J Med Sci 2025;45 (6):258-261
DOI: 10.4103/jmedsci.jmedsci_89_25

CASE REPORT

Pediatric Ocular Myasthenia Gravis Presenting with Alternating Ptosis and Gaze Palsy: A Case Report and Literature Review

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Pediatric ocular myasthenia gravis (OMG) is a rare autoimmune disorder characterized by fluctuating ptosis and ophthalmoparesis. We report the case of a 4-year-old female presenting with left upper eyelid ptosis, head tilting, and gaze palsy since 2 years old. Initial evaluation suggested a brainstem lesion owing to a positive Parinaud sign, with an 8.0-mm pineal cyst on magnetic resonance imaging. Follow-up imaging at 9 months revealed no progression. However, a positive ice-pack test combined with serologic positivity for anti-acetylcholine receptor antibodies and a decremental response to repetitive nerve stimulation confirmed the diagnosis of pediatric OMG. The patient partially and temporarily responded to combined pyridostigmine/prednisolone treatment. She subsequently underwent frontalis sling suspension surgery and achieved significant improvements in ptosis and visual function. This case highlights the diagnostic complexities of pediatric OMG and the importance of surgical intervention, specifically frontalis sling suspension, in the management of ptosis when pharmacological therapy is unsatisfactory.

Key words: Case report, frontalis sling suspension, ophthalmoparesis, pediatric ocular myasthenia gravis, ptosis, surgical intervention

INTRODUCTION

Myasthenia gravis (MG) is an autoimmune disorder affecting neuromuscular junctions, leading to fluctuating skeletal muscle weakness and fatigability, predominantly in the extraocular muscles. MG can manifest in ocular or generalized forms, with ocular symptoms such as ptosis and ophthalmoparesis often being the initial presentation.[1,2]

Pathogenic antibodies, mainly against acetylcholine receptors, lead to receptor degradation, complement activation, and impaired neuromuscular transmission. Pediatric cases, particularly those with prepubertal onset, often show isolated ocular symptoms, fewer thymic abnormalities, and a higher rate of seronegativity. The extraocular muscles are especially vulnerable, and some patients develop treatment-resistant ophthalmoplegia.[3]

Diagnosing pediatric ocular MG (OMG) is challenging because of symptom fluctuation overlap with other pediatric neuro-ophthalmic disorders and limited cooperation in young children. Differential diagnoses include congenital ptosis, brainstem lesions, mitochondrial disorders, and neurogenic syndromes, evaluated through clinical tests (e.g., ice-pack test), serological antibody assays, and electrophysiological studies.[4] Anti-acetylcholine receptor (AChR) antibodies positivity and repetitive nerve stimulation (RNS) decrement can confirm an OMG diagnosis.[1,4]

Despite diagnostic advances, literature on long-term outcomes and surgical strategies remains limited. Recent studies have enhanced our understanding of the clinical presentation and treatment responses in pediatric autoimmune OMG, and its incidence is higher in East Asia with geographic variation.[5,6] Pharmacological treatment is the primary approach, but several pediatric patients present with persistent ocular symptoms,

Received: May 01, 2025; Revised: May 19, 2025;
Accepted: June 02, 2025; Published: October 04, 2025
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How to cite this article: Tseng YS, Chien KH, Hu CF, Chen SJ. Pediatric ocular myasthenia gravis presenting with alternating ptosis and gaze palsy: A case report and literature review. J Med Sci 2025;45:258-61.

including ptosis and diplopia.[7] Similarly, despite pyridostigmine and corticosteroid therapy, nearly a third of patients experience refractory ptosis requiring additional intervention.[8]

Further, only few studies have discussed the surgical interventions for persistent ptosis in pediatric patients. A previous study demonstrated only an 8% revision in 25 pediatric eyelids treated with silicone slings for congenital ptosis and no signs of keratopathy,[9] and another study reported stable postoperative eyelid position over a 3-year follow-up in a child with congenital MG.[10] Similarly, another study described favorable outcomes in 23 MG cases, including 4 pediatric patients.[11]

Herein, we demonstrate the diagnostic complexities and management strategies in a pediatric case of OMG and emphasize the role of early surgical intervention.

