

## Short Commentary

### Hereditary Papulotranslucent Acrokeratoderma with Deafness

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A 6-year-old boy was referred to our department with a history of persistent eruptions on the palm, dorsal aspect of the finger/toe joints and knees for 6 months of age. The boy also had deafness and signs of mild pervasive developmental disorder since birth. The boy does not have a family history of similar dermatosis. On initial examination, multiple, translucent, skin-colored, smooth-surfaced, flat-topped, round or polygonal papules, 2-4 mm in diameter, were noted bilaterally and roughly symmetrically over both palms, especially at the thenar and hypothenar eminences, dorsal aspects of the interphalangeal joints of the fingers and toes, and over both knees (Figure 1a,b,c). There were no eruptions over the soles of the feet. Histopathologic examination of biopsy specimens from the left foot revealed striking convex epidermal hyperkeratosis, hypergranulosis, acanthosis and slight perivascular inflammatory infiltration in superficial dermis (Figure 2a). Examination of sections stained with Elastica van Gieson stain revealed no evidence of decrease or tearing of the elastic fibers. Hematologic and blood biochemical testing revealed no abnormalities. On the audiogram, both ears were rated as deaf (mean hearing level: 95dB on the right side and 129 dB on the left) (Figure 2b).



Figure 1a: Diffusely Distributed, Translucent, Skin-Colored Papules over the Left Palmar Thenar Eminence.



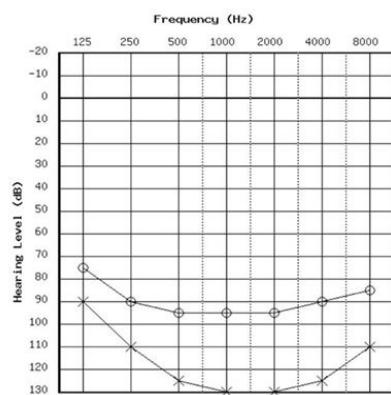
Figure 1b: Multiple Confluent, Translucent, Skin-Colored Papules Over the Dorsal Aspect of the Interphalangeal Joints of the Fingers.



Figure 1c: Conglomerate, Translucent, Skin-Colored Papules over the Dorsal Aspect of the Interphalangeal Joints of the Toes.



Figure 2a: Histologic Section of a Papule Showed Striking Convex Hyperkeratosis (Hematoxylin& Eosin Stain).



**Figure 2b:** Audiograms Showing Bilateral Deafness.

Hereditary Papulotranslucent Acrokeratoderma (HPA) is a rare disease inherited in an autosomal dominant manner, that was initially reported as a subtype of familial punctate keratoderma in 1973 by Onwukwe et al. [1-8]. Hereditary keratosis is known to be occasionally complicated by sensorineural hearing loss. We report a case of HPA complicated by sensorineural deafness.

A diagnosis of HPA was made, as the characteristics, distribution and histologic features of the eruptions were consistent with those described by Onwukwe et al. [1]. The boy was diagnosed as having HPA complicated by bilateral deafness and mild pervasive developmental disorder. Diseases which need to be distinguished from HPA include acrokeratoelastoidosis, punctate palmoplantar keratosis and dermatomyositis. In the present case, the clinical findings, blood test data and histopathological findings were useful for excluding these diseases.

In regard to the ichthyosis syndrome, reports of hereditary keratosis complicated by sensorineural hearing loss or deafness are available for Refsum's disease (MIM 266500), X-linked dominant ichthyosis (Conradi-Hunermann-Happle syndrome, MIM302960), KID syndrome (MIM 148210) and neutral lipid storage disease (Chanarin-Dorfman syndrome, MIM275630) [9]. In regard to conditions associated with palmoplantar keratosis, reports are

available on keratoma hereditaria mutilans (Vohwinkel's syndrome, MIM124500) [10] and palmoplantar keratosis associated with mitochondrial gene abnormalities [11]. To date, however, no case of HPA complicated by deafness has been reported, and the present case is the first such case.

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