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Pachydermoperiostosis (Touraine–Solente–Gole Syndrome) Presenting with Alopecic Scalp Nodules: A Case Report

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ABSTRACT

This report is to describe a rare case of Pachydermoperiostosis in a 19- year- old male presenting with swellings in his scalp. Following excision, pathological examination revealed Follicle structure containing a hair shaft destroyed due to inflammation and intertwined with caseous necrosis and surrounded by lymphohistiocytic inflammation.

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INTRODUCTION

Pachydermoperiostosis is a scarce genetic osteoarthrodermopathic disorder, also known as the primary idiopathic version of hypertrophic osteoarthropathy. The characteristic features of this disorder are acropachia (digital clubbing), pachyderma (hyperplasia of the soft tissues), hyperhydrosis, in addition to periosteosis (sketelatal changes) (1). The differential diagnosis includes acromegaly, severe insulin resistance, secondary hypertrophic osteoarthropathy, drug intake e.g. Minoxidil, Marfan's Syndrome, MacCune Albright Syndrome, syphilitic periosteosis psoriatic onycho-pachydermo-periosteosis (POPP), Paget's Disease, and hypothyroidism (1,2).

CASE PRESENTATION

A 19-year-old male was referred to the Department of Plastic Reconstructive and Aesthetic Surgery at Sutcu Imam University Hospital, Kahramanmaras, Turkey, complaining of swellings in his scalp of seven-month duration. The patient had coarse facial features, with forehead skin being thickened and thrown into folds and furrows, blepharoptosis, greasy skin (excessive sebaceous secretions) (Figure 1-3). Enlarged hands and feet, with digital clubbing were noticed as well. Physical examination of the scalp revealed cutis verticis gyrata with four soft mobile swellings, the largest approximately measured 1*0.5*0.5 cm and the smallest approximately measured 0.5*0.5*0.5 cm, the masses did not show transillumination.

