Erdheim-Chester disease

- Rare non-Langerhans cell, non-familial multisystemic histiocytosis, with widespread manifestations and of highly variable severity.
- Most common presenting symptom is bone pain.
- rare, non-inherited disease of middle age with a slight male predominance.
- Both Erdheim-Chester disease and LCH may coexist, and cases of double infiltration have been reported
- Leads to fibrosis and bone sclerosis

Erdheim-Chester Disease

- Rare lipidosis with the following features: *Age:* 50-70y
- Similar to Langerhans cell histiocytosis
- *Clinical:* Ranges from joint pain --> systemic involvement (heart, liver, spleen, pancreas, pericardium, lungs, adrenals, aorta, lymph nodes, bowel, orbit, and bone)
- *Histology:* Replacement of bone marrow by foamy lipid, histiocytes, and giant cells causing medullary fibrosis and osteosclerosis
- *X-ray:* Symmetric, appendicular skeleton >> axial skeleton Long bones invariably affected in the diaphysis and metaphysis with:
- - Patchy or diffuse increase in trabecular pattern
- - Medullary sclerosis
- - Cortical thickening

Clinical Issues

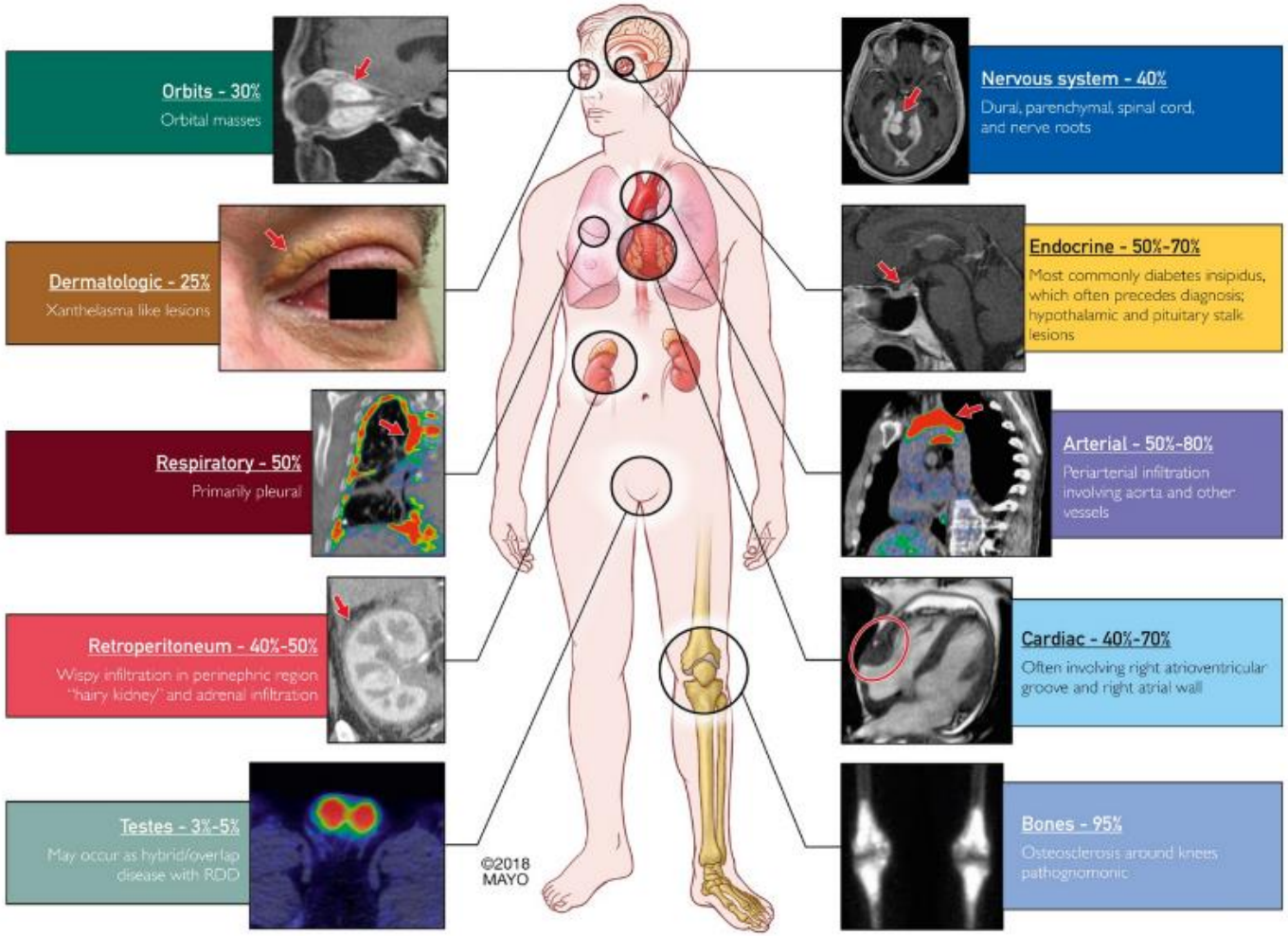
- Classic triad:
  - Bone pain
  - Exophthalmos
  - Diabetes insipidus
- Age range: 7-84 years; mean age: 53 years
- Chronic, progressive disease
- Prognosis worse with visceral involvement, especially cardiovascular and CNS

