Case Report

Facial diplegia, progressive bulbar weakness and tongue fasciculation without ocular involvement in myasthenia gravis: A case report

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ABSTRACT

Background: Juvenile Myasthenia Gravis (MG) is an immune-mediated disorder caused by antibodies against the post-synaptic membrane at the neuromuscular junction, typically presenting with fatigable weakness and ptosis. Although the clinical phenotypes of juvenile myasthenia gravis are similar to those in adults, several factors impact its management, including a broader range of differential diagnoses, atypical features, and higher rates of disease remission.

Case Report: We present a case of an adolescent boy with myasthenia gravis who experienced progressive bulbar weakness, facial diplegia, and tongue fasciculations without ocular involvement or significant limb issues, posing a diagnostic challenge. Due to bulbar involvement and aspiration pneumonia requiring hospitalization, a notable diurnal variation in secretion severity between morning and evening suggested a diagnosis of MG.

Conclusion: Recognition of myasthenia gravis with atypical features is crucial in preventing diagnostic delays and avoiding progressive disability and complications, such as myasthenic crises.

Keywords: Bulbar Palsy, Facial Diplegia, Juvenile Myasthenia Gravis, Tongue Fasciculation

INTRODUCTION

Juvenile Myasthenia Gravis (MG) is an immune-mediated disorder caused by antibodies against the post-synaptic membrane at the neuromuscular junction. The most common antibodies found are acetylcholine receptor antibodies (AChR), followed by muscle-specific kinase antibodies (MuSK). It typically manifests as muscle weakness that worsens with exertion [1]. The types can be ocular or generalized, depending on the involvement of muscles [2]. In over 50% of cases, the initial symptoms and signs are associated with weakness in the extraocular muscles, presenting as diplopia or ptosis. Although the clinical phenotypes of juvenile myasthenia gravis are like those in adults, several factors impact its management, including a broader range of differential diagnoses, atypical features, and higher rates of disease remission [1]. We present a case of an adolescent boy with atypical manifestations of progressive bulbar weakness, facial diplegia, and tongue fasciculations without ocular or significant limb involvement. This led to a broader differential diagnosis and ultimately resulted in a diagnosis of juvenile myasthenia gravis.

CASE REPORT

This case report is about a 14 years old boy, the third among five siblings, born to non-related parents with insignificant birth events and achievement of age-appropriate milestones. His parents noticed a change in his voice quality, which became hoarser after he turned 13, presuming it to be a pubertal change.

However, over a period of three months, they also observed that he had difficulty chewing food, particularly solids, along with occasional regurgitation, and his voice continued to become increasingly hoarse. Other than these symptoms, there was no history of seizures, focal deficits, behavioral changes, ptosis, walking difficulties, or any history of prostration.

The child had an initial encounter with his family physician, who, upon examination, found a conscious, oriented child with a hoarse voice and a weak gag reflex. An MRI of the brain was scheduled to investigate potential brainstem pathology, which yielded normal results. Subsequently, the child was referred to the neurology department for further evaluation.

A detailed neurological evaluation revealed progressive dysphagia that had advanced to liquids, along with infrequent to frequent choking episodes over the last six months, which had also led to aspiration pneumonia necessitating hospitalization. The gag reflex was weak, and there was bilateral facial weakness with lagophthalmos, difficulty blowing his cheeks, and pursing his lips, accompanied by the absence of bilateral nasolabial folds, indicating facial diplegia. The tongue was atrophied with fasciculations; however, there was no ptosis or eye muscle weakness. Muscle strength was 5/5 in all muscle groups except the neck flexors, which scored 3/5 according to the Medical Research Council (MRC) scale. Deep tendon reflexes were normally elicitable, with a flexor plantar response.

Differentials of motor neuron disease (MND), specifically bulbar amyotrophic lateral sclerosis (ALS), was considered in this case due to the presence of bulbar weakness, facial diplegia, an atrophied tongue with fasciculations, and elicitable reflexes.

Facioscapulohumeral dystrophy was initially considered based on the weak neck flexors and facial involvement, and the child was admitted for further workup. Although a nasogastric tube was passed to prevent aspiration, he developed pneumonia during the hospital stay. CSF analysis was normal, and creatine phosphokinase levels were within the normal range. Electromyography and nerve conduction studies revealed myopathy with secondary denervation. Chest radiography showed normal heart size but extensive pneumonic infiltrates, for which intravenous antibiotics and nebulization were initiated, along with the need for bubble CPAP.

