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Case Report

Touraine Solente Gole syndrome: The elephant skin disease

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ABSTRACT

Touraine Solente Gole syndrome is a rare hereditary syndrome of primary pachydermoperiostosis, with the characteristic triad of pachydermia (or elephant like skin), periostosis and acropachia. A 27-year-old patient presented with aesthetic deformity of forehead due to deep skin folds and coarsening of facial features due to progressive thickening of skin. Associated palmoplantar hyperkeratosis with broadened of finger and toe tips and digital clubbing were noticed. Dermatologic evaluation revealed cutis verticis gyrata of scalp, seborrhoeic hyperplasia of face and hyperhidrosis. Natural history of the disease and aetiopathogenesis were reviewed. Aesthetic correction of forehead through frontal rhytidectomy was attempted.

KEY WORDS

Clubbing; cutis verticis gyrata; frontal rhytidectomy; hypertrophic osteoarthropathy; pachyderma; periostosis

INTRODUCTION

Pachydermoperiostosis is a rare hereditary syndrome characterised by primary idiopathic hypertrophic osteoarthropathy (HOA) associated with thickened skin of face, seborrhoea and hyperhydrosis. The term 'cutis verticis gyrata' refers to an associated folded hyperplasia involving scalp skin. The disease process usually begins during adolescence with gradual thickening of facial skin, which ultimately resembles that of an elephant (pachyderm) and hence the name. A diagnosis of Touraine Solente Gole syndrome can be entertained only after various causes of secondary HOA are carefully excluded, since it accounts for only 3-5% of all HOA. Close follow-up of these patients for long-term complications like arthritis and myelofibrosis is recommended.

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CASE REPORT

A 27-year-old male presented with complaints of progressive thickening of facial skin for past 5 years. Thickened forehead skin subsequently developed deep transverse furrows [Figure 1]. Associated broadening of fingers and toes with painless clubbing of nails were noticed [Figure 2]. Recurrent episodes of pain and swelling over knee and ankle joints had occurred over the past 1 year, for which he was prescribed steroid and non-steroidal anti-inflammatory medications causing symptomatic relief. There were no significant complaints pertaining to cardiac respiratory or gastrointestinal systems, any history of fever or recent weight loss. No other member of his family exhibited similar complaints.

On examination, the patient showed coarse facial features with increased facial skin thickness, seborrhoeic hyperplasia, deepening of nasolabial folds and furrowing of forehead skin into multiple transverse folds. Scalp skin showed furrows oriented parallel to the sagittal plane, with normal hair distribution and no fixity of skin to the underlying calvarium [Figure 3].

On investigation, his haemogram, blood counts, erythrocyte sedimentation rate, peripheral smear, platelet counts, liver and renal function tests and serum electrophoresis were within the normal limits. Serum uric acid value was 4.8 mg/dl. Anti-nuclear antibody and rheumatoid factor were negative, with elevated C reactive protein value of 41.6 mg/L. Human growth hormone assay showed 1.34 ng/ml using chemiluminescence immunoassay method. Age and sex matched insulin-like growth factor 1 level was also within normal limits.

X-rays of long bones showed minimal skeletal changes with periosteal reaction involving diaphysis of fibula, without any evidence of joint effusions [Figure 5]. Magnetic resonance imaging showed diploic space enlargement in calvarium indicating marrow hyperplasia [Figure 4]. Sella turcica and pituitary glands were visualised normally. Skin biopsy showed increased pilosebaceous structures and minimal increase in perivascular and periappendageal inflammatory infiltration. Bone marrow biopsy showed minimal erythroid hyperplasia.

Surgical management

A frontal rhytidectomy through a browlift incision, with dissection into forehead in the subgaleal and subfrontalis planes was done. A spindle shaped excision of scalp skin and frontalis muscle behind hairline was performed. Skin was sutured without excessive tension, frontalis muscle was repaired with tacking sutures to periosteum. This procedure caused considerable improvement in

