RESEARCH PAPER.

JOHA
Journal Of Health Academics

REPORT OF A CASE AND REVIEW OF THE LITERATURE-COMPLETE PACHYDERMOPERIOSTOSIS

Gunarathne H.W.N.N[1], Wannigama E[2] ,Perera S[3], Wickramarathne C[1]

[1]Consultant Dermatologist Ministry of Health Sri Lanka, [2]Consultant Opthalmologist Ministry of Health Sri Lanka,[3] Post graduate trainee in Dermatology Ministry of Health Sri Lanka,

Corresponding Author: Gunarathne H.W.N.N email: niroshiwannigama@gmail.com

A 22-year-old male presented with extensive skin folding on the forehead, severe acne and scaly skin eruption with seborrheic distribution. No family history of similar condition or consanguinity noted. Examination revealed cutis vertices gyrate over the forehead, seborrhoea, acne and seborrheic dermatitis. Moreover, clubbing of his fingers and toes was noted and he had swollen knee joints. Radiological evaluation revealed hypertrophic osteoarthropathy. Skin biopsy findings showed sebaceous hyperplasia with thickening of the dermis with mucin deposition compatible with pachydermoperiostosis. However no other associations were found after through biochemical, haematological and radiological work up. primary PDP was diagnosed based on the typical clinical and radiological findings. The patient was referred to plastic surgical clinic for further management of cutis vertices gyrata where it was arranged Botox injections without surgical correction-but later we discussed with them regarding the nature of the disease and offered surgical correction for the patient. It was arranged the surgical correction instead of Botox.

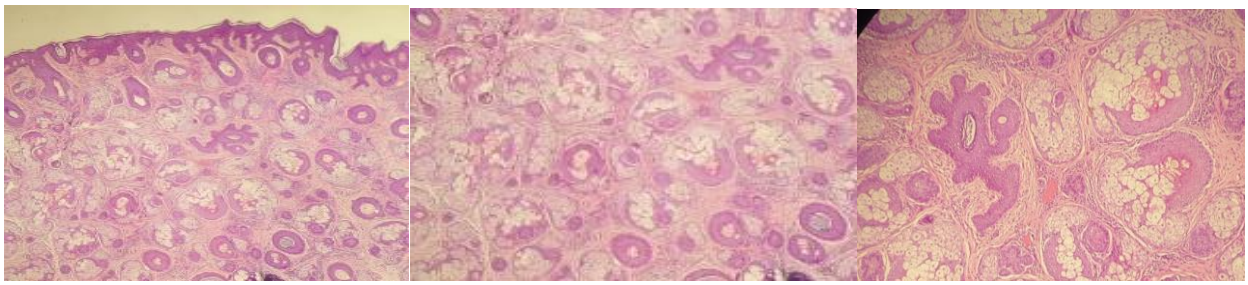

Figure 1: Skin biopsy findings showed sebaceous hyperplasia with thickening of the dermis with mucin deposition compatible with pachydermoperiostosis.

 2022 September 1

Academy of Health Research

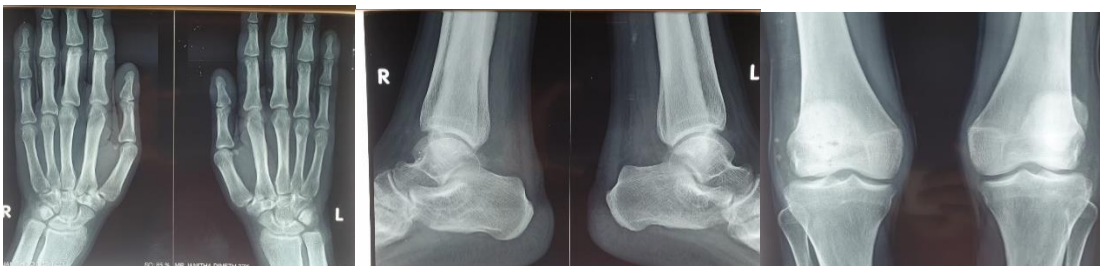

Figure 2: Radiological evaluation revealed hypertrophic osteoarthropathy.

It is important to arrange multidisciplinary approach for patients with uncommon diseases as this case in order to improve the ultimate patient outcome. It was arranged the management of active acne with oral doxycycline, topical adapalene and topical benzyl peroxide. Acne scars were managed with subcission, TCA cross and several cycles of TCA peels. Different mode of therapy was used as the patient was having acne scars with different morphology. Ablative laser was a better option but patient had financial constraints. It was offered him to above available options. Pachydermoperiostosis (PDP)is a hereditary disorder characterized by thickening of skin(pachydermia) and new bone formation (periostosis), has autosomal dominant inheritance and variable penetrance. Three forms of this entity have been identified: complete, incomplete, and the forme fruste. Clinical diagnosis can be made when at least two out of following four features are present- family history, hypertrophic skin changes, bone pain/radiographic changes, or clubbing. Management of this entity is symptomatic. Disease progression typically ceases after 10 years but patients may leave with disabling complications. The case presented here is a complete form of the syndrome, with most of the characteristic clinical and radiological features. Although this syndrome has a strong association with heredity, in this case, there was no report of relatives with similar characteristics.

