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# BENIGN HEREDITARY KERATODERMA: A FAMILY WITH THREE CASES AND REVIEW.

Medicine

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## ABSTRACT

In 1971 Brown was first to describe a condition of skin characterised by multiple tiny spiny projections on the palmo -planter surfaces called spiny keratoderma.1 Usually found in isolation but sometimes is associated with malignancies and other systemic diseases. We here in present a family, of which three members had spiny keratoderma. To the best of our knowledge it is the eighth case of hereditary keratoderma described in English literature.

## KEYWORDS

spines, keratoderma, keratin.

**MANUSCRIPT:**

65 year female of average built with underlying Type 2 Diabetes Mellitus and Hypertension was admitted in our department with features of community acquired pneumonia on left side. On routine clinical examination, we found presence of 1-2 mm brown spiny projections on palms and soles of patient (Fig 1). These were distributed uniformly over palmer aspects of hands including digits and soles but sparing dorsum of hands and feet. Patient has been having these spines since childhood and was not disturbing her routine work. These spines often used to regress in size on doing manual work. Her family history was significant in that her 30 year old son and 28 year old daughter had similar spines which appeared around 10 and 12 years of age. Both son and daughter had spiny projections present in palms and soles. They were plucking these spines regularly for cosmetic reasons.

On Systemic examination our patient had features of consolidation in left side of chest. Her CBC showed neutrophilic leukocytosis with a TLC 13.43 X $10^{9}$ . A ski gram of the chest revealed consolidation which resolved after receiving parenteral antibiotics. Kidney function tests, liver function tests, serum calcium and phosphorus were all normal. Patient was treated for community acquired pneumonia.

Histopathology of skin biosy was reported as normal lining epithelium with overlying hyperkeratosis (Fig 2). Patient was diagnosed as familial spiny keratoderma based on the presence of multiple family members affected by this disorder of keratinisation. During hospital stay patients investigations and imaging showed no features of any underlying systemic disease or malignancy. CT scan of chest and ultrsonography abdomen, pelvis were unremarkable.Patient ,her son and daughter are on close followup to look for emergence of any associated systemic disease or malignancy.

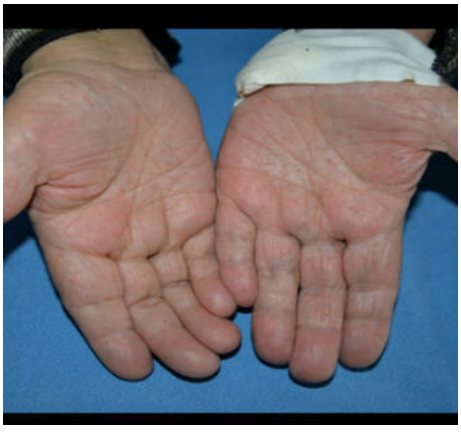

**Fig 1.** Spiny Projections On Palms.

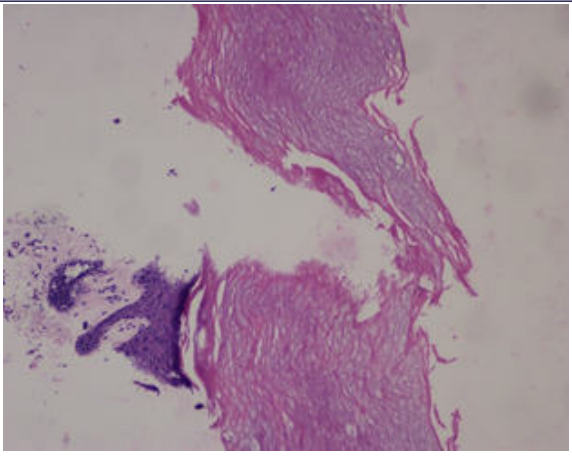

**FIG.2 HYPERKERATOSIS WITH UNDERLYING NORMAL EPIDERRMIS.**

**DISCUSSION:**

Spiny keratoderma is a rare skin disorder which manifests as keratotic, pinpoint papules on the palmer aspects of hands and soles. Both hereditary and acquired forms are known but familial hyperkeratosis has been described in 7 families up to the time this manuscript was compiled. ( Table 1).

