

# Medicine for Managers

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## Muscular Dystrophy

**Muscular dystrophy is a name that everyone has heard and all know that it results in muscle weakness. For the most part knowledge ends there. But the disorder, or rather the 60 or so disorders that are collectively known by the name, is very important. It affects just over one person in 1,000 and there are an estimated 70,000 sufferers in the UK.**

The various types vary in severity. They are caused by a defective gene within the DNA sequence. The significance of the abnormal gene is that an aspect of the creation of muscle cells does not occur correctly so that the muscles fail to work as they should.

Each type of muscular dystrophy is associated with a defect in a different gene. Despite being a genetic disorder only a minority of cases are inherited. More commonly the genetic error appears to occur as a result of a spontaneous mutation in a single family member.

The cardinal feature of muscular dystrophy is the muscle weakness which may first appear at any time from shortly after birth to middle age. It may appear as a floppy baby who does not meet the developmental milestones, an adolescent who becomes tired and increasingly weak or an older person who feels that strength is being lost.

The degree of muscle weakness and the speed with which it develops also depends on the type

of dystrophy. The muscle dysfunction may be associated with wasting and the muscles themselves may produce aches and pains. Weakness and disuse may be accompanied by the development of scarring within the muscles so that fixed deformities called contractures develop.

The most common form of muscular dystrophy, and perhaps the only type where the name is known, is **Duchenne Muscular Dystrophy** affecting one male in 3,500.

It causes weakness in the proximal muscles; that is those muscles in the upper arms and shoulders, the thighs and the buttocks.

It normally affects boys because it is an X-linked recessive characteristic (i.e. affects the male sex chromosome) ; girls may carry the gene but normally do not exhibit the condition.

It becomes apparent early in life as early as two to three months after birth. Muscle milestones are missed, the child cannot hop or run and

mobility declines to the extent that the young person is usually wheelchair-bound by the age of twelve.

Sufferers may be affected by a host of muscle-related symptoms as their ability to undertake even the simplest tasks is reduced. Men with the dystrophy do not usually survive beyond their thirties.

Making the diagnosis may have profound implications for the mother who may be carrying the gene should she have a further pregnancy. With modern genetic sampling it is possible to identify the defective gene in a proportion of pregnancies.

**Becker's Muscular Dystrophy** has the same characteristics as the Duchenne variety except that it is generally less severe and becomes apparent later. Symptoms often do not appear until the early twenties resulting in difficulty walking by the forties or fifties. Like Duchenne dystrophy it usually only occurs in boys although, again, the gene may be carried by the female.

**Emery-Dreifuss** dystrophy usually starts in childhood and affects the shoulders and arms. It may also affect the muscles in the lower leg causing falls. It progresses more slowly than other forms and a wheelchair may be needed only late in life. The condition may affect the electrical system of the heart necessitating a pacemaker.

**Limb Girdle Type** dystrophy affects the top of the arms and legs and there are many different types which display marked variations in

severity and muscles involved. The condition may affect males and females. Sometimes it is associated with heart muscle and respiratory muscle complications.

**Facio-scapulo-humeral** dystrophy usually affects the muscles of the face, shoulder and upper arm, hence the name. The degree of severity varies considerably with some patients experiencing virtually no symptoms whilst 10% end up in a wheelchair. It affects men and women. When noticeable it normally appears as facial weakness. It is unusual for this form of the disorder to reduce lifespan.

**Oculo-pharyngeal** dystrophy usually starts late in life and, as the name suggests, causes weakness in the eye (resulting in droopy eyelids) and difficulty in swallowing. After many years, mild arm and leg weakness may develop.

**Congenital Muscular Dystrophy** starts early in life and is recognised within a few weeks. However it has a very variable prognosis with some patients not progressing and the muscles gradually improving their strength whilst others suffer more severe features including fits and breathing difficulties resulting in a poor outlook. The condition is rare affecting about one in 50,000 people.

**The Diagnosis** of muscular dystrophy is often made clinically in the more severe forms with early onset but it may easily be missed in those which occur insidiously in later life. A blood test called **creatinine kinase** measures muscle breakdown and is commonly very high in muscular dystrophy. Otherwise a sample of the muscle is biopsied and reviewed under the

microscope or tested biochemically. Because of the inherited nature of the disorder, DNA testing has become available in recent years to confirm the diagnosis.

**Care and Management** of patients with muscular dystrophy depends on the nature and presentation of the condition. Weakness may be very variable but, obviously, it is the reduction in mobility with progressive weakness which needs to be managed and the patient provided with support wherever possible. Physiotherapy and exercise such as swimming may help to delay muscle deterioration and keeps the joints mobile.

Other techniques may involve splints for affected limbs, other walking aids and wheelchair provision. Some types of muscular dystrophy are sometimes treated with steroids and may temporarily slow the progress of the disease but the results may be disappointing.

Over time treatment is directed to relieving specific complications such as heart disorders, swallowing problems, maintaining language skills and also the use of surgery to correct symptoms such as tendon contractures, which prevent straightening of limbs, droopy eyelids and other consequences of muscle weakness.

Muscular dystrophy is a disorder which results from spontaneous changes in one or more genes resulting in variant forms or **mutations**. They result from the alteration in order of the units which create DNA or the deletion or insertion of sections of genes.

They are passed on from parent to child, one copy of any one gene being inherited from each parent. The condition can be inherited as:

- (a) **A recessive disorder** – in such circumstances, to **have the disorder**, defective genes must be inherited from both parents. If the gene is passed from only one parent, the child is a **carrier** and does not have the disorder. If the gene is not inherited from either parent, the child will be **healthy**.
- (b) **A dominant disorder** – in such circumstances it is necessary only to inherit the defective gene from one parent to **have the disorder**. This means that there is a one in two chance of inheriting the disease but there are no asymptomatic carriers because one copy of the gene is all that is required for symptoms.
- (c) **A sex-linked disorder** – Males and females are distinguished by one pair of sex chromosomes. In this pair a female has two **x** chromosomes and a male has one **x** and one **y** chromosome. Therefore if a foetus has a **y** chromosome from the male, the child too will be a male. If an **x** chromosome is inherited from the father the child will be female. Muscular dystrophy may be sex-linked on the **x** chromosome. The male has only one **x** and so, if transmitted, an offspring **male** will have the disease. The female has two **x** chromosomes and, if one is defective, the normal healthy chromosome

usually operates normally, meaning that a female either has no symptoms or only mild symptoms.

Alfredo Ferrari, son of Enzo Ferrari, was born in 1955 with Duchenne muscular dystrophy.

He designed the Ferrari 1.5 litre overhead cam V6 engine, which he never saw produced because he died in 1956 aged 24.

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