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Case Report

A mechanical engineer cannot open his fist - myotonia congenita

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ABSTRACT

Myotonia congenita is a rare congenital neurological disorder, not encountered by many physicians in their clinical practice. Two types are described Thomsen and Becker. The former autosomal dominant, presents at early age and less aggressive as in our patient. Backer is autosomal recessive more aggressive and with muscle weakness. In these disorders group of skeletal muscles contract longer and slow to relax due to mutation in CLCN1 gene leading to chloride channelopathy. They present with difficulty to initiate an action but can do it normally after repeated attempts. Hand and facial muscles are involved in Thomsen disease; they are not weak or atrophic but might have cramps. Diagnosis of Myotonia congenita is clinical and confirmed by electromyography. Many other clinical conditions have myotonia, dystrophic and non-dystrophic. Physiotherapy and membrane stabilizing drugs like anti-epileptics are of choice but with varying success. Genetic counseling is an important part of management of these cases.

Keywords: Difficulty in opening fist, Myotonia Congenita, EEG, Carbamazepine

INTRODUCTION

Myotonia congenita is a rare congenital disease with incidence of 0.3 to 0.6 per 100,000 populations. Highest is in Scandinavia up to 10 per 100,000. Male and female are equally effected.[1] Myo from Greek means muscle and tonus from Latin means tension. Autosomal dominant form described by Thomsen and recessive form described by Becker, diseases was named after them. Lack of sufficient functioning chloride channels due to mutation in the gene CLCN1 causes muscle fibre membrane more excitable and electrically active for longer periods.[2] This causes prolonged contraction and delayed relaxation of muscle fibres affected. Similar disease occurs in goats & horses.

CASE REPORT

Twenty four year old male patient, a mechanical engineer was brought to our hospital with complaints of inability to stand on legs for a long time, inability to open the closed fist quickly (Figure 1) and inability to open the mouth at the beginning of eating (Figure 2). He can take morning walks with friends but he had to warm up before joining them. He can take long walks but the most difficult part is climbing to the first floor to reach his study room. He used to struggle to take first bite of sandwich, albeit could eat easily rest of it. His friends complained frequently that he is holding hand shake too long. He never played games in college for fear of fall. As a child he used to have frequent falls and that fear persisted throughout. From the age of three all these complaints started. Though opening of mouth and fist are difficult to start with, normalised with repeated attempts. In spite of these difficulties he led normal life and completed Engineering with honours.

His parents came from low socioeconomic class; they have been working as Agriculture labourers. Their marriage was not consanguineous. None of their family

members has similar disease. His paternal uncle son who was born by consanguinity has mental retardation and bedridden since birth.

Blood pressure, temperature, pulse and built were normal. Tendon reflexes were normal; there were neither muscular hypertrophy nor any neurological deficit. Laboratory investigations were all normal except for a low vitamin D. As the history and clinical examination were pointing towards myotonia congenita we did an EMG which has shown myotonic potentials (Figure 3).

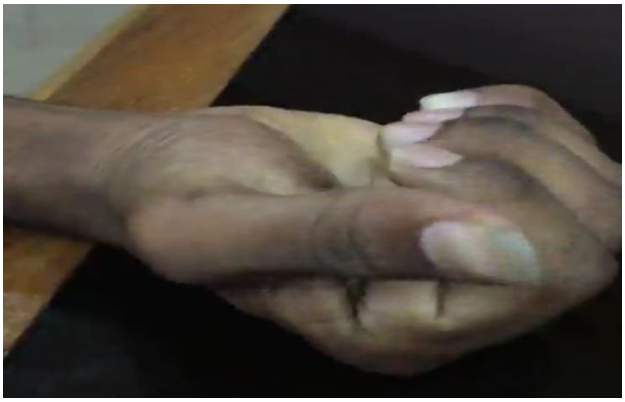

Figure 1: Difficulty to open the fist.