| Year of publication | Site | Author(s) | Cases in family |
|---|---|---|---|
| 2003 | USA | Asadi AK. Type I hereditary punctuate keratoderma[25]. | 2 |
| 2004 | India | Gupta R,Mehta S,Pandhi D,Sigal A. Hereditary punctuate palmoplantar keratoderma(PPK).(Brauer-Buschke-Fischer syndrome)[26] | 1 |
| 2009 | Spain | Susana Mallo, Ana I.Bernal, M. Ines Fernandez etal. Autosomal dominant punctuate palmoplantar keratoderma[27]. | 2 |
| 2012 | Canada | Ashey O Toole,Maureen O malley. Unilateral keratoderma in a mother and her son[28]. | 2 |
| 2012 | Spain | Grillo E,Perez-Garcia B Gonzalez-Garcia C etal. Spiky keratotic projections on the palms and fingers. Spiny keratoderma[29]. | 2 |

| | | | |
|---|---|---|---|
| 2012 | India | Pai VV, Kikkari NN, Athanikar SB etal.Type I punctuate palmoplantar keratoderma (Buschke-Fischer Brauer Disease) in a family. A report of two cases[30]. | 2 |
| 2015 | Canada | Alexander Kc Leung and Benjamin Beankin.Punctate palmo plantar keratoderma (Brauer-Buschke-Fischer syndrome) Report of a case and review of literature[31]. | 4 |
| 2019 | India | Present case | 3 |

**TABLE1. HEREDITARY KERATODERMA PUBLICATION LIST:**

The path physiology of spiny keratoderma is not entirely clear and several studies have pointed towards its causation to be to be aberrant hair formation. Kondo S, et al in their study found that keratins 6 and 16, which are markers of hyper proliferating cells, were over expressed in the keratotic projections of spiny keratoderma.[2] Keratins like 6 and 16 cause epidermal hyper -proliferation which produce keratotic projections on palms and soles.[3] Japanese investigators in a study concluded that spiny keratoderma represents aberrant hair formation on the palms and soles.[4] Patient can present to the physician as spiny projections on palms and soles or can be an incidental finding on clinical examination. Histopathology of skin biopsy shows a column of well-defined parakeratotic cells, with underlying hypogranulosis.[5] The spines resemble the spine of an old fashioned music box, corresponding to columns of keratotic materials over a hypo granular epidermis, also referred as music box spine.[6] Hereditary spiny keratoderma usually manifests between the ages of 12 and 50 years while as Acquired spiny keratoderma typically presents after the age of 50 years, although cases of acquired spiny keratoderma in patients as young as 35 years of age have been reported.[7,4] In the review of 28 spiny keratoderma patients, approximately 19% were hereditary.[8]The later is an autosomal dominant trait, benign and often found in isolation.[9] No association with systemic disease or malignancy has been reported with hereditary form of spiny keratoderma.[10] Acquired spiny keratoderma can precede or can be coincidental with associated malignancies.[8] Multiple systemic diseases and malignant conditions (Table 2) have been found to be associated with acquired form of spiny keratoderma and those patients with spiny keratoderma who present at an older age need proper evaluation and monitoring.[9] The connection between spiny keratoderma and the systemic diseases remains unclear, but since this dermatosis is likely under diagnosed, more reported cases will enable further understanding of this relationship.

Biopsy of skin reveal a compact column of hyperparakeratosis originating from the stratum corneum, and a hypo granular epidermis directly beneath it. The column is sharply demarcated from adjacent skin that consists of an orthokeratotic stratum corneum.[21] A close differential of spiny keratosis is porokeratosis which has similar histologic features. But spiny keratoderma does not show vacuolization and/or dyskeratosis of underlying spinous layer, cornoid lamella, or lymphocytic infiltration of the papillary dermis as seen in porokeratosis. Differentiation between the two is important as porokeratosis is associated with basal and squamous cell carcinoma.

Various modalities of treatment have been tried most of them with unsatisfactory results. However promising results have been obtained with 5-fluoruracil[22] , topical tacalcitol 0.002% ointment (an active form of vitamin D)[23] and acitretin[24] .Spines usually reappear after treatment is interupted. Besides the cosmetic problem of spiny keratoderma, the awareness and management of possible underlying malignancy and systemic conditions is important.

**Table 2. Causes Of Spiny Keratoderma.**

| | |
|---|---|
| 1. Hereditary[8] | |
| 2. Systemic diseases | Darier's Disease[11]<br>Chronic renal failure[12]<br>Myelofibrosis[13]<br>Type IV hyperlipoproteinemia[7]<br>Adult polycystic kidney disease with liver cysts[9] |
| 3. Malignancies | Rectal carcinoma[14]<br>Bronchial carcinoma[15]<br>Renal cell carcinoma[16]<br>Breast carcinomas[17]<br>Leukemia[18]<br>Squamous-cell carcinoma of the skin[19]<br>Melanoma[20] |

**CONCLUSION:**

Although Spiny keratoderma is a rare cosmetic skin disorder which can be managed with local medications, malignancies and systemic disorders can be associated with it which merit proper evaluation and close follow up.

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