Muscular Dystrophy

Frequently Asked Questions (FAQs)

INDIAN ACADEMY OF NEUROLOGY

A Public Information Initiative

Q. 1. What is muscular dystrophy?

Ans. Muscular dystrophies are a group of muscle diseases that are characterized by progressive loss of muscle mass and weakness. Crucial to muscle function are multiple muscle proteins that are needed to work in close association with each other. A defect in any muscle protein may impair functioning of other muscle proteins leading to weakness. Muscular dystrophies are caused by inheriting abnormal genes which in turn lead to defective or absent muscle protein formation. Muscular dystrophies are named according to the absent of defective protein: e.g., Duchenne muscular dystrophy (DMD) is due to absence of the protein dystrophin, sarcoglycanopathy due to abnormality of the protein sarcoglycan, and so forth.

Q.2. At what age can muscular dystrophy develop?

Ans. Certain muscular dystrophies like DMD develop in the first decade of life (disease only in boys); whereas others like limb-girdle muscular dystrophy can develop anytime between 10- 40 years of age (can occur in both males and females). Still others like, oculo-pharyngeal muscular dystrophy develop after 50 years of age.

Q. 3. What are the symptoms of muscular dystrophy?

Ans. Most patients with muscular dystrophy present with limb weakness (either in the hands or legs). These start gradually but progress over the years. Frequent falls in a child may be the earliest symptom of DMD. Most adults with muscular dystrophy manifest as difficulty in climbing up the stairs, getting up from a squatting position or difficulty in raising the arms above. Rarely, muscular dystrophies can manifest at the onset with breathing or swallowing abnormalities.

Myotonic muscular dystrophy is a peculiar type of muscular dystrophy where the affected patient has muscle weakness and myotonia (difficulty in rapidly releasing objects from the hands). Patients may also have facial weakness (difficulty in blowing air or spitting) and eye muscle weakness (drooping of eyelids).

Q.4. Do patients with muscular dystrophy have symptoms other than muscle weakness?

Ans. Certain muscular dystrophies can also lead to abnormal pumping of the heart (cardiomyopathy), heart rhythm abnormality, or skeletal abnormalities like, bent or rigid spine.

Q. 5. What are the common types of muscular dystrophy?

Ans. Some common types of muscular dystrophy are:

Duchenne Muscular Dystrophy

This develops in male children in the first decade of their life. They have abnormality of the Xp21 gene (as females have double copy of the X chromosome, they do not manifest this disease). Frequent falls, difficulty in climbing up the stairs and a waddling gait are the main features. Most of the affected children are wheelchair-bound by 12- 14 years of age.

Limb-Girdle Muscular Dystrophy

This represents a big group of muscular dystrophies with different disease types. Most individuals present between the ages of 20-40 years with progressive weakness in the hip or shoulder muscles.

Facioscapulohumeral Muscular Dystrophy

The affected individuals have weakness of the facial muscles, abnormal movement of the scapula on lifting the arms in front (winging of the scapulae), and hip muscle weakness.

Myotonic Muscular Dystrophy

In addition to muscle weakness (which can start as a weak hand-grip) and myotonia (inability to relax grip on an object), the involved persons may also have frontal balding, droopy eyes and early cataracts.

Q. 6. How is muscular dystrophy diagnosed?

Ans. Muscular dystrophy is suspected clinically when an individual has progressive muscle weakness, especially if there are other family members involved. However, the affected individual can be the only person in the family with the disease. Laboratory tests are done to confirm the presence of muscular dystrophy, e.g., elevated muscle enzyme CPK, abnormal EMG (electromyography) and muscle biopsy. Muscle biopsy helps in identifying the type of muscular dystrophy. Genetic testing is now available in many of the common muscular dystrophies like DMD, limb-girdle muscular dystrophies, etc.

Q. 7. How is muscular dystrophy treated?

Ans. Though the abnormal proteins in most of the muscular dystrophies are now known, yet a specific cure for most of these diseases remain elusive. Steroids are beneficial in patients with DMD. In most of the patients with muscular dystrophy, physiotherapy exercises for strengthening the muscles, as well as some breathing exercises are essential. Those with cardiac rhythm problems may need a pacemaker to be implanted.

Q. 8. Is there any recent medical breakthrough for patients with muscular dystrophy?

Ans. Research is proceeding at a fast pace throughout the world to try and find a cure for most of the muscular dystrophies. In those with DMD, genetic manipulation through a procedure called exon skipping is being tested. Through exon skipping, some functioning protein can be formed in those with DMD. In GNE myopathy, sialic acid is being tested in trials and appears promising.

Q. 9. Can I test prenatally, if my child will get muscular dystrophy?

Ans. This is possible for some of the dystrophies like DMD. Usually, during second or subsequent conceptions, couples seek help for prenatal diagnosis when one child has already been suffering from DMD. The genetic make-up of the child who has already suffered the disease is important for accurate prenatal diagnosis.

Disclaimer:
This brochure is for the general information of the public and the patients. People should not self-medicate themselves with the medicines and treatments mentioned here. Before taking any of the medications mentioned in the information brochure, please consult your neurologist.

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