

The Diagnostic Odyssey in Myasthenia Gravis

Patient #2: Diagnosis and management of MG in a young woman 24-year-old-female

Case contributor and commentary:

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Age 23-24

Initial symptoms:

- » Intermittent facial drooping that was greater on right vs left side. Drooping worsened in the evening or when eating or laughing
- » Family said she had “lost her smile”



PCP conclusion:

- » Diagnosed with Bell’s palsy

2 months later

Symptom progression:

- » Intermittent slurred speech that was worse in the evening
- » Difficulty swallowing both solids and liquids, with nasal regurgitation of liquids and cough after eating, especially later in the day



Management:

- » Began eating only soft foods

3 months later

Patient presented to the neuromuscular clinic for evaluation of slurred speech and loss of facial expression:

Diagnostic evaluation

Reported symptoms:

- » Episodic shortness of breath for 2 weeks prior to visit, which was worse with exertion and lasted a couple of days to a week
- » Intermittent blurred vision for the prior month
- » No drooping eyelids, double vision or limb weakness

Motor exam:

- » Severe weakness of neck flexion
- » Mild weakness of neck extension, bilateral shoulder abduction, elbow flexion and extension, hip flexion and knee extension
- » No muscle atrophy

Neurological exam:

Cranial nerves

- » Mild bilateral ptosis on forward gaze, worse with upgaze
- » Severe weakness of eyelid closure bilaterally
- » Weakness of cheek puff, tongue protrusion and jaw closure

Deep tendon reflexes:

- » Normal

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Electrodiagnostic testing

Repetitive nerve stimulation study of the nasalis and trapezius muscles showed a decrementing response



Confirmed the presence of abnormal neuromuscular transmission

Antibody testing

Elevated ACh receptor binding antibodies



Confirmed the diagnosis of MG

CT scan

No evidence of thymoma

Current treatment options for MG^{1,2} include:

Symptomatic Treatment:

1. Acetylcholinesterase inhibitors

Disease Modifying Treatments:

1. Corticosteroids (e.g. prednisone)
2. Non-steroid Immunosuppressants (e.g. mycophenolate mofetil and azathioprine)
3. Intravenous immunoglobulin (IVIG)
4. Plasma Exchange/Therapeutic Apheresis
5. Complement Inhibitors (e.g. eculizumab)
6. Thymectomy

Management based on MG diagnosis

Treatment recommendations:

- » Underwent a course of plasma exchange
- » After an extensive discussion about the risk of birth defects, she began prednisone followed by mycophenolate mofetil
- » Referred to thoracic surgery for thymectomy



Outcome:

- » Resolution of all symptoms except for lid ptosis and eyelid closure weakness

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Commentary: Common issues when treating young females with MG are:

- 1) Potential teratogenicity of some immune modulating drugs including mycophenolate mofetil
- 2) Potential value of thymectomy in improving the symptoms of MG and reducing the required immunosuppressant therapies

The benefit of thymectomy has been demonstrated in patients with seropositive MG¹⁻³

Guidelines and consensus statements recommend early thymectomy for:

- » Early-onset MG
- » MG in children
- » Patients with generalized MG who have anti-AChR antibodies and whose symptoms developed at the age of 50-65 years

Current evidence does not support thymectomy in patients with:

- » MG and anti-MuSK or anti-LRP4 antibodies
- » Ocular MG

Key learning points

- » Before starting immune modulating agents in young women of childbearing age, patients should be educated about the potential teratogenicity of these medications
- » Thymectomy should be considered in young women with MG unless medically contraindicated

References

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