

A CASE OF PERIMEMBRANOUS VENTRICULAR SEPTAL DEFECT ASSOCIATED WITH SINUS OF VALSALVA ANEURYSM MIMICKING MEMBRANOUS SEPTAL ANEURYSM

HYUNG RAE KIM, MD¹, SHIN-JAE KIM, MD, PHD¹, KYOUNG HOON LIM, MD¹, JONG MIN KIM, MD¹, JUN HO LEE, MD¹, YONG-GIUN KIM, MD¹, JONG-PIL JUNG, MD², AND SANG-GON LEE, MD, PHD¹
DEPARTMENTS OF ¹INTERNAL MEDICINE, ²CARDIAC SURGERY, ULSAN UNIVERSITY HOSPITAL, UNIVERSITY OF ULSAN COLLEGE OF MEDICINE, ULSAN, KOREA

Sinus of Valsalva aneurysms are rare. Sinus of Valsalva aneurysms are frequently associated with ventricular septal defect (VSD) and aortic regurgitation. They often remain asymptomatic until abruptly presenting with acute chest pain and heart failure secondary to rupture. Here, we describe a case of 20-year-old man who presented with chest pain with a history of VSD. Initial work-up concluded that the patient had VSD associated membranous septal aneurysm. Four years later, the patient presented with symptoms of heart failure. Work-up showed that the ruptured sinus of Valsalva aneurysm was the cause of symptoms. Due to its close proximity to the aortic annulus, sinus of Valsalva aneurysm should be differentiated from membranous septal aneurysm.

KEY WORDS: Sinus of Valsalva aneurysm · Ventricular septal defect · Membranous septal aneurysm · Heart failure.

INTRODUCTION

Sinus of Valsalva aneurysms, rare cardiac anomaly, are most often caused by weakness at the junction of the aortic media and the annulus fibrosus.¹⁾ Ruptured sinus of Valsalva aneurysms (RSVA) are frequently associated with other congenital defects, particularly with ventricular septal defect (VSD) and, aortic regurgitation (AR). The principal VSD associated with a RSVA is supracristal type.²⁻⁴⁾ The supracristal VSD is the predominant type reported in Asian countries, whereas the perimembranous VSD is often seen in Western countries.^{5,6)} Membranous septal aneurysms are bulging of the membranous portion of the interventricular septum below the aortic annulus in the right ventricle (RV). Sinus of Valsalva aneurysms must be distinguished from membranous septal aneurysms. However, sometimes it is difficult to differentiate the two because they occur anatomically adjacent to each other at the level of aortic annulus. Here, we report a case of sinus of Valsalva aneurysm mimicking membranous septal aneurysm associ-

ated with perimembranous VSD, that was ruptured later, causing severe right-sided heart failure.

CASE

A 20-year-old man visited our clinic for an evaluation of exertional chest pain. The patient had a history of VSD which had been seen at neonate. He had been checked regularly without events until the time of his presentation. Transthoracic echocardiography (TTE) and exercise treadmill test were performed. At parasternal short axis view, perimembranous VSD was detected with color Doppler imaging. There was a suspicious focal aneurysmal dilatation around the defect (Fig. 1, Supplementary movie 1 and 2). Left ventricular (LV) end-diastolic and end-systolic dimension were 52 mm and 31 mm, respectively. Qp/Qs was 1.2. Initially, these findings were considered clinically insignificant membranous septal aneurysm. The exercise treadmill test was negative. The patient was scheduled for a regular follow-up. However, the follow-up was

• Received: November 27, 2014 • Revised: April 15, 2015 • Accepted: May 19, 2015
• Address for Correspondence: Shin-Jae Kim, Department of Internal Medicine, Ulsan University Hospital, University of Ulsan College of Medicine, 877 Bangeojinsunhwando-ro, Dong-gu, Ulsan 682-714, Korea Tel: +82-52-250-8873, Fax: +82-52-251-8235, E-mail: kimsc226@gmail.com
• This is an Open Access article distributed under the terms of the Creative Commons Attribution Non-Commercial License (<http://creativecommons.org/licenses/by-nc/3.0>) which permits unrestricted non-commercial use, distribution, and reproduction in any medium, provided the original work is properly cited.

lost. Four years later, the patient visited emergency room due to nausea and vomiting lasting for one month. Continuous murmur is auscultated at left parasternal region. Mild pitting edema is presented at both lower legs. Abdomen computed tomography showed ascites and cutaneous edema without any evidence of cirrhosis of the liver, raising the possibility of right-sided heart failure. TTE showed enlargement of RV combined with depressed function. The LV was enlarged but the function of LV was preserved (Supplementary movie 3).

