Case Report

Melancholic Face Since Puberty; A Rare Case Report Of Primary Hypertrophic Osteoarthropathy

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Abstract

Pachydermoperiostosis or Touraine-Solente-Gole syndrome is a rare genetic disorder that follows autosomal dominant pattern of inheritance, wherein males are more commonly affected than females. The prevalence of the disease in the general population is about 0.16%. It is characterized by diffuse thickening of skin and subcutaneous tissue of distal extremities, coarsening of facial features, spade like enlargement of hand with clubbing of digits and excessive sweating. We hereby present a rare case report of pachydermoperiostosis with little skeletal involvement. The diagnosis was made by considering the clinical appearance of patient and ruling out other causes of hypertrophic osteoarthropathy. Primary hypertrophic osteoarthropathy constitutes about only 5% of all cases of hypertrophic osteoarthropathy; lack of awareness of this entity can lead to confusion and misdiagnosis.

Keywords: puberty,case report

Introduction

Pachydermoperiostosis or primary hypertrophic osteoarthropathy (PHO) or Touraine-Solente-Gole syndrome is a rare genetic disease predominantly affecting males with M:F ratio of 9:1(1). It follows autosomal dominant pattern of inheritance with variable penetration (2). Jajic et al in 1992 concluded that the prevalence of the disease in the general population is about 0.16% (3). It is thought to be due to homozygous mutations in the 15-hydroxyprostaglandin dehydrogenase gene. This is based on study conducted by Yuksel et al on six patients from three unrelated Turkish families with PHO (4). It was first described by 3 distinctive dermatologists, Touraine, Solente and Gole, as an inherited disorder, in 1935. Based on severity of skeletal involvement it was classified into 3 types i.e. (i). complete or classical type with periostosis and pachyderma, (ii). incomplete type without pachyderma, and (iii). the forme fruste variant with pachydermia with minimal skeletal changes (5).

Case Report

A 35-year-old male, from Odisha state of India, born out of second-degree consanguineous marriage presented to our out-patient department with chief complaints of exaggerated skin folds over face, heavy drooping lids and abnormal contour of finger tips and nails for the last 20 years. The patient also complained of shortness of breath, profuse sweating, progressive enlargement of hands and feet. He also had several episodes of vomiting and intermittent abdominal pain for which endoscopy was done and it showed erosive gastropathy. Patient did not have fever, joint pain, weight loss, photosensitivity, burning micturition, genital discharge, headache, oral ulceration and cough. He denied history of exposure. There was also no history of similar complaints in patient's family.

On examination there were marked skin folds on forehead, face, and eyelids. Clubbing of all the digits was also evident. Schamroth's sign was positive. General physical examination was otherwise unremarkable. Cardiovascular, respiratory, neurological, and per abdominal examination performed was also normal.

He was subsequently investigated extensively for acromegaly. Insulin-like growth factor-1 level, Serum growth hormone levels and Oral glucose tolerance test (OGTT) were normal. Thyroid function test was performed to rule out thyroid acropachy, it was found to be within normal limits. Rheumatoid factor and anti-cyclic citrullinated peptide were negative. Skin biopsy was performed from forehead area which showed no abnormality.

Complete blood count showed decreased haemoglobin level of 5 mg/dl for which multiple blood transfusions were done. Bone marrow aspiration revealed hypercellular marrow with erythroid hyperplasia with micronormoblastic erythropoiesis.

The x-rays of bilateral hands and feet revealed mild periostosis and hyperostosis of metacarpal and proximal phalanges. There was also generalised decreased bone density noted in bilateral feet. Soft tissue tumefaction was also seen especially in the distal phalanges of both hands and feet. Chest X-ray was normal. X-ray of head and neck revealed soft tissue tumefaction and was otherwise unremarkable.

Discussion

Hypertrophic osteoarthropathy is mainly described as two types based on aetiology i.e. primary and secondary forms. Pachydermop-eriostosis or the primary hypertrophic osteoarthropathy is an inherited disorder of unknown aetiology. It accounts for 5% of all cases of hypertrophic osteoarthropathy (6). The condition starts to appear at puberty and progresses insidiously over 5-10 years to involve skin and skeleton and remain unchanged throughout life (2). Due to the variable penetration of genes, the age and complaint of presentation varies based on severity of disease. In case of severe form, the patient

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presents in adolescence with complaint of joint pains. However, in mild form of disease, as in our case, the patient presents in third to fourth decade of life with skin changes and clubbing.

Other systemic involvement is usually absent in primary hypertrophic osteoarthropathy. The diagnosis is made based on clinical findings such as thickening of skin, periostosis and clubbing of digits and ruling out other systemic diseases such as pulmonary, cardiac, gastrointestinal, hepatobiliary, thyroid disorders or haematological malignancies that cause secondary hypertrophic osteoarthropathy (7). Therefore, pachydermoperiostosis is diagnosis of exclusion.

