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CASE REPORT

An Ocular Myasthenia Gravis: A Case Report

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Abstract

Myasthenia gravis (MG) is an autoimmune disease involving the postsynaptic receptors in the neuromuscular junction, characterized by weakness of the muscles. Ocular myasthenia gravis is a subtype of disease where weakness of oculomotor muscles usually occurs with the presence of ptosis. MG is considered a rare disease in pediatric age groups. Acetylcholinesterase inhibitors and immune-modifying medications are usually the mainstays of medication. We report here, a case of a 2-year-old that presented with ptosis and was diagnosed as ocular myasthenia gravis. Our case report describes the clinical presentation, diagnostic tests, and treatments followed.

Keywords: Myasthenia gravis, Ocular myasthenia gravis, Pediatric myasthenia gravis, Ptosis

Introduction

Myasthenia gravis (MG) is an autoimmune disorder that affects the acetylcholine receptors (AChR) and decreases the number of receptors in the neuromuscular junction.

Patients with ocular myasthenia gravis usually suffer from weakness in the eyelid elevator muscle, extraocular muscle, or orbicular muscle.[1]

In the pediatric age group though rare, there are three different types of myasthenia gravis: congenital, transient neonatal and juvenile myasthenia gravis, which is the most common type.

Early diagnosis is very important, especially in the pediatric age group. One-half to two thirds of these age groups are not easily diagnosed in the first year during the initial stages of symptoms.[2]

Case report

The clinical case we report here is of a 2-year-old Pakistani female, who presented to the Primary Health Clinic (PHC) accompanied by her father. She had a history of sudden onset of left eye ptosis with a drooped head for 4 months, which progressed to total ptosis for 1 week (Figure 1).

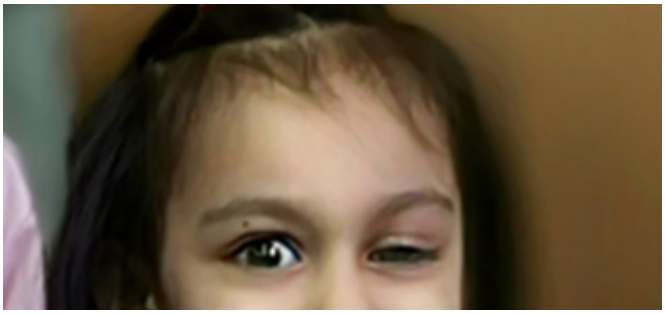

Figure 1: Image showing the presence of left eye ptosis

This was her first visit to the PHC, she was developmentally normal, and her immunization history was up to date with no history of previous hospital admission or surgery. Moreover, there was no significant family history of any autoimmune disease. On examination, there was total ptosis of the left eye, and the differential diagnosis was: myasthenia gravis, Duane syndrome, Horner syndrome, and third cranial nerve palsy.

Thereafter, the patient was then referred from the PHC to the ophthalmology department. Examination revealed that there was severe complete ptosis with poor levator function of eye movement (down and up), restricted elevation in the left eye, and restricted adduction and abduction movements. The cornea was clear. A rest test was done, and the patient was to sleep for 30 minutes in a dark room. After this they examined her again and ptosis was decreased indicating a positive test. The impression was that she had ocular myasthenia gravis, and the case was discussed with the pediatric neurologist. A neurological examination was conducted, and the patient was confirmed to present with left eye ptosis and anormal pupil. However, other cranial nerves examination was normal.

Diagnostic tests were obtained to confirm diagnosis. Magnetic Resonance Imaging (MRI) of the brain was found to be normal. The anti-acetylcholine receptors antibodies test was done and was found to be positive. Additionally, a test was done for muscle-specific kinase (MUSK) but was negative. Relatively, the patient's diagnosis confirmed of having ocular myasthenia gravis, she was started on pyridostigmine 7.5 mg every 4 hours per day. Unfortunately, she missed her follow-up appointments as she went back to her original country.

Discussion

Myasthenia gravis is a relatively rare autoimmune disease that affects the muscles due to a decrease in the Ach receptor numbers causing fatigability and weakness of the muscle.

About 40-70% of ocular myasthenia gravis patients will have antibodies that will destroy the Ach receptors leading to reduce transmission in the neuromuscular junction.

