Case report

Pachyonychia Congenita in both father and son

K L P D Seneviwickrama[1], A M S D Kumari[2], H M P Gunathilake[2]

Sri Lanka Journal of Dermatology, 2021, **22**: 46-48

Abstract

Pachyonychia Congenita (PC) is a rare geno-dermatosis due to mutations in one of five keratin genes. It primarily affects skin and nail. Here we report a case of PC in a 7 year old boy and his father.

Introduction

PC is an autosomal dominant inherited disorder of keratinization accounting into 10,000 cases world-wide. There is no gender predilection. Mutant keratin genes are responsible to form keratin in nails, palmoplantar skin, mucosae, and hair giving rise to clinical features of PC in corresponding sites. There are two main types of PC, PC-1 and PC-2. Affected individuals have normal life expectancy.

Case report

A 7 year old boy presented with a history of multiple asymptomatic skin colored papules all over the body for 9 months duration which slowly progress in size and number. He was the only child and the parents were non consanguineous. On questioning, father declared to have similar lumps on him since childhood. Examination of the child revealed pin point skin colored papules and cysts (Figure 1A) which were attached to the skin and scattered all over the body including genitalia (Figure2).

Some of his toenails were thickened with normal fingernails (Figure 4A). His hair, teeth and mucous membranes were normal. Systemic examination didn't reveal any abnormality.

Examination of his 47-year-old father revealed bigger nodules and cysts (Figure 1B) which were wide spread. His twenty nails were dystrophied and thickening noted with uplifting, and tenting of the distal edge and dystrophied (Figure 3) with discoloration of the nails which were lusterless, thickening of the nails with uplifting, and tenting of the distal edge of the nails were noted. The nails also showed subungual hyperkeratosis and adhesion of the nail plate to the underlying nail bed. There was hyper curvature of the transverse axis of all nail plates giving a "pinched shape" or "door wedge shape" to the free edge of the nail plate (Figure 4B). Needle puncturing of a papule showed thick, milky material giving clue to the steatocystoma. His hair, teeth and mucous membranes were normal. Considering the clinical features, clinical diagnosis of PC-2 in both father and child was made.

Discussion

PC is an autosomal dominant disorder of keratinization affecting primarily the skin and nail. 30-40% of cases are the result of new spontaneous mutation with no previous family history. The underlying genetic cause is a mutation in one of 5 keratin genes KRT6A, KRT6B, KRT6C, KRT16 or KRT17.

Figure 1.

[1]*Consultant Dermatologist*, [2]*Medical Officer in Dermatology, Sirimavo Bandaranaike Specialized Children's Hospital, Peradeniya, Sri Lanka.*

Figure 2.

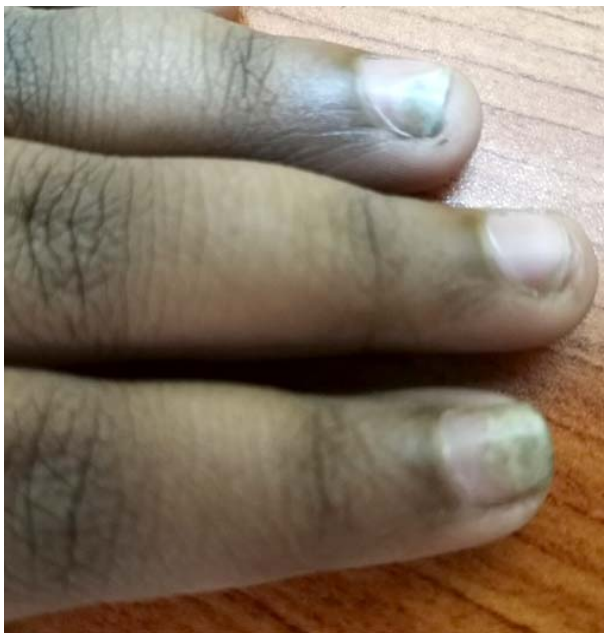

Figure 3.

