PACHYONYCHIA CONGENITA: CASE REPORT

Introduction:
Pachyonychia congenita (PC) is rare autosomal dominant genodermatoses caused by heterozygous mutations in any one of genes encoding differentiation-specific keratins KRT6A, KRT6B, KRT6C, KRT16, and KRT17. Clinical features include painful and debilitating plantar keratoderma, hypertrophic nail dystrophy, oral leukokeratosis, and variety of epidermal cysts, subdivided into two subtypes on basis of clinical presentation, PC-1 (Jadassohn–Lewandowski type) or PC-2 (Jackson–Lawler type).

Case Report:
1st Case:
A 38 Y/F with two daughters aged 21 and 17Y presented with complaints of thickened finger and toe nails, plantar pain, thickened yellow coloured skin of palms and soles since birth. On examination there was 20 nail dystrophy with palmo plantar keratoderma, palmoplantar hyperhidrosis, sebaceous cyst and hyperkeratosis on elbows and knees.

2nd Case:
A 29 Y/F with her 5 Y/F child presented with thickened finger and toe nails, plantar pain, thickened yellow coloured skin of palms and soles since birth. On examination there was 20 nail dystrophy with palmo plantar keratoderma, palmoplantar hyperhidrosis and hyperkeratosis on elbows and knees.

Discussion:
There is no specific therapy so main aim of treatment is to alleviate pain caused by plantar keratoderma. Patient was prescribed salicylic acid 12% ointment on nails and combination of salicylic acid and clobetasol ointment on palms and soles twice daily for 3 weeks initially. First case was given oral isotretinoin 20mg daily for 4 weeks. Mechanical removal of callosities is more effective. The "not too thick, not too thin" motto is essential in caring for keratoderma. Bleach baths can reduce onset of infections.

Conclusion:
This case is being presented because of its rarity, only 5000 to 10,000 cases reported worldwide till now. Even though there is lack of definitive treatment early diagnosis can improve the quality of life of these patients.