Cutaneous Cysts with Nail Dystrophy in a Young Female: A Classical Association

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Abstract

Pachyonychia Congenita (PC) refers to a group of autosomal dominant disorders with variable clinical presentations. While nail dystrophy and plantar keratoderma are the most consistent features in all the variants, a myriad of other manifestations has been observed. This report highlights a case of young female presenting with multiple asymptomatic cutaneous cysts associated with plantar keratoderma and nail dystrophy. Similar nail changes were evident in her son also. Such clinical presentation, in corroboration with histopathological evaluation of the cutaneous cyst prompted us to make a diagnosis of Pachyonychia Congenita type II.

Keywords: Autosomal dominant, pachyonychia congenita, plantar keratoderma, steatocystoma multiplex

What was known?

Pachyonychia Congenita is a rare, autosomal dominant genodermatosis characterized by dystrophy of the nails associated with nail bed and hyponychial hyperkeratosis, of which type II has been associated with mutations in K17.

Introduction

The term steatocystoma multiplex was coined by Pringle in 1899 after being described by Jamieson in 1873.[1] It is a nevoid malformation of the pilosebaceous duct, characterized by numerous, sebum-filled dermal cysts lined by stratified squamous epithelium with characteristic sebaceous glands in the cyst walls. It can be sporadic or inherited as an autosomal dominant trait. These lesions present as asymptomatic, yellow or skin-colored dermal papules or cysts located most commonly on the trunk, upper arms, scrotum, or chest. The disease can be classified as localized, generalized, facial, acral, and the suppurative types. The sporadic solitary lesions are known as steatocystoma simplex.[2] These lesions can become infected and suppurate, resulting in sinus formation and scarring. These lesions can also be found in syndromes such as Alagille syndrome and pachyonychia congenital type II (PC-II).
PC is a rare, autosomal dominant genodermatosis characterized by dystrophy of the nails associated with nail bed and hyponychial hyperkeratosis. Müller was first to highlight this entity in 1904 followed by reports published in 1905 by Wilson and in 1906 by Jadassohn and Pachyonychiacongenita.[3] Two main types of PC are recognized: (1) PC-I (Jadassohn Lewandowsky type) (2) PC-II (Jackson-Lawler type) (3) PC Tarda/type-III (Shafer-Brunauer type), characterized by a later-onset, ranging from late childhood to middle age associated with angular cheilitis, corneal dyskeratosis, and cataracts.[4] Another variety called Type IV also had been described which includes features of other three types in addition to laryngeal involvement and mental retardation.[5] In type II (Jackson–Lawler type), with less severe nail thickening and keratoderma than type I, a myriad of associations has been described: natal teeth, multiple epidermal cysts, sebocystomatosis, dry, lustreless and kinky scalp hair, eyebrows that stand straight out; and hamartomas. There is the absence of oral lesions.

**Case Report**

A 22-year-old married female patient presented with multiple asymptomatic yellowish to skin coloured cutaneous cysts over face, neck, and sternal region for the past 7–8 years. The cysts first appeared over the anterior aspect of the neck then gradually spread over the other areas [Figure 1]. She also had painful fissuring with crusting of plantar skin, especially over pressure areas [Figure 2]. She came with her 1.3-year-old male child with similar findings over the plantar skin [Figure 3]. The child had no skin colored cutaneous cyst hypertrophic, thickened, brittle nails were present in both mother and the child [Figures 4–6] with typical blackish brown discoloration and excess keratin deposition over nail bed [Figures 2 and 3]. She had brittle nail since childhood affecting only toe nails. However in case of her son, both finger and toe nails were affected since birth. She had a history of consanguineous marriage.

