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Case Report

What is Your Suspected Diagnosis? A Challenging Case of Unilateral Ptosis with Eyelid Edema in a Pediatric Patient.

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Abstract

Myasthenia gravis (MG) is an autoimmune disorder characterized by weakness and fatigability of skeletal muscle. Juvenile myasthenia gravis (JMG) is MG in patients under the age of 18. The incidence is estimated to be 5.9 - 8.7 per million person-years. In ocular JMG, a subtype of JMG, symptoms are isolated to eye muscles. This case report presents the diagnostic journey of a 3-year-old non-Asian female from Virginia, USA, who initially exhibited symptoms consistent with ocular JMG. The patient's early symptoms, including eyelid drooping and watery eyes, were initially misattributed to eye irritation and upper respiratory infection symptoms. However, suspicion for JMG emerged as her condition progressed to include bilateral ptosis, facial weakness, and other bulbar symptoms. The diagnostic process involved interdisciplinary collaboration and continued revisal of the differential diagnoses as symptoms evolved. The diagnosis was ultimately confirmed with a positive JMG panel, including elevated acetylcholine receptor (AChR) binding and modulating antibodies. This case highlights the importance of considering JMG in the differential diagnosis of ocular and bulbar symptoms, and the challenges associated with diagnosis of rare diseases. Tailored management strategies, including acetylcholinesterase inhibitors and potential immunosuppressive agents, should be considered based on individual patient needs.

Keywords : Myasthenia Gravis, Neuromuscular Disorders, Ptosis, Autoimmune, Facial Weakness.

INTRODUCTION

Myasthenia gravis (MG) is an antibody-mediated autoimmune disorder characterized by weakness and fatigability of skeletal muscle. The pathogenesis is often due to an antibody-mediated, immunological attack on the neuromuscular junction postsynaptic nicotinic acetylcholine receptor (AChR) [1]. Antibodies to muscle-specific kinase (MuSK) and leucine rich protein 4 (LRP4) have also been associated with MG [1]. MG in patients under the age of 18 is termed juvenile myasthenia gravis (JMG) [1]. The incidence is estimated to be 5.9 - 8.7 per million person-years [2]. JMG is more prevalent in Asian populations, with a greater proportion of prepubertal onset, between ages 5-10. In contrast to postpubertal patients and adults, prepubertal children show an equal male to female ratio [3]. JMG tends to have a better prognosis than adult-onset MG, with a higher rate of spontaneous remission [4].

In ocular JMG, a subtype of JMG, clinical manifestations are isolated to the levator palpebrae suprioris, orbicularis oculi, and extraocular muscles affecting eye movement [1]. Ocular JMG symptoms include painless eyelid droop (unilateral, bilateral, or alternating), double vision, dizziness, gait instability, and light sensitivity, typically worsening throughout the day and improving with rest [1]. Ocular JMG is the most frequent initial presentation in prepubertal JMG, with ptosis being the most common symptom [4]. Secondary generalized MG (SGMG) occurs when patients who initially presented with ocular symptoms develop weakness in other muscle groups [5]. The likelihood of symptom generalization is significantly lower in JMG, occurring in 21.9% of cases, compared to 50-80% in adult-onset MG [4].

We present the case of a 3-year-old non-Asian female whose initial symptoms were diagnostically challenging but ultimately found to be consistent with ocular JMG.

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CASE PRESENTATION

The female patient was born at 25 6/7 weeks gestation due to uterine infection; she spent 2 months in the NICU with an uncomplicated course. Her birth weight was 790 g. She had no retinopathy of prematurity or intraventricular hemorrhage. She walked at 18 months and received speech therapy, but has had normal development in all domains since. She has no prior surgeries, no known allergies, and no family history of autoimmune disease.

In early August, four months prior to diagnosis, the now 3 year old patient and her mother visited the PCP for evaluation of right eyelid droop of 3 day duration. The mother reported bilateral watery eyes and rhinorrhea. The right eyelid was not red or painful. However, the child was noted to be rubbing both eyes.

During the initial physical exam, the right upper eyelid was edematous and the right eye was more closed than the left. Sclera was clear and no discharge was noted. There was no conjunctival injection or tearing. Her face was otherwise symmetrical and the patient was able to raise the upper lid with upgaze.

In late August, ophthalmology evaluated the patient for right sided ptosis and found anisocoria with right sided miosis. No anhidrosis or left eye symptoms were reported. Due to concern for Horner's syndrome, neuroblastoma workup was initiated. MRI of the brain, C spine and chest was unremarkable with the exception of incidental pleural nodules. Abdominal US showed no sonographic abnormalities. XRay of the orbits revealed no fracture, dislocation, radiopaque foreign body, or soft tissue swelling and a midline nasal septum. Neuroendocrine testing showed normal Vanillylmandelic Acid and Homovanillic Acid. Laboratory work up, including thyroid stimulating hormone and free thyroxine, was normal. Anisocoria was suspected to be a natural variant.

In early September, the mother noted the ptosis had progressed to both eyes. Thus, the patient was re-evaluated by her PCP. Ptosis was reported as mild in the morning and progressively worse throughout the day. Her mother noticed the patient was lifting her chin up to see by the end of the day, due to impairment from ptosis. Physical exam was difficult to obtain but bilateral ptosis was noted. A JMG panel was ordered and the patient was referred to neurology.

Two weeks later, the patient had a neurology consultation. At this time the differential diagnoses included JMG, chronic progressive external ophthalmoplegia, and congenital myasthenic syndrome.

