Dermatology Practical & Conceptual

Erdheim Chester Disease: A Rare Entity from North India

Bhavya Swarnkar[1], Gouri Renuka Pushpanandan Anand[1], Somesh Gupta[1], Shipra Agarwal[2]

1 Department of Dermatology, Venereology and Leprosy, All India Institute of Medical Sciences, New Delhi, India
2 Department of Pathology, All India Institute of Medical Sciences, New Delhi, India

Key words: Erdheim Chester disease, non-Langerhans cell histiocytosis, setting sun, BRAF

Citation: Swarnkar B, Anand GRP, Gupta S, Agarwal S. Erdheim Chester disease: A rare entity from North India. *Dermatol Pract Concept.* 2023;13(3):e2023155. DOI: https://doi.org/10.5826/dpc.1303a155

Accepted: January 6, 2023; **Published:** July 2023

Funding: None.

Competing interests: None.

Authorship: All authors have contributed significantly to this publication.

Corresponding author: Dr Somesh Gupta, Department of Dermatology, All India Institute of Medical Sciences, Ansari Nagar East, New Delhi-110029 Phone: 9868899120 E-mail: someshgupta@hotmail.com

Patient consent was taken before sending the report for publication

Case Presentation

A man in his 40s presented with yellowish brown papuloplaques over bilateral eyelids for the past 3.5 years (Figure 1A). He complained of a mildly tender, skin-colored, cystic swelling over the left shin, diffuse pain over bilateral legs, polydipsia, and polyuria for the past 2 years.

Dermoscopy showed a 'setting sun' sign consisting of yellowish background with subtle erythema at the periphery. Increased pigment network and whitish areas were present. Branching linear vessels of variable thickness were also seen throughout the lesion (Figure 1B).

X-rays of the skull and extremities showed lytic lesions with sclerotic margin in bilateral femurs, tibia, (Figure 1C) fibula, radius, ulna, and skull (Figure 1D).

Skin biopsy showed features of xanthogranulomatous tissue response. Cells were immune-positive for CD68 but negative for S100 and CD1a. V600EBRAF mutation was positive.

Whole body PET scan showed increased FDG uptake over bilateral lower eyelids, hypermetabolic thickening involving the left renal pelvis extending to a calyceal system with left moderate and right mild hydronephrosis.

Serum osmolality was 292 mOsm/kg while urine osmolality was 93 mOsm/kg suggestive of diabetes insipidus. Based on history, examination, and investigations, we made the diagnosis of Erdheim Chester disease (ECD).

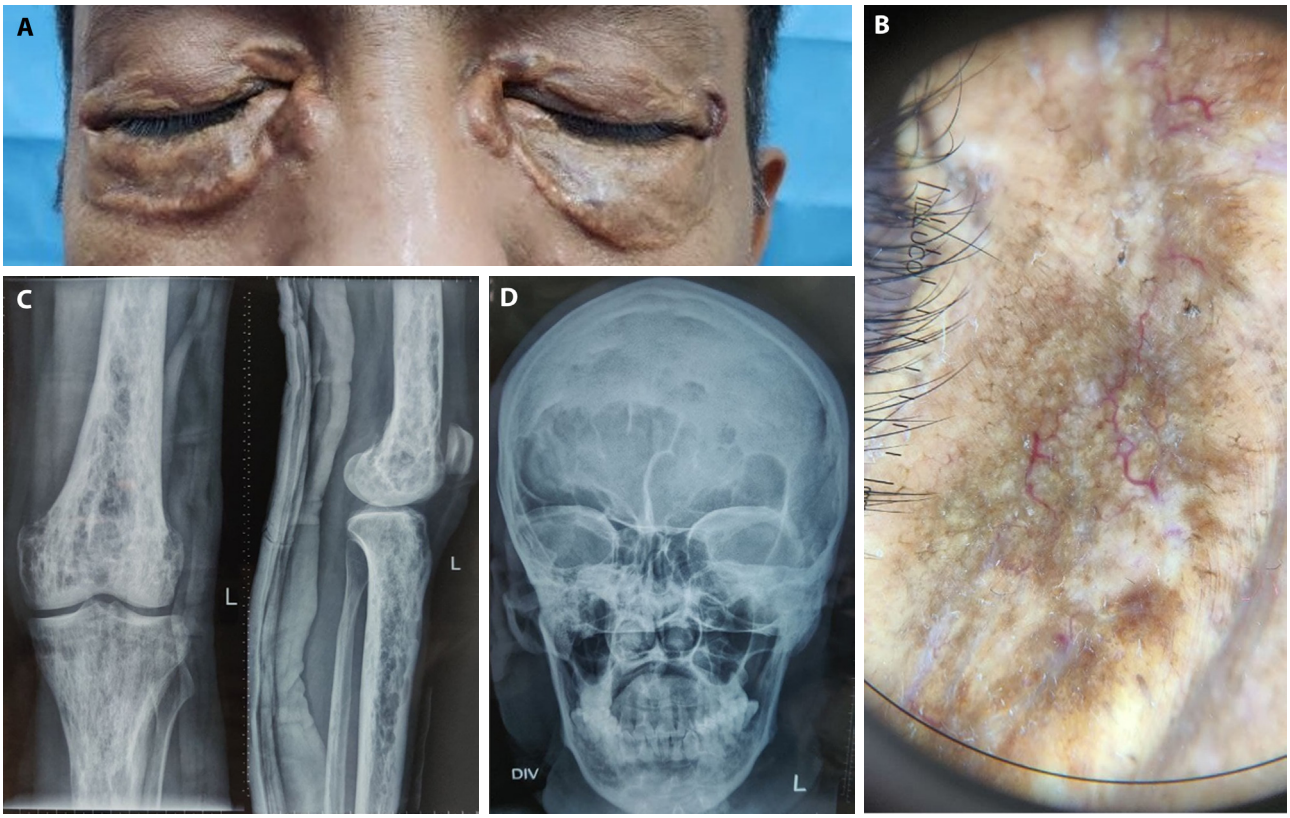

Figure 1. (A) Yellowish brown papuloplaques over bilateral eyelids. (B) 'Setting sun' sign, increased pigment network, whitish areas, and branching linear vessels (using hand held polarized dermatoscope IDS1100, ILLUCO, KOREA). (C) Lytic lesions with sclerotic margin in metaphyseal and diaphyseal region of femur and tibia. (D) Lytic lesions with sclerotic margin in skull.

Teaching Point

ECD should be ruled out in patients with periocular xanthelasma-like plaques with associated systemic symptoms like bone pain, and diabetes insipidus. Bony lytic lesions, as seen in our patient, have been reported uncommonly in around 5-30% of the cases of ECD [1,2]. We also described dermoscopy features.

References

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