FACT SHEET

Understanding Muscular Dystrophy

THIS FACT SHEET explains what muscular dystrophy is, the main types, who it affects and the cause. It covers symptoms and diagnosis, effects of the condition, treatment and offers care-giving tips.

What is Muscular Dystrophy?

Muscular dystrophy (MD) is a degenerative disorder that causes a person to become weaker and less mobile as groups of muscles in their body gradually waste away. There are more than 60 types of MD. The age at which muscle wasting begins and the muscle groups affected depends on the type of MD.

Who Develops MD?

About one person in 1000 develops MD and estimates suggest more than 25,000 people in Australia have it. MD affects people of all ages and can start at any age. It occurs in males and females in equal proportions. The exceptions are Duchenne MD and Becker MD, which occur in males only. Myotonic dystrophy is the most common type experienced by adults.

What Causes MD?

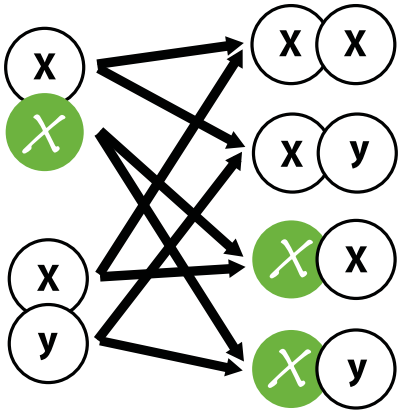
MD is a genetic disorder caused by a faulty gene that is generally inherited from one or both parents. Two thirds of all cases fall into this category. One third of cases, however, are caused by a faulty gene that is not inherited. This faulty gene mutates into an MD-causing agent of its own accord. Researchers do not yet know what causes the gene to mutate.

Genes exist in pairs in the cells of our body. They determine how our cells form, grow and behave. Each parent contributes one gene for each pair. There are three primary ways to inherit a faulty gene from parents that may – or may not – develop into MD:

- X-linked recessive inheritance
- Autosomal recessive inheritance
- Autosomal dominant inheritance

X-linked recessive: Each parent provides one sex chromosome to create the cell that becomes an embryo – an X chromosome or Y chromosome. Females have two X chromosomes, males have one Y and one X chromosome - the union creates either XX (a female child) or XY (a male child).

X-linked recessive inheritance is when the mother passes on one faulty X chromosome (See figure below). The resulting child, male or female, has a 50% chance of inheriting the faulty gene but only the male can develop MD from it. Because the female child has two X chromosomes, one healthy X can override one inherited faulty X. In this instance however, the female child is still a carrier



of the inherited faulty X and may pass it on to her children. The female child should have genetic testing to determine her status before having her own children.

Autosomal recessive: This type of inheritance is when both parents carry - and pass on - a recessive faulty gene. A recessive faulty gene is one that does not display symptoms, so the parents may not know

they carry it. Each of their offspring, regardless of gender, has a 25% chance of developing MD.

Autosomal dominant: In this case of inheritance, only one parent needs to pass on a faulty gene to affect their child (called a dominant gene). This faulty gene can come from either parent and can affect male or female children. The child has a 50% chance of developing MD.

Types of MD & Symptoms

The following are the most common types of MD that affect older adults. Symptoms vary according to the type.

Limb-girdle MD: This type can appear up to the age of about 30. First it affects the muscles of the hips and shoulders. It can progress rapidly or sometimes very slowly, allowing the person a normal life span.

Limb-girdle MD is caused by a recessive gene – both parents must pass it on. Each of their children has a 25% chance of developing the disease, a 50% chance of carrying the gene without developing the disease and a 25% chance of being completely free of the gene.

Fascioscapulohumeral MD: This usually appears from adolescence up to the mid-20s and the effects vary considerably. To begin, the person experiences problems with the muscles of the face and shoulder girdle. It may be difficult to raise their arms high and their shoulders might start to slope forward. This MD progresses very slowly. Most people have a normal life span but some eventually become quite disabled.

It is caused by a dominant gene - only one parent needs to pass it on for MD to occur.

The child of a parent with this condition has a 50% chance of inheriting the gene.