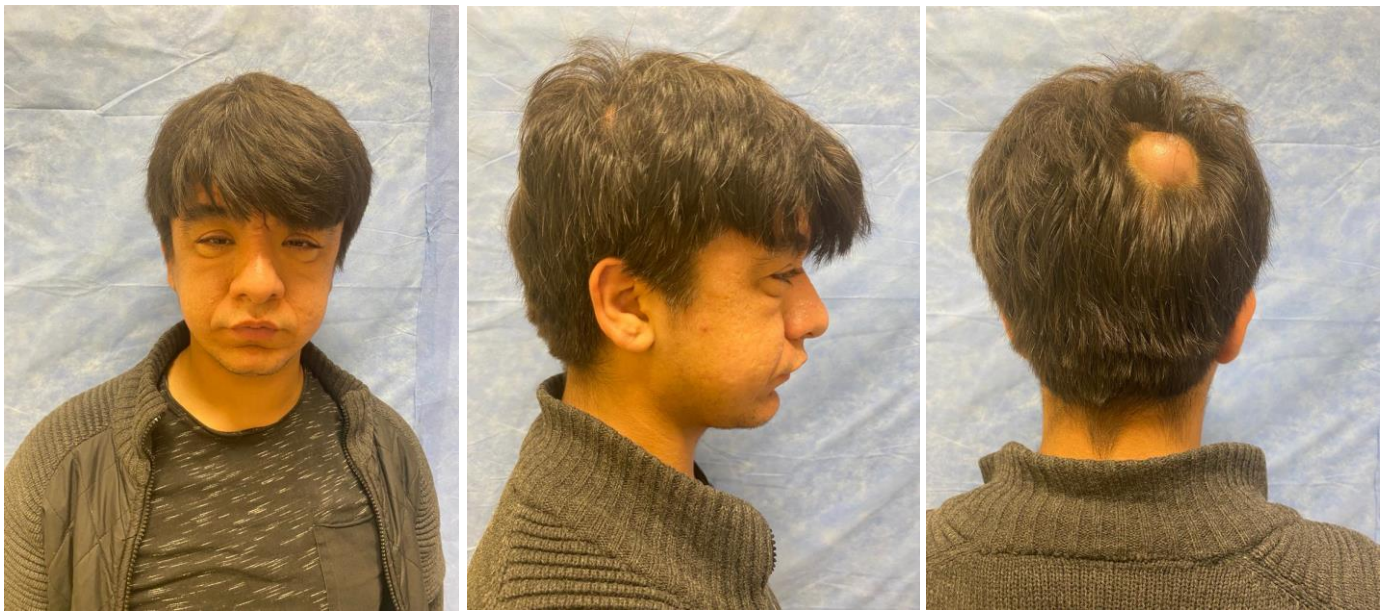

Figure 1-3. The patient had coarse facial features, with forehead skin being thickened and thrown into folds and furrows, blepharoptosis, greasy skin (excessive sebaceous secretions).

Ultrasonographic imaging revealed four thick- walled cystic lesions with the largest measuring 38*19mm, dense content with millimetric calcifications were observed.

Forwards, the patient was scheduled for excision. Under sterile conditions infiltration using 6 ml of lidocaine HCl 20mg/mcg epinephrine 0.0125 were diluted with normal saline 1:1, bicarbonate was added to the mixture in 1:10 proportion. The excised lesions were sent to the pathology lab for histopathological examination. And the incisions were sutured primarily using 3-0 Prolene sutures.

Pathological examination following PAS, Giemsa, and GMS staining showed necrotizing granulomatous reaction accompanied by prominent lymphocytic inflammation, lipogranuloma, fibrin and focal suppuration. Follicle structure (H/EX200) containing a hair shaft (arrow) destroyed due to inflammation and intertwined with necrosis (observed on the middle-left side of the follicle lumen) is seen in figure 4. Caseous necrosis and surrounding lymphohistiocytic inflammation (H/EX100) is seen in figure 5.

DISCUSSION

Pachydermoperiosteosis, also known as primary hypertrophic osteoarthropathy and AS Touraine-Solente-Gole syndrome was first described by Friedreisch in 1868 (1). This disorder shows an autosomal dominant inheritance with variable penetrance usually affecting pubertal males, with male to female ratio 9:1, family history is usually present in one third of the cases (3,4). Full manifestations are usually seen by around 20-30 years of age. The inheritance pattern is not fully recognized as some authors have proposed autosomal recessive inheritance pattern (5).

The usual presentation includes characteristic features of the complete form of this disorder are acropachia (digital clubbing), pachyderma (hyperplasia of the soft tissues),hyperhydrosis , in addition to periosteosis (sketelal changes). In the incomplete form cutis verticis gyrata is absent. Whereas in the forme fruste there is pachyderma with minimal or absent skeletal features i.e. periosteosis (6).

Other manifestations include, seborrhea (90%), acne, folliculitis, dilated pores, flushing, thickened eyelids leading to blepharoptosis, lower extremity oedema, and cutis verticis gyrata which was first described by Unna in 1907 as marked thickening of the skin of the forehead resembling the sulci and gyri of the cerebrum (6). When this disorder manifests in childhood, congenital heart disease, bronchiectasis, pneumonia and cystic fibrosis are seen as comorbidities, malignancies such as Hodgkin's Disease have been reported as well (6).

