J Inherit Metab Dis (2007) 30:830–831
DOI 10.1007/s10545-007-0648-1

IMAGES IN METABOLIC MEDICINE

Bilateral renal cell carcinoma development in long-term Fabry disease

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Received: 2 April 2007 / Submitted in revised form: 9 April 2007 / Accepted: 12 April 2007 / Published online: 12 May 2007

A 67-year-old patient was diagnosed with Fabry disease based on a compatible history and absent α-galactosidase A activity in white blood cells. The diagnosis was confirmed genetically (hemizygous for c.427G > A in exon 3). At the age of 33 years, the patient was started on haemodialysis for idiopathic end-stage renal failure. He experienced a cerebrovascular accident at age 42 years and received a renal transplant at age 47 years, which is functional to date (creatinine clearance 37ml/min without proteinuria). At 60 years of age, the patient developed bilateral multifocal renal cell carcinoma (RCC), of both the papillary (chromophilic) type and the eosinophilic variant of the clear cell (conventional) type; both subtypes were present bilaterally (Fig. 1). He underwent bi-lateral nephrectomy (staging T1aN0M0) and has been free of distant metastases since. The resection specimens did not show typical Fabry-related morphological changes, only nonspecific terminal glomerulonephritis with striking hyalinization and Munckenberg-type sclerosis of the vessels and without any nonsclerosed glomeruli.

Bilateral multifocal RCC has mainly been described in familial RCC, Von Hippel–Lindau disease, polycystic kidney disease and tuberous sclerosis. As this is the second report of bilateral RCC in Fabry disease (Blanco et al 2005), there could be a pathophysiological link between Fabry disease and RCC. Owing to earlier detection and improved treatment options for this patient population, their survival is increasing and this type of complication might become more important. Systematic radiological screening of Fabry patients for RCC therefore seems advisable, as might be (genetic) screening for Fabry disease in 'sporadic' bilateral RCC cases.

Communicating editor: Verena Peters

Competing interests: None declared

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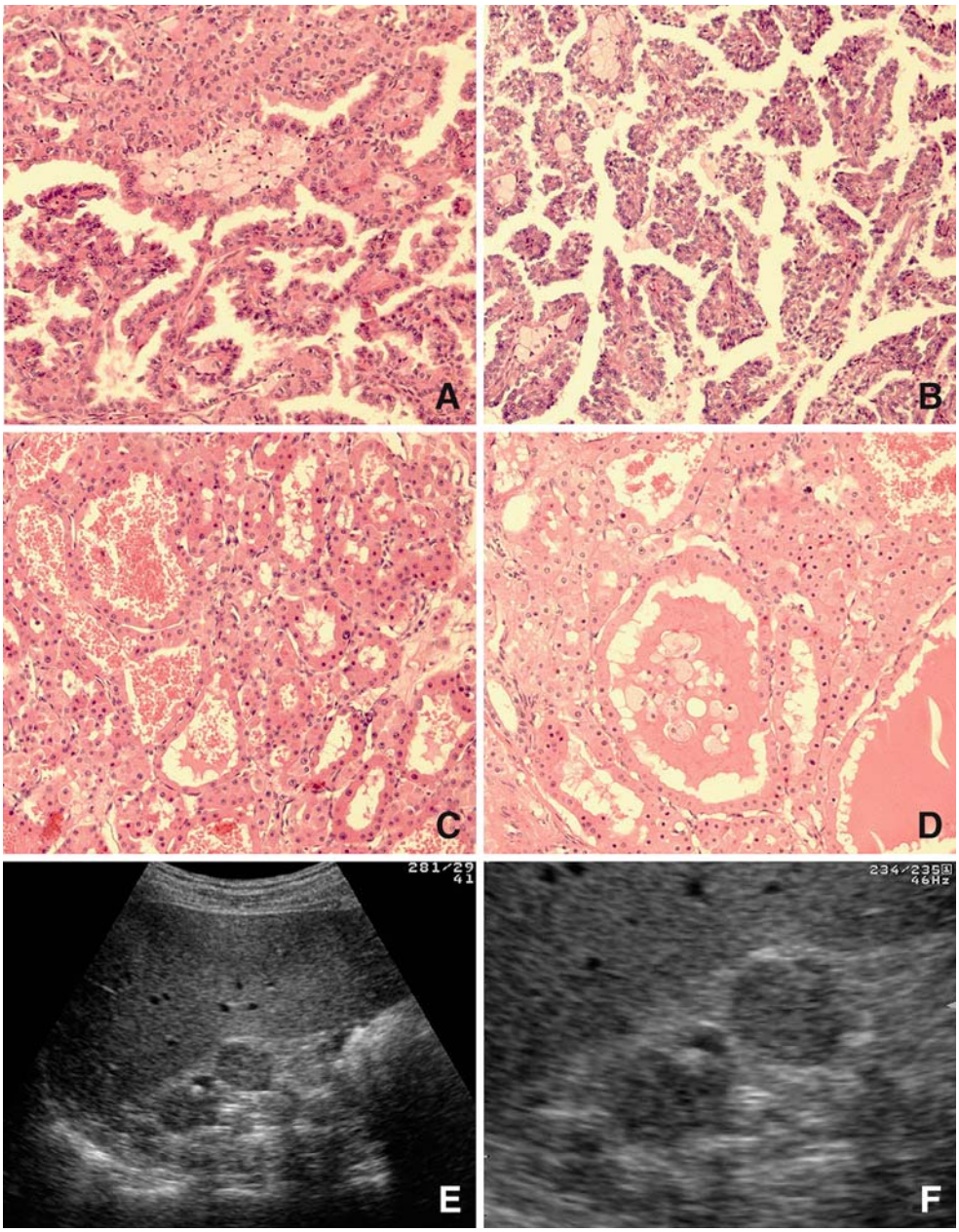

Fig. 1 (**A**) Papillary (chromophilic) renal cell carcinoma (left kidney). (**B**) Papillary (chromophilic) renal cell carcinoma (right kidney). (**C**) Eosinophilic variant clear cell (conventional) renal cell carcinoma (left kidney). (**D**) Eosinophilic variant clear cell (conventional) renal cell carcinoma (right kidney). (**A**–**D**: original magnification 100×, haematoxylin–eosin.) (**E**) Ultrasound image of the right kidney (lower half; upper half is the liver), showing the multiple tissue nodules morphologically identified as (**A**)–(**D**). (**F**) Close-up ultrasound image of the right kidney, showing the same tissue nodules

Reference

Blanco J, Herrero J, Arias LF, Garcia-Miralles N, Gamez C, Barrientos A (2005) Renal variant of Anderson–Fabry disease and bilateral renal cell carcinoma. *Pathol Res Pract* **200**: 857–860.