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Case Report

Dissecting Aneurysm in A Patient with Autosomal Dominant Polycystic Kidney Disease

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Autosomal dominant polycystic kidney disease (ADPKD) is primarily associated with renal failure, but it also causes systemic diseases, including cysts of other systemic organs and cerebral or visceral aneurysm. To make matters worse, life-threatening aortic diseases are associated with ADPKD in some cases. However, only a few reports of ADPKD-associated with thoracic aortic dissection have been published. Herein, we present a case of dissecting aneurysm in a patient with hypertension and ADPKD. He had been followed up for type B aortic dissection for six years. Preoperative creatinine level ranged from 2.1 to 2.4 mg/dl. We performed replacement of the thoracic aorta with prosthetic graft successfully, and postoperatively, dialysis was not required.

It is very important for us to recognize the relationship between ADPKD and thoracic aortic dissection, which can cause high mortality and morbidity rates.

Keywords: dissecting aneurysm, autosomal dominant polycystic kidney disease, life-threatening disease

Introduction

Autosomal dominant polycystic kidney disease (ADPKD) is a systemic disease that causes not only renal dysfunction, but also cysts of other systemic organs and cerebral or visceral saccular aneurysm.[1)] Additionally, vascular abnormalities develop in patients with ADPKD. Thoracic aortic dissection is a rare vascular abnormality, and a few cases of ADPKD-associated with thoracic aortic dissection have been reported previously. The scarcity of reports may also suggest that aortic dissection in patients with ADPKD is not always recognized by physicians. Although many articles on ADPKD address extrarenal manifestations of the disease, including visceral organ cysts or cerebral aneurysms, there is little information on ADPKD-associated aortic dissection, a life-threatening condition and the main determinant of mortality.

Case Report

A 44-year-old man was admitted to our hospital due to an operation for a thoracic dissecting aneurysm. Acute type B aortic dissection extending descending aorta distal to the left subclavian artery to iliac arteries bilaterally with the patent false lumen occurred 6 years previously, and subsequently watchful waiting had been done by cardiologists. His past history included hypertension and ADPKD. Hypertension was well-controlled with some oral drugs. The creatinine level ranged from 2.1 to 2.4 mg/dl preoperatively. As to his family history, sudden death by unknown cause happened to his father. His mother with hypertension on dialysis due to ADPKD died of subarachnoid hemorrhage. One younger sister had ADPKD without dialysis.

Physical examination was unremarkable, in particular the findings indicating Marfan syndrome were not recognized.

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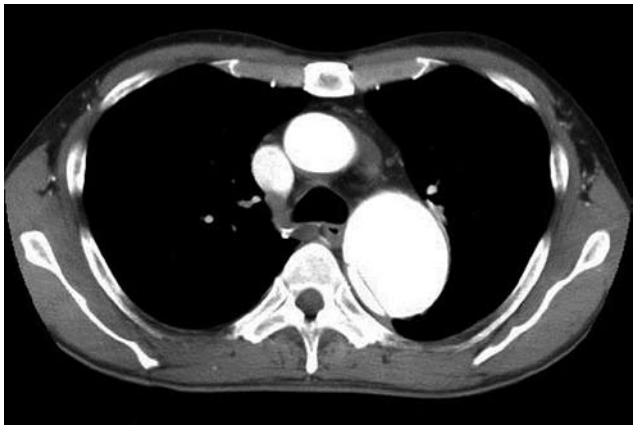

Fig. 1 Enhanced computed tomography shows thoracic dissecting aneurysm with the patent false lumen.

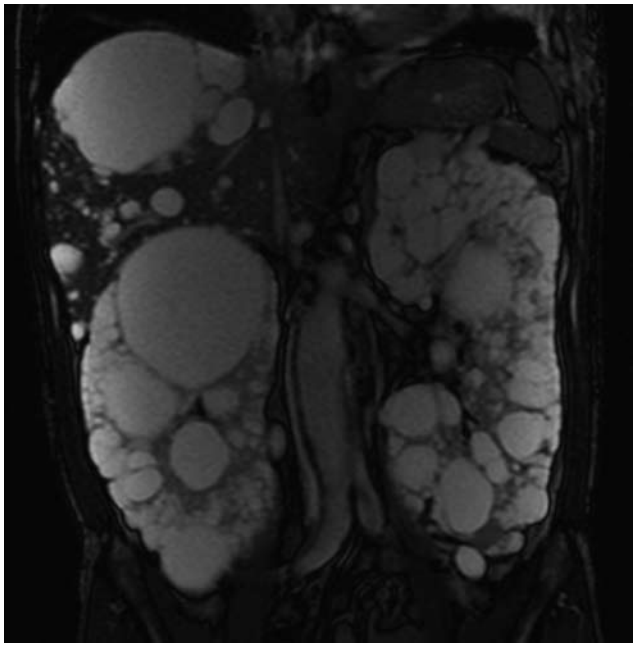

Fig. 2 Magnetic resonance imaging shows bilateral polycystic kidney and multiple cysts in the liver.

Preoperative computed tomography showed thoracic dissecting aneurysm in a maximum diameter of 6 cm with the patent false lumen (**Fig. 1**). Magnetic resonance imaging revealed bilateral polycystic kidneys larger than normal ones by 2 times and multiple cysts in the liver (**Fig. 2**).

