Pachydermoperiostosis

Alternative Names
PDP
Hypertrophic Osteoarthritis, Primary or Idiopathic

WHO International Classification of Diseases
Diseases of the musculoskeletal system and connective tissue

OMIM Number
167100

Mode of Inheritance
 Autosomal dominant; ? heterogeneity

Description
Pachydermoperiostosis is a rare developmental defect, first described by Friedreich in 1868 and recognized later in 1935 by Touraine, Solente and Golé as a familial disorder with three forms: complete (periostosis and pachydermia), incomplete (without pachydermia) and the forme fruste (pachydermia with minimal skeletal changes). It is inherited in an autosomal dominant fashion with variable expressivity and penetrance; autosomal recessive forms have also been reported. It is characterized by digital clubbing, cylindrical thickening of the legs and forearms, hyperhidrosis, sebaceous gland over activity and symmetrical irregular periosteal ossification predominantly affecting the distal ends of long bones. The syndrome is associated with thickening and furrowing of the facial features, deep nasolabial folds, a corrugated scalp, and often greasy skin of the face and the scalp.

Pachydermoperiostosis usually begins soon after puberty, progresses for five to ten years, and remains unchanged throughout life. The syndrome has a pronounced predilection for males, with a male to female ratio of 7:1.

Epidemiology in the Arab World

Tunisia
Until year 1977, two cases of pachydermoperiostosis have been reported in the Tunisian population (Haddad et al., 1977).

United Arab Emirates
Afify el al. (1986) reported the case of a 27 year old United Arab Emirates national male patient, consulting for pain over the body, easy fatigability, flatulent dyspepsia, and facial, hand and feet appearances which had been coarsening for the last 10 years. The patient was born to consanguineous parents and was the only member of the family affected. Clinical examination revealed thickening and folding of the skin of his face, eyelids were thick with folds and furrows, hands and feet were spade-like with marked clubbing of both fingers and toes, skin was rough on the palms and soles with hyperhidrosis, and arms and legs were cylindrical. Skeletal x-rays showed proliferative periostitis especially in the diaphyses of tibia, fibula, radius and ulna of the long bones. The metacarpal, metatarsal and proximal phalanges showed the same periosteal thickening, as well as some thickening of the bone cortex. Afify el al. (1986) suggested that this characteristic is a case of primary pachydermoperiostosis.

References

Contributors
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