Cardiovascular and Metabolic Science www.ancam.org.mx

Clinical case

Vol. 36 No. 3
July-September 2025

doi: 10.35366/121371

Right coronary sinus fistula for ruptured aneurysm to the right atrium in a patient with Noonan syndrome

Fístula del seno coronario derecho por aneurisma roto hacia aurícula derecha en una paciente con síndrome de Noonan

Oziel Vega-Nañez,*,‡ Fernando Manuel Calderón-Osorio,*,§ Eleazar Muruato-Ontiveros,*,¶ José Luis Vázquez-Torres,*,‖ Efrén López-Rodríguez*,**

Keywords:
vascular fistula, Noonan syndrome, ruptured aneurysm, mutation.

Palabras clave:
fístula vascular, síndrome de Noonan, aneurisma roto, mutación.

ABSTRACT

Introduction: an aneurysm at the level of the sinuses of Valsalva is a pathology of congenital or acquired origin. The presence of a fistula can also be asymptomatic or express itself at any age with different manifestations. **Case presentation:** 34-year-old female patient carrier of Noonan syndrome (NS); surgical history of ventricular septal defect closure. Since the patient was 27 years old, she presented episodes of dyspnea. Cardiopulmonary auscultation revealed the presence of a continuous murmur in the second right parasternal intercostal space. A transthoracic echocardiogram was performed with findings of a fistulized right coronary aneurysm. Surgery was performed where a fistula from the right coronary sinus to the right atrium was observed, proceeding to closure. **Conclusions:** a fistula from a coronary sinus to a cardiac cavity is a rare presentation of a ruptured aneurysm, so it highlights the presentation of this pathology in a patient with a RASopathy related to cardiac defects. Diagnosis requires an adequate approach. However, in some cases, the exact location of the defect can be determined during surgery. Early diagnosis and treatment are crucial.

RESUMEN

Introducción: *un aneurisma a nivel de los senos de Valsalva es una patología de origen congénito o adquirido. La presencia de fístulas, pueden así mismo, ser asintomáticas o expresarse a cualquier edad con distintas manifestaciones.* ***Presentación del caso:*** *paciente mujer de 34 años, portadora de síndrome de Noonan (SN); antecedente quirúrgico de cierre de comunicación interventricular. Desde los 27 años presenta episodios de disnea. La auscultación cardiopulmonar destaca la presencia de soplo continuo en segundo espacio intercostal paraesternal derecho. Se realiza ecocardiograma transtorácico con hallazgos de un aneurisma coronario derecho fistulizado. Se llevó a cabo cirugía donde se observa fístula de seno coronario derecho a aurícula derecha, procediendo a cierre.* ***Conclusiones:*** *una fístula de un seno coronario hacia una cavidad cardíaca es una presentación poco frecuente de un aneurisma roto, por lo que destaca la presentación de esta patología en una paciente con una RASopatia relacionada con defectos cardíacos. El diagnóstico requiere un adecuado abordaje. Sin embargo, en algunos casos, la localización exacta del defecto se consigue en la cirugía. Su diagnóstico y tratamiento temprano es crucial.*

* Hospital de Especialidades No. 71, Instituto Mexicano del Seguro Social (IMSS).
‡ Cardiology resident.
§ Cardiologist, Imageologist.
¶ Cardiothoracic Surgeon.
‖ Interventional Cardiologist.
** Echocardiographer Cardiologist.

Received:
01/03/2025
Accepted:
07/23/2025

Abbreviation:
MAPK = Mitogen-Activated Protein Kinase.
NS = Noonan syndrome.