Replacement of the thoracic aorta with prosthetic graft was performed under deep hypothermic circulatory arrest via left thoracotomy approach.

Although postoperative wound infection occurred, he was discharged in a good condition after healing of wound infection.

Pathological finding of the aortic wall revealed the dissection of media alone. No findings such as aortitis or cystic medial necrosis were recognized.

Discussion

ADPKD is the most common of the inherited renal cystic diseases, and it is characterized by the development of renal cysts and various extrarenal manifestations. ADPKD occurs worldwide in all races and in 1 out of 400–1000 individuals.[2] Age-adjusted ratios greater than unity (1.2–1.3) indicate that the disease is more progressive in men than in women.[2] In more than 50% of patients with ADPKD, renal function declines progressively, end-stage renal failure develops, and patients require the renal replacement therapy.[3]

ADPKD is a systemic disease resulting from mutations in either the polycystic kidney disease-1 (PKD-1) or PKD-2 genes. PKD-1 and PKD-2 mutations account for 85%–95% and 5%–15% of ADPKD cases, respectively.[3]

PKD-1 encodes the polycystin-1 protein, and PKD-2 encodes polycystin-2. These proteins function independently or as a complex to regulate transcription, apotosis, differentiation, and cell-matrix interactions in fetal and adult epithelial cells. Disruption of the proteins leads to malfunction of these processes and, therefore, malfunctions of epithelial cells. The extrarenal manifestations of ADPKD, including cysts of the liver, pancreas, spleen and other organs, cerebral or visceral saccular aneurysm and diverticulosis of the colon[1] reflect the generalized abnormalities in collagen and extracellular matrix induced by the PKD-1 and PKD-2 mutations. In particular, polycystin-1 plays an important role in maintaining the structural integrity of blood vessels.[3]

Extrarenal cardiovascular manifestations

As mentioned above, more than 50% of patients with ADPKD will eventually require renal replacement therapy. However, with the availability of the renal replacement therapy, cardiovascular complications, including aneurysm and cardiac valvular diseases have become the main causes of death in patients undergoing renal replacement therapy.[4] Hypertension is a common, early complication that develops in 50%–70% of patients with ADPKD with a rapid progression to the end-stage renal failure and an increasing cardiovascular complications.[4,5]

1. Cardiac manifestations

Cardiac valvular diseases are well-established extrarenal

complications in patients with ADPKD. Leier et al. have reported that 18% of patients with ADPKD and 27% of autopsy cases had one or more cardiovascular diseases.[1)]

Mitral valve prolapse is the most common cardiac manifestation, and it is detected in up to 26% in patients with PKD-1 based on echocardiographic analyses. The prevalence of hemodynamically significant mitral regurgitation (grade 2 to 3) was 13%. With regard to the prevalence of valvular diseases, another study showed that 26% of patients with ADPKD had mitral valve prolapse, 31% had mitral regurgitation, 8% had aortic regurgitation, 15% had tricuspid regurgitation and 6% had tricuspid valve prolapsed.[6)]

2. Vascular manifestations

Vascular manifestations of ADPKD include intracranial aneurysms and dolichoectasias, thoracic aortic and cervicocephalic artery dissections and coronary artery aneurysms.[2)] These conditions are caused by alterations in the vasculature directly linked to mutations in PKD-1 or PKD-2.[7)]

ADPKD-associated vascular manifestations, such as aortic root dilatation, coarctation of aorta, abdominal aortic aneurysm are also associated with the mutation in PKD-1.[3,7,8)]

Among the vascular manifestations of ADPKD, relatively few cases of thoracic aortic dissection have been published in English literature,[8–17)] when compared with reports of intracranial aneurysms, which are well-established extrarenal manifestations of ADPKD and occur in approximately 6% of patients without a family history of aneurysms and in 16% of those with such a family history.[18)]

Marfan syndrome is a well-known systemic connective tissue disorder that leads to life-threatening cardiovascular complications. Although ADPKD also results in systemic abnormalities in collagen and the extracellular matrix, ADPKD is not recognized as a risk factor for life-threatening cardiovascular outcomes, including thoracic aortic dissection. Interestingly, there is a report on familial clustering of aortic dissection in ADPKD.[17)]

A case of sudden death caused by a dissecting thoracic aortic aneurysm was reported to alert physicians to the presence and possibility of an aortic dissection in patients with ADPKD and risk of sudden cardiac arrest.[11)]

Some authors have warned the medical community of the presence of aortic dissection in patients with ADPKD and concomitant hypertension.[10,11,13)] Through these reports, physicians must pay attention to the possibility of devastating entity behind ADPKD.

Early and effective treatment of hypertension is important in patients with ADPKD to slow the progression of renal failure and prevent the cardiovascular complications that lead to high mortality and morbidity.[2)] In patients with ADPKD and concomitant hypertension, aortic dissection is thought to develop because of hypertension and the reduced structural integrity of vessels caused by mutations in PKD-1.[3,7,8)]

However, once thoracic aortic dissection occurs, conventional surgical or medical treatments are required, and these treatments should be based on the classification of aortic dissection.

Disclosure Statement

Fukunaga N and other co-authors have no conflict of interest.

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