

Atypical Cutaneous Manifestations of Dyskeratosis Congenita: A Case Report from Yemen

Abstract

Dyskeratosis congenita is a rare genodermatosis characterized by a classic triad of reticulate skin pigmentation, nail dystrophy, and mucosal leukoplakia, along with a predisposition to malignancy. We report the case of a 15-year-old boy who presented to our dermatology clinic with the characteristic triad and several rare findings. Unfortunately, genetic testing was not available, so the diagnosis was based solely on clinical findings. To the best of our knowledge, only one case with the additional findings of atrophic scars, reticulated atrophy, and hypopigmented macules has been reported in the literature to date, and no cases of reticulate pigmentation of the helix have yet been described.

Introduction

Dyskeratosis congenita (DC), also known as Zinsser-Engman-Cole syndrome, is a rare multisystem genodermatosis with an estimated incidence of approximately 1 per million people, it was originally described by Zinsser in 1906.[1,2] DC is considered the archetypal telomere biology disorder (TBD) and is characterized by the classic triad of oral leukoplakia, nail dystrophy, and reticulate skin pigmentation.[3] Although X-linked DC due to *DKC1* mutations is an important form, DC/TBDs have multiple inheritance patterns, including autosomal dominant and autosomal recessive forms. Most cases are associated with defects in the *DKC1* gene, which encodes the dyskerin protein involved in telomere maintenance.[4] DC is most commonly inherited in an X-linked recessive disorder; therefore, males are affected approximately three times more often than females. Approximately 80% of DC cases exhibit associated bone marrow failure.[5]

Case report

A 15-year-old boy presented to the dermatology clinic with dystrophic nails and dyspigmentation of the skin and tongue. He was short for his age, and was wearing a diaper due to a urethral stricture that had recently been surgically corrected.

On cutaneous examination, a lacy, reticulated hyperpigmentation pattern was identified on the neck, upper chest, and upper abdomen, occasionally admixed with confetti-like hypopigmented macules. In addition, all 20 nails were dystrophic, hypoplastic, or absent, with dorsal pterygia. The patient's tongue was covered with white reticulated plaques (Figure. 1A–D) and (Figure 2A–D). Ophthalmologic examination revealed epiphora (excessive tearing) of the left eye.



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Alshami MA^{1*}, Alshami AM², Alshami HM¹ and Lutf RM¹

¹Department of Dermatology, Faculty of Medicine and Medical Sciences, Sana'a University, Sana'a, Yemen

²Department of Conservative Dentistry, Faculty of Dentistry, Sana'a University, Sana'a, Yemen

*Address for Correspondence

Mohammad Ali Alshami, Department of Dermatology, Faculty of Medicine and Medical Sciences, Sana'a University, Sana'a 1064, Yemen. E-mail Id: mohammadalshami62@gmail.com

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The patient also exhibited multiple oval-shaped atrophic scars in an oblique linear distribution over the right flank (Figure 2C). Larger hypopigmented macules were present on the upper back and nape of the neck, with smaller ones on the anterior neck (Figure 2B), (Figure 3C). The patient also had adermatoglyphia of both palms and early graying of the scalp hair (Figure 1D), (Figure 3D).

The initial differential diagnoses included reticulate

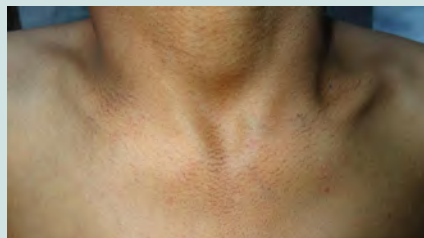


Figure 1A: Lacy reticulate hyperpigmentation on the upper chest and anterior aspect of the neck.



Figure 1B: Larger areas of reticulate, partially confluent hyperpigmentation admixed with hypopigmented macules over the right axilla and upper arm.



Figure 1C: Near-complete loss of nails of the third and fourth digits of the right hand (anonychia) and partial loss of nails of the second and fifth digits (micronychia), with dorsal pterygia and wrinkling of the skin over the distal phalanges.



Figure 1D: Right palmar hyperkeratosis with superimposed guttate hypomelanotic macules and loss of dermatoglyphics (adermatoglyphia).