INTRODUCTION

An aneurysm at the level of the sinuses of Valsalva is a pathology of either congenital or acquired origin, with an incidence from 0.14 to 3.5% of patients undergoing cardiac surgery. Regarding its congenital origin, it seems to be related to defects during the formation of the cardiac loop in the embryonic period, during the fusion of the arterial cones to form both infundibula. An inadequate fusion in this area would result in an area of weakness that would

How to cite: Vega-Nañez O, Calderón-Osorio FM, Muruato-Ontiveros E, Vázquez-Torres JL, López-Rodríguez E. Right coronary sinus fistula for ruptured aneurysm to the right atrium in a patient with Noonan syndrome. Cardiovasc Metab Sci. 2025; 36 (3): 166-171. https://dx.doi.org/10.35366/121371

undergo progressive dilation. The same origin would have the interventricular communication that sometimes accompanies it. Associated cases of aortic coarctation and bileaflet aorta have been reported.[1] The clinical presentation is variable, presenting even in acute heart failure, when they are ruptured, and according to the site where the fistula develops, and may cause sudden death, secondary to open rupture of the pericardium, myocardial infarction, or atrioventricular block. The presence of a fistula can also be asymptomatic or express itself at any age with different manifestations.[2]

The case of a young patient with Noonan syndrome (NS) with a history of surgical closure of a ventricular septum defect in childhood, with a right coronary sinus aneurysm, probably congenital, which was ruptured, with a fistula to the right atrium, causing precapillary pulmonary hypertension.

CASE PRESENTATION

34-year-old female patient with the following important antecedents: smoking and ethylism denied; carrier of Noonan syndrome; surgical history of ventricular septal defect closure at 4 years of age.

Since the patient was 27 years old, presented episodes of dyspnea on small efforts and palpitations, and was kept under expectant management until the patient was 34 years old, reinitiating the approach due to further deterioration of her functional class. Physical examination revealed hypertelorism and eyelid ptosis, short stature, and mild neurocognitive deficit; cardiopulmonary auscultation revealed the presence of a continuous grade IV/VI murmur in the second right parasternal intercostal space.

A transthoracic echocardiogram was performed with findings suggestive of fistulized right coronary aneurysm, a high velocity flow was found in the subaortic region (at 10-11 hour clockwise in short axis projection) which is directed towards the right cavities *(Figure 1)*, with a coronary flow pattern on continuous Doppler interrogation, with a maximum velocity recorded of 6. 2 m/s, maximum gradient of 154 mmHg, dilated right ventricle (basal diameter 39 mm, medium 30 mm, longitudinal 90 mm). Angiotomography was performed with the presence of a right coronary aneurysm type defect communicating to the right cavities *(Figure 2)*.

With these findings, an invasive approach by hemodynamics was performed, where the aortogram showed evidence of a fistula at the level of the right coronary sinus draining into the right cavities, resulting in a QP:QS of 1.66, in addition to precapillary pulmonary hypertension (> 51 mmHg mean pulmonary pressure and 3.8 Wood units of PVR) *(Figure 3)*.

Surgery was performed where a fistula from the right coronary sinus to the right atrium above the septal valve of the tricuspid was observed, proceeding to closure in two planes and placement of a pericardial patch in the right atrium *(Figure 4)*. There were no postoperative complications, and the clinical course was uneventful.

