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Case Report

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Monoarthralgia, Facial morphology with Clubbed fingers and toes sans systemic involvement: Primary hypertrophic osteoarthropathy

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Abstract

Abstract: Formation of new bone subperiosteally with clubbing and synovial effusions are symptoms of hypertrophic osteoarthropathy. It could be secondary or primary. We describe a 26-year-old man who presented with right knee discomfort and digital clubbing that began in adulthood. X-ray findings revealed subperiosteal new bone growth and periosteal thickening. Secondary causes were not found in any of the investigations.

Keywords: Digital clubbing, Hypertrophic osteoarthropathy, periosteal thickening, subperiosteal new bone formation.

INTRODUCTION

Periostitis with new bone formation subperiosteally with fingers and toes clubbing and arthralgia form the principle depictions of Hypertrophic osteoarthropathy (HOA). Two main types seen are-primary (pachydermoperiostosis) and secondary [1]. Usually the autosomal dominant inheritance forms the mainstay of primary and secondary type encompasses Fallot's tetralogy , lung cancer, IBS and malignancies of ovary [2].

Genes like HPGD and SLCO2A1 are frequently related with primary type resulting in prostaglandin accumulation namely PGE2 [3].

SLCO2A1 mutation has overall male preponderance presenting with anaemia like features but enteropathy like characteristics are more seen in females and whereas HPGD mutation have no definite gender predominance. Patients with SLCO2A1 mutation thus need a careful evaluation before treatment with selective COX-2 inhibitors taking into the account the gastro-intestinal complications [4].

CASE REPORT

A 26-year-old male presented to the OPD with a longstanding fingers and toes enlargement. He also complained of arthralgia in right knee with no morning stiffness or any other joint involvement. Above-mentioned complaints gradually progressed over 8 years being noticed first at adolescence.No gastrointestinal complaints have been noted. No significant family history was obtained.

On physical examination, forehead showed deep furrows and prominent nasolabial folds with digital clubbing in bilateral hands and feet.Right knee demonstrated tenderness with no significant swelling and terminally restricted movements.

Laboratory tests except for Alkaline phosphatase -208 IU/L (30-120 IU/L) which was slightly above normal range ,including rheumatoid factor ,anti-cyclic citrullinated peptide and IGF-1 (to rule out acromegaly), were insignificant . Bilateral hand with forearm radiographs showed periosteal thickening (Fig. 3).

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X ray right knee was insignificant with ultrasound showing minimal effusion (~ 4 ml) with no signs of arthritis.Ultrasound aspiration of fluid was done and sent for analysis which came back unremarkable.

A chest radiograph and arterial blood gas analysis was normal ruling out any secondary cause. 2D-Echocardiography was found within normal limits. Genetic testing revealed variant in homozygosis in SLCO2A1 gene and thus diagnosis of Primary hypertrophic osteoarthropathy (PHO) was made.Patient was started on Etoricoxib 90 mg/day supplemented by ice pack application and relief was obtained after 4 weeks of treatment.

