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Case Report

Paediatrics Section

A Great Masquerader: Acute Respiratory Failure as the Only Initial Presentation for Juvenile Myasthenia Gravis

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ABSTRACT

Myasthenia Gravis (MG) is most commonly caused by antibodies to acetylcholine receptor (AChRs). MG can present as respiratory failure during the late course of the disease, known as a myasthenia crisis. Mostly, this crisis develops in adult patients and associated with some intercurrent illness like infections. Sometimes, MG can present isolated respiratory failure as an initial presentation of disease, which was a rare presentation. This presentation will create diagnostic dilemmas among treating clinicians. Here, the present authors report a case of about an eight-year-old female with juvenile MG. She presented with isolated respiratory failure as her presenting manifestation. The child was diagnosed as MG based on history and electro-diagnostic tests. Child was recovered with supportive therapy, steroids and pyridostigmine. So, it is important to consider MG in cases of unexplained respiratory failure.

Keywords: Electro-diagnostic test, History, Neuromuscular junction

CASE REPORT

An eight-year-old female born of second-order to non-consanguineous parents, with uneventful perinatal history and normal development for age, presented to Emergency Room (ER) with severe respiratory distress, since morning. Further questioning did not reveal any probable exposure to pesticides or other toxin exposure. She denied symptoms of extremities weakness, diplopia and swallowing difficulties. She had similar complaints one week back and was treated with mechanical ventilation for four days and Intravenous (IV) dexamethasone. At that time, Magnetic Resonance Imaging (MRI) brain with spine and Cerebro Spinal Fluid (CSF) analysis found no abnormality. CSF analysis was done on day 2 of admission.

On examination, the child was alert, conscious and had tachypnea, tachycardia, SpO_2 88% at room air, normal blood pressure and paradoxical breathing, no ptosis, no motor weakness and normal deep tendon reflexes. In ER, child was stabilised with emergency intubation and mechanical ventilation. Arterial blood gas analysis revealed mild metabolic acidosis, with normal anion gap.

Routine investigation comprising of complete blood count, urinalysis, serum electrolytes, liver and renal functions were performed and found to be under normal limits. Chest radiograph showed flat diaphragm without any parenchymal changes. Hence, it was suspected that this respiratory failure may be due to neuromuscular disorder.

Nerve Conduction Study (NCS) with Repetitive Nerve Stimulation (RNS) showed significant decremental response in facial muscle. The child was treated with IV Methylprednisolone (30 mg/kg/day) and nasogastric Pyridostigmine (0.5 mg/kg/dose, 6th hourly). She gradually improved and was extubated on fourth day of admission. Anti-Cholinesterase Receptor (AChR) antibodies were not detected. She was discharged one week later, with oral Prednisolone (1mg/kg/day) and Pyridostigmine (0.5 mg/kg/dose, 6th hourly).

On follow-up, the child developed steroid side effects, hence tapering of steroids was started. So, prednisolone dose (0.5 mg/kg/day) was decreased and azathioprine (1 mg/kg/day once a day) was initiated. Based on the electrophysiological evidence and improvement with immunotherapy, the diagnosis of Juvenile MG presenting as isolated respiratory failure was confirmed. Since last one year on follow-up, the child had no recurrence of symptoms.

DISCUSSION

Myasthenia gravis is an autoimmune disease of the nervous system that causes abnormal muscular weakness [1]. The disease onset occurs in childhood or adolescence in approximately 11%-24% of the cases; though it arises at any age, peaking in the third and sixth decades [2]. Juvenile MG is one of the subtypes of paediatric MG [2]. These patients can present only with ocular symptoms or generalised weakness with or without dysphagia and bulbar weakness. Usually, MG was diagnosed based upon history along with electrodiagnostic tests, serological tests and treated with anticholinesterase inhibitors and immunotherapy [3,4].

Most commonly, JMG presents with ptosis associated with ophthalmoplagia, strabismus; the symptoms usually progress to include the limb muscles [1,2]. Myasthenic crisis occurs due to respiratory failure which results from diaphragmatic or intercostal muscle weakness. This respiratory failure can manifest during late course of disease or association with intercurrent illness like infections in about 3 to 8% of cases, known as a myasthenia crisis [5]. Gracey DR et al., reported profile of 288 myasthenia patients over a period of 2 years duration. In their series, 22 (7.6%) patients developed respiratory failure needing mechanical ventilation [6]. However, in the present case, isolated respiratory failure as the initial presenting symptom is very unusual presentation.

In literature, some cases of MG with respiratory failure as an initial manifestation have been reported [7-10]. Dushay KM et al., reported a case with similar history in adult patient and the patient was recovered with pyridostigmine and immunotherapy [7]. In a study on adult patients, respiratory failure as an initial symptom was observed in 14 to 18% of the patients [8]. In literature, paediatric case reports were rare. However, this patient showed no other neurological symptoms associated with MG like ptosis, dysphagia. There was no evidence of respiratory cause. There were no parenchymal or pleural changes on chest X-ray except flat diaphragm. The present clinicians were unable to find any cardiac causes and echocardiogram was normal. Drugs or trauma was ruled out by history. Hence, neuromuscular disease was suspected, especially Guillain-Barre Syndrome (GBS) and MG [9]. During previous admission, CSF analysis was done on day 2 of symptoms, which was found to be normal. Also, she was completely normal for 2 days after discharge, hence GBS was

ruled out. Electrophysiological diagnostic studies showed evidence of MG; also AChR antibodies were not detected.

To diagnose MG, the combination of a thorough history, repeated physical examination, and electrophysiological investigations as well as antibody samples are helpful clues [4]. MuSK antibodies positive MG is rare in children and associated with more severe disease. It mostly affects facial and bulbar weakness and frequently causes respiratory crises. In MuSK positive MG, females are predominantly affected and most paediatric cases have been reported within adult series [11,12]. The present case can be a possibility of Anti MuSK antibody positive MG. AntiMuSK antibody titres could not be performed due to financial constraints and there was already sufficient evidence to diagnose the child of MG.

Most patients with juvenile MG that require maintenance therapy are treated with anticholinesterase agents with or without a variety of immunosuppressive medications like prednisolone and azathioprine. Pyridostigmine is recommended as an initial intervention. Plasmapheresis and IV gamma globulin (IVIG) are generally reserved for more refractory patients or for those in myasthenic crisis [13,14]. In present case, the child improved with immunotherapy and Pyridostigmine. However, she developed side effects to oral steroid. So, Azathioprine was started as maintenance therapy along with Pyridostigmine.

CONCLUSION(S)

The present case report revealed that acute respiratory failure can be an initial presentation of MG for a patient in crisis. Even though antibodies were not determined, an electrodiagnostic studies confirmed the diagnosis of MG. The diagnosis of MG was confirmed seeing the clinical response of the patient to Corticosteroids and Pyridostigmine. This case suggests that one should consider MG in a case of unexplained respiratory failure.

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