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Neurology

A RARE CASE OF MYOTONIA CONGENITA: BECKER'S DISEASE

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ABSTRACT Myotonia congenita is a non-dystrophic inherited disorder due to CLCN1 gene mutation. Becker disease is the most common form causes attacks of weak muscles in legs and arms and men are prone to affect than female population .We describe a case of 13 year old male patient presented with the history of feeling of muscle tightness while initiating movements from age 7 and also had a history of development of musculature like a body builder-progressively. Patient was diagnosed as muscular dystrophy and was taking treatment accordingly. On examination muscles of arm, forearm, thighs, calf, shoulders, abdomen, and back were hypertrophied. Powers of the muscles were normal. On further laboratory investigations CPK level was found to be elevated and other routine blood test was found to be normal. Electromyography and muscle biopsy obtained and diagnosed as Becker's myotonia congenita .Patient was managed with the help of mexiletine and phenobarbitone. Follow up carried out after 6 months of initiation of treatment and had improvement of symptoms.

Summary: Myotonia congenita is rare congenital disorder often remains undiagnosed or misdiagnosed. Proper clinical evaluation and investigations is needed for making the diagnosis .With initiation of medications adequate symptomatic improvement can be provided to the patient.

KEYWORDS : Myotonia congenita, Becker disease, Thomson disease

INTRODUCTION

Myotonic disorders are a heterogeneous group of genetic disorders that are unified by the factor myotonia .Myotonias are classified into dystrophic and non-dystrophic. Myotonia congenita (MC) is a non dystropic specific inherited disorder characterized by inability of skeletal muscles to relax after voluntary movements They are characterized by reduced sarcolemmal chloride conductance due to CLCN1 gene mutation and exaggerated response to stimulation (hyper excitability).[1,2,4] As a result affected individuals have difficulty in relaxing certain muscles after contracting them (myotonia), muscle stiffness (rigidity) and abnormal enlargement of musculature (hypertrophy)resulting in a –herculean- or body builder like appearance.[1,2,3] Myotonic symptoms occur when attempting to move certain muscles after rest. The disorder may be transmitted as either as dominant (Thomsen disease) or recessive form (Becker disease)[4,5]. Thomsen disease was initially described by Thomsen in 1876 through a detailed description of his own disability and that of his family members. This disease general examination demonstrates generalized muscular hypertrophy, action myotonia more pronounced in hands than eyelids. Becker disease is inherited as an autosomal recessive trait and was described by Becker PE in 1950 which is usually presents during the period of 4-12 years of age and symptoms similar to Thomsen disease ,but myotonia tends to be more severe in lower limbs and proximal muscles and with disease progression muscles of arms ,face are eventually affected . Men are more prone to be affected than women population.[5,6,7]This case is Becker type myotonia congenita and the reported incidence of this disorder ranging from 0.3 to 0.6 per 100,000 people [7].

In this case report, we will describe about a patient who was misdiagnosed as Becker's muscular dystrophy and on further evaluation it was diagnosed as myotonia congenita.

CASE REPORT

A 13 year old boy was presented to hospital with history of feeling of tightness of muscle while initiating movements from the age of 7 years onwards. He noticed the problems more during daily activities like standing from sitting position, climbing the steps, elevating the upper limbs above the level of head, initiating brushing, taking weights like bucket of water. He also describes difficulty in unfolding the fist after taking heavy objects. He gives the history that the tightness was getting relieved by around 15-20 seconds on taking rest or while continuing activities and he is able to continue the activity normally as his friends of same age. Also he gives the history of development of musculature like a body builder-progressively from the age of around 8 year although he was getting defeated by his friends in physical activities like arm wrestling. Patient was diagnosed as muscular dystrophy and was taking treatment accordingly. On examination muscles of arm, forearm, thighs, calf, shoulders, abdomen, and back were hypertrophied. Powers of the muscles were normal. While checking power the muscles were becoming hard, with rod like feel and it was getting relieved by itself and the muscle was becoming soft to firm after around 15-20 seconds. He was able to stand from sitting position with the support of hands only. When he was asked to climb the steps, he was able to climb the initial steps slowly only and after climbing 5-6 steps, he was able to continue it without difficulty. He was having hand grip myotonia and percussion myotonia.

