Touraine Solente Gole Syndrome – The disease and associated Tongue fissuring

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INTRODUCTION
Pachydermoperostitis (PDP) defined by the presence of digital clubbing, pachyderma (thickening of the facial skin and/or scalp), and periostosis (swelling of periarticular tissue and subperiosteal new bone formation) is a rare, inherited, familial disorder first described in 1868 and later characterized by Touraine, Solente and Gole in 1935 to represent the primary form of hypertrophic osteoarthropathy (HOA)[1]. The precise incidence of this osteodermopathic syndrome is unknown[1]. Inherited in an autosomal dominant fashion with variable expression and penetration three forms of PDP have been recognized[1]. A complete form which includes clubbing, pachyderma and periostosis, an incomplete form lacking the dermatological manifestations, and a forme fruste with minimal-to-absent skeletal changes. PDP begins as clubbing usually during adolescence, followed by progressive changes in the skeleton and skin over the next 5-20 years, resulting in significant morbidity and then remains unchanged for life[1]. PDP must be recognized early because of the social stigmata linked to its cutaneous manifestations[2] and its uniformly good prognosis if treated[3]. We herein report a case of primary HOA with thickened fissured tongue, a yet un-described manifestation of the disease and discuss the social cum clinical relevance of identifying this relatively uncommon disorder.

CASE REPORT
A 30 year old male with a history of tuberculosis 7 years back presented with cough and mucopurulent sputum production for the past 1 year which had increased over the past week. On examination he showed thickened, furrowed, greasy skin of the face (seborrhea), accentuation of facial folds, hypertrophy of eyelids and bilateral mechanical ptosis (Fig.1). The tongue was fissured similar to a scrotal tongue (Fig.2). Scalp was grooved and thickened typical of cutis verticis gyrata. Clubbing was noted in the digits and toes (Fig.3,4) accompanied by swelling of the knee joints bilaterally and distal aspect of both lower limbs in a columnar fashion (Fig.5) along with plantopalmar thickening. A lipoma was noted on the back. Auscultation of the chest revealed crackles with scattered rhonchi over the entire right and left lower hemithorax respectively. Examination of other systems was unremarkable. There was no history of frequent colds, nasal secretion, respiratory allergies, or recurrent pneumonic infections. Questioning, further revealed the presence of similar clubbing sans other florid manifestations in his sister. Both developed their respective manifestations insidiously around the age of 15 to 17 years.

Investigations revealed a near normal hemogram except for a slight raise in the WBC count (n=15,000 cells/mm3) with predominant neutrophils (84%). Gram staining of the sputum showed predominant gram negative organism and E.coli was grown on culture. Chest radiography showed reticulonodular opacities with ectatic changes encompassing the right upper and middle zones. Honeycombing of the right upper and middle lobes with thin walled sacculations involving the left upper lobe - changes consistent with cystic bronchiectasis, were noted on HRCT.
X ray films of the wrist and ankles showed soft tissue swelling with irregular periosteal proliferation cum cortical thickening of the long bones ,metacarpals(Fig.6) and metatarsals and acroosteolysis ( Fig.7). Xray examination of the knees (Fig.8) revealed periostosis of the long bones along with increased joint space suggestive of knee effusions.

ESR, C-reactive protein (CRP), blood chemistry (electrolytes, liver enzymes, BUN, creatinine, uric acid, glucose, proteins, lipids, thyroid hormones, growth hormone, PTH, iron levels and immunoglobulins) and semen analysis were normal . Sputum AFB, blood cultures, sweat chloride test, RPR, VDRL,ELISA for HIV, ANA, anti ds DNA and rheumatoid factor were negative. A non inflammatory synovial fluid with neutrophil count < 500 /mm3 were obtained on knee joint aspiration.

Quantitative α₁-antitrypsin levels, delta F508, transepithelial nasal potentials and bone scintigraphy could not be carried out due to economic constraints.

Secondary causes having been excluded primary HOA or Touraine Solente Gole syndrome with bronchiectasis secondary to tuberculosis was diagnosed.