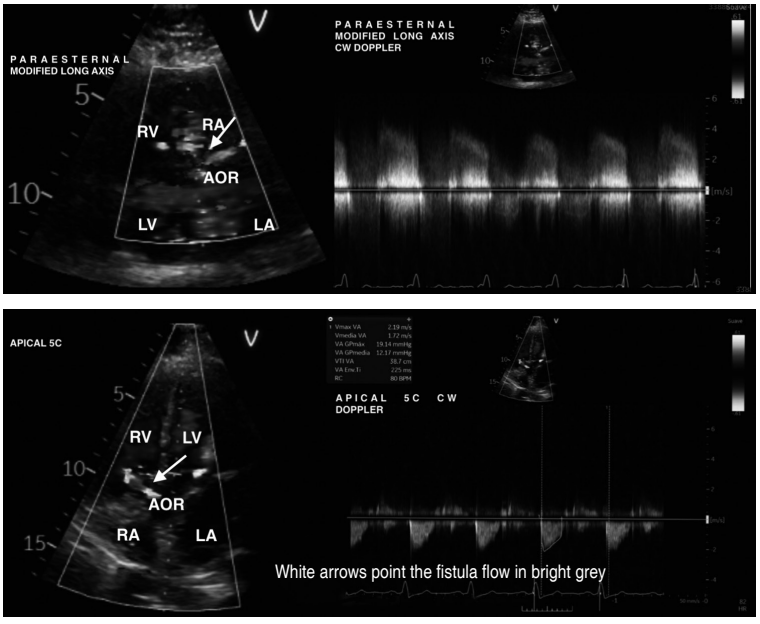

Figure 1: Transthoracic echocardiogram. Upper images in the parasternal long axis, lower images in the four-chamber apical axis showing a fistulized right coronary aneurysm (white arrows), with high-speed flow in the subaortic region, which is directed towards the right cavities.
AOR = aortic root. LA = left atrium. LV = left ventricle. RA = right atrium. RV = right ventricle.

DISCUSSION

The RASopathies are a group of disorders caused by a germline mutation in one of the

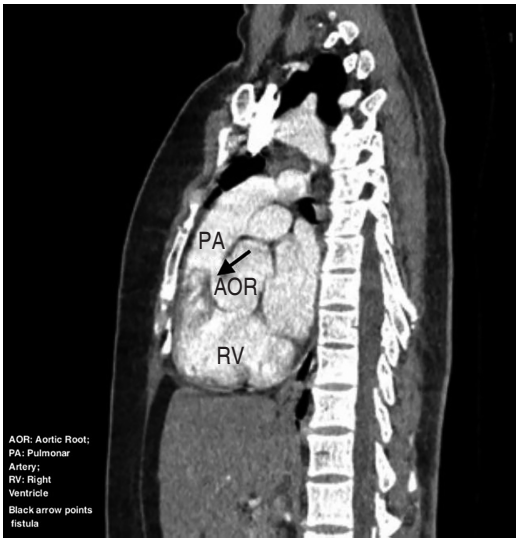

Figure 2: Angiotomography showing a right coronary aneurysm type defect that communicates to the right cavities.
AOR = aortic root. PA = pulmonar artery. RV = right ventricle.

genes encoding a component of the RAS/MAPK (Mitogen-Activated Protein Kinase) pathway, a signal transduction system that regulates cell growth, division, and differentiation. These disorders, including neurofibromatosis type 1, Noonan syndrome, cardiofaciocutaneous syndrome, Costello syndrome, and Legius syndrome, among others, have overlapping clinical features due to RAS/MAPK dysfunction. Although several of the RASopathies are very rare, collectively, these disorders are relatively common.[3] No generally accepted definition and definitive delineation of RASopathies exist, so far.[4]

Noonan syndrome is characterized by a high clinical and genetic heterogeneity, with variable and age-varying involvement of multiple organs and systems. Because of this variability, it is essential that physicians involved in its care are familiar with its manifestations and aware of follow-up recommendations, including monitoring of growth and development.[5]

Essential phenotypic features of NS include facial anomalies, short stature, congenital cardiac anomalies, coagulation disorders, and a variable degree of cognitive delay. In the face, one can observe a broad forehead, hypertelorism, ptosis, epicanthal fold, high labial philtrum, and labial ridges with accentuated upward angulation, low-set and posteriorly rotated ears, arched eyebrows or with a superior vertex angle, light blue iris, wide neck, and low posterior hairline. In the thorax, pectus excavatum inferior/carinatum superior and separated areolas are common; in 50-80% of cases there are congenital cardiac anomalies, mainly pulmonary stenosis (20-50%) and hypertrophic cardiomyopathy (20-30%), but also atrial and ventricular septal defects and tetralogy of Fallot. There is considerable clinical and genetic heterogeneity in NS, and a much larger series of patients needs to be studied.[6]

Sinus of Valsalva aneurysm, a congenital or acquired cardiac defect present in approximately 0.09% of the general population, often presents as an incidental finding during cardiac imaging. However, an echocardiogram is the standard imaging technique for such findings; cardiac computed tomography angiography has been increasingly utilized.[7]

