www.mednet.gr/archives
ARCHIVES OF HELLENIC MEDICINE: ISSN 11-05-3992

CASE REPORT
ΕΝΔΙΑΦΕΡΟΥΣΑ ΠΕΡΙΠΤΩΣΗ

A 62-year-old woman with interatrial defect type sinus venosus

ARCHIVES OF HELLENIC MEDICINE 2023, 40(3):418–422
ΑΡΧΕΙΑ ΕΛΛΗΝΙΚΗΣ ΙΑΤΡΙΚΗΣ 2023, 40(3):418–422

G.M. Aquino,[1]
S.B. Cobra,[1]
V.M. dos Santos,[2]
O.L.R. Almeida[1]

[1]Institute Hospital of Base of Federal District, Brasília-DF
[2]Armed Forces Hospital and Catholic University, Brasília-DF, Brazil

Γυναίκα 62 ετών με μεσοκολπικό έλλειμμα τύπου φλεβοκόλπου

Περίληψη στο τέλος του άρθρου

The atrial septal defects are among the most common congenital cardiac anomalies diagnosed among adult individuals and may affect approximately 25% of children, characterized by interatrial communication that often persists long standing undetected. The sinus venosus defect responds to between 5 and 10% of the diverse anatomical types. Spontaneous closure of small-diameter defects usually occurs before 5 years of age, but the sinus venosus and ostium primum types do not evolve with this kind of closure. Patients with enlarged right heart chambers may manifest clinical symptoms of the right or global cardiac dysfunction, arrhythmias, thromboembolic episodes, and pulmonary arterial hypertension (12% in the corrected and 34% in uncorrected defects). Routine evaluation by echocardiography increases the possibility of early diagnosis. The report of a 62-year-old female with a late diagnosis of a sinus venosus type defect aims to enhance the suspicion index of primary health workers about this condition.

Key words

Arrhythmia
Atrial septal defect
Echocardiography

Submitted 22.5.2022
Accepted 4.6.2022

Atrial septal defect (ASD) is one of the most prevalent congenital heart anomalies found in adults, occurs in approximately 25% of children, is characterized by an opening at the interatrial septum, and can be undiagnosed for several decades.[1–20] ASDs may occur either like an isolated phenomenon or associated with some hereditary condition like Down's, Noonan's, Treacher-Collins', Turner's, and Thrombocytopenia-absent radius syndromes, and maternal exposure to rubella or drugs like cocaine.[14] An 80-year-old woman was reported with an ASD of 5.0 mm and left-to-right shunt besides a Chiari network in the right atrium (RA) near the oval window.[13] The elderly patient had long-standing atrial fibrillation and pulmonary hypertension but the heart anomalies were not suspected early, despite regular specialized care.[13] This is an example of the diagnostic challenges and late identification of an ASDs; however, while six or more decades ago the early diagnosis of congenital heart diseases (CHDs) were scarce, the detection of these anomalies is currently done even *in utero*.[15]

Anatomical types of ASDs include ostium secundum (approximately 75%), ostium primum (15%), sinus venosus (5–10%), and coronary sinus (<1%) due to a hole in the wall between the left atrium (LA) and the coronary sinus with interatrial communication.[9,14] The spontaneous closures of ASDs with small diameters are more frequently described before the age of 5 years. The sinus venosus and ostium primum types do not have spontaneous closure, while the ostium secundum type measuring 3.0 to 8.0 mm may close in 12% to 82% of cases.[2] Late presentations of ASD may occur due to the insidious development of right ventricular remodeling, with enlargement of the right heart chambers; patients may present with symptoms related to right or global myocardial dysfunction, arrhythmias, or thromboembolic events.[1,2,5,12] Among 1,877 adults with congenital heart disease followed up for 5 years in the European Union, 896 were diagnosed with ASD, with a prevalence of pulmonary arterial hypertension (PAH) between 12% and 34% for corrected and uncorrected defects, respectively. PAH was diagnosed when the sPAP was ≥40 mmHg, and Eisenmenger's syndrome was detected in 15 patients. The syndrome was more prevalent in patients with cyanosis (17% vs 3% in non-cyanotic).[4] The widespread echocardiogram (Echo) studies yielded an increased number of the early diagnoses and favorable outcomes of the ASDs.[16]

