Muscular dystrophy

Summary

- Muscular dystrophies are inherited muscle conditions.
- These conditions cause weakness and wasting of the muscles.
- There is currently no cure for any of the 30 types of muscular dystrophy.
- The symptoms of different muscular dystrophies may vary.

Muscular dystrophy is the name given to a group of inherited neuromuscular disorders. These disorders cause weakness and wasting of the muscles. This muscle wastage gets worse over time, and is not reversible.

There are more than 30 different types of muscular dystrophy. Most are caused by changes to genes involved in providing strength to the muscle structure.

There are around 30 other neuromuscular conditions that can be confused with muscular dystrophy due to their having similar symptoms. However, these conditions are caused by a different mechanism.

What causes muscular dystrophy?

Many genes help to make the proteins that protect muscle fibres from damage. Muscular dystrophy occurs if one of these genes does not work properly.

Each type of muscular dystrophy is caused by a different change in a gene. Some of these gene changes are inherited from a parent. Some of them are new changes that occur early in development. These are known as spontaneous or ‘de novo’ gene changes.

Spontaneous gene changes are not inherited, but they can be passed on to the next generation.

Types of muscular dystrophy

There are more than 30 different types of muscular dystrophy. The main types are:

- Duchenne muscular dystrophy
- Becker muscular dystrophy
- congenital muscular dystrophy
- limb-girdle muscular dystrophy
- facioscapulohumeral muscular dystrophy
- myotonic dystrophy
- oculopharyngeal muscular dystrophy
- Emery-Dreifuss muscular dystrophy.

Some of these types of muscular dystrophy are further divided into sub-types. For example, there are more than 20 types of limb-girdle muscular dystrophy.

From one type of muscular dystrophy to another, there is variation in:

- the severity of the muscle degeneration
- which muscles degenerate.

Duchenne and Becker muscular dystrophy
Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD) are often discussed together because they cause similar patterns of weakness and are inherited in the same way. BMD is less severe than DMD.

These conditions are caused by an alteration in the *DMD* gene. This gene is responsible for the production of a large protein (dystrophin) which provides a scaffold structure to muscle fibres. This protects them from damage during muscle contraction. Without this protein the muscles gradually break down. DMD mostly affects boys, but girls can be affected too.

**Symptoms of Duchenne muscular dystrophy**

Duchenne muscular dystrophy is not usually noticeable before the age of two or three. Symptoms and signs include:

- delayed walking age
- frequent falls, difficulty rising up from the ground or going up hills or stairs
- difficulty running and jumping
- well-developed or excessively large calf muscles. Other muscles are poorly developed
- a waddling walk
- a sway-back (‘lordosis’)
- a tendency to stand and walk on the forefoot, with the heel off the ground. This is often called ‘toe-walking’.

**Effects of Duchenne muscular dystrophy**

Duchenne is one of the most severe forms of muscular dystrophy. It can lead to:

- wheelchair use – generally begins from around nine years of age. Total dependence on a wheelchair occurs around the early teens
- restriction of joint motion – caused by contractures, which are shortenings of the muscles and tendons. The ankles are usually affected first, and the hips and knees last
- scoliosis – a curvature of the spine. Corrective surgery is usually required
- difficulty breathing – caused by weakness of the muscles associated with breathing. In some cases, mechanically assisted breathing helps in the latter stages
- heart problems – in older boys, the dystrophic process can start to affect the heart muscle
- intellectual difficulties – a minority of boys with Duchenne muscular dystrophy have a learning disability that can affect their school work
- early death – most affected people only survive into their 20s. Small numbers survive only into their teens or reach more than 30 years.

**Symptoms of Becker muscular dystrophy**

The rate of muscle degeneration in people with Becker muscular dystrophy (BMD) varies greatly between individuals. Signs and symptoms of the condition are similar to those of DMD, but are usually milder and more varied.

**Effects of Becker muscular dystrophy**

Becker muscular dystrophy is less severe than Duchenne muscular dystrophy. Its features include:

- People with BMD can still walk at 16 years. Some can continue to walk until early adulthood or into advanced age.
- Many affected people may survive up to middle age. Some people with BMD have lived beyond 80 years.
- Scoliosis seldom occurs.
- The effect on lung function is less severe than for DMD.
- Heart trouble is less frequent, although it is occasionally serious.

**Facioscapulohumeral dystrophy (FSH)**
Facioscapulohumeral dystrophy (FSH) is a form of muscular dystrophy that affects the face and shoulders. It is generally considered less serious than other forms of muscular dystrophy.

