ABOUT DUCHENNE MUSCULAR DYSTROPHY

WHAT IS DUCHENNE MUSCULAR DYSTROPHY?

• Duchenne muscular dystrophy (DMD) is a rare and severe form of muscular dystrophy primarily affecting males.
• It is caused by a mutation in the dystrophin gene located on the X chromosome. Dystrophin is important for muscle cell stability.
• Muscles to become weaker over time until it affects the whole body.
• One in 5,000 boys are born with DMD worldwide.
• The DMD gene mutation can be passed from the mother, or can happen spontaneously.
• A woman with a genetic change in one of her two copies is said to be ‘a carrier’.
• DMD is a progressive disease. First, walking becomes difficult. Then, other motor functions follow until it ultimately affects the ability to breathe as well as the function of the heart.

WHAT ARE EARLY SIGNS OF DUCHENNE MD?

• Signs of DMD usually are recognized when kids are very young. However, diagnosis often takes place at the age of 4 or even later.
• Symptoms include muscles getting weaker over time, trouble walking, falling down a lot, and being slower to reach functional milestones like crawling or walking.
• The missing protein also has a function in the brain, so learning- and behavior issues can also be part of the disease.
• The diagnosis typically involves a series of clinical evaluations, physical examinations, and genetic testing.

HOW DO YOU CARE FOR A PERSON WITH DUCHENNE MD?

• The current standard of care for DMD includes treatment with corticosteroids, giving physical therapy, taking care of the heart, and should be provided by a multidisciplinary team.
• The 2018 Care Considerations provide clinicians with the Standard of Care to be implemented when caring for people living with DMD.
• People with DMD need to be extra careful if they have a fall or bump into something because they are at a higher risk for Fat Embolism Syndrome (FES) in case of a fracture.

IS THERE A CURE OR TREATMENT FOR DUCHENNE MD?

• There are currently 7 (conditionally) approved treatments for DMD that slow down disease progression.
• There is no definitive ‘cure’ for DMD. Muscle tissue that is lost cannot be restored.
• Research efforts are focused on developing disease-modifying therapies, for example gene therapy, exon skipping, and gene editing techniques, to restore or add the missing or defective dystrophin protein.