VOLUME - 10, ISSUE - 04, APRIL - 2021 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

Neurology

MYOTONIC DYSTROPHY TYPE1 WITH SERO NEGATIVE MYASTHENIA IN THE SAME PATIENT

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ABSTRACT Myotonic Dystrophy type1 (DM1) is the most common adult muscular dystrophy with autosomal dominant pattern of inheritance, characterized by muscle atrophy, distal weakness, myotonia and dystrophic changes in non-muscular tissues[1]. The coexistence of Myasthenia Gravis and Myotonic Dystrophy in the same patient is rare. The literature review showed only a few cases; probably mildly affected individuals escape detection. We present a 46yrs old women with Myotonic dystrophy features with fatiguable weakness and went into respiratory failure following acute respiratory illness and suspected myasthenia due to mild diurnal variation of symptoms and was given anticholinesterase and the patient recovered from respiratory failure and she was further investigated for myasthenia. Her serology was negative for myasthenia and also CT chest shows no thymoma but was clear positive symptoms in neostigmine test and decremental response in repetitive nerve stimulation test (RNST).The patient improved remarkably after treatment with anticholine esterase and steroids.

KEYWORDS : Myotonic dystrophy type1, Myasthenia gravis, Thymoma, RA(Rheumatoid arthritis),Respiratory failure.

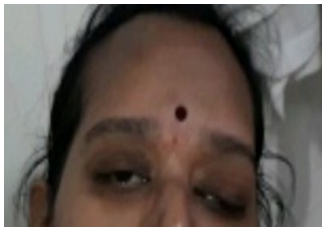
INTRODUCTION:

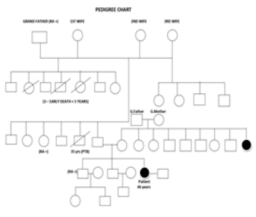
Type1 Myotonic dystrophy is distinguished by an autosomal dominant pattern of inheritance with a high level of penetrance, typically have characteristic facies, frontal baldness, temporal wasting, ptosis, facial distinctive appearance known as "hatchet face"[1,2]. Myotonia is perhaps early feature in this disorder. Other features include gonadal atrophy, infertility, cataracts, cardiac arrhythmias and type2 diabetes mellitus. This disease caused by an increase number of CTG, trinucleotide repeats of > 50 in the myotonin protein kinase(DMPK) gene on chromosome 19q[1].Myasthenia Gravis (MG) is an autoimmune disease characterised by fluctuating muscle weakness and fatiguability of voluntary muscles due to post synaptic impairment of neuromuscular transmission[3]. The simultaneous occurrence of both DM1 and MG is rare. We present a patient with Myotonic dystrophy proven with genetic testing with sero negative myasthenia without thymoma responded with anticholinesterase and steroids.

CASE REPORT:

A 46 –year-old, female with primary infertility, initially presented with immense body pains following viral fever in 2007(when patient was 33yrs old),following which she had progressive distal weakness of both lower limbs and later in 6 months she had distal weakness of both upper limbs and also associated with difficulty in getting up from squatting position, later she found to be having severe vitD deficiency which was treated, her pains subsided but weakness was progressing ,associated with wasting and she had frontal baldness, temporalis wasting&hirsutism.She noticed difficulty in opening hand grip after making tight fist.Percussion myotonia of thenar and tongue muscles was noted. Then she was suspected to have myotonia and was further evaluated and EMG study demonstrated diffuse runs of myotonic discharges, short duration motor unit action potentials and early recruitment consistence with myotonic dystrophy and was confirmed after 3yrs of onset of symptoms. Later she developed diabetes type2, ptosis, ophthalmoplegia, neck flexor wasting, distal limbs weakness and wasting, grip myotonia, bilateral foot drop and waddling gait. Serum creatine kinase was within normal limits. Cardiac and endocrine evaluation disclosed no abnormalities. No cataracts were detected upon slit lamp examination. After 10 yrs she developed sudden onset of respiratory illness and became unconscious and was treated for 3wks in our ICU care and improved with antibiotics and bronchodilators but within a day after improvement she became again breathlessness and had arrhythmia SVT which was reverted with injections and later she was discharged with o2 concentrator and bipap support. She was on bipap support complete day with increasing generalized weakness for which she was admitted again in a year and suspected myasthenia because of fatiguable weakness and dysarthria and was tested for serum AchR ab's ,which was negative, and was given anticholine esterase , clinical improvement in voice and weakness was seen and genetic testing was done and showed myotonic dystrophy type 1 gene (DMPK)positive. Now patient is on bipap only in night time and was regular follow up and since 1yr patient do not have excursions of weakness and breathlessness.

Figure 1. The Patient Has Bilateral Ptosis With Fatiguable Weakness





DISCUSSION:

Myotonic dystrophy is considered as the most common of the muscular dystrophies in the general population, the possibility of its coexistence in the same patient with myasthenia gravis estimated at 1 in 40 million [1] .Although both diseases have specific clinical and laboratory features, they share some similarities which generate difficulty in diagnosis of both entities in the same patient. Schoen et al[4]. Reported a 13yrs girl with DM1 and MG who had previously been diagnosed with MG after a positive tensilon test and electro diagnostic testing. She also had myotonic discharges on EMG in addition to bulbar muscle wasting and myotonia

on exam. Benito-Leo´n[1] and colleagues also reported a 61-year old woman with fatiguable weakness who was found to have a decremental response on repetitive stimulation, an elevated AChR antibody level, and myotonic discharges on EMG,350 CTG repeats in the DMPK locus[5].Myotonic dystrophy type 1 has highly variable clinical manifestations. Patients may be asymptomatic, have minimal features (such as cataract or asymptomatic myotonia), show moderate or severe facial and distal limb muscle wasting and weakness, or have a severe congenital disorder[6]. It is due to an unstable trinucleotide repeat expansion containing cytosine-thymidine-guanosine [CTG]n, located in the region of chromosome 19q13.3 [7,8].Myasthenia gravis is an autoimmune disease characterised by fluctuating weakness and fatiguability of voluntary muscles due to post synaptic impairment of neuromuscular transmission. Acetylcholine receptor binding antibodies are the first choice of assay for confirming a diagnosis of myasthenia gravis [7,8]. Although both diseases have specific clinical and laboratory presentations that permit appropriate diagnosis individually, they present with some common features that can make differentiation of the two diseases very difficult[9,10]. Both can present with weakness with predominantly facial and hand/wrist involvement. Ptosis is more marked in myasthenia gravis, and has the unique characteristic of hourly fluctuations[11,12].

Our patient with myotonic dystrophy proven with genetic testing had frequent admissions with respiratory failure and when clearly examined had mild diurnal variation of symptoms and was evaluated for myasthenia and proved with sero negative myasthenia without thymoma responded with anticholinesterases and steroids. After daily anticholine esterase therapy patient improved clinically and was regular follow up .Now since 1yr patient don't have excursions of weakness and breathlessness .This patient demonstrates that even very rare diseases can occur in the same patient, sharing similar signs and symptoms, so even in the presence of a straight forward diagnosis a complete search for other conditions is necessary with a thorough follow-up.

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