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Little droop, big challenge: A child's journey with ocular myasthenia gravis

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Abstract
Context: Ocular myasthenia gravis (OMG) is a rare neuromuscular disorder that presents significant diagnostic and therapeutic challenges, particularly in pediatric cases. It can mimic other neurological disorders, leading to misdiagnosis and delayed treatment. Early identification and intervention are crucial in preventing progression and improving patient outcomes.
Purpose: This case report aims to highlight the diagnostic approach and management of ocular myasthenia gravis in a pediatric patient. It underscores the importance of comprehensive clinical evaluation and early treatment initiation to enhance the quality of life in affected children.
Methods and Material: A 7-year-old female presented with a one-year history of progressive ptosis, more pronounced in the right eye, with diurnal variation. A detailed history was taken, followed by a thorough ophthalmological examination and specialized tests, including the fatigue test, ice pack test, and repetitive nerve stimulation (RNS) test. Serological confirmation was achieved through the detection of elevated acetylcholine receptor antibodies.
Results and Conclusion: Clinical and diagnostic findings confirmed seropositive ocular myasthenia gravis. Treatment with oral pyridostigmine and corticosteroids led to significant improvement in ptosis and reduction in diurnal variation. This case emphasizes the necessity of early recognition and tailored management strategies for pediatric ocular myasthenia gravis. Continuous monitoring is essential to detect potential progression to generalized myasthenia gravis and to adjust therapy accordingly.

Keywords: Ocular myasthenia gravis, ptosis, neuromuscular disorder, childhood neurological disorders

Introduction

Ocular myasthenia gravis (OMG) is a rare yet significant neuromuscular disorder that presents diagnostic and therapeutic challenges, particularly in paediatric cases [1]. OMG is more common in children of Asian descent, with a prevalence of 3-9.1 cases per million in the pediatric population [2]. This article presents the case of a 7-year-old girl with progressive ptosis and explores the clinical approach, diagnostic intricacies, and management strategies involved in ocular myasthenia gravis. Understanding this condition is critical, as its symptoms can mimic other neurological disorders, leading to potential misdiagnosis and delayed treatment.

Materials and Methods

The evaluation of the patient was started with a detailed history taking. The patient was a seven-year-old female who presented with a one-year history of progressive drooping of both upper eyelids, more pronounced in the right eye than the left. Her symptoms had a gradual onset and demonstrated a diurnal variation, worsening by evening and with increased exertion. Her perinatal history was uneventful, with no significant past medical or family history of neuromuscular disorders. This prompted us to do a full ophthalmological examination with ptosis evaluation and other tests to rule out various ocular motility disorders.

Results

On examination, she exhibited compensatory chin elevation and head tilt, suggestive of an attempt to improve her field of vision. Best-corrected visual acuity (BCVA) in both eyes was measured at 6/9. Ptosis evaluation revealed severe right-sided ptosis and moderate left-sided ptosis. Additional findings included:

Table 1: Ocular examination

	Right eye	Left eye
BCVA	6/9	6/9
LIDS	Severe ptosis	Moderate ptosis
IOP	14.3mmHg	15.6 mm Hg
Anterior segment	Within Normal Limit	Within Normal Limit

Table 2: Ptosis evaluation

	Right eye	Left eye
MRD 1	-2 mm	-1 mm
MRD 2	5 mm	5 mm
LPS function	1 mm	3 mm
Palpebral fissure height	3 mm	4 mm

Table 3: Specific tests for ocular myasthenia gravis

Test	Inference
Fatigue test	Positive for both eyes
Ice pack test	Positive for both eyes, improvement in ptosis noted in Right eye: 2 mm Left eye: 3 mm
Repetitive Nerve Stimulation (RNS) test	Decremental response in B/L orbicularis oculi muscle s/o post synaptic neuromuscular junction transmission defect.
Serum acetylcholine receptor antibody	1.6 nmol/L (Positive).

All these tests were a hallmark feature of myasthenia gravis and thus confirmed seropositive ocular myasthenia gravis. After confirming this diagnosis, the patient was initiated on a combination of oral steroids and anticholinesterase inhibitors which included:

1. **Oral pyridostigmine 15mg QID for 2 weeks:** A cholinesterase inhibitor used to enhance neuromuscular transmission.
2. **Oral prednisolone 20mg once alternate day for 5 days followed by tapering over 2 weeks:** Low-dose corticosteroids were administered to suppress the autoimmune response and prevent disease progression.

After planning the treatment course, regular follow-up visits were scheduled to assess response to treatment and monitor potential side effects. Over several weeks, significant improvement in ptosis was observed, with reduced diurnal variation and decreased need for compensatory head tilt.

Fig 1: Pre treatment ptosis

Fig 2: Post treatment (One month follow up)

Discussion

Approximately 20% patient with myasthenia gravis have involvement of only extraocular muscles [3]. It is called the great masquerader owing to its varied clinical presentations [4]. Serum acetylcholine antibody test is vital in diagnosis of myasthenia gravis and is positive in 80% of cases [5]. Early diagnosis and initiation of treatment is key to improving quality of life as complications resulting from delay of treatment can be lethal. The ice pack test and fatigue test remain invaluable bedside tools for rapid assessment, particularly in resource-limited settings [6].

Pediatric Myasthenia Gravis specifically poses a unique challenge as this condition is more frequently seen in adults. Hence, pediatric cases require special considerations [7]

- **Atypical presentation:** Unlike adults, pediatric patients may present with subtle signs such as chin elevation or intermittent ptosis.
- **Treatment tolerance:** Long-term steroid use poses risks of growth suppression and osteoporosis, necessitating a balanced approach.
- **Risk of generalization:** Up to 50% of pediatric ocular myasthenia gravis cases may progress to generalized myasthenia gravis within two years, emphasizing the need for close surveillance.

We should always be meticulous during or evaluation and examination as misdiagnosis can lead to unnecessary interventions. Conditions such as congenital myopathies and mitochondrial disorders can present similarly, underscoring the need for confirmatory electrophysiological and serological testing.

Conclusion

This case highlights the importance of maintaining a high index of suspicion for ocular myasthenia gravis in children presenting with ptosis and diurnal variation. Comprehensive evaluation, including fatigue testing, ice pack test, and serological markers, is essential for accurate diagnosis. Early initiation of therapy can lead to significant symptomatic improvement, reducing functional impairment and enhancing the patient's quality of life. Ongoing research into pediatric-specific treatment strategies will further refine the management of this challenging condition.

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