

SCHOOL HEALTH MANUAL

MUSCULAR DYSTROPHY

MAINE SCHOOL HEALTH ADVISORY COMMITTEE

with input from

MAINE DEPARTMENT OF EDUCATION

MAINE DEPARTMENT OF HUMAN SERVICES

AND OTHER RELATED ORGANIZATIONS

COMMENTS

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Comments may be given to members of the School Health Advisory Committee or sent to the below.

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MUSCULAR DYSTROPHY

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Muscular dystrophy is the general designation for a group of chronic, hereditary diseases characterized by the progressive degeneration and weakness of voluntary muscle.

Contrary to the widespread 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. The most common form of muscular dystrophy strikes children between the ages of two and six.

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. Generally, the more rapid the progression and the more widespread and disabling the deterioration.

As muscles deteriorate, patients become weaker. In the severe forms of the disease, patients lose the power of ambulation and are confined to wheelchairs, and eventually to bed. In such cases, they are finally unable to carry out the simplest activities of everyday life. They cannot combat intercurrent infections, and death usually results from respiratory disease; it also may be precipitated by involvement of heart muscle.

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. Because other neuromuscular disorders are not always distinguishable from various forms of muscular dystrophy on clinical grounds alone, a diagnosis of dystrophy may require direct evidence of muscle degeneration. Examination of a muscle biopsy is the most definitive procedure for confirming the presence of such degeneration. Seen microscopically, dystrophic muscle has a characteristically abnormal appearance, showing wide variation in fiber diameter and infiltration with fat and connective tissue. Electromyography is also a valuable diagnostic tool as is the measurement of various serum enzymes.

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. The thesis of a high spontaneous mutation rate in Duchenne dystrophy, which has been advanced to explain these sporadic cases, is now being questioned by certain investigators.

The basic defect in any hereditary disorder resides in the patient's genetic material. In muscular dystrophy, through pathogenetic processes that are not completely understood, the genetic defect finds expression in the biochemical and structural degradation of muscle cells. Extensive research has established numerous biochemical abnormalities in muscular dystrophy, but these appear to be secondary to the disease process rather than its immediate cause.

Among the abnormalities are defects in the surface and interior membranes of the muscle cell, as well as in the surface membranes of the blood cells. Such evidence suggests to many researchers that generalized membrane defects are implicated in the pathogenesis of muscular dystrophy, but these appear to be secondary to the disease process rather than its immediate cause.

Presently, there is no known treatment that will arrest or reverse the dystrophic process, but medical management can increase mobility, maximize independence in daily activities, and ease

the patient's discomfort. The use of orthopedic devices and physical therapy can keep patients ambulatory longer, minimize crippling contractures, and prevent or delay curvature of the spine.

Levels of the enzyme creatine kinase (CPK) and certain other enzymes are significantly elevated in the blood serum of female carriers of Duchenne dystrophy. Laboratory tests measuring the serum levels of these enzymes are 70-80 percent effective in identifying the carrier state in Duchenne muscular dystrophy. There are no tests at the present time for carrier detection in other types of dystrophy. Carrier testing for Duchenne muscular dystrophy is available at MDA clinics.

DUCHENNE MUSCULAR DYSTROPHY

Clinical onset of Duchenne muscular dystrophy typically occurs in boys between the ages of two and six; occasionally soon after birth. The disease starts so insidiously that it may go unnoticed for many months or even years. The child may have difficulty climbing stairs and rising from sitting or lying positions. There is a tendency to fall frequently. Later, the child may develop a waddling gait. Serum levels of creatine kinase (CPK), a muscle enzyme, are elevated in Duchenne muscular dystrophy even before clinical signs of the disease appear. These high CPK levels in blood serum are a consequence of abnormal muscle cell membrane function, which results in the loss of CPK and other enzymes from dystrophic muscle.

A distinctive characteristic of Duchenne muscular dystrophy, although not invariable, is the seeming enlargement (pseudohypertrophy) of calf muscles, caused by deposition of fat and connective tissue. Progression is rapid with no remission and is marked by wasting of proximal muscles - particularly in the pelvic girdle - followed by involvement of the pectoral muscles, and finally of all muscle groups. In this, the most common and severe type of muscular dystrophy, few patients survive their early 20's. Antibiotic therapy and other advances in dealing with pulmonary complications have been major factors in extending life expectancy.

The hereditary pattern in Duchenne muscular dystrophy is of the recessive X-linked type, in which the defective gene is transmitted by the mother, almost exclusively to male children. There is a 50% probability that any male child of a female carrier will develop Duchenne muscular dystrophy and a 50% probability that any female offspring will be a carrier of the defective gene. Carriers of Duchenne dystrophy are themselves unaffected.

BECKER MUSCULAR DYSTROPHY

Clinical onset of Becker muscular dystrophy occurs between the 5th and 25th years in males. The first clinical signs are similar to those of Duchenne dystrophy but considerably less severe. Pseudohypertrophy of calf muscles is common, and CPK levels can be as grossly elevated in cases of Becker muscular dystrophy as in Duchenne.

Progression, in contrast to that in Duchenne muscular dystrophy, is comparatively slow, although it follows a similar course, with progressive weakness and wasting of the pelvic and later the pectoral muscles. However, weakness is less pronounced than in Duchenne and the disease much more benign in its consequences. The patient often lives out a normal life span. The hereditary pattern in Becker muscular dystrophy is X-linked recessive.

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 disability. Progression

is sometimes quite slow and sometimes fairly rapid - although never as rapid as in the Duchenne type. When progression is slow, patients may have a normal life span.

The hereditary pattern is 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% probability of being clinically affected, a 50% probability of being normal but carrying the defective gene, and a 25% 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 patient to patient, as well as in the age of onset. 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 patients 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% probability of incidence among offspring - male or female.

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 involvement 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 is autosomal dominant: the defective gene may be inherited from either side of the family, with 50% probability of incidence among offspring.

CONGENITAL MUSCULAR DYSTROPHY

In this less familiar form of muscular dystrophy, the most progressive and active phase of muscle degeneration takes place during the fetal period, and the disease is already manifest at birth. Its essential features include hypotonia, muscle weakness, and contractures - all present at birth - and some functional improvement during childhood, with little or no progression thereafter. The pattern of inheritance is probably autosomal recessive.