CASE REPORT

Asymptomatic Buschke-Ollendorff Syndrome Presenting with Yellowish Papules: A Case Report

Monira El Waseef,[1] MD, Shaimaa Farouk,[1] MD, Hayder Al-Darraji,[2] MD, Ammar Al-Dujaili,[2] MD
Belal Muwafak,[2] MD

[1]*Cairo Hospital for Dermatology and Venereology (Al Houd Al Marsoud), Ministry of Health and Population, Cairo, Egypt*
[2] *Egyptian board of Dermatology*

ABSTRACT

Buschke-Ollendorff Syndrome (BOS) is a rare genetic disorder characterized by the presence of connective tissue nevi and osteopoikilosis. Asymptomatic individuals may seek medical advice for nonspecific dermatological findings such as yellowish papules, which can lead to incidental diagnosis. This case report highlights the diagnostic approach to a patient with asymptomatic BOS presenting with cutaneous lesions, emphasizing the importance of clinical evaluation and radiographic imaging.

KEYWORDS: Buschke–Ollendorff, Asymptomatic, Syndrome, osteopoikilosis

INTRODUCTION

Buschke-Ollendorff Syndrome (BOS) is a rare genetic autosomal dominant disorder. It is primarily associated with connective tissue nevi termed (dermatofibrosis lenticularis disseminate) and osteopoikilosis, a benign sclerotic bone dysplasia. This syndrome is caused by mutations in the LEMD3 gene, which plays a crucial role in regulating TGF-β and BMP signaling pathways that influence bone and connective tissue development. While these features are often asymptomatic, the dermatological manifestations may raise cosmetic or diagnostic concerns, leading patients to seek medical advice.[1] This report discusses the presentation, diagnostic workup, and clinical management of a patient with BOS presenting with yellowish papules.

CASE PRESENTATION

A 17-year-old female presented to the dermatology clinic with asymptomatic, sudden onset and persistent yellowish papules over upper extremities of one year duration. with no associated pain, pruritus, or systemic symptoms. There was no personal or family history of similar skin changes, skeletal abnormalities, or significant medical conditions.

Physical examination revealed multiple, well-defined, firm, yellowish papules measuring 2–5 mm in diameter, distributed symmetrically over upper extremities (Fig. 1 A, B, C). The skin overlying the lesions was smooth and non-tender. No other cutaneous abnormalities were noted, with no evidence of systemic involvement.

Dermoscopic examination of yellowish skin

Correspondence: *Dr. Shaimaa Farouk: E-mail: dr.shaimaafarouk@gmail.com ,Tel.: +201096244898. ORCID ID: 0000-0003-2415-2511*

papules revealed whitish scare like papule with atrophic appearance (Fig. 1 D).

