Case Report

Pierre Marie-Bamberger Syndrome: A unique case report

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Abstract

Hypertrophic osteoarthropathy (HOA) was described by Friedreich in 1868. It is a rare condition with variable presentations including clubbing of the toes and fingers, arthralgia with edema, bilateral ptosis, thickening of the skin and leonine facies. Bone and periarticular tissue proliferation leads to expansive extremities. It is a distant effect disorder in various neoplasms (paraneoplastic syndrome); often associated with lung neoplasm. We report a case of a 36 year old Pakistani male smoker, presenting with bilateral joint pains, shortness of breath and grade 4 digital clubbing on general examination.

Introduction

Hypertrophic osteoarthropathy (HOA) clinically presents with clubbing of the fingers and toes, increase in the size of extremities, arthralgia and swelling in the joints. A characteristic feature of HOA is symmetric periostitis which mainly affect the long bones of both upper and limbs. The disease can be either primary (pachydermoperiostosis) or secondary. The primary form, or idiopathic, is a rare familial autosomal dominant disease, corresponding to 3-5% of all cases. In majority of the cases, the disease is secondary to a pulmonary tumour, hence called hypertrophic pulmonary osteoarthropathy (HPO)[1, 2]. We present a case of 36 years old Pakistani male, who presented with generalized malaise and lethargy. The patient complained chronic pain in joints of hands and axial skeleton and was later diagnosed to have non-small cell carcinoma, squamous cell cancer of lungs.

Case Report

We here in present a case of a Pakistani 36 year's old male, presenting in clinic with generalized malaise and lethargy. Patient was stable with BP 130/90, pulse rate 74/min, respiratory rate of 16/min and afebrile. Patient gave history of bilateral chronic pain in joints of hands and axial skeleton. Pain in his hand joints started almost 5 years back, dull aching in character without any morning stiffness, worse on movement, relieved on taking acetaminophen and rest. There was swelling associated with the joint pain but no fever or chills. Patient also reported of exacerbation of pain in joint of hand on motion and had been consuming over the counter painkillers for these arthritis like symptoms for last 5 years on and off. He also complained of shortness of breath and occasional non-productive cough. He is known smoker for past 16 years with history of 24 pack years and has complained of chronic bronchitis for many years. There was also a positive family history of lung cancer.

On general examination, a grade 4 clubbing of the digits was seen with excessive proliferation of skin. There was visible swelling of small joints of hands and elbows with no calor, warmth (a cardinal feature of inflammation) on palpation. There was severe pain on movement of interphalangeal joints. Initially a provisional diagnosis of degenerative joint disease was made, although a positive family history of lung cancer and smoking habit, a paraneoplastic syndrome was considered a differential for diagnosing the case. Patient was referred to radiologist for the radiographs of hands and the chest. The hand radiograph showed periosteal reaction around the shafts of all visible bone, (Figure 1).

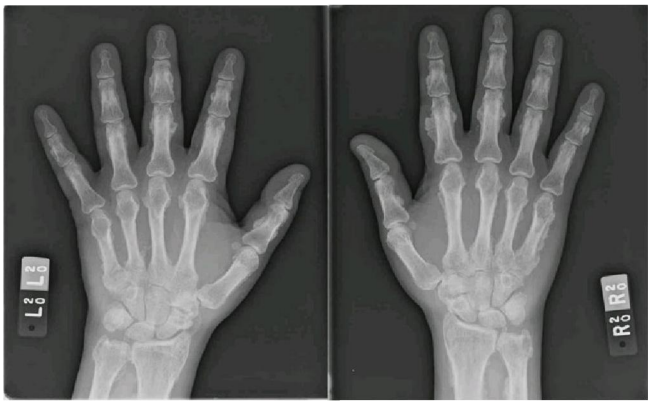

Figure 1. X-ray bilateral hand PA view. Periosteal reaction is seen around the shafts of all visible bones with no cortical break. No fracture is seen. Findings are consistent with hypertrophic pulmonary osteoarthropathy.

The chest radiograph showed a right mid-zone focal lesion, suggestive of malignancy (Figure 2). The lung biopsy was done, which showed a non-small cell carcinoma, squamous cell cancer. A diagnosis of Pierre Marie-Bamberger Syndrome, rare condition was made. Patient was advised to continue NSAIDS for symptomatic management and was referred to surgery department for treatment of primary cause.

