Muscular dystrophy

Summary

- Muscular dystrophies are inherited muscle diseases that lead to progressive weakness and irreversible wasting of muscle tissue.
- There is no cure for any of the 60 neuromuscular disorders.
- The symptoms of different muscular dystrophies may vary.

Muscular dystrophy is the name given to a group of neuromuscular disorders that cause progressive and irreversible weakness and wasting of the muscles.

There are more than 30 different types of muscular dystrophy. They are genetic conditions, which means that they are caused by an alteration within the genetic makeup. The genes causing the majority of cases of muscular dystrophy have been identified and the function of these genes have been studied. Many of the genes are involved in providing strength to the muscle structure.

There are approximately 30 other neuromuscular disorders that are often confused with muscular dystrophy, because they also cause muscle weakness. However, in these conditions, the muscle weakness is usually not caused by a problem within the muscles themselves, but by the nerves or motor neurons controlling them, or the supply of energy to the muscles. These include spinal muscular atrophy, neuropathies such as Charcot-Marie-Tooth disease, myasthenia gravis, and the myopathies (including metabolic, mitochondrial and inflammatory myopathies).

Causes and cure research into muscular dystrophy

Each of the approximately 30 muscular dystrophy diseases has a separate cause, and there is no cure. Medical research has led to a greater understanding of muscular dystrophy and potential treatments are beginning to emerge. Several of these are now in clinical trial. However, the discovery of a cure for one type of muscular dystrophy may not necessarily help in curing another type.

Diagnosis of muscular dystrophy

Diagnosis before the age of two or three (before symptoms are very obvious) is possible through:

- a blood test – this will show very high levels of a protein called creatine phosphokinase (CPK)
- 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.

Genetic counselling for muscular dystrophy

Genetic counselling is available to help people understand the hereditary nature of the disorder and the probable risk of them having a child with muscular dystrophy. Counsellors can help couples make an informed decision about having children and discuss options such as prenatal and preimplantation genetic diagnosis.

Symptoms and support needs vary

betterhealth.vic.gov.au
People affected by muscular dystrophy have different degrees of independence, mobility and carer needs. These needs will vary within each type of muscular dystrophy and between types. The most severe conditions cause major disability and shorten life expectancy, while the milder conditions do not present any symptoms until later in life and progress more slowly.

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.

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
- oculopharangeal 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.

There is considerable variation in the severity and distribution of muscle degeneration among the various types of muscular dystrophy.

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. Becker muscular dystrophy is less severe than Duchenne muscular dystrophy.

These conditions are caused by an alteration in the dystrophin gene, which usually only affects boys. This gene is responsible for the production of a large protein which provides a scaffold structure to muscle fibres, protecting them from damage during muscle contraction. Without this protein the muscles gradually break down.

Symptoms of Duchenne muscular dystrophy

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

- frequent falls, difficulty rising up from the ground or going up hills or stairs
- 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.

**Effects of Becker muscular dystrophy**

Becker muscular dystrophy is less severe than Duchenne muscular dystrophy. Its features include:
- People with Becker muscular dystrophy 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 affected people have lived beyond 80.
- Scoliosis seldom occurs.
- The effect on lung function is less severe.
- 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 more benign (less serious) than other forms of muscular dystrophy.

This condition gets its name from the areas of the body that are affected most often, being the muscles in the face (facio-), around the shoulder blades (scapulo-), and in the upper arms (humeral). Some muscle groups on one side of the body are stronger than they are on the other side of the body.

Typical features of FSH include:
- The eyes appear to be slightly open when the affected person is sleeping. This is due to weakness of eye closure muscles.
- There are 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, 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 frequent problem. Changes also occur in the eyes, although this seldom affects vision. However, people with FSH should have their eyes checked regularly.

Other difficulties 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.

Those most severely affected by FSH first feel its effects in infancy or early childhood, and are unable to walk by adolescence or early adulthood. At the other end of the spectrum, even an experienced doctor would find it difficult to tell that a person had the condition.

**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. Unlike the other muscular dystrophies, the muscle weakness is accompanied by myotonia (delayed relaxation of muscles after contraction) and by a variety of abnormalities in addition to those of muscle. The disorder is also known as Steinert’s disease and dystrophia myotonica.