He was started on phenytoin sodium 300mg per day along with Vitamin D replacement. He was entrusted to physiotherapy. He is gradually improving.

Figure 2: Difficulty to open the mouth.

DISSCUSION

Our patient presented with symptomatic myotonia congenita from the age of three. The disease is non-progressive, non-dystrophic and without much muscle weakness pointing to autosomal dominant Thomsen disease.

He also suffered from loss of balance and used to be rigid after a fall as a child. Facial and upper limb muscle was predominantly involved. There is a warming effect. There is no muscle hypertrophy, atrophy or weakness. His intellectual functions are normal. All these feature points to Thomsen disease. Through autosomal dominant, there is no family history in this case; sporadic cases were also described instead of familial clustering. Myotonia doesn't cause pain and may cause cramps, this patient used to feel leg cramps on prolonged standing. Except for frequent falls he did not have early symptoms of childhood like difficulty in swallowing, gagging and difficulty in opening eyes after a cry (Von Graefe's sign).

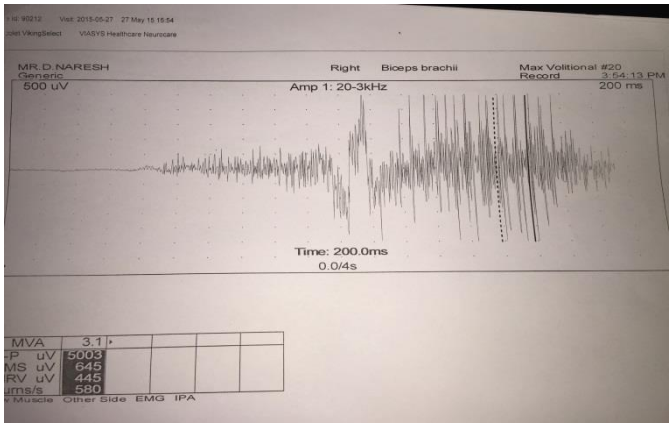

Figure 3: EEG showing myotonic potentials.

Myotonia is diagnosed by presence of classical clinical features and electromyography. EMG demonstrated a repetitive discharge of electrical impulses (action potentials) after a forceful contraction. Genetic studies will be contributory if available. Muscle biopsy changes are nonspecific useful to exclude other diseases. Myotonia has to be differentiated on clinical grounds from other myotonic disorders. These include sodium channel myotonia hyperkalaemia with periodic paralysis, potassium aggravated myotonia, Paramyotonia congenita. Potassium channel disorder like Andersen – Tawil syndrome (long QT) is also contender.[3]

Myotonic muscular dystrophies type I and type II can be excluded by clinical grounds and if required by muscle biopsy. Myotonia produced by thyroid disorder will require thyroid function tests to be differentiated. Certain drugs like statin, fibrates, chloroquine and cyclosporine are also implicated in myotonia and careful drug history is paramount importance.

Treatment of Thomsen and Becker myotonia is symptomatic. Some people may not require treatment as risk of the medications outweigh the benefits.[4] Physical therapy and proper exercise plan may alleviate the symptoms of myotonia. Extra caution may be required when administrating the anaesthesia as they may produce prolonged paralysis or hyperthermia sometimes.[5]

Drugs that are used to reduce muscle stiffness and other symptoms of myotonia are membrane stabilizers like anticonvulsants and muscle relaxants. Phenytoin, acetazolamide, mexelestine, carbamazepine, quinine

sulphate and dantroline were all tried. There were reports of success with antihistamine trimeprezine. There are favourable reports with use of carbamezepine[6] and mexiletine.[7]

CONCLUSION

Myotonia congenita is rare disease but we may come across one in our clinical practice. Prior knowledge of the disease helps to spot it with ease. Diagnosis is clinical, Electromyography is confirmatory. Drugs may be beneficial in some individuals, physiotherapy and exercise schedule are useful for all patients. Genetic counselling is essential part of management of these congenital diseases.

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