



Case Report

Ruptured Sinus of a Valsalva Associated with Autosomal-Dominant Polycystic Kidney Disease in a young male

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ABSTRACT

Autosomal-dominant polycystic kidney disease (ADPKD) is a systemic hereditary disorder characterized by localized cellular proliferation and extracellular matrix abnormalities resulting in both renal and extra renal manifestations, with multiple structural flaws such as renal cysts, cerebral and aortic aneurysms, annulo-aortic ectasia, and valvular insufficiency states. Association between Autosomal-dominant polycystic kidney disease (ADPKD) and ruptured sinus of Valsalva aneurysm (SVA) has rarely been documented before. We herein report a case of Autosomal-dominant polycystic kidney disease (ADPKD) who presented with shortness of breath, was diagnosed as a case of ruptured sinus of Valsalva aneurysm (SVA) and defect was closed successfully.

KEYWORDS:ADPKD, Sinus of Valsava aneurysm, young male.

INTRODUCTION

Aneurysm of the aortic sinus, also known as the sinus of Valsalva, is comparatively rare. But its presence usually found in either the right (65–85%) or in the non-coronary (10–30%) sinus and rarely in the left (< 5%) sinus [1]. This type of aneurysm may be asymptomatic, if unruptured.

An aneurysm of the aortic sinus may rupture due to infective endocarditis involving the aortic wall and tertiary-stage syphilis or in rare conditions as Autosomal-dominant polycystic kidney disease (ADPKD). In ADPKD due to defective extracellular protein "polycystin", structural integrity of the aortic valve cusps is compromised, resulting in sinus of Valsalva aneurysm (SVA).

CASE REPORT

35 years old male, a diagnosed case of ADPKD since 2014 with baseline creatinine of 3 mg presented with dyspnea on

exertion (NYHA II) (Fig 1). On physical examination, the patient's blood pressure was 146/86mm of mercury with a regular pulse of 80 per minute. His respiratory rate was 9 per minute, and was afebrile. The carotid pulse was hyper dynamic. The jugular venous pressure was normal. The lung fields were clear to auscultation.

There was a palpable left parasternal thrill, with a forceful but non displaced apical impulse. The first and second heart sounds were normal. A continuous murmur (grade 4/6) was heard at the left lower sternal border. Abdomen was non tender, bilateral kidneys were palpable.

Transesophageal echocardiogram (TEE) showed right atrial enlargement, moderate tricuspid regurgitation, and normal global left ventricular systolic function. A discrete jet of continuous turbulent flow was seen emanating from the non coronary sinus of Valsalva and terminating in the right atrium(Fig 2).

