World Duchenne Muscular Dystrophy Day

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Why is in news? World Duchenne Muscular Dystrophy Day observed by Department of Empowerment of Persons with Disabilities (DEPwD)

World Duchenne Muscular Dystrophy Day is observed on **7th September** each year to raise awareness about Duchenne muscular dystrophy.

The purpose of the day is to support efforts that will improve the quality of life for those who have dystrophinopathies through education, advocacy, and social inclusion.

This year's World Duchenne Muscular Dystrophy Day **theme is Duchenne: Breaking Barriers**.

A **rare degenerative illness** called Duchenne muscular dystrophy.

It was first described by the French neurologist Guillaume Benjamin Amand Duchenne in the **1860**.

It is characterized by **progressive muscle degeneration and weakness due to the alterations of a protein** called **dystrophin** that helps keep muscle cells intact.

It is a **multi-systemic condition**, affecting many parts of the body, which results in deterioration of the skeletal, heart, and lung muscles.

The dystrophin gene is found on the X-chromosome, it primarily affects males, while females are typically carriers.

One in five thousand boys are born with the condition.

Symptoms:

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.

First, walking becomes challenging, then other motor functions follow, and finally, breathing and heart function are affected because the heart is also a muscle.

The affected child might have difficulty jumping, running, and walking.

Other symptoms include enlargement of the calves, a waddling gait, and lumbar lordosis (an inward curve of the spine).

Later on, the heart and respiratory muscles are affected as well.

Learning and behavioral problems may potentially be a symptom of the condition because the missing protein also has a purpose in the brain.

Treatments: Presently available treatments are **gene therapy, exon skipping, stop codon read-through and gene repair.**