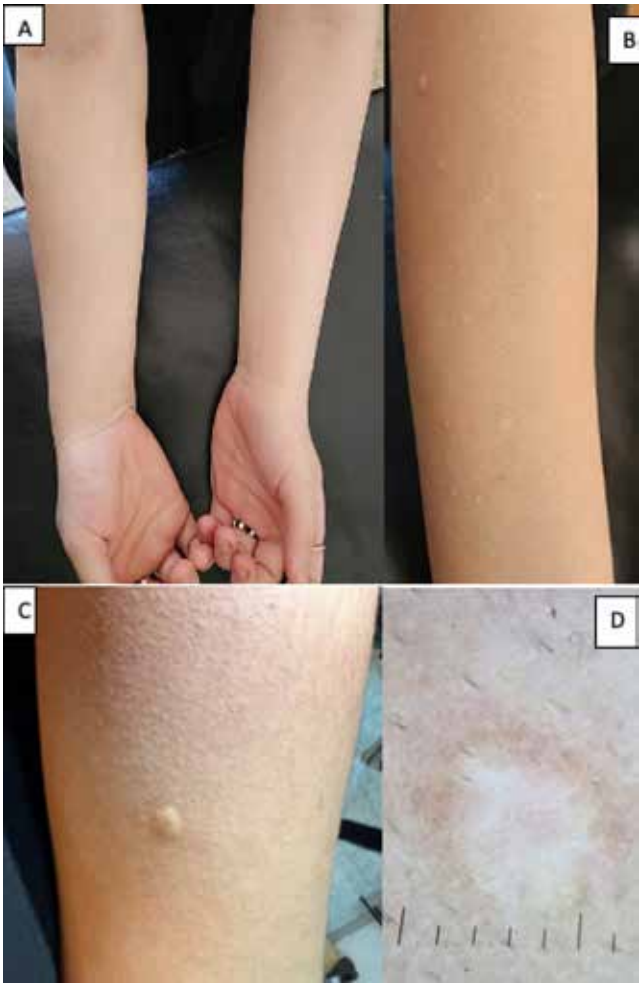

Fig. 1 A 17-year-old female presented with A, B,C multiple, well-defined, firm, yellowish papules measuring 2–5 mm in diameter, distributed symmetrically over upper extremities. On Dermoscopy examination D reveled whitish scare like papule with atrophic appearance.

A skin biopsy of one of the papules revealed increased spaces in between collagen fibers and a reduction in elastic fibers, consistent with a diagnosis of connective tissue nevus or Elastomas (Fig. 2).

Routine laboratory tests, including inflammatory markers, calcium, and alkaline phosphatase, glycated hemoglobin was within normal limits.

Radiographic imaging, including plain X-rays of forearms and hands, revealed multiple small, well-demarcated areas of increased bone density and tiny sclerotic focus seen at capitellum (bony island) characteristic of osteopoikilosis (Fig. 1).

Diagnostic Workup: Given the clinical suspicion of a connective tissue disorder, a skin biopsy was performed. Histopathological examination revealed thickened collagen bundles interspersed with elastin fibers, consistent with connective tissue nevi. Radiographic imaging, including plain X-rays of forearms and hands, revealed multiple small, well-demarcated areas of increased bone density, characteristic of osteopoikilosis, Genetic testing confirmed a pathogenic mutation in the LEMD3 gene, establishing the diagnosis of Buschke-Ollendorff syndrome.

DISCUSSION

BOS is a rare connective tissue disorder with an estimated prevalence of less than 1 in 20,000 individuals. It is characterized by two hallmark features: connective tissue nevi and osteopoikilosis. Connective tissue nevi typically manifest as asymptomatic, firm, yellowish papules or plaques, commonly located on the trunk, extremities, or buttocks. Osteopoikilosis, often an incidental radiological finding, appears as multiple sclerotic foci in cancellous bone, particularly in the pelvis, long bones, and spine.[2]

The pathogenesis of BOS involves mutations in the LEMD3 gene, which encodes an inner nuclear membrane protein that negatively regulates bone morphogenetic protein and transforming growth factor-β signaling pathways. This dysregulation leads to the abnormal connective tissue and bone findings characteristic of the syndrome.[3]

While BOS is generally asymptomatic, its recognition is essential to differentiate it from other conditions with similar dermatological or radiological features, such as pseudoxanthoma elasti-

