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A Rare Case of Becker Disease in a 7 Year Old Boy

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ABSTRACT

Becker Disease is an autosomal recessive version of the rare congenital disorder called Myotonia Congenita. Due to the rarity of Becker Disease, the genetic and pathological basis of this disease have not been studied well and possible diagnostic methods and techniques are yet to be explored. The existing method of diagnosis of such a case is predominantly dependent on the clinical examination. More work and studies need to be done on the diagnostic aspect of this disease to discover newer diagnostic methods for this disease, which are more reliable and specific. In order to develop better diagnostic methods for the disease, more cases of Becker disease need to be reported, with details of clinical and structural abnormalities. Here, we present the case report of a seven year old boy who has a history and clinical findings suggestive of Becker disease. Identification of this and similar cases of Becker disease can help us better understand this disease and hopefully one day help us develop a treatment for it.

MeSH WORDS

Becker Disease, muscular hypertrophy, muscle spasms, muscle weakness.

INTRODUCTION

Myotonia Congenita is a rare genetic disease and it affects 1 in 100,000 individuals.[1] Its symptoms begin to appear in early childhood and are characterized by defects in the skeletal muscle membrane which leads to delayed muscle relaxation, stiffness and weakness of the muscles. It has two subtypes, an autosomal dominant type (Thomsen Disease) and an autosomal recessive type (Becker disease).[2] The symptoms are more severe in the recessive form of this disease than the dominant form, where the transient weakness and stiffness of the muscle is more severe especially after prolonged periods of rest.[3] These patient also suffer from painless muscle stiffness, which can be relieved by activity (a method known as the warm-up phenomenon).[4] Moreover, these individuals have muscular hypertrophy, specially in the limbs, which gives them an athletic look.[3] The diagnosis of Becker disease is dependent on clinical symptoms and signs like muscle stiffness, delayed muscle relaxation, difficulty swallowing and alleviation of stiffness after

brief exercise.[3] Electromyogram in these patients shows myotonic bursts.[3] Sequence coding CLCN1 gene can help identify 95% of cases.[3] The genetic mutations responsible for Becker disease have been identified to occur in CLCN1 gene, which encodes for a chloride channel in a normal muscle membrane.[3] This gene resides on chromosome 7q35.[5] It is responsible for the formation of voltage dependent chloride channels, which in turn control the movement of chloride ions into and out of cells.[5] This abnormality therefore leads to all the symptoms mentioned above. However, as one particular study suggests that there can be as many as 130 different mutations in the CLCN1 gene that are responsible for this condition, this diversity in mutations could be the reason why no two cases of Becker disease present in the exact same manner.[6] Hence it is essential to determine the most common mutations and then to investigate the association of those mutations to their particular clinical presentations. In order to achieve this goal more studies need to be conducted after receiving samples from new confirmed cases of Becker Disease.

CASE PRESENTATION

Here we present the case of a 7 year old boy who presented to Aga Khan University Hospital, Karachi Pakistan, in 2016, with complains of difficulty in walking and weakness of muscles, specifically in the lower limbs. His symptoms were first noticed by the father, who stated that the patient has had walking difficulties since he was 5 years old. On inquiring, it was reported that the patient suffered from episodes of muscle stiffness after prolonged episodes of rest and he was unable to walk in a straight line.

On examination, the patient appeared to be restless and weak. He had weakness of the proximal muscles of hips and thighs bilaterally. No muscle wasting, no hypertonia and no atonia was observed. However, the pathognomonic hypertrophy of calf muscle seen in patients of myotonia congenita was also observed in this patient. Eye examination revealed that he was also hyperopic.

Electromyography studies showed diffuse myotonia in all muscles examined in the upper and lower extremities. Evaluation of voluntary motor unit action potentials was hampered by the presence of profound myotonia. There was no definite evidence of myopathy. Blood tests showed that Vitamin D levels were 20.3ng/ml.

Based on patient history, clinical examination and electromyography test, the patient was diagnosed to be suffering from Becker disease. The parents were educated about this debilitating disease. Due to the non-availability of any significantly effective treatment, the patient was advised to get regular physiotherapy and prescribed Vitamin D supplements.

DISCUSSION

Myotonia Congenita, a musculoskeletal disorder, was first identified with seminal studies by Bryant, Lipicky and colleagues in 1960.^[6] It was in the 1990s, when the first CLCN1 gene mutations were discovered and along with it the autosomal dominant and recessive components of this disease were identified.^[7] CLCN1 gene in the body is responsible for normal production of proteins that which controls the chloride channels in the skeletal muscle fiber, which then assist in normal muscle contraction and relaxation.^[8] Mutations in this gene hence leads to alteration in the chloride channels, which in turn reduces the movement of ions across the membrane.^[9] It is believed that mutations in the CLCN1 gene are responsible for both autosomal recessive (Becker disease) and dominant versions (Thomsen disease) of Myotonia Congenita. In fact one study has identified as many as 130 mutations in the CLCN1 gene.^[6] Hence the precise mutations responsible for Becker and Thomsen diseases are yet to be discovered.

In order to treat this condition it is important to

determine the extent of this disease. A genetic counselor and neurologist should be taken on board to decide how progressed the disease is and what treatment regimen should be used.^[3] A few pharmacological treatments are being tested for Myotonia Congenita. A randomized double blinded trial showed that mexiletine reduced muscle stiffness in 59 patients with Myotonia and 34 patients out of these suffered from Myotonia Congenita.^[9] It has also been suggested that other compounds, such as quinine, dantrolene, acetazolamide, carbamezapine or phenytoin,^[10] can be tried to reduce the symptoms. However, looking at the symptoms of this condition anesthetic agents and depolarizing muscle relaxants should be used with care.^[3] One study also reported that beta-antagonist propranolol worsen the symptoms of Myotonia Congenita, hence it should be avoided.

We have reported the first case of Becker disease (an autosomal recessive form of Myotonia Congenita) from Pakistan. This is a rare disease that should be reported so that more research can be conducted on its genetic basis and for the discovery of its treatment.

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Prem Chand; concept, data collection, data analysis, manuscript writing, manuscript review
Rozmen Badruddin Hussain; data collection, data analysis, manuscript writing, manuscript review
Fazal M Arain; data analysis, manuscript writing, manuscript review