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Case Report

Aortic Arch Aneurysm in Polycystic Kidney Disease Patients: An Unusual combination

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Abstract

Autosomal dominant polycystic kidney disease (ADPKD) is a systemic disease characterized by the formation of multiple cysts in several organs. Combination of ADPKD, multiple aortic aneurysms, which has rarely been reported. Here we present a case of a middle-aged gentleman who was diagnosed to have recurrence of multiple aortic aneurysms with polycystic kidney disease as a risk factor.

Keywords: polycystic kidney disease; aortic aneurysms; vascular disease

Introduction

Aortic aneurysm is a rare vascular disease. Most aortic aneurysm are degenerative in origin and atherosclerosis and hypertension, aortitis secondary to infections or inflammatory disorders are other aetiology of Aortic aneurysm. Autosomal dominant polycystic kidney disease (ADPKD) is a systemic disease characterized by the formation of multiple cysts in several organs. The formation of aneurysms accompanying this disease is being increasingly reported in the literature, and mutations in PKD-1 and PKD-2 are suspected in this etiology. Although the association between ADPKD and multiple coronary arteries aneurysms (CAA) was reported several times, we are presenting a case with the combination of ADPKD, multiple aortic aneurysms, which has rarely been reported. Here we present a case of a middle-aged gentleman who was diagnosed to have recurrence of multiple aortic aneurysms with polycystic kidney disease as a risk factor.

Case Presentation

In 2018, 67-year-old gentleman, was evaluated elsewhere for hoarseness of voice and he was incidentally diagnosed to have aortic arch aneurysm. He was a known hypertensive for last 3 years and a chronic smoker. He was a known case of CKD and his ultrasound revealed bilateral enlarged kidneys with multiple cysts of varying sizes, suggestive of polycystic kidney disease. (Figure 1)

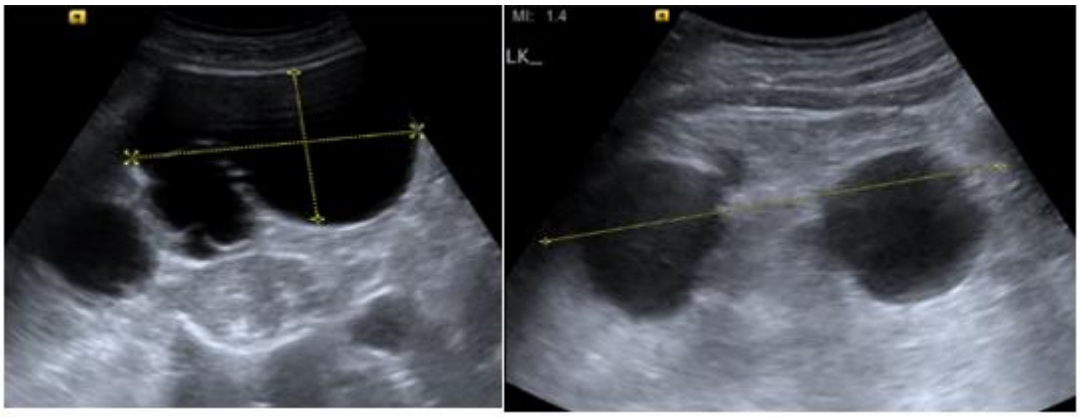

Figure 1: Ultrasound Showing bilateral enlarged kidneys with multiple cysts of varying sizes

When evaluated in our hospital, he was found to have left vocal cord palsy and CT neck and chest and CT aortogram confirmed an aortic arch aneurysm and left renal artery stenosis. He underwent CAG and PAG on 27/10/18 which showed minor coronary artery disease and saccular aneurysm of the thoracic aorta (Figure 2).

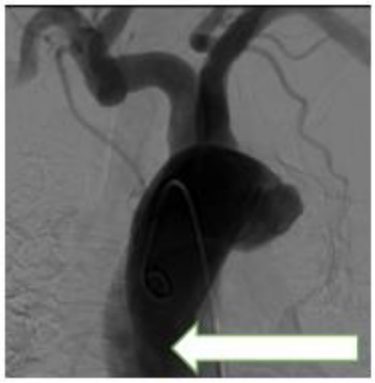

Figure 2: PAG showing saccular aneurysm of the thoracic aorta

He underwent endovascular repair of distal aortic arch saccular aneurysm using a fenestrated endograft done in 2018.

