American Journal of Case Reports

e-ISSN 1941-5923
© Am J Case Rep, 2026; 27: e953540
DOI: 10.12659/AJCR.953540

Received: 2026.03.24
Accepted: 2026.07.02
Available online: 2026.07.31
Published: 2026.XX.XX

Eisenmenger Syndrome After Delayed Atrial Septal Defect Closure at High Altitude: A Case Report

Authors' Contribution:
Study Design A
Data Collection B
Statistical Analysis C
Data Interpretation D
Manuscript Preparation E
Literature Search F
Funds Collection G

ABCDEF 1 Angelica Guaman-Gavilema
BEF 1 Domenica Villacres-Alarcon
CDEF 1 Andres Arteaga-Arellano
DEF 1 Roberto Arregui-Costales
CDEF 2,3 Jordan Llerena-Velastegui
DEF 2,3 Ruth Jimbo-Sotomayor
G 2,3 Xavier Sanchez

1 Hospital Axxis, Quito, Ecuador
2 Pontificia Universidad Católica del Ecuador, Quito, Ecuador
3 Centro de Investigación para la Salud en América Latina (CISeAL), Quito, Ecuador

Corresponding Author: Ruth Jimbo-Sotomayor, Pontificia Universidad Católica del Ecuador, Avenida 12 de Octubre 1076 y Vicente Ramón Roca, 170525, Quito, Ecuador, Phone: +593 99 500 5177, e-mail: rejimbo@puce.edu.ec

Financial support: None declared
Conflict of interest: None declared

Patient: Female, 46-year-old
Final Diagnosis: Eisenmenger syndrome following delayed surgical repair of atrial septal defect
Symptoms: Chest pain • cyanosis • dyspnea • fatigue • orthopnea
Clinical Procedure: —
Specialty: Cardiology

Objective: Congenital defects / diseases

Background: Eisenmenger syndrome develops from untreated or late-repaired cardiac shunts, causing irreversible pulmonary-vascular remodeling and systemic symptoms. Although timely repair has nearly eradicated Eisenmenger syndrome in high-income regions, delayed diagnosis in middle-income countries, especially when combined with chronic high-altitude hypobaric hypoxia, such as at approximately 2800 m (9186 ft) in the Ecuadorian Andes, can accelerate pulmonary hypertension through hypoxic vasoconstriction and erythrocytosis. We report a case of Eisenmenger syndrome 2 decades after surgical atrial septal defect closure.

Case Report: A 46-year-old woman living at high altitude presented with severe cyanosis and heart failure 22 years after atrial septal defect closure. She had marked hypoxemia and secondary erythrocytosis. Echocardiography showed suprasystemic pulmonary artery pressure, severe right-chamber dilation, and reduced right ventricular systolic function. Right-heart catheterization confirmed Eisenmenger physiology with severe pulmonary-vascular resistance and low cardiac index. Chest computed tomography showed marked pulmonary artery enlargement without thromboembolic disease. Multidisciplinary management included bosentan 125 mg twice daily, tadalafil 10 mg once daily, nocturnal oxygen, diuretics, and anticoagulation, resulting in symptomatic stabilization and discharge on day 4. Transplant evaluation was initiated.

Conclusions: Delayed atrial septal defect repair at high altitude can lead to irreversible Eisenmenger syndrome, for which current therapy is palliative. Early defect recognition and closure, congenital heart screening, and access to pulmonary vasodilators and transplant programs are imperative in resource-constrained mountainous regions.

Keywords: Atrial Septum • Eisenmenger Complex • Hypertension, Pulmonary • Hypoxia

Full-text PDF: https://www.amjcaserep.com/abstract/index/idArt/953540

Indexed in: [PMC] [PubMed] [Emerging Sources Citation Index (ESCI)] [Web of Science by Clarivate]

Introduction

Eisenmenger syndrome is a severe complication of untreated or late-repaired congenital heart disease, characterized by irreversible pulmonary arterial hypertension (PAH), reversal of longstanding left-to-right shunts, and systemic cyanosis [1]. Progressive pulmonary-vascular remodeling ultimately leads to chronic hypoxemia and multiorgan complications, including secondary erythrocytosis, arrhythmias, coagulopathy, and heart failure [1,2]. Although early repair of significant shunts during childhood has markedly reduced the incidence of Eisenmenger syndrome in high-income countries, the condition remains clinically relevant in low- and middle-income regions where congenital defects are often diagnosed or corrected later than recommended [1,2]. This report describes a 46-year-old woman residing at approximately 2800 m (9186 ft) above sea level in the Ecuadorian Andes who developed Eisenmenger physiology 2 decades after late surgical atrial septal defect closure, highlighting the consequences of delayed intervention and the additional hemodynamic burden imposed by chronic high-altitude hypoxemia.

