Intracranial Aneurysm and Recessive Polycystic Kidney Disease

The Third Reported Case

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Objective: To highlight the possible association of intracranial aneurysm with autosomal recessive polycystic kidney disease.

Design, Setting, and Patient: To our knowledge, this association has been reported only twice in the medical literature. We herein report the case of a 21-year-old man with autosomal recessive polycystic kidney disease, presenting with subarachnoid hemorrhage secondary to a ruptured intracranial aneurysm, at our institution.

Results: In the presence of only 3 cases in the medical literature, one might conclude they are a simple coincidence. However, should this association exist, such as with the dominant form, then the neurologic prognosis and even the life of young patients may be at stake.

Conclusions: Given the devastating consequences of intracranial bleeding in young patients, early neurologic screening may be warranted.


Intracranial aneurysms occur in patients with autosomal dominant (AD) polycystic kidney disease (PKD) in 5% to 10% of cases. The association of intracranial aneurysm with autosomal recessive (AR) PKD is extremely rare. To our knowledge, it has been reported only twice in the medical literature. We herein report the case of a 21-year-old man with ARPKD, presenting with subarachnoid hemorrhage secondary to a ruptured intracranial aneurysm.

REPORT OF A CASE

A 21-year-old man was admitted to the emergency department for severe sudden occipital headache. In his childhood, he had multiple episodes of pulmonary infections. At the age of 6 years, fiberoptic gastroscopy for recurrent hematemesis showed esophageal varices that were managed with sclerotherapy. The diagnosis of PKD was suspected when he was 10 years of age, while he was being investigated for hepatosplenomegaly with impaired renal function. Renal ultrasonography revealed the polycystic kidneys, and liver biopsy showed hepatic fibrosis, with obstruction of the small bile ducts. Thus, the diagnosis of ARPKD was confirmed. Abdominal ultrasonography performed in both parents were normal; however, they were first-degree cousins and had lost 2 of their children. The autopsy of 1 of these children, aged 3 months, showed hepatic fibrosis and polycystic kidneys.

The child was then lost to follow-up until recently, when he was brought to the emergency department following the sudden onset of intense occipital headache with photophobia, nausea, and vomiting. Physical examination revealed a 55-kg young man with a relative failure to thrive, measuring 155 cm. Results from neurologic examination were normal, with a Glasgow coma score of 15. The abnormal results from laboratory examinations included hemoglobin level of 8.3 g/dL (to convert to grams per liter, multiply by 10.0), platelet count of 50,000/mm³, blood urea nitrogen level of 122.12 mg/dL (to convert to millimoles per liter, multiply by 0.357), creatinine level of 571 mmol/L, sodium bicarbonate level of 7 mEq/L (to convert to millimoles per liter, multiply by 1.0), and prothrombin time at 36%. The results of the rest of the laboratory examinations were all within the normal limits. A noninjected brain scan showed a right parenchymal parietal hematoma with surrounding edema (Figure, A). For tech-
Technical reasons, the scheduled angiographic magnetic resonance imaging was replaced by an injected 64-slice angiographic scan that revealed a 5.7-mm aneurysm of the right anterior choroid artery (Figure, B). Two days following the injected scan, the patient developed acute renal failure (blood urea nitrogen, 124.1 mg/dL; creatinine, 598 μmol/L) with oligo-anuria and electrolyte disturbances requiring hemodialysis. Surgical clipping of the aneurysm was performed under general anesthesia on day 3. The patient was extubated at the end of the procedure, without any neurologic sequelae. The rest of the hospital stay was uneventful. An arteriovenous access for hemodialysis was surgically created, and the patient was discharged from the hospital on day 12 with scheduled follow-up in nephrology.

**COMMENT**

There are 2 genetically and clinically different forms of PKD. Autosomal dominant PKD is quite common among the general population (0.1% to 0.25%), but it is often asymptomatic until adulthood.1,2 The second very rare form is ARPKD, typically diagnosed in childhood, with evolution into end-stage renal failure during the first decade of life in 50% of patients.3,4 The liver usually shows signs of sclerosing cholangitis, with portal vein hypertension, esophageal varices, and evolution toward end-stage cirrhosis. Other features of ARPKD include pulmonary infections, feeding disorders, Potter facies, oligohydramnios, low implanted ears, micrognathia, and growth disorders.3

The association between intracranial aneurysms and the dominant form of PKD occurs in 5% to 10% of cases.1,4 This association is exceedingly rare in cases of recessive PKD; it has been reported only twice in the medical literature.3,5 Reported in 1999, the first patient was a 31-year-old woman diagnosed as having ARPKD at the age of 14 years.5 Her renal function deteriorated progressively until the age of 29 years, when she received a kidney transplant. During a routine follow-up for recurrent headaches at the age of 31 years, 4 intracranial aneurysms were discovered on neuroimaging and were successfully clipped. Reported in 2001, the second patient was a 2.5-year-old girl without any family history of PKD.3 She was diagnosed as having ARPKD and moderate renal failure while being investigated for pulmonary infection. Her liver was enlarged with cystic dilatation of the biliary ducts. Magnetic resonance angiography of the brain performed for recurrent vomiting episodes showed multiple saccular and fusiform intracranial aneurysms. This patient was followed up clinically until the age of 4 years. She was reported to be growing well, with no neurologic symptoms.4 The patient we herein describe is the third reported occurrence of ARPKD and intracranial aneurysm. However, he differs from both previously reported patients. This is the first male patient and the only case with a ruptured aneurysm and intracranial hemorrhage. Additionally, this patient had a single aneurysm, as opposed to the other cases with multiple intracranial aneurysms.

In the presence of only 3 cases in the medical literature, one might conclude that the coexistence of intracranial aneurysms and ARPKD in these cases were a simple coincidence. However, should this association exist, such as with the dominant form, then the neurologic prognosis and even the life of these young patients may be at stake. In the presence of the dominant form of PKD and given the devastating consequences of aneurysmal rupture, systematic screening of the brain is currently recommended when patients reach the age of 30 years and must be repeated every 5 to 10 years.1 These recommendations are mainly based on the risk for intracranial bleeding. Although the overall risk for rupture of an associated aneurysm is similar to that of the general population (0.5%-2%), this rupture occurs 10 years earlier (median age, 41 years) in cases of ADPKD.1 Rupture occurs even before age 21 years in 10% of cases. Occasionally, rupture is an inaugural and catastrophic event in ADPKD.1 In addition, multiple aneurysms occur in 24% to 31% of patients with ADPKD compared with only 16% in the general population.1 The question is whether these recommendations can be applied to patients with the recessive form in the absence of formal studies on large numbers of patients. The potentially life-threatening complications of intracranial bleeding in very young patients argue strongly in favor of the systematic early detection of cerebral an-

![Figure. Images of patient's brain. A, Brain computed tomographic scan showing the intracranial bleeding (arrow). B, Angiographic study showing the 5.7-mm aneurysm (arrow).](https://jamanetwork.com/asset//jamanetwork.com/asset/)
eurysms. The scarcity of reported cases may be owing to the low incidence of ARPKD and the short life expectancy of these patients. Early systematic neurologic screening of patients with ARPKD may increase the number of cases diagnosed as having intracranial aneurysms and provide new material for clinical and genetic studies.

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