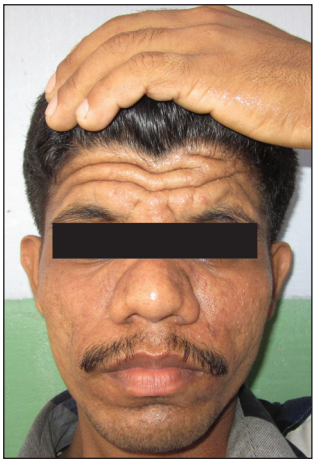

Figure 1: Pre-operative photograph showing forehead furrows, coarse facial features and acral enlargement of left hand

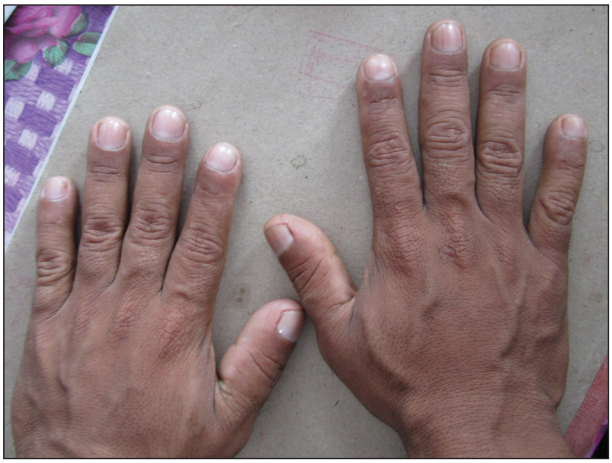

Figure 2: Hands showing bilateral acral enlargement and clubbing of digits

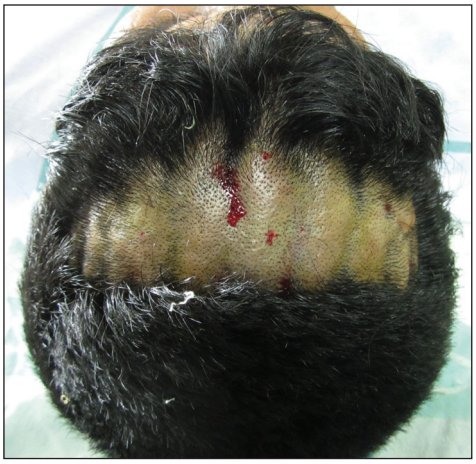

Figure 3: Shaved area of scalp skin with cutis verticis gyrata

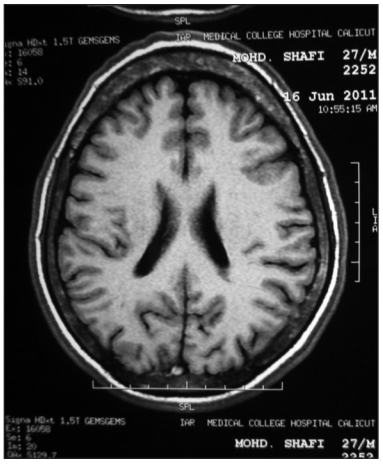

Figure 4: Magnetic resonance imaging scan showing diploeic widening in calvarium

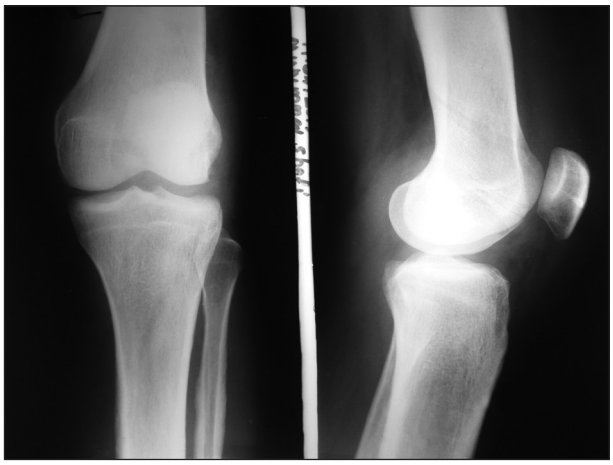

Figure 5: Plain radiograph of fibula showing periostosis

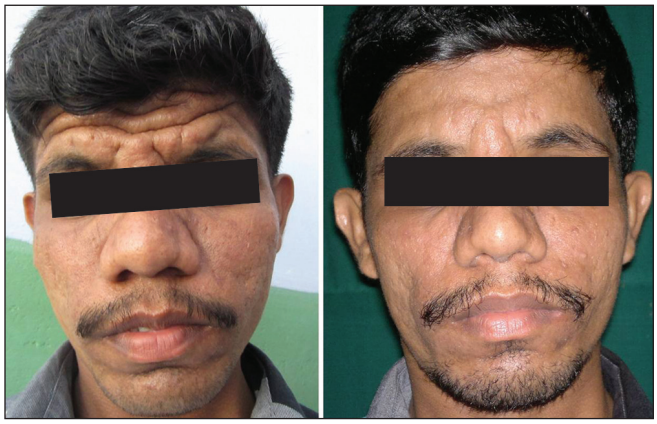

Figure 6: Pre- and post-operative photographs, 4 weeks after frontal rhytidectomy

his forehead furrows, though he is expected to require revision surgeries in future with progression of the disease [Figure 6].

DISCUSSION

Pachydermoperiostosis is a rare hereditary disorder characterised by primary HOA associated with thickening of scalp and facial skin, seborrhoea and hyperhidrosis. In later stages, the disease is epitomised by extensive digital clubbing, disfiguring pachydermia, mechanical ptosis and arthritis. The condition was first described by Nikoleus Frederich in 1868 as a familial case of HOA.[1] In 1890, Pierre Marie described it as '*osteoarthropathie hypertrophiante pneumique*'. Touraine, Solente and Gole recognised the entity as a primary form of HOA in 1935 and proposed a classification into the complete form-with pachydermia, clubbing and periostosis; the Fruste form-with prominent pachydermia and minimal skeletal changes and an Incomplete form-with no pachydermia.[2] Cutis verticis gyrate as described by Unna in 1906 is a frequent association of pachydermoperiostosis.

Touraine Solente Gole syndrome is a rare entity described in many races, but precise incidence is unknown. It occurs predominantly in males and inheritance is attributed to an autosomal dominant gene with variable expressivity. Radiologic survey of asymptomatic family members have yielded positive results in 25-38% cases.