A notable observation was the difference in the severity of secretions between morning and evening hours, indicating a diurnal variation. Thus, an atypical presentation of Myasthenia Gravis (MG) was considered. Due to worsening respiratory distress, the child had to be shifted to the intensive care unit for ventilatory support. Following stabilization and after informed consent, a neostigmine test was performed in the intensive care unit, resulting in significant improvement in neck flexor muscle strength. The repetitive nerve stimulation test showed a decrement of >10% (Figure 1). Acetylcholinesterase antibodies were tested and found to be positive (>120), confirming the diagnosis of immune-mediated Myasthenia Gravis. His Myasthenia Gravis Activity of Daily Living scale (MG-ADL) score was 13. MRI evaluation of the thymus revealed no hyperplasia.

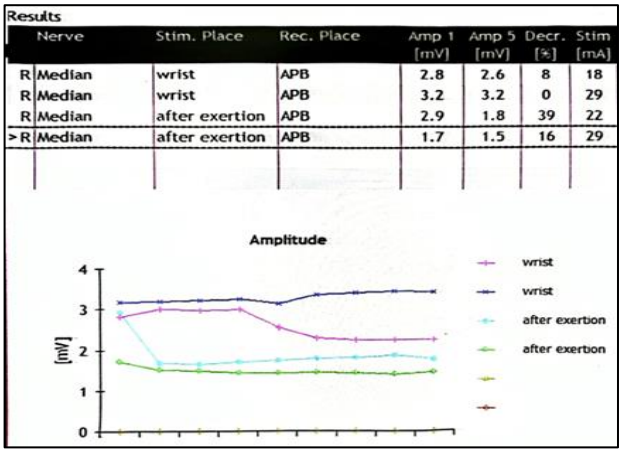

Figure 1: Repetitive nerve stimulation test showing decrement of >10%

He was started on pyridostigmine and low-dose steroids, coupled with five sessions of plasmapheresis and intensified management of pneumonia. After the plasmapheresis sessions, the intensive care team was able to wean him off the ventilator. Pyridostigmine was continued along with low-dose steroids. The nasogastric tube was removed as he was able to eat orally without any

choking episodes, and his MG-ADL score at discharge decreased to 3 as shown in figure 2.

At the three-month follow-up, he is ambulatory in all settings, has returned to school, and has experienced a reversal of tongue fasciculations. Although his voice remains hoarse as part of the disease, there is no longer any difficulty with feeding.

