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Case Report

Progressive Clubbing and Periostosis in Primary Hypertrophic Osteoarthropathy: A Case Report

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Abstract

Background: *Primary hypertrophic osteoarthropathy (PHO), also known as pachydermoperiostosis, is a rare genetic disorder characterized by the triad of digital clubbing, periostosis of long bones, and pachydermia. It accounts for only 3–5% of all hypertrophic osteoarthropathy cases.*

Case Presentation: *We report the case of a 22-year-old male who presented with progressive clubbing of the fingers and toes, periosteal new bone formation causing painless bony swelling of multiple joints, and coarsening of facial skin (pachydermia) over two years. There was no evidence of underlying cardiac, pulmonary, or gastrointestinal disease. Imaging revealed diffuse periosteal thickening of the distal long bones. After exclusion of secondary causes, a diagnosis of primary hypertrophic osteoarthropathy was established.*

Conclusion: *This case highlights the importance of recognizing PHO in young patients with clubbing and periostosis to differentiate it from secondary causes and avoid unnecessary investigations.*

Keywords: *Primary hypertrophic osteoarthropathy; Pachydermoperiostosis; Clubbing; Periostosis; Pachydermia*

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Introduction

Hypertrophic osteoarthropathy (HOA) is a syndrome characterized by digital clubbing and periosteal reaction of long bones, often accompanied by arthralgia or arthritis. HOA may be secondary to underlying systemic disease or primary (idiopathic). Primary hypertrophic osteoarthropathy is rare, typically begins in adolescence or early adulthood, and more commonly affects males. The classical triad consists of pachydermia, periostosis, and digital clubbing.

Case Presentation

A 22-year-old Bangladeshi man presented with a 2-year history of thickening and furrowing of the skin over his forehead and scalp, along with 1.5 years of painless swelling of both knees and wrists and progressive clubbing of the fingers and toes. There was no history of fever, weight loss, chronic cough, dyspnea, cyanosis, or known cardiac disease. No family history of similar illness was reported.

On examination, pachydermia with prominent furrowing over the forehead and scalp was present. Grade 4 digital clubbing was present in both fingers and toes. Non-tender bony swelling of the knees and wrists was noted without joint effusion or restriction of movement. No lymphadenopathy, cyanosis, or signs of systemic disease were found. There were no features suggestive of acromegaly.

Investigations showed mildly elevated ESR and CRP. Chest X-ray and echocardiography were normal. Radiographs demonstrated periosteal thickening of long bones. MRI of the pituitary gland was normal. No evidence of underlying pulmonary, cardiac, or gastrointestinal disease was detected.

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Picture was taken in 2022

Current Picture

Figure 1: Facial profile showing pachydermia with forehead furrowing and scalp folds.

Based on clinical findings and exclusion of secondary causes, a diagnosis of primary hypertrophic osteoarthropathy was made. The patient was counseled regarding the benign nature of the condition and treated symptomatically with NSAIDs, with advice for regular follow-up.

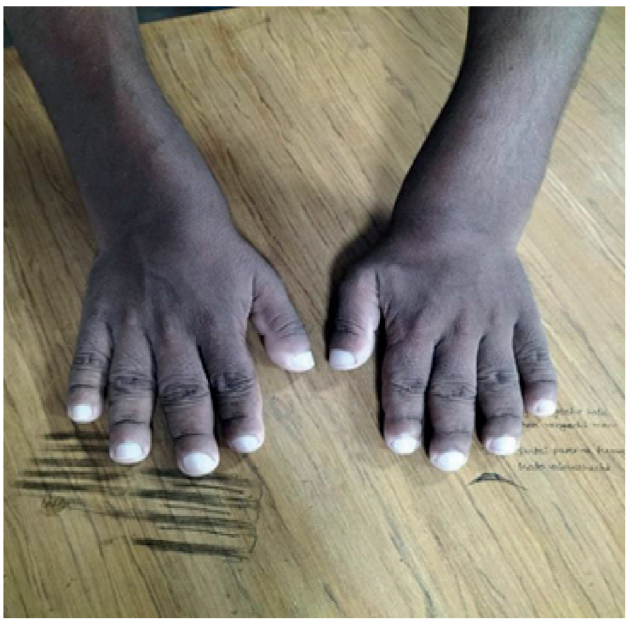

Figure 2: *Grade 4 digital clubbing of the fingers.*

Table 1. Key Investigations and Results

Investigation	Result
Complete blood count	Within normal limits
ESR, CRP	Mildly elevated
Liver & renal function tests	Normal
Thyroid function	Normal
Rheumatoid factor, ANA	Negative
Chest X-ray	Normal
Echocardiography	Normal
Pituitary MRI	Normal
X-rays of long bones	Periosteal thickening consistent with periostosis

Differential Diagnosis

Differential diagnoses considered included secondary hypertrophic osteoarthropathy due to pulmonary or cardiac disease, acromegaly, thyroid acropachy, chronic infection, and rheumatologic disorders. These were excluded based on normal cardiopulmonary evaluation, absence of systemic symptoms, and normal pituitary imaging.