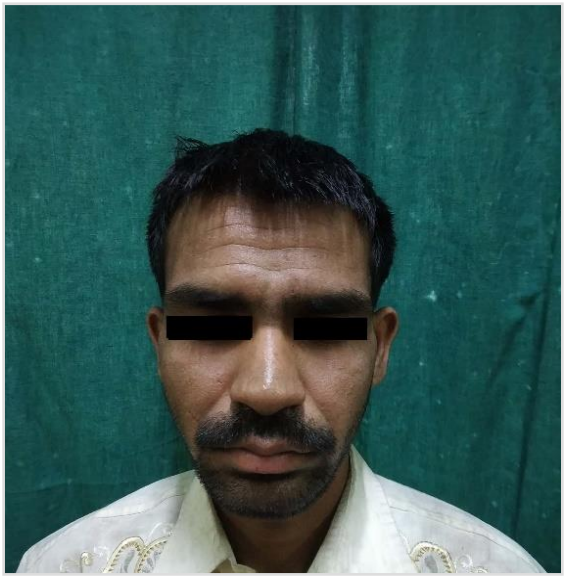

Figure 1: Deep furrows over forehead, prominent nasolabial folds

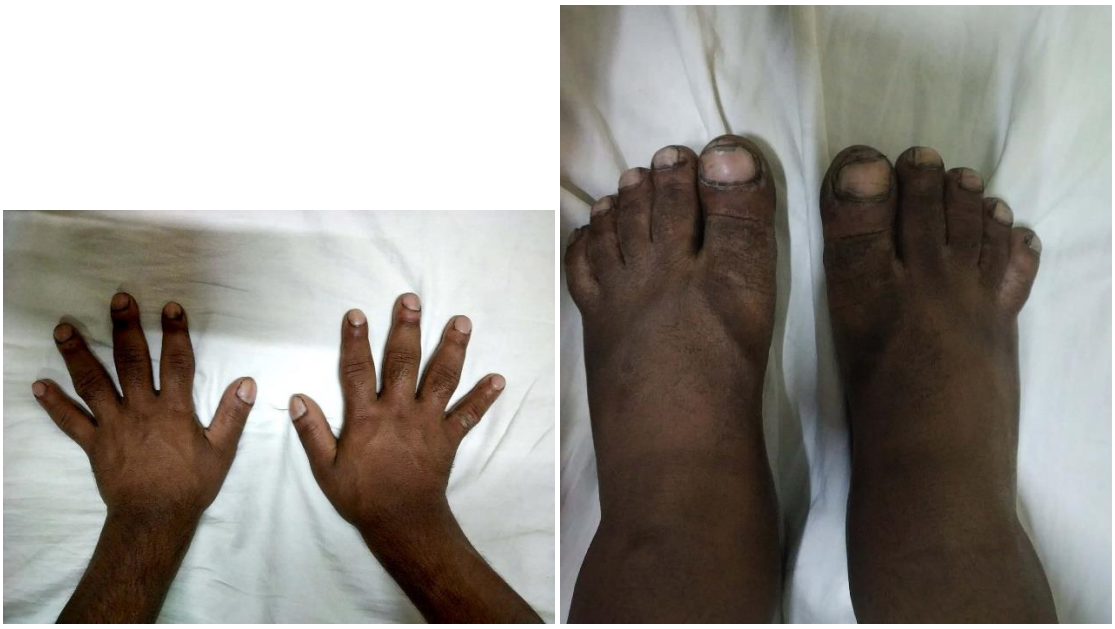

Figure 2: Digital clubbing

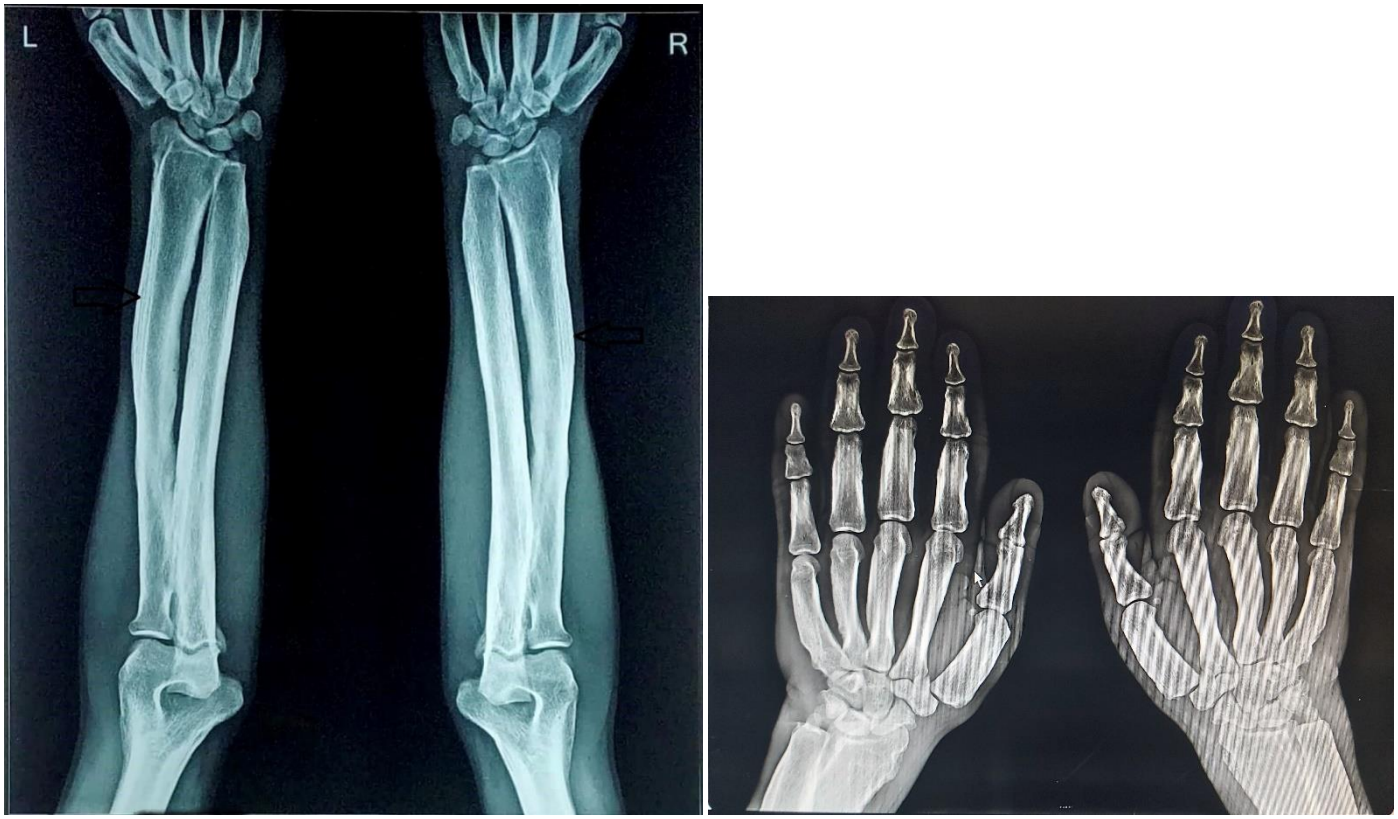

Figure 3: Subperiosteal new bone formation in both Radial shaft (arrow) and periostosis in both metacarpals

DISCUSSION

Periostitis with new bone formation subperiosteally with fingers and toes clubbing and arthralgia form the principle depictions of Hypertrophic osteoarthropathy (HOA). Two main types seen are- primary (pachydermoperiostosis) and secondary [1]. Usually the autosomal dominant inheritance forms the mainstay of primary and secondary type encompasses Fallot's tetralogy , lung cancer ,IBS and malignancies of ovary [2].

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Facial morphology and digital clubbing often points towards hypertrophic osteoarthropathy with usually no symptoms at all or with mild symptoms.If symptomatic ,they show a sluggish progression over months or years.PHO is a great mimicker of rheumatological conditions and other common causes are to be ruled out before ascertaining the PHO as the cause [5].

No definitive treatment is available at present and NSAIDs and ice pack applications are used to manage symptoms like arthralgia and bone pain.[6] COX-2 inhibitors can be safely used if there is no enteropathy like features and predisposition to gastritis [7].

CONCLUSION

Hypertrophic osteoarthropathy though usually seen as secondary to cardiac and pulmonary ailments should be evaluated thoroughly to delineate primary type and consequent genetic testing and watchful monitoring of symptoms warranting treatment should be undergone.

Conflict of Interest

None declared.

Financial Support

None declared

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