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...
lesions are most commonly xanthelasma-like, although nonfacial cutaneous xanthoma-like lesions as seen in our patient may also occur. 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.

**Study Funding**
No targeted funding reported.

**Disclosure**
Dr. Budhram, Dr. Rech, Dr. Peikert, Dr. Okuno, and Dr. Go report no disclosures. Dr. Dubey received research support from Center for Multiple Sclerosis and Autoimmune Neurology and Grifols Pharmaceuticals. Dr. Dubey has consulted for UCB Pharmaceuticals. All compensation for consulting activities is paid directly to Mayo Clinic. Dr. Tobin receives research funding from Mallinckrodt Pharmaceuticals and from the Mayo Clinic Center for MS and Autoimmune Neurology. Go to Neurology.org/N for full disclosures.
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Neurology 2021;96:e1590-e1592 Published Online before print November 9, 2020
DOI 10.1212/WNL.0000000000011159

This information is current as of November 9, 2020

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