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, r**equirement 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.