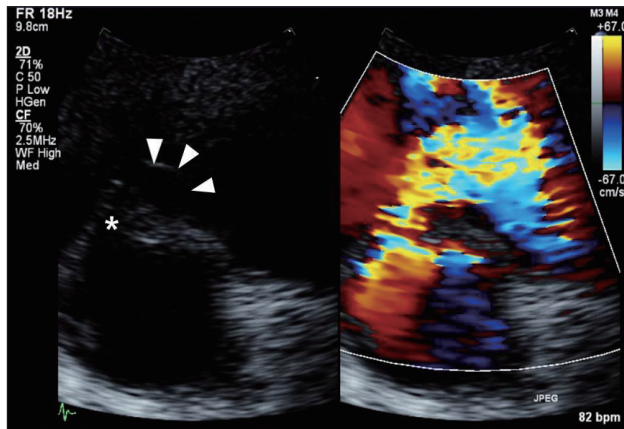


Fig. 1. Initial transthoracic echocardiography. At parasternal short axis view, perimembranous ventricular septal defect (star) was detected with 2 dimensional and color Doppler echocardiography. Focal aneurysmal dilatation (arrowheads) was noted around the defect.

However, the interventricular septum was flattened toward LV. At parasternal view, the width of VSD was wider than the width shown in the previous study (Supplementary movie 4). Color Doppler imaging showed turbulent systolic jet with a large proximal isovelocity surface area through the defect (Fig. 2C, Supplementary movie 5). There was definitely an elongated aneurysm of the right sinus of Valsalva with the tip ruptured (Fig. 2A, B, and C, Supplementary movie 6). Fortunately, the aortic valve had normal morphology without aortic regurgitation (Supplementary movie 7). Continuous wave Doppler revealed the diastolic jet flow from the aortic root to the RV through the ruptured aneurysm and systolic jet flow through VSD (Fig. 2D). The patient was referred to the department of cardiac surgery for open heart surgery. After median sternotomy, enlarged RV and prominent appendage of right atrium was observed. Aortotomy revealed ruptured aneurysm with windsock appearance which was closed with Dacron patch (Fig. 3A). However, the aortic valve was normal and left untouched. Subsequent right ventriculotomy exposed the perimembranous VSD sized about 1 cm which was also closed with Dacron patch (Fig. 3B). The weaning of cardiopulmonary bypass pump was successful. Postoperative course was uneventful. Follow-up echocardiogram after one year showed no evidence of residual shunt or AR and showed normalization of LV and RV size. The functions of both ventricles were well preserved.