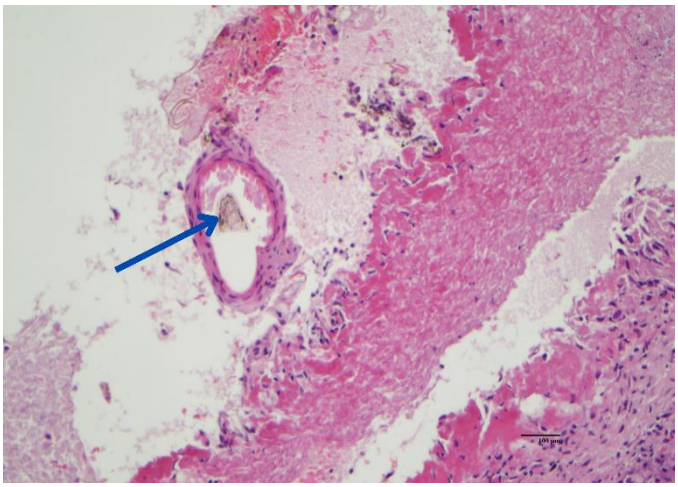

Figure 4. Follicle structure (H/EX200) containing a hair shaft (arrow) destroyed due to inflammation and intertwined with necrosis (observed on the middle-left side of the follicle lumen) is seen.

Several pathophysiologies have been proposed so far, Mauritz-Lavin suggested that this disease is a result of an increase in the proliferation of fibroblasts hence increasing the proliferation of

collagen fibers. Wegrowski suggested that the dysregulation of the matrix deposits and the increase in decorin protein result in this disease (7). It is also thought that as a result of defective selective uptake across the plasma membrane by solute carrier organic anion transporter family member 2A1, and/ or intracellular degradation by 15-hydroxyprostaglandin dehydrogenase result in increased levels of prostaglandin E2 which induce cytokine- mediated tissue remodeling and vascular stimulation, resulting in excessive sweating, acro-osteolysis, periosteosis. Arthritis, as well as pachyderma (8). The massive growth of the fibrillar structures of the dermis and subcutaneous tissues result in intimate skin cohesion, the ostia of hair follicles become expanded and horny masses accumulate. Mature sweat and sebaceous glands increase, periosteal ossification can be seen, hyperplasia of superficial synovial cells, marked thickening of the vasculature may occur this may have devastating consequences especially when involving the vasculature of the internal organs, myelofibrosis is not uncommon in such patients (5).

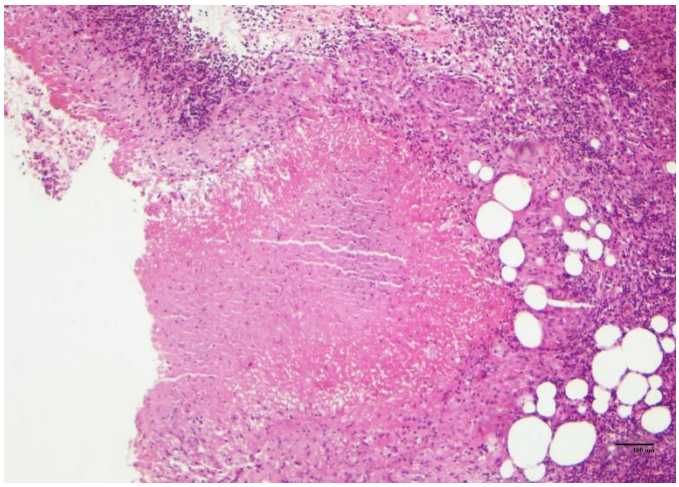

Figure 5. Caseous necrosis and surrounding lymphohistiocytic inflammation (H/EX100) is seen.

Definitive treatment has not been established yet, however, symptomatic treatment includes NSAIDs, over the counter analgesics, intravenous bisphosphonates, when arthralgia is resistant to NSAIDs treatment Tamoxifen has been reported to be effective (1).

Cosmetic treatment includes rhytidectomy on the forehead furrows to improve the facial features in patients with pachydermoperiostosis as suggested by Taneja et al.(9).

To sum up, the diagnosis of pachydermoperiosteosis is challenging, and is mainly established by clinical and radiological investigations (10). Yet, the other spectrum of differential diagnosis must be kept in mind so as not to miss especially secondary hypertrophic osteoarthropathy which might have definitive treatment options.

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None to declare.

Conflict of Interest

None to declare.

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