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Erdheim-Chester disease presenting as bilateral proptosis

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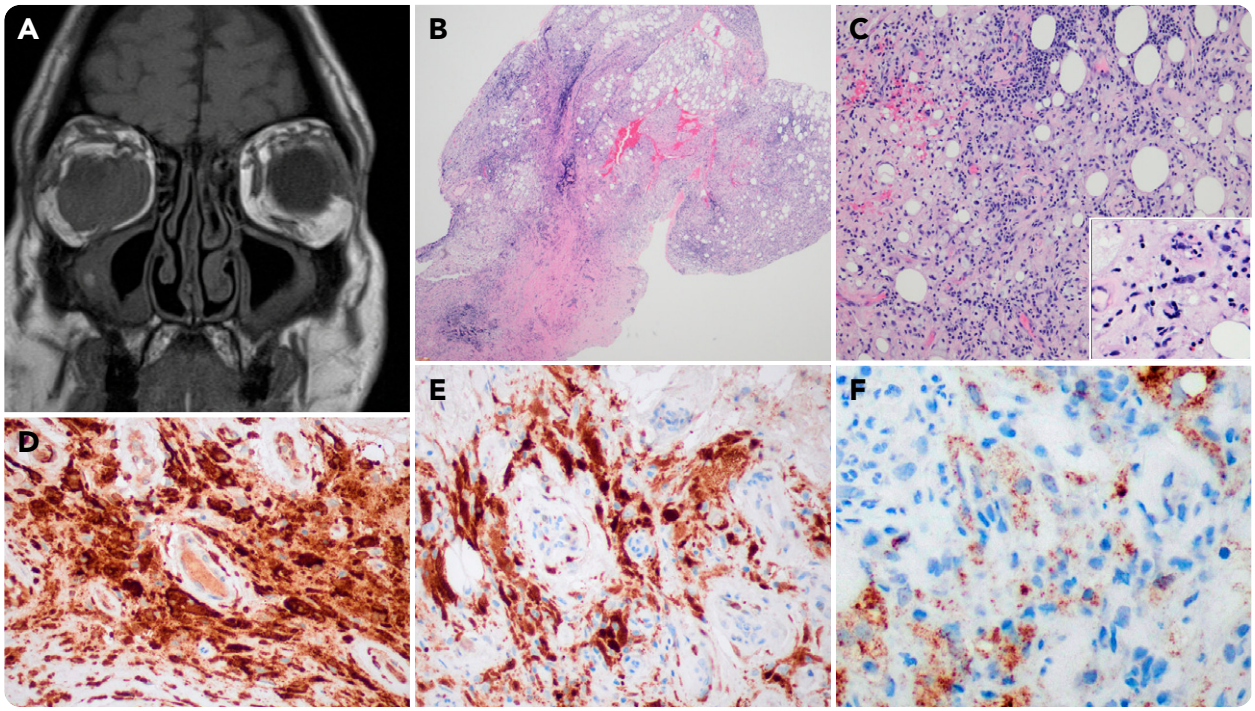

A 59-year-old man presented with upper eyelid lesions and bilateral proptosis. Magnetic resonance imaging of the head (panel A) revealed bilateral enhancing intraconal masses encasing the optic nerves. A right orbital biopsy showed a diffuse, histiocytic infiltrate involving fibroconnective tissue with scant inflammatory cells, including few eosinophils (panel B; hematoxylin-eosin [H&E]; original magnification ×4 objective; panel C; H&E, original magnification ×20 objective; inset; original magnification ×40 objective). The foamy, multivacuolated histiocytes were admixed with scattered Touton-type giant cells. By immunostaining, the histiocytes were positive for CD68, CD163 (panel D; H&E, original magnification ×40 objective), factor XIIIA (panel E; H&E, original magnification ×40 objective), BRAF V600E (panel F; H&E, original magnification ×40 objective), and negative for S100 and CD1a, supporting the diagnosis of Erdheim-Chester disease (ECD).

ECD is a multisystem, clonal non-Langerhans cell histiocytic neoplasm. ECD has myriad presentations, including bone pain, endocrinopathy, and neurological symptoms. Ocular involvement, seen in 25% of patients, is classically characterized by bilateral proptosis. A high degree of suspicion is required in assessing morphologically bland histiocytic lesions, where positivity for BRAF V600E and factor XIIIa coupled with negativity for CD1a and S100 supports a designation of ECD. Treatment options include interferon-α and *BRAF* inhibitors, like vemurafenib.

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