Extraskeletal manifestations in > 50%

- Hypothalamus-pituitary axis: diabetes insipidus (DI)
- Orbit: retroorbital soft tissue mass → exophthalmos
- Retroperitoneum: particularly perirenal
  - May lead to hydronephrosis
- Lung: dyspnea
- Heart/pericardium
- Liver/spleen: hepatosplenomegaly
- Skin: xanthomas

KEY FEATURES OF ERDHEIM-CHESTER DISEASE

The illustration depicts clinical and radiographic features with frequencies and descriptions.



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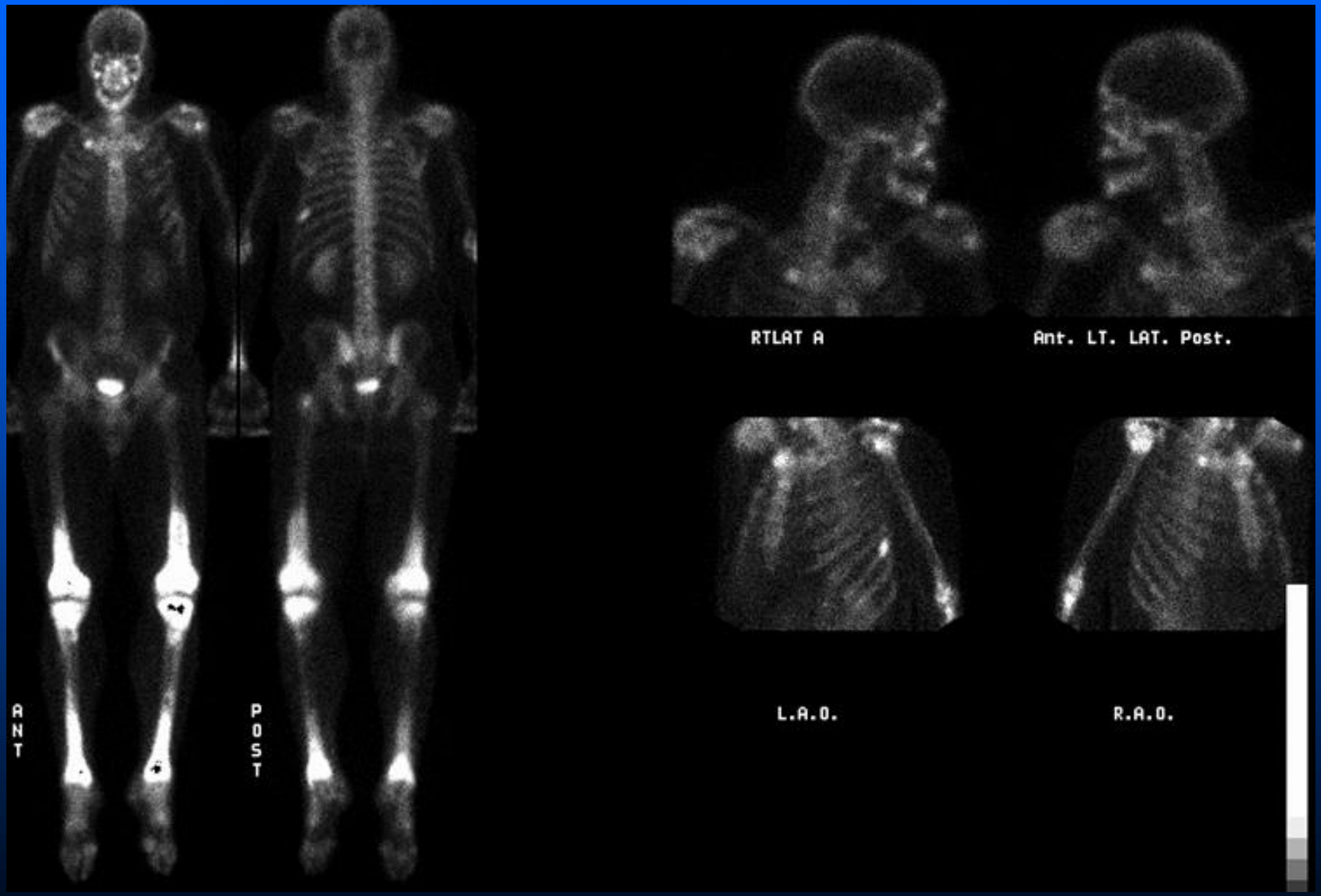
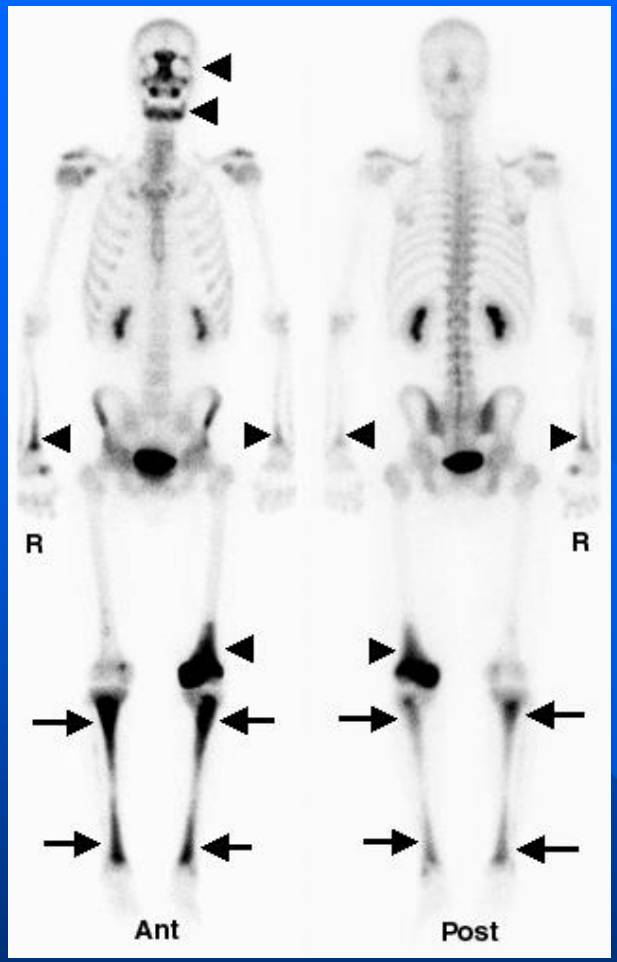
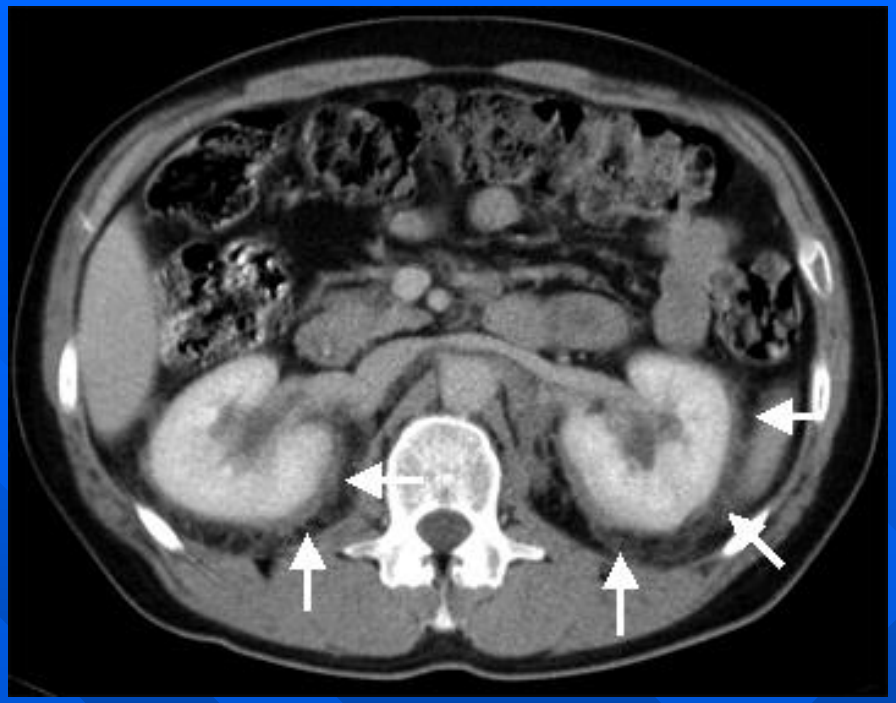
Erdheim-Chester Disease



