CLINICAL IMAGE

Chronic kidney disease caused by tuberous sclerosis complex: lymphangioleiomyomatosis diagnosed in an adult woman

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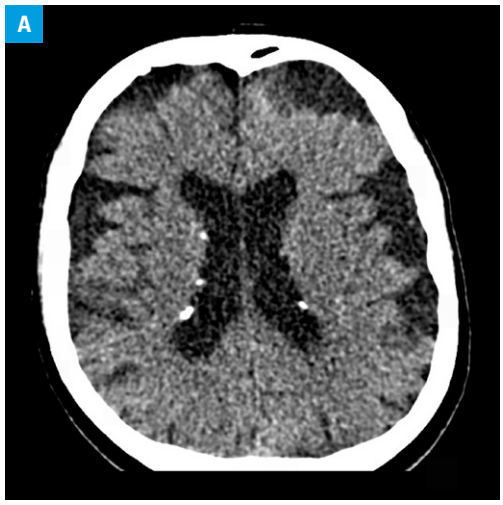
Tuberous sclerosis complex (TSC) is an autosomal dominant genetic disorder characterized by multiple benign tumors, and rarely malignant neoplasms of the skin, brain, eyes, heart, lung, liver, and kidney. Its incidence is 1 in 5000 to 10 000 live births with a high prevalence of lymphangioleiomyomatosis (LAM) in adults.[1] Rarely, it contributes to chronic kidney disease (CKD). Most patients with kidney angiomyolipomas in the Tuberous Sclerosis Registry were asymptomatic (82%).[2]

A 47-year-old woman after right-sided nephrectomy due to bleeding angiomyolipoma, with CKD stage G3bA2, hypertension, diabetes mellitus type 2, epilepsy was admitted to our department to continue the diagnostic workup and prepare her for renal replacement therapy.

In the past, ultrasonography of the abdomen revealed kidney cysts. Soon after, she underwent an episode of a life-threatening abdominal bleeding from angiomyolipoma leading to right-sided nephrectomy in 1992. Since the operation, a progression of CKD has been observed. In 2010 and 2012, computed tomography (CT) of the head performed due to epilepsy demonstrated multiple calcified subependymal nodules in the walls of the lateral ventricles, typical for tuberous sclerosis (FIGURE 1A). In subsequent magnetic resonance imaging (MRI), cortical / subcortical tubers were found, also characteristic of TSC.

On admission, she had chronic fatigue, sporadic dyspnea during physical effort, and oliguria. Physical examination showed skin lesions: ungual fibromas around the toenail (FIGURE 1B), hypomelanic macules on extremities, shagreen patches around the nose (FIGURE 1C), fibrous plaques on the abdomen. Blood test results showed



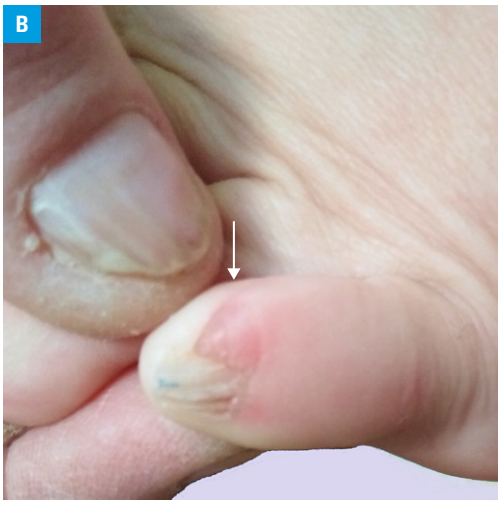


FIGURE 1 Imaging of an adult woman with tuberous sclerosis: **A** – computed tomography of the head showing calcified subependymal nodules; **B** – ungual fibromas around the toenail (arrow)