Figure 2A: Confetti-like hypopigmented macules with lacy reticulated hyperpigmentation over the submental area and anterior aspect of the neck.

acropigmentation of Kitamura, dyschromatosis symmetrica hereditaria, Naegeli–Franceschetti–Jadassohn syndrome, dermatopathia pigmentosa reticularis, and Dowling–Degos disease. Although mucosal lichen planus was considered a differential diagnosis of the oral lesions, the other cutaneous findings were

inconsistent with this diagnosis. Based on the characteristic clinical findings, the patient was diagnosed with DC. The patient fulfilled the diagnostic criteria for DC, meeting three major criteria—reticulate pigmentation, nail dystrophy, and oral leukoplakia—and three minor criteria—early graying of hair, urethral stricture, and short stature [6,7].

Laboratory investigations revealed a white blood cell count of $20.1 \times 10^9/L$, with 75.8% neutrophils and 17.9% lymphocytes, a platelet count of $485 \times 10^9/L$, and a hemoglobin level of 10.9 g/dL. A punch biopsy was declined by the patient's parent. These routine laboratory investigations did not indicate bone marrow involvement,

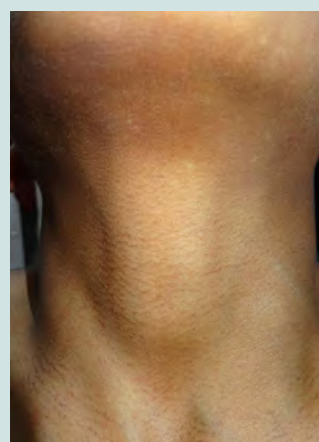


Figure 2B: Confetti-like hypopigmented macules with lacy reticulated hyperpigmentation over the submental area and anterior aspect of the neck.



Figure 2C: Three atrophic scars in an oblique linear distribution over the right flank.



Figure 2D: Fine, lacy, reticulated pigmentation over the nose, cheeks, and upper lip.



Figure 3A: Lacy reticulate pigmentation over the chest and abdomen, along with two atrophic macules on the right lower abdomen.

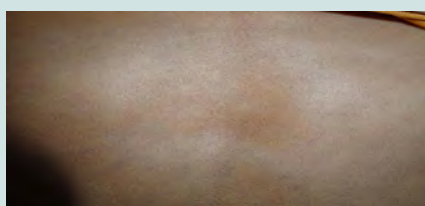


Figure 3B: Lacy reticulate pigmentation over the back, producing the impression of a reticulated atrophic lesion.

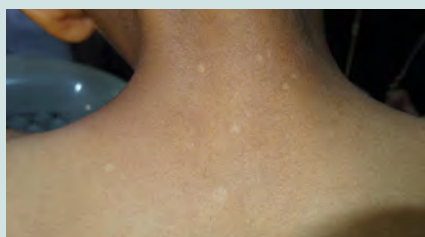


Figure 3C: Large hypopigmented macules over the upper back and nape of the neck.



Figure 3D: Dense reticulate hyperpigmentation of the right helix accompanied by premature graying of the scalp hair.

which occurs in up to 80% of cases and may progress to bone marrow failure, necessitating hematopoietic cell transplantation. This apparent leukocytosis with neutrophilia was considered to be due to bacterial infection, while the anemia was attributed to malnutrition,

which is common in our country. Hematological consultation was advised, but the parents refused a bone marrow aspiration/biopsy.

No abnormalities were identified in the neurological, pulmonary, orthopedic, or gastrointestinal systems. A genetic test for a mutation in the *CTCI*, *DKC1*, *TERC*, *TERT*, *TINF2*, *NHP2*, *NOP10* and *WRAP53* genes, all essential for proper telomere function, would have confirmed the diagnosis of DC; however, this test was unavailable at our institution. The patient was scheduled for follow-up visits every 6 months but was lost to follow up.

Discussion

The present case expands the clinical spectrum of DC by demonstrating several uncommon cutaneous manifestations in addition to the characteristic mucocutaneous triad.[8] Specifically, the patient exhibited multiple oval-shaped atrophic scars arranged in an oblique linear distribution over the right flank, a finding that has been described only once previously, by Milgrom in 1964. Larger hypopigmented macules were also present on the upper back and nape, with smaller macules on the anterior neck and submental region; these findings have been reported only once previously, by Ward et al. in 2017.[9] This case is unique in that the patient exhibited not only the typical mucocutaneous triad but also several rare manifestations. To the best of our knowledge, atrophic scars, reticulated atrophy (Figure 3B), and hypopigmented macules have each been described only once previously in the literature. Furthermore, reticulate pigmentation of the helix, as observed in this patient, has, to the best of our knowledge, not been previously reported, as a Medline search revealed no results.

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