CASE PRESENTATION

A 62-year-old female patient was admitted to the cardiology service in June 2020 complaining of palpitations associated with back pain which started one day ago. The electrocardiogram performed previously in another service, on the same day, revealed a supraventricular tachycardia. Her previous history did not identify arterial hypertension, diabetes mellitus, or coronary artery disease, but she had an arrhythmia without a clarified cause for 3 years. On physical examination, she was tachycardic (heart rate [HR]: 171 bpm), hypotensive (blood pressure [BP]: 90×60 mmHg), with a saturation of 92%, irregular heart rhythm, without murmurs, and unremarkable lung and abdominal examinations. There was no edema, fever, or cyanosis, and the pulses were palpable and symmetrical. She was admitted with the diagnoses of stable tachycardia and narrow QRS, and incessant atrial tachycardia because the reversal after adenosine, metoprolol, and amiodarone was not successful. Sinus node dysfunction was suspected and the investigation continued. Transthoracic echocardiography TTEcho (fig. 1) showed the ejection fraction: 65% (normal: 55–70%), LA volume: 70 mL/m^2 (normal: 34 mL/m^2), LA volume index (VI): 43.9 mL/m^2 (normal: ≤39.0 mL/m^2), right atrium volume index (RA VI): 125.4 mL/m^2 (normal: ≤41.0 mL/m^2), DDVE: 4 mm (normal: 3.8–5.4 mm), DSVE: 2.6 mm (normal: 2.3–3.6 mm); and yielded the diagnoses of moderate LA increase, marked tricuspid regurgitation, arterial pulmonary hypertension (48 mmHg), mild pericardial effusion, right ventricular (RV) systolic dysfunction (RV systolic excursion velocity: 8 cm/s and tricuspid annular plane systolic excursion: 1.45 mm), enlarged right chambers, and a "D shape" interventricular septum paradoxical movement towards the left ventricle. Computed tomography (CT) showed no filling defects in pulmonary arterial branches indicating thromboembolism; the diameters of the pulmonary arteries were increased (right: 2.9 cm and left: 2.8 cm) due to hypertension, and pericardial fluid (fig. 2). During the investigation, the hypothesis of Ebstein's anomaly was raised and the cardiac magnetic resonance with contrast showed an accentuated atrial dilatation, significant increase in the RV (223 mL), preserved global and segmental function of both ventricles (right ventricular ejection fraction [RVEF]: 54% and left ventricular ejection fraction [LVEF]: 69%). A tricuspid regurgitant jet due to failure in the coaptation of the leaflets was visualized, with a topical tricuspid valve insertion plane, which discarded the hypothesis of Ebstein's anomaly. In addition, there was an image of late myocardial enhancement, consistent with the absence of myocardial fibrosis or necrosis. During hospitalization, the patient had heart rate control only with a maximum dose of beta-blocker associated with amiodarone. The electrocardiogram showed ectopic atrial rhythm with a heart rate of 75 bpm, after the clinical compensation. The transesophageal Echo with Doppler (fig. 3) was the main tool for diagnosis, revealing this ASD of the sinus venosus type, with 2.4 cm in major diameter, and the predominant flux from left to right, with the Qp/Qs: 5.3; in addition, the diagnosis of accentuated tricuspid regurgitation secondary to the ASD was confirmed. After improvement of her general condition, the patient remained under ambulatory control.