Congenital heart disease-associated PAH represents an important subgroup of Group 1 pulmonary hypertension and is associated with worse outcomes than several other PAH etiologies [1,2]. In Latin America, congenital heart diseases account for approximately 35% to 50% of PAH cases reported in regional registries [3]. In Ecuador, limited epidemiological data and restricted access to specialized cardiopulmonary care may contribute to delayed presentation with advanced disease. Eisenmenger syndrome commonly develops during the third or fourth decade of life in patients with unrepaired or residual shunts. In the present case, atrial septal defect repair was performed at 24 years of age, beyond the period during which irreversible pulmonary-vascular remodeling may already be established. Current guidelines recommend early closure of significant septal defects and consider shunt closure contraindicated once Eisenmenger syndrome has developed because removal of the "pop-off" shunt may precipitate acute right-heart failure [4]. Accordingly, this case underscores the importance of timely congenital heart disease detection and intervention to prevent progression to irreversible pulmonary-vascular disease.

Management of established Eisenmenger syndrome is primarily supportive because pulmonary-vascular changes are usually irreversible. Advances in PAH-targeted therapy have improved functional status and exercise capacity, particularly with endothelin receptor antagonists and phosphodiesterase-5 inhibitors [1]. The BREATHE-5 trial demonstrated that bosentan improved hemodynamics and 6-minute walk distance in Eisenmenger syndrome patients without worsening systemic oxygen saturation [5]. Nevertheless, long-term prognosis remains poor, with contemporary registries reporting a 5-year mortality of approximately 25% to 30%, especially among patients with severe ventricular dysfunction or profound hypoxemia [1,6]. Heart-lung transplantation remains the only definitive treatment, but is rarely feasible because of donor scarcity and limited access to advanced transplant programs. These challenges are further amplified in resource-limited settings and may be compounded by chronic hypobaric hypoxemia in patients living at high altitude. At this altitude, reduced barometric pressure lowers inspired and alveolar oxygen tension; in a susceptible pulmonary-vascular bed, persistent hypoxic pulmonary vasoconstriction and erythrocytosis can further increase pulmonary arterial pressure and right ventricular afterload.

Overall, this case emphasizes that Eisenmenger syndrome remains an important clinical entity in adults with late-treated congenital heart disease and illustrates the complex interaction between delayed repair, pulmonary-vascular disease, and chronic high-altitude exposure. It also highlights the need for earlier diagnosis of congenital heart defects and improved access to specialized cardiopulmonary care in developing regions.

Case Report

A 46-year-old mestizo woman, with no prior medical history and long-term residence at approximately 2800 m (9186 ft) above sea level in Quito, Ecuador, in the Ecuadorian Andes, first experienced exertional dyspnea and fatigue at 17 years of age. This high-altitude exposure was clinically relevant because chronic hypobaric hypoxia may amplify pulmonary vasoconstriction and secondary erythrocytosis in patients with pulmonary-vascular disease. A transthoracic echocardiogram at that time demonstrated a non-restrictive atrial septal defect, yet the lesion remained unrepaired. Late surgical closure was finally undertaken at 24 years of age without peri-operative complications; however, pulmonary-vascular resistance had already become irreversibly elevated, and over the ensuing 2 decades she developed progressive cyanosis and exertional limitation, culminating in a diagnosis of severe PAH in 2022.

Right-heart catheterization in 2024 revealed suprasystemic pulmonary artery pressures (systolic 106 mm Hg), a pulmonary-vascular resistance of 23 Wood units, and a cardiac index of 1.48 L min^{-1} m^{-2}, confirming advanced Eisenmenger physiology. Additionally, baseline spirometry and hemoglobin-adjusted diffusion capacity were within normal limits, and a 6-minute walk distance of 456 m (68% of predicted) underscored significant though submaximal functional reserve.

