CORRESPONDENCE

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Response to comment on "Birt-Hogg-Dubé syndrome in Korean: clinicoradiologic features and long term follow-up"

Joo Hee Lee[1], Min Joo Jeon[1], Joon Seon Song[2], Eun Jin Chae[3], Jin-Ho Choi[4], Gu-Hwan Kim[5], and Jin Woo Song[1]

Departments of [1]Pulmonary and Critical Care Medicine, [2]Pathology, [3]Radiology, and [4]Pediatrics, [5]Medical Genetics Center, Asan Medical Center, University of Ulsan College of Medicine, Seoul, Korea

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Correspondence to
Jin Woo Song, M.D.
Department of Pulmonary and Critical Care Medicine, Asan Medical Center, University of Ulsan College of Medicine, 88 Olympic-ro 43-gil, Songpa-gu, Seoul 05505, Korea
Tel: +82-2-3010-3993
Fax: +82-2-3010-6968
E-mail: jwsong@amc.seoul.kr
https://orcid.org/0000-0001-5121-3522

Thank you for your interest in the article entitled "Birt-Hogg-Dubé syndrome in Korean: clinicoradiologic features and long term follow-up" and your comments. As you commented, we cited two papers as references that presented a lower incidence of skin lesions in Asian patients with Birt-Hogg-Dubé (BHD) syndrome compared with Caucacian patients [1,2].

In a study by Kunogi et al. [1], a folliculin (*FLCN*) germline mutation was found in 23 of the 36 patients by denaturing high performance liquid chromatography. The remaining 13 patients were examined by a real-time quantitative polymerase chain reaction, and a genomic deletion was found in two patients. Accordingly, an *FLCN* germline mutation was found in 25 of the 36 patients with multiple lung cysts. All patients were screened for skin lesion by dermatologists and skin lesions were detected in seven (28%) patients, and typical skin lesion (fibrofolliculoma) was confirmed in only one patient (4%) by skin biopsy. In another study by Furuya et al. [2], 76 of 156 *FLCN* mutation carriers (48.7%) had skin papules or tumors; however, typical fibrofolliculomas were confirmed in six (3.8%) by skin biopsy.

In Asian patients with BHD syndrome, low histologic confirmation rate may be due to inconspicuous skin lesions. In a study by Furuya et al. [2], with the exception of one individual, none of these patients cared or consulted dermatologists because skin manifestations were not their major complaint. Even though other biopsied papules macroscopically looked like fibrofolliculomas, histologic diagnosis varied and included perifollicular fibromas and sebaceous hyperplasia. Therefore, in the case of histologically unidentified skin lesions, the likelihood of fibrofolliculoma will be low. This also suggest that lung and kidney lesion may be more informative than fibrofolliculomas as diagnostic criteria for BHD syndrome in Asian populations [2].

Although one recent paper mentioned in the correspondence suggested a relatively higher incidence of skin lesions in Japanese patients with BHD syndrome [3], in addition to previous Japanese papers, recent papers including Korean and Chinese patients also suggested low incidence of typical skin lesions in Asian patients with BHD syndrome (20% in Korean and 7.4% in Chinese) [4,5].

We appreciate your interests in our article and we hope that our explanation has addressed your questions.

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Conflict of interest

No potential conflict of interest relevant to this article was reported.

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