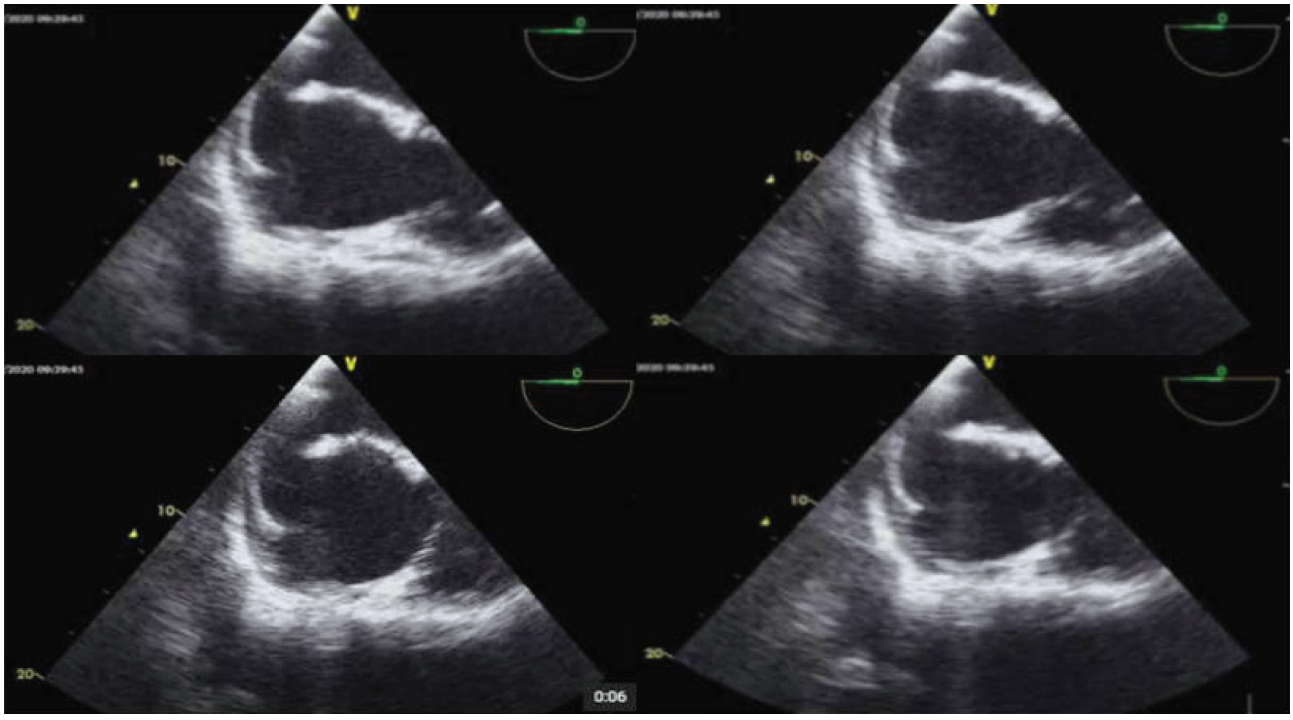

Figure 1. Transthoracic echocardiography revealed dilation of right chambers, right ventricular dysfunction, and paradoxical movement of the interventricular septum with bulging to the left ventricle, tricuspid regurgitation, and pulmonary hypertension.

