MDA website for a detailed factsheet about myotonic dystrophy. [http://www.mda.org.au/Disorders/Dystrophies/MYT.asp]

Q78. **What are polymyositis and dermatomyositis?**

A. Polymyositis is an inflammatory disorder of muscle. It occurs in acute and chronic forms. When accompanied by a rash it is called dermatomyositis. These conditions are not hereditary, the symptoms are similar to those of muscular dystrophy and they can often be treated successfully with medication that suppresses the body's immune response. Some cases respond to plasmapheresis (plasma exchange) therapy.

Q79. **What is amyotrophic lateral sclerosis (ALS)? Does is mimic muscular dystrophy?**

A. ALS is quite different to muscular dystrophy. It is a rapidly progressive neuromuscular disorder of adults, resulting in both muscle weakness and spasms that may ultimately cause difficulty in swallowing, speaking and breathing. Many clinical variations are present, depending upon which parts of the brain stem, spinal cord, or nerves are involved. ALS is also known as Motor Neurone Disease (MND) or Lou Gehrig's disease.

Q80. **Is spinal muscular atrophy (SMA) a form of muscular dystrophy?**

A. No, these neuromuscular conditions are diseases of the spinal cord that secondarily cause muscle wasting. There are four different types of SMA with varying degrees of severity. Type 1 (also known as Werdnig-Hoffmann disease) has an onset typically in early infancy; the condition may even be apparent at birth. In contrast, type 3 (also known as Kugelberg-Welander disease) usually occurs in adolescents and young adults. Please see the MDA website for detailed SMA factsheets. [http://www.mda.org.au/Disorders/Atrophies/SMA.asp]

Q81. **What is congenital muscular dystrophy?**

A. These are types of muscular dystrophy with symptoms typically present at birth. Symptoms include generalised muscle weakness, with possible joint deformities from shortening of muscles. There are at least 30 different types - many types progress very slowly; some shorten life span.

Q82. **What is Charcot-Marie-Tooth disease? Can it be confused with muscular dystrophy?**

A. Charcot-Marie-Tooth disease (CMT) is a disease of the peripheral nervous system. It is characterised by progressive muscular wasting particularly of the legs, foot deformity, and weakness of selective muscles in the hands. Its onset is usually in adulthood, although children can be affected. It is seldom confused with muscular dystrophy. Please see the MDA website for a detailed CMT factsheet. [http://www.mda.org.au/Disorders/Peripheral/CMT.asp]

Q83. **Is benign congenital hypotonia a type of muscle disease?**
A. Yes, it is a condition, distinguished by weakness and floppiness at birth without accompanying findings of classical muscular dystrophy. This diagnosis is rarely given these days because it is now possible to diagnose children more accurately with other conditions. However, the term benign congenital hypotonia is still used occasionally to describe children with mild muscle weakness who appear to have a favourable outcome and in whom no other diagnosis can at this stage be made.

**Q84. What are the congenital myopathies? The metabolic myopathies?**

A. The congenital myopathies are a group of diseases of muscle characterised by weakness that may be symptomatic at birth, although some do not appear until later in childhood. Examples of congenital myopathies include nemaline myopathy, myotubular myopathy and centronuclear myopathy. The metabolic myopathies are caused by a specific enzyme deficiency or other disorder of muscle metabolism. In these conditions the muscles are unable to produce enough energy to work properly. Examples of these diseases are phosphorylase deficiency; acid maltase deficiency, debrancher enzyme deficiency and carnitine palmitoyltransferase deficiency. Congenital and metabolic myopathies are usually passed down through families.

**Q85. What is distal muscular dystrophy? Oculopharyngeal muscular dystrophy?**

A. Distal muscular dystrophy is a rare disorder that initially and primarily involves the small muscles of the hands and feet. Oculopharyngeal muscular dystrophy is a rare condition that usually appears in adulthood and is slowly progressive. Muscles of the eyes and pharynx are affected.

**Q86. What is myasthenia gravis?**

A. Myasthenia gravis is an autoimmune condition that affects the structure at the junction of the nerves and the muscles – the neuromuscular junction. Symptoms include muscle weakness, fatigue and ptosis (drooping of the eyelids). Drug therapy is used in treating this condition. Plasma exchange (plasmapheresis) has also been effective in the treatment of selected cases. Please see the MDA website for a detailed myasthenia gravis factsheet. [http://www.mda.org.au/Disorders/NMJ/MG.asp]

**Q87. What is Friedreich's ataxia? Is it a primary disease of muscle?**

A. Friedreich's ataxia is not a primary disease of muscle like muscular dystrophy. It is an inherited condition that causes progressive damage to the nervous system resulting in symptoms ranging from muscle weakness and speech problems to heart disease. Ataxia (lack of muscle coordination) results from the degeneration of nerve tissue in the spinal cord and of nerves that control muscle movement in the arms and legs.

**Q88. Can I/my child be included in a clinical trial?**

A. To take part in a clinical trial you generally have to live close to the clinic that is conducting the trial and comply with certain inclusion and exclusion criteria. There may be many different criteria which are very specific to each clinical trial. If you or your child would like to take part in a clinical trial at any time in the future, the best way forward is to discuss this with your consultant at the hospital. It is also very important to remember that a
drug going through a clinical trial is not a treatment and there are risks involved. More information about clinical trials is available on the MDA website. [http://www.mda.org.au/trials/index2.asp]

Q89. What does the future hold? Is there hope?

Although there is no cure for muscular dystrophy today, research is making big strides forward so that for the first time promising treatments are now being tested in clinical trial for several different types of muscular dystrophy. This brings with it hope that a treatment may be just around the corner.

