A Case Of Pachydermoperiostosis And Overriding Forth Toes

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Citation

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Abstract

Primary hypertrophic osteoarthropathy is a disorder with clubbing of digits, of thickened skin and periosteal new bone formation prominently in distal ends of the limbs. We present a case with characteristic components of the disease, bilateral congenital overriding of the forth toes, a finding described for first time in association with the disease, and a negative pathergy test, detect neutrophil hypereactivity.

INTRODUCTION

Hypertrophic osteoarthropathy (HOA) can be primary or secondary. Primary hypertrophic osteoarthropathy which is also referred to as pachydermoperiostosis (PDP) is an autosomal dominant disorder with clubbing of digits, hyperhidrosis of thickened skin and periosteal new bone formation prominently in distal ends of the limbs [$_1$]. Autosomal recessive inheritance also seems to occur [$_1$]. Two forms of the primary disease are distinguished, familial and idiopathic. To establish a diagnosis of PHO, finger clubbing and periostosis of long bones must be present. In addition absence of any signs and symptoms of reasons causing secondary HOA is a must to construct the diagnosis of the primary form [$_2$]. Three forms of the primary one are defined: complete with a clinical picture of pachydermia and changes in the joints and bones, incomplete with the evidence of bone changes and the so-called forme fruste with the prominent manifestations of pachydermia and absent or mild bone changes [$_3$]. We present our case with clinic, radiological and laboratory findings of PDP with the overriding of bilateral forth toe

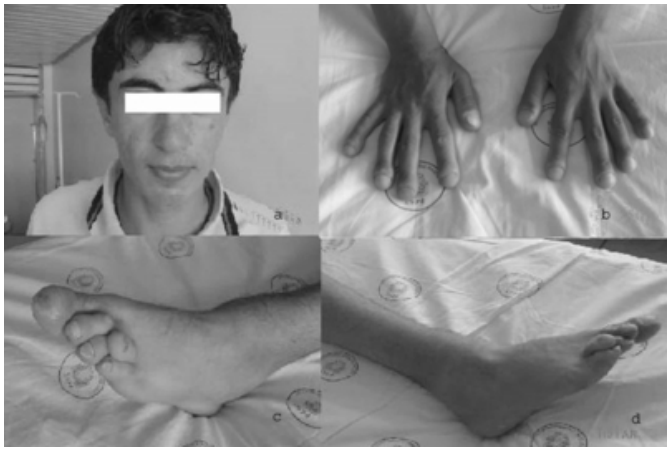
CASE REPORT

A 17 year old boy admitted to Internal Medicine Department of Dicle University Hospital, with recurrent swelling and pain of both knees and ankles. The patient was also complaining enlargement of extremities which was learned from the family to be proportional to the growth of the body. He has also observed enlargement of wrists and ankles and overriding of bilateral forth toe from childhood. He had palmar hyperhidrosis but no trauma and operation history. His past medical history revealed nothing else. But his family history revealed a sister with same symptoms, but no diagnosis. His sister refused to admit hospital for diagnostic tests.

Clubbing of the digits (figure 1b), enlargement of distal ends of the wrists and ankles (figure 1c,d), were detected on physical examination. Overriding of bilateral forth toe (figure 1c,d), congenital, was observed. No swollen joint was detected. Thyroid, liver, and spleen were normal on physical examination. No masses and enlarged lymph nodes were observed. Cardiac and lung examinations were normal. Palmar hyperhidrosis, pachydermia, oiliness facial skin, seborrhea and acne-like changes on face (figure 1a), and palmoplantar hyperkeratosis were found on dermal examination.

Figure 1

Figure 1: Characteristic facial findings (a), clubbing of fingers (b), enlarged distal ends of lower extremities and overriding of the both forth toes (c,d).

