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

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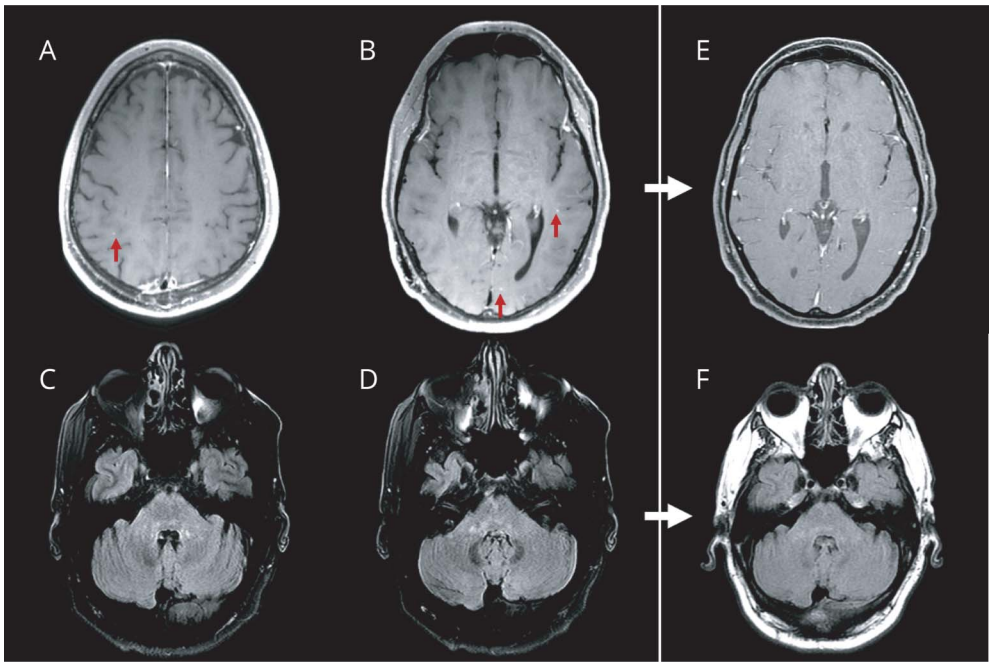
Neurology® 2021;96:e1590-e1592. doi:10.1212/WNL.0000000000011159

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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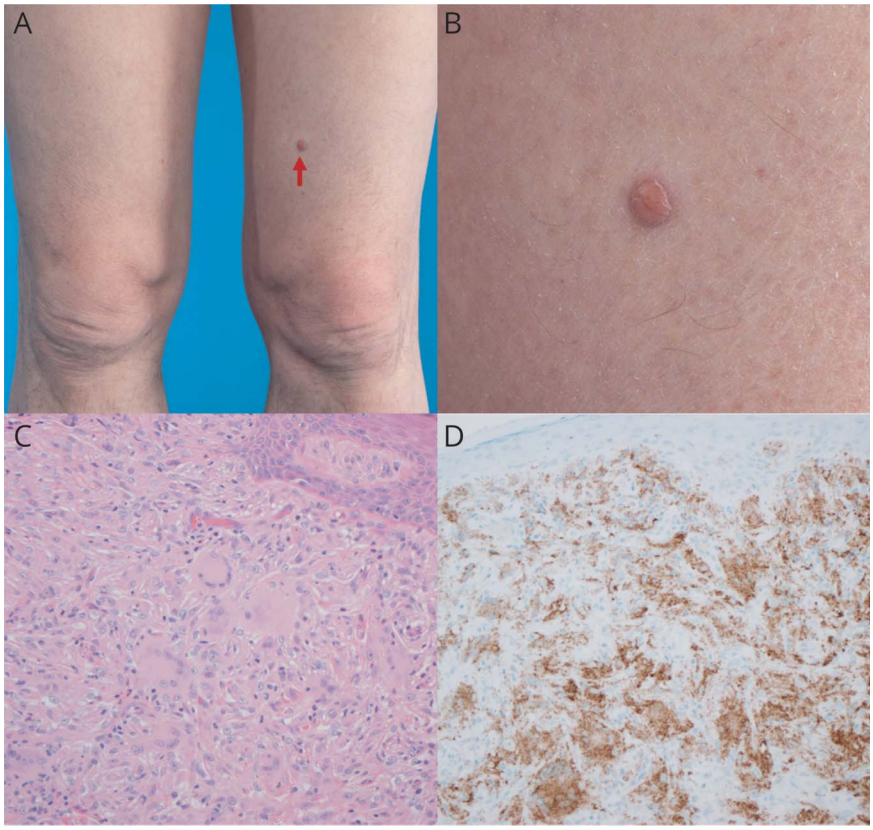
Figure 2 Lesional Skin Biopsy in Erdheim-Chester Disease



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.

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.

Appendix Authors

Name	Location	Contributions
Adrian Budhram, MD	Mayo Clinic, Rochester, MN	Drafted manuscript, analyzed and interpreted the data, composed the figures
Karen L. Rech, MD	Mayo Clinic, Rochester, MN	Interpreted the data, revised the manuscript for intellectual content
Johann M. Peikert, MD	Mayo Clinic Health System, Eau Claire, WI	Interpreted the data, revised the manuscript for intellectual content
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W. Oliver Tobin, MB BCh, BAO, PhD	Mayo Clinic, Rochester, MN	Designed and conceptualized study, analyzed and interpreted the data, revised the manuscript for intellectual content, study supervision

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Neurology 2021;96;e1590-e1592 Published Online before print November 9, 2020
DOI 10.1212/WNL.0000000000011159