Clinical Reasoning: A 16-year-old girl with fixed unilateral grimace

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SECTION 1

A 16-year-old girl was referred from the epilepsy clinic for urgent admission to the long-term EEG monitoring unit. She had a prior history of both epileptic and nonepileptic seizures of multiple types, but presented to the clinic for evaluation of a new type of movement, present for 1 month. Over that time period her home antiseizure regimen (levetiracetam and topiramate) had been serially increased with no relief. She described (and demonstrated) a tonic spasm of the right lower face which had a sudden onset and lasted 30–90 seconds. In retrospect, her parents reported small facial twitches which preceded the full-blown spasms by 2 months. These spells

appeared to worsen with stress and attention. Short episodes were not painful, but as the episodes became more frequent she developed progressively worsening masseter pain. On the day of admission, she experienced these spasms every 5–10 minutes, more frequently during the examination. Between episodes, her neurologic examination had normal results, with no sensory or motor changes to the face. The video on the *Neurology*® Web site at www.neurology.org provides an example.

Questions for consideration:

1. What is your differential for this facial spasm?
2. How would you evaluate this movement?

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SECTION 2

At the top of our differential were true seizures and nonepileptic events, including pseudoseizures. Review of the first day's awake EEG supported our initial clinical suspicion; her facial spasms had no electrographic correlate. On the first morning following admission, we began a discussion with her mother regarding nonepileptic seizures. On that day, the patient had 2 nontraumatic falls which her

parents agreed were identical to her previously characterized nonepileptic events. Upon review of her overnight EEG (figure 1), however, we were forced to rethink her presentation and expand our differential.

Question for consideration:

1. What diagnoses would you now include in your differential, and what can be excluded?

Figure 1 A typical EEG during the facial spasm

Overnight EEG illustrating a typical facial spasm. Sleep spindles (arrow) can be observed in the first 3 seconds of the recording, indicating that this event occurred from stage II sleep.

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SECTION 3

The spasm captured in this segment of the patient's overnight EEG was identical to those seen during wakefulness, though they occurred less frequently and were clinically of lesser severity. The episodes were not associated with any particular phases of sleep and generally did not result in arousals. This tracing also demonstrates that the facial spasm erupted from stage II sleep, excluding the possibility that the events were psychogenic. This finding led us to expand our differential to include movement disorders. She had no sensations of building internal tension or compulsion and the movements were not suppressible, making tics unlikely. Her painful masseter involvement led us to consider hemimasticatory spasm. We discarded this diagnosis because her events did not result in forced jaw closure, a universal feature in hemimasticatory spasm. Leading our differential diagnosis at that time were oromandibular dystonia (OMD) and hemifacial spasm (HFS).

Involuntary spasm of the pharyngeal, masticatory, or lingual muscles may result in deviation, closure, or opening of the jaw¹; this cluster of dystonic movements is referred to as OMD. Patients often experience spread of the dystonic spasm to the muscles of the eyelids, nose, neck, or vocal cords, producing blepharospasm, bruxism, trismus, risus sardonicus, dysphagia, dysarthria, or conceivably any other combination of abnormal orofacial movements. These

are often painful. As with many other dystonias, certain activities or facial positions can elicit the dystonic movements, and idiosyncratic sensory tricks may relieve them. Our patient had no clear inciting factors and suffered only embarrassment at the episodes, no discomfort or dysfunction. The typical age at onset in OMD is 40 to 70 years, though inherited cases (usually associated with DYT1 dystonia) may present in childhood or adolescence.² Of relevance to our patient, OMD may occur from sleep; this feature is most common in secondary cases caused by drug exposure.

HFS is a series of unilateral facial movements which most commonly begin in the orbicularis oculi and spread over months to years to involve the muscles of the ipsilateral lower face.³ Many patients experience provocation or exacerbation in certain situations (driving, conversation) or following particular stimuli (laughing, emotion, and social stressors). HFS is classically characterized by brief segmental myoclonus, not the near-tetanic rictus seen in our patient. Likewise, our patient's spasms involved the masseter, risorius, and zygomaticus major from onset, without progressive spread. The typical case of HFS is thought to be due to compression of the peripheral portion of the seventh cranial nerve, most commonly by ectatic vessels shortly after the nerve exits the inferior pons. These vessels are often too small to visualize with MRI or CT. Irritation of the facial nerve nucleus can also cause HFS, though less commonly. HFS typically begins in the fifth or sixth decade; rare presentations in early life are most commonly associated with multiple sclerosis.^{4,5} Given this possible structural cause for her spasms, we ordered a nonsedated MRI. Simultaneously we started a medication empirically for her putative movement disorder; following a single dose of this medication, her spasms decreased in frequency by 90%, and were completely absent by the following day. Serial MRIs (figure 2) suggested the eventual diagnosis, performed 90 minutes following treatment initiation and 2 days after starting therapy.

This focal area of focal swelling and restricted diffusion suggests a limited differential diagnosis.

