CASE REPORT

MYOTONIA CONGENITA

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ABSTRACT

Myotonia congenita is a rare genetic disease that affects chloride channels and cause transient stiffness in the muscles. It has two sub types according to the genetic phenotyping.

1. Thomson disease
2. Becker's disease

Becker's disease is an autosomal recessive sub type that affects more commonly leg muscles and the onset of symptoms manifest slightly in older age. We are reporting a case of Becker disease in 16yrs old boy having delayed relaxation of muscles noticing his symptoms for the last 8 years.

Key words: *Myotonia congenita, Autosomal recessive, Thomson disease, Becker disease.*

Introduction

Myotonia congenita is a genetic channelopathy that affects limbs and facial muscles. The defect is actually in the chloride channels. The hallmark of the disease is the delayed relaxation of muscle after voluntary contraction and relieved by repeated contraction "Warm up effect".[1,2] It has two sub types:

1. Autosomal dominant Thomson disease.
2. Autosomal recessive Becker's disease

These two subtypes are similar except transient weakness occur in Becker's disease.[1] In Becker disease more commonly limbs are involved mostly leg muscles, prone the patient to fall and the onset is slightly in older age. Here we report a case of Becker disease in a 16yrs old male who came with delayed relaxation of muscles of hands and peri ocular muscles which improved with repeated contraction.

Case Report

A 16 years old boy, farmer by occupation, well built admitted to our Hospital on 2/01/2014 with complaints of delayed relaxation of muscles after voluntary contraction particularly hands i.e. could not extend the fingers after shaking hand for 25 - 30 seconds (Transient weakness in muscles) and face e.g. eye lids Orbicularis oculi i.e. could not open eyes after forceful closing of eyes (Von Grafe sign) which he had noticed for the last 8yrs, also had difficulty in initiating movement more pronounced in the morning after prolong rest at night and also after a slight rest even for a few minutes. There was no other compliant e.g. Cataract, abnormal cardiac conduction (arrhythmias) and endocrine dysfunction except these transient weakness and delayed relaxation of muscles. General physical examination was normal except muscular hypertrophy probably because of his laborious work. All the other systems were normal. All the laboratory investigations e.g. Blood CP, RFTs, Serum electrolytes, Serum cretinine kinase ECHO Cardiograph were normal. There was no history of taking any medications for this problem or any other disease. History of same sort of problems present in paternal cousin as well as his elder sister.

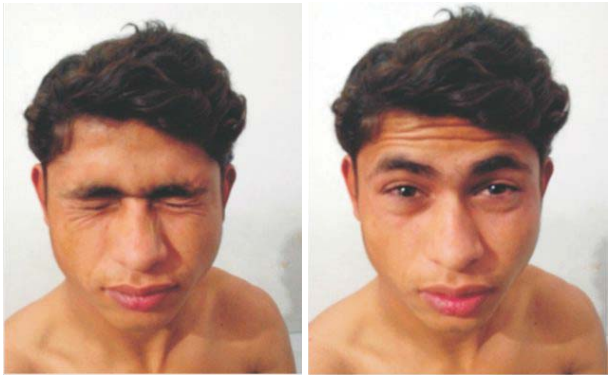

Fig 1 & 2: Von Grafe sign (Eye opening after forceful closing)

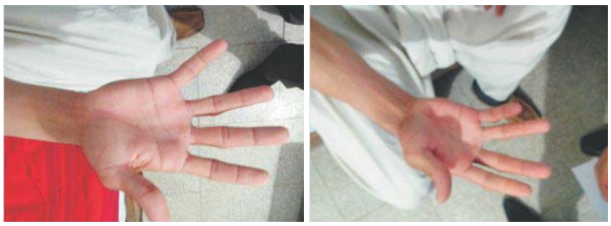

Fig 3 & 4: Delayed relaxation of Hand small muscles after contraction

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Discussion

Myotonia congenita is a genetic channelopathy that affects muscle. Finnish heritage disease, common in ethnic Finns in Finland, northern Scandinavia.[3,4] The prevalence is 1: 10,000 and affects 1: 100,000 worldwide. The hallmark of the disease is the delayed relaxation of muscle after voluntary contraction and relieved by repeated contraction "Warm up effect".[5] The muscles are usually hypertrophic.[6] The disease is caused by mutation in part of gene CLCN 1 encoding CLC 1 Chloride channel resulting in muscle fiber membrane causing an unusually exaggerated response to stimulation (Hyperexcitability).[1,7] The diagnosis is usually made clinically and relevant family history laboratory investigations are almost always unremarkable and not help in making diagnosis. Although in some patients Serum Cretinine Kinase is elevated.[6] Two types of Myotonia Congenita exist Autosomal dominant (THOMSON DISEASE) that affect more commonly hands small muscles, face and eye lids and onset of symptoms is usually in infancy and early childhood. The other one is Autosomal recessive (BEKER DISEASE) affecting more commonly leg muscles making the patient to fall with onset at slightly older age.[1,7,8] The disease is aggrevated by some factors e.g. inactivity, cold weather, pregnancy. Depolarizing muscle relaxants e.g. Suxametonium,[9] Adrenaline and Diuretics can worsen symptoms .

All the clinical signs were present in our patient e.g. Delayed relaxation of hand muscles (cannot extend the fingers after shaking hand. Difficulty in eye opening after strenuous contraction (von Grafe sign). Stiff movements that improves when they are repeated (Warm up effect).[5] Although there are no complications present e.g. Aspiration Pneumonia, frequent choking, chronic joint problems and injuries due to fall. As the disease is Genetic and at present is not causing hindrance in his routine work and patient has not developed any of the complications mentioned above. Since the complications with treatment over weigh the benefits. So the patient was discharged after diagnosis with the advice not to take bath in deep water as it can endanger his life. The patient was warned against taking Depolarizing muscle relaxants e.g. Suxamethonium,[9] Adrenaline, Diuretics, Beta agonists and Beta antagonists with caution.

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