CLINICAL VIGNETTE

Hemifacial Myokymia in the Setting of Multiple Sclerosis (MS) – An Under-Recognized Diagnosis and Treatment

Lauren Matsuno, Julian Landaw, MD, Miguel Lemus, MD and Carol Lee, MD

Case Description

A 37-year-old male with no significant past medical history presents to the emergency department (ED) for three weeks of left-sided facial twitching. The patient describes the twitching as a pulling sensation along the lower cheek and lower eyelid that keeps him awake at night. The twitching is constant but fluctuates in intensity throughout the day. He initially presented to the ED for the same problem 10 days prior and was prescribed a course of Acyclovir for a presumed diagnosis of Bell's palsy but noted no improvement of his symptoms. Subsequently, he saw his primary care physician who ordered an MRI brain with and without contrast, which showed abnormal enhancement of the left facial nerve and white matter hyperintensities in pattern concerning for multiple sclerosis (MS). The patient was advised to go to the ED for a neurology evaluation. His temperature was 97.8 F, pulse 72, respiratory rate 19, blood pressure 147/73 mmHg, and his oxygen saturation was 100%. His physical exam was remarkable for twitching movements of the left lower cheek and left lower eyelid. The rest of the neurological exam were normal.

At his initial presentation, the patient's laboratory test results were within normal limits. An MRI of the brain with and without contrast (Figure 1) showed multiple FLAIR hyperintensities in a pattern concerning for demyelinating disease (bilateral periventricular white matter, right pontine tegmentum, left lateral midbrain), as well as a 3 mm area of enhancement in the left internal auditory canal near cranial nerves VII/VIII. There was no evidence of cervical cord signal abnormality or abnormal enhancement. An MRI of the thoracic spine with and without contrast (Figure 2) showed small foci of signal hyperintensity at T11 and T12 concerning for demyelinating disease. Cerebral spinal fluid analysis showed elevated protein at 58 mg/dL.

The neurology service was consulted and it was decided to admit the patient for a course of intravenous methylprednisolone 1,000 mg daily for 3 days. The patient followed up in the neurology tele-health clinic two weeks after discharge. He reported continued twitching of the left side of his face and experienced no benefit to the prescribed steroids or antispasmodics. He was offered a trial of Baclofen, a suggested trial of Gabapentin, and a referral for Botox. For his multiple sclerosis, he agreed to initiate Ocrelizumab for his significant inflammatory disease after discussing the risks and benefits of various therapeutic options.

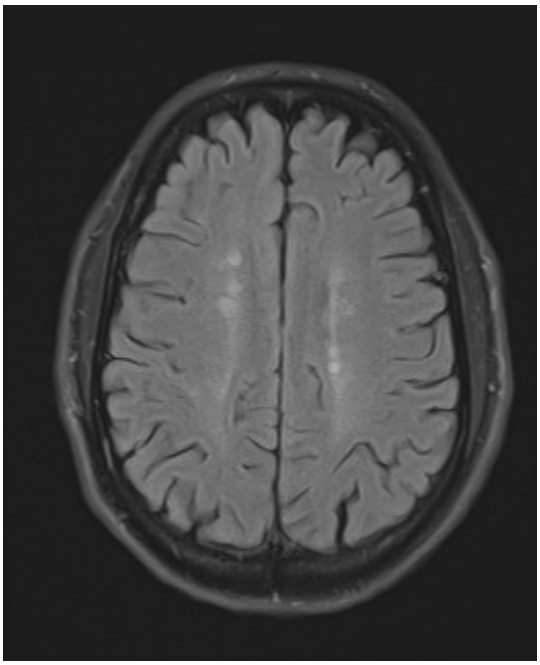

Figure 1: An MRI of the brain with and without contrast showed multiple FLAIR hyperintensities in a pattern concerning for demyelinating disease

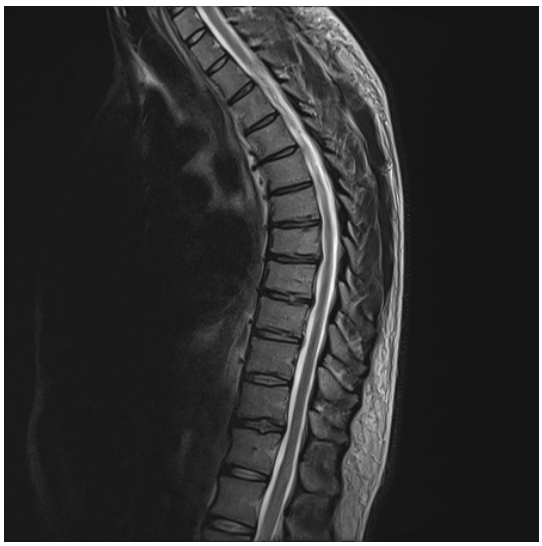

Figure 2: An MRI of the thoracic spine with and without contrast showed tiny foci of signal hyperintensity at T11 and T12 concerning demyelinating disease.

