



Muscular Dystrophy

Symptoms or Behaviors

Each form of muscular dystrophy is caused by a defect in a gene. All forms are hereditary conditions, with the genetic defect transmitted by one parent in some forms of the disease and by both parents in other types.

Many thousands of Americans suffer from muscular dystrophy. Almost 2/3 of these cases are children. Symptoms may appear any time between birth and adolescence. However, muscular dystrophy can strike anyone at any age. Some forms develop in the teens; others in the twenties, or even middle age.

About the Disorder

There are four main types of muscular dystrophy: Duchenne, Facioscapulo-humeral, Limb-Girdle, and Myotonic.

Duchenne Muscular Dystrophy usually develops in boys between the ages of two and six. Early signs are often overlooked. 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 the muscle enzyme CPK are elevated in Duchenne Muscular Dystrophy even before clinical signs of the disease appear.

A distinctive characteristic of Duchenne Muscular Dystrophy is the seeming enlargement of calf muscles, caused by the 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. The hereditary pattern in Duchenne MD is of the recessive X-linked type, in which the mother transmits the defective gene almost exclusively to male children. There is a 50% probability that any female offspring will be a carrier of the defective gene.

Facioscapulo-Humeral Muscular Dystrophy usually occurs in early adolescence, occasionally as late as the mid-20's, and sometimes in infancy. There is marked variability in the severity and onset of symptoms from patient to patient. Initial involvement occurs in the muscles of the face and shoulder girdle. There is a resulting lack of facial mobility (difficulty closing eyes and whistling, unlined face even in middle age); difficulty in raising arms over the head, and a characteristic forward slope of the shoulders. The progression is very slow as a rule, with plateaus of significant duration. Average life span is rarely shortened, although patients may suffer considerable disability. Trunk and leg muscles may become involved, and the person may be unable to walk. The hereditary pattern is autosomal dominant. In this form of inheritance, a single gene derived from one parent transmits a trait. The carrier of a dominant disease gene usually suffers from the same disorder. There is a 50% probability of incidence among offspring – male or female.

Limb-Girdle Dystrophy occurs anywhere from the first to the third decade of life. It begins in either the shoulder muscles or muscles of the lower trunk and upper legs. The symptoms vary with part of the body affected. When the shoulders are affected, the individual may have difficulty raising arms and lifting objects, and have drooping shoulders. If the legs and trunk are affected, the student may waddle, have frequent falls, and/or have difficulty rising from the floor, and climbing stairs. The progression of the disease varies. If it begins in the shoulder muscles, it is generally slower. Sometimes the progression is very rapid. The course is unpredictable, but most become severely disabled and unable to walk in middle age. Life span is usually shortened. The hereditary pattern is autosomal recessive. Unless both parents carry the gene, each offspring has a 25% probability of being completely free of the hereditary defect. Sons and daughters are equally at risk.

Myotonic Muscular Dystrophy, also known as Steinert's disease, may occur at any age including infancy, but is most frequent between 20 and 35. The first signs are weakness of the fingers, hands, forearms, feet and lower legs. Facial muscle weakness (drooping lids, jaw) may also be initially apparent. There may be stiffness in the hands and feet, especially after being chilled. The individual may have difficulty relaxing his/her grip, and may also trip and fall frequently. The hereditary pattern is autosomal dominant; the defective gene may be inherited from either side of the family. There is 50% probability of incidence among offspring.

Other types of muscular dystrophy include Becker MD, Congenital Dystrophy, Distal MD, Ophthalmoplegic MD and Oculopharyngeal Dystrophy. Becker MD is similar to Duchenne but starts later in life and is less severe. The life span may be normal. Congenital Dystrophy takes place during the fetal period and the disease is already manifested at birth. The essential features include hypotonia, muscle weakness, and contractures. Distal MD involves the small muscles of the extremities, and is the rarest subgroup. Ophthalmoplegic MD usually shows in adulthood. Extraocular muscles are involved initially and the muscles used in swallowing tend to become affected. Oculopharyngeal usually appears in adulthood. It affects the eyes and throat. It progresses slowly but steadily

Educational Implications

- Extra set of books at home
- Evacuation plan
- Rest periods if needed
- DAPE, OT, PT
- Assistive devices (computer, pencil grips, eating utensils, Hoyer lifts, etc.)
- Extra time for assignments
- Pupil support assistant to assist with ADL's

Resources

Facts About Muscular Dystrophy, Muscular Dystrophy Association

Muscular Dystrophy, MDA

What Everyone Should Know About Muscular Dystrophy, MDA