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A Rare case of Myotonia Congenita Subtype in Indian Population – a Case Report

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Abstract:
Congenital Myotonia is rare genetic disorder in which abnormality of normal voluntary skeletal muscle fibres causes an unusual exaggerated response to stimulation. Affected individuals have difficulty relaxing certain muscles after contracting them (myotonia), muscle stiffness (rigidity) and associated symptoms. Thomsen and Becker types myotonia congenita are thought to affect males and females in relatively equal numbers. The reported incidence of these disorders is 1 per 10,00,000 people in the general population. Thomsen disease which is the autosomal dominant type is more prevalent than the autosomal recessive Becker type. We present here a case report of a 15 yr old boy who came with the complaints of difficulty in walking which was gradually progressive. Clinical Examination were found to have findings typically associated with myotonia congenita. Complete Blood count, Liver and renal function tests were found to be normal. Serum Creatine Kinase was found to be elevated. Electromyography was done to confirm the diagnosis. Supportive treatment was given. Genetic counseling was given to the affected patient families and proven to be beneficial. Acknowledgement of this disease provides greater insight for physicians and providing early interventional therapies thereby reducing morbidity.

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I. Introduction

Thomsen and Becker types Myotonia congenita are thought to affect males and females in equal numbers. Thomsen disease was more common than Becker, the autosomal recessive one.

Thomsen type myotonia congenita was initially described in 1876 by a Danish physician (Thomsen J). The disease affected the physician himself as well as multiple family members in several generations. According to reports in the medical literature, Thomsen disease tends to occur over multiple generations, usually without skipping generations. However, some rare exceptions have been noted in which skipped generations have been shown. In individuals with Thomsen disease, the severity is more associated with males typically more affected than females.

The autosomal recessive form, designated Becker type myotonia congenita, was later described by another investigator (Becker PE). Becker type myotonia congenita has been reported in multiple siblings of unaffected parents. As noted above, in some of these cases, parents of children with Becker disease have been closely related by blood (consanguineous).

II. Case Report

A 15 year old boy came to the outpatient department with the chief complaints of difficulty in walking for the past 4 years. The difficulty in walking was insidious in onset and gradually progressive. The patient had difficulty in climbing stairs and difficulty in rising up from bed. There were no difficulty in standing, combing and raising hand, sensory symptoms, loss of consciousness, bowel and bladder disturbances, autonomic disturbances, dizziness and vertigo. Past history yields no significant hospitalization, surgeries and infections in the past. His Maternal uncle also had similar illness in the past. Antenatal, Natal and Postnatal history is

uneventful. Developmental Milestones were achieved according to his appropriate age and time. The patient was immunized according to NIP schedule.

Upon General examination, vitals were stable and signs such as pallor, icterus, cyanosis, clubbing, lymph adenopathy and pedal edema were not present. Neurological Examination was done. Motor examination was as follows. Muscle tone is found to be normal, power is decreased minimally and bulk is increased in all four limbs. Calf and Biceps muscle hypertrophy was significant. Deep tendon reflexes were decreased and plantar response showed withdrawal from stimulus. Waddling type of gait is seen in this patient. Gowers sign was Positive**. *Percussion Myotonia sign was positive***. Sensory examination, higher mental Functions and cerebellar Function tests were unremarkable. Other systems were normal.

Complete blood count:

Hemoglobin	14.0 g/dl
RBC	4.98 million/cu.mm
WBC	4800 cells/cu.mm
HCT	39.4%
MCV	79.1fL
MCHC	35.5 g/dl
Platelet count	298000/cu.mm

Renal function test

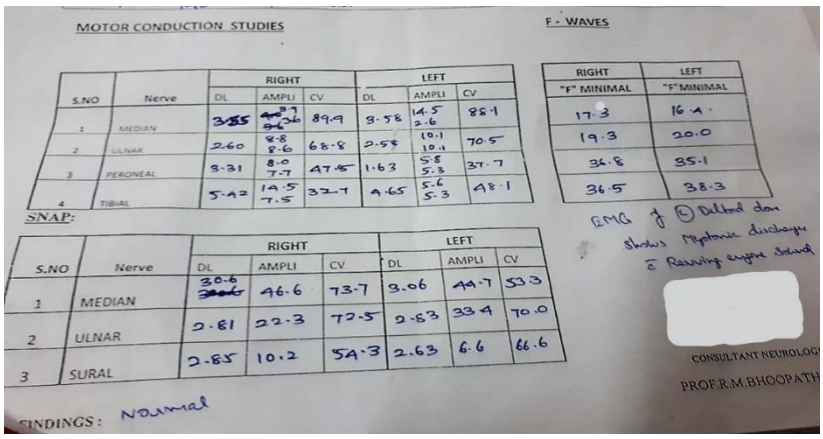
Random blood sugar	80 mg%
Urea	25 mg%
Creatinine	0.7 mg%
Sodium	139 meq/L
Potassium	3.9 meq/L

Liver function test

Serum bilirubin	0.6 mg/dl
SGOT	41 U/L
SGPT	20 U/L
Serum ALP	354 U/L
Total protein	7.0 g/dl
Albumin	4.2 g/dl
Globulin	2.8 g/dl
Creatine Kinase	254 U/L

Electromyography was done and found to have repetitive discharge of electrical impulses (action potentials) after forceful contraction. These are called revving engine myotonic discharges, characteristic of myotonia congenita.

Nerve conduction studies were found to be normal.



III. Discussion:

In most cases of Becker disease, the myotonia is initially apparent in muscles of the legs. With disease progression, arms, trunk and face are eventually affected. Enhancement of symptoms by cold exposure are common. Abnormal muscle hypertrophy is typically present individuals with Becker disease and this particularly striking, resulting in an even more pronounced body builder type appearance. Malignant Hyperthermia like cases are associated with Becker disease in some cases.

Thomsen and Becker diseases are generally considered to be "Channelopathies". They are linked to "CLCN1" gene located in chromosome 7q35. Researchers suspect that certain mutations of CLCN1 gene, which codes for voltage gated chloride channels in the sarcolemmal membrane resulting in reduced numbers or insufficient functioning of chloride channels causing an impaired ability to maintain normal muscle excitability and associated myotonia. Myotonia congenita may be diagnosed from infancy or early childhood to adulthood, based upon a thorough clinical evaluation, a detailed patient and family history, various specialized tests, and genetic analysis, if available. In patients with Myotonia congenita, clinical examination may reveal an inability to quickly release the hand grip, sustained muscle contraction after direct muscle percussion (Percussion myotonia), and other characteristic findings. In addition, specialized testing that records electrical activity in skeletal muscle at rest and during muscle contraction (electromyography) typically demonstrates a repetitive discharge of electrical impulses (action potentials) after forceful contraction (Revving Engine myotonic Discharges). EMG testing may reveal myotonic discharges in some (particularly male) carriers (subclinical myotonia).

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