Discussion

Multiple sclerosis (MS) is a chronic autoimmune inflammatory demyelinating disease of the central nervous system.[1,2] The disease commonly afflicts young women in a 2:1 ratio compared to men and has onset between the ages of 20-50 years old. Individuals are typically of Northern European descent.[1] There are multiple forms of MS based on symptom presentation. The most common is the relapsing-remitting pattern, in which symptoms alternate between worsening episodes (relapse) and less severe episodes (remission).[2] The exact etiology of MS is unknown but the disease itself is related to the presence of inflammatory plaques causing demyelination and axonal degeneration leading to neurologic disability.[1] Common signs and symptoms include constitutional symptoms, muscle weakness, ataxia, sensory loss, spasticity, vision disturbances (e.g. acute optic neuritis), bladder and bowel incontinence, and cognitive dysfunction.[1-4] These symptoms depend on where the plaques are located in the brain and spinal cord. Other less common presenting symptoms include fatigue and other movement disorders.[2,4] Lastly and most pertinent, there have been several documented cases of facial myokymia and hemifacial spasms that were the presenting signs of MS.[5-7]

Hemifacial myokymia refers to the spontaneous, persistent, involuntary, vermicular, rhythmic, fine "twitching" movements of unilateral facial muscles beneath the skin.[1,5-10] More specifically, if the movements are intermittent, spastic, and non-sustained, then these movements are called hemifacial spasms. Hemifacial spasms classically originate unilaterally in the orbicularis oculi, and often involve other facial muscles innervated by the facial nerve (e.g. perioral muscles, platysma).[11] In contrast, if movements are sustained contractions, these are referred to as Spastic Paretic Hemifacial Contracture (SPHC).[1,5] SPHC also involves the orbicularis oculi muscle, especially of the lower eyelid, and can propagate across the rest of the face. Myokymia that is isolated to the eyelid alone is often seen in young healthy subjects. These cases are self-limiting to a few weeks, and are typically not associated with more serious disease.[9]

The most common primary cause of hemifacial myokymia is the vascular compression of the facial nerve at its exit/entry point into the posterior cranial fossa, which can increase the risk of demyelination injury.[5,9] This is particularly the case if the vessels are enlarged, aberrant and/or ectactic, most commonly the superior cerebellar, anterior inferior cerebellar or vertebral artery.[1,9] Secondary causes of hemifacial myokymia include brainstem demyelinating lesions such as multiple sclerosis. Other causes include other demyelinating disorders like acute inflammatory demyelinating polyradiculoneuropathy, brainstem neoplasms at the cerebellopontine angle, stroke, trauma, arteriovenous malformations, infections (cysticercosis), structural abnormalities, parotid tumors, and Bell's palsy.[1,5,9] Furthermore, transient facial myokymia may be due to benign causes such as excess caffeine intake, fatigue, anxiety, exercise, eye muscle fatigue, and magnesium deficiency.[1,9]

Among the cases of multiple sclerosis patients who present with facial myokymia and hemifacial spasms, initial imaging noted associated demyelinating lesions in the ipsilateral pontine tegmentum involving the postnuclear, postgenu portion of the facial nerve.[1,5,7,9,12] However, there are also numerous reports that found evidence of new demyelination lesions of the ipsilateral facial nerve itself at the time of the event on imaging.[5,6,9] It is important to note that lack of visualized lesions on imaging do not confirm that the lesions does not exist, as they may eventually develop or they may have resolved by the time of imaging.

Conclusion

This case highlights various clinical challenges and pitfalls in the diagnosis of MS with clinical symptoms of unilateral facial twitching, also known as hemifacial myokymia. Like other cases, our patient had initial conservative management or alternative diagnosis prior to further imaging that suggested MS. It is important to recognize that facial myokymia may be benign in presentation, but these movements may also be due to a concerning underlying condition, especially if symptoms persist for a long period of time. Other patients with known diagnosis of multiple sclerosis have had facial myokymia persisting for months, and hemifacial spasm persisting for up to 9 years, even with treatment.[5] Patients with multiple sclerosis may have other persistent manifestations including MS patients with spastic paretic hemifacial contracture with mild ipsilateral facial paresis.[13,14] Although rare, it is important to recognize that sustained symptoms of facial contracture or spasms may be a sign of MS.