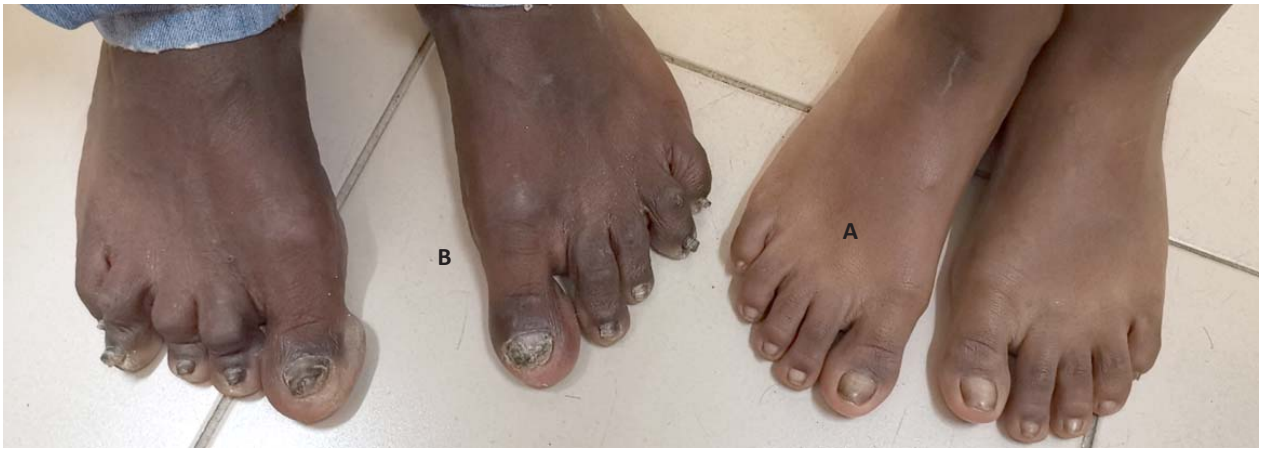

Figure 4.

PC was first described by the Muller in 1904 followed by Jadassohn and Lewandowsky in 1906. There are 2 main sub-types named PC-1, the Jadassohn-Lewandowsky type and PC-2 Jackson Lawler type.

Hyperkeratosis of the nail bed is the first sign to appear in majority of cases within first two years of life. Onset of focal palmoplantar keratoderma is seen when child starts walking. Focal keratoderma may give rise to severe pain when walking. The disease severity varies even within families.

In PC-1 about 90% of patients have palmoplantar keratoderma. Nail changes, wedge – like thickening of distal nail plate in fingers and toes are the secondly most common finding. Oral leukokeratosis resembling candidiasis[1] can be seen on tongue and oral mucosa. Keratosis pilaris and follicular keratosis on knees and

elbows are seen in some patients. Laryngeal involvement may produce hoarseness[2]. Squamous cell carcinoma may arise from areas which undergo repeated ulceration.

PC-2 shares clinical features with PC-1, but with less severe palmoplantar involvement. Oral mucosal lesions are absent in PC-2. Natal teeth are seen in majority. Association of cutaneous cysts is a feature in PC-2 which helps to distinguished PC-2 from PC-1[3]. They include epidermoid cysts and steatocysts. Extensive cysts and recurrent infections of these cysts in flexural locations cause significant disability to the patients. In some families with PC, like in index case present only with nail changes and cysts.

Treatment include liberal use of emollients together with keratolytics, oral acitretin may help in some cases temporarily. Surgical excision may attempt, but recurrences are anticipated. Over the past few years, a number of reports have shown the planta injections of botulinum toxin reduce or even eliminate pain, blistering and callosities in patients[4].

References

1. Mawhinney H, Creswell S, Beare JM. Pachyonychia congenita with candidiasis. C*lin Exp Dermatol* 1981; **6**: 145-9.
2. Stieglitz JB, Centerwall JW. Pachyonychia congenita (Jadassohn-Lewandowsky syndrome): a seventeen member, four-generation pedigree with unusual respiratory and dental involvement. *Am J Med Genet* 1983; **14**: 21-8.
3. Alan D, Peter H, Albert C. Harper's Text book of Pediatric Dermatology. 3rd edition. Oxford Blackwell Scientific Publications. Chapter 120; 21.
4. Koren A, Sprecher E, Reider E, Artzi O. *Br J Dermatol*. 182; **3**: 671-7.