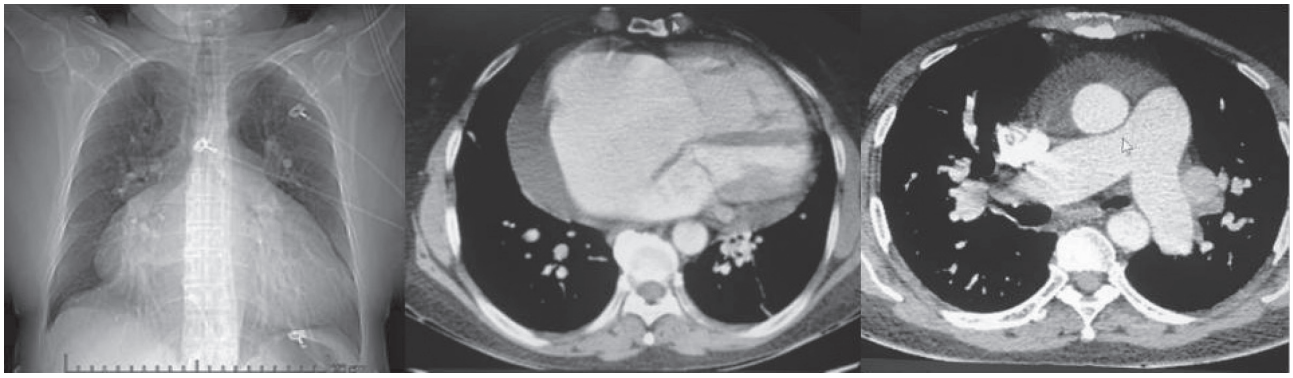

Figure 2. Chest computed tomography (CT) showed dilated pulmonary arteries, enlarged right chambers, pericardial effusion and absence of thromboembolism.

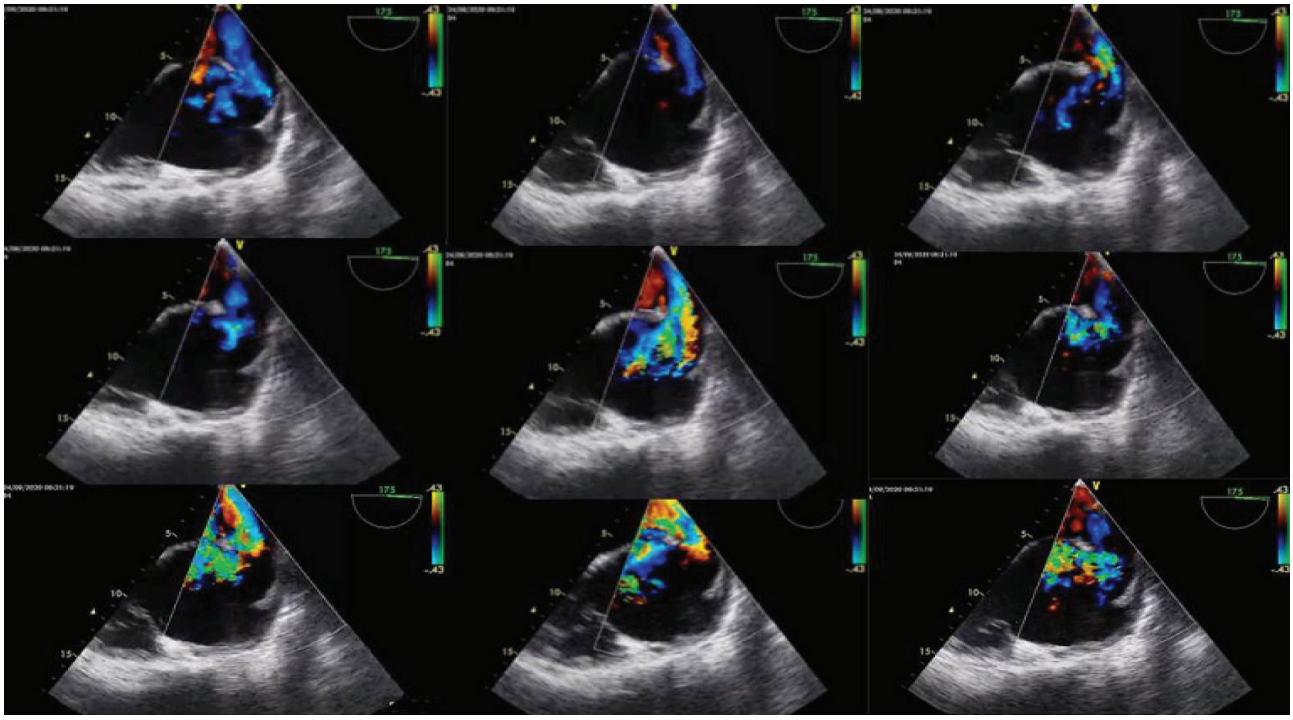

Figure 3. Transesophageal echocardiography confirmed the ASD of the sinus venosus type, the predominant flux from left to right, and accentuated tricuspid regurgitation.