In late September, the JMG panel returned positive, with elevated titers for the AChR Binding Antibody and AChR Modulating Antibody. Elevated titers of the AChR Binding Antibody have a high specificity for JMG, making it an appropriate clinical diagnostic test [6]. Recent studies have also found clinical significance in evaluating AChR Blocking Antibody and AChR Modulating Antibody titers [6]. This was done using a radio immunosuppression assay.

In early November, additional symptoms had emerged: right sided lip drooping, "hoarse voice", and coughing while drinking from a straw. The mother did not report any vision changes, chewing fatigue, choking, arm/leg weakness, difficulty breathing, or decreased exercise tolerance. The patient had no difficulty climbing stairs, running, or jumping. Examination showed prominent ptosis with right > left, mild right lower facial weakness upon activation, and hypophonia. No proptosis or scleral injection were noted. Eye movements were conjugate. As history, exam, and lab work were consistent with JMG, symptomatic management with the acetylcholinesterase inhibitor, Pyridostigmine, was started. Future consideration of steroid or steroid sparing disease modifying agents was planned as needed.

At the follow up appointment in mid November, the patient was noted to have significant improvement in symptoms with medication. She no longer choked on water, and both her voice and ptosis had improved, though facial droop remained. No diurnal fluctuation of symptoms was noted under medication.

DISCUSSION

JMG is rarely the primary differential diagnosis in cases of unilateral ptosis with eyelid edema, often resulting in extensive workups and delayed diagnosis. While this patient ultimately developed non-ocular symptoms, this patient presented initially with symptoms consistent with ocular JMG. The progression of this case underscores the diagnostic challenges of rare diseases.

The patient's initial presentation with unilateral eyelid drooping and watery eyes, was misattributed to eye irritation and upper respiratory infection. Subsequently her ptosis and normal variant anisocoria created concern for Horner's syndrome and led to neuroblastoma workup. Development of bilateral ptosis worsening throughout the day ultimately raised suspicion for ocular JMG, further supported by the presence of AChR antibodies. Ultimately, this patient exhibited bulbar symptoms of hoarse voice, coughing while drinking, and mild right lower facial weakness, prompting a revised diagnosis of generalized JMG. Notably, asymmetric facial weakness is uncommon [4].

Ocular JMG can be mistaken for conditions such as Chronic Progressive External Ophthalmoplegia, Grave's Ophthalmopathy, and Congenital Myasthenic Syndrome. Accurate diagnosis of any rare disease requires thorough evaluation, broad differential consideration, and close monitoring for symptom progression.

Early recognition is critical. Despite the more favorable

prognosis of JMG compared to adult-onset MG, both JMG and adult-onset MG patients share a similar risk of myasthenic crisis—a life-threatening complication marked by respiratory failure [1]. While deaths from myasthenic crises are rare, children are considered to be at higher risk [1]. Primary treatment options include corticosteroids, immunoglobulins, and plasma exchange [1]. While this patient did not develop respiratory compromise, she had involvement of muscle groups important in airway protection and nutrition, as evidenced by her bulbar symptoms.

Diagnosis is further complicated by the high seronegativity rate in the JMG population; 36 - 50% of patients may lack detectable antibodies [7]. In this case AChR antibodies supported the diagnosis, but many pediatric patients with similar presentations may remain seronegative, contributing to delays in definitive diagnosis.

Histological and in vitro thymic changes have been linked to AChR seropositive MG, highlighting the thymus's potential role in the disease's pathogenesis. In JMG, the most common thymic abnormality is thymic hyperplasia [1]. Thymic imaging is recommended following diagnosis [1]. This patient's MRI chest imaging did not find thymic abnormalities.

JMG management includes symptomatic treatment with acetylcholinesterase inhibitors, which increase the neuromuscular junction concentration of acetylcholine by inhibiting neurotransmitter breakdown. This patient responded well to the acetylcholinesterase inhibitor, Pyridostigmine.

Diagnosis required collaboration among ophthalmology, radiology, and neurology, emphasizing the importance of interdisciplinary care.

The age and ethnicity of the patient are relevant factors in understanding this case presentation in the larger epidemiological context of JMG. The ethnicity and age of onset of this patient both deviate from the typical patient population associated with JMG. Race and ethnicity may influence the clinical phenotype, pathophysiology, and treatment response in JMG, likely reflecting underlying genetic and immunologic variation across populations [8]. The consideration of gender identity is not relevant given the patient's young age but should be acknowledged in the future as gender identity may influence healthcare decisions and disease experiences.

CONCLUSION

This case report functions as a teaching tool highlighting the multidisciplinary approach needed in diagnosing a rare disease. Diagnosis required recognizing variable presentations of a rare disease, which could easily be conflated with other diseases. This report outlines essential steps in the diagnostic process, including pivotal imaging and laboratory tests. It also highlights the risk of diagnostic errors resulting from premature diagnostic closure and stresses the importance of continually revising the differential diagnosis as symptoms evolve. Furthermore, it emphasizes the critical need for interdisciplinary collaboration and effective communication among healthcare professionals to ensure accurate diagnosis and optimal management of rare diseases.

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Ethical Approval and Informed Consent Statements

Our institution does not require ethical approval for reporting individual cases or case series. Written informed consent was obtained from a legally authorised representative for anonymised patient information to be published in this article.

Contributors Statements

Thisha Thiagarajan, BS conceptualized and designed the manuscript, drafted the initial manuscript, and critically reviewed and revised the manuscript.

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