Review of: "A retrospective two centre study of Birt-Hogg-Dubé syndrome reveals a pathogenic founder mutation in FLCN in the Swedish population"

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Potential competing interests: The author(s) declared that no potential competing interests exist.

Dear Editor,

Thank you very much for sending me the article regarding analysis of Birt Hogg Dubé syndrome in Swedish population. Taking into account the rarity, and huge underdiagnosis of the disease authors collected one of the largest group of patients in Europe, but not the largest (vande Beek I et al.). I would like to congratulate the authors.
However, some aspects of presented population require revision.
What was the observation period?
How long was the diagnosis delay?
There is disproportion between number of patients with pneumothorax and number of patients with detected lung cysts , 40%(75/186) had at least one episode of spontaneous pneumothorax and 26% (49/186) had pulmonary cysts in imaging control prior or after the genetic testing. Please explain this.
How many cases had X- ray picture, and how many had HRCT examination?
Did you perform lung function assessment?
Did all presented patients have an MRI of abdomen at the time of genetic testing?
How often the follow-up visit has been performed?
How many visits/patient were noticed in the time of follow-up?
Did you observe patients with clinical symptoms of BHDS (fibrofolliculoma, lung cysts and/ of kidney tumor) but without detected mutation of *FLCN?*

van de Beek I, Glykofridis IE, Wolthuis RFM, et al. No evidence for increased prevalence of colorectal carcinoma in 399 Dutch patients with Birt-Hogg-Dubé syndrome. Brit J Cancer. 2020; 122: 590–594.