

# Erdheim–Chester Disease and Palatal Tremor

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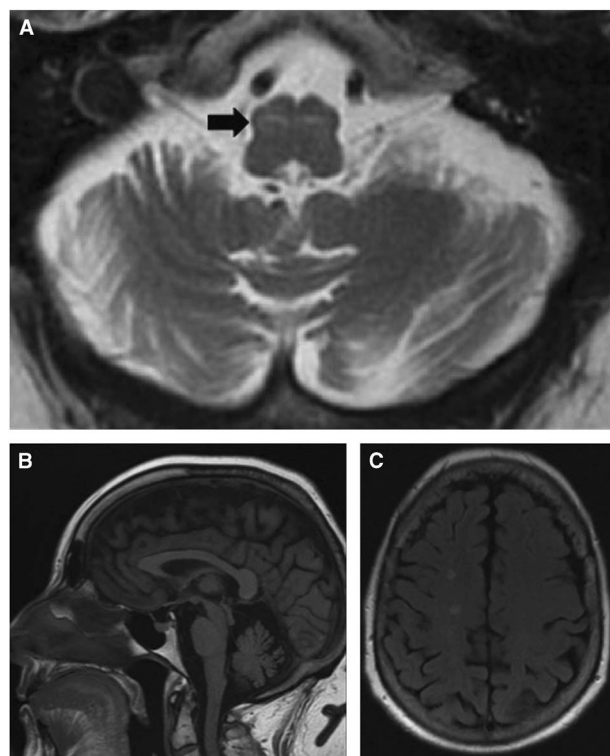
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A 53-year-old woman presented to neurology with progressive imbalance, dysphagia, and clicking of her ears over two years. Two years prior to onset of neurologic symptoms, she developed a right breast mass followed by additional soft tissue masses composed of foamy histiocytes with fibrosis, leading to a pathologically confirmed diagnosis of Erdheim–Chester disease (ECD), previously reported in detail.<sup>1</sup> Following pathology diagnosis, long-bone X-rays revealed typical diaphyseal and metaphyseal osteosclerosis.<sup>1</sup> She did not have diabetes insipidus. Peripheral blood BRAF<sup>V600E</sup> mutation was negative. Neurologic examination was remarkable only for palatal tremor (Video 1; see Supplementary Materials), symmetric limb ataxia, and ataxic gait. Magnetic resonance imaging of the brain demonstrated bilateral T2 hyperintensity of the inferior olivary nucleus (ION), moderate cerebellar atrophy, and several nonspecific T2 hyperintensities in the cerebral white matter without intracranial gadolinium enhancement (Figure 1).

ECD is a rare hematologic disorder of histiocytes with multifocal involvement, including skeletal (96%), retroperitoneal (68%), cardiac (64%), central nervous system (51%), orbital (25%), and hypophyseal (23%) manifestations.<sup>2</sup> Among ECD patients with neurologic involvement, about 40% experience symptoms of a cerebellar syndrome with such features as ataxia, nystagmus, and dysarthria.<sup>3</sup> In this cohort of 66 ECD patients with neurologic manifestations, 45% exhibited pyramidal signs, while 47% had diabetes insipidus.<sup>3</sup> Neurologic symptoms may arise from extra-axial meningeal mass lesions or intra-axial infiltration. In a cohort with brain imaging, 44% had infiltrative cerebral lesions primarily in the brainstem and cerebellum, including within the Guillain–Mollaret triangle.<sup>3</sup>

The Guillain–Mollaret triangle, or the dentato–rubro–olivary circuit, links the contralateral dentate nucleus with the ipsilateral red nucleus and the ION.<sup>4</sup> Disruption of the circuit can be clinically associated with palatal tremor, ataxia, and ocular manifestations.<sup>4</sup> Pathological causes include degenerative disorders (Alexander’s disease), infarction, demyelination, and trauma.<sup>5</sup> Following damage to the Guillain–Mollaret triangle, ION T2 hyperintensity appears at approximately 1 month followed by ION hypertrophy at 6 months, which persists for 3–4 years, though T2 hyperintensity may remain indefinitely.<sup>5</sup>

We suspect that histiocytic infiltration of the structures composing the Guillain–Mollaret triangle resulted in ION radiographic changes and palatal tremor. Despite frequent infiltration of the brainstem and cerebellum, palatal tremor has not previously been described in ECD.



**Figure 1:** Bilateral ION T2 hyperintensity (A, arrow) consistent with symptomatic palatal tremor associated with moderate cerebellar atrophy (B) and nonspecific cerebral white matter T2 hyperintensities (C).

## DISCLOSURES

Natalie E. Parks and R. Allan Purdy hereby declare that they have no conflicts of interest to disclose.

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**SUPPLEMENTARY MATERIAL**

To view the supplementary materials for this article, please visit <https://doi.org/10.1017/cjn.2017.259>.

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