DISCUSSION

The 62-year-old woman here reported had the confirmed diagnosis of sinus venosus type of ASD, manifested by palpitations due to supraventricular tachycardia. Although younger people may have no symptoms of ASD, the stress test may show below-normal ability.[3,5] With advancing age, overt symptoms of fatigue, exercise intolerance, dyspnea, palpitation, and manifestations of heart failure become prevalent and the survival rate is reduced. Arrhythmias are the major cause of hospitalization in adults with congenital heart disease and are also responsible for increased morbidity and mortality in these patients, observed in 21% of adults over 40 years of age, with increasing frequency over time.[1,2,9] The most common emergencies in these patients are arrhythmias, infections, heart failure, cerebral ischemia, or aortic root changes; supraventricular arrhythmias, such as atrial fibrillation, flutter, and tachycardia, are more frequent than ventricular arrhythmias.[1] The onset of ischemic heart disease and other comorbidities associated with decreased left ventricular compliance (arterial hypertension, aortic valve stenosis, and aging)

lead to increased left-to-right shunt through the defect, which further exacerbates symptoms and leads to clinical deterioration.[1,14] As a whole, the risk for the development of severe PAH in patients with ASD is less than 10%, but it is higher (approximately 14%) in cases of sinus venosus defect.[12] Patients with this defect who have very dilated pulmonary arteries, Echo images of abnormal hemodynamics, and peripheral oxygen saturation less than 93% on exertion, should be considered at risk for postoperative complications with poor outcomes.[12] Based on the natural history, the mortality rates of ASDs is up to 0.7% per annum during the first two decades of life, and rise in the next decades; 25% of patients die before the age of 27 years, 50% by 36 years, 75% by 50 years, and 90% by 60 years.[2]

The diameter of defects is classified as small (<3 mm), moderate (3 mm to 8 mm), and large (>8 mm), and is considered the main factor for spontaneous closure, and exceeding rare is the spontaneous closure of very large ASDs.[19] Mechanisms of closure are not well clear, including local shear stress and disordered blood flow dynamics, adaptive migration of endothelial cells, myocardial regeneration, fibroblast migration, and extracellular matrix deposition, as structural support for the long-term closures.[19] The largest defects with significant systemic to pulmonary shunts and oxygen desaturation require intervention. If indicated, surgical closures should be done before 25 years of age, when are more effective, safer, and related to normal life expectancy.[5] The closure of the ASD after the age of 40 years can reduce the morbidity and mortality in comparison with sole medical treatment,[11] but episodes of atrial tachyarrhythmia (mainly flutter or atrial fibrillation) remain high after the closures done in adulthood. Risk factors include atrial arrhythmia before the closure procedure and the age at closure over 40 years.[6,15] Percutaneous closures of small ASDs have favorable results and can be the first option for treatment of the ostium secundum type ≤6 mm.[3] Studies show improvement in symptoms and capacity to perform exercises, a decrease in the volumes of the RA and right ventricle, and control of pulmonary hypertension in most patients; but the benefits are lesser for the individuals over 60.[6,8,14–20] The symptomatic improvement and increase in the six-minute walking distance, in addition to less invasive procedures, justify the closure of the ASDs in elderly individuals.[7,8,10] A prospective cross-sectional study to evaluate the physical activity and self-efficacy levels in Greek young people with operated CHDs (13% ASDs) included 76 patients, 60.5% male aged 10 to 17 years and 78 controls, 51.3% male aged 10 to 14 years.[15] Authors concluded that the operated group has a sedentary lifestyle, which in adulthood can be associated health-related risks in comparison with the group of healthy controls. Besides, higher levels of self-efficacy propitiated higher levels of physical activity.[15]

The Echo is easily performed and the assessment of ventricular function, both systolic and diastolic, can be accurate in most CHD, to evaluate the pathophysiology of the lesions and possible implications. Transthoracic Echo has more limitations in adults, but transesophageal Echo can frequently clarify the most complex pathologies, in addition to being useful for the evaluation of elderly people with a poor echocardiographic window. Echo evaluations can disclose the exact anatomic location, size, and degree of hemodynamic repercussion (Qp/Qs) of the defects,[1,3,14,20] and associated anomalies or complications like PAH. In significant volumetric overloads of the right ventricle, there is a paradoxical movement of the interventricular septum. Magnetic resonance imaging (MRI) is also utilized in adults with CHD.

