Case Report

Myotonia Congenita – Shortage of Chloride Channels

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ABSTRACT

Autosomal recessive myotonia congenita (Becker diseases) caused by mutations in the CLCN1 gene. It is characterized by muscle stiffness during sustained muscle contraction and variable degree of muscle weakness that tends to improve with repeated contractions. In this study we discussed about the clinical features and pathophysiology of different myotonic disorders, treatment availability for diseases. This is rare and genetical disorder, this is also known as chloride chanallopathies.

Keywords: CLCN1 gene, chloride channel, dystrophy, myotonia congenita.

INTRODUCTION

Myo: Muscle, Tonia: Tension Congenital: defect by birth. Actually it is a rare and genetically heterogeneous syndrome. In clinical myotonia is failure of muscle relaxation after contraction of muscle[1]. Electrical myotonia is spontaneous discharge of muscle fibres which increase or decrease in both frequency and amplitude on electromyography (EMG)[2]. Drugs like cholesterol lowering agents, cyclosporine, and others, in pompe disease inflammatory myopathies leads to electrical myotonia[1]. Clinical myotonia symptoms like painless muscle stiffness and some are with pain[2]. The location of stiffness depends upon the disorder but mainly seen in parts of eyelids, mouth, hands, and proximal legs[3-5], cold, stress, and exercise, and symptoms worse during pregnancy and menstruation[3]. Mytonia can be revealed by fallowing method, by asking the patient to grip and relax their arm and blinking of their eyes[6].

Myotonia can be classified into dystrophic or non dystrophic. it is characterized by fixed weakness, dystrophic changes on muscle biopsy, and fixed weakness and dystrophic changes are less common in non dystrophic myotionias (NDM)[7]. Recent studies reveal that structural muscle changes on magnetic resonance imaging and ultrasound imaging of some patient[8-9].

CASE REPORT

The subject name was xxx 6yrs old boy, the subject joined in the hospital with these clinical futures like difficulty in getting up from sitting position, holding of object is hard with both hands, muscle stiffness, hyperhydrosis were observed in patient.

Medical examination results in goiter (+), deep tendon reflex, hypertrophied muscle.

Lab investigation

Reviewed that serum electrolytes were normal, serum creatine phosphokinase was observed to be 184 U/L

Treatment

Acetazolimide (it is a carbonic and hydrase inhibitor, it inhibit accumulation of carbonic acid.) carbamazepine (It is a sodium channel blocker, it binds preferentially to voltage –gated sodium channels in their inactive conformation, which prevents repetitive and sustained firing of an action potential) phenytoin (involve in voltage –dependent blockade of membrane sodium channels resulting in a reduction in sustained high –frequency neuronal discharges.) gabapentin (it was designed to mimic the neurotransmitter GABA it doesn't, however, bind to GABA receptors it involves in inhibition of alpha2-delta subunit of voltage- gated calcium channels)

Genetic Counselling

Myotonia congentia may be autosonal recessive (Beckers diseases) or an autosomal dominant (Thomsen diseases). In the individual with autosomal dominant have50% chance of inheriting the pathogenic variant. Autosomal recessive is 25% chance of being affected, a 50% of chance of being an asymptomatic carrier, and nearly 25% of subject is being unaffected and not a carrier. The molecular genetic testing reveals two pathogenic variants in CLCN1. Molecular genetic testing is done in the case of high rate of inheritance in the family.

DISCUSSION

MYOTONIC CONGENITA (MC)

It is also known as chloride chanalopathy, typically dived into two types

1) Thomsen diseases
2) Becker diseases

Over 150 mutations in the CLCN1 gene have been reported[10].

In 1876 Thomsen was the first person who affected with the Thomsen diseases, he described his own disability[11].In first decade it is mainly seen in legs more than arms, hands and face[3,4,12]. Disease severity is widely in family but penetrance is incomplete[13]. Worsen pain observed after rest and removal of is seen after slight movements. Myotonia can also affect muscle of mastication and swallowing[3,4]. Myotonia is more pronounced in hands than eyelids and percussion myotonia[3,4].

Beckers is usually seen in between the age of 4 and 12 years, and onset in adulthood is also seen [11,12]. Symptoms are similar to the Thomsen diseases but myotonia tends appear more in lower limbs and proximal muscle and men are more affected than women[12]. It last for seconds to minutes, is sometimes seen after sustained bouts of myotonia and patients often have difficulty moving if startled suddenly[4,11]. It is similar to Thomsen disease , no symptomatic features, and lifespan is normal.

Laboratory investigation are normal in both in dominant and recessive MC anyway mild elevation is ck can be seen[11].electromyography needle (EMG) shows wide spread of myotonic discharges and myotonic motor units can be seen in weak muscles if it is not masked by myotonia. Genetic testing and muscle biopsy are rarely done due commercial availability [11].

CONCLUSION

The myotonia disorder is a heterogeneous group of diseases that result in clinical and electrical myotonia. In this case report it is an autosomal recessive myotonia congenita caused by a heterogeneous mutation in the in genotype of CLCN1 gene.

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