Case Report

Juvenile myasthenia gravis

Abstract

Juvenile myasthenia gravis is a rare disorder acquired in childhood, representing 10% to 15% of all cases of myasthenia gravis. Like the adult form, it is generally characterized by an autoimmune attack on acetylcholine receptors at the neuromuscular junction. Most patients present with ptosis, diplopia, and fatigability. More advanced cases may also have bulbar problems and limb weakness and may progress to paralysis of the respiratory muscles.

Key words: Acetylcholine receptors antibodies, opthalmoplegia, ptosis

Meenakshi Kalyan, Shubhangi A. Kanitkar, Anu N. Gaikwad

Department of Medicine, Padmashree Dr. D.Y. Patil Medical College Hospital and Research Centre, Pune, Maharashtra, India

Address for the Correspondence: Dr. Meenakshi Kalyan, Department of Medicine, Padmashree Dr. D. Y. Patil Medical College Hospital and Research Centre, Pune, Maharashtra, India. E-mail: drmeenakshi.kalyan@gmail.com

Access this article online

Website: www.ijmedph.org

DOI: 10.4103/2230-8598.144137

Quick response code:

INTRODUCTION

Juvenile myasthenia gravis (JMG) is an acquired, autoimmune disease occurring before age 16 years.[1] It is a rare disorder of the childhood which accounts for less than 10-15% with an incidence of 1-5 per million per year.[2] There is a lack of epidemiological data from India and only few case reports are from Asia. Pre-pubertal children tend to have more ocular presentations whereas post-pubertal cases have more generalized disease and female preponderance.[3] Detection of acetylcholine receptor (AChR) antibodies supports the diagnosis of JMG. Early recognition and management of JMG prevents the progression of symptoms and results in low morbidity and mortality. We report a 15-year-old boy with JMG.

CASE REPORT

A 15-year-old schoolboy presented with drooping of both eyelids, restriction of eye movements and diplopia with diurnal variation worse in evening since 4 months, painless weakness in all four limbs since 2 months that was gradual onset and progressive. He had difficulty in raising arms above the head, writing and getting up from the squatting position. He also had nasal voice since 3 months. All symptoms worsened over the day. No history of facial, neck and truncal muscle weakness, dysphagia, sensory symptoms or bladder and bowel involvement. No history of breathlessness. There was no significant family history. General physical examination was within normal limits with respiratory rate of 14 per minute. Single breath count was 25. Neurological examination revealed dysarthria, bilateral partial ptosis [Figure 1], total opthalmoplegia in both eyes, power of grade 4/5 in all 4 limbs at all joints, hypotonic in all 4 limbs, all deep tendon reflexes were absent and bilateral plantars flexors. Fundus examination was normal. Pupils size and reaction to light were normal. Gag reflex was present. Sensory system was normal. Rest of the systemic examination was unremarkable. Investigations revealed complete blood count, liver and renal function tests, serum electrolytes, thyroid function tests were normal. Serum acetylcholine receptor antibodies were 5.73 nM/l (Normal: 0.0-0.25 nM/l). Anti MuSK (muscle kinase) antibodies were negative. Chest X-ray, MRI brain, contrast-enhanced CT chest were normal. A muscle biopsy was nonspecific. Thymus gland scientigraphy was performed using 10 mCi Tertofosmin intravenous injection and spect imaging done at 40 min and 2 hrs post injection which was suggestive of moderate increased tracer uptake in thymus gland [Figure 2]. Electromyography revealed decremental response with repetitive nerve stimulation suggesting neuromuscular junction disease. On the basis of above findings, diagnosis of myasthenia gravis was made. The patient was put on pyridostigmine, steroids and azathioprine with marked improvement of extra ocular movements and muscle power. He is under consideration for thymectomy.

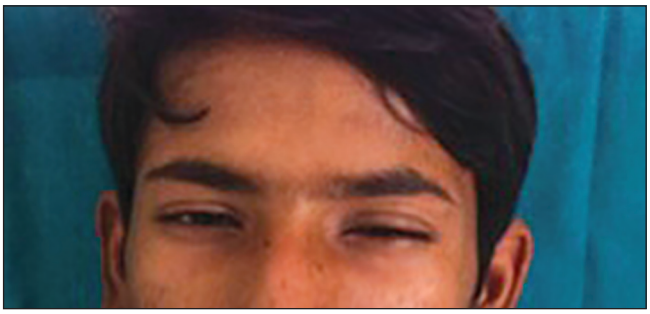

Figure 1: Bilateral partial ptosis

DISCUSSION

Autoimmune JMG is an uncommon disorder in the pediatric population, characterized by fatigable weakness due to antibody-mediated destruction of the AChR at the neuromuscular junction. Children typically present with ocular symptoms (ptosis, diplopia, ophthalmoplegia) but can also present with generalized weakness or bulbar symptoms (facial weakness, voice change, difficulty in chewing or swallowing). About 50-69% of JMG patients are seropositive (AChR antibodies), compared to 80% of adult patients. In both age groups, generalized MG has a higher seropositivity rate than pure ocular MG.[4] The differential diagnosis of JMG includes congenital myopathies, congenital myasthenic syndromes, toxins, hypothyroidism, mitochondrial myopathies, multiple sclerosis and brainstem tumors.[5] The diagnosis is based upon clinical signs and symptoms, with laboratory and electrophysiological studies used for confirmation. Although thymoma in children is rare, the thymus must be imaged (usually by CT) once JMG has been diagnosed. Most mediastinal tumors in the pediatric population are either neurogenic in origin (33%) or lymphomas (41%). Primary thymic lesions (such as thymic cysts, thymolipomas, and thymic hyperplasia) represent only 2.5% of mediastinal tumors, while thymomas comprise about 1%.[6] The Myasthenia Gravis Foundation of America (MGFA) clinical classification divides MG into five main classes and several subclasses, which is designed to identify subgroups of patients who share distinct clinical features or severity of disease that may indicate different prognoses or responses to therapy.[7]

MGFA clinical classification

Class I

Any ocular muscle weakness.
May have weakness of eye closure.
All other muscle strength is normal.

