The Importance of Clinicopathological Correlation: A Case of Xanthoma Disseminatum

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ABSTRACT

Xanthoma disseminatum (XD) is a rare disease that presents with pruritic papules, classically involving the extensor surfaces and mucosal areas. We present a case of XD that was misdiagnosed as eruptive xanthomas and eventually self-resolved. Self-resolving XD may be within a spectrum of adult-onset juvenile xanthogranuloma. However, as there may be systemic consequences of XD, it is critical to ensure these patients obtain a complete evaluation and close follow-up.

INTRODUCTION

Xanthoma disseminatum (XD) is a very rare, non-familial non-Langerhans cell histiocytosis.¹-² Classically, XD presents as yellow-brown or red-brown papules on the face, trunk, and flexural areas.¹-⁵ Mucous membrane involvement may also occur in 40%-60% of cases.¹-⁵ This disease is more common in men (2.4:1 ratio) and younger patients, and laboratory findings usually show normolipidemic or slightly hyperlipidemic blood levels.¹-⁴ The diagnosis of XD is based on clinical suspicion, in combination with laboratory and pathology results. Here we describe a misdiagnosed case of XD, as an example to highlight the importance of clinicopathological correlation with xanthomatous eruptions.

CASE REPORT

A 70-year-old male presented with a four-month history of firm red pruritic papules on the face, neck, trunk, bilateral axillae, and bilateral thighs (Figures 1A, 1B, 1C and 1D). The patient reported no oral or genital lesions and took no chronic medications. The patient was otherwise healthy and asymptomatic. Previously, the patient was seen at an outside clinic where the diagnosis of eruptive xanthomas was made based on a right thigh biopsy consistent with xanthomas.

The clinical findings did not show typical eruptive xanthomas or extensor involvement. Laboratory testing showed mildly elevated cholesterol 291 (normal: <200) and low-density lipoprotein 223 (normal: <100); beta-2 microglobulin, protein electrophoresis, complete blood count, HbA1C, and comprehensive metabolic panel were normal.
XD is a rare non-Langerhans cell histiocytosis that distinctively involves the flexural areas and mucous membranes.\textsuperscript{1-2, 5} The pathogenesis is understood to be a reactive proliferative histiocytosis with secondary lipid deposition.\textsuperscript{5-6} XD is associated with upper airway lesions, CNS involvement of hypothalamus and pituitary causing mild diabetes insipidus, and rarely with plasma cell dyscrasias and monoclonal gammopathy.\textsuperscript{1-2,5} XD has a variable prognosis, with three subtypes: a) self-resolving, as in our patient, b) persistent disease, and c) rare systemic progression with organ dysfunction and CNS involvement.\textsuperscript{4,6}

There is no consensus on XD treatment; options include surgical removal, carbon dioxide laser treatment, cryotherapy, intralesional steroids, 2-chlorodeoxyadenosine, and electrocoagulation.\textsuperscript{1,3,5} Surgical removal and carbon dioxide laser have shown the best cosmetic outcomes, though 2-chlorodeoxyadenosine has recently demonstrated efficacy.\textsuperscript{1,5} Radiation therapy is effective for airway obstruction, while cyclophosphamide is used for mucosal lesions.\textsuperscript{1,5} Our patient had no evidence of mucosal lesions or airway obstruction and declined any intervention.

The differential diagnosis of XD includes adult-onset juvenile xanthogranuloma, eruptive xanthomas, as well as other xanthomatous diseases. The prognosis of each disease varies and, thus, can impact the management of these patients.

Histologically, XD resembles juvenile xanthogranuloma, which has a benign
prognosis.\textsuperscript{1} However, the disease typically affects children under the age of 2 and self-

![Figure 2](image)

**Figure 2** Histopathology of a right thigh biopsy, which showed a superficial dermal nodule of CD68-positive foamy histiocytes, as well as a few Touton giant cells seen in the enlarged image.

resolves; adult-onset juvenile xanthogranuloma is rare.\textsuperscript{1} Our patient was initially misdiagnosed with eruptive xanthomas, which, unlike XD, are associated with hypertriglyceridemia and commonly involve the extensor surfaces.\textsuperscript{3}

**CONCLUSION**

Overall, XD is a rare disease that can be difficult to distinguish from adult-onset juvenile xanthogranuloma; the latter may be within a spectrum of self-resolving XD. However, the consequences of XD can be systemic, and in a minority of cases, life-threatening. Therefore, it is critical to ensure these patients obtain a complete laboratory and imaging evaluation to rule out systemic involvement and close follow-up.

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