Teaching NeuroImages: Brain and Skin Involvement in Erdheim-Chester Disease

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Figure 1 Brain MRI in Erdheim-Chester Disease

Axial T1-weighted postgadolinium imaging demonstrates punctate enhancing foci supratentorially (A, B, red arrows). Axial T2-fluid-attenuated inversion recovery (FLAIR) imaging demonstrates patchy pontine and middle cerebellar peduncle T2 hyperintensity (C, D). On repeat imaging 4 months after initiation of vemurafenib, punctate gadolinium enhancement has resolved (E), while pontine and middle cerebellar peduncle T2-FLAIR hyperintensity has nearly resolved (F).

A 61-year-old man developed progressive gait imbalance and papular skin lesions over 9 months. He reported fatigue, but no other constitutional symptoms. Neurologic examination revealed gait ataxia. Brain MRI (figure 1, A–D) demonstrated punctate enhancing foci supratentorially and patchy pontine T2 hyperintensity; no pituitary or orbital abnormality was reported. Skin biopsy (figure 2) revealed a histiocytic neoplasm that stained negative for CD1a, weakly positive for S100, and positive for CD163, factor 13A, and BRAF V600E, compatible with Erdheim-Chester disease.¹ Bone scan and body PET did not demonstrate osseous, cardiac, lung, or retroperitoneal involvement. Vemurafenib was prescribed for Erdheim-Chester disease with brain and skin involvement.² Brain involvement occurs in up to half of patients with Erdheim-Chester disease and most often manifests radiographically as pontine or cerebellar T2 hyperintensity, although multifocal punctate enhancement has also been reported.^{3,4} Skin involvement is observed in approximately one-quarter of patients and

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Figure 2 Lesional Skin Biopsy in Erdheim-Chester Disease

Biopsy of papular skin lesion (A, B) shows dense dermal infiltrate of histiocytes and multinucleated giant cells (C, 200× original magnification). By immunohistochemistry, histiocytic infiltrate is positive for BRAF V600E mutation (D, 200× original magnification).

lesions are most commonly xanthelasma-like, although nonfacial cutaneous xanthoma-like lesions as seen in our patient may also occur.⁵ On follow-up 4 months later, the patient's skin lesions had flattened, gadolinium enhancement had resolved, and pontine T2 hyperintensity had nearly resolved (figure 1, E and F), although gait imbalance persisted.

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Karen L. Rech, MD	Mayo Clinic, Rochester, MN	Interpreted the data, revised the manuscript for intellectual content
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