Details of the Procedure:

1.Endograft preparation was done

Three fenestrations were made using circular thermal cautery instruments as follows:

IA: 9mm in diameter, located between stent segments 2 and 3, mostly behind the 12 o'clock line.

LCCA: 8mm in diameter, located between stent segments 3 and 4, mostly anterior to the 12 o'clock line, and with its centre 14mm away from the IA fenestration centre.

LSA: 8mm in diameter, located between stent segments 4 and 5, mostly anterior to the 12 o'clock line, and with its centre 15mm away from the LCCA fenestration centre.

The procedure was performed under general anesthesia with endotracheal intubation.

Vascular access sites and sheaths:

LFA: 7F short sheath. Changed to 18F after endograft deployment.

RFA: 7F sheath.

RFV: 6F short sheath changed later to 8F 63cm long curved sheath.

LCCA: 3F Balton sheath with valve system.

LBA: 4F,11 cm Cordi's sheath

RBA: 4F,11 cm Cordi's sheath

2. Setting up the platform before endograft deployment: A Lunderquist wire was parked in the ascending aorta on a 6F JR catheter and a VOTT catheter was taken over it to perform angiogram. A Terumo Glide wire was passed through the left common cartoid artery sheath into the ascending aorta. This was snared and exteriorised from the right common femoral artery sheath using a 7F EV3 Ensnare. This artery-to-artery loop was used to delimit the posterior boundary of the LCCA artery to guide endograft deployment.

3. Fenestrated Endograft deployment and stenting of arch arteries:

A Medtronic Valiant Captiva (VAMF 3838C200TE mm) endograft with fenestrations made for the three arch branches was introduced from the LFA over the 0.035" Lunderquist extra-stiff guidewire. The very proximal portion of the endograft was deployed and pacing rate was initiated at 180/minutes to achieve a pulse pressure < 10 mm of Hg and systolic pressure at approximately 60 mm of Hg. The Freeflo zone of the graft was released after the intial two stent struts were exposed. The graft was then fully deployed, and pacing was discontinued.

Post procedure patient was kept in Intensive care unit for one day and later shifted to ward and subsequently discharged without an eventful stay.

He was under regular OPD follow up. He was doing well till 2023 when he had new onset hematemesis and diagnosed to have CLD ,probably alcohol related and underwent banding and plain his CT angiogram revealed retrograde filling of the aneurysmal sac from the distal end of the attachment of the stent (type 1B endoleak) and partial eccentric thrombus of the fusiform aneurysm of the distal part of the descending thoracic / proximal abdominal aorta at the level of the diaphragmatic hiatus, measuring 7 x 4.5cm.(Figure 3)

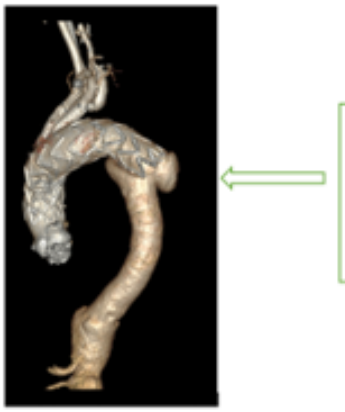

Fig 3: CT angiogram showing fusiform aneurysm of the distal part of the descending thoracic/proximal abdominal aorta at the level of the diaphragmatic hiatus.

There was another eccentric partial thrombus of the fusiform aneurysm of the abdominal aorta below the origin of the bilateral renal arteries extending to just above the bifurcation into the bilateral common iliac arteries, the largest dimension measuring 5.1 x 4.3cm (Figure 4)

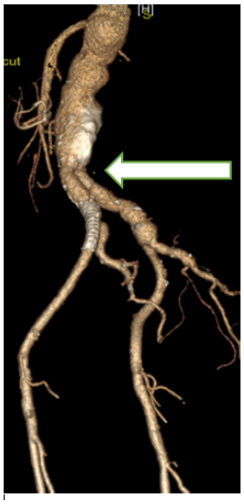

Fig 4: CT angiogram showing fusiform aneurysm of the abdominal aorta below the origin of the bilateral renal arteries extending to just above the bifurcation into the bilateral common iliac arteries.