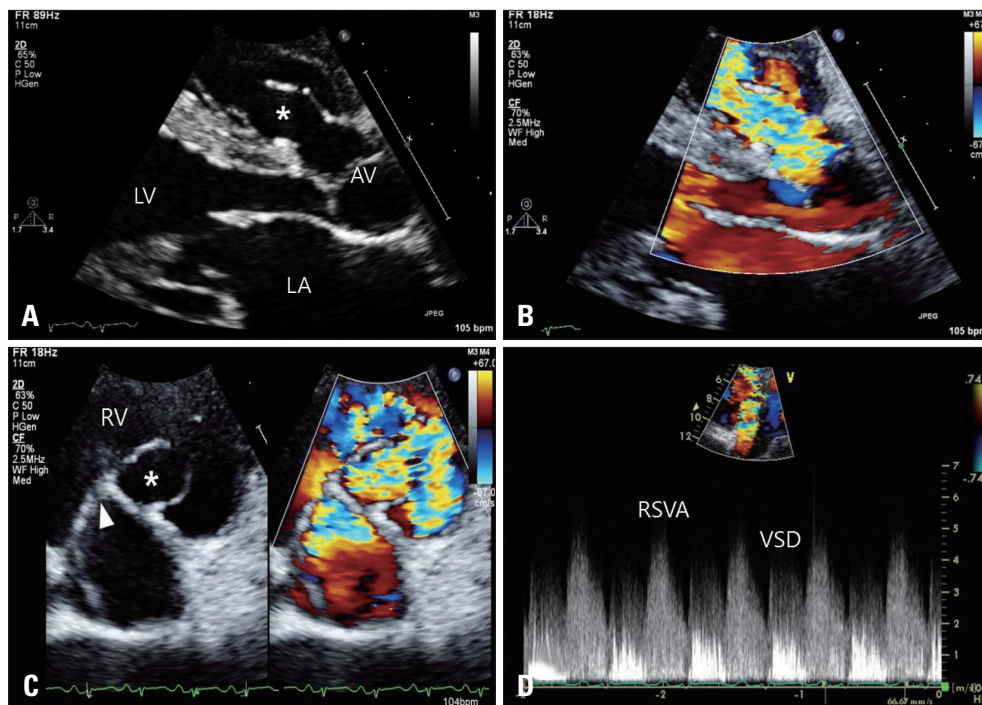


Fig. 2. Transthoracic echocardiography performed at the emergency room. A: At parasternal long axis view, 2-dimensional imaging showed the "windsock" appearance dilatation of right sinus of Valsalva (star) and its tip was ruptured. B: Color Doppler imaging showed diastolic jet flow through the ruptured right sinus of Valsalva aneurysm from the aortic root to the right ventricle. C: At parasternal short axis view, 2-dimensional and color Doppler imaging showed the RSVA (star) and VSD (arrowhead). D: At apical 5 chamber view, continuous wave Doppler showed systolic jet from VSD and diastolic jet from RSVA clearly. AV: aortic valve, LA: left atrium, LV: left ventricle, RV: right ventricle, RSVA: ruptured sinus of Valsalva aneurysm, VSD: ventricular septal defect.

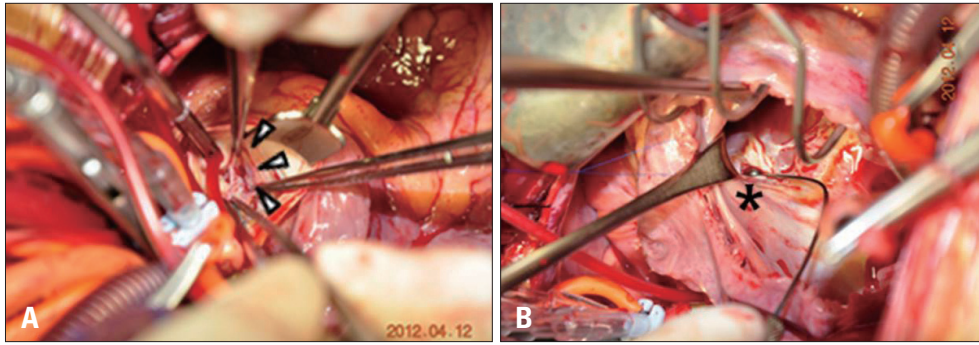


Fig. 3. Photographs of surgical repair. A: After aortotomy, there was small defect at the tip of sinus of Valsalva aneurysm (8 mm sized, arrowheads). B: After right ventriculotomy, probe indicated the perimembranous ventricular septal defect (1 cm sized, star).

DISCUSSION

Sinus of Valsalva aneurysms are very rare, with incidence rate ranging from 0.1% to 3.5% of all congenital heart diseases.⁷⁾ Sinus of Valsalva aneurysms occur three times more often in males, with the highest incidence in Asian populations.⁸⁾ Most sinus of Valsalva aneurysms arise from the right or the noncoronary sinuses. They commonly rupture into the RV or right atrium.

