A 53-year-old woman was referred to our hospital because of an increasing number of facial skin papules on her cheeks and ears. A biopsy was taken from several of these papules. The location of these lesions combined with the histology confirmed the diagnoses fibrofolliculomas (Fig. 1A and B). Except for facial papules since the age of 26 years, her medical history was unremarkable. The familial history was negative for renal cancer and spontaneous pneumothorax. She never smoked. Her father died of pancreatic cancer at the age of 73 years and her mother of cardiac failure at age 85. Based on the histology and location of the skin lesions, the patient was clinically diagnosed as having Birt–Hogg–Dubé syndrome (BHD). Therefore, she was referred for CT abdomen with i.v. contrast, which showed a solid interpolar tumour in the lower pole of the right kidney, with a diameter of 19 mm, classified as T1N0Mx (Fig. 2). The slices through the basal parts of the lungs revealed multiple lung cysts (Fig. 3).
Partial nephrectomy followed and a clear cell tumour, Fuhrmann grade 2, was found. This combination of skin fibrofolliculomas, renal cell cancer and cysts in the basal parts of the lungs is typical for BHD, an autosomal dominantly inherited cancer disorder, caused by pathogenic FLCN mutations.

Sequencing of the FLCN gene showed a pathogenic splice site mutation in exon 12 (c.[1301-7_1304:delCinsGA]), which confirmed the clinical diagnosis at the DNA level.

Afterwards we identified four additional FLCN mutation carriers in her family. The family data are summarized in Fig. 4. Magnetic resonance imaging (MRI) showed no local recurrence of the removed renal tumour, respectively early detection of a new renal tumour during the 18-month follow-up after surgery. Lifetime frequent renal MRI will be performed in this patient. As the prevalence of renal cancer in BHD patients after initial renal imaging has been described in up to 27% of cases, BHD should be considered when facial fibrofolliculomas are diagnosed and consequently relatives should be encouraged to be screened for genetic testing.

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