Long-term oxygen supplementation was initiated at 37 years of age together with sildenafil 25 mg twice daily, furosemide 40

Indexed in: [PMC] [PubMed] [Emerging Sources Citation Index (ESCI)] [Web of Science by Clarivate]

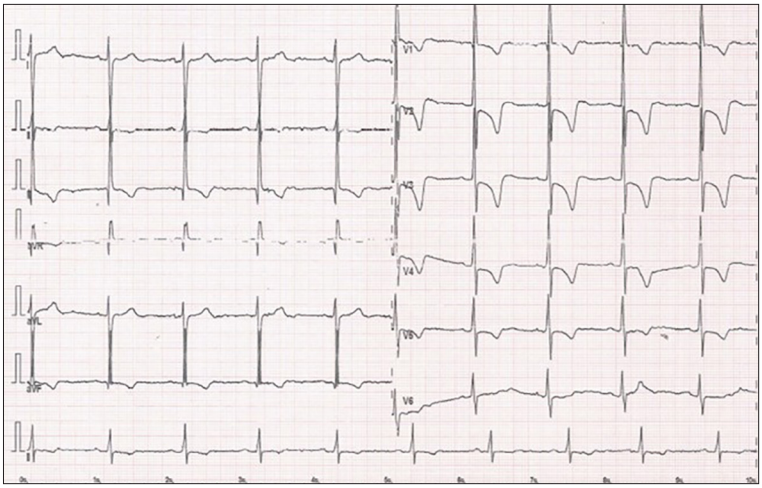

Figure 1. Resting 12-lead electrocardiogram (ECG; 25 mm/s, 10 mm/mV) showing sinus bradycardia (58 beats per minute), PR interval of 208 ms, QRS complex duration of 94 ms with rsR' pattern in V1-V3, right-axis deviation (+142°), R wave in V1, deep S wave in lead I/aVL, a peaked P wave of 132 ms in lead II, QT interval and heart-rate-corrected QT interval (QT/QTc) of 444/435 ms, and ST-segment depression with T-wave inversion in V1-V5, indicating right atrial and right ventricular hypertrophy/overload in Eisenmenger syndrome.

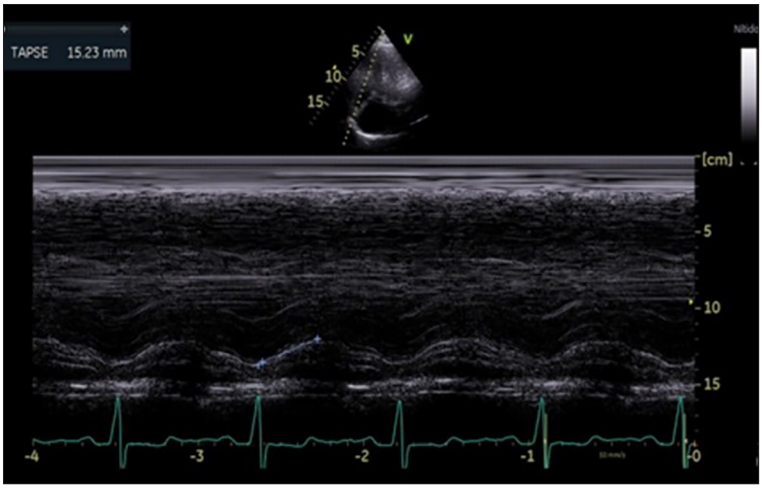

Figure 2. Apical 4-chamber transthoracic echocardiogram with M-mode across the lateral tricuspid annulus showing tricuspid annular plane systolic excursion (TAPSE) of 15.23 mm, indicating reduced right ventricular systolic function; a simultaneous electrocardiogram (ECG) delineates the systolic phase.

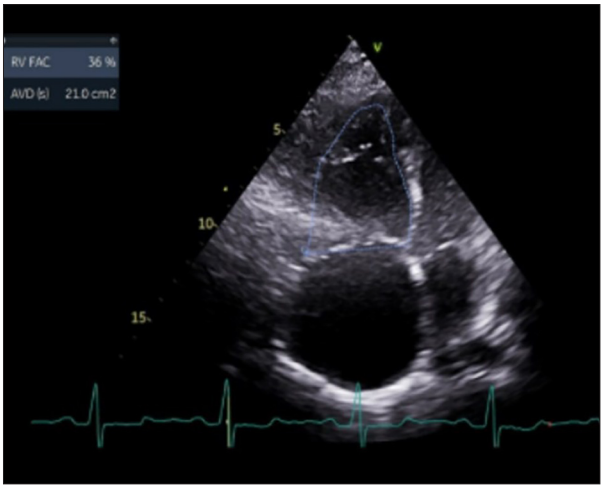

Figure 3. Apical 4-chamber transthoracic echocardiogram demonstrating right ventricular dilation with a fractional area change of 36%, confirming mild systolic impairment.

