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Case Report

# Primary Hypertrophic Osteoarthropathy in End Stage Renal Disease Patient with Marked Anemia. A Case Report

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## Abstract

Primary or idiopathic hypertrophic osteoarthropathy also known as pachydermoperiostosis is a rare disease characterized by clubbing of the digits, enlargement of the extremities and synovial joint effusions and a variety of skin abnormalities resulting from cutaneous hypertrophy. Other associated features are hypertrophic gastropathy, peptic ulcer and gynecomastia. Eyelid ptosis which is caused by thickened eyelids (blepharoptosis) is a less common association with this disease. We report a rare case of a patient with a complete form of pachydermoperiostosis with bilateral ptosis, clubbing, other skin manifestations and marked erythropoietin resistant anemia.

**Keywords:** Primary Hypertrophic Osteoarthropathy (PHOA); Clubbing; Skin Abnormalities; Erythropoietin

## Introduction

Primary hypertrophic osteoarthropathy also known as primary pachydermoperiostosis or Touraine- Solente- Gole syndrome was initially described by Friedreich in 1868 and letter on by Touraine., *et al*. in 1935 who classified this condition as familial disorder with three forms: complete (periostosis and pachyderma), incomplete (without pachyderma), and forme fruste (pachyderma with minimal skeletal changes). This rare disease occurs in absence of any other condition involving other systems of the body [1].

This disease is a familial or idiopathic and account for 3% - 5% of all cases of HOA and affects persons of all races. In Arab world we found 18 cases of primary hypertrophic osteoarthropathy distributed in different regions and this is the very rare case we are reporting from Saudi Arabia. This disease has an autosomal dominant pattern of inheritance but autosomal recessive and X- linked mutations may also be present. This disease has a bimodal peak of onset that occurs in patient younger than one year and in patients who are around puberty, i.e., approximately age of 15 years [2].

HOA has a marked predominance in males, with male to female ratio of 9:1. This disease is described in many races and precise incidence of this syndrome is unknown but estimated prevalence of the disease is 0.16% [3]. There is no current treatment available for disease itself but we can manage certain complications of disease effectively by medical or surgical means. We report a case of complete HOA with marked anemia that presented in our out-sourcing dialysis center.

## Case Report

A 41- year-old man recently diagnosed as End Stage Renal Disease secondary to chronic glomerulonephritis with no family history of note, was found to have marked digital clubbing, profuse sweating of palms and soles, progressive enlargement and widening of hand fingers, wrist, toes, feet, ankle joints with swelling and ptosis. He also has thickening of skin on face and scalp.

Physical examination revealed pallor, advanced clubbing of all fingers, enlargement of hand and wrist joints without signs of

inflammation. There was also clubbing of toes, widening of feet ankles and knee joints. He has thickening of forehead fold with evidence of pachyderma. Cardiopulmonary and abdominal examination were unremarkable. He received high doses (12000 three times/week post dialysis) of Erythropoietin injection to improved his hemoglobin levels but failed to achieved his target levels.

Laboratory results reveled mean hemoglobin levels 7 gram/dl (10-12 gram/dl in dialysis population), total leukocytes count 7000/$mm^3$ (4000-11000), platelets 280x$10^9$/l, Serum ferritin 600ng/ml with transferrin saturation of 30%, blood urea 45mmol/L (2.6-7.5), serum creatinine 900mmol/l (less than 88), parathyroid hormone level 500 pg/ml, serum bicarbonate 23mmol/l (22-28), serum potassium 4 mmol/l (3.5-5.1). liver function test, serum albumin, C reactive protein, ESR, rheumatoid factor, antinuclear antibodies and blood cultures within normal limits. His chest X- ray, electrocardiogram and echocardiogram were normal.

## Discussion and Conclusion

Hypertrophic osteoarthropathy (HOA) manifests as changes in the bone and skin. It is characterized by digital clubbing, arthralgia, periostitis and subperiosteal new bone formation in tubular bone. It could be either primary or secondary. The primary familial form is also known as pachydermoperiostosis, is hereditary and manifested in different ways. Although the exact prevalence is unknown, one study reported a prevalence of 0.16%. it is more commonly present in male population as compared to female (9:1). and is considered to be familial (25-40% of cases) [4], but in our case no family member of this patient has similar disease.

The secondary form of HOA can be caused by variety of systemic diseases including cyanotic heart disease, cystic fibrosis, bronchiectasis, tuberculosis, hepatic disease gastrointestinal diseases including inflammatory bowel disease, and certain malignancies (Hodgkin lymphoma, nasopharyngeal carcinoma, and chronic myeloid leukemia) [5]. the exact etiology of HOA is unknown, but possible mechanisms have been suggested. The increase or decrease in cytokines such as prostaglandin E2 have been suggested hormonal, immunological, neurological and thrombotic mechanisms [6].

In primary form, mutation in HPGD encoding 15 hydroxy prostaglandin dehydrogenases, whose enzyme activity is NAD+ dependent has been shown to be responsible for this syndrome [7].

Primary hypertrophic osteoarthropathy has a bimodal presentation, with one peak in the first year of life and another at age 15 years. It develops gradually from adulthood. It usually begins as clubbing during adolescence, followed by progressive changes in the skeleton and skin over the next 5-20 years. The diagnosis of HOA is based on the presence of at least two of the four criteria set by Borochowitz- a history of familial transmission, pachyderma, digital clubbing and skeletal manifestations such as pain or sign of radiographic periostitis [3]. Our patient has complete form of the syndrome with the presence of most of the clinical features and radiological findings.

The most common clinical symptoms of PHOA include clubbing (89%), radiographic periostitis of the distal long bones periostosis. Swelling of periarticular tissue and subperiosteal new bone formation reveled by radiography (97%), pachyderma, coursing of skin, seborrhea (90%), acne folliculitis, palmo-planter hyperhidrosis (44-67%), partial ptosis and cutis vertices gyrata (42%) [8]. Anemia is such patient is usually multifactorial caused by gastrointestinal bleeding, myelofibrosis and serum inhibitors of erythropoiesis [9]. In our ESRD patient we thoroughly investigated causes of resistant anemia (Acute/chronic blood loss, iron and vitamin deficiency, infections, inflammation, secondary hyperthyroidism, any obvious malignancy etc.) but could not able to find any definitive cause although he was on maximum doses of intravenous erythropoietin injection i.e. 450 iu/kg/week.

The diagnosis of PHOA is clinical on the basis of digital clubbing and distal periostitis of long bones associated with pachyderma. There is no definitive diagnostic test for this condition. X rays of tubular bones reveal periosteal new bone formation which supports the diagnosis.

The medical treatment of PHOA is symptomatic. Non-steroidal anti-inflammatory drugs (NSAIDs), corticosteroids, tamoxifen citrate, risedronate and colchicine used to relieve pain [10]. This condition is associated with significant reduction in quality of life however, prognosis is good.

## Conflict of Interest

None declared.

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