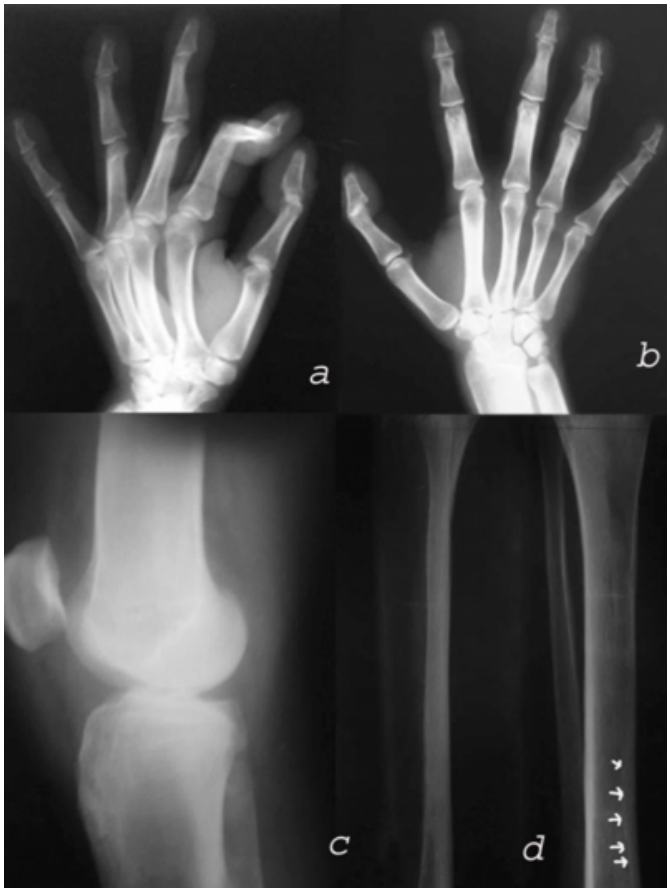


On laboratory evaluation tests of liver, kidney, thyroid, glucose, hemoglobin, platelet, leukocyte count, erythrocyte sedimentation rate (ESR), C-reactive protein, were normal. Rheumatoid factors and antinuclear factors were negative. On ECG normal sinus rhythm observed. Blood smear revealed normal differential count and no other abnormalities. Upper gastrointestinal endoscopy, performed to detect duodenal ulcers, hypertrophic gastritis [4], showed no pathology.

Radiological examination showed that periosteal new bone formation in long bones (figure 2d), metacarpals, and metatarsals (figure 2a,b). Also, acro-osteolysis in hands (figure 2a, b) and soft tissue swelling in hands, feet, and knees (figure 2c), were detected but sacroiliac films, PA lung scans and 2-dimension echocardiographias were normal.

Figure 2

Figure 2: Periosteal new bone formation in the metacarpal bones and phalanx and acroosteolysis in distal phalanx of both hands (a,b), soft tissue swelling at knee (c), new bone formation of tibia (d arrows)



A differential diagnosis was constructed to exclude the causes of secondary hypertrophic osteoarthropathy (lung, heart or liver disease, thyroid and rheumatologic disorders). After taking into consideration the familial occurrence of clubbing of the digits, characteristic physical signs, laboratory and radiological findings, a diagnosis of the complete form of primary hypertrophic osteoarthropathy was established. NSAID treatment was given.

DISCUSSION

In addition to the digital clubbing, the clinical manifestations of PDP include hypertrophic skin changes, such as pachydermia (thickening of the facial skin), seborrhea, folliculitis and, less commonly, cutis verticis gyrata, as well as the osteoarticular manifestations related to periostosis [2].

In our patient the diagnosis of complete form of familial PDP was based on familial occurrence and long history of clubbing, enlargement of distal end of long bones, hyperostosis on x-rays and characteristic skin changes.

Secondary HOA reasons were ruled out before the diagnosis was made.

Heredity reported in 72 % of the cases in a study [4] as in our case. Both acro- osteolysis in hands [4] and cutis verticis gyrata [2] are rare components of the disease. The former one was demonstrated very well in our case but the latter one was not present. Also, congenital overriding of the forth toe, in association with pachydermoperiostosis, was found in our case.

PDP may be associated with sacroiliitis, psoriasis, rheumatoid arthritis, duodenal ulcers, hypertrophic gastritis [4], gynecomastia [3], anemia, myelofibrosis [5], juvenile polyps, gastric cancer [6], spondylolisthesis [7], and cutaneous squamous carcinomas [8]. In all these cases the associated diagnosis are after the PDP diagnosis so they are not in group of HOA secondary to the other diseases. In our case all possible associations are searched no one was found. It was suggested that combined tumors may not be incidental. Chromosome instability [8] and chromosomal mosaicism [6] have been accused for tumor association. Chromosomal analysis was normal and no tumor was found in our case.

NSAIDs have been routinely used for skeletal symptoms. Also, colchicine provided a beneficial therapeutic response in both inhibiting increased chemotactic activity and in reducing tissue edema in a study [9]. In our case pathergy test performed on the flexor aspect of the forearm by the technique [10] to detect neutrophil hypereactivity. Pathergy test, which may be used as an easy guide to use colchicine, result was negative

CONCLUSION

In conclusion we say that PDP must be present in the list of differential diagnosis for patients with long term clubbing of digits especially the familial ones after ruling out secondary reasons of HOA and overriding toes may be associated with PDP.

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