VSD and AR are frequently associated with RSVAs. The incidence rate of associated aortic sinus aneurysms and VSD ranges from 34.6% to 82.4%, with higher rate in Asian countries, including Korea.³⁾⁴⁾⁸⁻¹²⁾ Principal VSD associated with RSVAs is supracristal type.²⁻⁴⁾ Supracristal VSD is the predominant type seen in Asian countries, whereas perimembranous VSD is often seen in Western countries.⁵⁾⁶⁾ In the present case, RSVAs were associated with perimembranous VSD which was suspected on echocardiography based on the 11 o'clock direction of shunt flow, which was confirmed by the surgeon. The size of VSD was larger than measured with TTE. This could be due to the shape of VSD that was ovoid, which might have induced error in the measuring of the defect size. In patients with asymmetric defects like the present case, real-time three-dimensional echocardiography is a complementary tool to assess the size and morphology of defects more accurately.¹³⁾

The incidence rate of associated sinus of Valsalva aneurysm and AR ranges from 20% to 64.7%.³⁾⁸⁾¹¹⁾ It is well known that supracristal VSD is the preceding pathology.¹¹⁾ Anatomically, most sinus of Valsalva aneurysms are congenital in origin, caused by the lack of fusion between the media of the aorta and the annulus fibrosus of the aortic valve.¹⁾ Hemodynamically, the flow through VSD produces Venturi effect, a tendency for the related aortic sinus and cusp to pull away from closure. These two mechanisms induce AR, and AR begets AR. Surgical treatment of AR in addition to repairing the RSVAs is needed in some patients. The incidence of associated perimembranous VSD and AR is 2.4 times less common compared to that of associated supracristal VSD and AR.¹⁴⁾ Eapen et al.¹⁵⁾ suggested echocardiographic features of perimembranous VSD susceptible to AR, presence of aortic cusp override, aortic cusp movement abnormalities, and presence of

color flow mapping across ventricular septum. In the present case, the perimembranous VSD was not associated with AR. There was no finding of aortic cusp override or aortic cusp movement abnormality, decreasing the possibility of associated AR despite the strong jet flow across ventricular septum.

Echocardiography is necessary for the definitive diagnosis. The accuracy of TTE is about 75%.¹⁶⁾ TTE shows the site and size of aneurysm, the direction of the rupture, which helped us find the associated VSD, fluttering of tricuspid valve, and shunt flow.¹⁶⁾¹⁷⁾ In case TTE fails to show the accurate anatomical structure, transesophageal echocardiography (TEE) images of various angles such as RV outflow tract, sinus of Valsalva and aortic root, and color Doppler help us assess the exact flow velocity and direction through RSVAs or VSD.¹⁸⁾

The membranous septal aneurysms are bulging of the membranous portion of the interventricular septum below the aortic annulus in the RV. Membranous septal aneurysms, associated with VSD in 19% of cases,¹⁹⁾ play a role of spontaneous VSD closure in children. Sinus of Valsalva aneurysms must be distinguished from membranous septal aneurysms because these two distinct pathologies lead to different results. Sinus of Valsalva aneurysm leads to rupture causing serious symptoms like the current case, whereas membranous septal aneurysms lead to spontaneous VSD closure in selected cases, especially in children.²⁰⁾ However, these two distinct pathologies sometimes are difficult to differentiate, especially in poor echo window, because they occur anatomically adjacent to each other at the level of aortic annulus. Sinus of Valsalva aneurysms are located above the aortic annulus (Fig. 4A), whereas membranous septal aneurysms are located below the annulus (Fig. 4B). Therefore, multiple views are helpful to differentiate these two pathologies. With only a single image at the parasternal short axis view, it is difficult to differentiate whether the aneurysm is located above or below the annulus. In the present case, at initial exam, the operator rashly concluded that the focal aneurysm around the VSD was membranous septal aneurysm based on the parasternal short axis view. If the operator had a close look at the aneurysm on multiple views, the aneurysm would have been found to be located above the annulus, the diagnosis would have changed.