Question for consideration:

1. What is your next diagnostic step?

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Figure 2 MRI before and after treatment

Diffusion-weighted MRI from (A) a day with multiple facial spasms per hour and (B) 2 days later, following resolution of the abnormal movements. Arrowheads in (A) indicate the area of greatest edema and restricted diffusion. The DWI abnormality in the same area of cortex has nearly resolved in B.

SECTION 4

The patient's MRI demonstrated a focal area of restricted diffusion in the contralateral motor cortex. There was no evidence of abnormal enhancement on postcontrast images at this site or elsewhere within the brain parenchyma. Further, there were no ectatic vessels or focal lesions in the brainstem, and no other evidence of an inflammatory or demyelinating lesion was seen. The differential diagnosis for an area of highly restricted focal area of edema and restricted diffusion, without perfusion abnormalities, mass lesion, or magnetic resonance angiography evidence of infarct, includes postictal swelling, focal encephalitis, and neoplasm. CSF examination revealed a normal cell count, differential, and protein. Cytologic examination was also normal, with no malignant cells.

We had chosen to treat her putative movement disorder with phenytoin due to its potency and efficacy in a variety of paroxysmal movement disorders.⁶ Carbamazepine might also have been effective but lacks a readily available IV preparation. Her immediate and complete response to phenytoin, coupled with the MRI localization to the area of her motor cortex controlling the face, argued that these movements were in fact seizures. We repeated her MRI 2 days after starting phenytoin, with interval decrease in events from >50 per day to complete cessation, and noted that the area of prior restricted diffusion had resolved almost completely, although the gyrus appeared slightly swollen relative to the surrounding cortex. This tiny strip of cortex, measuring 3.2 cm³, was substantially below the 22 cm³ required to detect an abnormality using 19-electrode scalp EEG⁷ (improves to 6 cm³ with 128-electrode EEG). This corresponds to the better-known estimate that 6 cm² of cortical surface is required to generate a surface EEG potential,^{8,9} recently revised upward to >10 cm².

Thus, despite the unusual appearance of her movements, our distraction with her past history of nonepileptic events, the lack of EEG correlate, and the absence of any postictal phenomena, we arrived at her final diagnosis: epilepsy partialis continua affecting only the facial portion of the motor strip. She remains free of these facial spasms on low-dose phenytoin. This case serves as a reminder that clinically robust seizures can arise from quite restricted

areas of cortex, too small to be detected by scalp EEG recordings.

AUTHOR CONTRIBUTIONS

Jeff Waugh, MD/PhD: author of the primary manuscript, prepared figures. Sanjay Prabhu, MBBS: prepared figures, editor of manuscript. Blaise Bourgeois, MD: editor of manuscript. Sanjeev Kothare, MD: editor of manuscript.

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DISCLOSURE

Dr. Waugh serves on the editorial board of the *Journal of Pediatric Biochemistry*. Dr. Prabhu reports no disclosures. Dr. Bourgeois serves on the editorial board of *Epilepsy Research*; holds a patent on and receives royalties for a technology that allows patient-specific early seizure detection based on EEG recording in patients with epileptic seizures; receives publishing royalties for *Pediatric Epilepsy, 3rd edition* (Demos, 2008); and receives research support from Lundbeck Inc./Ovation. Dr. Kothare interprets video-EEGs and routine EEG in the Division of Clinical Neurophysiology at Children's Hospital Boston; and has received research support from Eisai Inc. and the NIH.

REFERENCES

1. Balasubramaniam R, Rasmussen J, Carlson L, Van Sickle J, Okeson J. Oromandibular dystonia revisited: a review and a unique case. *J Oral Maxillofac Surg* 2008;66:379–386.
2. Jankovic J. Etiology and differential diagnosis of blepharospasm and oromandibular dystonia. *Adv Neurol* 1988;49:103–116.
3. Jankovic J. Peripherally induced movement disorders. *Neurol Clin* 2009;27:821–832.
4. Telischi F, Grobman L, Sheremata W, Apple M, Ayyar R. Hemifacial spasm: occurrence in multiple sclerosis. *Arch Otolaryngol Head Neck Surg* 1991;117:554–556.
5. Nociti V, Bentivoglio A, Frisullo G, et al. Movement disorders in multiple sclerosis: causal or coincidental association? *Mult Scler* 2008;14:1284–1287.
6. Dressler D, Benecke R. Diagnosis and management of acute movement disorders. *J Neurol* 2005;252:1299–1306.
7. Ferree TC, Clay MT, Tucker DM. The spatial resolution of scalp EEG. *Neurocomputing* 2001;38–40:1209–1216.
8. Cooper R, Winter A, Crow H, Walter W. Comparison of subcortical, cortical and scalp activity using chronically indwelling electrodes in man. *Electroencephalogr Clin Neurophysiol* 1965;18:217–228.
9. Tao J, Ray A, Hawes-Ebersole S, Ebersole J. Intracranial EEG substrates of scalp EEG interictal spikes. *Epilepsia* 2005;46:669–676.

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