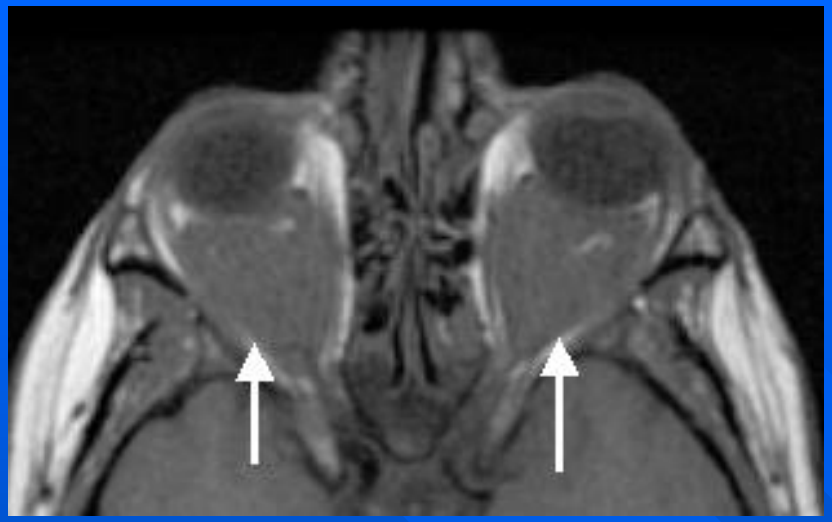

a.
b.
c.





