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A RARE CASE OF MYASTHENIA GRAVIS WITH COEXISTING MUSCULAR DYSTROPHY

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ABSTRACT

Myasthenia gravis (MG) is an autoimmune disease in which antibodies are directed against postsynaptic membrane of neuromuscular junction, resulting in muscle weakness and fatigability. We report a rare case of an 11 years old boy who was a known case of myasthenia gravis presented with progressive weakness and wasting of facial and limb musculature and was found to have coexisting muscular dystrophy most like facioscapulohumeral muscular dystrophy (FSHD).

Key Words: Myasthenia gravis(MG), muscular dystrophy, facioscapulohumeral dystrophy (FSHD).

INTRODUCTION

Myasthenia gravis is a chronic disease characterized by rapid fatigability of striated muscle. The most common cause is an immune mediated neuromuscular blockade. In its juvenile form ptosis and some degree of extraocular muscle weakness are the earliest and most constant signs. Rapid fatigue of muscle is characteristic feature of myasthenia gravis that distinguish it from other neuromuscular disorders⁽¹⁾. Previous authors have observed rare coexistence of myasthenia gravis with different types of Muscular dystrophy including myotonic dystrophy⁽²⁾, limb girdle muscular dystrophy⁽³⁾ and familial facioscapulohumeral muscular dystrophy (FSHD)^(4, 5, 6). However most of these cases are seen in the adult population.

CASE SUMMARY

An 11 years old boy known case of myasthenia gravis was admitted in pediatric intensive care Unit (PICU) with complain of progressive weakness and wasting of face and limbs since last 4 months and respiratory distress since last 4 days. He was diagnosed as case of myasthenia gravis 2 years back when he presented with bilateral ptosis, loss of neck holding, and ophthalmoplegia. His EMG done at that time was normal but NCS showed a decremental response on repeated nerve stimulation. He was put on anticholinesterase inhibitors, initially for few months he showed some improvement but later on his weakness progressively increases to involve all four limb but more marked in upper limb. On examination he was conscious, oriented but emaciated child with obvious facial wasting and bilateral winging of

scapula. He was having bilateral ptosis and ophthalmoplegia. He was vitally stable child with GCS of 15/15. He had reduced tone, diminished reflexes and power of 2/5 in all four limbs with non specific babinski bilaterally. Her chest had right sided crepitation with wheeze. Rest of examination was unremarkable. He was admitted to PICU because he developed type 2 respiratory failure due to pneumonia and was put on non-invasive mechanical ventilation. He was given anticholinesterase inhibitors and IVIG to treat myasthenic crises but patient showed no improvement. All his routine examination including CBC, renal profile. Liver function test, serum electrolytes, CRP, blood and urine culture were normal. CXR shows right sided infiltrates. His CPK level was found to be 160 u/l and LDH 1044 u/l. Tensilon test was refused by the parents. His muscle biopsy was done under local anesthesia from right deltoid muscle which showed variable sized muscle fibers with internalization of nuclei, increase connective tissue infiltration and degeneration of muscle fibers suggesting muscular dystrophy. The patient progressive weakness was due to this muscular dystrophy that's why it's not responding to any anticholinesterase inhibitors or IVIG. However by intensive care, physiotherapy, non invasive ventilation and antibiotics patient pneumonia settled and he was off ventilator on 20th day of admission. He was discharged with power of 4/5 in lower limb and 3/5 in upper limb (wheel chair dependant). At discharge he was given anticholinesterase inhibitors, multivitamin and advised physiotherapy. His parents were counseled about the disease and advised proper follow up but they are non compliant to follow up. So on basis of clinical features, EMG showing decremental response with muscular dystrophy finding on muscle biopsy he was

diagnosed as a case of myasthenia gravis with coexisting muscular dystrophy most likely facioscapulohumeral dystrophy.

DISCUSSION

Juvenile Myasthenia gravis is a rare autoimmune disorder of childhood which is characterized by the neuromuscular blockade leading to rapid fatigability of striated muscle. It is one of the few neuromuscular disease in which EMG is diagnostic. A decremental response is seen on repetitive nerve stimulation. Therapeutic options includes cholinesterase inhibiting drugs, steroids, thymectomy. In resistant cases plasmapheresis, IV immunoglobulins and rituximab (monoclonal antibody to B cell CD20 antigen) may be helpful⁽¹⁾. Fascioscapulohumeral dystrophy is a third most common inherited muscular dystrophy presented with slowly progressive weakness and wasting of facial, shoulder girdle and sometimes limb musculature⁽⁷⁾. Bulbar muscles are typically spared in FSHD. The clinical severity is wide ranging from asymptomatic to wheel chair dependant. Serum CK and other enzymes vary greatly ranging from normal to elevation in several thousands. EMG reveal non specific myopathic changes. Diagnostic molecular testing in idvisual cases and within families is indicated for prediction. Muscle biopsy distinguish it from other neuromuscular disorders. No effective pharmacological treatment is available yet⁽⁸⁾. The coexistence of MG and FSHD has also been described previously. Asadollahi et al described coexistence of FSHD and MG in a 70 years old man^(4, 5, 6). There was another report by Mcconigal et al describing occurrence of MG in a 56 years old known patient of FSHD⁽⁵⁾. Sansone et al reported 69 yaers old known case of FSHD presented with sudden deterioration of muscle weakness with bulbar involvement and was found to have concurrent MG⁽⁶⁾. Our case is a rare coexistence of FSHD with MG as no single report had been observed in pediatric population yet. Theoretically it is related to breaking of immune tolerance to acetylcholine receptors as a result of muscle fibre degenera-

tion⁽⁹⁾. Other theory could be role of immune mechanism in pathogenesis of FSHD but these patients donot benefit from prednisolone treatment⁽¹⁰⁾.

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Author's Contribution:

Dr. Sommayya Aftab: Study concept and design, data collection, data analysis, manuscript writing, manuscript review

Dr. M Haroon Hamid: Data collection, data analysis, manuscript writing, manuscript review

Dr. Tipu Sultan: Study concept and design, data analysis, manuscript writing, manuscript review