In conclusion, the aim of this report on the sinus venosus type of ASD in an adult is to highlight the diagnostic challenges and the unsuspected evolution of a congenital defect by decades, which could have decreased the expectancy of life without its early closure.

ΠΕΡΙΛΗΨΗ

Γυναίκα 62 ετών με μεσοκολπικό έλλειμμα τύπου φλεβοκόλπου

G.M. AQUINO,[1] S.B. COBRA,[1] V.M. DOS SANTOS,[2] O.L.R. ALMEIDA[1]

[1]Institute Hospital of Base of Federal District, Brasília-DF, [2]Armed Forces Hospital and Catholic University, Brasília-DF, Βραζιλία

Αρχεία Ελληνικής Ιατρικής 2023, 40(3):418–422

Τα ελλείμματα του κολπικού διαφράγματος είναι από τις πιο κοινές συγγενείς καρδιακές ανωμαλίες που διαγιγνώσκονται μεταξύ των ενηλίκων, μπορεί να επηρεάσουν περίπου το 25% των παιδιών και χαρακτηρίζονται από μεσοκολπική επικοινωνία η οποία συχνά επιμένει για χρόνια παραμένοντας μη ανιχνεύσιμη. Το έλλειμμα του φλεβοκόλπου αφορά στο 5–10% των διαφόρων ανατομικών τύπων. Η αυτόματη σύγκλειση των ελλειμμάτων μικρής διαμέτρου συμβαί-

νει συνήθως πριν από την ηλικία των 5 ετών, αλλά οι τύποι του φλεβοκόλπου και του ostium primum δεν κλείνουν με αυτόν τον τρόπο. Ασθενείς με διευρυμένες δεξιές καρδιακές κοιλότητες μπορεί να εκδηλώσουν κλινικά συμπτώματα δεξιάς ή ολικής καρδιακής δυσλειτουργίας, αρρυθμίες, θρομβοεμβολικά επεισόδια και πνευμονική αρτηριακή υπέρταση (12% στα διορθωμένα ελαττώματα και 34% στα μη διορθωμένα). Η αξιολόγηση με υπερηχοκαρδιογράφημα αυξάνει την πιθανότητα έγκαιρης διάγνωσης. Περιγράφεται η περίπτωση μιας γυναίκας 62 ετών με καθυστερημένη διάγνωση ελλείμματος τύπου φλεβοκόλπου με στόχο την ενίσχυση του δείκτη υποψίας των εργαζομένων στην πρωτοβάθμια υγεία για τη συγκεκριμένη πάθηση.