This condition gets its name from the areas of the body that are affected most often – the muscles:
- in the face (facio-)
- around the shoulder blades (scapulo-)
- in the upper arms (humeral).

Some muscle groups on one side of the body are stronger than those on the other side.

Onset of FSH can occur in infants, but symptoms may appear at any time from childhood until someone is in their 50s.

Typical facial features of FSH include:
- eyes appear to be slightly open when the person is sleeping due to weakness of eye closure muscles
- fewer than usual facial lines due to age. This characteristic facial appearance is more noticeable when the muscles are in use, for example, during speech.

Muscle weakness in the shoulders and arms may lead to:
- ‘winging’ of the shoulder blades – the shoulder blades stick out backwards, especially when the arms are held forward
- reduced muscle bulk between the shoulder blades
- difficulty with, or an inability to raise the arms. Some people first notice a problem in sport, for example, serving at tennis
- weakened ability to bend and straighten the elbow, which is due to weakness of the upper arm muscles.

The selective pattern of muscle weakness in the lower limbs and back may include:
- foot drop due to weakness of muscles in the front of the leg
- weakened ability to straighten the hip joints – the knees may also be affected
- lordosis (sway-back) – in severe cases, the abdomen may stick out and the shoulders may be excessively held back.

**FSH muscular dystrophy difficulties**

In severe early-onset FSH, deafness is a common symptom. Changes also occur in the eyes, although this seldom affects vision. However, people with FSH should have their eyes checked regularly.

Other issues faced by people with FSH include:
- trouble combing hair, hanging out washing and reaching high shelves, due to an inability to raise the arms
- a tendency to trip due to foot drop
- a tendency for one or both knees to give out
- difficulty with stairs and steps.

Due to the understated and variable nature of FSH symptoms, health professionals are sometimes challenged to identify the condition. This is where genetic testing can quickly assist with a diagnosis.

**Progression of FSH**

On average, FSH muscular dystrophy progresses slowly and the level of severity eventually seems to plateau (level off). In very mild cases, it may not be possible to detect that the disease is progressing. People affected by FSH of ‘average severity’ usually retain the ability to walk and have a normal life span.

**Myotonic dystrophy**

Myotonic dystrophy is the most common adult form of muscular dystrophy. (It is also known as Steinert's disease and dystrophia myotonica.) Unlike the other muscular dystrophies, the muscle weakness is accompanied by

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myotonia (delayed relaxation of muscles after contraction) and by various other non-muscular symptoms.

The first muscles to be affected by weakness are those of the face, neck, hands, forearms, and feet. Myotonic dystrophy can affect the tissues and organs of many body systems.

The effects can include:
- cardiac disease
- cataracts
- testicular atrophy – the testes become smaller and may stop functioning
- difficulty breathing and adverse reactions to anaesthesia
- difficulty in swallowing (dysphagia)
- digestive problems
- excessive daytime sleepiness
- learning difficulties
- diabetes
- thyroid dysfunction.

Onset of myotonic dystrophy
Fifty per cent of people with myotonic dystrophy show visible signs by about twenty years of age. But a significant number do not develop clear-cut symptoms until after age fifty.

When myotonic dystrophy is suspected (because it is present in other members of the family), careful examination may reveal typical features before obvious symptoms appear.

Myotonic dystrophy is inherited in an autosomal dominant manner. This means that the condition can be caused by inheriting just one copy of the altered gene.

We inherit one copy of each gene from our mother and father. Therefore, when a person with myotonic dystrophy goes on to have children, there is a 50 per cent chance that each child will have myotonic dystrophy.

This condition exhibits an unusual pattern called ‘anticipation’ when passed down between generations. ‘Anticipation’ is a term used by health professionals to describe when symptoms of a condition appear at an earlier age and often at an increased severity.

Effects of myotonic dystrophy
The course of myotonic dystrophy varies widely, even in the same family. There are people with the disorder whose symptoms are so mild they hardly know the condition is present. Whatever muscle weakness they experience is something they assume to be normal and adapt to.

In some cases, the only symptom may be a cataract. Nevertheless, these people do have myotonic dystrophy and can transmit a more severe version of the condition to their children.