Class II

Mild weakness affecting other than ocular muscles.
May also have ocular muscle weakness of any severity.

IIa: Predominantly affecting limb, axial muscles, or both.

May also have lesser involvement of oropharyngeal muscles.

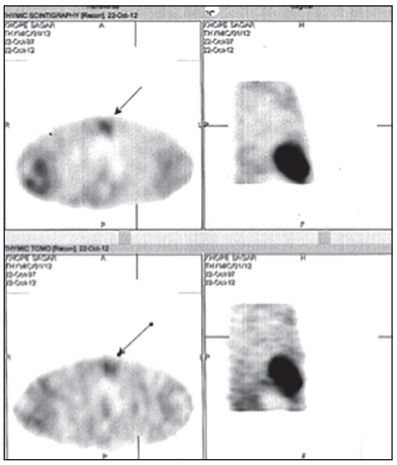

Figure 2: Thymus gland scientigraphy: Spect tomographic images in different views reveal physiological tracer concentration in myocardium and moderate increased tracer concentration in retrosternal region

IIb: Predominantly affecting oropharyngeal, respiratory muscles, or both.

May also have lesser or equal involvement of limb, axial muscles, or both.

Class III

Moderate weakness affecting other than ocular muscles.
May also have ocular muscle weakness of any severity.

IIIa: Predominantly affecting limb, axial muscles, or both.

May also have lesser involvement of oropharyngeal muscles.

IIIb: Predominantly affecting oropharyngeal, respiratory muscles, or both.

May also have lesser or equal involvement of limb, axial muscles, or both.

Class IV

Severe weakness affecting other than ocular muscles.
May also have ocular muscle weakness of any severity.

IVa: Predominantly affecting limb and or axial muscles.
May also have lesser involvement of oropharyngeal muscles.

IVb: Predominantly affecting oropharyngeal, respiratory muscles, or both.

May also have lesser or equal involvement of limb, axial muscles, or both.

Class V

Defined by intubation, with or without mechanical ventilation, except when employed during routine postoperative management. The use of feeding tube without intubation places the patient in class IVb.

This patient fits into class IIa MGFA classification.

Treatment consists of anticholinesterase drugs like pyridostigmine (30-90 mg every 6 hourly) to be given first with oral corticosteroids

(prednisone 15-20 mg/day). When long-term immunosuppression is necessary, azathioprine is recommended to allow tapering the steroids to the lowest possible dose whilst maintaining azathioprine. Cyclosporine A, mycophenolate mofetil and cyclophosphamide are used for severe cases. Plasma exchange is recommended in severe cases to induce remission and in preparation for surgery. Intravenous immune globulin and plasma exchange are effective for the treatment of MG exacerbations. For patients with non-thymomatous MG, thymectomy is recommended as an option to increase the probability of remission or improvement. It is considered an appropriate procedure for many patients with generalized MG between puberty and 55 years of age. If possible thymectomy should be postponed until puberty because of the importance of the gland in the development of the immune system, but JMG is also quite responsive. The remission rate after thymectomy is approximately 35% provided it is done in the first year or two after the onset of the disease and another 50% will improve to some extent.[8] Once thymoma is diagnosed, thymectomy is indicated irrespective of MG severity. The course of the illness is extremely variable. The long-term outlook for children with myasthenia is better than it is for adults.

REFERENCES

1. Nikolic DM, Nikolic AV, Lavrnic DV, Medjo BP, Ivanovski PI. Childhood onset myasthenia gravis with thymoma. Paediatr Neurol 2012;46:329-31.
2. Evoli A. Acquired myasthenia gravis in childhood. Curr Opin Neurol 2010;23:536-40.
3. Heckmann JM, Hansen P, Van Toorn R, Lubbe E, Janse van Rensburg E, Wilmshurst JM. The characteristics of juvenile myasthenia gravis among South Africans. S Afr Med J 2012;102:532-6.
4. Chiang LM, Darras BT, Kang PB. Juvenile myasthenia gravis. Mus Nerve 2009;39:423-431.
5. Finnis MF, Jayawant S. Juvenile myasthenia gravis: A paediatric pespective. Autoimm Dis 2011;2011: 404101.
6. Shetty PK, Balaiah K, Nayak G, Shetty PK. Thymoma with myasthenia gravis in adolescent. Online J Health Allied Sci 2010;9:16.
7. Jaretzki A 3rd, Barohn RJ, Ernstoff RM, Kaminski HJ, Keesey JC, Penn AS, *et al*. Myasthenia gravis: Recommendations for clinical research standards. Ann Thoracic Surg 2000;70:327-34.
8. Ropper AH, Samuels MA. Myasthenia gravis and related disorders of the neuromuscular junction: In Adam's and Victor's Principles of Neurology. 9th ed. USA 2009. p.1414.

How to cite this article: Kalyan M, Kanitkar SA, Gaikwad AN. Juvenile myasthenia gravis. Int J Med Public Health 2014;4:520-2.

Source of Support: Nil, **Conflict of Interest:** None declared.