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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.
Pachyonychia congenita with corneal dystrophy

Dear Editor,

Pachyonychia congenita (PC) is a group of rare autosomal dominant keratinizing disorders caused by mutation in one of the four keratin genes, KRT6A, KRT6B, KRT16 and KRT17. We report the case of a 9-year-old boy with a family history negative for dermatological diseases presenting to our department with marked vision loss and gross thickening nails (Fig. 1). Other features included scant eyebrow, thinning hair and hyperkeratosis on the soles, which appeared 4 years prior. The patient reported pain while walking. His medical history revealed photophobia, corneal and ciliary congestion in both of the eyes. According to the examination, his eyes visual acuity was 0.25, with a large number of new blood vessels from neighboring areas into the substantia propria layer and former elastic substrate layer of the cornea, with only pupil area translucent. Widely, epithelium geographic ulcer and hyperkeratosis of the cornea were observed, with flusress dyeing positive. The anterior chamber depth was normal, pupils 3 mm in diameter and sensitive to light. The lenses were transparent, however, the vitreous body and fundus were fuzzy. Oral, laryngeal and dental changes, pigment anomaly, follicular hyperkeratosis, cutaneous cysts and hyperhidrosis were not detected. No abnormalities on physical, neurological and laboratory examinations were noticed. This report is the first detailed described case of PC with corneal dyskeratosis. The study was approved by the Third Military Medical University Review Board. After the informed consent was obtained from the patient, mutation analysis was performed on this young boy by sequencing the whole exons and the exon–intron boundaries of the four keratin genes, as described by Wilson et al.1 However, no mutation was detected.

Pachyonychia congenita is usually a rare inherited genodermatosis. Numerous subdivisions of pachyonychia congenita have been suggested. According to the classification devised by Feinstein et al.,2 leukokeratosis of the cornea and cataracts can be classified as PC-3, and hair anomalies and alopecia are typical features of PC-4. Leachman et al.3 analyzed clinical, pathological and genetic data from the published work and two research registries. They proposed that corneal dystrophy appears to be a specific clinical feature of PC-2. However, there was no detailed clinical report on corneal changes in PC patients till today. Eliason et al.4 surveyed 254 individuals with confirmed keratin mutations regarding their experience with PC, but found no PC patients with corneal dystrophy. They recommended the elimination of the terms “PC-1” and “PC-2” and proposed replacement with notation of the specific keratin defect, such as PC-6a, PC-6b, PC-16, PC-17 and PC-U, which means absence of a known PC keratin gene mutation. According to our findings, the boy could be classified as being PC-U.

Figure 1. (a) Thickening of all fingernails and toenails and palmoplantar hyperkeratosis. (b) Corneal hyperemia, dystrophy, ulcer and dyskeratosis. (c) Scant eyebrow and hair.

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Conflict of interest: None declared.

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