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We hope that making available the relevant information on Pachyonychia Congenita will be a means of furthering research to find effective therapies and a cure for PC.
Figure 1. A) At the age of 33 years: appearance of premature aging. B) Many lax papules and plaques on her abdomen. C) Red urticarial vasculitis-like plaques on the right forearm. D) Elastic fibers were diffusely decreased in all layers of the dermis (elastic tissue strain, ×100). E) Elastic fibers are shortened and fragmented only within the inflammatory infiltrate of neutrophils around the vessels and between the collagen bundles (elastic tissue strain, ×200). F) At present: therapy after rhytidectomy and blepharoplasty.

the progression of cutis laxa. How to hold back progression of the disease is still a challenge.


A novel mutation (p.Arg94Gly) of keratin 17 in a Chinese family with steatocystoma multiplex

Steatocystoma multiplex (SM) is an uncommon, autosomal dominant disorder affecting the pilosebaceous unit, characterized by widespread sebum-containing dermal cysts that may occasionally rupture, become inflamed, and heal with scarring (SM suppurativum) [1]. SM was demonstrated to
be caused by mutations in the keratin 17 (KRT17) gene, which is also associated with pachyonychia congenital type 2 (PC-2) [1, 2]. Here, we report a Chinese SM family with a novel mutation in KRT17.

The proband was a 28-year-old Chinese man who was referred to our clinic for multiple cysts. When he was 17, multiple papules and cysts developed on his face, chest and axilla. These cysts gradually grew larger and more extensive, varying in size from 0.1 to 5 cm in diameter. Some cysts were repeatedly infected and discharged yellowish, oily material. Physical examination showed multiple soft cysts and nodules on his face, nape, trunk, groin, and thighs (figure 1A) with atrophic scars. The nail, palmoplantar areas, mouth, tongue and teeth were normal. Histologic examination of a subcutaneous cyst revealed that the cyst wall was composed of several layers of epithelial cells accompanied by sebaceous gland lobules (figure 1C). There were 7 affected members among 15 people in the pedigree (figure 1D). Other patients in this family had similar symptoms, the females were less affected and their lesions were found only in the axilla (figure 1B).

After informed consent, genomic DNA was extracted from all available family members. Exon 1 of the KRT17 gene was amplified by polymerase chain reaction (PCR) with specific primers, to avoid pseudogene contamination, as described previously [3]. The amplified products were purified and directly sequenced. A novel heterozygous mutation (c.280C>G) in the helix initiation motif of the KRT17 gene was found, which resulted in a predicted substitution mutation, R94G (figure 1E). This mutation was verified through the same PCR and sequencing in other family members and 50 unrelated controls.

The keratin pairs K17 and K6B are expressed in nail bed, palmoplantar epidermis (restricted), epidermal appendages, sebaceous glands, hair shafts, other epithelia and wound healing [3]. Keratins consist of an alphahelical rod domain with four helical segments (1A, 1B, 2A, 2B). To date, almost all mutations in PC-2 and SM have
been reported within the highly conserved helix boundary domains at either end of the alpha-helical rod domain of keratin [4].

In total, 18 mutations in KRT17 have been reported (http://www.interfil.org). Most KRT17 mutations were found in PC-2 families and only 4 mutations were reported in SM families without nail changes. In PC-2 and SM, the majority of the mutations were found in the helix initiation motif of K17 with mutations at two codons, N92 and R94, accounting for more than 50% cases. This area is highly conserved and even subtle amino acid changes are thought to disrupt intermediate filament formation and assembly [2, 3].

Although SM and PC-2 patients share the same K17 mutation, R94C and R94H, some have the typical PC-2 nail dystrophy and epidermal cysts, whereas others have only SM without nail changes [1, 2, 4, 5]. It is suggested that the variable phenotype is not due to the specific K17 mutation alone, but also depends on a combination of other factors, e.g. androgenic stimulation. Sebum synthesis could be stimulated by androgen. Androgen could also increase downstream PPAR expression, which in turn transduces the terminal differentiation of sebocytes [6]. In this study, the female patients had mild SM lesions, only in their axilla, which supports that androgenic stimulation might be an important factor in SM.

In summary, we report a novel missense mutation of K17 in a Chinese SM family. This may help in expanding the molecular mutation spectrum of K17 in SM.


Lipedematous scalp with heterochromia of scalp hair in a boy

Lipedematous scalp (LS) is a rare condition characterized by a thickened and boggy scalp, due to hyperplasia of the subcutaneous fat layer, without any hair loss. To date, twenty-two cases have been reported in English with a predilection for middle-aged women [1-4]. Herein, we report a case of LS with heterochromia of scalp hair in a boy.

An 11-year-old Chinese boy presented with an eight-month history of a circumscribed area of lighter hair on the vertex with gradually asymptomatic thickening of the underlying scalp. There was no history of trauma or hair dyeing. The family history was also unremarkable. Physical examination revealed a circumscribed, round patch (4 cm × 4 cm in size) of lighter hair on the vertex without shortening or loss of hair (figure 1A). On palpation, the underlying scalp was found to be thickened, soft and boggy compared to the unaffected areas. No inflammation or color change was observed. Histopathological examination of a punch biopsy specimen from the affected scalp revealed an irregular epidermis, a mild perivascular lymphocytic inflammatory infiltrate in the dermis and marked hyperplasia of the subcutaneous fatty tissue. The number and distribution of terminal hair follicles were normal. No mucinous material was observed (figure 1B). Magnetic resonance imaging (MRI) displayed diffuse thickening of the subcutaneous fat layer at the vertex with a maximum thickness of 11 mm, whereas unaffected scalp measured 5 mm (figure 1C).

Figure 1. A) A circumscribed patch of lighter hair emerging from the vertex whorl. B) Scalp biopsy from the affected area shows a mild perivascular lymphocytic inflammatory infiltrate in the dermis and marked hyperplasia of the subcutaneous fatty tissue and normal morphology of the hair follicles (HE stain). C) Sagittal T2-weighted MR image shows thickening of the subcutaneous fat tissue at the vertex (arrows indicate the affected area).