HEALTH EDUCATION CORNER

MUSCULAR DYSTROPHY

Overview

Muscular dystrophy is the name given to a group of inherited muscle diseases that cause progressive degeneration and weakness of the muscles. Muscular dystrophy can occur at any age. People affected by neuromuscular disorders have different degrees of independence, mobility and carer needs. Each of the approximately 60 neuromuscular disorders has a separate cause.

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

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.

To obtain more details on the specific types of Muscular Dystrophy you should consult the website.

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.

Source

The information in this article has been sourced from the Muscular Dystrophy Australia website.