In most cases, weakness and muscle wasting starts with certain muscles and slowly progresses to the point of some physical inability. As it progresses it moves beyond the muscles originally involved to those of the shoulders, hips, and thighs. This muscle weakness rarely becomes severe until fifteen to twenty years after the onset of symptoms.
The older a person is when muscle weakness is first noticed, the slower it progresses and the less serious the condition and effects.

Congenital muscular dystrophy
The congenital muscular dystrophies (CMDs) are a group of conditions that vary in severity and rates of progression. Congenital means ‘from birth’. In most cases of congenital muscular dystrophy, the initial symptoms are present at birth or in the first few months.
Babies with CMD often have low muscle tone or floppiness and may have reduced movements. Other common signs are contractures (tightness) in the ankles, hips, knees and elbows. Some babies may also have trouble breathing and feeding. Some improvement often occurs in childhood and the disease shows little or no progression.

There are at least five different types of CMD, which are caused by variations in different genes. Both parents usually carry the altered gene but are unaffected by the condition. The affected child inherits two copies of the altered gene – one from each parent.

Late onset muscular dystrophies

Many people think that muscular dystrophy is exclusively a childhood condition. However, it can occur at any point in life.

As well as myotonic dystrophy and FSH, there are three other types of muscular dystrophy that can occur later in life:

- limb-girdle muscular dystrophy – which involves slow to fairly rapid progressive muscle deterioration of the proximal (back of the body) muscles of the pelvis and shoulders. People with limb-girdle muscular dystrophy have generally inherited the altered gene from both parents. This type usually occurs in the first to third decades of life and can involve a normal life span if the muscle deterioration progresses slowly
- ophthalmoplegic muscular dystrophy – which is fairly rare and affects the extraocular (eye) muscles, leading to drooping eyelids. Eventually, the muscles associated with swallowing may be affected. It usually occurs in adulthood
- distal muscular dystrophy – which is the rarest of the muscular dystrophies, although it is comparatively more common in Sweden. It affects the small muscles of the arms and legs.

Diagnosis of muscular dystrophy

Diagnosis before the age of two or three is possible through:

- a blood test – to check the levels of a protein called creatine phosphokinase (CPK). CPK is produced by damaged muscle, so levels are very high in people with muscular dystrophy
- genetic testing – if it is known that a condition runs in a family, a test to detect the genetic change can be performed on the DNA in the blood
- a muscle biopsy – removal of a small piece of muscle tissue for examination under a microscope
- electromyography (EMG) – checks the health of the muscles and the nerves that control the muscles. It involves inserting a very thin needle into the muscle.

Early diagnosis of muscular dystrophy will enable the most appropriate management of the condition from a young age.

Treatment for muscular dystrophy

There is no cure for muscular dystrophy.

To help ease discomfort, reduce joint contractures, and prevent or delay scoliosis, physiotherapists offer advice on stretches and exercises, and the prescription of orthoses and other orthopaedic devices. Occupational therapists also provide advice on sitting positions and activities. Such treatment can keep affected people walking for longer and maximise independence in daily living.

For some types of muscular dystrophy, medication can help manage the symptoms of the condition. For example, boys with Duchenne muscular dystrophy are usually prescribed corticosteroids, which can delay the need for a wheelchair by several years on average. However, the risk of side effects needs to be considered.

Genetic counselling for muscular dystrophy

If your child or another family member has been diagnosed with muscular dystrophy, or if it runs in your family, it may be helpful to speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing
emotional support, they can help you to understand muscular dystrophy and what causes it, how it is inherited, and what a diagnosis means for your child's health and development, and for your family.

Genetic counsellors are trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.

**Genetic testing for muscular dystrophy**

If muscular dystrophy runs in your family, a genetic counsellor can explain what genetic testing options are available to you and other family members. You may choose to visit a genetic counsellor if you are planning a family – to find out your risk of passing the condition on to your child, or to arrange for prenatal tests. Read more about genetic testing for muscular dystrophy.

**Victorian Clinical Genetics Services (VCGS)** provides genetic consultation, counselling, testing and diagnostic services for children, adults, families, and prospective parents.

The **Genetic Support Network of Victoria (GSNV)** is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by muscular dystrophy.

**Where to get help**

- Your **GP (doctor)**
- A **genetic counsellor**
- **Muscular Dystrophy Australia** Tel. (03) 9320 9555
- **Muscular Dystrophy Foundation Australia - support near you**
- **Victorian Clinical Genetics Services (VCGS)** Tel. 1300 118 247
- **Genetic Support Network of Victoria (GSNV)** Tel. (03) 8341 6315

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