Pathologic clues for the diagnosis of Erdheim Chester Disease (ECD)

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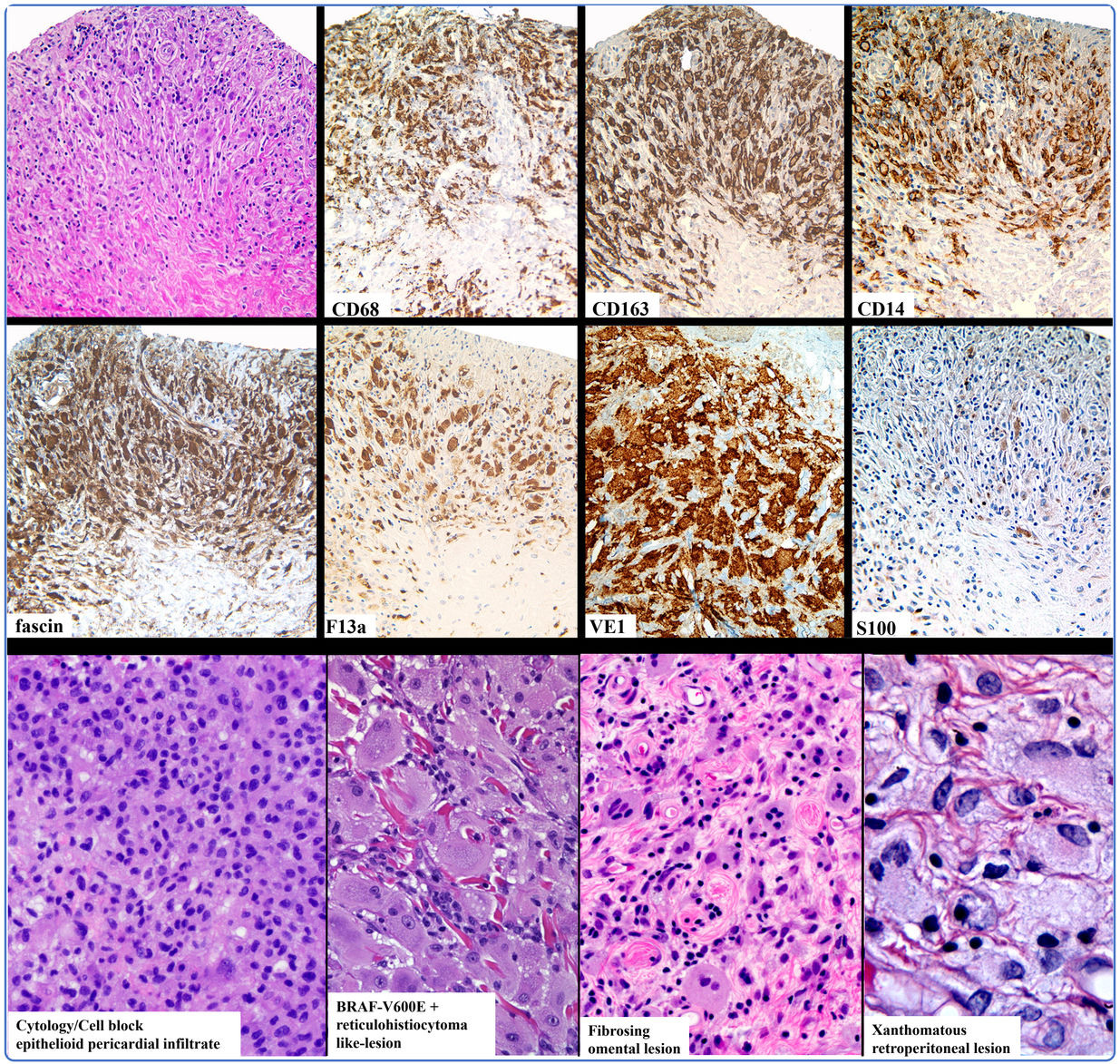
on behalf of the Erdheim Chester Disease (ECD) Global Alliance

What is ECD?

- A clonal proliferation of histiocytes that has a xanthogranuloma (XG) phenotype with a distinctive clinicoradiographic presentation.
- ECD now recognized as a distinct entity in the updated 2016/2017 WHO tumors of hematopoietic and lymphoid tissues.
- Accumulating data support ECD as a clonal inflammatory myeloid neoplasm, similar to LCH.
- Proposed "L" (Langerhans) grouping in the 2016 *Blood* revised classification of histiocytoses and neoplasms of the macrophage-dendritic cell linages.



Varied morphology:

Epithelioid, xanthomatous, or spindled histiocytes, +/- fibrosis

Immunophenotype of XG family:

- CD68 (granular cytoplasmic)
- CD163 (surface/cytoplasmic)
- CD14 (surface)
- Fascin and Factor XIIIa (cyto)
- VE1 (dark cytoplasmic granular) in *BRAF*-V600E mutated cases
- Little to negative S100
- Negative CD1a and Langerin

Ancillary molecular testing:

- Mutations in *BRAF*, *MAP2K1*, *NRAS*, *PIK3CA*, and gene fusions identified
- *BRAF* testing to include sensitive methods (<1% allele) for accurate detection, especially given the variable low content of clonal histiocytes in some cases.

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