

Pachydermoperiostosis - A Rare Syndrome With Rare Surgical Associations

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Citation

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Abstract

Pachydermoperiostosis is a rare syndrome with rare surgical associations and with diverse clinical and radiological features. Though diagnosis can be made on basis of clinical and radiological features, it is often missed due to variable presentations. This is a rare condition often mimicking some other well known conditions like acromegaly and the leonine facies seen in leprosy. This case is unusual because of the lack of family history and the late age of presentation. A case of pachydermoperiostosis associated with inguinal hernia and peptic ulcer disease, which is a rare association to a rare syndrome, is reported

CASE HISTORY

A 26 year old male, non smoker and non alcoholic presented with left inguinoscrotal swelling, right upper abdominal pain relieved on food intake, progressive enlargement of both hands and foot, accompanied by excessive sweating of face and extremities and also pain in multiple joints (knee, ankle and wrist). History of right herniorraphy done ten years back. No history of similar condition in family and consanguinity. On examination leonine facies, thickening and furrowing of forehead skin (figure-1), gross enlargement of forearm, hand, leg and foot (figure-2), clubbing (figure-6) of all fingers and toes was found. Mild tenderness presented in the right hypochondria. A left uncomplicated incomplete indirect inguinal hernia was also present.

X-ray of bones revealed symmetrical subperiosteal new bone formation with acroosteolysis of phalanges of both hand and foot (figure-3 and 4a,b), and an upper gastrointestinal endoscopy revealed gastritis and duodenitis. A skin biopsy revealed pachydermia. Growth hormone and IGF-1, thyroid profile, serum calcium, phosphorus, sodium and potassium, liver function test and renal parameters were normal. Rapid plasma reaction was negative. Computed tomography of the brain (figure-5), chest and abdomen were normal.

A complete anesthesia workup for a left hernioplasty was done. The post operative period was uneventful. The patient was started on proton pump inhibitors and cox-2inhibitors.

Figure 1



DISCUSSION

Pachydermoperiostosis (PDP) is a rare genetic disorder that affects both bones and skin⁽¹⁾. Other names are idiopathic hypertrophic osteoarthropathy or Touraine-Solente-Gole syndrome⁽²⁾. In 1868, PDP was first described by Friedreich as 'excessive growth of bone of the entire skeleton'. Touraine, Solente and Golé described PDP as the primary form of bone disease hypertrophic osteoarthropathy in 1935

and distinguished its three known forms⁽¹⁾.

The complete form occurs in 40 % of the cases and can involve all the symptoms but mainly pachydermia, periostosis and finger clubbing. This is also referred to as the full-blown phenotype^(1,7). The incomplete form occurs in 54 % of the cases and is characterized by having mainly effect on the bones and thereby the skeletal changes. Its effect on the skin (causing for instance pachydermia) is very limited^(1,7). The fruste form occurs in only 6 % of the cases and is the opposite of the incomplete form. Minor skeletal changes are found and mostly cutaneous symptoms are observed with limited periostosis^(1,7). The cause of these differentiating pathologies is still unknown^(1,7).

It is mainly characterized by pachydermia (thickening of the skin), periostosis (excessive bone formation) and finger clubbing (swelling of tissue with loss of normal angle between nail and nail bed)^(1,3). PDP is a rare genetic disease⁽¹⁾. At least 204 cases of PDP have been reported^(1,7). The precise incidence and prevalence of PDP are still unknown⁽¹⁾. A prevalence of 0.16% was suggested by Jajic et Jajic⁽⁸⁾. This disease affects relatively more men than women^(1,4). After onset, the disease stabilizes after about 5-20 years. Life of PDP patients can be severely impaired^(1,5). Currently, symptomatic treatments are NSAIDs and steroids or surgical procedures^{[5][6]}.

The association to the surgical conditions is very rare and reports of PDP associated with gastric involvement have been described in families⁽⁹⁾ and in sporadic cases. the Gastro intestinal symptoms appear usually in the twenties and are severe, with gastric and duodenal ulcer⁽⁹⁾. The levels of serum pepsinogen I and II can be found elevated. In our case the patient had severe gastritis and duodenitis which is to be a rare association.

Considering the fact, in 1994 Combemale et al. did an ultra structural study of pachydermoperiostosis, electron microscopy revealed irregular calibre of collagen fibers⁽¹⁰⁾ and in 2006, Szczesny et al., studied the etiology of inguinal

hernia. He stated that there is difference in the caliber of individual fibers and disrupted ground matter-to-fiber ratio⁽¹¹⁾. This supports the theory linking connective tissue alterations with the etiology of the hernia. Correlating these above theory the formation of hernia in our patient who had bilateral inguinal hernia can be considered and PDP, one of the rarest syndromes causing connective tissue alteration can be considered as a rarest etiology for inguinal hernia. To our knowledge this might be the first case to be presented with such a rare association to a rare syndrome. We mainly stress upon a criteria is that even a common conditions like inguinal hernia can be associated with a rare syndrome like this, hence a close clinical observation and a clear history and examination to be elicited and carried out in all cases.

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