

## 양측성 신장 종양 환자에서 시행한 유전자 검사를 통해 진단된 Birt-Hogg-Dubé 증후군 1예

서울대학교병원 신장내과<sup>1</sup>, 서울대학교병원 비뇨기과<sup>2</sup>, 서울대학교병원 영상의학과<sup>3</sup>  
서울대학교병원 병리과<sup>4</sup>, 서울대학교 어린이병원 소아과<sup>5</sup>

박석우<sup>1</sup>, 박세훈<sup>1</sup>, 강은정<sup>1</sup>, 곽철<sup>2</sup>, 윤순호<sup>3</sup>, 이경분<sup>4</sup>, 정해일<sup>5</sup>, 김연수<sup>1</sup>, 이하정<sup>1</sup>

### A Case of Bilateral Multifocal Renal Tumors Diagnosed as Birt-Hogg-Dubé Syndrome Confirmed by Genetic Analysis

Seokwoo Park<sup>1</sup>, Sehoon Park<sup>1</sup>, Eunjeong Kang<sup>1</sup>, Cheol Kwak<sup>2</sup>, Soon Ho Yoon<sup>3</sup>  
Kyungboon Lee<sup>4</sup>, Hae Il Cheong<sup>5</sup>, Yon Su Kim<sup>1</sup>, Hajeong Lee<sup>1</sup>

Department of Internal Medicine<sup>1</sup>, Seoul National University College of Medicine,  
Seoul National University Hospital, Seoul, Korea

Department of Urology<sup>2</sup>, Seoul National University College of Medicine, Seoul, Korea

Department of Radiology<sup>3</sup>, Seoul National University College of Medicine and Institute of  
Radiation Medicine, Seoul National University Medical Research Center, Seoul, Korea,

Department of Pathology<sup>4</sup>, Seoul National University College of Medicine,

Seoul National University Hospital, Seoul, Korea

Department of Pediatrics<sup>5</sup>, Seoul National University Children's Hospital, Seoul, Korea

Birt-Hogg-Dubé syndrome (BHD) is an autosomal dominantly inherited cancer susceptibility syndrome characterized by skin fibrofolliculomas, pulmonary cysts and spontaneous pneumothorax, and renal cancers. The syndrome is caused by various germline mutations of the FLCN gene located in 17p11.2 encoding folliculin. In this study, we described a case of 50-year-old-woman who presented with flank pain. A CT scan of her abdomen and pelvis showed multifocal tumors with heterogenous enhancement in both kidneys including a 3.5 cm enhancing mass on the upper pole of the right kidney. To differentiate a metastatic cancer from primary kidney tumors, we performed PET-CT which showed no other hypermetabolic lesion suggestive of a primary site of metastatic cancer, leading us to suspect a primary renal cancer. Her chest CT showed a few thin-walled oval and lentiform cysts up to 1.5 cm at bilateral lower lungs. A needle biopsy of 3.5 cm mass on the right kidney revealed that the mass was chromophobe type renal cell carcinoma. Moreover, cystic changes of lungs were compatible with the findings in BHD. Genetic analysis performed to confirm the diagnosis of BHD recognized a single nucleotide deletion c.1557delT in exon 14. Considering the fact that renal cancers might be the principal determinant of life expectancy in BHD-affected patients, this report is noticeable as the mentioned mutation has not been associated with renal cancer before.

**Key Words:** Birt-Hogg-Dubé, 증후군, 신장암, 폐낭종

Birt-Hogg-Dubé syndrome, Renal cancer, Lung cysts