Aneurysms of the sinus of Valsalva usually develop due to weakness of the aortic wall that is part of the coronary sinus; the right one is most frequently affected, through which a saccular aneurysm gradually and progressively protrudes into a cardiac cavity, usually the right ventricle. The acquired ones are secondary to trauma, endocarditis, or syphilis, or to aging itself. Congenital are the most frequent, secondary to failure in the fusion of the middle layer of the aorta with the fibrous skeleton of the heart, which is the basis for its development.[8]

The right coronary sinus is most frequently affected, followed by the non-coronary sinus and rarely the left coronary sinus, which are associated with other defects, including ventricular septal defect in 30 to 60% and aortic valve anomalies, such as aortic insufficiency (20 to 30%), bicuspid valve (10%), aortic stenosis (6.5%), as well as such as pulmonary stenosis (9.7%), coarctation (6.5%), patent ductus arteriosus (3.2%), tricuspid regurgitation (3.2%) and interatrial defect.[9] It is a rare pathology. It has been reported in 0.09% in a series of autopsies, in 0.14 to 0.23% in Western surgical studies, and in 0.46 to 3.5% in Eastern studies.[10]

From an embryological point of view, aneurysmal dilation of one of the sinuses of Valsalva results from an incomplete fusion of

the valvular part of the septum in its distal part. It is, therefore, related to defects of the membranous septum of the ventricular septum, as occurs more frequently with the type of interventricular communications that are of this type, a membranous defect.[11]

Our patient has a history of a congenital ventricular septal defect. As mentioned, this defect is the most common associated lesion, followed by aortic insufficiency, which is often associated with a ventricular septal defect or a bicuspid aortic valve. The patient's sinus of Valsalva aneurysm was detected in adulthood, presenting with a rupture with a fistula towards the right atrium.

According to the study group of De Bakey et al., anomalies of the sinus of Valsalva can be classified into three groups:[12]

1. Sinus aneurysm.
2. Aneurysm with fistula.
3. Fistula.

The patient belongs to group 2, draining into the right atrium, which is the second common site (60%). Other sites where fistulization can

