

# Duchenne Muscular Dystrophy

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**In news**– Researchers in India are working on developing an affordable treatment for a rare and incurable genetic disorder called Duchenne Muscular Dystrophy(DMD) with over 5 lakh cases in the country.

## Key updates-

- **The current therapeutic options available to treat DMD are minimal and highly expensive treatment** with costs shooting up to Rs 2-3 crore per child a year and are mostly imported from abroad, accelerating dosing costs and putting them out of reach for most families.
- **The Indian Institute of Technology (IIT), Jodhpur has established a research centre for DMD in collaboration with Dystrophy Annihilation Research Trust (DART), Bengaluru** and the All India Institute of Medical Sciences (AIIMS) Jodhpur.
- The centre aims to develop affordable therapeutics for this rare and incurable genetic disorder.
- **The researchers are working on affordable therapeutics for DMD** and enhance the efficacy of Antisense Oligonucleotide (AON)-based therapeutics.
- According to researcher, the AON-based therapeutics' idea is to hide or mask specific exons (a segment of a DNA or RNA molecule containing information coding for a protein) in a gene sequence.
- "In DMD patients, one or more exons can be masked with specific molecules called AON or molecular patches. Due to these challenges, DMD patients need personalised medicine.
- Currently, the research team is also working on reduction of AON based therapeutic dose through new molecular tags.
- Until recently, boys with DMD usually did not survive

much beyond their teen years. However, with advances in cardiac and respiratory care, life expectancy is increasing.

### **What is Duchenne Muscular Dystrophy?**

- It is a severe type of muscle weakness that usually begins at an early age and worsens quickly, may soon have a new strategy of treatment through genetic regulation.
- **DMD is an X-linked recessive muscular dystrophy affecting roughly one in 3,500 boys, which causes gradual loss of muscle tissue** and function eventually leading to wheelchair dependency at **approximately the age of 12 years**, requirement for assisted ventilation at approximately the age of 20 years and eventually premature death.
- **Currently, there is no cure for DMD**, but improvements in integrative treatment can slow down the disease progression and thereby, extend the life expectancy of DMD patients.
- **Patients with DMD have different forms of mutations at varying positions of the protein**, resulting in the production of functionally compromised dystrophin ORF.
- **According to scientists, muscle weakness is the principal symptom of DMD.**
- **It can begin as early as age 2 or 3, first affecting the proximal muscles** (those close to the core of the body) and **later affecting the distal limb muscles** (those close to the extremities).
- Usually, the lower external muscles are affected before the upper external muscles. The affected child might have difficulty jumping, running, and walking.
- **Other symptoms include enlargement of calves, a waddling gait**, and lumbar lordosis (an inward curve of the spine). Later on, heart and respiratory muscles are affected as well.

- **Progressive weakness and scoliosis result in impaired pulmonary function**, which can eventually cause acute respiratory failure.