Differentials considered were: Mycotic aneurysms, non-infective/ inflammatory etiologies. To rule out infective causes with blood cultures and syphilis screen ELISA was sent and turned out to be negative. The more likely differential in this case had seemed to be non-infective/ inflammatory etiologies. He also underwent PET - CT which did not show any extra abnormalities. His lab investigations are shown in table 1.

Discussion

Autosomal dominant polycystic kidney disease (ADPKD) is a common disorder, occurring in approximately 1 in 1000 live births.(1) Approximately 78 percent of families with ADPKD have an abnormality on chromosome 16 (*PKD1* locus) . Most of the remaining families (14 percent) have a different defect that involves a gene on chromosome 4 (the *PKD2* locus), while a minority of families have a defect in the *GANAB* gene, encoding the glucosidase II alpha subunit, the *ALG9* gene or the *DNAJB11* gene[2]. Relentless cyst growth substantially enlarges both kidneys and culminates in renal failure. Patients with ADPKD also have vascular abnormalities; intracranial aneurysms (IAs) are found in ∼10% of asymptomatic patients during screening and in up to 25% of those with a family history of IA or subarachnoid haemorrhage. As the genes responsible for ADPKD—PKD1 and PKD2—have complex integrative roles in mechanotransduction and intracellular calcium signalling, the molecular basis of IA formation might involve focal haemodynamic conditions exacerbated by hypertension and altered flow sensing. Other vascular aneurysms and anomalies—including aneurysms of the aorta and coronary arteries, cervicocephalic and thoracic aortic dissections, aortic root dilatation and cerebral dolichoectasia are less common and screening is not usually indicated.[3] The mechanism by which specific PKD mutations predispose to a vascular phenotype remains unclear. Although certain mutations might increase the risk of developing an aneurysm, not all individuals from high-risk families will experience vascular complications. Unfortunately, very few systematic studies of the incidence and prevalence of vascular abnormalities other than IA in patients with ADPKD exist and validated recommendations for screening and follow up cannot be made. PKD1 or PKD2 haploinsufficiency, alterations in intracellular calcium signalling, modifier genes and alterations in TGF-β signalling could contribute to aneurysm formation. Upregulation of TGF-β signalling in the setting of Pkd1 haploinsufficiency suggests a potential mechanism that will require further exploration.[3]

The major extrarenal complications of ADPKD are

The major extrarenal complications of ADPKD are:

- Cerebral aneurysms
- Hepatic and pancreatic cysts
- Cardiac valve disease
- Colonic diverticula
- Abdominal wall and inguinal hernia
- Seminal vesicle cysts

Valvular abnormalities of unclear clinical significance can be detected by echocardiography in 25 to 30 percent of patients with ADPKD [4] An association between abdominal aortic aneurysms and ADPKD has been proposed. However, a study that compared 139 patients with ADPKD and 149 controls was unable to demonstrate by ultrasonography an increase in either aortic diameter or the incidence of aneurysm formation in the patients with ADPKD and they concluded that although aortic aneurysms do not appear to be an intrinsic feature of ADPKD, there may be some increase in risk in patients with uncontrolled hypertension.[5]

Vascular manifestations of ADPKD like intracranial aneurysms and dolichoectasias, thoracic aortic and cervicocephalic artery dissections and coronary artery aneurysms are caused by alterations in the vasculature directly linked to mutations in PKD-1 or PKD-2[6] ADPKD-associated vascular manifestations, such as aortic root dilatation, coarctation of aorta, abdominal aortic aneurysm are also associated with the mutation in PKD-1[7,8]

In our patient he had recurrence of multiple aortic aneurysms. Other aetiologies like

Aortitis due to infectious or inflammatory etiologies were ruled out. His blood pressure and cholesterol level were under control. Mutation of ADPKD-related genes may predispose to coronary abnormalities, especially aneurysms. Although substantial progress has been made in the diagnosis and treatment of vascular anomalies—particularly IAs—in patients with ADPKD, important questions remain. More research is required to define which patients are at risk of vascular anomalies especially aortic aneurysms and their complications and to better understand the mechanisms of aneurysm formation, including the role of inflammation and the interactions of ADPKD genetic mutations with TGF-β and other signalling pathways.

Conclusion

In conclusion, we report a case of recurrence of aortic aneurysms associated with ADPKD and emphasize the importance of strict control of blood pressure and regular follow-up of the cardiovascular system.

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