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. Consequently, myotonic dystrophy may present itself in what one expert has called a “bewildering variety of ways”.

The effects can include cardiac disease, cataracts, testicular atrophy, respiratory impairment and adverse reactions to anaesthesia, difficulty in swallowing (dysphagia) and other gastrointestinal tract involvement, mental disorders (including mental retardation), excessive output of insulin and abnormal carbohydrate metabolism, and excessive sleeping.

**Onset of myotonic dystrophy**

Fifty percent of those with the disorder show visible signs by about twenty years of age, but a significant number do not develop clear-cut symptoms until after age fifty. However, when myotonic dystrophy is suspected (because it is present in other members of the family), careful examination may reveal typical abnormalities before obvious symptoms appear.
Myotonic dystrophy is transmitted from generation to generation by men or women who themselves have inherited the defective gene and have the disease. Because the defective gene is dominant, only one myotonic dystrophy gene derived from either the father or mother, is required to produce the disease in an offspring. There is a 50-50 chance that, if one parent has the disease, such transmission will occur. The technical term for this mode of inheritance is 'autosomal dominant'.

The severity of the condition tends to get worse with successive generations in a family. Geneticists call this phenomenon ‘anticipation’. This can lead to a very severe form of myotonic dystrophy with onset in infancy.

**Effects of myotonic dystrophy**

The course of myotonic dystrophy varies widely, even in the same family. On the one hand, there are people with the disorder whose symptoms are so mild they hardly know anything is wrong. Whatever muscle weakness they experience is something they take for granted and adapt to.

In some cases, the only symptom may be a cataract. Nevertheless, these people do have myotonic dystrophy and can transmit a serious case of the disease to their children.

For the most part, weakness and wasting slowly progress to the point of some disability, moving beyond the muscles originally involved to those of the shoulders, hips, and thighs. As a rule, disability 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 is the progression and the less serious the consequences.

**Congenital muscular dystrophy**

The congenital muscular dystrophies (CMDs) are a very mixed group of conditions with varying degrees of severity and rates of progression. Congenital means 'from birth' and in most cases of congenital muscular dystrophy, the initial symptoms are present at birth or in the first few months.

Babies with congenital muscular dystrophy 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 alterations 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 disorder. However, it can occur at any point in your life.

As well as myotonic dystrophy, FSH and Becker MD, three other types that can occur later in life include:

- limb-girdle muscular dystrophy
- ophalmoplegic muscular dystrophy
- distal muscular dystrophy.

**Limb-girdle muscular dystrophy**

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 involves:
• the proximal (back of the body) muscles of the pelvis and shoulders
• slow to fairly rapid progressive muscle deterioration
• a possible normal life span, if the muscle deterioration progresses slowly.

**Ophthalmoplegic muscular dystrophy**

This form of muscular dystrophy 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**

This is the rarest of the muscular dystrophies, although it is comparatively more common in Sweden. It affects the small muscles of the extremities (arms and legs).

**Where to get help**

• Your doctor
• Muscular Dystrophy Association Tel. (03) 9320 9555

**Things to remember**

• Muscular dystrophies are inherited muscle diseases that lead to progressive weakness and irreversible wasting of muscle tissue.
• There is no cure for any of the 60 neuromuscular disorders.
• The symptoms of different muscular dystrophies may vary.

---

Content on this website is provided for information purposes only. Information about a therapy, service, product or treatment does not in any way endorse or support such therapy, service, product or treatment and is not intended to replace advice from your doctor or other registered health professional. The information and materials contained on this website are not intended to constitute a comprehensive guide concerning all aspects of the therapy, product or treatment described on the website. All users are urged to always seek advice from a registered health care professional for diagnosis and answers to their medical questions and to ascertain whether the particular therapy, service, product or treatment described on the website is suitable in their circumstances. The State of Victoria and the Department of Health & Human Services shall not bear any liability for reliance by any user on the materials contained on this website.

For the latest updates and more information, visit www.betterhealth.vic.gov.au

**Copyright © 1999/2019** State of Victoria. Reproduced from the Better Health Channel (www.betterhealth.vic.gov.au) at no cost with permission of the Victorian Minister for Health. Unauthorised reproduction and other uses comprised in the copyright are prohibited without permission.

---

Muscular dystrophy