Erdheim-Chester Disease: An Unusual Case

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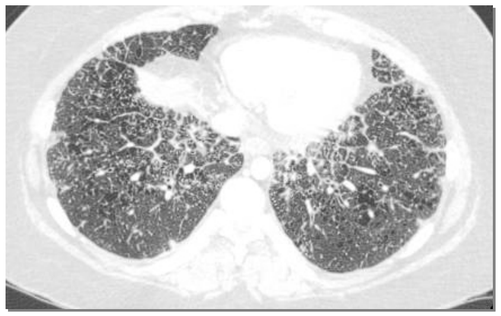
Case History

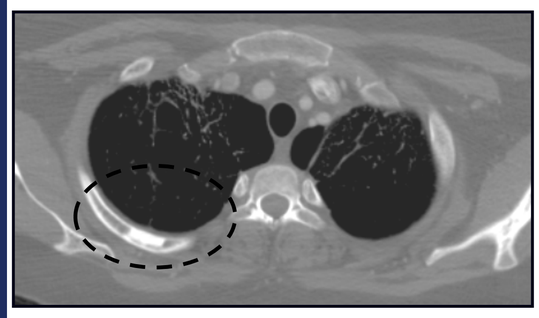
This 25-year-old woman had carried a diagnosis of diabetes insipidus since age five and had been treated with desmopressin since her teenage years.

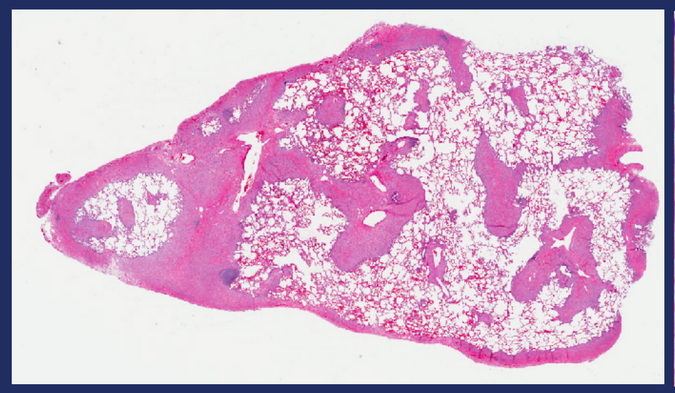
She presented three months prior to surgical lung biopsy with dyspnea on exertion which progressively worsened. She was noted to have bilateral pulmonary infiltrates and CT scan was showed diffuse pleural thickening, upper lobe bullous/emphysematous changes, marked lower lobe diffuse septal thickening, and coalescent linear parenchymal consolidation.

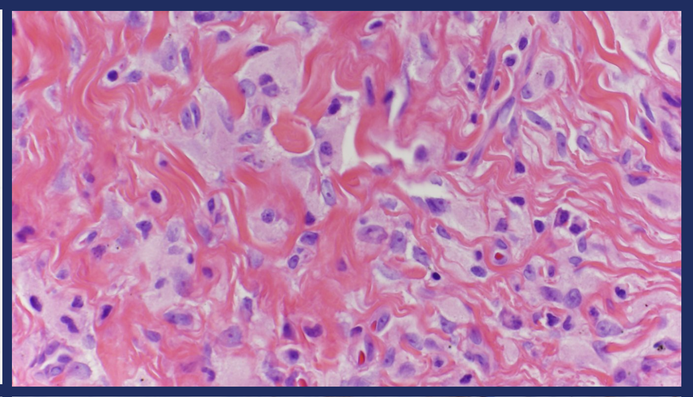


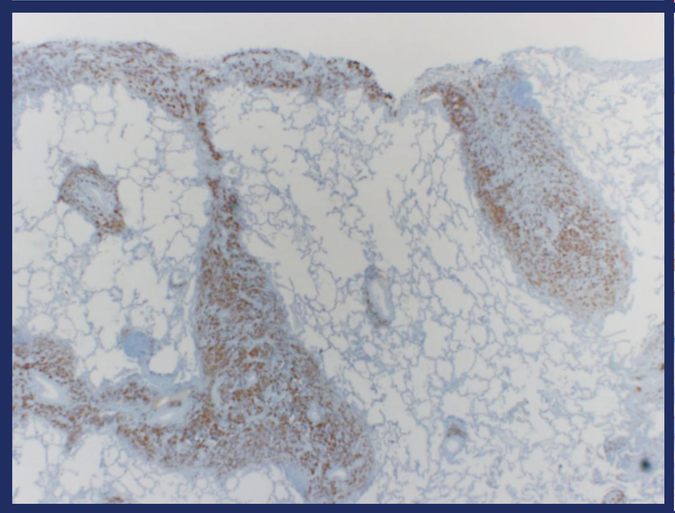
Surgical lung biopsy (center panels) showed a fibrosing process exquisitely limited to lymphatic routes in the pleura, septa and along bronchovascular bundles. The appearance was strongly suggestive of Erdheim-Chester disease.

The CT scan was re-reviewed and in retrospect a region of bony sclerosis was noted in one of the ribs and this finding was considered consistent with Erdheim-Chester disease.









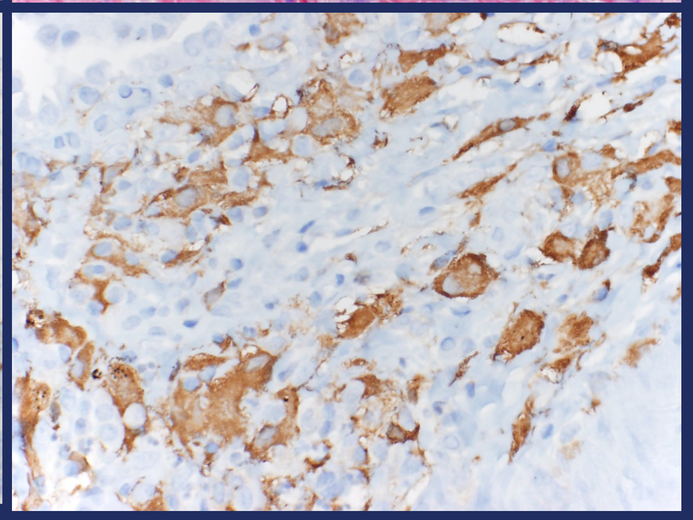


Figure Legends

Upper left: Scanning power view of the lung biopsy shows dense eosinphilic fibrosis outlining lymphatic routes in the pleura, septa, and along bronchovascular bundles. H&E

Upper right: High power view of the infiltrate in the fibrous tissue shows histiocytes with abundant pale cytoplasm in between the collagen fibers. H&E

Lower left: Scanning power view of Factor XIIIa staining highlights the positive histiocytes in the infiltrate as well as the distribution of the process.

Lower right: High power view of the Factor XIIIa positive histiocytes.

Additional findings on immunostains: CD68 positive, S-100 negative

Follow-up

- Three years post lung biopsy diagnosis the patient had persistent and progressive lung disease and was being evaluated for lung transplantation.

Discussion

This case of ECD has a number of unusual aspects:

1. Presentation in a young female
2. Symptoms of presumed CNS involvement with diabetes insipidus since age 5
3. Presentation with pulmonary symptoms
4. Radiologic confirmation of the diagnosis on the basis of bone windows on chest CT scan

Histologic findings on a lung biopsy are nearly unique to this condition and allow the diagnosis to be strongly suspected by the pathologist.

Factor XIIIa is not a specific stain for this condition but it is a useful support to the diagnosis and highlight the infiltrating cells.

References

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