[3] Onset is usually in adolescence as enlargement of distal extremities with clubbing causing spade like hands and feet with cylindrical arms and legs due to circumferential diffuse periosteal ossifications. Bone scans show a higher uptake of Technitium 99 methylene-diphosphonate especially at the ends of tibia, fibula, radius and ulna. The skin and bone changes progress over next 5-20 years and then remain unchanged throughout life. Coarsening and furrowing of facial skin with deep nasolabial folds and mechanical ptosis result in a characteristic facial expression of weariness and despair. Increased seborrhoeic activity (in 90% cases) and troublesome hyperhidrosis (in 44-67%)[3] are frequent associations. Rare presentations with myelofibrosis and Empty sella have also been reported.[4]

A diagnosis of Primary HOA can be made only after exclusion of secondary pachydermoperiostosis with rapid bone changes and painful clubbing which may occur as a manifestation of severe pulmonary diseases such as adenocarcinoma of bronchus, pleural mesothelioma, bronchiectasis, gastric carcinoma or cyanotic heart diseases.[5] Other differential diagnoses include — acromegaly, thyroid acropachy, psoriasis, rhematoid arthritis and hematodermias. Patients with predominantly cutaneous manifestations may also have to be differentiated from the rare hyperelasticity disorders such as Ehler Danlos syndrome, cutis laxa, Meretoga's syndrome, Marfan's syndrome and pseudoxanthoma elasticum, which may cause forehead furrows.

Our patient essentially manifested a complete form of Touraine Solente Gole syndrome with predominant cutaneous changes in the form of deep furrows of forehead, prominent nasolabial folds, cutis verticis gyrata, seborrhoea and hyperhidrosis. He had early skeletal deformities of hands and feet in the form of acral enlargement of digits, clubbing and palmolplantar hyperkeratosis. Osteoarthropathic changes have been controlled with the early administration of steroids and non-steroidal anti-inflammatory drugs. Colchicine and

biphosphonates for articular symptoms and isotretinoin to improve skin changes have also been described with variable results.[5] Surgical management is limited to frontal rhytidectomy[6] for forehead furrows, blepharoplasty for ptosis correction and soft-tissue reduction in digital enlargements. Our patient had considerable aesthetic improvement with frontal rhytidectomy though he may require repetition of such procedures in future with further progression of the disease.

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