Diagnosis of tuberous sclerosis in a patient who presented with polycystic kidney disease

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A 19-year-old mentally retarded man presented with congestive heart failure, renal failure (serum creatinine 12.8 mg/dl), anaemia (haematocrit 25%), and severe hypertension. His mother initially denied any prior medical history. A CT scan demonstrated (Figure 1A) very massive kidneys (19-cm in length, large arrows) with huge (4–5 cm, small arrows) cysts, that were considered diagnostic of autosomal dominant polycystic kidney disease (PCKD). However, the age of renal failure was inconsistent with autosomal dominant PCKD and there was no family history of renal insufficiency. Although he might have had autosomal recessive disease, further examination revealed a skin lesion consistent with adenoma sebaceum (Figure 1B). His mother subsequently revealed a history of seizure at age 5 that she had forgotten. A CT scan of the brain showed multiple subependymal tubers (Figure 1C) which confirmed the diagnosis of tuberous sclerosis.

Similar to PCKD, tuberous sclerosis can present solely as chronic renal failure with large cystic kidneys. While angiomyolipomas are typically described, the kidneys can, as in this case, have an identical appearance to those with PCKD. Patients with tuberous sclerosis present younger than those with PCKD, and often as mentally retarded patients with epilepsy and adenoma sebaceum. Von Hippel Lindau (VHL) disease is also included in the differential diagnosis of polycystic kidneys, with a similar age of onset of cysts to PCKD but hypertension and deterioration of renal function are rare in VHL [1].

References
Fig. 1. (A) A CT scan showing very massive kidneys (see large arrows) with huge cysts (see small arrows). (B) Photograph showing skin lesion consistent with adenoma sebaceum. (C) A CT scan of the brain showing multiple subependymal tubers (see arrows).