CASE REPORT

A 4-year-old female presented with left upper eyelid ptosis and intermittent left head tilt for several months. She initially received oral corticosteroids and Vitamin B12 at 1 year and 3 months of age with transient improvement. However, at 2 years and 5 months of age, ptosis recurred and worsened, accompanied by bilateral upward gaze palsy and increased left head tilt [Figure 1a]. She also experienced frequent falls while riding a balance bike, which prompted further evaluation. Neurological examination revealed a positive Parinaud's sign, raising concerns regarding brainstem lesions and intracranial tumors.

Brain magnetic resonance imaging (MRI) at 2 years and 6 months of age identified an 8.0-mm pineal cyst, raising the suspicion of a pineal tumor [Figure 2]. However, follow-up imaging 9 months later showed no interval change, reducing the likelihood of a neoplastic process. Later, the ptosis alternated sides and continued to worsen over the following months. By 3 years and 5 months of age, the patient was admitted because of further deterioration, particularly afternoon fatigue and visual impairment.

Ophthalmological examination at this stage revealed reduced visual acuity (OD/OS 6/10, 6/15). Levator function was normal (12 mm), but marginal reflex distance 1 (MRD1) was only 1 mm, OS. A 30-s ice-pack test improved MRD1–4 mm, OS. Given these findings, OMG was strongly suspected. Visual evoked potentials (VEP) revealed prolonged P100 latencies, suggestive of bilateral prechiasmatic optic neuropathy.

Serology confirmed positive anti-AChR antibodies (4 nmol/L, normal range < 0.5 nmol/L), whereas antibodies for muscle-specific tyrosine kinase, aquaporin-4, and myelin oligodendrocyte glycoprotein were negative. RNS of the right facial nerve showed 10%–24% decrement, consistent with neuromuscular junction dysfunction. These findings contributed to the final diagnosis of pediatric OMG.

Treatment was initiated with pyridostigmine (3.5 mg/kg/day, later increased to 5.2 mg/kg/day) and prednisolone (0.4 mg/kg/day), but ptosis persisted, prompting a surgical approach. At 3 years and 11 months of age, she underwent frontalis sling suspension surgery with silicon rods. Postoperatively, MRD1 improved to 4 mm, OS, and head tilting decreased; however, mild ocular deviation persisted.

On the 8th day post-surgery, visual acuity partially improved to OD/OS 6/10 and 6/7.5, and ptosis almost resolved [Figure 1b]. Continued treatment with pyridostigmine 5.2 mg/kg/day was recommended, while the prednisolone dose was tapered gradually to 0.2 mg/kg/day. At the 6-month follow-up visit, the patient remained free of ptosis. Levator function was preserved postoperatively, with complete eyelid closure and no evidence of exposure keratopathy. The patient remains under follow-up to assess long-term functional outcomes and monitor potential progression to generalized MG. Further ophthalmic evaluation is planned to clarify the cause of the prior VEP findings and assess for any residual or coexisting optic pathway dysfunction.

DISCUSSION

Diagnosing pediatric OMG is challenging due to overlapping signs. Our patient's initial presentation – ptosis, gaze palsy, and

Figure 1: Photographs of the patient. (a) First clinic visit (2 years old), showing left upper eyelid ptosis and head tilting; (b) At 3 years 11 months, 8 days after frontalis sling suspension surgery, showing improved eyelid position and reduced head tilting

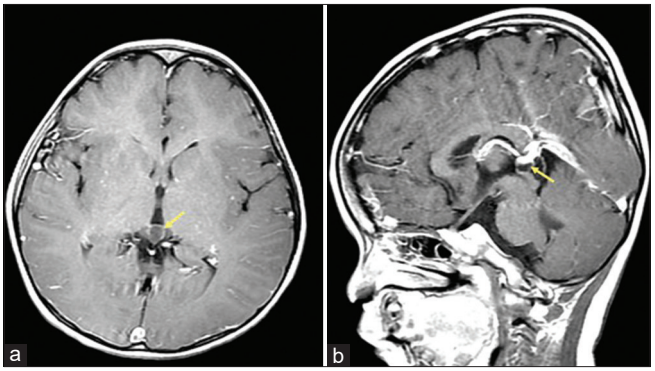

Figure 2: Brain magnetic resonance imaging performed at 2 years and 6 months of age. (a) Axial T1-weighted image showing a hypodense 8.0-mm pineal cyst. (b) Sagittal T1-weighted image demonstrating the pineal cyst with no evidence of compression or surrounding mass effect