On examination, multiple, smooth, well-defined, round to oval cutaneous cysts, without a punctum were detected, between the diameter of 2 and 5 mm. Presence of oily material was detected on incising one of the cysts. However, both mother and child had no oral mucosal lesion. Systemic examination and family history were unremarkable. KOH microscopy and culture of nail clippings were done to rule out onychomycosis. Fine-needle aspiration cytology from the cutaneous cyst revealed few squamous cells in an oily background. The differential diagnoses of steatocystoma and multiple epidermoid cysts were considered and a biopsy was performed. On histopathological examination, the tissue specimen was lined by keratinized, stratified, squamous epithelium. The dermis revealed a cyst which was lined by flattened, stratified, squamous epithelium without a granular layer and with a cellular eosinophilic cuticle over its surface. The sebaceous gland lobules were lying close to the cyst wall[5] [Figures 7 and 8]. All other investigations such as complete hemogram, urine analysis, and fasting blood sugar were within the normal limits. Based on the history, clinical features and laboratory reports a diagnosis steatocystoma multiplex in PC-II (Jackson-Lawler type) in mother and son were made. The patient was prescribed oral Isotretinoin 20 mg/day and few lesions of steatocystoma multiplex over cosmetically sensitive sites were treated with radiofrequency ablation. The further surgical intervention was declined by the patient. White soft paraffin cream was prescribed for planter keratoderma.

**Discussion**

PC is a rare genodermatosis. Since 1904, only 300 cases have been reported. The form of PC with widespread pilosebaceous cysts was first described by Jackson and Lawler in 1951. Feinstein et al. (1988) first proposed a classification of PC into four overlapping subtypes. In 1995, Dahl et al. proposed a simplified diagnostic module in the form of characteristic nail changes (major criteria), in conjunction with at least one minor criteria autosomal dominant inheritance, palmoplantar keratoderma, leukokeratosis oris, follicular keratosis, bullae on palms or soles, and orlaryngeal leukokeratosis. [4] PC-II has been associated with mutations in K17. Keratin 17 (K17) is a differentiation specific keratin expressed in the nail bed, hair follicle, sebaceous gland, and other epidermal appendages. Unlike PC-I, where pathogenic mutations have been detected either in K16 or its expression partner K6A, no expression partner has been reported till date.
for K17, and its mutation has been described in PC-II or familial steatocystoma multiplex. Therefore, it can be perceived that K17 can be considered as a genetic marker in PC-II and familial steatocystoma multiplex. Available literature depicted that mutation reports of two separate studies conducted in 1997 and 1998, respectively, dealt with total seven cases of PC and two cases of familial steatocystoma multiplex only. These studies revealed that while K17 mutation was the only common factor in all of the cases, clinical variations have been significant. Thickening of nails have been a consistent feature in both PC-II and familial steatocystoma multiplex in all the cases barring one. Plantar keratoderma, natal teeth, acneiform eruptions, pili torti, milia, flexural abscess, blistering of feet, and hyperkeratosis of buccal mucosa were other clinical features detected.\[5,6\] To the best of our knowledge, the present case appears to be the first from Indian subcontinent where two successive members of the family presented with features of PC-II [Figure 9 pedigree], in addition to steatocystoma multiplex in the mother. Due to logistic limitations, mutation analysis was out of bounds, but the clinical features were fairly suggestive toward the diagnosis of PC-II with the possible K17 mutation. This report is purported to highlight a classical but rare association of steatocystoma multiplex with nail hypertrophy and plantar keratoderma. Such an association should warrant a mutation analysis and detailed history and examination for other family members.

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**Conflicts of interest** There are no conflicts of interest.

**What is new?**

Association of steatocystoma multiplex with nail hypertrophy and plantar keratoderma warrants a mutation analysis and detailed history and examination of other family members.

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**Figures and Tables**
Figure 1

Lesions of steatocystoma multiplex in mother
Figure 2

Planter keratoderma in mother
Figure 3

Planter keratoderma in baby
Figure 4

Toe nail changes in baby
Figure 5

Finger nail changes in baby
Figure 6

Nail changes in mother
Figure 7

Histopathological picture of the cutaneous cystic lesion of mother showing features suggestive of steatocystoma multiplex magnification for this histological image was H and E, ×100
Figure 8

Histopathological picture of the cutaneous cystic lesion of mother showing features suggestive of steatocystoma multiplex magnification for the histological image was H and E, ×400
From pedigree chart, it was evident that parents and siblings of the female were not affected as well as no other 1st degree relatives of her husband's family were affected.