

CASE REPORTS

Beyond Muscle Weakness: Sleep Dysfunction and Increased Intracranial Pressure in Pediatric Myasthenia Gravis

Humza Siddiqi, MD¹, Liz Lezama, MD², Anna Wani, MD²

¹ Family and Community Medicine, The University of Texas Southwestern Medical Center, ² Sleep Medicine, The University of Texas Southwestern Medical Center

Keywords: Myasthenia gravis, MuSK antibodies, Pediatric sleep, Restless legs syndrome, Intracranial hypertension, Papilledema, Hypersomnolence, Neuromuscular disorders

<https://doi.org/10.65738/001c.155381>

Texas Family Medicine Research Journal

Vol. 1, Issue 3, 2026

1. Introduction

Myasthenia gravis (MG) is an autoimmune disorder characterized by fluctuating skeletal muscle weakness due to impaired neuromuscular transmission. Anti-muscle-specific kinase (MuSK) antibodies account for a minority of MG cases and are associated with bulbar involvement and increased risk of respiratory crises. Pediatric MuSK-positive MG remains uncommon, and data describing its relationship with sleep disorders are limited. We present a pediatric case of MuSK-positive MG with sleep dysfunction and elevated intracranial pressure, illustrating an under-recognized clinical overlap between neuromuscular, sleep, and cerebrospinal pressure dysregulation.

2. Case Presentation

A 9-year-old Mediterranean female with anti-MuSK antibody-positive MG and ophthalmoparesis was referred to the pediatric sleep clinic for a one-year history of insomnia, restless sleep, and daytime somnolence. She exhibited mouth breathing, morning headaches, and frequent daytime naps at school. Prior to presentation, she had trialed melatonin, discontinued due to nightmares. Examination showed BMI 26.3 kg/m², Mallampati IV airway, and 4+ tonsillar hypertrophy. Ferritin was 25 ng/mL, meeting pediatric RLS criteria; oral iron was initiated.

Polysomnography demonstrated normal sleep efficiency and architecture, without evidence of sleep-disordered breathing or significant periodic limb movements. Actigraphy confirmed normal average sleep duration. Despite ferritin improvement to 58 ng/mL, symptoms persisted, and gabapentin was started for refractory RLS. The patient demonstrated improvement in restless legs symptoms and overall sleep quality following initiation of gabapentin; however, the subsequent development and persistence of primary intracranial hypertension complicated the clinical picture and contributed to ongoing sleep difficulties.

Her disease course was complicated by recurrent myasthenic crises prior to her initial sleep evaluation. After starting gabapentin, she experienced her first hospitalization for viral gastroenteritis and mycoplasma infection, during which gabapentin was held. The second hospitalization occurred for suspected resistant mycoplasma pneumonia/bacteremia, diagnosed by bronchoalveolar lavage, and required intubation; following recovery, gabapentin was resumed, though adherence was inconsistent given these significant admissions. Approximately three months later, she was admitted twice for severe headaches, at which time ophthalmologic assessment revealed bilateral papilledema on OCT RNFL imaging. MRI confirmed mild papilledema, and lumbar puncture demonstrated an opening pressure of 22 cmH₂O. These findings met diagnostic criteria for primary intracranial hypertension (PIH), and the diagnosis was established at this time. She started on acetazolamide 250 mg twice daily. Due to incomplete adherence and persistent papilledema, repeat lumbar puncture three months later again showed an opening pressure of 22 cmH₂O. Doses of acetazolamide were titrated up to 750 mg twice daily with mild subsequent improvement. At her last follow-up visit in October, she continued to experience debilitating headaches, disrupted sleep, weight loss, and daytime somnolence. At that time, gabapentin was increased to 200 mg nightly, and hydroxyzine and magnesium were added to her regimen.

3. Discussion

Myasthenia gravis (MG) is an autoimmune disorder of the neuromuscular junction characterized by fluctuating skeletal muscle weakness mediated by antibodies against postsynaptic targets, including the acetylcholine receptor and muscle-specific kinase.^{1,2} Although MG is well described in adults, pediatric MG remains uncommon and may present with heterogeneous and atypical features, contributing to diagnostic delay.¹ Recent advances in pediatric MG management emphasize individualized, targeted immunotherapies and multidisciplinary care.²

Neurologic symptoms in MG are often attributed to neuromuscular weakness; however, non-motor manifestations may coexist and warrant independent evaluation. Primary intracranial hypertension (PIH), also referred to as idiopathic intracranial hypertension, is characterized by elevated intracranial pressure in the absence of structural brain abnormalities or cerebrospinal fluid pathology.³⁻⁵ Contemporary pediatric consensus guidelines emphasize early recognition of PIH to prevent irreversible visual impairment and highlight the importance of neuro-ophthalmologic assessment when children present with headaches, visual changes, or papilledema.^{3,4}

While PIH is increasingly recognized in pediatric populations, its association with autoimmune neuromuscular disorders such as MG is rarely reported.^{5,6} Isolated case reports describe the coexistence of MG and pseudotumor cerebri, often in the context of systemic inflammatory or autoimmune conditions, underscoring the rarity of this clinical combination.⁶ To date, pediatric cases documenting both conditions concurrently remain exceedingly limited in the literature.