Table 1: Comparison of pediatric ptosis cases involving frontalis sling surgery

	Our case	Bradley *et al*., 2001[10]	Carter *et al*., 1996[9]
Pediatric cases	1	1	14
Age at surgery	3 years 11 months	6 years	Mean 1.5 years
Diagnosis	Ocular MG	Congenital MG	Congenital ptosis
Presentation	Alternating ptosis, head tilting, gaze palsy	Bilateral ptosis, ophthalmoplegia	Ptosis, chin-up head posturing, occlusion of visual axis
Medical therapy before surgery	Pyridostigmine, low-dose prednisolone	Pyridostigmine, 3,4-DAP	Not MG-specific; general pediatric indication
Surgery	Frontalis sling with silicon rod	Frontalis sling with autogenous fascia lata	Frontalis sling with silicone rod
Preoperative MRD1	1 mm (OS)	Not specified	0–1 mm
Postoperative MRD1	4 mm (OS)	Good position at 3 months	Good to excellent in 92%
Follow-up duration	6 months	3+ years	Mean 16 months
Exposure keratopathy	None observed	None reported	Transient in 1 child, no chronic cases
Recurrence	None within 6 months	None at 3+ years	2 sling (8%) recurrence

MG=Myasthenia gravis; MRD1=Marginal reflex distance to upper lid; DAP=Diammonium phosphate

Parinaud sign – mimicked brainstem pathology. The detection of an 8.0-mm pineal cyst further complicated the diagnostic process. However, serial MRI showed no progressive changes, and the subsequent worsening of ptosis, particularly later in the day, warranted the reconsideration of a different diagnosis.

A positive ice-pack test reduced MRD1, and subsequent AChR antibody positivity and RNS decrement established the diagnosis of OMG. Prompt diagnosis is key to prevent disease progression and associated visual complications.[1,8]

While our patient showed limited response to pyridostigmine and steroids, persistent ptosis necessitated surgical intervention. Frontalis sling suspension, although typically used for congenital or neurogenic ptosis, may benefit MG cases with suboptimal response to medical therapy. Till date, no guideline has defined the optimal surgical timing in pediatric OMG; however, preoperative evaluation – especially of Bell's phenomenon and exposure keratopathy risk – is essential.[10] In this case, surgery at 2 years and 8 months after onset resulted in a stable eyelid position without exposure keratopathy, supporting early surgical intervention in selected pediatric patients. The alternating nature of ptosis observed in our patient, while uncommon, may reflect asymmetric fatigability rather than structural differences and should prompt careful monitoring. The observed improvement in visual acuity may be attributed to the relief of the mechanical occlusion of the visual axis caused by prolonged ptosis, thereby facilitating better visual input. We have summarized and compared published pediatric ptosis surgery cases in Table 1.

This case report has some limitations. First, this was a single case study, which limits its generalizability. In addition, further imaging such as chest computed tomography was not performed to evaluate thymic pathology, although thymectomy is generally not indicated for pediatric OMG.[1] The absence of standardized quality-of-life or functional outcome measures also limits the evaluation of surgical success. The follow-up period was relatively short, and longer-term monitoring is essential given the risk of recurrence and potential progression to generalized MG in up to 23% of cases as previously reported.[7] Future studies should explore the long-term surgical outcomes and refine the criteria for surgical intervention in pediatric MG.

CONCLUSION

This case highlights the diagnostic complexities and therapeutic considerations in pediatric OMG, particularly when presenting with alternating ptosis with waxing, waning, and gaze palsy. Early recognition and treatment are essential to prevent functional impairment. Surgical intervention should be considered in cases of persistent ptosis despite optimal medical therapy. Future research should focus on the long-term outcomes of surgical interventions and optimal treatment strategies for pediatric OMG.

Declaration of patient consent

The study was conducted in accordance with the Declaration of Helsinki and approved by the Institutional Review Board of Tri-Service General Hospital with approval number B20251506; approval date 2025/3/21. The authors certify that they have obtained all appropriate patient consent forms. In the form, the legal guardian has given her consent for images and other clinical information to be reported in the journal. The guardian understands that her name and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

Acknowledgments

We would like to thank Editage (www.editage.com.tw) for English language editing.

Data availability statement

The data that support the findings of this study are available from the corresponding author, Chen, upon reasonable request.

Financial support and sponsorship

The study was supported by grants from Tri-Service General Hospital (TSGH_C01_114027) to SJ Chen.

Conflicts of interest

There are no conflicts of interest.

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