The coarse facial features of pachydermoperiostosis are also seen in acromegaly and scleromyxedema. Absence of clubbing in latter two differentiates it from pachydermoperiostosis (8). In case of thyroid acropachy, features of hyperthyroidism are present; wrist and ankle joints are not involved. Other systemic involvements such as congenital heart disease, carcinoma, bronchopulmonary suppuration etc also needs to be ruled out before making the diagnosis of primary hypertrophic osteoarthropathy. However, secondary hypertrophic osteoarthropathy also has subtle radiological evidence of new bone formation with fine opacities seen only in diaphysis (9).

We hereby report a rare case 35-year-old male with of forme fruste variant (type 3) of pachydermoperiostosis. All clinical and laboratory investigations were suggestive of PHO. Patient also had erosive gastropathy, severe anaemia with bone marrow failure which is in concordance with findings of favus et al (10). Patient was concerned about the sombre look on his face, thereby he was referred to plastic surgery department for the corrective procedure. Genetic counselling was done and he was called for follow up to look out for extensive skeletal involvement.

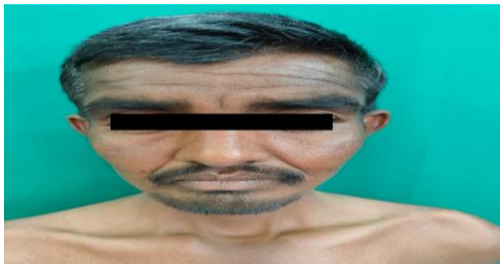

Figure 1: Anterior view of face showing worried look of face due to exaggerated skin folds

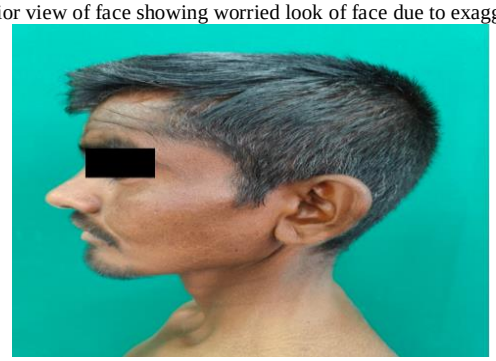

Figure 2: Lateral view of face showing exaggerated normal skin folds

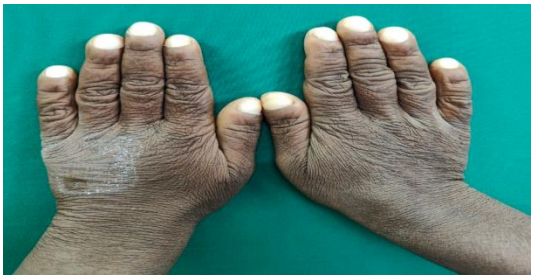

Figure 3: Grade 4 clubbing in all fingers of hand

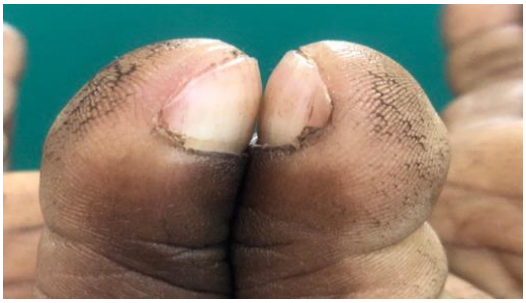

Figure 4: Schamroth's sign- positive

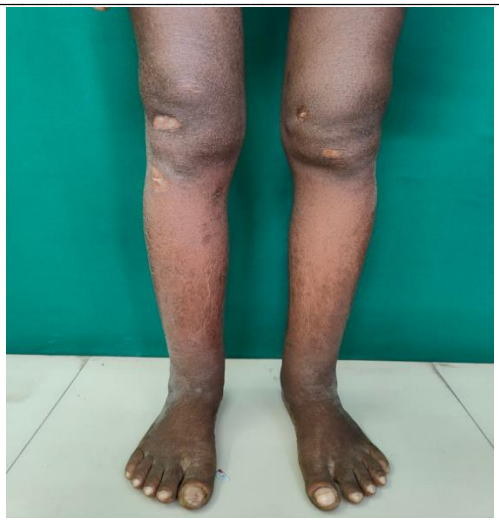

Figure 5: Slight swelling at knee joint and clubbing of all toes

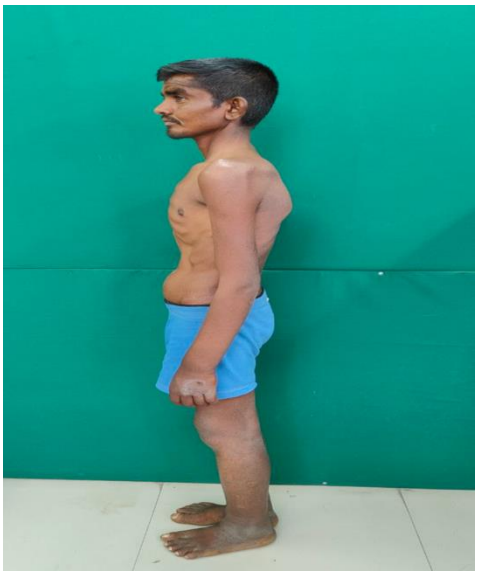

Figure 6: Side view of patient showing mild swelling at wrist, knee and ankle joint