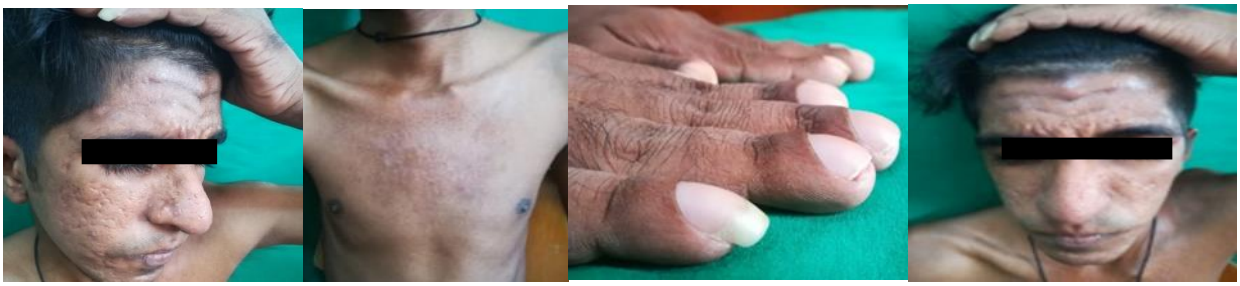

Figure 3: Visible external clinical feature of Pachydermoperiostosis

Pachydermoperiostosis (PDP) or primary hypertrophic osteoarthropathy (PHO) is a rare hereditary disease that was first described in 1868.(1) Its actual incidence is unknown,(2) but as evidenced by the MEDLINE search, a total of 286 reported cases were found. Adolescent males are predominantly affected, with male-to-female ratio of approximately 7:1.(3)

Increased levels of prostaglandin E_2 (PGE2) resulting from defective degradation due to the implicated gene mutations, HPGD and SLCO2A(1), appear to contribute to the pathogenesis of PDP(4). The severity of pachydermia and associated histological changes have been correlated with serum PGE_2 levels and SLCO2A(1) genotypes. PGE2 can mimic the activity of osteoblasts and osteoclasts, which may be responsible for the acro-osteolysis and periosteal bone formation(5). Moreover, the prolonged local vasodilatory effects of PGE2 may explain digital clubbing(6)(7). Although it is known that one-third of PDP patients have a family history(3), the present patient did not have relatives with suspected characteristics. A normal level of IGF-1 is strong evidence that the patient does not have acromegaly, which is an important differential diagnosis(8).

Generally, onset of pachydermoperiosteosis is noted at the adolescent age and its long-term persistence could create various disabilities and deformities. This situation simply directs to a state which has a sociocultural hidden psychologic impact. Therefore, it is necessary to arrange early detection which helps to integrate the patients into a well-organized rehabilitation care programme. As pachydermoperiosteosis is identified as an autosomal dominant condition, providing early genetic counselling can be considered as the most beneficial preventive strategy. As it is not possible to revert skin and bone changes, it is essential to.

REFERENCES

1. Sandoval ARH, Flores-Robles BJ, Llanos JC, Porres S, Dardón JDP, Harrison RM. Cutis verticis gyrata as a clinical manifestation of Touraine-Solente-Gole' syndrome (pachydermoperiostosis). BMJ Case Rep. 2013 Jul;2013.
2. Lee S, Park SY, Kwon HJ, Lee C-H, Kim O-H, Rhee Y. Identification of the Mutations in the Prostaglandin Transporter Gene, SLCO2A1 and Clinical Characterization in Korean Patients with Pachydermoperiostosis. J Korean Med Sci. 2016 May;31(5):735–42.
3. Abdullah NRA, Jason WLC, Nasruddin AB. Pachydermoperiostosis: a rare mimicker of acromegaly. Endocrinol diabetes Metab case reports. 2017;2017.
4. Nakazawa S, Niizeki H, Matsuda M, Nakabayashi K, Seki A, Mori T, et al. Involvement of prostaglandin E2 in the first Japanese case of pachydermoperiostosis with HPGD mutation and recalcitrant leg ulcer. Vol. 78, Journal of dermatological science. Netherlands; 2015. p. 153–5.
5. Uppal S, Diggle CP, Carr IM, Fishwick CWG, Ahmed M, Ibrahim GH, et al. Mutations in 15-hydroxyprostaglandin dehydrogenase cause primary hypertrophic osteoarthropathy. Nat Genet. 2008 Jun;40(6):789–93.
6. Kim HJ, Koo KY, Shin DY, Kim DY, Lee J-S, Lee MG. Complete form of pachydermoperiostosis with SLCO2A1 gene mutation in a Korean family. Vol. 42, The Journal of dermatology. England; 2015. p. 655–7.
7. Coggins KG, Coffman TM, Koller BH. The Hippocratic finger points the blame at PGE2. Vol. 40, Nature genetics. United States; 2008. p. 691–2.

 2022 September 1

Academy of Health Research