

ERDHEIM-CHESTER DISEASE (ECD)

A rare histiocytic neoplasm

Radiology findings are vital to diagnose and guide life-saving treatments.



DEFINITION

- A rare histiocytic disorder with infiltrates throughout the body's organs and tissues
- A multi-system disease affecting virtually any combination of organ systems, including ophthalmic/periorbital, pulmonary, cardiovascular, renal, musculoskeletal, dermatologic, hypothalamic/pituitary, and central nervous system
- A rare disease with a critical need for prompt diagnosis to improve patient health outcomes
- Now classified as a blood cancer, diagnosed through clinical and radiologic findings, biopsy, and molecular (e.g., BRAF V600E and other mutation) testing

PROTEAN CLINICAL PRESENTATIONS

- Depends on organs involved
- Generalized symptoms of bone pain, fevers, night sweats, weight loss, fatigue, and/or weakness are often present and can be severe
- Findings may include diabetes insipidus, ataxia, dysarthria, diplopia, proptosis, dyspnea on exertion, xanthelasma, and renal failure
- Typical onset between 40 and 70 years of age, although documented cases in all age groups
- Slight preponderance of males

TYPICAL RADIOLOGY FINDINGS

- Bilateral cortical sclerosis of the long bones involving the diaphyseal regions
- Strong bilateral long bone uptake of radioactive tracer on 99mTc bone scintigraphs or PET scans
- Encasement of organs - "hairy kidney," "coated aorta," retroperitoneal fibrosis, right atrial mass, pericardial thickening or effusion

PATHOLOGY FINDINGS

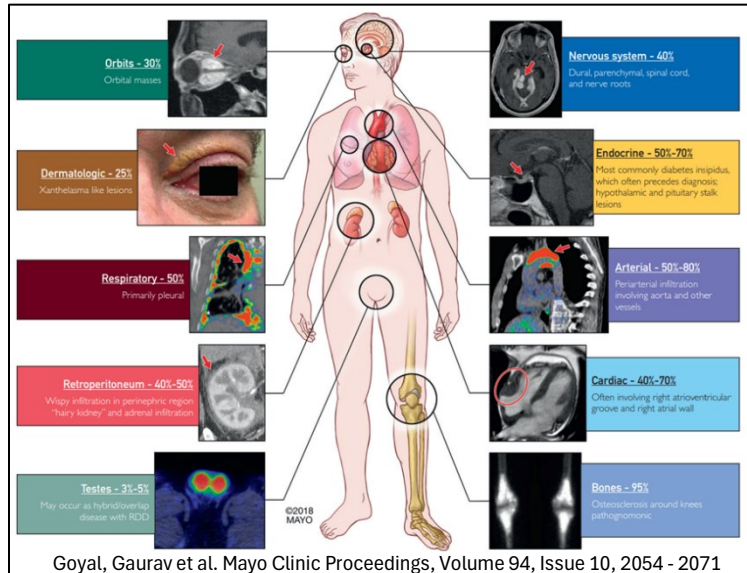
- Infiltration by foamy or lipid-laden, epithelioid or spindled histiocytes, with associated fibrosis and/or inflammatory background; foam cell finding not always present
- Touton giant cells may be present
- Immunohistochemistry: ECD histiocytes are XG family phenotype:

CD68+	Factor 13a+
Fascin+	CD163+
S-100+/-	CD1a-

- *BRAF*V600E mutations in >50% of patients
- Other MAPK pathway alterations (e.g. MAP2K1, KRAS), including kinase fusions, in nearly all patients without *BRAF*V600E mutation

KEY POINTS FOR RADIOLOGISTS

1. Diagnosis requires multidisciplinary input in which radiology is key, along with clinical, histopathological, and molecular findings.
2. Imaging is crucial to diagnosis, evaluating the extent of disease, identifying amenable biopsy targets, and evaluating treatment response.
3. Whole body FDG-PET/CT including distal extremities (vertex to toes) is the preferred diagnostic scan, as it allows for evaluation of metadiaphyseal osteosclerosis of the knees as well as other organ involvement.
4. Bilateral, symmetric metadiaphyseal osteosclerosis of the long bones of lower extremities is a pathognomonic finding.
5. “Hairy kidney” is a characteristic retroperitoneal finding due to diffuse bilateral infiltration leading to stellate pattern of perinephric soft tissue thickening.
6. “Coated aorta” is a characteristic vascular finding with circumferential periarterial soft tissue thickening.
7. Hypothalamic-pituitary axis is commonly involved and diabetes insipidus often precedes imaging findings on MR brain.
8. Cardiac involvement most often involves the right atrium and atrioventricular groove (pseudotumor).



Goyal, Gaurav et al. Mayo Clinic Proceedings, Volume 94, Issue 10, 2054 - 2071

Want to Learn More? Contact an ECD radiologist.

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