REFERENCES

1. **Hertz R, Espinosa J, Lucerna A, Stranges D**. Multiple Sclerosis Presenting with Facial Twitching (Myokymia and Hemifacial Spasms). *Case Rep Neurol Med*. 2017;2017:7180560. doi: 10.1155/2017/7180560. Epub 2017 Sep 17. PMID: 29075542; PMCID: PMC5623781.
2. **Loma I, Heyman R**. Multiple sclerosis: pathogenesis and treatment. *Curr Neuropharmacol*. 2011 Sep;9(3):409-16. doi: 10.2174/157015911796557911. PMID: 22379455; PMCID: PMC3151595.
3. **Huang WJ, Chen WW, Zhang X**. Multiple sclerosis: Pathology, diagnosis and treatments. *Exp Ther Med*. 2017 Jun;13(6):3163-3166. doi: 10.3892/etm.2017.4410. Epub 2017 Apr 28. PMID: 28588671; PMCID: PMC5450788.
4. **Klocke S, Hahn N**. Multiple sclerosis. *Ment Health Clin*. 2019 Nov 27;9(6):349-358. doi: 10.9740/mhc.2019.11.349. PMID: 31857931; PMCID: PMC6881109.
5. **Marin Collazo IV, Tobin WO**. Facial Myokymia and Hemifacial Spasm in Multiple Sclerosis: A Descriptive Study on Clinical Features and Treatment Outcomes. *Neurologist*. 2018 Jan;23(1):1-6. doi: 10.1097/NRL.0000000000000163. PMID: 29266036.

6. **Cousins O, Harikrishnan S**. PO121 Continuous facial myokymia as the presenting symptom of multiple sclerosis. 2017;88:A43-A44. Available at: https://jnnp.bmj.com/ content/88/ Suppl_1/A43.4.
7. **London F, Hadhoum N, Zéphir H, Vermersch P, Outteryck O**. Continuous hemifacial myokymia as the revealing symptom of demyelinating disease of the CNS. *Mult Scler Relat Disord*. 2017 Jan;11:10-11. doi: 10.1016/j.msard.2016.11.001. Epub 2016 Nov 9. PMID: 28104248.
8. **Sedano MJ, Trejo JM, Macarrón JL, Polo JM, Berciano J, Calleja J**. Continuous facial myokymia in multiple sclerosis: treatment with botulinum toxin. *Eur Neurol*. 2000;43(3):137-40. doi: 10.1159/000008152. PMID: 10765052.
9. **Palasí A, Martínez Sánchez N, Bau L**. Mioquimias palpebrales unilaterales como forma de presentación de una esclerosis múltiple. *Neurología* (Barc., Ed. impr.). 2013:187-94. (Article in Portuguese.) Available at: https://pesquisa.bvsalud.org/portal/resource/pt/ibc-111650?lang=en.
10. **Andermann F, Cosgrove JB, Lloyd-Smith DL, Gloor P, Mcnaughton FL**. Facial myokymia in multiple sclerosis. *Brain*. 1961 Mar 1;84(1):31-44. doi.org/10.1093/brain/ 84.1.31.
11. **Chaudhry N, Srivastava A, Joshi L**. Hemifacial spasm: The past, present and future. *J Neurol Sci*. 2015 Sep 15;356(1-2):27-31. doi: 10.1016/j.jns.2015.06.032. Epub 2015 Jun 16. PMID: 26111430.
12. **Jacobs L, Kaba S, Pullicino P**. The lesion causing continuous facial myokymia in multiple sclerosis. *Arch Neurol*. 1994 Nov;51(11):1115-9. doi: 10.1001/archneur. 1994.00540230053012. PMID: 7980106.
13. **Fastré S, Hanson P, London F**. Spastic paretic hemifacial contracture related to multiple sclerosis: a rare and under-recognized entity. *Acta Neurol Belg*. 2017 Dec;117(4):927-929. doi: 10.1007/s13760-017-0817-4. Epub 2017 Jul 20. PMID: 28730530.
14. **Sarrigiannis P, Tsakanicas C, Anagnostouli M, Karandreas N**. Spastic paretic hemifacial contracture (SPHC) in a patient with multiple sclerosis. A clinical, EMG and neuroimaging study. *Neurophysiol Clin*. 2004 Oct;34(3-4):147-51. doi: 10.1016/j.neucli.2004.07.006. PMID: 15501684.