On laboratory investigations his CPK level was found to be 198u/dl. Other routine blood investigations including serum calcium and phosphate were within normal limits.

Figure :1

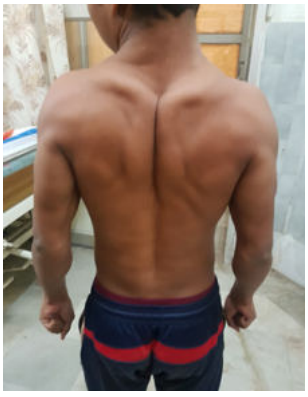

Figure:2

Differential Diagnosis

1. Duchene / Becker's Muscular Dystrophy

Both DMD and BMD present with muscular weakness and muscular hypertrophy (pseudo hypertrophy). Duchene muscular dystrophy is present at birth but usually become apparent by the age of 3-5 years. Loss of power is progressive with predilection of proximal muscles and neck flexors .Leg more severely involved than arm. By the age of 12 years, they become wheelchair dependent.

Becker's muscular dystrophy is a milder form of DMD and closely resembles it. Proximal muscles are weak especially of lower extremities. Hypertrophies of calf muscles are prominent. Age of onset of weakness is between age of 5 and 15.

Limb Girdle Muscle Dystrophy

LGMD manifests as progressive weakness of pelvic and shoulder muscles. Onset ranges from late in 1st decade to 4th decade.

Fukuyama Congenital Muscle Dystrophy

Onset is at birth. Characterized by hypotonia, joint contractures generalized muscle weakness, hypertrophy of calf muscles, seizures and mental retardation. EMG shows myopathy pattern and MRI shows hydrocephalous and frontal hypo myelination.

Myotonia congenita is diagnosed by clinical evaluation, a detailed personal and family history, various specialized tests and genetic analysis if available.

Our patient's electromyography was obtained and demonstrates repetitive discharge of electrical impulses (action potentials) after forceful contractions (myotonic discharges). Muscle biopsy- reveals minimal abnormal changes (e.g. Muscle fiber enlargement in hypertrophied muscle) and myotonia was confirmed. ECG, ray chest, echocardiogram, ultrasound abdomen, complete ophthalmic checkup were done and were within normal limits. Patient was treated with mexiletine 200 mg once daily and phenobarbitone 30 mg once daily. Patient was followed up after 6 months of initiation of treatment, was having improvement of symptoms. The other family members were screened clinically and were normal.

DISCUSSION

Thomsen and Becker type's myotonia congenita are considered "ion channel diseases" or "channelopathies", occur due to the abnormalities in the flow of certain ions across muscle cell membranes. Symptoms typically begin in childhood and vary from person to person which includes muscle stiffness, muscle weakness and attacks of stiffness brought on by movement after rest. Becker disease is most common and severe, generalized form; while Thomsen disease is a rare and milder form. Becker type myotonia congenita is inherited as an autosomal recessive trait. Recessive genetic disorders occur when an individual inherits the same abnormal gene for the same trait from each parent[1,2] The risk for two carrier parents to both pass the defective gene and, therefore, have an affected child is 25% with each pregnancy. The risk to have a child who is a carrier like the parents is 50% with each pregnancy. The chance for a child to receive normal genes from both parents and be genetically normal for that particular trait is 25%.According to reports in the medical literature, parents of several individuals with Becker disease have been closely related by blood (consanguineous). With closely related parents, there may be an increased likelihood that both carry the same recessive disease gene, which increases the risk that their children may inherit the two genes essential for the development of the disease.[5,6,7] Treatment of Thomsen and Becker types myotonia congenita is directed toward the specific symptoms that are apparent in each individual, which may include include mexiletine, dantrolene or acetazolamide and carbamazepine. Early intervention is essential to ensure that affected children reach their potential. Special services that may be beneficial include special social support, physical therapy, and/or other medical, social, and/or vocational services. Genetic counseling will be of benefit for affected individuals and their families. Other treatment for this disorder is symptomatic and supportive.

CONCLUSION

Myotonia congenita is rare congenital disorder often remains undiagnosed or misdiagnosed. Proper clinical evaluation and investigations is needed for making the diagnosis .With initiation of medications adequate symptomatic improvement can be provided to the patient.

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