Q. What is muscular dystrophy?

Ans. Muscular dystrophy (MD) refers to a group of more than 30 genetic diseases that cause progressive weakness and degeneration of skeletal muscles. Muscular dystrophies are progressive, and affected individuals eventually lose the ability to walk.

Q. What are the symptoms of muscular dystrophy?

Ans. The clinical feature varies according to the type of MD. Delayed attainment of walking**,** recurrent falls, difficulty in climbing stairs and getting up from sitting posture are common complaints. Often these children are clumsy while walking and not able to run as peers and easily fatigued. Children with muscular dystrophy can have enlarged calf or pain in calf muscles. Toe walking and foot deformities are common.

Q. What causes muscular dystrophy?

Ans. All muscular dystrophies are inherited disorders and involve a mutation in the genes that program proteins critical to muscle integrity. Many muscular dystrophies are familial, meaning there is other family members affected with the disease.

Q. How common is muscular dystrophy?

Ans. MD occurs worldwide, affecting all races. Its incidence varies, as some forms are more common than others. It's most common form in children, Duchenne muscular dystrophy affects approximately 1 in every 3,500 to 6,000 boys.

Q. How does muscular dystrophy cause muscle weakness?

Ans. Our muscles are made up of thousands of muscle fibers. Each muscle fiber is an individual muscle cell that contains several proteins involved muscle fiber contraction, relaxation and membrane stabilization. Lack of a particular protein or dysfunctional protein leads to muscle fiber damage. Because of ongoing muscle fibers damage, our muscles tend to get weak, thin and short.

Q. What are the types of muscular dystrophies?

Ans. There are >40 types of muscular dystrophies. Most common muscular dystrophies with childhood-onset are Duchenne muscular dystrophy, Becker muscular dystrophy, and congenital muscular dystrophy. Other less common conditions are Limb-girdle muscular dystrophy, Emery dreifuss muscular dystrophy and Facioscapulo-humeral muscular dystrophy. Muscular dystrophies vary by the age of onset, pattern of involvement and rate of progression.

Q. What is Duchenne muscular dystrophy?

Ans: Duchenne muscular dystrophy is the most common childhood form of MD, as well as the most common of muscular dystrophies accounting for approximately 50% of all cases. Duchenne muscular dystrophy primarily affects boys; they present with frequent falls, difficulty in climbing stairs noted from 4-5 years of age and loss of ambulation by ten years of age.

Q. What are the other complications of muscular dystrophy?

Ans. Muscular dystrophy also affects our other organs, like heart, bones, respiration, and brain. The heart muscles involvement may present with rhythm abnormalities or heart failure. Respiratory muscle weakness in muscular dystrophy may require ventilator support.

Q. How are the muscular dystrophies diagnosed?

Ans. Diagnosis of muscular dystrophy is based on the history and examination findings. Several investigations can help in diagnoses such as creatine kinase, nerve conduction study and electromyography. The diagnosis of muscular dystrophy is confirmed by muscle biopsy, and genetic testing.

Q. How are the muscular dystrophies treated?

Ans. There is no specific treatment for muscular dystrophies. However, available treatments are aimed at keeping the person independent for as long as possible and prevent complications that result from weakness, reduced mobility, and cardiac and respiratory difficulties. Treatment may involve a combination of approaches-

- Physical therapy and regular muscle stretches
- Orthotic devices
- Drug therapy
- Surgical correction of contractures.

Regular muscle stretching and exercise is necessary to maintain the muscle strength and prevention of contractures. Drug therapy may help in reducing muscle degeneration. Corticosteroids may reduce the rate of progression of weakness in Duchenne muscular dystrophy. Orthotic devices such as ankle-foot orthosis, night splints help in improving mobility.

Q. What are the home modifications to help the patient with muscular dystrophy?

Ans. The environmental modification can help the patient in daily activity. Supporting bars in the bathroom, removal of obstacles in the home, ramp in place of stairs may help the patients with muscular dystrophy.

Q. What is the prognosis?

Ans. The prognosis varies according to the type of muscular dystrophy, and the speed of progression. Some types are mild and progress very slowly, allowing normal life expectancy, while others are more severe and result in functional disability and loss of ambulation.

Q. What research is being done?

Ans. Although muscular dystrophies are not curable at present, however research related their natural history, immunomodulation therapy, and gene therapy is ongoing. Exon skipping therapy has been approved in some countries for the treatment of Duchenne muscular dystrophy.