Duchenne Muscular Dystrophy (DMD)

There are a variety of muscular dystrophies, with Duchenne Muscular Dystrophy being the most severe and common disorder found in childhood. A milder form with a later onset is called Becker Muscular Dystrophy. Duchenne Muscular Dystrophy has an early onset between ages 3-5 years and primarily affects males due to its X-linked recessive inheritance. These disorders cause progressive muscle weakness and atrophy that ultimately debilitates patient mobility.



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Mechanism

X-linked Recessive
X-suit with Recessive-chocolate
This condition is inherited in an X-linked recessive pattern meaning that the gene associated with this condition is located on the X chromosome. Because males only have one X chromosome, one altered copy of the gene is enough to cause the condition. Females, on the other hand, have two X chromosomes, making it unlikely that they will have two altered copies of the gene in order to inherit the condition. So remember, males are much more likely to inherit this disorder. The defect affects dystrophin, a protein involved in anchoring skeletal muscle cells to the basement membrane.

Signs and Symptoms

Muscle Weakness
Weak-drooping-muscle
Skeletal muscles become progressively weaker. This weakness may result in chronic limb contractures due to limited use.

Motor Problems
Motor
Children present with early motor problems, including a delay in walking, running, and climbing stairs.

Gower Sign
Growling-growler
This is a classic maneuver that children with muscular dystrophy employ in order to stand up. Due to lower limb muscle weakness, a child will align the torso parallel to the ground with hands and knees on the ground. Then, the child pushes up with their arms, essentially walking their hands backwards toward the lower limbs. The hands then push off the lower limbs, and the lower back muscles contract in order to erect the torso. These children often present with lordosis due to excessive use and bearing of weight of the lumbar region of the back.

Large Calves
Large Calves
Pseudohypertrophy of the calves is common in this disease. The pseudohypertrophy seen in this disease is due to fatty and connective tissue deposits replacing the muscle of the calves.

Waddling Gait
Waddling Gate
Weakness in the gluteus medius and maximus muscles in the legs and buttocks causes an inability to stabilize the pelvis when walking. These patients present with a classic waddle walk or gait.

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Loss of Ambulation

Loss of Footprints

In Duchenne muscular dystrophy, the child often loses the ability to ambulate on their own by age 12. These children become wheelchair-dependent and then bedridden. Suspect Becker Dystrophy if the child is still ambulating independently past 13 years of age.

Consideration

Cardiac and Respiratory Failure

Dead Heart and Lungs

The disease progresses to affect the heart and respiratory muscles (e.g., the diaphragm). The patient may ultimately require respiratory support as they lose the muscle strength to breathe on their own.

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