This case is notable for the coexistence of MuSK-positive pediatric MG and PIH, reinforcing the need for clinicians to maintain a broad differential diagnosis when evaluating neurologic symptoms that cannot be fully explained by MG alone. Headache, visual complaints, and sleep disturbance should prompt consideration of intracranial pathology and adherence to established pediatric PIH diagnostic guidelines.³⁻⁵

The presence of sleep dysfunction further complicates the clinical picture and may contribute to symptom burden and reduced quality of life. Although sleep-related complaints are increasingly recognized in children with neurologic and autoimmune conditions, they remain underexplored in pediatric MG populations. The overlapping symptomatology highlights the importance of a multidisciplinary approach involving neurology, ophthalmology, sleep medicine, and primary care to ensure comprehensive evaluation and management.

This case contributes to the limited pediatric literature describing the coexistence of MG and PIH and underscores the importance of avoiding diagnostic anchoring. Increased awareness of this rare association may facilitate earlier diagnosis, guide appropriate workup, and ultimately reduce morbidity in affected children.

4. Conclusions

Pediatric MuSK-positive MG may present with complex, overlapping neurological and sleep manifestations beyond typical respiratory involvement. The co-occurrence of RLS and intracranial hypertension in this case underscores the need for comprehensive sleep evaluation in MG and vigilance for secondary contributors such as PIH when headaches or visual symptoms emerge.

IRB Statement

Ethical review and approval were waived for this single-patient case report as per institutional policy at the University of Texas Southwestern Medical Center.

Informed Consent

Written informed consent for publication was obtained from the patient's parent/legal guardian.

Data Availability

No new datasets were generated or analyzed in this report.

Author Contributions

Conceptualization, L.L., A.W., and H.S.; data curation, L.L.; writing—original draft preparation, L.L.; writing—review and editing, A.W. and H.S.; supervision, H.S. All authors have read and agreed to the published version of the manuscript.

Conflicts of Interest

The authors declare no conflict of interest.

List of Abbreviations

- MG – Myasthenia Gravis
- **MuSK** – Muscle-Specific Kinase
- PIH – Primary Intracranial Hypertension
- RLS – Restless Legs Syndrome
- OSA – Obstructive Sleep Apnea
- OCT **RNFL** – Optical Coherence Tomography Retinal Nerve Fiber Layer
- BMI – Body Mass Index

Submitted: November 16, 2025 CDT. Accepted: January 15, 2026 CDT. Published: March 24, 2026 CDT.

REFERENCES

1. Sanders DB, Wolfe GI, Benatar M, et al. International consensus guidance for management of myasthenia gravis. *Neurology*. 2016;87(1):1-7. doi:[10.1212/WNL.0000000000002790](https://doi.org/10.1212/WNL.0000000000002790). PMID:27358333
2. Ramdas S, Painho T, Vanegas MI, Famili DT, Lim MJ, Jungbluth H. Targeted treatments for myasthenia gravis in children and adolescents. *Pediatr Drugs*. 2024;26:719-740. doi:[10.1007/s40272-024-00649-3](https://doi.org/10.1007/s40272-024-00649-3)
3. Mehta V, Moran M, Amin B, et al. Consensus recommendations for the assessment and management of idiopathic intracranial hypertension in children and young people. *Arch Dis Child*. 2024;109(8):654-658. doi:[10.1136/archdischild-2023-326545](https://doi.org/10.1136/archdischild-2023-326545)
4. Alder Hey Children's NHS Foundation Trust. *Guideline on Diagnosis and Management of Idiopathic Intracranial Hypertension in Children*. Alder Hey Children's NHS Foundation Trust; 2024.
5. Aylward SC, Reem RE. Pediatric intracranial hypertension. *Pediatr Rev*. 2018;39(3):121-129. doi:[10.1542/pir.2016-0226](https://doi.org/10.1542/pir.2016-0226)
6. Fein AS, Trejo-Bittar HE, Shende MR, Scalzetti EM, Ko MW. Castleman disease presenting with pseudotumour cerebri and myasthenia gravis: a case report and literature review. *Neuroophthalmology*. 2019;43(3):185-191. doi:[10.1080/01658107.2018.1484932](https://doi.org/10.1080/01658107.2018.1484932). PMID:31312243