Λέξεις ευρετηρίου: Αρρυθμία, Κολπικό διαφραγματικό ελάττωμα, Υπερηχοκαρδιογραφία

References

1. ATTIE F, ROSAS M, GRANADOS N, ZABAL C, BUENDÍA A, CALDERÓN J. Surgical treatment for secundum atrial septal defects in patients >40 years old. A randomized clinical trial. *J Am Coll Cardiol* 2001, 38:2035–2042
2. CAMPBELL M. Natural history of atrial septal defect. *Br Heart J* 1970, 32:820–826
3. CHAMIÉ F, CHAMIÉ D. Percutaneous closure of small ostium secundum atrial septal defects. *Rev Bras Cardiol Invasiva* 2014, 22:264–270
4. ENGELFRIET PM, DUFFELS MGJ, MÖLLER T, BOERSMA E, TIJSSEN JGP, THAULOW E ET AL. Pulmonary arterial hypertension in adults born with a heart septal defect: The Euro Heart Survey on adult congenital heart disease. *Heart* 2007, 93:682–687
5. GEVA T, MARTINS JD, WALD RM. Atrial septal defects. *Lancet* 2014, 383:1921–1932
6. GIARDINI A, DONTI A, FORMIGARI R, SPECCHIA S, PRANDSTRALLER D, BRONZETTI G ET AL. Determinants of cardiopulmonary functional improvement after transcatheter atrial septal defect closure in asymptomatic adults. *J Am Coll Cardiol* 2004, 43:1886–1891
7. HANNINEN M, KMET A, TAYLOR DA, ROSS DB, REBEYKA I, VONDER MUHLL IF. Atrial septal defect closure in the elderly is associated with excellent quality of life, functional improvement, and ventricular remodelling. *Can J Cardiol* 2011, 27:698–704
8. HUMENBERGER M, ROSENHEK R, GABRIEL H, RADAR F, HEGER M, KLAAR U ET AL. Benefit of atrial septal defect closure in adults: Impact of age. *Eur Heart J* 2011, 32:553–560
9. KARIMZADEH S, SOLOGASHVILI T, MYERS PO, HACHULLA AL, LADOR F, BOUCHARDY J ET AL. Sinus venosus and unroofed coronary sinus defects: Two cases diagnosed and treated in adulthood. *Cardiovasc Med* 2019, 22:w02074
10. KHAN AA, TAN JL, LI W, DIMOPOULOS K, SPENCE MS, CHOW P ET AL. The impact of transcatheter atrial septal defect closure in the older population: A prospective study. *JACC Cardiovasc Interv* 2010, 3:276–281
11. KONSTANTINIDES S, GEIBEL A, OLSCHEWSKI M, GÖRNANDT L, ROSKAMM H, SPILLNER G ET AL. A comparison of surgical and medical therapy for atrial septal defect in adults. *N Engl J Med* 1995, 333:469–473
12. LOPES AA, MESQUITA SMF. Atrial septal defect in adults: Does repair always mean cure? *Arq Bras Cardiol* 2014, 103:446–448
13. MANETSKA O, TARCHENKO I. Chiari network in patient with interatrial septal defect and pulmonary hypertension. *Galician Med J* 2020, 27:E202027
14. MENILLO AM, LEE LS, PEARSON-SHAVER AL. Atrial septal defect. In: *StatPearls* [Internet]. StatPearls Publishin, Treasure Island, FL, 2022
15. MOSCHOVI D, KAPETANAKIS EI, SFYRIDIS PG, RAMMOS S, MAVRIKAKI E. Physical activity levels and self-efficacy of Greek children with congenital heart disease compared to their healthy peers. *Hellenic J Cardiol* 2020, 61:180–186
16. NAYOR M, MARON BA. Contemporary approach to paradoxical embolism. *Circulation* 2014, 129:1892–1897
17. SCHUSSLER JM, ANWAR A, PHILLIPS SD, ROBERTS BJ, VALLABHAN RC, GRAYBURN PA. Effect on right ventricular volume of percutaneous Amplatzer closure of atrial septal defect in adults. *Am J Cardiol* 2005, 95:993–995
18. VELDTMAN GR, RAZACK V, SIU S, EL-HAJJ H, WALKER F, WEBB GD ET AL. Right ventricular form and function after percutaneous atrial septal defect device closure. *J Am Coll Cardiol* 2001, 37:2108–2113
19. WANG SY, WELCH TD, ELFENBEIN A, KAPLAN AV. Spontaneous closure of a secundum atrial septal defect. *Methodist Debakey Cardiovasc J* 2018, 14:60–62
20. YALONETSKY S, LORBER A. Comparative changes of pulmonary artery pressure values and tricuspid valve regurgitation following transcatheter atrial septal defect closure in adults and the elderly. *Congenit Heart Dis* 2009, 4:17–20

Corresponding author:

V. dos Santos, Armed Forces Hospital, Estrada do Contorno do Bosque, Cruzeiro Novo, 70.658-900, Brasília-DF, Brazil
e-mail: vitorinomodesto@gmail.com;
https://orcid.org/0000-0002-7033-6074