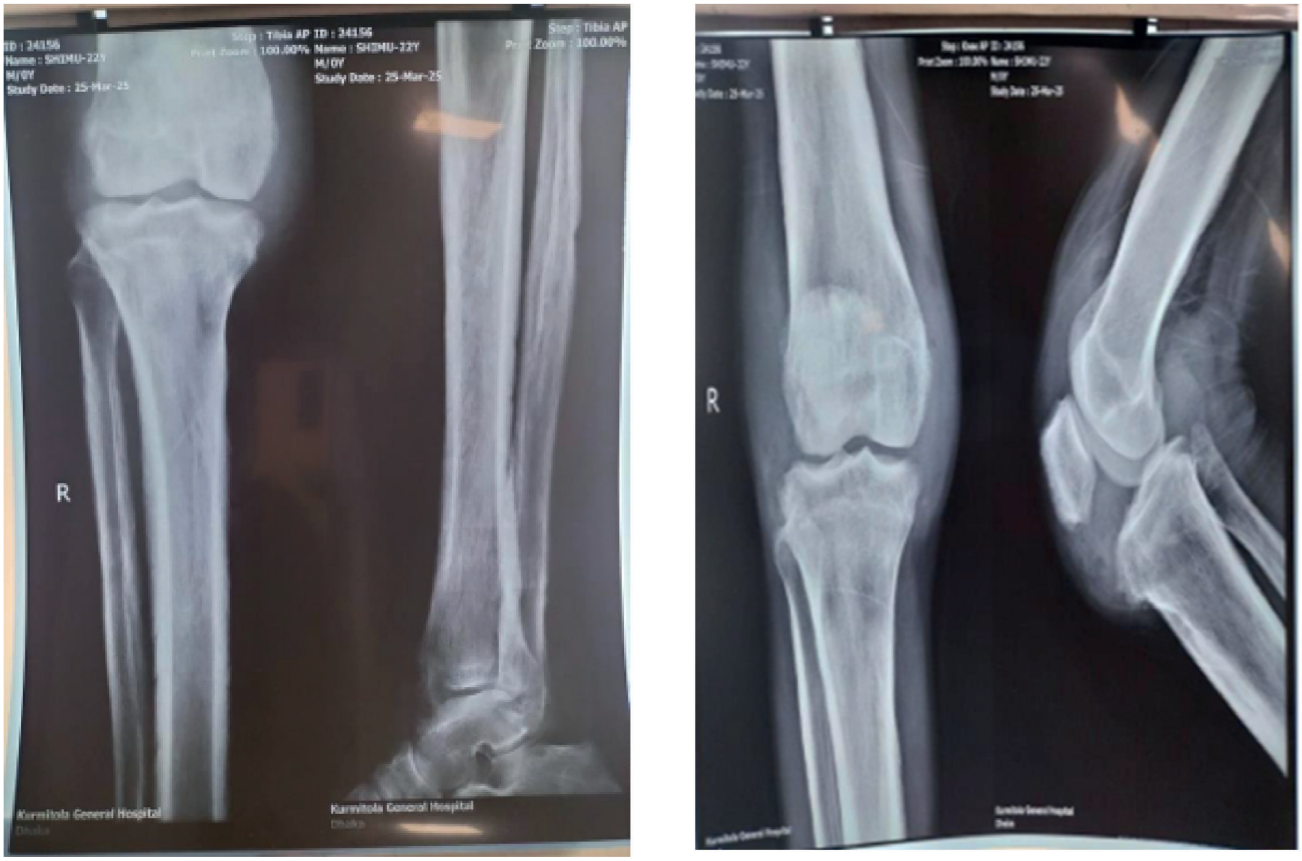

Figure 3: Radiograph demonstrating periosteal new bone formation and cortical thickening

Discussion

Primary hypertrophic osteoarthropathy is a rare hereditary disorder characterized by digital clubbing, periostosis, and pachydermia. It commonly presents in adolescence or early adulthood with male predominance. The disorder is linked to elevated prostaglandin E2 levels due to mutations in genes involved in prostaglandin metabolism (e.g., HPGD, SLCO2A1). Diagnosis requires recognition of clinical features and exclusion of secondary causes such as lung malignancy, chronic pulmonary disease, and cyanotic heart disease.

