



TOURAINÉ-SOLENTE-GOLÉ SYNDROME: AN UNUSUAL CASE OF DIGITAL CLUBBING AND HYPERHIDROSIS IN A TEENAGER.

Shishir Shouri¹, Shivangi^{2*}, Sabavath Arun³, Amber Kumar⁴, Shikha Malik⁵

¹MBBS, Junior Resident, Department Of Pediatrics, All Institute Of Medical Sciences, Bhopal, Madhya Pradesh, India – 462020.

^{2*}MBBS, Junior Resident, Department Of Pediatrics, All Institute Of Medical Sciences, Bhopal, Madhya Pradesh, India – 462020.

³MBBS, Junior Resident, Department Of Pediatrics, All Institute Of Medical Sciences, Bhopal, Madhya Pradesh, India – 462020.

⁴Additional Professor, Department Of Pediatrics, All Institute Of Medical Sciences, Bhopal, Madhya Pradesh, India – 462020.

⁵Professor, Department Of Pediatrics, All Institute Of Medical Sciences, Bhopal, Madhya Pradesh, India – 462020.

***Corresponding Author:** Shivangi,

*MBBS, Junior Resident, Department of Pediatrics, All Institute of Medical Sciences, Bhopal, Madhya Pradesh, India – 462020, Email: sinha.shivangi54820@gmail.com

Abstract:

Primary hypertrophic osteoarthropathy (PHO), characterised by digital clubbing, periostosis, pachydermia, and hyperhidrosis, is also known as pachydermoperiostosis (PDP). It is a rare genetic condition with three clinical presentations. Here, we describe a 14-year-old boy presenting with recurrent episodes of arthralgia, profuse perspiration, and increasing swelling of his fingers and toes. On examination, all digits exhibited grade 4 clubbing, hyperhidrosis, and anemia. Secondary causes of clubbing and hyperhidrosis were excluded. Although the clinical characteristics met the requirements for PDP, genetic testing was not carried out because of monetary limitations. Male preponderance and varying inheritance patterns characterize PDP, which usually first appears in adolescence, develops over ten years, and stabilizes. Thyroid acropathy and acromegaly are few of many differential diagnoses. This instance emphasizes how critical it is to conduct a comprehensive assessment and identify PDP early, especially in situations when genetic testing is not available.

Introduction:

Primary Hypertrophic Osteoarthropathy, also known as pachydermoperiostosis (PDP), a rare syndrome, was first reported in 1935 by Touraine, Solente and Golé¹. It is characterised by features including digital clubbing, thickened facial skin, excessive sweating (hyperhidrosis), and abnormal new bone formation that often presents with joint deformities. The condition usually presents in one of three clinical forms namely, complete form (pachydermia, periostosis, clubbing), incomplete form (periostosis and clubbing only) or forme fruste (pachydermia with minimal or no bone involvement). Case report:

A 14-year-old male child, first born of non-consanguineous marriage with insignificant past and family history presented with complaints of fingers and toes swellings for one-year, excessive sweating for 6 months, and arthralgia without any history of chronic drug usage. Examination revealed appropriate anthropometry for age, pallor, grade 4 clubbing of fingers and toes, and hyperhidrosis (Figure 1). Laboratory investigations showed anemia with microcytic and hypochromic picture on peripheral smear with normal iron profile. Secondary causes of hyperhidrosis and clubbing like hyperthyroidism, cardiac conditions causing clubbing, infectious, metabolic conditions, chronic obstructive pulmonary disease, asthma, malignancies, inflammatory bowel disease and auto immune conditions were ruled out. Henceforth, the phenotype of clubbing, arthralgia, hyperhidrosis were fulfilling the clinical criteria of PDP. However, genetic tests could not be done due to financial issues.

Discussion:

Digital clubbing, periostosis of long bones, and skin thickening (pachydermia), especially cutis verticis gyrata, are characteristic features of pachydermoperiostosis (also referred to as Touraine-Solente-Golé syndrome), a rare and genetic form of primary hypertrophic osteoarthropathy. It usually initially manifests during adolescence, progresses for next ten years, and then stabilizes². The first reported case enumerating this syndrome were the Hagner brothers, which was described by Freidreich in 1868³. However, it was first identified as a separate entity in 1935 by Touraine-Solente-Golé. With autosomal dominant inheritance, the disorder can frequently be familial and has a male predominance. Hyperhidrosis, coarse facial features, arthritis, and, in rare cases, anemia from bone marrow invasion are among the clinical characteristics. Radiologically, cortical thickening and the growth of new bone in the periosteum are noted. Acromegaly, thyroid acropachy, and subsequent hypertrophic osteoarthropathy are instances for differential diagnosis. The exact pathogenesis causing PDP is elusive. This condition is complex to diagnose because its symptoms often overlap with those of other illnesses, such as acromegaly, and its precise pathophysiology is still unknown^{4,5}. PDP should be taken into account by clinicians when treating individuals who present these specific features since it can aid in precise diagnosis and suitable therapy.

Here, without genetic testing, a genetic etiology is strongly suspected based on the clinical phenotype which is known to have variable inheritance patterns, including autosomal dominant, autosomal recessive, and incomplete forms. At present, the patient has exhibited only a limited subset of symptoms. With the possibility of phenotypic progression, the patient has been placed under close clinical surveillance to monitor for any evolution of the disease.

Conclusion:

Pachydermoperiostosis is a rare and under recognised condition appearing in a variety of ways. The necessity of a thorough clinical evaluation and the exclusion of secondary factors are highlighted by this instance. Clinical diagnosis is frequently made, particularly in cases when genetic testing is not possible. In this self-limiting illness, early detection and follow-up are essential for controlling progression and preventing pointless investigations.



(a)

(b)

Figure 1: Hyperhidrosis and grade 4 clubbing.

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