

Supplementary Table 1. *FLCN* haplotypes were reconstructed using four single nucleotide polymorphisms in 60 Korean individuals (56 normal control individuals and four patients with Birt-Hogg-Dubé syndrome)

Number of haplotypes	Haplotype	Count
H1	ATTT	13
H2	ATCC	12
H3	ACTT	4
H4	ACCC	35
H5	GTTT	16
H6	GTCC	17
H7	GCTT	18
H8	GCCC	5

Supplementary Table 2. Haplotypes of the *FLCN* gene reconstructed in patients with the c.1539-2A>G variant

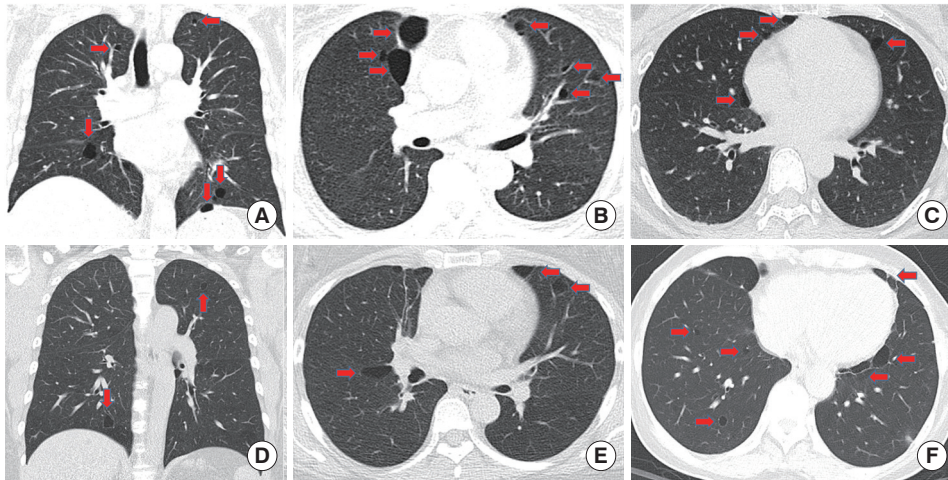
ID	Alleles				Haplotypes*
	rs56926918	rs56153623	rs2072652	rs2075659	
1	A/A	C/T	C/T	C/T	H1, H4
3.I.2	A/A	C/T	C/T	C/T	H1, H2

*H1, haplotype 1: ATTT; H2, haplotype 2: ATCC; H4, haplotype 4: ACCC.

Supplementary Table 3. Pulmonary function test results from patients with Birt-Hogg-Dubé syndrome

ID	FEV ₁ /FVC	FVC (L)	FVC (%pred)	FEV ₁ (L)	FEV ₁ (%pred)	DL _{CO} adj (mL/mmHg/min)	DL _{CO} adj (%pred)	DL _{CO} /VA adj (mL/mmHg/min/L)	DL _{CO} /VA adj (%pred)	Interpretation	
										Spirometry	Diffusing capacity
1	0.78	3.09	98	2.4	101	18.3	83	4.42	112	Normal ventilatory function	Normal diffusing capacity
2	0.70	2.72	92	1.89	88	12.7	69	3.21	84	Normal ventilatory function	Mildly decreased diffusing capacity
3.1.2	0.80	1.61	67	1.26	76	13.4	75	4.86	126	Mild restrictive pattern	Mildly decreased diffusing capacity
4	0.70	3.15	101	2.21	93	NT	NT	NT	NT	Normal ventilatory function	NT
Mean ± SD	0.75 ± 0.05	3 ± 0.71	80 ± 15.46	2 ± 0.50	90 ± 10.47	15 ± 3.05	76 ± 7.02	4 ± 0.85	107 ± 21.39		
All										1/4 (25%)	2/3 (67%)

Abbreviations: FEV₁, forced expiratory volume in one second; FVC, forced volume vital capacity; %pred, the percentage of predicted values; DL_{CO} adj, diffusing capacity for carbon monoxide, adjusted for hemoglobin concentration; DL_{CO}/VA adj, the ratio of diffusing capacity for carbon monoxide to alveolar volume, adjusted for hemoglobin concentration; NT, not tested.



Supplementary Fig. 1. Low-dose chest computed tomography imaging in patients with Birt-Hogg-Dubé syndrome. (A) and (B) Patients with the *FLCN* c.1539-2A>G variant. Bilateral multiple irregularly shaped cysts (red arrow) are seen in the lower and upper lobes; (C) The first daughter of the proband (A) and (B) with the identical *FLCN* variant as the proband. Bilateral multiple-round cysts are seen in the upper lobes (D) and (E). Another patient has the *FLCN* c.1539-2A>G variant. Bilateral multiple irregularly shaped cysts are seen in the lower and upper lobes; (F) A patient with the *FLCN* c.1215C>G variant. Multiple bilateral cystic lesions are seen in the lower lobes.