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Inherited Disorder of Progressive Muscular Weakening, Typically In Boys: A Case Report of Duchene Muscular Dystrophy Patient

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Abstract

Duchenne Muscular dystrophies are a category of hereditary diseases characterised by increasing muscular weakening and degeneration, particularly in skeletal and cardiac muscles. This is more common in guys. It is caused by a mutation in the DMD gene, which can be passed down through families in an X-linked recessive pattern. It frequently affects people from families with no known history of the disease. Mutations in the dystrophin gene can be inherited or occur spontaneously during germ line transmission. Dystrophin deficiency causes DMD. Dystrophin is a protein that helps maintain muscle cells intact by helping to the preservation of the cell membrane of muscle fibres. Duchene muscular dystrophy affects males who do not produce the dystrophin protein in their muscles. Duchene muscular dystrophy is an x-linked recessive genetic mutation that affects two-thirds of the population (mother is the carrier) and one-third of the population (sporadic new mutation). Muscle dystrophy is distinguished from other muscle problems such as neuropathy by a microscopic sample of tissue that indicates the nerve/ neuromuscular junction is unaffected and only the muscle tissue is damaged. Duchene muscle dystrophy (DMD) and Becker muscular dystrophy are two types of muscular dystrophy caused by genetic mutations known as dystrophinopathies. DMD is not seen in females because the Duchene mutatic gene is inactivated by random X inactivation (Lyon hypothesis). Patients are generally asymptomatic if x inactivation is performed on the functioning gene rather than the faulty gene.

Keywords: NIL

Introduction

Case Report

Chief Complaint:

The patient, a two-and-a-half-year-old male, had been walking in an unsteady manner for the previous six months. He went to the hospital because his upper body shook from side to side when he walked with his hands on his knees from a squat.

Physical Examination:

The near-end of the lower limbs exhibited mild atrophy, gastrocnemius muscle hypertrophy, muscular tonus of all four limbs was low, tendon reflex was reduced, and the CPK value was 741U/L (by Rosalki technique, the normal value: 10200U/L). Skeletal muscle exhibited evident atrophy degeneration, as evidenced by a biopsy of muscle tissue. A cranial CT scan revealed that everything was fine. The posterior tibial nerve was shown to be normal using somatosensory evoked potentials. III4, the proband III1's sibling, showed a symptom that was identical to III1. II3 is the proband's uncle (his mother's brother), and III7 is the proband's cousin (the son of his mother's sister). II3 was dead at 23 years old, also had limitation of activity before he died. III7 is three and half years, he had symptom one year ago.

Patient's clinical analysis:

CPK was 741 U/L (typical value: 102000 U/L, according to the Rosalki technique). Somatosensory evoked potential test interpretation: posterior tibial nerve was normal. Biopsy of muscle: skeletal muscle exhibited significant atrophy degeneration, cranial CT was normal. The most specific test for muscular dystrophy is a Creatine Phosphokinase (CPK) measurement (MD). CPK levels that are elevated are suggestive of muscle disorders caused by enzyme leakage from muscle cells. Muscle atrophy is most often seen in those who are temporarily disabled, such as being confined to a wheelchair (for eg: when the patient is confined to a wheelchair). When no tumours, blood clots, fractures, or other abnormalities are shown on a CT scan, the results are deemed normal. SEP monitoring of the posterior tibial nerve is commonly utilised to monitor the spinal cord during scoliosis operations and other surgical treatments in which the spinal cord is at danger. Although drugs, surgery, and other therapies temporarily alleviate muscular weakness, it has recently been shown that directly associating the dystrophin gene is more successful. As a result, in gene therapy, mutations at the gene level are seen. DMD is mostly caused by a frame shift mutation. The frame changes when a nucleotide is lost owing to DNA damage or replication mistakes. Therefore different codons are read resulting in a non-functional protein. In the frame shift mutation type causes a stop codon to appear in the middle which results in an incomplete non-functional dystrophin

For diagnosis, what need to be examined?

A doctor generally begins diagnosing muscular dystrophy by obtaining a patient's and family's medical history and doing a physical examination. These may teach us a lot, including the pattern of weakness. Even before any sophisticated diagnostic procedures are performed, the history and physical examination go a great way toward determining the diagnosis.

Ck Level

Doctors frequently prescribe a CK level test early in the diagnostic procedure. Creatine kinase (CK) is an enzyme that leaks from injured muscle. When high CK levels are detected in a blood sample, it typically indicates that muscle is being damaged as a result of an aberrant process such as muscular dystrophy or inflammation. A extremely high CK level indicates that the muscles themselves (rather than the nerves that regulate them) are the likely source of the weakness, however it does not specify the type of muscle disease.

Genetic Testing

Genetic testing is examining the DNA of any cells (typically blood cells) to determine whether a mutation in the dystrophin gene exists and, if so, where it is located. In the United States, DNA testing for dystrophin mutations is readily accessible. For additional information, speak with your MDA clinic physician or genetic counsellor. Also, read The Genie's Out of the Bottle: Genetic Testing in the Twenty-First Century for more information on acquiring a clear genetic diagnosis.

Discussion

How to treat this disease

Standard Therapies

There is currently no cure for DMD. Treatments are tailored to the exact symptoms that each person is experiencing. Physical therapy and active and passive exercise should be used to increase muscular strength and avoid contractures. Some people may require surgery to address contractures or scoliosis. Contractures can be prevented with the use of braces. Mechanical assistance (such as canes, braces, and wheelchairs) may be required to facilitate walking (ambulation). Individuals with DMD are treated with corticosteroids as a standard of treatment. These medicines reduce the course of muscular weakening and postpone the loss of ambulation by 2-3 years in those who are afflicted. Prednisone and deflazacort are two popular corticosteroid medications used to treat people with DMD (which is not available in the United States). Exondys 51 (eteplirsen) injection was authorised by the FDA in 2016 to treat DMD, making it the first medication to do so. Exondys 51 is only for individuals with a verified dystrophin gene mutation that is susceptible to exon 51 skipping, which affects around 13% of the population with DMD. Sarepta Therapeutics manufactures Exondys 51. Emflaza (deflazacort) was authorised by the FDA in 2017 for the treatment of DMD patients aged 5 and above. PTC Therapeutics distributes Emflaza.

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Clinical Testing and Work Up

Children with DMD should be checked for cardiac involvement on a frequent basis. Severe respiratory distress may necessitate the use of a ventilator to aid breathing in certain people. Affected individuals and their families may benefit from genetic counselling.

Conclusion

Duchenne muscular dystrophy (DMD) is a muscular disease that results in progressive muscle loss. DMD mostly affects men, however in rare cases, it can also affect women. DMD is characterised by progressive weakening and loss (atrophy) of both skeletal and cardiac muscle. Problems learning to sit, stand, or walk, as well as delayed ability to sit, stand, or walk, are early indicators.. The majority of children with DMD use a wheelchair by their early teens. Heart and respiratory problems can begin in youth and escalate to serious, life-threatening complications. Mutations in the DMD gene cause DMD (DNA variations).

DMD is an X-linked recessive illness that can afflict people with no family history of the disease. DMD is diagnosed using symptoms, a clinical examination, and the results of a biopsy to remove a small piece of muscle for examination under a microscope. The results of genetic testing might also help confirm the diagnosis. Although there is no cure for DMD, there are treatments that can help control symptoms.

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