A Family with Autosomal Recessive Generalised Myotonia with Herculean Appearance

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Abstract
A 28-year-old male had history of stiffness in limb muscles, with hypertrophy of most muscle groups and both action and percussion myotonia. We report a very interesting rare family of brothers and sister of myotonia congenita, conforming to autosomal recessive transmission (Becker’s variety) with Herculean appearance.

Introduction
Disorders of voltage-gated ion channels are responsible for a diverse group of neurological and muscular diseases. These diseases are now called “channelopathies.” The “channelopathies” are characterized by increased or decreased excitability of nerve or muscle. Disorders of ion channels can be toxin-mediated, immune-mediated or genetically determined.

Myotonia congenita is an uncommon hereditary disease of skeletal muscles that begins in early life, and is characterized by myotonia and muscular hypertrophy. Prevalence is between 0.2 to 7.3 per 100000 worldwide and incidence among Asians is not well established. Mutations in the skeletal muscle chloride channel (CLCN-1) on chromosome 7q35 cause both autosomal dominant (Thomsen’s variant) and autosomal recessive (Becker’s variant) myotonia congenita. Patients with myotonia congenita frequently appear “muscular” because of their prominent muscle hypertrophy and classically described as “Herculean features.” Myotonia manifests as generalized stiffness, especially after rest. Muscle wasting and weakness can occur late.

We report a family of ten brothers and one sister of myotonia congenita conforming to autosomal recessive transmission (Becker’s variety).

Case Report
A 28-year-old Muslim male, born of a consanguineous marriage, complained of tightness of whole body, difficulty in releasing firmly held object from hands and palm maintaining the posture of incomplete opening since childhood. History of falls was present whenever someone pulled or pushed him acutely. After few minutes of starting of work or exercise his difficulty became lesser in magnitude. His symptoms did not increase on exposure to cold. He had history of cramping in bilateral calf and thigh regions.

On examination, thighs, calf muscles and proximal muscles in the upper limbs were hypertrophied and there was evidence of grip, tongue and chest muscle myotonia. No other neurological deficit was present. Getting up from a sitting posture or climbing stairs was accomplished with difficulty and initiation of movement was slow which improved as he continued to walk. On examining his parents and eleven siblings, his seven brothers and one sister were found to be well built with hypertrophied thigh, upper limb and trunk muscles and there was evidence of grip and percussion myotonia (Figure 1). The parents had no neurological abnormality. The family history was suggestive of autosomal recessive pattern of inheritance.

Discussion
Our patient was diagnosed as autosomal recessive myotonia congenita based on the young age of onset, family history, muscular hypertrophy rather than dystrophy, characteristic EMG findings and genetic testing.

Myotonia congenita is a non-progressive, non-dystrophic myotonic disorder. Both the autosomal dominant (Thomsen’s disease) and the autosomal recessive (Becker’s disease) forms of myotonia congenita are linked to CLCN-1 gene on chromosome 7q35 encoding the skeletal muscle chloride channel. In Thomsen’s disease muscle strength is normal, but in Becker’s, which is usually more severe, there may be muscle weakness. Symptoms often begin in the childhood between the ages of 4 and 12 years, in form of muscle stiffness with difficulty in starting to run or walk briskly with progressive muscle hypertrophy causing “Herculean appearance.” In our patients, weakness used to be worsened by cold and improved by activity so that the patients could “work off” the slowness with continued exercise. Sun et al reported two siblings and a first degree cousin from a consanguineous marriage who demonstrated wasting of muscles and contractures instead of muscle hypertrophy. Our patients had no weakness or contractures. Most of the patients have eyelid myotonia which was not found in our patients.

Many patients will not require treatment and learn that the symptoms improve with activity, but some patients require drugs such as phenytoin, carbamazepine, acetazolamide,
Myotonia congenita being a non progressive disorder carries a good prognosis and subjects may live up to adult life. Care must be taken with the use of depolarizing muscle relaxants such as suxamethonium during preoperative anesthesia, because they can cause life-threatening muscle spasms and ventilation difficulties.

Till date very few cases are reported from India. The aim of this report is to make the readers aware of this entity which can be easily controlled with medication and prevented by genetic counseling. We also wish to emphasize the irony that some of the individuals with good muscular physique might be suffering from neurological disorder like Becker myotonia.

**Conclusion**

Myotonia congenita should be suspected in a child presenting with myotonia, generalized hypertrophy of skeletal muscles with a non-progressive course and characteristic electromyography findings. Genetic study is confirmatory. Other members of the family of an affected child should undergo detailed screening for diagnosis and genetic counseling. Phenytoin, carbamazepine, acetazolamid, quinine, mexiletine or tocainide may provide symptomatic relief.

**References**