Touraine-Solente-Gole syndrome

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Pachydermoperiostosis (PDP) presents as idiopathic clubbing and periostosis. It is a genetically determined disease. It has been classified into 2 forms, primary idiopathic and secondary with varied cardiopulmonary and neoplastic conditions. The principal organs affected are skin and skeletal system. The skin presents as coarse facial features, sebaceous hyperplasia, thickened furrows on scalp (cutis verticis gyrata) and forehead. The skeletal features include thickening of the terminal ends of the phalanges, clubbing of the digits, periosteal new bone formation especially at the end of long bones, which are increased in diameter at ankle and wrist by periosteal proliferation. Males are predominantly affected.

A 48-year-old male presented to our clinic following a blunt trauma to the right leg. On examination he was found to have coarse features with folded skin on the face and dorsum of hands. Prominent furrows were visible on the forehead. Radiological examination of the injured right leg **Figure 1** showed no bony injury but prominent subperiosteal ossification over the lower end of tibia. The toes were thickened in diameter with skin on the dorsum of feet being acanthotic. The scalp did not show changes and features of cutis verticis gyrata. There was no family history of similar features or history of hyperhidrosis. A diagnosis of acropachyderma with pachyperiostitis (Pachydermoperiostitis) was made and the patient was investigated for any other systemic disorders, none other than skeletal or cutaneous abnormalities were noted.

Figure 1 - Radiograph showing prominent subperiosteal ossification around the lower end of tibia and fibula.

Pachydermoperiostosis was first described in 1891 and has 2 varied forms: primary, an autosomal dominant condition with variable expressibility with no underlying disease, and secondary with a variety of cardio-respiratory and gastrointestinal chronic or neoplastic diseases.[1] Pachydermoperiostosis mainly affects the skin and skeletal system. Digital clubbing and radiographic periostosis are the most common manifestations. Pachydermoperiostosis appears to have a bimodal onset, first occurring in the first year of life and then at adolescence. In our case the patient noted skin changes and clubbing in his late twenties. Males are predominantly affected with 9:1 male to female ratio. The absence of a family history, in our case may suggest autosomal mutation. The etiopathology of the disease is unknown. Deposition of acid mucopolysaccharides and some fibrillar material in the dermis along with increased levels of osteocalcin in serum have been observed.[1] Increased activity of serum growth factors such as platelet-derived growth factors are also suspected.[2] Endocrinal and chromosomal abnormalities have also been postulated. No endocrine abnormality was detected in our case.

The most common manifestations of primary PDP are cortical thickening and periosteal changes (97%), digital clubbing (89%), coarse features (60%), seborrhea (33%), and cutis vertices gyrat (24%).[3] Primary PDP may present in 3 ways: 'complete' form exhibiting all the cutaneous and skeletal manifestations, 'incomplete' form defined by absence of cutis verticis gyrata, as in our case or 'forme fruste' characterized by presence of one or more cutaneous findings with no, or minimal, osseous anomalies. The differential diagnosis of PDP includes acromegaly, thyroid acropachy, and syphilitic periostosis.

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