Porokeratosis (PK), first described in 1983, is a chronic parakeratotic skin disorder. Clinical types include porokeratosis of Mibelli (PM), disseminated superficial PK, disseminated superficial actinic porokeratosis (DSAP), facial PK, and pychotropica porokeratosis (PP). Dermoscopic images of PK show an obvious annular margin with scales and an atrophied center.\(^1\) Wood’s lamp pictures of PK present a diamond necklace-like structure.\(^2\) Gene mutations in mevalonate pathway enzymes, such as mevalonate kinase (\(MVK\)), phosphomevalonate kinase (\(PMVK\)), mevalonate decarboxylase (\(MVD\)), and farnesyl diphosphate synthase (\(FDPS\)), may be involved in the pathogenesis of PK.\(^3,4\) In the present study, five sporadic patients with PK were recruited. Dermoscopic features were investigated, and genetic testing was conducted.

Sporadic patients with PK were diagnosed on the basis of typical clinical and histopathological features. PK is clinically characterized by a ring-like structure with an elevated border and atrophic center. Pathologically, PK is characterized by a parakeratotic column in the epidermis. The lesions selected for dermoscopic imaging were approximately 0.5 cm in diameter. The lesions were imaged under original magnification ×50 and then photographed under a dermoscope with white light. Venous blood was collected from all subjects to detect \(MVK\), \(PMVK\), \(MVD\), and \(FDPS\) genes. Meanwhile, blood samples from 100 controls with normal phenotype were collected to rule out polymorphism. Genomic DNA was isolated from whole blood using a whole-blood genomic DNA extraction kit (Aidlab Inc., Beijing, China). \(MVK\), \(PMVK\), \(MVD\), and \(FDPS\) sequences were obtained from the National Biologic Information Center gene pool and University of California Santa Cruz database. Primers were designed through Primer Premier 5. Polymerase chain reaction (PCR) was performed in ABI GeneAmp 9700 PCR amplifier (Applied Biosystems, Foster, USA). Bidirectional DNA sequencing was used to validate the detected gene mutations.

Among the five sporadic patients, two suffered from DSAP, two manifested facial PK, and one was diagnosed with PM. The male-to-female ratio was 2:3. The onset age ranged from 10 years old to 67 years old. Disease duration ranged from 3 years to 39 years. One patient suffered from light pruritus. Two patients presented with accompanying seborrheic keratosis. Many treatments, such as cryotherapy, retinoic acid ointment, and carbon dioxide laser treatment, exerted no significant effect on PK. The dermoscopic appearance of two patients with DSAP presented a brown ring-like structure with a raised border and scar-like center. Numerous scales were located on the border [Figure 1a]. The dermoscopic results for two patients with facial PK exhibited a black circular structure with a slightly raised border and atrophic center [Figure 1b]. The dermoscopic images from one patient with PM showed a brown annular structure with a ridged border and scar-like center [Figure 1c]. A novel \(MVK\) splicing mutation, designated as IVS4+1G>A (NM_000431.3), was detected in one patient with facial PK [Figure 1d].

The onset age of sporadic PK is in accordance with that reported in the literature.\(^5\) PK is a chronic keratinization disorder with many different clinical types. DSAP and PM have generally been known by dermatologists. Facial PK, a rare variant, was detected in one patient with facial PK [Figure 1d].

Address for correspondence: Dr. Shi Lian, Department of Dermatology and Venereology, Xuanwu Hospital, Capital Medical University, Beijing 100053, China E-Mail: drlianshi@sina.com

This is an open access article distributed under the terms of the Creative Commons Attribution-NonCommercial-ShareAlike 3.0 License, which allows others to remix, tweak, and build upon the work non-commercially, as long as the author is credited and the new creations are licensed under the identical terms.
patients with PK had at least one mutation in four genes in the mevalonate pathway. This study only found a novel *MVK* splicing mutation, which was designated as IVS4+1G>A, in a sporadic patient with facial PK possibly because PK can be caused by other factors, such as sunlight exposure. The novel *MVK* gene mutation (IVS4+1G>A) may result in the absence of 52 amino acids (GPAEPGYRSVVGAAPRGGLQLRRGLGVSGSSPP DCVRGDPPAEGRGLRQQ) or two alpha helixes that are encoded by exon 4. The abnormal *MVK* structure caused by *MVK* gene mutations may negatively affect the mevalonate pathway. Finally, the growth of keratinocytes was abnormal and PK was caused. Zhang et al. also discovered a novel *MVK* splicing mutation, which was designated as IVS4+2T>A, in a patient with PP.

In conclusion, dermoscopic characteristics of PK may not be correlated with clinical type but with lesion sites. However, this hypothesis deserves further study. A novel *MVK* splicing mutation (IVS4+1G>A) has expanded the database of *MVK* mutations and may help elucidate the pathogenesis of PK.

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and attempts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship
This study was supported by a grant from the Beijing Natural Science Foundation (No. 7163216).

Conflicts of interest
There are no conflicts of interest.

REFERENCES
1. Nicola A, Magliano J. Dermoscopy of disseminated superficial actinic porokeratosis. Actas Dermsosifiliogr 2016. doi: 10.1016/j.ad.2015.09.025. [Epub ahead of print].
2. Sun R, Chen H, Zhu W, Lian S. Wood’s lamp image of porokeratosis. Photodermatol Photoimmunol Photomed 2017;33:114‑6. doi: 10.1111/phpp.12285.
3. Zhang Z, Li C, Wu F, Ma R, Luan J, Yang F, et al. Correction: Genomic variations of the mevalonate pathway in porokeratosis. Elife 2016;5:e14383. doi: 10.7554/eLife.14383.
4. Li M, Li Z, Wang J, Ni C, Sun Z, Wilson NJ, et al. Mutations in the mevalonate pathway genes in Chinese patients with porokeratosis. Eur J Dermatol Venereol 2016;30:1512‑7. doi: 10.1111/jdv.13653.
5. Gutierrez EL, Galarza C, Ramos W, Tello M, De Paz PC, Bobbio L, et al. Facial porokeratosis: A series of six patients. Australas J Dermatol 2010;51:191‑4. doi: 10.1111/j.1440-0960.2009.00616.x.