Myotonic MD: Myotonic MD most often occurs between 20 and 35 years of age but about half those who develop the condition have symptoms before the age of 20. A significant number have no clear-cut symptoms until they are over 50.

Myotonia causes a delayed relaxation of muscles after contraction – trouble relaxing a hand grip, for example. Facial muscles are the first affected but other early signs include weakness in the hands, feet and neck muscles. Myotonic MD progresses slowly, having significant effects after about 15 or 20 years. It can involve other parts of the body, such as the central nervous system (brain, optic nerves, spinal cord), the glands that secrete hormones and the eye muscles.

It is caused by a dominant gene. Each child of a parent with the condition has a 50% chance of inheriting the gene.

Diagnosis

A neurologist (a specialist in conditions that affect the brain and nervous system) will take a detailed, personal history and family history to determine the pattern of the disease, such as the age at which symptoms first appeared in you or your family. A muscle biopsy is also necessary, which involves removing a small piece of muscle under general or local anaesthetic that is sent to a laboratory for examination. Gene tests and a "CK test" that measures blood enzyme levels are common procedures.

Genetic testing: It is generally possible to do tests at any age (even before birth) to determine if you are a carrier or likely to develop MD. A doctor would first conduct an examination for early signs of MD.

Testing to determine if a person is a carrier usually occurs near adulthood, so that he or she can make informed decisions about whether or not to have children.

Counselling: Because the results of genetic testing can be upsetting and create difficult decisions, anyone who chooses to have it must first see a genetic counsellor. Counsellors will map out your family tree to identify the pattern of MD inheritance, determine your risk level and discuss thoroughly with you the possible outcomes of testing.

To enquire about testing, contact the Victorian Clinical Genetics Service located at the Royal Children's Hospital: telephone (03) 9345 5157

Treatment

There is no known cure for MD and no treatment that can stop its progression.

However, that a great deal can be done to ease discomfort and help a person maintain their quality of life, mobility and independence.

What is the Outlook?

MD is a degenerative condition – the muscles become gradually weaker – but the rate at which this happens varies considerably between the many types. Forms of MD that occur in adults often move slowly and the person may have years of quality life ahead or an average life expectancy.

As a general rule, the younger the person is when the disease first appears, the more rapidly it progresses and the more widespread and disabling its effects. In the most aggressive forms, a person becomes unable to walk and confined to a wheelchair. Adult forms of MD tend to result in slower degeneration. It is generally accepted that the older the person is at diagnosis the slower the degeneration process. Gradually the body also loses the ability to fight infections and death can result from a simple chest infection.

Caring for Someone with MD

Assembling experts: A GP can monitor the person's general fitness, mobility and breathing but some types of MD involve other medical conditions, such as heart problems, cataracts or diabetes, that require specialist care. Assembling a good team of experts is important. Your diagnosing doctor and the MD Association can help.

Rehabilitation team: An alert rehabilitation team plays a vital role by stepping in to prevent problems before they occur. The team will support carers in looking after someone at home for as long as possible.

Members can suggest ways to avoid complications and advise on aids, equipment and services that make life easier to manage. (See Fact Sheet 13: Adapting Your Home; Fact Sheet 14: Lifting and Moving).

Physiotherapists, for example, can advise on the use of orthopaedic devices, such as splints that prevent or delay curvature of the spine and that minimise painful muscle contractures.

Team members can also provide counselling for the person or family and can help to organise respite care when needed.

Fatigue: People with MD tire easily so it's important to plan activities in advance and allow for regular rest periods (See Fact

Sheet 11: Managing Fatigue). A person may need help from an occupational therapist to learn how to conserve energy, simplify work or establish a regular sleeping pattern.

Speech and swallowing difficulties: If the muscles of speech are affected, the speech pathologist on the team can provide strategies and advise on electronic aids to help the person communicate as well as possible. Speech therapists also help to manage difficulties in eating and swallowing. (See Fact Sheet 8: Eating and Swallowing Problems.)

Body comfort and mobility: Treatments such as massage and hydrotherapy have helped many people remain more comfortable and mobile. Discuss these options with your physiotherapist or occupational therapist. (See Fact Sheet 5: Assessing Alternative Treatments).

Contact

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