The exact incidence of PHO is not well established, but one study estimated its prevalence to be around 0.16% in the general population, highlighting its rarity. Clinically, PHO may present with varying degrees of the classic triad. Our patient exhibited the complete form of the disease, having all three major features (clubbing, periostosis, and skin changes). In contrast, some patients have an incomplete form (prominent bone and joint manifestations with minimal or no skin involvement), and a few have a fruste form (mostly skin changes with only mild skeletal findings. Historically, Touraine, Solente, and Golé in 1935 classified pachydermoperiostosis into these three forms. The complete form comprises roughly 40% of cases, the incomplete form about 54%, and the fruste (mild) form approximately 6%.

Genetic and Pathophysiological Insights: Primary hypertrophic osteoarthropathy is usually familial and can be inherited in an autosomal dominant or recessive manner. Research over the past decade has identified two key genes associated with PHO. Mutations in the HPGD gene (15-hydroxyprostaglandin dehydrogenase) cause an autosomal recessive form of PHO (sometimes referred to as PHOAR1). HPGD encodes an enzyme responsible for the degradation of prostaglandin E, (PGE,). Loss-of-function mutations in HPGD lead to elevated levels of PGE, in the body, which is thought to drive the periostosis and other features of PHO by stimulating bone formation and vascular endothelial growth factor (VEGF) production. Another set of mutations occurs in the SLCO2A1 gene, which encodes a prostaglandin transporter; these mutations cause another form of PHO that can be autosomal recessive (often termed PHOAR2) or even a milder autosomal dominant variant (PHOAD). SLCO2A1 mutations are particularly associated with prominent skin manifestations and gastrointestinal involvement in PHO, and

many reported cases from East Asia (especially China and Japan) have been linked to SLCO2A1 defects. Our patient's clinical profile (male, early adulthood onset, classical triad) is consistent with these genetic forms, though genetic testing was not performed in our setting. In a recent case report from China, a young male with similar features was confirmed to have compound heterozygous mutations in SLCO2A1. Genetic testing can thus be used to definitively diagnose PHO and differentiate between HPGD-related and SLCO2A1-related subtypes, which may have implications for family counseling. Diagnosing PHO requires a high index of suspicion, especially once more common causes of clubbing have been ruled out. In practice, the diagnosis is made by recognizing the combination of clubbing, periostosis, and typical skin changes, and by excluding secondary etiologies.

Management:

There is no definitive cure for primary hypertrophic osteoarthropathy; management is mainly supportive and symptomatic. The arthralgia or bone pain in PHO can be alleviated with NSAIDs (nonsteroidal anti-inflammatory drugs), which may also inhibit prostaglandin E, synthesis (thus targeting the mediator believed to drive the disease). In some cases, coxib drugs (selective COX-2 inhibitors such as etoricoxib or indomethacin) have been reported to reduce periostosis and improve joint symptoms, presumably by reducing PGE, levels. Another therapeutic approach reported in the literature is the use of bisphosphonates – for example, pamidronate or zoledronic acid – which can help with bone pain and may slow the progression of periosteal bone formation. A recent case report of an adolescent with PHO demonstrated significant improvement in joint pain and swelling after treatment with intravenous zoledronic acid, along with NSAIDs. No disease-modifying therapy is yet established for PHO; however, some studies have explored agents targeting bone turnover or vascular factors (for instance, tamoxifen or interferon-α in isolated cases), but with limited evidence of benefit. For cutaneous manifestations, particularly severe pachydermia of the forehead and scalp, plastic surgery or dermatologic treatments (e.g. retinoids) may be considered to improve cosmetic appearance. Excessive sweating (hyperhidrosis) can be managed with topical therapies or medications like anticholinergics if it causes discomfort. In our patient, the disease was relatively early in its course and primarily causing cosmetic and mild musculoskeletal symptoms; thus, we opted for NSAIDs for pain relief and observation. We educated him about the benign nature of PHO – unlike secondary HOA, it is not indicative of an internal malignancy – and emphasized the importance of follow-up to monitor disease progression or any emerging complications.

Conclusion

Primary hypertrophic osteoarthropathy is an uncommon but distinctive cause of clubbing and periostosis that clinicians should keep in mind. This case illustrates the classical presentation of PHO with digital clubbing, periosteal new bone formation, and skin changes in a young adult with no underlying systemic disease. Early recognition of this idiopathic condition is important to differentiate it from secondary hypertrophic osteoarthropathy and other mimics such as acromegaly. Establishing the correct diagnosis helps avoid extensive investigations and inappropriate treatments. Although PHO is a benign (non-life-threatening) condition, its progressive nature can impact the patient's quality of life; hence, supportive management and regular follow-up are advised.

Acknowledgments

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Patient Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images.

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