Figure 1: Ultrasonography showing bilateral enlarged kidneys with multiple cysts

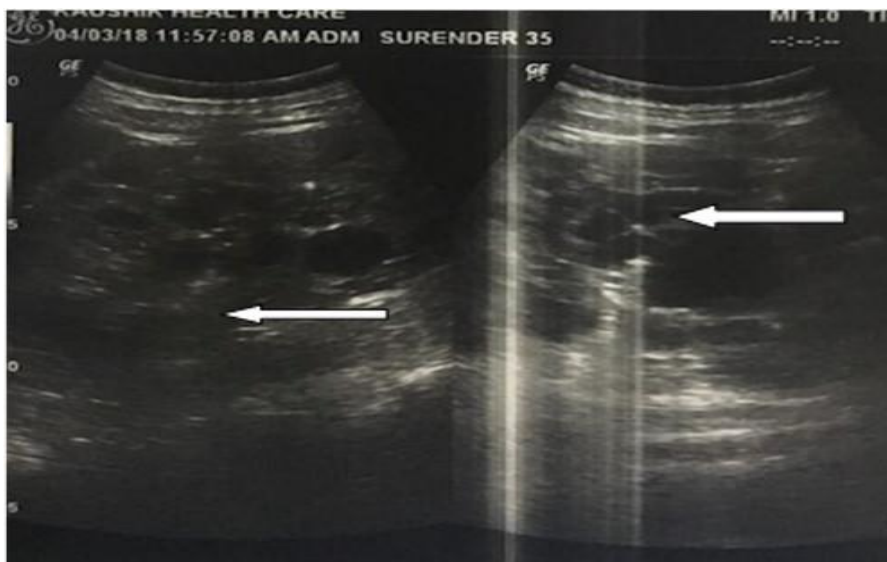


Fig 2. Ruptured sinus of Valsalva aneurysm (SVA) with jet turbulent flow into right atrium (enlarged right atrium) right side of the figure shows colour across ruptured sinus of Valsalva and right atrium

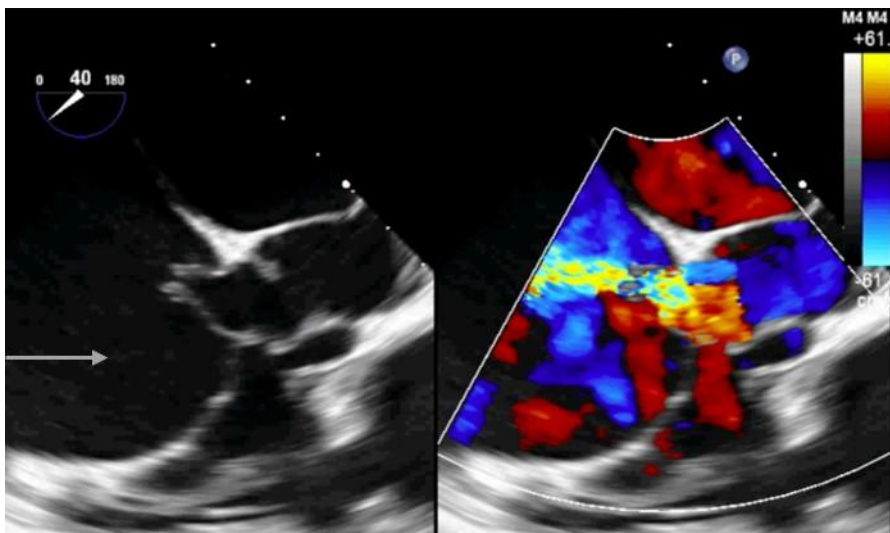
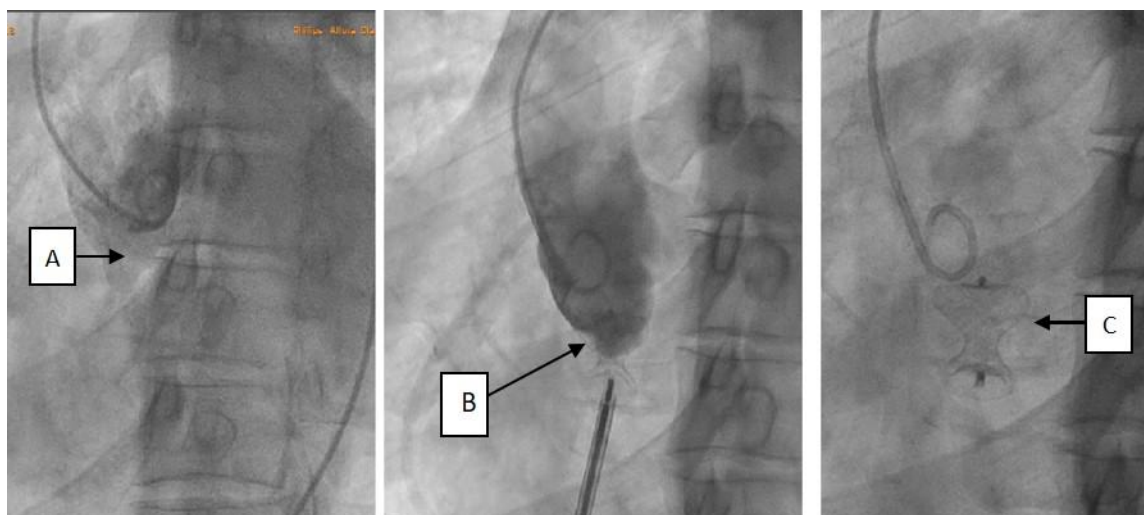


