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Paediatrics

UNILATERAL TRAUMATIC PTOSIS TURNED OUT TO BE JUVENILE OCULAR MYASTHENIA GRAVIS

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ABSTRACT Juvenile myasthenia gravis is a rare disorder with an incidence of 0.1 per one lakh of which ocular myasthenia gravis is reported in 10-35% cases. Juvenile myasthenia gravis is challenging, due to variations in serological testing, technical difficulty while performing electrophysiological testing, risk of high ocular morbidity when it overlaps with critical periods of vision development. Mainstay of treatment is acetyl cholinesterase inhibitors and immunosuppression. Once the underlying disease is stabilised, surgery can be considered if ptosis persists. Hence any clinical suspicion warrants early involvement of paediatric neurologist and an ophthalmologist. We report a rare case of 4 year old girl whose initial symptom of ocular myasthenia gravis, acute right ptosis with characteristic diurnal variation was manifested after local peri-orbital trauma. Repetitive nerve stimulation showed decremental response. She responded to pyridostigmine and ptosis improved gradually.

KEYWORDS : Juvenile ocular myasthenia gravis, traumatic ptosis, ocular morbidity, acetylcholinesterase inhibitors

INTRODUCTION:

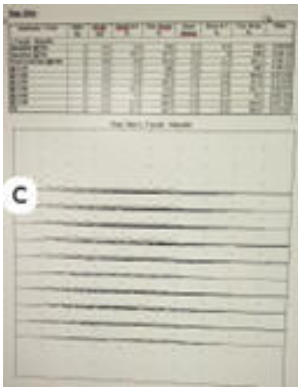
Juvenile myasthenia gravis is a rare disorder with an incidence of 0.1 per one lakh. It is an autoimmune disorder due to auto antibodies against neuromuscular junction proteins. Ocular myasthenia gravis is reported in 10-35% of Juvenile Myasthenia gravis cases[3]. Primarily it is a diagnosis made clinically by a classical pattern of fluctuating weakness and fatiguability. Juvenile Ocular Myasthenia gravis is challenging to manage due to variations in serological testing, technical difficulty while performing electrophysiological testing, risk of high ocular morbidity as it overlaps with critical periods of vision development. Since it mimics various ocular motility disorders and some of which are life threatening ones, MRI brain imaging is necessary. Currently, there is paucity of evidence based guidelines in the management of Juvenile myasthenia gravis. Hence the purpose of reporting this rare case is to emphasise the need for clinicians to be aware of this entity as it carries higher comorbidity.[1,3,4]

CASE REPORT:

We report a rare case of 4 year old girl who was referred by a local clinician. Her chief complaint was sudden onset right sided ptosis after an accidental trauma to peri-orbital area with jowar stick, 2 weeks prior to the visit. At presentation she had no signs of local injury, no diplopia, no restricted eye movements, did not have any generalised muscle weakness. But she reported diurnal variability in ptosis and ptosis got better after waking up from sleep or after naps every time. Ophthalmologist was consulted, visual acuity and pupillary examination were within normal limits. MRI brain was normal. Electrophysiological Studies, Repetitive nerve stimulation (RNS) showed decremental response. Serum antibody testing (Anti Ach and Anti Musk) were negative. CT chest ruled out thymoma. She was started on pyridostigmine at 5mg per kg body weight in 4 divided doses and was taught ocular exercises. Gradually, improvement was noticed in ptosis during followup.

A) Right sided ptosis at the initial Presentation

B) Improvement in ptosis after starting the treatment, during followup at 8 weeks



C) Decremental response noticed in RNS study

DISCUSSION :

Definitions : Myasthenia gravis is an autoimmune disorder where antibodies are directed at postsynaptic membranes of neuromuscular junction. If it occurs in < 19 yrs of age, it is called juvenile myasthenia gravis. If myasthenia gravis is restricted to ocular muscles without being generalised, it is called ocular myasthenia gravis.

Juvenile myasthenia gravis is a rare disorder of childhood. The distinguishing features of juvenile myasthenia gravis are presence of isolated ocular symptoms, low rates of antibody positivity, high chances of remission. There are other subtypes of childhood myasthenias and important one being congenital myasthenia syndrome without any autoimmune basis that needs to get differentiated. Congenital myasthenias usually present in infancy with positive family history. Genetic analysis and muscle biopsy help in the diagnosis. The other close differential diagnoses are mitochondrial

disorders, myopathies, Guillain Barre syndrome, Acute Disseminated Encephalomyelitis, brain stem tumours, botulism.[3,6]

Most frequent ocular manifestations are clinical features are fluctuating ptosis, ophthalmoplegia, diplopia, strabismus. These symptoms if persistent can cause amblyopia in children. They also develop generalised muscle weakness, fatiguability of limb and bulbar muscles. Hence they can present with dysphagia, dysphonia, proximal limb muscle weakness which is more pronounced through the day and improves with rest. So children have the risk of choking or aspiration or chest infections. If there is impairment of respiratory muscles, that leads to an emergency called myasthenic crisis. It requires ventilatory support, Plasmapheresis or Intravenous Immunoglobulin therapy.

Investigations : Antibodies that are to be tested are anti Ach and anti Musk antibodies. But in pediatric age group, they will have low affinity antibodies to receptors, which the standard assays cannot detect. Seronegative myasthenia gravis is more common. Electrophysiological testing with Repetitive nerve stimulation is valuable in suspected Juvenile myasthenia gravis. It characteristically shows a decremental response in amplitude of motor action potentials by >10% by fourth or fifth stimulation. Single fibre electromyography has 97% sensitivity, but technically more difficult in children[5].

Medical treatment : Though there is higher rate of spontaneous remission, there is need for early appropriate treatment to avoid long term physical and ocular morbidity. There is general lack of evidence based guidelines or consensus on ocular myasthenia gravis treatment, most of the randomised control trials are recruited from generalised myasthenia gravis patients. Initial first line therapy is with acetyl cholinesterase inhibitors (pyridostigmine) and oral steroids. If not responsive, Second line immunosuppression is with Azathioprine, Mycophenolate mofetil, cyclosporine, Tacrolimus, Rituximab. Pyridostigmine is more effective in alleviating ptosis than diplopia. Ocular myasthenia gravis may not respond to steroids due to underlying ultrastructural pathological muscle degeneration.[2,6]

Surgical treatment : Even after optimal medical treatment, if the ptosis is stable for at least 2 years, ptosis surgery is chosen based on levator function. Thymus has a crucial role in acetyl choline receptor antibody production. But therapeutic value of thymectomy in patients with ocular myasthenia gravis are still controversial due to lack of studies proving its benefit.[7]

CONCLUSION :

Ocular Myasthenia gravis is a focal form of Juvenile myasthenia gravis with its characteristic clinical, serological and treatment aspects. It is called a great masquerader as the clinical features mimic various other conditions. In young children, it is particularly important to distinguish and establish the diagnosis for correct treatment options and for prognostication. Once the underlying disease is medically managed and if ptosis still persists, surgery can be considered. Thus ocular JMG requires close involvement of ophthalmologist and pediatric neurologist to prevent complications like amblyopia leading to ocular morbidity.

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