Clin Exp Nephrol
DOI 10.1007/s10157-013-0876-7

IMAGES IN NEPHROLOGY

Arterial aneurysms: autosomal dominant polycystic kidney disease, Marfan syndrome or both?

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Received: 5 June 2013 / Accepted: 18 September 2013

Keywords Autosomal dominant polycystic kidney disease · Marfan syndrome · Arterial aneurysm

A 31-year-old male with Marfan syndrome was hospitalized for gross haematuria and abdominal pain. He was diagnosed with Marfan syndrome at 26 years of age because of his family history (paternal inheritance) and his manifestations. Contrast-enhanced computed tomography of the abdomen revealed an aneurysm of the abdominal aorta (Fig. 1a) and of the hepatic artery (Fig. 1b), as well as multiple hepatic and bilateral renal cysts with increased kidney size (Fig. 1b), suggesting a diagnosis of autosomal dominant polycystic kidney disease (ADPKD). A positive family history was not elicited; however, his mother died at 38 years of age due to a ruptured dissecting aneurysm of the thoracic aorta, a feature rarely reported in ADPKD [1, 2]. To clarify the diagnosis of ADPKD, a genetic analysis was performed, and revealed a nonsense mutation in exon 4 of the *PKD2* gene (c.916C>Tp.R306X).

The association between ADPKD and Marfan syndrome is rare, and is reported in few cases in the literature [3]. Although renal cysts are the cardinal manifestations of ADPKD, it presents multiple extra-renal manifestations, encompassing both cystic involvement of the organs and

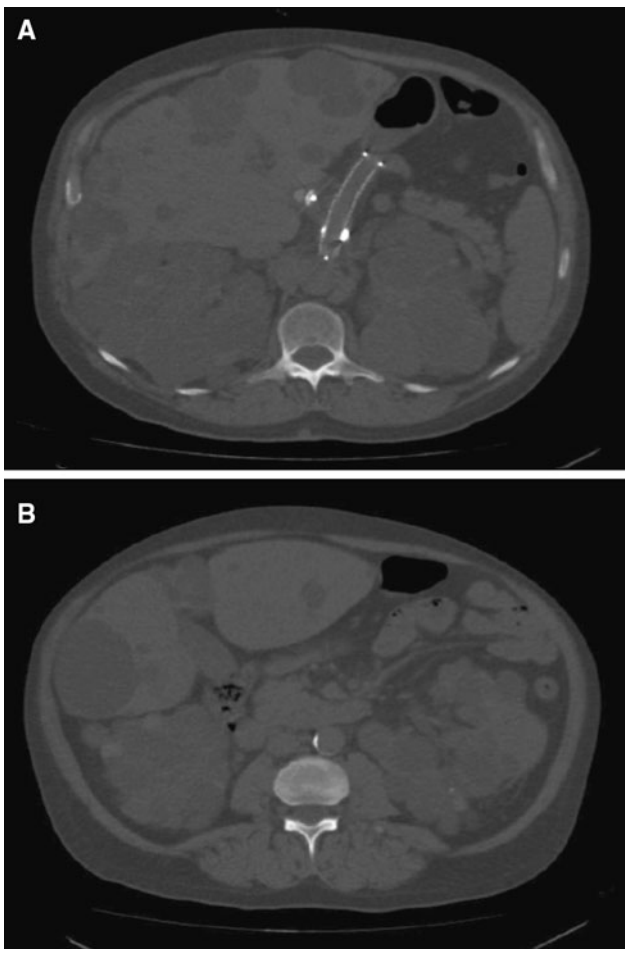

Fig. 1 **a** Contrast-enhanced computed tomography scan of the abdomen, showing an aneurysm of the abdominal aorta; **b** contrast-enhanced computed tomography scan of the abdomen, showing an aneurysm of the hepatic artery, and multiple hepatic and bilateral renal cysts with increased kidney size

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Published online: 10 October 2013

connective tissue abnormalities, such as artery aneurysms [4]. These associations have led authors to hypothesize that a defect in connective tissue may underlie the pathogenesis of ADPKD [5].

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