Dermatology Practical & Conceptual

A Case of Xanthoma Disseminatum in a Progressive Form With Bladder Involvement Effectively Treated With 2-Chlorodeoxyadenosine

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Introduction

Xanthoma disseminatum (XD) is a rare non-Langerhans cell histiocytosis that is difficult to treat [1]. In rare progressive forms of the disease, organ dysfunctions may occur [2]. We report a case of XD with bladder involvement, with an excellent response to 2-chlorodeoxyadenosine.

Case Presentation

A 47-year-old male patient presented with a 5-year history of numerous skin lesions on the oral mucosa, head, neck, trunk, and upper limbs (Figure 1, A and C). His personal and family history was unremarkable. He denied any systemic symptoms. Dermatological examination demonstrated numerous symmetrical yellow to brown papules and nodules that enlarged into plaques and tumoral lesions, especially in the intertriginous areas. Laboratory tests were normal except for moderate leukocytosis and a slightly elevated erythrocyte sedimentation rate. Electrocardiography and chest X-ray were also normal.

Histopathologic examination revealed diffuse dermal infiltration by histiocytic cells, foamy cells, and Touton-type giant cells. Immunohistochemical studies showed positive staining for CD68, factor XIIIa, and fascin, and negative staining for S100 (Figure 2, A-C). The patient was diagnosed with XD based on clinicopathological and immunohistochemical findings.

Abdominal ultrasound examination for possible systemic involvement revealed grade 2 hydronephrosis in the right kidney and an irregular 18 mm echogenic mass behind the bladder that prevented the opening of the right ureter. The mass in the bladder was also confirmed by an abdominopelvic CT scan. Magnetic resonance imaging of the brain did not find any abnormality and serum protein electrophoresis did not show any M band.

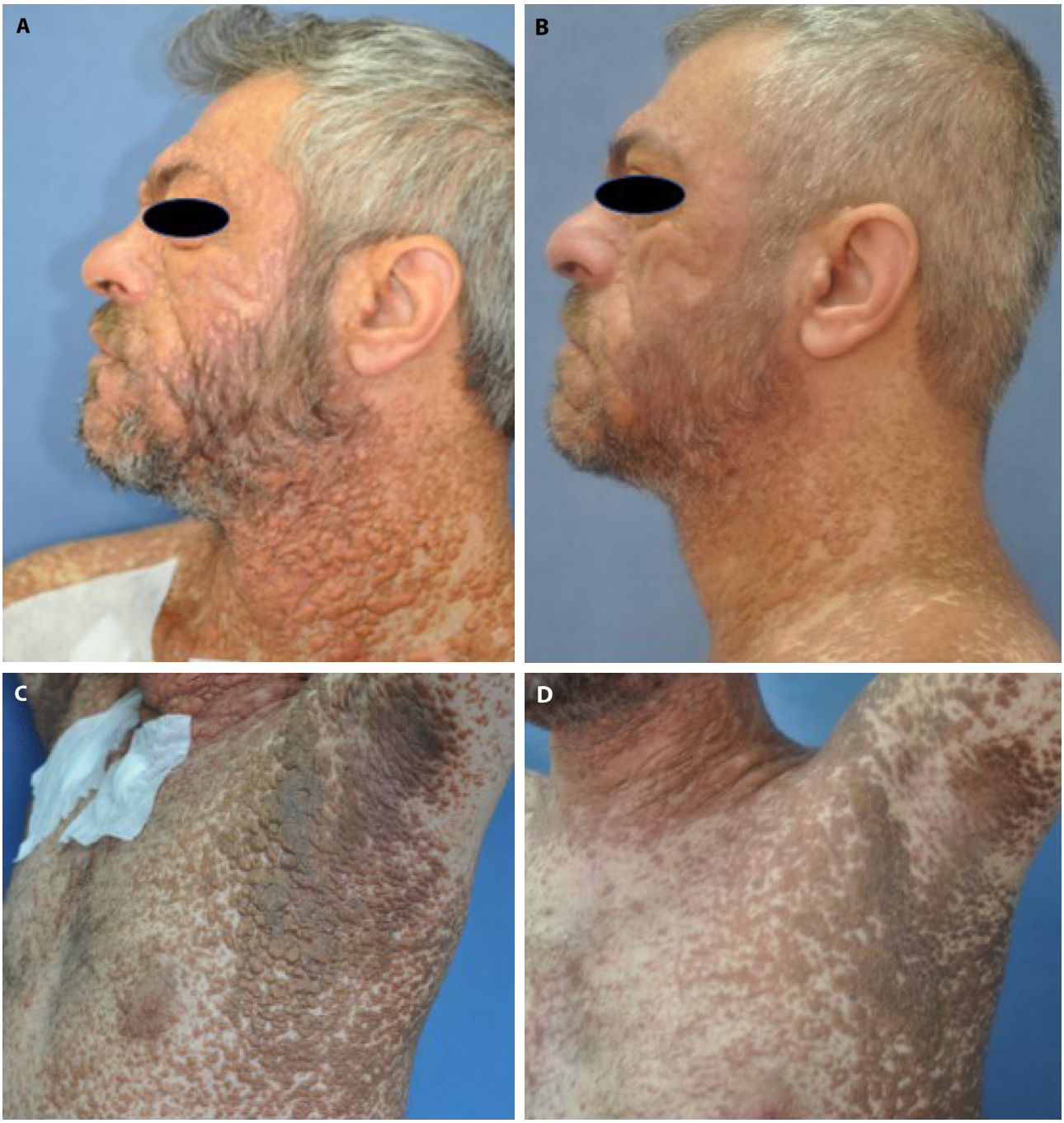

Figure 1. The appearance of lesions before (A,C) and after (B,D) treatment with 2-chlorodeoxyadenosine.

