Congenital myopathy: Genetic causes, clinical manifestations, and treatment options.

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Introduction

Congenital myopathy is a group of inherited muscle disorders that present at birth or in early infancy. These conditions are characterized by muscle weakness and structural abnormalities in the muscle fibers. Unlike other types of muscular dystrophies, congenital myopathies are typically non-progressive or only slowly progressive. This overview will cover the pathophysiology, symptoms, diagnosis, causes, treatment, and living with congenital myopathies [1].

Congenital myopathies are caused by genetic mutations that affect the proteins involved in muscle contraction and structure. These mutations lead to abnormalities in muscle fibers, which can be detected through muscle biopsies. The main types of congenital myopathies include: Nemaline Myopathy: Characterized by the presence of rod-like structures called nemaline bodies in the muscle fibers. Mutations in genes such as ACTA1, NEB, and TPM3 are commonly associated with this type [2].

Central Core Disease: Characterized by the presence of central cores (areas lacking oxidative enzyme activity) in the muscle fibers. Mutations in the RYR1 gene are most commonly implicated. Centronuclear Myopathy: Characterized by the central location of nuclei in the muscle fibers. Mutations in genes such as MTM1, DNM2, and BIN1 are associated with this type [3].

Multiminicore Disease: Characterized by multiple small areas (cores) of structural abnormalities in the muscle fibers. Mutations in genes like SEPN1 and RYR1 are often involved. Congenital Fiber Type Disproportion: Characterized by an imbalance in the size of different types of muscle fibers. Mutations in genes such as ACTA1 and TPM3 are commonly associated [4].

The symptoms of congenital myopathies can vary widely depending on the specific type and severity of the condition. Common symptoms include: Muscle Weakness: Typically affects the muscles closest to the center of the body (proximal muscles) such as the hips, shoulders, and neck. Weakness may also be present in the facial muscles and respiratory muscles. Hypotonia: Reduced muscle tone or "floppiness" is often observed in infants [5].

Delayed Motor Milestones: Infants and children may experience delays in achieving motor milestones such as sitting, crawling, and walking. Respiratory Problems: Weakness of the respiratory muscles can lead to breathing difficulties and an increased risk of respiratory infections [6].

Feeding Difficulties: Weakness of the facial and throat muscles can result in difficulties with sucking, swallowing, and feeding. Skeletal Abnormalities: Scoliosis (curvature of the spine), joint contractures, and hip dislocations may occur due to muscle weakness and imbalance [7].

Congenital myopathies are caused by mutations in genes that are critical for muscle function. These mutations can be inherited in various patterns:Autosomal Dominant: Only one copy of the mutated gene is needed to cause the disease. This pattern often results in a milder form of the condition. Autosomal Recessive: Both copies of the gene must be mutated for the disease to manifest. This pattern often results in a more severe form of the condition. X-Linked: The mutated gene is located on the X chromosome. Males are typically more severely affected, while females may have milder symptoms or be carriers [8].

Physical Therapy: To maintain muscle strength, flexibility, and joint mobility. Tailored exercise programs can help prevent contractures and scoliosis. Occupational Therapy: To assist with daily activities and recommend adaptive devices to improve independence and safety. Respiratory Support: For individuals with respiratory muscle weakness, non-invasive ventilation or other respiratory support may be needed. Regular monitoring of respiratory function is important [9].

Nutritional Support: Ensuring adequate nutrition is crucial, especially if feeding difficulties are present. Nutritional counseling and the use of feeding tubes may be necessary in severe cases. Orthopedic Interventions: Use of braces, orthotic devices, and surgical interventions to correct skeletal abnormalities such as scoliosis and joint contractures. Medications: In some cases, medications such as acetylcholinesterase inhibitors may be used to improve muscle function. Experimental therapies targeting specific genetic mutations are also being investigated [10].

Conclusion

Congenital myopathy is a group of genetic muscle disorders that present at birth or in early infancy, characterized by muscle weakness and structural abnormalities in muscle fibers. While there is no cure, advances in medical care and supportive

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therapies have improved the management and prognosis of these conditions. A comprehensive approach to treatment, including physical therapy, occupational therapy, respiratory support, and nutritional management, can help individuals with congenital myopathy maintain function and quality of life. Ongoing research holds promise for more effective treatments and a deeper understanding of this challenging condition. Through continued efforts in scientific research, patient care, and advocacy, the outlook for those living with congenital myopathy continues to improve.

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