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Saudi Patient with Keutel Syndrome, a Rare Disease

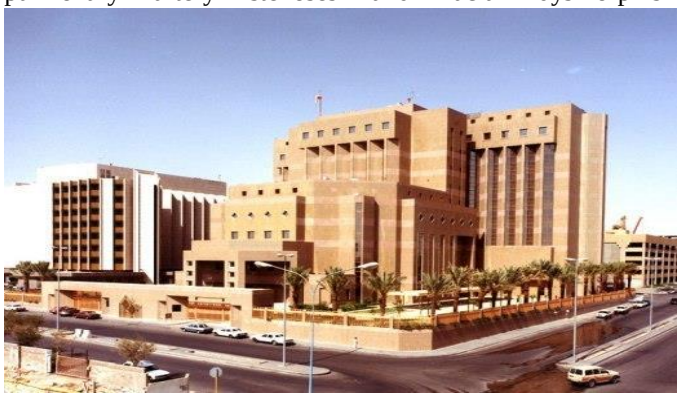
Nouf Mohammed Aleid

Prince sultan military medical city, Saudi Arabia

Abstract

22 years old female, presents with 2 years history of lax skin on neck and axilla, also patient complain of hardening of both ears. No History of bleeding, or poor wound healing, or tendency to bruise easily. Her sister has same complaint. Her parents are relatives. On examination, patient found to have lax skin in neck and both axillae, no other skin changes. Both ears are firm on palpation. The diagnosis of psuedoxanthma elasticum was suspected and skin biopsy was taken from left axilla which showed: thickened calcified elastic fibers in the dermis, favors the diagnosis of pseudoxanthoma elasticum. Genetic testing: showed homozygous mutation in the matrix Gla protein (MGP) (c.94+1G>A) .So the patient diagnosed as Keutel Syndrome. The patient was referred to ophthalmology , ENT, Plastic, Pulmonolgy and gastroenterology.

Keutel syndrome is a rare autosomal recessive disease caused by mutations in the gene encoding the matrix Gla protein (MGP, located at 12p13.1-p12.3). Less than 30 cases have been reported in the literature so far, with the majority of patients being diagnosed during childhood. It is characterised by diffuse cartilage calcification, brachytelephalangism, peripheral pulmonary artery stenoses and facial dysmorphism.



Biography:

Nouf Mohammed aleid is a Dermatology consultant at Prince sultan military medical city. She is currently working at Saudi board of dermatology and she done her Hair and Nail fellowship from university of Miami.

Speaker Publications:

1. "Scalp Itch: A Systematic Review"
2. "Common Allergens Identified Based on Patch Test Results in Patients with Suspected Contact Dermatitis of the Scalp"
3. "Medical comorbidities in patients with lichen planopilaris, a retrospective case-control study".
4. "Patch test nails".

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