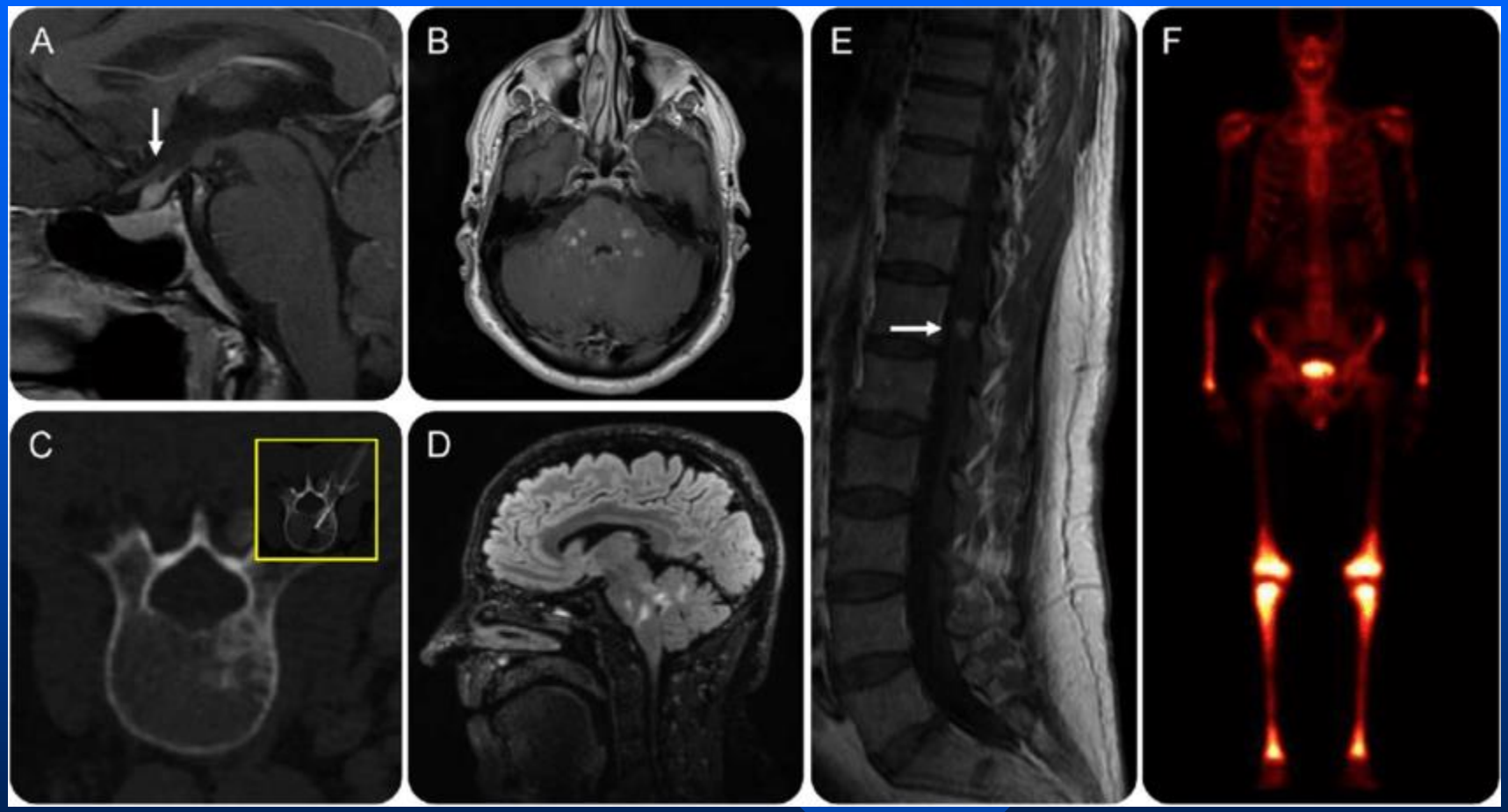
Typical findings on bone scintigraphy are bilateral symmetric uptake of bone seeking radiopharmaceutical within the metadiaphyses of the appendicular skeleton

Perirenal



Plain radiographs of the involved bones demonstrate ostesclerosis of the diaphyses and the metaphyses with sparing of the epiphyses. This pattern is considered to be

pathognomonic of ECD


A
B
C
D
E
F