Erdheim-Chester Disease and Neuroendocrine Gastrointestinal Neoplasms

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Erdheim-Chester Disease and Neuroendocrine Gastrointestinal Neoplasms

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Erdheim-Chester disease (ECD) is a rare, non-Langerhans histiocytosis, mainly affecting adults ages 40 - 70. This clonal disorder is marked by recurrent BRAFV600E mutations in over 68% of patients and presents as uncontrolled inflammation of multiple organ systems. This pattern of cytokine expression and protein kinase signaling results in pro-inflammatory recruitment of histiocytes. Only 500 cases have been described in the literature with longstanding uncertainty about etiology and classification. While data supporting treatment is limited, IFN-α and pegylated IFN-α are considered first-line options with Kinase inhibitors demonstrating dramatic improvement in a small number of cases.

A 65 year-old female presented with new onset, transient neurologic symptoms of ataxia, headaches, right-sided weakness, and dysarthria. A brain MRI demonstrated multiple enhancing supratentorial and infratentorial lesions with calvarium involvement. Right temporal lobe and dural masses were also noted and biopsied. Pathology described foamy histiocytes with tissue demonstrating a BRAFV600E mutation, which is consistent with ECD. Having predominantly progressive CNS and osseous involvement rendered the patient eligible for a phase II study of Vemurafenib therapy. She tolerated the treatment with clinical improvement over one year. She then presented with melena, was found to have a clean-based antral ulceration on EGD, and was treated conservatively. Several months later, a PET scan revealed a new subepithelial duodenal mass with FDG avidity. Patient underwent an endoscopic ultrasound guided fine needle aspiration (Figure 1). Pathology revealed a low grade neuroendocrine neoplasm and is currently pending endoscopic removal (Figure 2).


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REFERENCES:


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