Histopathological and immunohistochemical findings of the specimen obtained through transurethral resection of the bladder were also consistent with XD (Figure 2, D and E).

We initiated 2-chlorodeoxyadenosine (cladribine) 0.14 mg/kg/d for five consecutive days, repeated monthly. The treatment was well tolerated, and no serious side effects developed. After three cycles of treatment, improvement in skin lesions was remarkably evident, with more flattening and fading, although a complete resolution was not achieved (Figure 1, B and D). Also, the tumoral lesion in the bladder and hydronephrosis also completely regressed in the control CT scan. No new lesions developed during our follow-up of 48 months.

Conclusions

Xanthoma disseminatum is characterized by erythematous, reddish-brown papules, plaques, and nodules that are typically symmetrical and tend to coalesce in intertriginous areas [1,2]. Histopathologic findings include diffuse dermal infiltration by histiocytes and Touton giant cells. Immunohistochemical evaluation shows histiocytes staining positive with CD68, CD163, factor XIIIa, and fascin, and negative with S-100 and CD1a (Langerin) [3 4].

Although XD is generally considered a benign disease, in extremely rare progressive forms, mechanical mucosal complications and progressive organ dysfunction can cause

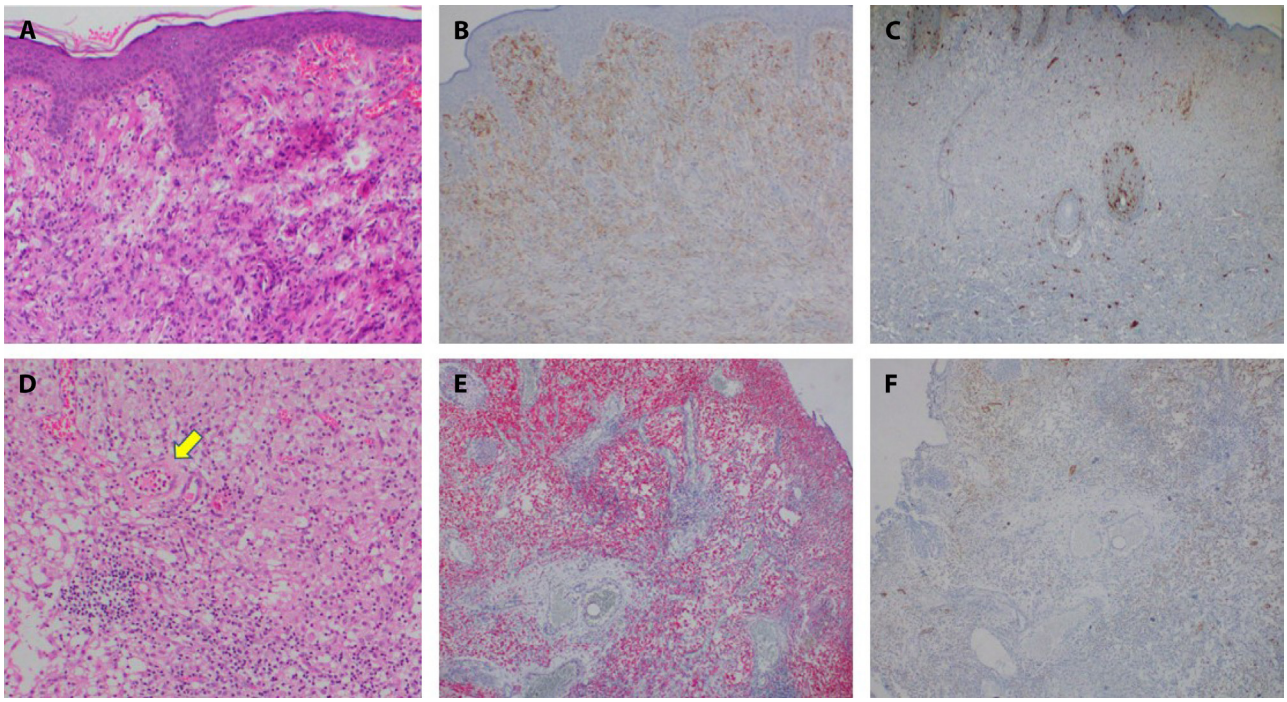

Figure 2. Histopathological appearance of skin and bladder biopsy specimens showing intense dermal infiltration of histiocytes and foamy cells (A and D, respectively) with Tauton giant cells (D, yellow arrow). H&E original magnifications ([(A] × 40; [D] × 100). Immunohistochemical examination, the cells in the skin and bladder stained positive for CD68 (B and D, respectively) and negative for CD1a (C and E, respectively). Original magnification x40 for B,C,E and F.

significant morbidity and mortality [2]. Many treatments including steroids, azathioprine, cyclophosphamide, electrocoagulation, or surgical excision, have been tried in XD, but recently the most satisfactory results have been obtained with 2-chlorodeoxyadenosine [5,6].

The excellent response to 2-chlorodeoxyadenosine was remarkable in this case with the progressive form of XD with bladder involvement. We emphasize the need for appropriate treatment with careful evaluation of the disease and close follow-up to prevent significant morbidity in progressive disease.

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