CLINICAL SNAPSHOT

Hornstein–Knickenberg Syndrome: Potential Diagnosis at a Glance

Figure 1: The patient's facial fibrofolliculomas

Figure 2: Coronal high-resolution computed tomography of the thorax in a female patient with numerous pulmonary cysts of varying size, predominantly basal

A 67-year-old woman, a multimorbid ex-smoker with heart failure, was transferred to our department for weaning from mechanical ventilation after suffering acute respiratory insufficiency due to bilateral pneumonia. Intubation had been followed by bilateral pneumothorax. Weaning was successful. There were four main differential diagnoses for the thin-walled pulmonary cysts seen on thoracic computed tomography: lymphangioleiomyomatosis, pulmonary Langerhans cell histiocytosis, lymphocytic interstitial pneumonia, and Hornstein–Knickenberg syndrome (also known as Birt–Hogg–Dubé syndrome), which is associated with facial fibrofolliculomas. Facial fibrofolliculomas had been histologically confirmed in our patient 20 years previously, but in spite of her recurring episodes of spontaneous pneumothorax the diagnosis of Hornstein–Knickenberg syndrome had never been made. This autosomal dominant hereditary syndrome, caused by a mutation in the *folliculin* gene, is associated with an elevated risk of benign and malignant tumors of the kidney: renal cell cancer ensues in approximately 20% of cases, usually from the age of 50 years onward. The patient and her relatives were advised accordingly and recommended to undergo genetic counseling. Renal neoplasia was not present at the time of diagnosis.

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Conflict of interest statement: The authors declare that no conflict of interest exists.

Translated from the original German by David Roseveare.

Cite this as: Klein M, Koch M, Pfeifer M: Hornstein–Knickenberg syndrome: potential diagnosis at a glance. *Dtsch Arztebl Int* 2021; 118: 554.
 DOI: 10.3238/arztebl.m2021.0070