Abstract
A case of Pachydermoperiostosis (PDP) presented to us in rheumatology clinic with complaints of enlargement and broadening of bilateral hands and feet, grade IV digital clubbing, coarsening of facial features, excessive sweating of the palms, soles during summers.

Introduction
Pachydermoperiostosis or primary hypertrophic osteoarthropathy (HOA) is a rare hereditary disorder that is characterised by digital clubbing, pachydermia, and periostosis.

Case Report
A 30 year old male presented with complaints of enlargement of distal phalanges and distal toes along with excessive sweating from palms and soles for approximately 13 years. There was no history of fever, cough, dyspnea, palpitation, bluish discolouration of tongue, arthralgia, backache, hematochezia, oral ulcers, weight loss or bleeding tendency. The patient also reported the same problems with his father.

On general physical examination there was furrowing of cheeks (Figure 1), widening of bilateral wrist and ankle joints (Figure 2) which were non-tender. There was associated enlargement of bilateral distal phalanges and distal toes with grade IV digital clubbing seen (Figure 2).

Systemic examination was clinically unremarkable.

On investigation his haemoglobin, ESR, leucocyte count and platelet count were within normal limits. Peripheral blood film showed normocytic normochromic picture. Liver, kidney and thyroid tests were normal. Blood sugar was within normal range. Urine examination was also within normal range. Insulin-like growth factor 1 (IGF1) was done to rule out other causes of digital clubbing and both were normal.

Patient’s father aged 70 years and all the three sons aged 11 years, 7 years and 3 years were also examined.

Patient father’s general examination also showed coarse facial feature (Figure 1), widening of wrists and ankles associated with enlargement of bilateral hands and feet which were non-tender and digital clubbing grade IV. Systemic examination was unremarkable.

All the three sons of the patient showed no coarsening of facial features, no widening of ankles and wrists or enlargement of hands and feet.

Chest X-ray and 2D-Echo was done to rule out other causes of digital clubbing and both were normal.

The final diagnosis was pachydermoperiostosis.

Discussion
Pachydermoperiostosis or primary HOA is a rare hereditary disorder that was first described in 1868. Only 204 cases have been reported. It is characterized by digital clubbing, pachydermia (thickening of the facial skin and/or scalp), and periostosis (swelling of periarticular tissue and subperiosteal new bone formation). Pachydermoperiostosis or primary hypertrophic osteoarthropathy is associated with pain, polyarthritis, cutis verticis gyrata, seborrhea, eyelid ptosis, and hyperhidrosis.

Touraine et al described three forms of pachydermoperiostosis or primary HOA:
1. A complete form with pachydermia and periostitis,
2. An incomplete form with evidence of bone abnormalities but lacking pachydermia, and
3. A forme fruste with prominent pachydermia and minimal-to-

Fig. 1: Coarse facial features in patient and his father

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absent skeletal changes.

The patient under discussion had complete form of pachydermia and periostosis with no associated conditions mentioned above.

Following findings may be present in a patient of pachydermoperiostosis:

1. Skin: Pachydermia, coarse skin, oily skin, eczema, thick hand and feet, furrowing, leonine facies, cutis verticis gyrate, increased secretion of sebum, sebornheic hyperplasia, keloid formation
2. Bone: Periostosis, acroosteolysis, myelofibrosis, thick toe and finger bone, widening of bone formation
3. Digital clubbing
4. Hyperhidrosis
5. Eye: Drooping of eyelids, thick stratum corneum
6. Joints: Arthralgia, joint effusion
7. Muscle discomfort
8. Decreased facial and pubic hair
9. Peripheral vascular stasis
10. GI involvement: peptic ulcer, chronic gastritis, Crohn’s disease.

Causes

The pathogenesis of PDP is still not fully understood. However several theories have been suggested:

1. The neurogenic theory proposes that stimulation of the vagus nerve leads to vasodilation, increased blood flow and PDP. 3
2. The humoral theory proposes that mediators such as growth factors or inflammatory mediators are increased, leading to fibroblast proliferation and PDP. 1,3
3. HPGD gene (4q33-q34) mutations have been identified. The gene encodes 15-hydroxyprostaglandin dehydrogenase (15-PGDH) which is the main enzyme of prostaglandin (PG) degradation. 4 Patients with homozygous mutations have chronically elevated PG E2 levels. Deficiency of the prostaglandin transporter (SLC02A1) has been characterized as the main cause of primary hypertrophic osteoarthropathy. 5

It is usually inherited as autosomal dominant model with incomplete penetrance and variable expression, both autosomal recessive and X-linked inheritance have been suggested in some PDP families. One-third of these patients have a positive family history. In our patient’s case his grandfather (deceased), father and all his uncles reported to have the same complaints.

The age of onset is often at puberty. 1
In the present case also clubbing started at the age of 15.

PDP may be associated with sacroiliitis, psoriasis, rheumatoid arthritis, duodenal ulcers, hypertrophic gastritis, gynecomastia, anemia, myelofibrosis, juvenile polyps, gastric cancer, spondylolisthesis, and cutaneous squamous carcinomas but our patient was found to be associated with none.

Nonsteroidal anti-inflammatory drugs (NSAIDs) or corticosteroids are used for skeletal symptoms. Also, Colchicine provides a beneficial response in both inhibiting increased chemotactic activity and in reducing tissue edema. Vagotomy may improve the articular pain and swelling associated with PDP. Plastic surgery may improve the appearance of the face and scalp by excising redundant skin and correcting the cutis verticis gyrate.

References