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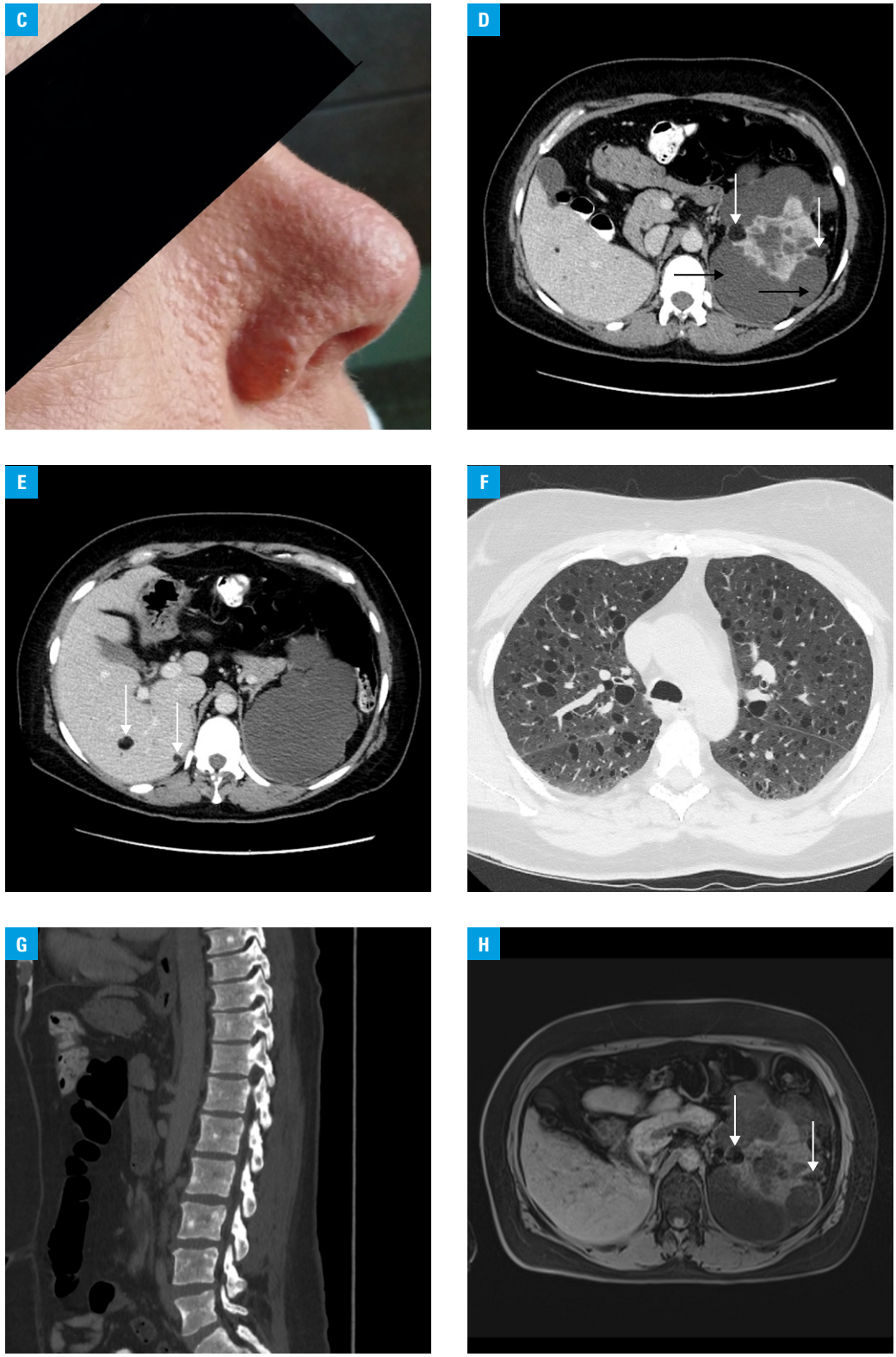

FIGURE 1 Imaging of an adult woman with tuberous sclerosis: **C** – shagreen patches around the nose; **D** – computed tomography of the abdomen, venous phase, showing the left kidney with numerous peripheral cysts (black arrows) and a few angiomyolipomas (white arrows); **E** – computed tomography of the abdomen, venous phase, showing liver angiomyolipomas (arrows); **F** – computed tomography of the chest showing lymphangioleiomyomatosis; **G** – computed tomography showing osteosclerotic foci in the spine; **H** – magnetic resonance of the abdomen, fat saturation sequence, showing angiomyolipomas of the kidney (arrows). The authors obtained informed consent from the patient.

a high level of creatinine of 311 μmol/l (reference range, 44–80 μmol/l), estimated glomerular filtration rate of 15 ml/min/1.73 m^2 (reference range >60 ml/min/1.73 m^2), urea nitrogen level of 19.9 mmol/l (reference range, 2.76–8.07 mmol/l), and parathormone level of 604 pg/ml (reference range, 14.9–56.9 pg/ml). Abdomen CT revealed the left kidney measuring 270 × 142 × 143 mm

with numerous peripheral cysts and a few lesions with fat attenuation, typical for kidney angiomyolipomas as well as a few liver angiomyolipomas (FIGURE 1D and 1E).

Chest CT revealed pulmonary tissue pattern characteristic of LAM (FIGURE 1F). It also confirmed numerous small osteosclerotic foci in the spine that may occur in TSC (FIGURE 1G).

Angiomyolipomas, even fat-poor with no macroscopic foci of fat, may be well visualized on MRI.[3] A month later, during the second hospitalization in our department, MRI of the abdomen confirmed angiomyolipomas of the kidney and liver (FIGURE 1H).

The whole clinical presentation led to the diagnosis of TSC with LAM. The patient had pulmonary consultation and received permanent ambulatory care due to dyspnea and LAM. After consultations and screening tests in CKD stage 4/5, she was registered on the national waiting list for a kidney transplantation.

Usually, TSC is diagnosed in childhood or in young adults. Most commonly, the disease affects the brain (90%) and the kidneys, with angiomyolipomas occurring in 80% and renal cystic disease in 50% of patients. Pulmonary involvement, especially LAM, is the third most common symptom of TSC, affecting 35% of female patients with TSC.[1] Skeletal osteosclerotic foci in tuberous sclerosis were also described in the literature.[4] Moreover, approximately 40% of patients with TSC experience a premature decline in glomelural filtration rate in the absence of angiomyolipoma bleeding or interventions.[5]

Our patient, despite the presence of skin, head, kidney, and pulmonary lesions typical for the genetic disease, had the final diagnosis at the age of 47.

ARTICLE INFORMATION

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CONFLICT OF INTEREST None declared.

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