Muscular Dystrophy Tasmania

Facts about Later Onset Muscular Dystrophy

At what age do signs of dystrophy appear?

Contrary to the wide spread notion that muscular dystrophy is an exclusively childhood disorder, clinical onset may occur at any point in the life span. The different types of the disease vary in the age at which muscle wasting becomes manifest and in the muscle groups first affected.

This fact sheet deals mainly with those muscular dystrophies of later onset. The Muscular Dystrophy Association produces a separate pamphlet on the early onset disorders namely Duchenne and Becker Muscular Dystrophy.

Does the rate of progression vary?

Degeneration of muscle in muscular dystrophy is a continuing process, with considerable variation in its rate and severity among the different forms of the disease.

As a rule, it can be said that the earlier the clinical signs appear, the more rapid the progression and the more widespread and disabling the deterioration.

As muscles deteriorate, people become weaker. In the severe forms of the disease, people lose the power to walk and need to use wheelchairs for mobility. In such cases, they are finally unable to carry out the simplest activities of everyday life. They cannot combat other infections, and death usually results from respiratory disease; it also may be hastened by involvement of heart muscle.

How is a diagnosis of muscular dystrophy established?

The age of onset, distribution and severity of muscle weakness, and the pattern of inheritance indicated by a family history provide essential information in the diagnosis of muscular dystrophy.

Examination of a muscle biopsy is the most definitive procedure for confirming the presence of degeneration. Degeneration of the abnormal gene is now possible for some forms of MD.

Electromyography is also a valuable diagnostic tool, as is the measurement of various serum enzymes.

Is muscular dystrophy always inherited?

It is now well established that all forms of muscular dystrophy are hereditary conditions with the genetic defect transmitted by one parent in some forms of the disease and by both parents in other types. However, there are many cases of muscular dystrophy in families with no known history of the disease. This is explained, in Duchenne dystrophy by a high spontaneous mutation rate, in other words the genetic fault has begun in the affected person.

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Is there any treatment for muscular dystrophy?

At this time, there is no known treatment that will arrest or reverse the dystrophic process, but medical management can increase mobility, maximise independence in daily activities, and ease discomfort. The use of orthopedic devices and physiotherapy, for example, can keep people walking longer, minimise crippling contractures, and prevent or delay curvature of the spine.

What are the specific forms of muscular dystrophy?

The following descriptions summarize the major characteristics of the various types of Muscular Dystrophy (MD). Duchenne Muscular Dystrophy and Becker Muscular Dystrophy are discussed in a separate pamphlet.

Limb-Girdle Muscular Dystrophy

Clinical onset of the disease occurs anywhere from the first to the third decade of life. The initial muscles affected are the proximal muscles of the pelvic and shoulder girdles.

The progression of Limb-girdle Muscular Dystrophy varies considerably, as does the degree of dis ability. Progression is sometimes quite slow and sometimes fairly rapid although never as rapid as in the Duchenne type. When progression is slow, people may have a normal life span.

The hereditary pattern in Limb-girdle Muscular Dystrophy is usually autosomal recessive. Unless both parents carry the defective gene, none of their children will manifest the disease. When both parents carry the gene, each offspring has a 25 percent probability of being clinically affected, a 50 percent probability of being normal but carrying the defective gene, and 25 percent probability of being completely free of the hereditary defect. Sons and daughters are equally at risk.

Facioscapulohumeral Muscular Dystrophy

Clinical onset of the disease usually occurs in early adolescence, occasionally as late as the mid-20's and sometimes in infancy. There is marked variability in the severity of symptoms from person to person, as well as in the age of onset. As indicated by the name of the disease, initial involvement occurs in the muscles of the face and shoulder girdle. There is a resulting lack of facial mobility, difficulty in raising arms over the head, and a characteristic forward slope of the shoulders. The progression of Facioscapulohumeral Muscular Dystrophy is very slow as a rule, with plateaus of significant duration. Average life span is rarely shortened, although people may suffer considerable disability.

The hereditary pattern is autosomal dominant. In this form of inheritance, a trait is transmitted by a single gene derived from one parent. The carrier of a dominant disease gene usually suffers from the disorder. There is a 50 percent probability of incidence among offspring - male or female. If there is no family history of the condition, then it may have begun in the affected person, or that person might have a parent affected so mildly that the condition is difficult to detect

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Myotonic Muscular Dystrophy

Clinical onset of Myotonic Muscular Dystrophy, also known as Steinert's disease, may occur at any age including infancy, but is most frequent between 20 and 35. Myotonia (delayed relaxation of muscles after contraction) and facial weakness are among the earliest and most characteristic features of Myotonic Dystrophy. Weakness of the feet and hands is another common early sign, as is weakness of the anterior muscles of the neck.

The progression of the disease is typically slow. Disability rarely becomes severe until 15 to 20 years after the onset of symptoms. A distinctive characteristic of Myotonic Dystrophy is involve ment of other parts of the body - such as the central nervous system, smooth muscles, endocrine glands and eyes, in addition to the voluntary musculature.

The hereditary pattern in Myotonic Muscular Dystrophy is autosomal dominant: the defective gene may be inherited from either side of the family. There is a 50 percent probability of incidence among offspring.

Ophthalmoplegic Muscular Dystrophy

Progressive weakness of the muscles around the eyes leads to droopy eyelids and difficulty in moving the eyes around. This condition is also known as Progressive External Ophthalmoplegia. It appears that this condition is most frequently associated with a genetic mutation affecting a small amount of DNA that is not transmitted in the chromosomes but rather in the mitochondria. The mitochondria are subcellular organelles, which are responsible for energy production, and each mitochondria has a small sequence of DNA which can develop mutations similar to those seen in the chromosomes which are contained in the nucleus of the cell. It appears that an individual obtains his or her mitochondria from his or her mother's egg and so disorders associated with abnormal ties can also be responsible for more diffuse muscle weakness, a condition known as mitochondrial myopathy.

Distal Muscular Dystrophy

The chief distinguishing characteristic of Distal Muscular Dystrophy is the initial and primary involvement of the small muscles of the extremities. The resulting impairment is frequently confused with Charcot-Marie-Tooth disease, a disorder of peripheral nerve. Distal Muscular Dystrophy is the rarest subgroup of the dystrophies. In Sweden, however, its incidence is comparatively high and as yet unexplained. The pattern of inheritance is autosomal dominant.

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