A case of Erdheim-Chester Disease (ECD) with gastrointestinal involvement

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Background

Erdheim-Chester disease (ECD): a rare form of non-Langerhans cell histiocytosis derived from a monocyte-macrophage lineage.

Pathophysiology: Somatic mutations of BRAF or other components of the MAPK signaling pathway are present in more than 50% of patients with ECD [1]. Pathology shows histiocytic infiltration with sheets of foamy histiocytes on biopsy (Figure 1).

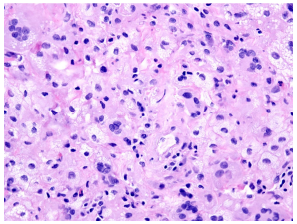

Figure 1: Soft tissue sample showing foamy (xanthomatous) histiocytes with surrounding fibrosis

Presentation: ECD is a multisystem disease that can encompass a wide spectrum of disorders.

Skeletal involvement, the most common clinical manifestation, occurs in the form of multifocal sclerotic lesions in the long bones of lower limbs (Figure 2).

Other common clinical features include diabetes insipidus, neurologic symptoms, constitutional symptoms, circumferential thickening of the aorta, and retroperitoneal fibrosis [2].

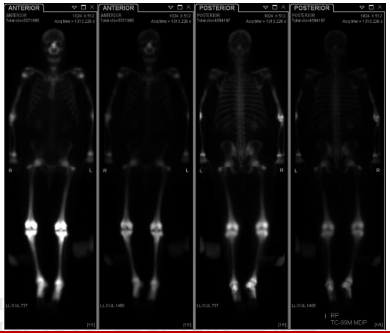

Figure 2: Bone scan showing diffuse increased uptake within osseous structures and periarticular surfaces

Case: 35 y.o. M with abdominal pain

PMH: retinitis pigmentosa, hypertension, diabetes insipidus, bilateral ureteral obstruction status post bilateral nephrostomy tubes, avascular necrosis of the left hip. ECD diagnosed 1 month prior to presentation.

HPI: 4-day history of nausea, vomiting, diarrhea, and abdominal pain. He denied flatus for several days.

PE: abdomen was soft, distended, and had mild tenderness to palpation. Patient was hypertensive to 152/91. Otherwise unremarkable.

Imaging: CT findings suggestive of small bowel obstruction and acute enteritis (Figures 3 & 4)

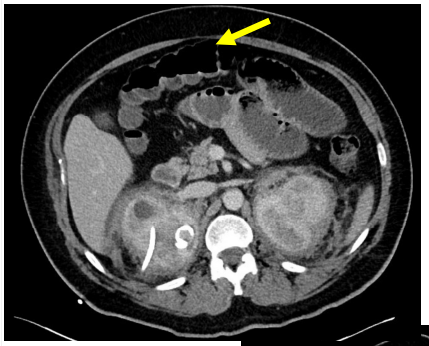

Figure 3: numerous dilated loops of proximal small bowel with air fluid levels

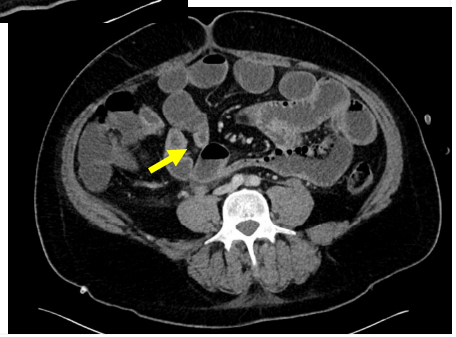

Figure 4: Enhancing wall thickening of the ileum

The patient was initially managed conservatively for small bowel obstruction with a nasogastric tube and bowel rest.

By the fourth day of hospital stay, the patient still reported minimal flatus.

He received a dose of prednisone and was taken to the OR that day for diagnostic laparoscopy, during which he was found to have no small bowel obstruction or adhesions. An inflamed jejunum and ileum were noted.

Post-operative course was uncomplicated; he was discharged the following day.

Conclusion

This case illustrates a classic presentation of ECD: an adult male with long bone involvement (avascular necrosis of the left hip) and diabetes insipidus.

Unique to this case was jejunal and ileal involvement with associated small bowel obstruction.

Prednisone is recommended for management of ECD, but the patient might have also benefited from INFa, which is the first-line treatment for ECD[2]. Furthermore, steroid treatment may be contraindicated in ECD patients with GI involvement [3].

While ECD can present in a variety of multisystemic manifestations, the most common presenting symptom is bone pain, especially of the lower limb [5].

References

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