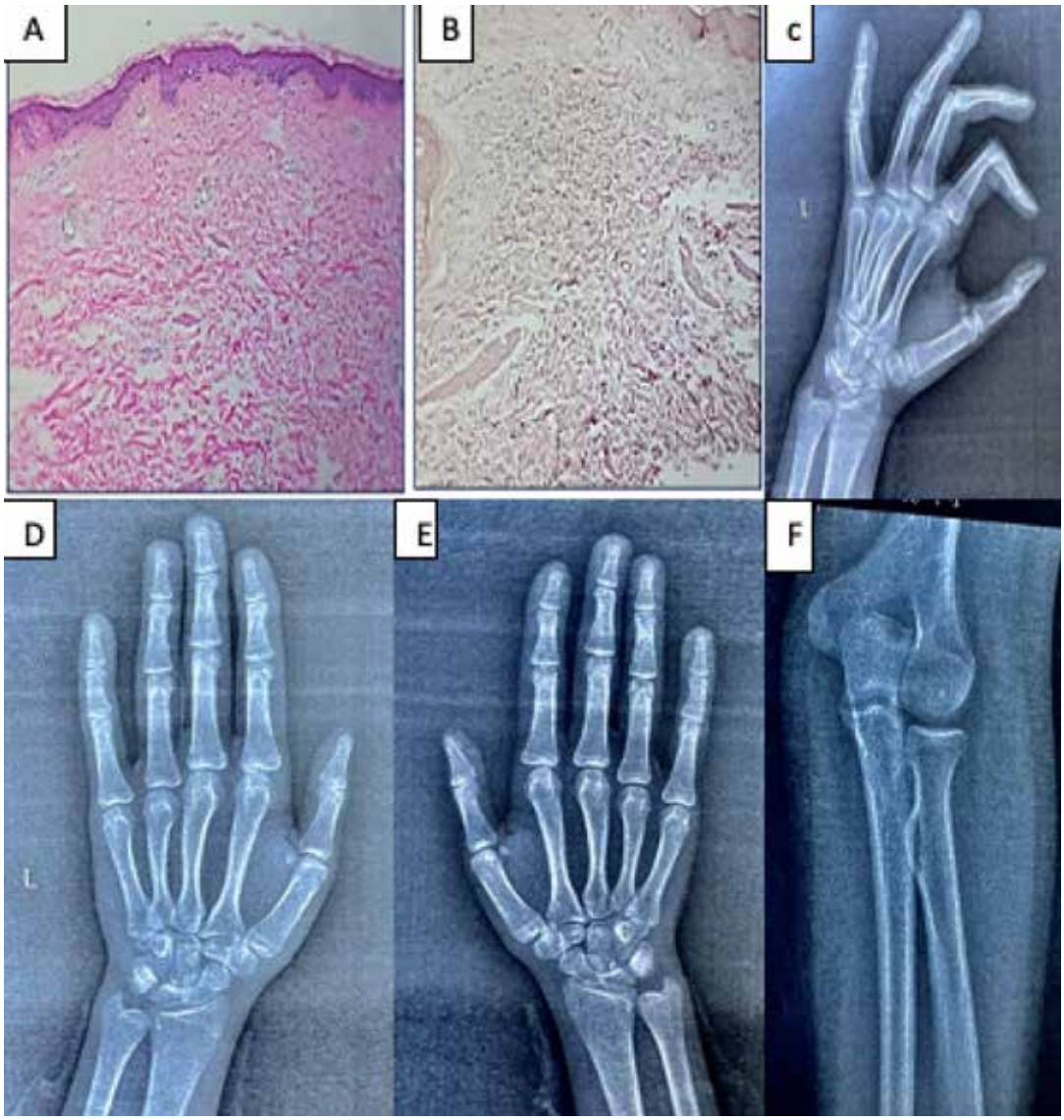

Fig. 2 A skin biopsy of one of the papules revealed (A) basked wave stratum corneum, Dermis showed increase spaces in collagen fibers, (B) Orcin stain showed highlighted the elastic fibers which were decreased in amount and fragmented C, D,E ,F plain X-rays of forearms and hands, revealed multiple small, well-demarcated areas of increased bone density and tiny sclerotic focus seen at capitellum (bony island).

cum, dermatofibroma, or osteoblastic metastases. Genetic counseling is recommended for affected individuals and their families, given the autosomal dominant inheritance pattern.[4]

Management of BOS is primarily supportive, focusing on patient education and reassurance regarding the benign nature of the condition. Regular follow-up is advised to monitor for potential complications, although these are rare.

CONCLUSION

This case highlights the significance of considering BOS in the differential diagnosis of incidental osteopoikilosis, even in asymptomatic individuals. Genetic testing is essential for definitive diagnosis, particularly in the absence of clinical manifestations. Increased awareness of asymptomatic presentations will aid in timely identification and management of this rare syndrome. Early diagnosis facilitates patient education, ge-

netic counseling, and long-term surveillance to prevent potential complications.

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