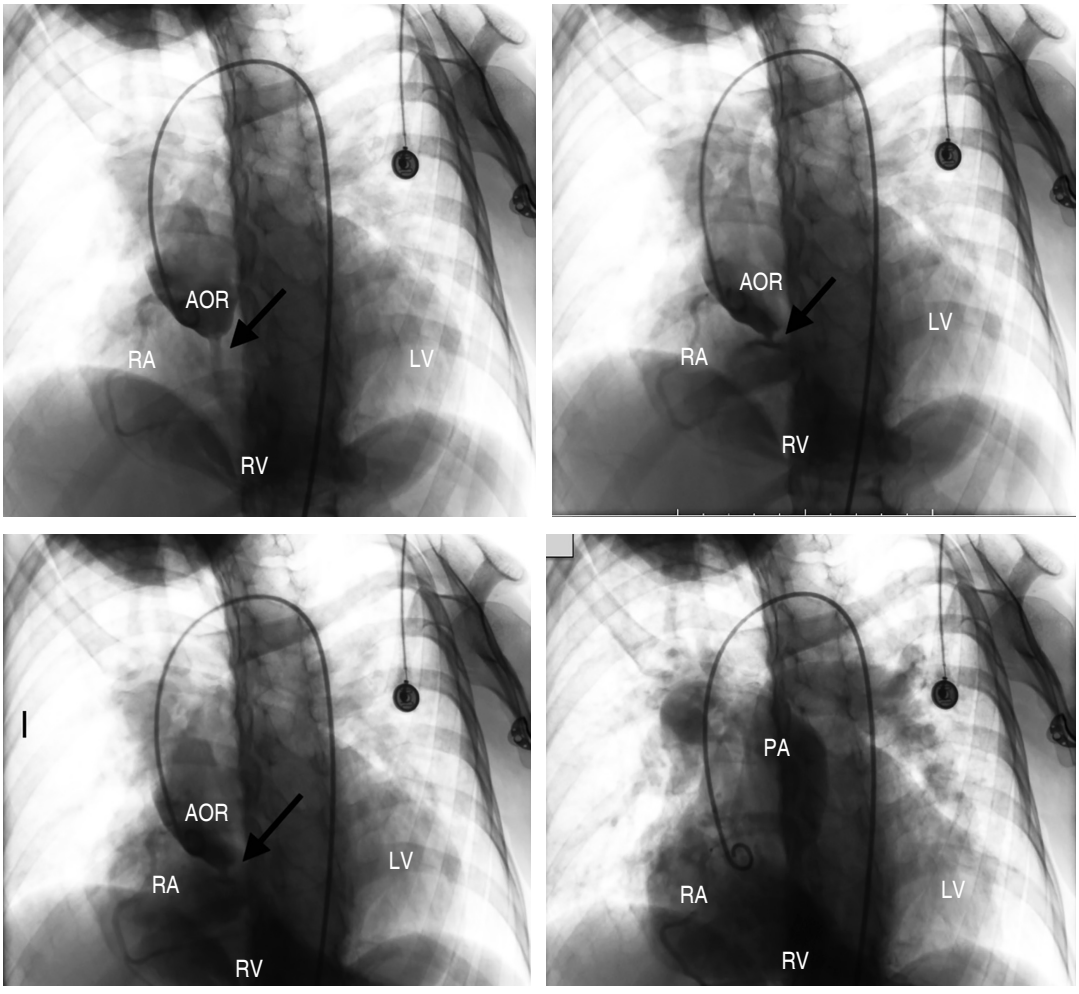

Figure 3: Aortogram in different phases of the cardiac cycle that shows a fistula at the level of the right coronary sinus that drains into the right cavities (black arrows). In the lower right box, the passage of contrast medium into the pulmonary circulation is observed.
AOR = aortic root. LV = left ventricle. PA = pulmonar artery. RA = right atrium. RV = right ventricle black arrow points fistula.

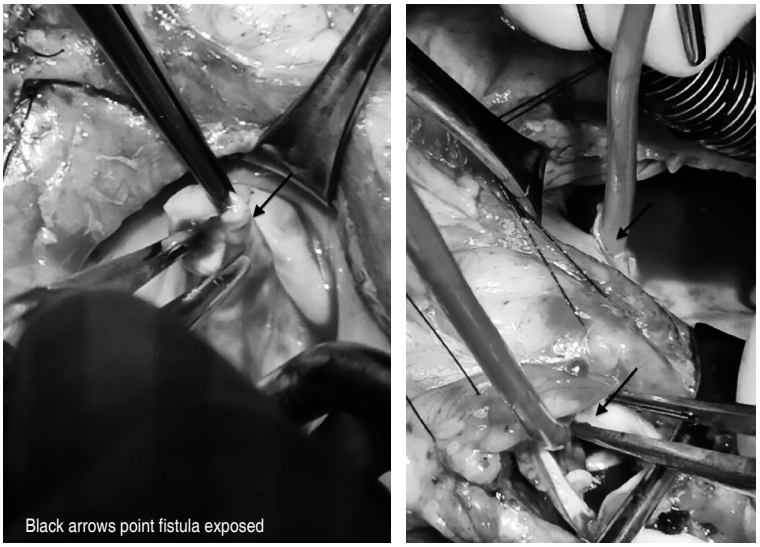

Figure 4: Images obtained during surgery. In the left image, adequate exposure of the fistula is achieved, observed from the right atrium. In the right image, a crossed fistula is observed, extending the from the right atrium (above) to the aorta (below).

occur are the right ventricle (60%), left atrium (6%), left ventricle (4%), or pericardium (1%). It is relevant to mention that the rupture of acquired aneurysms is usually towards the pericardium or the pleural space and is generally fatal.[9] A fistula of the sinus of Valsalva was reported after a Ross procedure.[13]