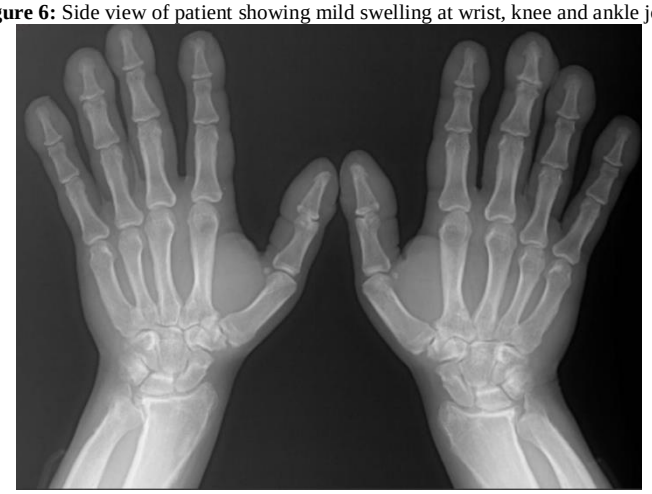

Figure 7: X-ray of hand showing soft tissue tumefaction and mild periostosis at distal phalanges

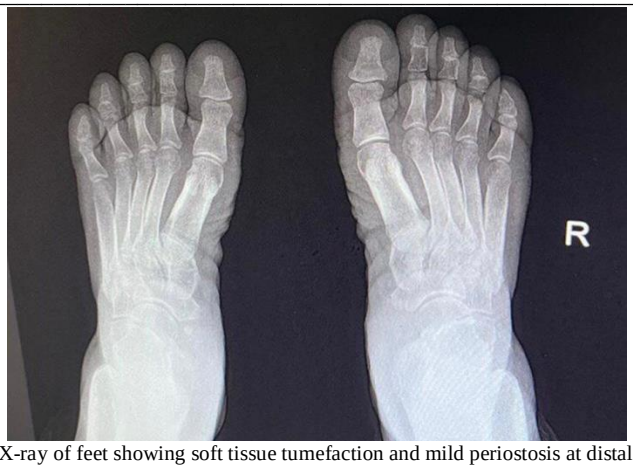

Figure 8: X-ray of feet showing soft tissue tumefaction and mild periostosis at distal phalanges

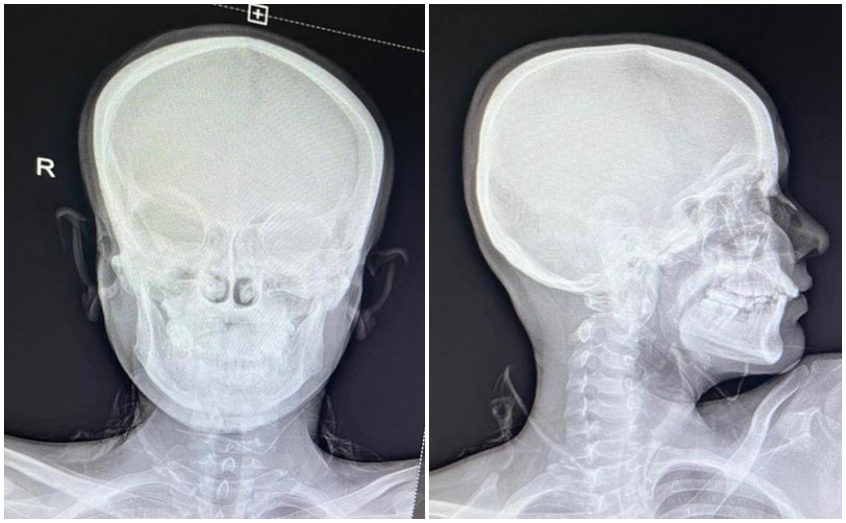

Figure 9: X-ray of head and neck-AP and lateral view showing soft tissue tumefaction

Conclusion

Pachydermoperiostosis is a rare entity. There is limited literature on its prevalence and maybe overlooked due to lack of knowledge about this entity.It forms an important clinical differential diagnosis for hypertrophic osteoarthropathy. it is usually missed due to its variable presentation.

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