Knowledge about how to manage the symptoms of the conditions is also increasing so that quality of life is much improved compared with previous generations. For conditions that limit life expectancy such as DMD, better care standards have also increased length of life by more than 10 years over the past 30 years.

Q90. How can I keep up-to-date with research and clinical trial news?

Keep an eye on the MDA website – all major research developments will be reported here. A summary of research news can also be found in our newsletter – Research Gateway - which is published 3 to 4 times per year. You can get regular updates by becoming a friend of the MDA Facebook page [www.facebook.com/MuscularDystrophyAustraliaPage].

Other good sources of research news are the other major muscular dystrophy charities around the world such as the Muscular Dystrophy Campaign in the UK [http://www.muscular-dystrophy.org/research/news] and the Muscular Dystrophy Association in the USA. [http://mda.org/research/news]

Q91. I’ve read on the internet that a cure for muscular dystrophy has been found, how can I find out if it’s true?

Contact the MDA – our Scientific Communications Officer will be able to help (details at the end of this page). Unfortunately all too often false hope is raised by over enthusiastic journalists who don’t have a real understanding of the subject matter. Although there are many promising advances happening in research labs all around the world, it could still be some time yet before it is translated into a successful treatment for patients. Also see Q90 above for reputable sources of research news that will help put advances in perspective and keep expectations realistic.

Q92. I am a full time carer; how can I get support/respite?

The MDA holds four week-long camps per year as well as short weekend breaks throughout the year. These programs give children and adults with muscular dystrophy a chance to get away and mix with their peers in a fun and relaxed environment while giving their carers a much needed break. More information about respite can be found on the MDA website. [http://www.mda.org.au/Respite/Respite.asp]. There are also Carer Support Groups, contact the MDA for more information (details at the end of this page). It may also be useful to discuss respite options with your clinician.
Q93. Is camp just for kids?

NO!!! Camp is for everyone. In the past campers up to 76 years of age have enjoyed CampMDA.

Q94. I’ve seen a clinic advertising stem cell therapy for muscular dystrophy – should I give it a try?

It is completely understandable that people with serious, debilitating and often life threatening neuromuscular conditions are willing to try anything that might offer them a treatment for their condition. However, the clinics offering so called “stem cell therapies” are unregulated and ask for large sums of money without any evidence of their safety or effectiveness. As such, they may be at best ineffective, or at worst dangerous and potentially fatal. There are numerous reports of people suffering serious complications including several deaths after treatment at unregulated stem cell clinics. Although there have been some promising results in animals using stem cells for neuromuscular conditions, there have not yet been any clinical trials in humans. It will probably be some time yet until the work on stem cells currently ongoing in the lab can be translated into treatments in the clinic. For more information about stem cells, download the Australian Stem Cell Handbook.

Q95. What can I do to minimise discomfort at night?

There are a many different options such as special mattresses which should be discussed with your neurologist and/or occupational therapist.

Q96. Where do I get an electric wheelchair from, how long is the waiting list for one and is there any funding I can access to pay towards this.

The Victorian State-Wide Equipment Program (SWEP) provides disabled people with subsidised aids and equipment. The waiting time depends on what equipment is required and the urgency of the request. [http://swep.bhs.org.au/]

Q97. Is there funding for home modifications?

SWEP also provides funding for home modifications up to a maximum of $4000 (per person per life time) and requires an assessment by an occupational therapist.


Q98. Where can I get a Wheelchair Accessible Van or get my van modified.

There are many modified vehicle providers; it is a good idea to shop around for a solution that suits you. The RACV [https://www.racv.com.au/wps/wcm/connect/racv/Internet/Primary/road+safety/advice+_+information/drivers+with+a+disability/links] has a list of links that is a good place to start. Funding for the vehicle or modifications is available through the Vehicle Modification Subsidy Scheme (VMSS). [http://swep.bhs.org.au/vmss]
Q99. What is available after my son leaves school and does not want to attend further study but wants to do something away from home?

The Department of Human Services [http://www.dhs.vic.gov.au/for-individuals/disability/community-life-and-jobs] funds a range of services and supports that can assist people with a disability to become more independent and participate in their local community. Day services are one option - activities organised or provided directly by community service organisations to provide the opportunity to learn new skills and participate in the community. [http://www.dhs.vic.gov.au/for-individuals/disability/community-life-and-jobs/day-supports]

Q100. I’m planning a visit to Melbourne and I’m worried about accessibility for my wheelchair, where can I find information and maps?

Getting about large cities can be daunting, even more so if you are affected by a neuromuscular condition and rely on physical assistance. The MDA website has links to easy-to-read mobility maps which are feature rich with important information. [http://www.mda.org.au/Information/Access/Maps.asp]

Many local council websites also have “Mobility Maps” available.

Q101. What can I do to help bring treatments closer to reality?

Over recent times, significant headway has been made in the understanding and development of potential treatments for muscular dystrophy by research scientists. MDA supports the National Muscular Dystrophy Research Centre which is currently developing therapeutic approaches which show great promise. But all this takes money - and plenty of it!

Please consider the difference your financial contribution will make to the cutting edge research at the National Muscular Dystrophy Research Centre. We do rely on private and corporate donations to underwrite the research program.

To contribute toward the fight against muscular dystrophy you can make a donation on the MDA’s Secure Server. [http://www.mda.org.au/SupportMD/Donation/Donation.asp]

Alternatively you could do your bit by taking part in one of the many MDA fundraising events. [http://www.mda.org.au/SupportMD/HelpMD.asp]

Further information

Please contact Muscular Dystrophy Australia

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