Usually, the aneurysm is asymptomatic until it ruptures, usually between the second and third decades of life. The clinical picture may be acute chest pain or heart failure. The physical examination of patients with a ruptured aneurysm typically presents a continuous murmur in the mesocardium; however, only an expulsive murmur or an expulsive murmur combined with a diastolic murmur may be heard.[14]

Cardiovascular imaging plays a crucial role in the assessment and management of aortic root and thoracic aorta ectasia and aneurysms. Sinus of Valsalva aneurysms are rare entities. Unique complications associated with sinus of Valsalva aneurysms make them different from traditional aortic root aneurysms. Established guidelines on the diagnosis and management of sinus of Valsalva aneurysms are lacking.[15]

Regarding diagnosis, angiography was considered the gold standard for diagnosis, however, less invasive techniques such as transthoracic and transesophageal echocardiography are the most used currently with a diagnostic certainty of 75 and 90% respectively for ruptured and unruptured aneurysms, allowing a more precise localization of the affected sinus, of the fistula to the cavities, in the identification of other cardiac alterations, in the delimitation of the size and morphology of congenital aneurysms as well as in the identification of aneurysm prolapse through a ventricular septal defect.[16]

In transthoracic echocardiography, the demonstrated diastolic flow reversal in the descending thoracic aorta occurs due to a significant left-to-right shunt from the aortic root to the right-sided cardiac chamber, creating similar hemodynamic physiology as severe aortic regurgitation.[17]

This condition, left to its evolution, can lead to symptoms of pulmonary hypertension, heart failure, and myocardial ischemia. The patient had already presented in a symptomatic context, corroborating precapillary pulmonary hypertension through hemodynamics.[18] This is due to excessive blood flow to the right cavities, which overloads them. The increase in pulmonary arterial pressure and blood flow causes pulmonary edema and right heart failure, which can lead to a fulminant course and premature death.[19]

Timely surgical intervention is imperative to address these complications once the diagnosis is confirmed. The prognosis following surgical repair of SoVA is generally favorable. Early diagnosis and prompt surgical intervention are essential for enhancing survival rates. The reported preoperative mortality rate stands at 7%, with a long-term survival rate of approximately 63% in extensive case series.[20]

The repair reduces the risk of the aforementioned complications, improving life expectancy. Successful repair has been achieved via transcatheter methods, using septal occlusion devices, ductal occluders, Amplatzer vascular occluders, and even Rashkind umbrellas. Due to the rarity of this pathology, no clinical trials have been carried out that compare the superiority of different surgical techniques. The most commonly used is the «double exposure technique», in which both the aorta and the fistula end chamber are

explored. The aneurysmal sac is removed, and the resulting defect is repaired by direct suture or patch closure. Currently, the 10-year survival rate after surgical repair is 90%.[18]

CONCLUSIONS

A fistula from a coronary sinus to a cardiac cavity is a rare presentation of a ruptured aneurysm, so it highlights the presentation of this pathology in a patient with a rasopathy related to cardiac defects. Diagnosis requires an adequate approach, ranging from experience in physical examination to studies such as echocardiography and cardiac catheterization. However, in some cases, the exact location of the defect can be determined during surgery. Early diagnosis and treatment are crucial, since free evolution can lead to various complications, including pulmonary hypertension, as in the case of the patient. If complications occur, they can significantly impact the quality of life and life expectancy. For this reason, its diagnostic and therapeutic approach requires specialized centers with experience in congenital heart disease.

