GENETICS AND GENODERMATOSES

RENAL TUMORS AND FACIAL PAPULES: A LATE DIAGNOSIS OF A RARE GENODERMATOSIS

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Background: BHD syndrome is an autosomal-dominant, adult-onset condition caused by germline mutations of the folliculin (FLCN) gene. Clinical features include fibrofolliculomas and trichodiscomas, pulmonary cysts, spontaneous pneumothoraces, and renal tumors, most commonly chromophobe and mixed chromophobe/oncocytic variants.

Observation: A 70-year-old Caucasian woman was referred to the urology clinic for evaluation of bilateral renal masses discovered on abdominal CT-scan during a work-up for chronic abdominal pain. Her past medical history was positive for hypothyroidism and atrial fibrillation. A renal biopsy demonstrated a mixed tumor with elements of oncocytoma and chromophobe renal cell carcinoma. This rare histologic finding raised suspicion for Birt-Hogg-Dubé syndrome (BHD). The patient was referred to dermatology for a skin exam, which revealed numerous small, whitish-grey, dome-shaped papules on her nose and cheeks. Crops of these papules were also noted on both her flanks. These asymptomatic lesions started appearing in her thirties and caused her great distress due to their cosmetic appearance. She had undergone laser-resurfacing treatments, but they were discontinued after the lesions continued to recur. Interestingly, she recalled her father having the same facial papules. Shave biopsies of the papules demonstrated fibrofolliculomas, which along with trichodiscomas, are suggestive of BHD syndrome. She denied a history of spontaneous pneumothoraces, but a CT-scan of the thorax revealed four pulmonary cysts. Genetic testing involving sequence analysis was positive for a pathogenic mutation of the FLCN gene. Her renal tumors are being managed by active surveillance, given their indolent nature.

Key message: Fibrofolliculomas and trichodiscomas present as small, whitish-grey, dome-shaped papules of the head, neck, and upper body. They are the most common initial manifestation of BHD with an onset typically in the third decade of life. Their presence should alert the physician to this rare genodermatosis.