Fig 3. Aortic root angiogram showing leakage of contrast into right atrium. (A) Aortic root angiogram contrast opacification of right atrium, (B) aortic root angiogram showing duct occluder and (C) device in situ



Patient was taken for device closure of RSOV. The procedure was performed under local anaesthesia with transesophageal echocardiography (TEE) guidance. Aortic root angiography was done in the left anterior oblique (LAO) and right anterior oblique (RAO) to better delineate the anatomy. Size of RSOVA was measured at its aortic end both on the TEE and angiography. The Cocoon Duct Occluder of size 18/16 was selected, after confirming that there was no encroachment on coronary arteries and no significant aortic regurgitation (AR) as seen on TEE and angiography the device was released from the delivery cable (Fig 3). Thereafter, aortogram depicts successful deployment of 18/16 Cocoon Duct Occluder with no flow across the defect.

DISCUSSION

ADPKD is the most common cystic disease of the kidney, with an estimated prevalence ranging from 1:400 to 1:1000. ADPKD has been associated with renal and liver cysts; colon diverticula (80%); hernias (25%); cardiac valve (tricuspid, mitral, aortic) regurgitation; mitral and tricuspid valve prolapse; Annulo aortic ectasia; and intracranial, aortic, and coronary arterial aneurysms [2].

The gene defect mapped to chromosome 16p13.3 accounts for 85% of cases. The resulting defective extracellular protein "polycystin" is thought to be causative for cysts and aneurysms in ADPKD. Defective extracellular polycystin is part of lamina lucida of the basal lamina which is responsible for structural integrity of the aortic valve cusps. This results in loss of continuity between aortic valve endothelium and trigonumfibrosum. This abnormal sinus of Valsalva tissue after repeated stress becomes fibrotic and dilates resulting in SVA [3].

Ruptured sinus of Valsalva aneurysms (RSOVAs) are more common in males than females (male: female, 4:1). Aneurysms are more commonly seen in right coronary sinus (RCS) (70%), followed by non-coronary sinus (29%), and rarely involve left coronary sinus (1%) [4]. Aneurysms of RCS may rupture into the right ventricle or right atrium, whereas aneurysms of non-coronary sinus (NCS) rupture into the right atrium. There are some case reports where aneurysms have been reported into the pulmonary artery, left ventricle, left atrium, or pericardial cavity [5].

Rupture usually manifests as a sudden onset of chest pain and acute heart failure in most of the cases. Death usually occurs within 1 year of untreated RSOVA [5]. Aneurysms of sinus of Valsalva are rare, and, hence, the experience

remains limited. Surgery has been the gold standard treatment for RSOVA. However, the morphological features of RSOVA make it amenable to percutaneous closure. First device closure of RSOVA was done in 1994 [6]. Thereafter clinicians have tried variety of occluders to close RSOVA.

Many case reports have been published using patent ductus arteriosus (PDA) occludes, VSD occludes, atrial septal defect (ASD) occludes, Gianturco coils, Rashkind umbrella, etc. However, PDA occluders are currently favored. The most vital step in closure of RSOVA is accurate angiographic identification of cusps. Unless the anatomy is clearly defined on angiography, one should not proceed for device closure, as chances of failure or complications remain high. Residual shunt, development of aortic regurgitation (AR) or worsening of AR, coronary ostial encroachment, infective endocarditis and failure to deploy are some of the important complications associated with device closure in RSOVA. This rare case report reiterates the association of ADPKD with SVA, The risk of rupture and another case of successful device closure of RSOV.

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