REFERENCES

1. Caballero J, Arana R, Calle G et al. Aneurisma congénito del seno de Valsalva roto a ventrículo derecho, comunicación interventricular e insuficiencia aórtica. Rev Esp Cardiol. 1999; 52: 635-638.
2. Muñoz OE, Fortich MF, Velásquez O. Aneurisma del seno de Valsalva disecando hacia el septo interventricular en una paciente embarazada. Rev Colomb Cardiol. 2015; 22 (6): 321-325.
3. Hebron KE, Hernandez ER, Yohe ME. The RASopathies: from pathogenetics to therapeutics. Dis Model Mech. 2022; 15 (2): dmm049107.
4. Zenker M. Clinical overview on RASopathies. Am J Med Genet C Semin Med Genet. 2022; 190 (4): 414-424.
5. Carcavilla A, Suárez-Ortega L, Rodríguez Sánchez A et al. Síndrome de Noonan: actualización genética, clínica y de opciones terapéuticas [Noonan syndrome: genetic and clinical update and treatment options]. An Pediatr (Engl Ed). 2020; 93 (1): 61.e1-61.e14.
6. Allanson JE. Noonan syndrome. Am J Med Genet C Semin Med Genet. 2007; 145C (3): 274-279.
7. Weinreich M, Yu PJ, Trost B. Sinus of valsalva aneurysms: review of the literature and an update on management. Clin Cardiol. 2015; 38 (3): 185-189.
8. Goldberg N, Krasnow N. Sinus of Valsalva aneurysms. Clin Cardiol. 1990; 13 (12): 831-836.
9. Galicia M, Marín B, Mercado O, Espinoza S, Martínez M, Villal-Pando E. Aneurisma del seno de Valsalva roto. Informe de casos y revisión de la literatura. Cir Cir. 2009; 77: 473-477.
10. Vautrin E, Barone-Rochette G, Baguet JP. Rupture of right sinus of Valsalva into right atrium: ultrasound, magnetic resonance, angiography and surgical imaging. Arch Cardiovasc Dis. 2008; 101 (7-8): 501-502.
11. Marroush TS, Boshara AR, Botros B et al. Rupture of sinus of Valsalva aneurysm: two case reports and a concise review of the literature. Heart Lung. 2018; 47 (2): 131-135.
12. Kanzaki Y, Terasaki F, Fuji S et al. A sinus of Valsalva-right atrium fistula without aneurysm formation. Intern Med. 2010; 49 (8): 749-751.
13. Williams-Phillips S. Post Ross procedure aortic right sinus of Valsalva fistula to right ventricle. West Indian Med J. 2011; 60 (6): 669-673.
14. Adams JE, Sawyers JL, Scott HW Jr. Surgical treatment of aneurysm of aortic sinus with aorticoatrial fistula. Surgery. 1957; 41: 26-42.
15. Xu B, Kocyigit D, Betancor J et al. Sinus of Valsalva aneurysms: a state-of-the-art imaging review. J Am Soc Echocardiogr. 2020; 33 (3): 295-312.
16. Garrido MA, Oliver RJM, González AE, Mesa GJM, Benito F, Sobrino JA. Ecocardiografía transesofágica multiplanar en la valoración preoperatoria de fístulas de senos de Valsalva en cavidades derechas. Rev Esp Cardiolern. 2002; 55 (1): 29-36.
17. Doost A, Craig JA, Soh SY. Acute rupture of a sinus of Valsalva aneurysm into the right atrium: a case report and a narrative review. BMC Cardiovasc Disord. 2020; 20 (1): 84.
18. Jaswal V, Kumar V, Thingnam SKS, Puri GD. Surgical repair of ruptured sinus of Valsalva aneurysm: 13-year single center experience. J Card Surg. 2021; 36 (4): 1264-1269.
19. González HOR, Torres CJA, Hernández del Río JE. Aneurisma del seno de Valsalva roto con fístula a ventrículo derecho. Presentación de un caso clínico y revisión de la literatura. Insuficiencia Cardiaca. 2017; 12 (3): 134-141.
20. Sari NMAW, Dewangga MSY, Wibhuti IBR et al. Ruptured sinus of Valsalva aneurysm coexisted with congenital ventricular septal defect: a case series. Egypt Heart J. 2023; 75 (1): 90.

Declaration of patient consent: the patient and tutor provided consent for the publication of the case, with personal data omited.

Funding: no financial support was received for this study.

Conflict of interest: the authors declare no conflict of interest.

Correspondence:
Fernando Manuel Calderón-Osorio
E-mail: drcalderoncardio@gmail.com