Intravenous antibiotics (cefotaxime/gentamicin) for ten days followed by an oral antibiotic (Amoxycillin) for a month, bronchodilators and chest physiotherapy resulted in significant improvement of the respiratory symptoms. No specific therapy was instituted for the osteodermopathic manifestations of primary HOA as they were refused by the patient.
Figure 3
Figure 3- Clubbing of digits

Figure 4
Figure 4 – Clubbing of toes

Figure 5
Figure 5- Swelling of the distal aspect of both lower limbs in a columnar fashion

Figure 6
Figure 6- Cortical thickening of metacarpals
DISCUSSION

Toraine Solente Gole syndrome is a diagnosis of exclusion characterized by cutaneous and skeletal manifestations of HOA sans the inciting pathosis seen in secondary forms[1]. Having excluded alternate causes in our patient a diagnosis of primary HOA or Toraine Solente Gole syndrome was made. Bronchiectasis in the patient was an incidental occurrence secondary to Tuberculosis and not the primary inciting event as evidenced by the time of onset of symptoms. The prominent cutaneous manifestations in the form of thickened, furrowed, seborrhic skin of the face, accentuation of facial folds, hypertrophy of eyelids and bilateral mechanical ptosis in conjunction with absent skeletal tenderness, slow insidious onset of bone changes and a family history of clubbing in his sister further support the diagnosis of primary HOA[1].

An interesting observation in our case was the presence of a thick fissured tongue. This association of a thick fissured tongue with primary HOA has not been reported earlier to the best of our knowledge. Tongue fissuring may be an independent manifestation of primary HOA or incidental finding. Both the conditions have a familial clustering[1,4] with an autosomal dominant pattern of inheritance[1,5]. The rare association may be attributed to the vascular pattern of the tongue which may prevent the lodging of megakaryocytes as opposed to their preferential lodging in the digits which has been proposed as a plausible theory for skin thickening in HOA[1]. Tongue fissuring could also be an incidental finding in our case with no relation to HOA as 1-5% of the population have fissured tongue with no clinical significance[4]. A systematic study of a larger population of patients with primary HOA is required before any conclusions can be drawn.

Yet another rare but not unknown manifestation in our case was acroosteolysis of distal phalanges of the fingers[6]. Clubbing in conjunction with osteolysis helps to differentiate primary HOA from all other causes of acroosteolysis except Cheney’s syndrome[6].

Familial clustering of PDP is found in 25-38% of cases with a male to female ratio of 9:1[1]. Males are more severely affected [1]. The truncated manifestations of PDP in the patients sister point towards the possibility of an X linked pattern of inheritance[7].

The etiology of primary HOA is still unclear with two widely floated theories a) Neurogenic[1,8]- neural reflexes initiated by vagal stimulation lead to vasodilation, increased blood flow, HOA and b) Humoral[1,8] - mediators which include various growth factors like platelet derived growth factors, epidermal growth factor, transforming growth factor and vascular endothelial growth factor have been found to be increased in patients with HOA leading to fibroblast proliferation and subsequent fibrosis.

PDP being a relatively uncommon condition, the disfigurements in facial and skeletal appearances have been confused with leprosy and syphilis[2]. These two conditions have widespread psychological and social problems especially in developing countries making the differentiation from HOA imperative[9].

Pain in PDP has routinely been dealt with NSAID’s[1]. Colchicine inhibits neutrophil chemotaxis and tissue edema
thereby improving joint symptoms, pachyderma and folliculitis[1]. Steroids and pamidronate by their inhibition of osteoclasts and antiresorptive effects have been tried with promising results for rheumatological symptoms of HOA[1,10]. Retinoids by decreasing procollagen mRNA in fibroblasts have shown improvements in pachyderma, seborrhea, acne, folliculitis and cutis verticis gyrata[1].

CONCLUSION

Clinicians need to be aware of primary HOA because of improved outcomes with early treatment and also to avoid a misdiagnosis of syphilitic perostosis or leprosy which have widespread social implications in developing countries. The appearance of thick tongue with fissuring may be an independent manifestation of Toraine Solente Gole syndrome.

References

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