

DMD is the most lethal, can be controlled with right treatment

05 June 2017 | Views

DMD is an inherited disease(X linked disorder recessive). However there is a possibility for a family to have a son with the disease, with no any previous family history. Dr. Aashish Chaudhry Orthopedic Surgeon and Managing Director, Aakash Healthcare shares his views on DMD



Inherited diseases are the ones which are caused due to DNA abnormalities. Duchenne muscular dystrophy (DMD) is a genetic disorder characterized by progressive muscle degeneration and weakness. All forms of Muscular Dystrophies are rare, but Duchenne Muscular Dystrophy (DMD) is the commonest of all and the most lethal. DMD is caused by an absence of a special kind of protein called 'Dystrophin'. The disease primarily affects boys, but in rare cases it can affect girls also.

DMD is an inherited disease(X linked disorder recessive). However there is a possibility for a family to have a son with the disease, with no any previous family history. It is possible to diagnose DMD in antenatal period by detailed DNA analysis and/or fetal muscle biopsy, but the facility is not available at all places and most of the patients can't afford it. However, it is advised that couples with positive cases in the family (X linked recessive patients), should at least try and attempt prenatal diagnosis of DMD.

Common signs and symptoms of DMD include:

- The calf muscles may appear enlarged & bulky in patients with DMD
- Children with DMD have difficulty climbing stairs, getting up from floor or running, and may seem clumsy and fall often
- They may also experience difficulty in raising their arms
- May experience pain and altered sensation in their legs, and often experience cramps. If any of the symptoms can be seen in the child, consulting a health expert is must.

Lack of dystrophin can weaken the muscle layer in heart (myocardium), resulting in a condition called cardiomyopathy. DMD may create serious damage to heart over the years, and can become life-threatening. At times, DMD causes respiratory problems, as the disease weakens the muscles that operate the lungs. Also it may cause learning disability in children, and in rare cases the child may have serious mental retardation.

Diagnosis of DMD starts with investigating the history of the parents. Physical examination of the child is done in order to understand the complexity of the disease. First, the CK level of blood is being tested. CK known as creatine kinase is an enzyme that leaks out of damaged muscle cells. If test reports shows high CK level in blood that means muscles are ruptured due to some abnormal process.

Further tests involve muscles biopsy i.e the surgical process of taking a small piece of muscle from the patient and checking it under microscope. By examining the tissue, the doctors can specifically diagnose the disease.

There is no known cure for Duchenne Muscular Dystrophy. However the use of available treatments can control the symptoms and can help in improving the quality of life. Steroids are given to slow down the loss of muscle strength. Exercises can help build skeletal muscle and keep the cardiovascular system healthy. However, not undergoing any physical activity leads to excessive weight and obesity. And obesity is harmful for bones and joints. Speech therapy is often needed.

Bringing up a child with any disability is not a problem if the parents understand and take the child through proper diagnosis and treatment. Always take proper guidance and medical assistance from your health expert.