Duchenne Muscular Dystrophy

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ABSTRACT

CLINCAL PRESENTATION & EXAM: Duchenne muscular dystrophy (DMD) is an inherited, neuromuscular disorder that affects approximately 1 in 5000 live male births. It is primarily characterized by muscular degeneration due to dystrophin deficiency, a protein found in muscles. There are several varieties of muscular dystrophy disorders—another common one is Becker muscular dystrophy, caused by a truncated, but partially functional DMD gene. DMD begins during childhood, with symptoms starting between 3 and 5 years of age, and it gradually progresses throughout the lifespan of an individual. The first apparent physical sign is an abnormal, waddling-like motion while walking, indication of a weak, unsupportive hip extensor. The patient has great difficulty performing regular ambulatory movements. Patients with DMD display the Gowers' sign, a signal of muscle weakness, where one's hands and limbs are utilized to get into an upright position from a crouched or squatting position. **ANATOMY & PATHOLOGY**: The dystrophin protein functions to connect the cytoskeleton of each muscle fiber to the underlying basal lamina. At its onset, DMD predominantly affects proximal muscles, especially in the lower extremities of the body, spreading to the more distal muscles throughout the body as the disorder progresses. Muscles deceptively seem large and defined in people with the degenerative disease, for muscle is replaced by adipose and connective tissues. Muscle volume continues to decrease over a lifespan, leading to a reduction in motor abilities. In addition to skeletal muscle degeneration, respiratory capabilities are reduced due to deterioration of respiratory lung muscles. Similarly, signs of cardiomyopathy are visible in DMD patients, usually beginning around age 10. **DIAGNOSTIC TESTING & CONSIDERATIONS**: Genetic DNA testing via multiplex polymerase chain reaction (PCR) is utilized to analyze possible locations of mutations on the large, mutation-prone DMD gene. Most cases of DMD are due to mutations resulting from deletions of one or more of the 79 coding exons from the DMD gene. Alternatively, if results from genetic testing are inconclusive, physical diagnosis is performed through a muscle biopsy, during which dystrophin protein levels are quantified. Another method of diagnosis is by testing the creatine kinase levels in a patient; higher levels of creatine kinase are symptoms of muscular dystrophy. Patients are especially prone to being diagnosed with several psychosocial conditions, including autism spectrum disorder, attention-deficit hyperactivity disorder (ADHD), and obsessive-compulsive disorder (OCD). **TREATMENT & RETURN TO ACTIVITY**: Effective Duchenne muscular dystrophy treatment options are available, although there is no comprehensive cure for this dystrophic disorder. In general, bone health can be maintained through regular nutritious intake of calcium and vitamin D. Corticosteroids are pharmaceutical medications that slow muscle degeneration, while orthopedic therapy provides extra support to weakened muscles. Gene therapy is a groundbreaking alternative treatment that restores function of the dystrophin gene by modifying the reading mechanism as to "skip" over mutated exons along the gene. Pertaining to activity, affected patients should be cautious of muscle overexertion. Incorporation of low to moderate aerobic activity into one's lifestyle is suggested.