TSC2/PKD1 contiguous gene deletion syndrome

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A 16-year-old White female patient presented with newly diagnosed hypertension and intermittent bilateral flank discomfort. Her family history was notable for autosomal dominant polycystic kidney disease in her mother, who progressed to kidney failure at the age of 38 years. On physical examination, the patient's blood pressure was 150/95 mm Hg, and multiple reddish facial papules, consistent with angiofibromas, were noted. Laboratory studies showed normal kidney function and C-reactive protein level, with no evidence of hematuria.

Abdominal magnetic resonance imaging revealed bilaterally enlarged multicystic kidneys and 2 parenchymal masses (Figure 1): a 66 × 61 × 52-mm right interpolar lesion and a 47 × 47 × 46-mm upper pole lesion in the left kidney. Both demonstrated mixed signal intensities on T2-weighted images, consistent with fat, fluid, and soft tissue components, and suggestive of angiomyolipomas. Brain magnetic resonance imaging identified multiple cortical tubers (Supplementary Figure S1).

Genetic testing in the patient and her mother revealed a 160-kb heterozygous interstitial deletion at 16p13.3, encompassing the entire *TSC2* gene and exons 3–46 of *PKD1*, confirming *TSC2/PKD1* contiguous gene deletion syndrome. This rare disorder combines features of tuberous sclerosis complex and autosomal dominant polycystic kidney disease. Tuberous sclerosis complex may manifest with cutaneous signs (facial angiofibromas, hypomelanotic macules, and shagreen patches), neurologic involvement (cortical tubers, subependymal nodules or giant cell astrocytomas, epilepsy, developmental delay, and autism spectrum disorder), and renal abnormalities (angiomyolipomas and multiple cysts).

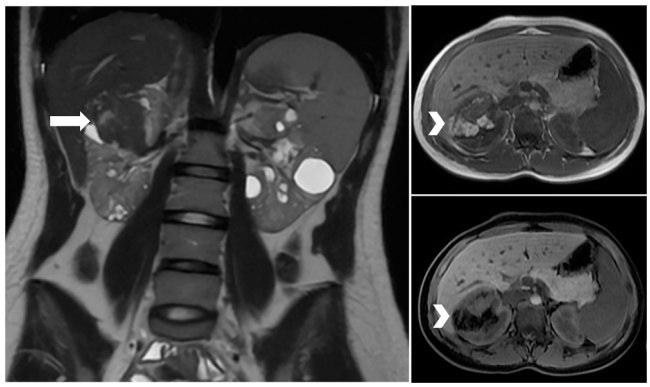

Figure 1 | Abdominal magnetic resonance imaging: coronal T2-weighted image reveals multiple simple cystic lesions in both kidneys. A mixed-signal right upper pole mass is also identified (arrow). Transverse T1 and fat-suppressed T1 images reveal fat content in the right upper pole lesion, appearing in bright signal on T1 and disappearing on fat suppression (arrowheads).

TSC2/PKD1 contiguous gene deletion syndrome is associated with early-onset kidney failure and necessitates multidisciplinary management. In this patient, selective arterial embolization of the 2 angiomyolipomas was proposed. In addition, treatment with mammalian target of rapamycin inhibitors was being considered because of its potential to reduce angiomyolipoma size and address other manifestations of tuberous sclerosis complex.

DISCLOSURE

All the authors declared no competing interests.

Supplementary material is available online at www.kidney-international.org.