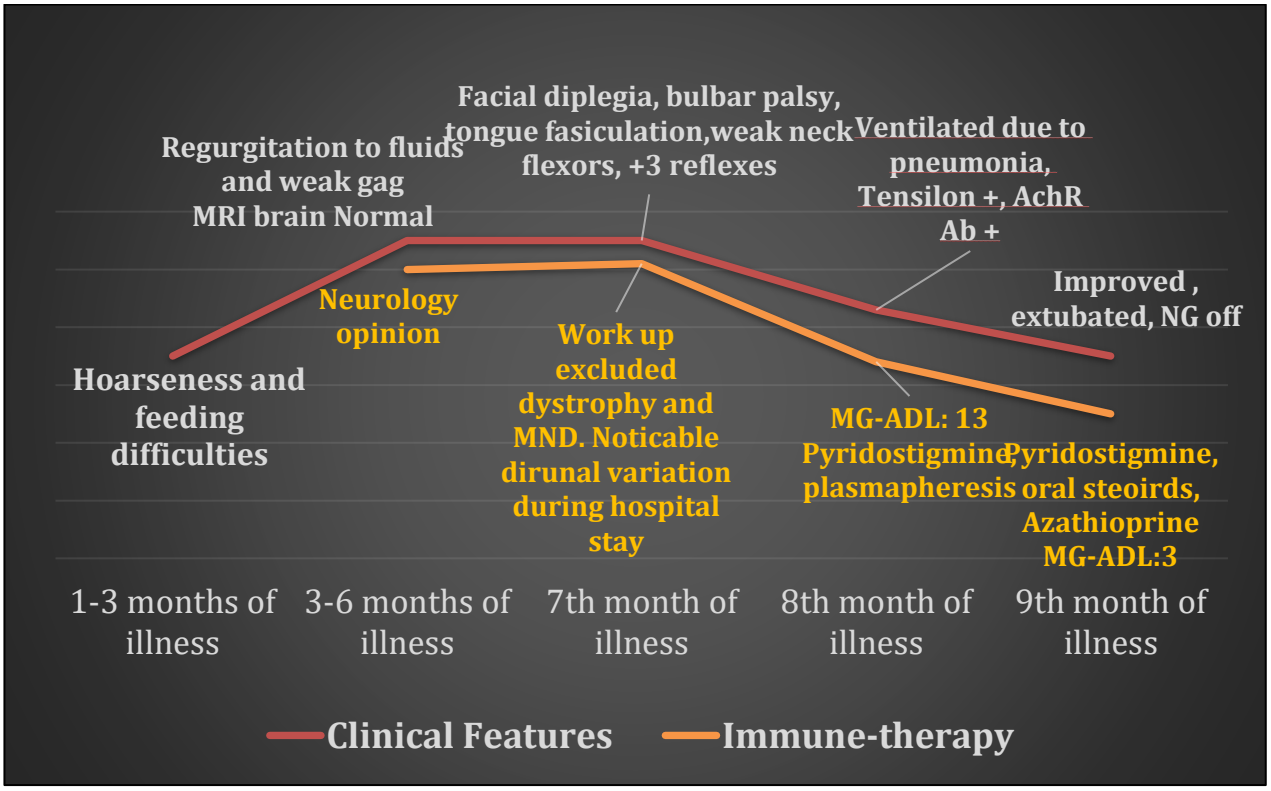

Figure 2: Timeline of events

DISCUSSION

We endeavored to explore the etiological factors that could lead to progressive bulbar palsy, facial diplegia, and isolated neck flexor weakness, ultimately resulting in the diagnosis of juvenile-onset myasthenia gravis in our adolescent patient. Such an unusual presentation is rare, with only a few case reports globally documenting similar instances, predominantly in adults and rarely in children [3, 4].

Rodolico et al. reported several atypical presentations of MG, including foot drop, asymmetric distal upper limb weakness, dropped head, acute facial diplegia, and limb-girdle type weakness [5]. Mukhtiar et al. described four toddlers with myasthenia gravis, ranging from congenital to immune-mediated MG. One patient manifested with facial diplegia and was initially managed as a Miller Fisher variant, highlighting the overlapping features of diverse neurological disorders [6]. Our patient exhibited an atypical and amalgam of features, including bulbar involvement and bilateral facial and neck flexor weakness without any ptosis, further expanding the scope of the disease. The heightened susceptibility of extraocular muscles in myasthenia gravis has been proposed to stem from variations in acetylcholine receptor isoform expression, safety factor, firing frequency, and sensitivity to complement deposition [7].

Tongue weakness is another unusual presentation in MG, as seen in our patient, who exhibited resultant fasciculations and atrophy due to the long-standing disease. This symptom is typically observed in older patients and is often associated with anti-MuSK antibodies in reported cases [8]. However, Krishnan et al. described a young woman with AChR antibody-positive MG presenting with tongue atrophy, which improved after treatment with pyridostigmine and steroids [9]. Our patient also tested positive for AChR antibodies, but the most characteristic feature is his young age.

CONCLUSION

Our case report underscores that myasthenia gravis (MG) may manifest with bulbar involvement and facial diplegia, even in the absence of ocular symptoms. In such overlapping instances, it is imperative to conduct neurophysiological examinations and electrodiagnostic tests to exclude

neuromuscular junction disorders, thereby combating diagnostic delays. Given the scarcity of reported cases with similar presentations, we advocate for mandatory screening of acetylcholine receptor antibodies in patients with bilateral facial diplegia, bulbar weakness, and tongue atrophy.

Consent to Publication: Author(s) declared taking informed written consent for the publication of clinical photographs/material (if any used), from the legal guardian of the patient with an understanding that every effort will be made to conceal the identity of the patient, however it cannot be guaranteed.

Authors Contribution: The authors confirm contribution to the paper as follows: **JA**: Concept, Analysis, approving final manuscript, **AM**: Analysis, approving final manuscript, **TS, MZR**: Literature review, drafting, approving final manuscript, **SA, NS**: Supervision, critical revision, editing, approving final manuscript.

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