The most commonly affected muscle is the extraocular muscle, as it has a less number of ACH receptors.[1]

There are three types of MG in children:

1. Congenital transient myasthenia gravis: This occurs due to passage of the Ach antibodies from the pregnant women with myasthenia gravis to the fetus through the placenta. [1] Neonates will develop a weakness of respiratory muscle, apnea, weak crying at birth and pharyngeal and respiratory muscle weakness. They usually need respiratory support immediately after delivery. Hypotonia and extraocular muscle fatigability is also seen as a presentation.[1-3]
2. Transient neonatal myasthenia gravis: the etiology of this type is an abnormality in the neuromuscular junction either as pre- or post-synaptic structure abnormalities or functional abnormalities which cause an abnormality in Ach release. The symptoms are usually noticed at the age of 2 years.[1]
3. Juvenile myasthenia gravis: This is the commonest type. It occurs due to a blockage of Ach receptors affecting the age group between 0 to 19 years. However, it is more often seen between 2 to 5 years of age, as in our case. Sometimes it will occur after an infection. It is divided into ocular myasthenia gravis and systemic or generalized myasthenia gravis.[1]

Ocular myasthenia gravis is characterized by a weakness of the oculomotor muscle for 2 years duration without becoming generalized as in our case. [4] It can be present at any age, they are usually present with ptosis and sometimes present with diplopia. These symptoms become worse as the day progress. [3] On the other hand, systemic myasthenia gravis can affect any skeletal muscle of the body.[1]

Myasthenia gravis is less common in children than in adults.[5] The incidence in children is 3-9.1 cases per million per year.[1] Observed more commonly in girls than boys, it occurs in around 1.3 to 1 in the prepubertal age group and 1.8 to 1 peripubertally.[5]

Ocular myasthenia gravis is more common in Asian children, which is similar to our patient's ethnic group.[1] The genetic factors play an important role

in myasthenia gravis. Some studies have shown that 1-7.1% of patients with myasthenia gravis have a family history of the disease.[6]

There are many diagnostic tests such as the Anti-AchR antibodies test, which was done on our patient and found to be positive. However, sometimes in children, these Anti-AchR antibodies will not be present making it difficult to differentiate it from congenital myasthenia gravis. Up to 40% of children without AchR antibodies are found to have another antibody against another neuromuscular junction protein such as muscle-specific kinase (Musk) but in our patient it was negative.[4]

Electromyography (EMG) is considered, especially if the Anti-AchR antibodies test and (MUSK) test are negative.[3,4]

In addition, pharmacological investigation plays a role in the diagnosis of myasthenia gravis. For example, a tension test uses an edrophonium infusion, which causes a cholinergic effect by increasing the concentration of the neurotransmitter at the neuromuscular junction. The weakness of the muscle will be resolved and this can be recorded.[4] In addition, a neostigmine test can play a role in the diagnosis, and the pyridostigmine test which can be used to diagnose children less than 1 year.[1]

About 70% of patients with myasthenia gravis may have thymic hyperplasia. Thus, it is important to take a chest computed tomography (CT) scan to rule out a thymoma,[7] which is more common in Juvenile myasthenia gravis and is rare to occur in prepubertal children.[4]

The management of a child with myasthenia gravis should be delivered by a multidisciplinary team comprising of a pediatrician, pediatric neurologist, physiotherapist, psychologist, and speech therapist.[4] The acetylcholinesterase inhibitors can be used as the first line of treatment, such as the pyridostigmine, which had been used in our case. Immunosuppressive therapy also, can be effective such as corticosteroid or azathioprine which is a purine analog that suppresses the B and T cell proliferation. Some studies have shown that using a combination of corticosteroids with azathioprine helps in reducing the progression of ocular myasthenia gravis to systemic myasthenia gravis.[4]

Mycophenolate mofetil (MMF) is an inosine monophosphate dehydrogenase inhibitor that can also be used in AchR seropositive patients which includes treating children from the age of 11 years. It can also reduce the symptoms by blocking purine synthesis. In addition, Rituximab which is a monoclonal antibody medication can be used to treat Juvenile myasthenia gravis.[4] In the presence of thymoma, thymectomy should be carried out.[3]

Since December 2019 the COVID-19 global pandemic affected the world.[8]

Unfortunately, multisystemic inflammatory syndromes in children were rarely recognized by the WHO and CDC during the pandemic. Several cases of MG developed within weeks after COVID-19 infection. One of the possible etiological factors is that the virus enters the cell and increases the production of antibodies and perhaps it damages T-cell mediated immunity. The study showed that all patients tested positive for acetylcholine receptors antibodies.[9]

Children with myasthenia gravis are more prone to COVID-19 complications. It was found that less than 50% of children had severe respiratory infections using only non-invasive ventilation.[8]

Some medications used during COVID-19 management such as hydroxychloroquine may worsen the MG manifestations but, on the other hand, using steroids proved effective in reducing the mortality rate among those groups. Data suggests that patients with myasthenia gravis should continue their medications during the COVID-19 pandemic.[10] Also, the CDC recommends that patients with myasthenia gravis should be vaccinated with any FDA-authorized COVID-19 vaccines.[11]

Ocular myasthenia gravis is an autoimmune disease that is uncommon in the pediatric age group. Management is the same in all age groups, but in children, it can progress to generalized myasthenia gravis. Thus, close monitoring and follow up's are highly recommended.

Ethics and consent

Consent for publication of this case was signed by patient's father, including accompanying image of the patient.

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