Intracranial aneurysm in autosomal dominant polycystic kidney disease

Intracranial aneurysm are at least twice as common in patients with autosomal dominant polycystic kidney disease (ADPKD) than in the general population.

In ADPKD, 10% to 15% of patients develop intracranial aneurysms.

Screening

Autosomal dominant polycystic kidney disease (ADPKD) is a risk factor for formation of intracranial aneurysms (IAs), though the ideal screening and treatment strategies in this population are unclear.

Systematic screening of intracranial aneurysm (ICAN) with Three dimensional magnetic resonance imaging based on Time-of-flight magnetic resonance angiography is recommended for patients with ADPKD, particularly for adult patients (≧30 years) or patients with a positive family history of hemorrhagic stroke or ICAN [1)].

Treatment

With appropriate management, coil embolization may be safe and effective for intracranial aneurysms in ADPKD. There is a concern about contrast induced nephropathy in patients with CKD stage 5 or high serum Cr level (>2.0 mg/dL) [2)].

Case series

Wilkinson et al., from the Department of Neurosurgery, University of Michigan, Ann Arbor, performed a retrospective analysis of all patients with autosomal dominant polycystic kidney disease (ADPKD) and IAs at a single center from 2000 to 2016.

Forty-five patients with ADPKD harboring 71 aneurysms were identified, including 11 patients with subarachnoid hemorrhage (SAH). Of 22 aneurysms managed with observation, none ruptured in 136 yr of clinical follow-up. Thirty-five aneurysms were treated with open surgery and 14 with an endovascular approach. Among treated aneurysms, poor neurologic outcome (modified Rankin scale >2) was seen only in patients presenting with SAH (17% SAH vs 0% elective, P = .06). Acute kidney injury (AKI) was also significantly associated with SAH presentation (22% SAH vs 0% elective, P = .05). Neither procedural complications nor AKI were associated with treatment modality. Among 175 yr of radiographic follow-up in patients with known IAs, 8 de novo aneurysms were found, including 3 that were treated. Of 11 patients with SAH, 7 ruptured in the setting of previously known ADPKD, including 2 with prior angiographic screening and 5 without screening.

Poor outcomes occurred only with ruptured presentation but were equivalent between treatment modalities. Screening is performed only selectively, and 64% (7 of 11) of patients presenting with SAH had previously known ADPKD [3)].

Flahault et al., from Centre Hospitalier Sainte-Anne and Hôpital Necker-Enfants Malades, Paris, France, evaluated in a single-center cohort of 495 consecutive patients with ADPKD submitted to targeted intracranial aneurysm screening. Cerebral magnetic resonance angiography was proposed to 110 patients with a familial history of intracranial aneurysm (group 1), whereas it was not our intention to propose it to 385 patients without familial risk (group 2). Magnetic resonance angiography results, intracranial aneurysm prophylactic repair, rupture events, and cost-effectiveness of intracranial aneurysm screening strategies were retrospectively analyzed. During a median follow up of 5.9 years, five non-fatal intracranial aneurysm ruptures occurred (incidence rate 2.0 (0.87-4.6)/1000 patients-year). In group 1, 90% of patients were screened and an intracranial aneurysm was detected in 14, treated preventively in five, and ruptured in one patient despite surveillance. In group 2, 21% of patients were screened and an intracranial aneurysm was detected in five, and treated preventively in one. Intracranial aneurysm rupture occurred in four patients in group 2. Systematic screening was deemed cost-effective and provides a gain of 0.68 quality-adjusted life years compared to targeted screening. Thus, the intracranial aneurysm rupture rate is high in ADPKD despite targeted screening, and involves mostly patients without familial risk factors. Hence, cost-utility analysis suggests that intracranial aneurysm screening could be proposed to all ADPKD patients [4].

Chauveau et al., from the Hôpital Necker, Paris, France, conducted a retrospective study on 77 ADPKD patients from 64 families presenting with ruptured (N = 71) or unruptured (N = 6) aneurysm. Information was collected on kidney disease, intracranial aneurysm and family history. Linkage to PKD1 locus was examined by five probes to obtain informative flanking markers. Within one year prior to rupture, blood pressure was normal in 29% of the patients. At the time of rupture, mean age was 39.5 years (range 15 to 69), renal function was normal in half of the patients and 11% were on renal replacement therapy. The ruptured aneurysm was usually located on the middle cerebral artery. Additional intact aneurysms (1 to 6) were detected in 31% of the patients. Surgical or endovascular treatment was performed in 54 (76%) patients whereas 17 (24%) had medical management only. Rupture of ICA was fatal in seven (10%) patients. On long-term follow-up 27 (38%) were left with severe disablement. Five patients bled from another aneurysm 2 days to 14 years after initial rupture. Only two of six patients with unruptured aneurysm alone were treated on a prophylactic basis. No clinical marker associated with aneurysm was found. A family history of aneurysm rupture was demonstrated in 10 (18%) kindreds. Linkage to the PKD1 locus was established in two of three tested families [5].

1)
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