Article

PAMJ Clinical Medicine

Images in clinical medicine

Homozygous type 2 familial hypercholesterolemia

Shamma Khamis Almheiri, Tarek Mohamed Ibrahim

Corresponding author: Shamma Khamis Almehairi, Dermatology Department, Rashid Hospital, Dubai, United Arab Emirates. shammabin3bed@gmail.com

Received: 23 Oct 2023 - **Accepted:** 30 Nov 2023 - **Published:** 12 Dec 2023

Keywords: Xanthoma, familial hypercholesterolemia, genetics, dermatology, cardiology

Copyright: Shamma Khamis Almheiri et al. PAMJ Clinical Medicine (ISSN: 2707-2797). This is an Open Access article distributed under the terms of the Creative Commons Attribution International 4.0 License (https://creativecommons.org/licenses/by/4.0/), which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.

Cite this article: Shamma Khamis Almheiri et al. Homozygous type 2 familial hypercholesterolemia. PAMJ Clinical Medicine. 2023;13(34). 10.11604/pamj-cm.2023.13.34.42028

Available online at: https://www.clinical-medicine.panafrican-med-journal.com//content/article/13/34/full

Homozygous type 2 familial hypercholesterolemia

Shamma Khamis Almheiri[1,&], Tarek Mohamed Ibrahim[1]

[1]Dermatology Department, Rashid Hospital, Dubai, United Arab Emirates

[&]**Corresponding author**

Shamma Khamis Almehairi, Dermatology Department, Rashid Hospital, Dubai, United Arab Emirates

Image in medicine

A 59-year-old woman with a history of hypertension and hypercholesterolemia was admitted as a case of Non-ST-elevation myocardial infarction (NSTEMI). Urgent coronary artery angiography was performed and revealed subtotal occlusion of the left main coronary artery and the left anterior descending coronary artery, which indicated the need for an urgent coronary artery bypass grafting (CABG). Dermatology service was consulted prior to the surgery in view of a generalized skin lesion. The patient reported that the skin lesion started to appear at the age of nine and progressed as the patient got older. Four family members including one sibling, two nephews, and one niece also had the same skin lesions, all of whom passed away in their twenties. On examination, multiple tendinous xanthomas

were seen on the bilateral hands (A) and feet (B). Webspace plane xanthoma on the right hand, located between the index and the middle fingers (A). Plaques and papules of plane xanthomas on the arms (C) and legs, including the antecubital fossae (D). Lipid profile revealed total cholesterol of 840 mg/dL, LDL of 792 mg/dL, and normal triglycerides. A diagnosis of homozygous type 2 familial hypercholesterolemia was made. It is an autosomal dominant disorder characterized by pure hypercholesterolemia. Homozygotes develop xanthomas during their childhood, have a very high LDL level reaching 800 mg/dL or above, and develop pathognomonic plane xanthomas of the webspaces and the antecubital fossae.

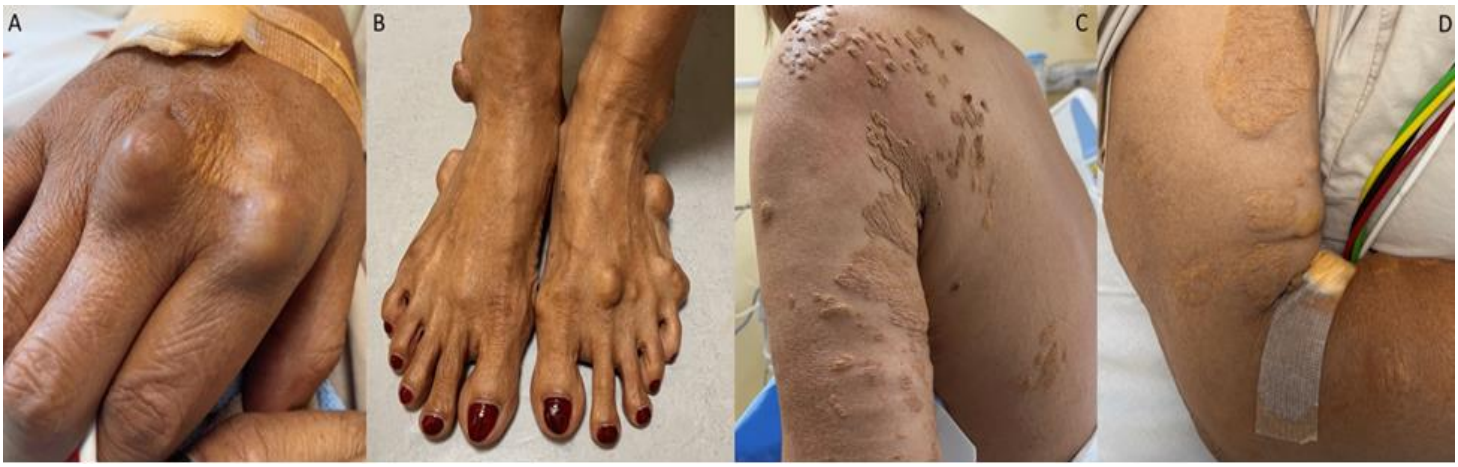

Figure 1: (A) tendinous xanthomas located on the index and middle fingers of the right hand with an interdigital webspace plane xanthoma situated between them; (B) multiple large tendinous xanthomas involving both feet; (C) multiple papules and plaques of plane xanthomas on the left arm and extending to involve the left shoulder area and the back; (D) multiple plaques of plane xanthomas on the right arm involving the right antecubital fossa