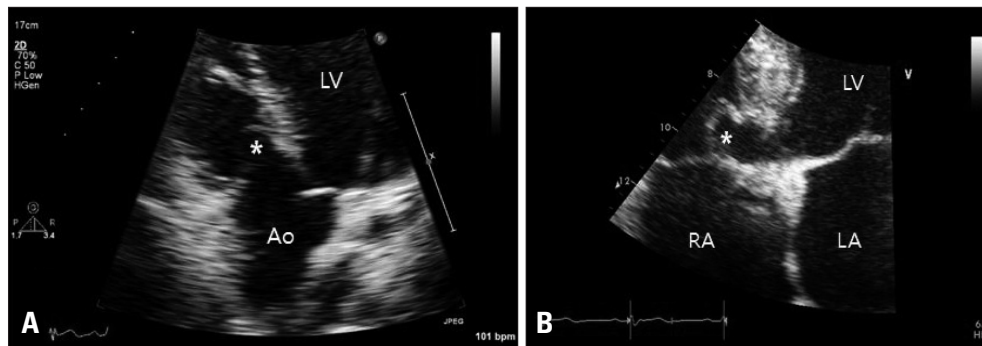


Fig. 4. Comparative images between sinus of Valsalva aneurysm and membranous septal aneurysm. A: Current case of sinus of Valsalva aneurysm at apical 5 chamber view showed aneurysmal dilatation (star) above the aortic annulus. B: A case of membranous septal aneurysm at apical 4 chamber view showed aneurysmal dilatation (star) below the aortic annulus. Ao: aorta, LA: left atrium, LV: left ventricle, RA: right atrium.

In conclusion, in young patients with unexplained chest pain, sinus of Valsalva aneurysm could be suspected. Echocardiography should be performed. Careful TTE with multiple windows and TEE in selected cases are needed to confirm the diagnosis. Sinus of Valsalva aneurysms should be differentiated from membranous septal aneurysms because these two pathologies occur anatomically adjacent to each other at the level of aortic annulus. Unruptured or ruptured sinus of Valsalva aneurysms with VSD should be repaired surgically as early as possible. Before going to the operating room, it should be determined whether AR is present. If AR is present, surgical treatment of AR in addition to repairing the RSVA is needed.

SUPPLEMENTARY MOVIE LEGENDS

Movie 1. Transthoracic echocardiography image at initial presentation showing the suspicious aneurysmal dilatation around the aortic annulus at parasternal long axis view.

Movie 2. Transthoracic echocardiography images at initial presentation showing the suspicious aneurysmal dilatation around the aortic annulus at parasternal short axis view.

Movie 3. Transthoracic echocardiography image at emergency room at parasternal long axis view.

Movie 4. Transthoracic echocardiography image at emergency room showing ventricular septal defect.

Movie 5. Side by side 2D and color Doppler image at parasternal short axis view showing turbulent jets toward the right ventricle through the ventricular septal defect and the ruptured sinus of Valsalva aneurysm.

Movie 6. Transthoracic echocardiography image at emergency room showing elongated aneurysm of the right sinus of Valsalva with the tip ruptured.

Movie 7. Transthoracic echocardiography image at emergency room showing normal morphology of aortic valve without aortic regurgitation.

REFERENCES

1. Edwards JE, Burchell HB. *The pathological anatomy of deficiencies between the aortic root and the heart, including aortic sinus aneurysms.* *Thorax* 1957;12:125-39.