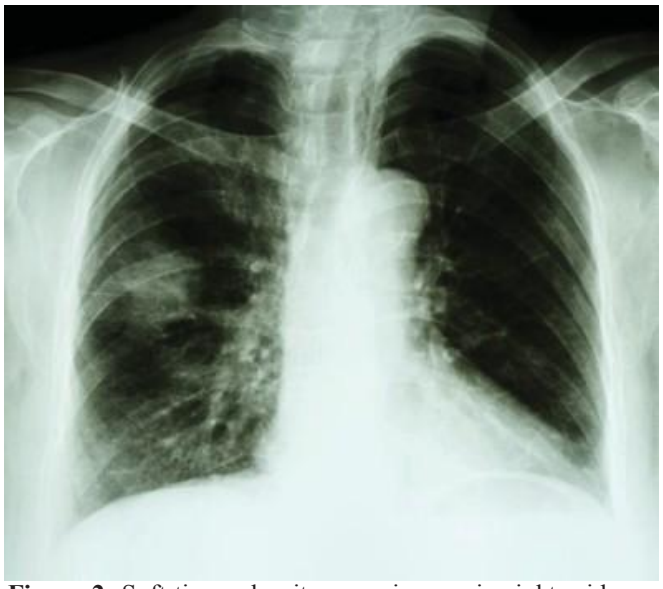

Figure 2. Soft tissue density mass is seen in right mid zone with irregular indistinct margins suggestive of malignancy.

Discussion

HOA is a syndrome manifesting bony deformities and multi-organ involvement[3]. The major work on the syndrome is done in 1889 by Bamberger followed by further research by Pierre Marie in 1890, hence the name Pierre Marie–Bamberger (PMB) disease was coined[4]. PMB is a rheumatologic disease with variable presentations. The common features include digital clubbing, increase in size of the limbs secondary to bone and periarticular tissue proliferation, arthralgia with edema, bilateral eyelid ptosis, leonine facies, and thickening of the skin. Histological examination of tissue biopsy shows hyperplasia of the adjacent subcutaneous tissue[5, 6]. PMB is etiologically classified into primary (hereditary or idiopathic) or secondary disease. Touraine et al divided the primary disease into three forms: complete, incomplete, and fruste form. The complete and fruste variety both present with pachydermia along with skeletal abnormalities. However digital clubbing and periostosis is seen in complete form[7]. Incomplete form cases do not exhibit pachydermia. Secondary HOA is associated with an underlying multiorgan involvement and often has a fatal course. HPO is an uncommon paraneoplastic syndrome which is frequently associated with pulmonary tumor; the actuarial incidence of HPO is, however, not well known[8].

Our patient in the reported case initially presented with bilateral joint pains in the hands with increased pain on movement. He was on over the counter pain medications for chronic arthritis. On general physical exam he had grade 4 clubbing in hands, thickened skin, edema and joint pain on movement of the hands which are all features of HPO. Considering shortness of breath, smoking history and digital clubbing in our reported case we suspected Hypertrophic Osteoarthropathy (HPO) secondary to lung pathology and sent the patient for chest X-ray. Further tests and biopsy revealed lung carcinoma.

Simple clubbing of the fingers should be differentiated from osteoarthropathy. The preliminary alteration in the architecture of limbs of patients with pulmonary HPO is an overgrowth of vascular connective tissue involving structures in the distal part of the limb. The new tissue formed lies over the periosteum and osteogenesis takes place beneath it. Surrounding the joints, the newly formed tissue gives the appearance of periarthritis, though there are no specific articular changes. The nail beds invasion gives rise to clubbing of the fingers. Osteoarthropathy is commonly secondary to pulmonary disease, but it may be associated with thoracic lesions without pulmonary involvement[9].

In the end summarizing the treatment of secondary hypertrophic osteoarthropathy (HPO) we consider the option of treating underlying primary cause like resection of tumor, chemotherapy, radiotherapy etc. and supportive management including bisphosphonates, NSAIDS and vagotomy[4].

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