2. Michael DF, William HP. *A sinus of valsalva fistula.* In: Alexander RW, Sclant RC, Fuster V, editors. *Hurst's the heart, arteries and veins.* 9th ed. New York: McGraw-Hill;1998.
3. Lee TH, Lee DW, Cho JY, Hyun MC, Lee SB. *Clinical features and surgical results of ruptured sinus of valsalva aneurysm.* *Korean J Pediatr* 2006;49:287-91.
4. Moon KS, Choi RK, Lim DS, Park HS, Hong SK, Lee YT, Hwang HK. *Clinical characteristics in patients with ruptured aneurysm of sinus of Valsalva.* *Korean Circ J* 2000;30:183-90.
5. De Bakey ME, Diethrich EB, Liddicoat JE, Kinard SA, Garrett HE. *Abnormalities of the sinuses of valsalva. Experience with 35 patients.* *J Thorac Cardiovasc Surg* 1967;54:312-32.
6. Sanchez HE, Barnard CN, Barnard MS. *Fistula of the sinus of Valsalva.* *J Thorac Cardiovasc Surg* 1977;73:877-9.
7. Takach TJ, Reul GJ, Duncan JM, Cooley DA, Livesay JJ, Ott DA, Frazier OH. *Sinus of Valsalva aneurysm or fistula: management and outcome.* *Ann Thorac Surg* 1999;68:1573-7.
8. Chu SH, Hung CR, How SS, Chang H, Wang SS, Tsai CH, Liao CS, Tseng CD, Tseng YZ, Lee YT, et al. *Ruptured aneurysms of the sinus of Valsalva in Oriental patients.* *J Thorac Cardiovasc Surg* 1990;99:288-98.
9. Norwicki ER, Aberdeen E, Friedman S, Rashkind WJ. *Congenital left aortic sinus-left ventricle fistula and review of aortocardiac fistulas.* *Am Thorac Surg* 1977;23:378-88.
10. Burakovskiy VI, Podsolkov VP, Sabirov BN, Nasedkina MA, Alekian BG, Dvinyaninova NB. *Ruptured congenital aneurysm of the sinus of Valsalva. Clinical manifestations, diagnosis, and results of surgical corrections.* *J Thorac Cardiovasc Surg* 1988;95:836-41.
11. van Son JA, Danielson GK, Schaff HV, Orszulak TA, Edwards WD, Seward JB. *Long-term outcome of surgical repair of ruptured sinus of Valsalva aneurysm.* *Circulation* 1994;90(5 Pt 2):II20-9.
12. Joo SJ, Koh KG, Kim YH, Park YB, Choi YS, Seo JD, Lee YW, Park JH, Suh KP. *Clinical observation on ruptured aneurysm of the sinus of Valsalva.* *Korean Circ J* 1987;17:149-58.
13. Acar P, Abadir S, Aggoun Y. *Transcatheter closure of perimembranous ventricular septal defects with Amplatzer occluder assessed by real-time three-dimensional echocardiography.* *Eur J Echocardiogr* 2007;8:110-5.
14. Schmaltz AA, Schaefer M, Hentrich F, Neudorf U, Brecher AM, Asfour B, Urban AE. *{Ventricular septal defect and aortic regurgitation-pathophysiological aspects and therapeutic consequences}.* *Z Kardiol* 2004;93:194-200.
15. Eapen RS, Lemler MS, Scott WA, Ramaciotti C. *Echocardiographic characteristics of perimembranous ventricular septal defects associated with aortic regurgitation.* *J Am Soc Echocardiogr* 2003;16:209-13.
16. Dev V, Goswami KC, Shrivastava S, Bahl VK, Saxena A. *Echocardiographic diagnosis of aneurysm of the sinus of Valsalva.* *Am Heart J* 1993;

- 126:930-6.
17. Terdjman M, Bourdarias JP, Farcot JC, Gueret P, Dubourg O, Ferrier A, Hanania G. *Aneurysms of sinus of Valsalva: two-dimensional echocardiographic diagnosis and recognition of rupture into the right heart cavities.* *J Am Coll Cardiol* 1984;3:1227-35.
 18. Wang KY, St John Sutton M, Ho HY, Ting CT. *Congenital sinus of Valsalva aneurysm: a multiplane transesophageal echocardiographic experience.* *J Am Soc Echocardiogr* 1997;10:956-63.
 19. Choi M, Jung JI, Lee BY, Kim HR. *Ventricular septal aneurysms in adults: findings of cardiac CT images and correlation with clinical features.* *Acta Radiol* 2011;52:619-23.
 20. Beerman LB, Park SC, Fischer DR, Fricker FJ, Mathews RA, Neches WH, Lenox CC, Zuberbuhler JR. *Ventricular septal defect associated with aneurysm of the membranous septum.* *J Am Coll Cardiol* 1985;5:118-23.