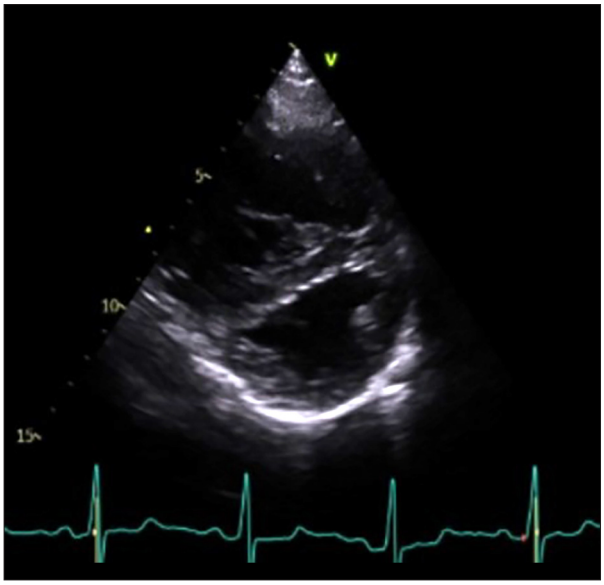

Figure 4. Parasternal short-axis transthoracic echocardiogram revealing leftward septal shift and a D-shaped left ventricle, reflecting marked right ventricular dilation.

Indexed in: [PMC] [PubMed] [Emerging Sources Citation Index (ESCI)] [Web of Science by Clarivate]

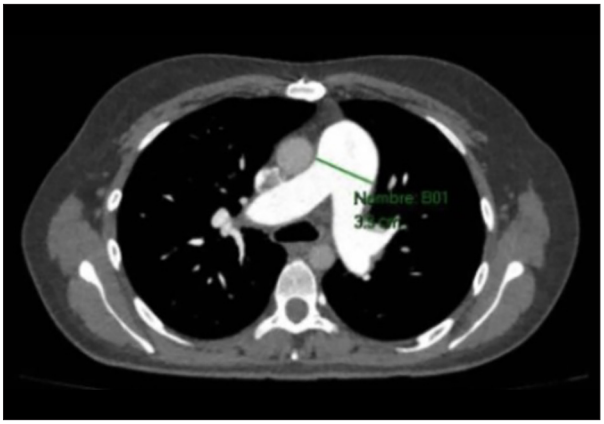

Figure 5. Axial chest computed tomography angiography shows dilatation of the main pulmonary artery to 3.9 cm, remodeling of the right ventricular outflow tract, and a right-to-left shunt, confirming advanced type 1 pulmonary hypertension (Eisenmenger syndrome).

mg daily, spironolactone 25 mg daily, and rivaroxaban 20 mg daily. Despite this regimen, her New York Heart Association (NYHA) functional class deteriorated inexorably, and at 46 years she presented to the emergency department with NYHA class IV symptoms—minimal-exertion dyspnea, orthopnea, oppressive chest pain, and profound hypoxemia (peripheral oxygen saturation [SpO_2] 77% on room air).

On admission she appeared markedly cyanotic, with perioral discoloration, grade I jugular venous distension, diminished peripheral pulses, digital clubbing, hepatomegaly, and a high-frequency decrescendo diastolic murmur consistent with a Graham-Steell murmur, accompanied by a palpable parasternal thrill. Furthermore, laboratory testing showed secondary erythrocytosis (hemoglobin 16.7 g/dL) and indirect hyperbilirubinemia; arterial blood gas analysis demonstrated respiratory alkalosis with hypoxemia (arterial oxygen partial pressure [PaO_2] 71 mm Hg, arterial carbon dioxide partial pressure [$PaCO_2$] 30 mm Hg, pH 7.44).

Electrocardiography showed sinus bradycardia with right-axis deviation and a right ventricular strain pattern (**Figure 1**). Transthoracic echocardiography demonstrated severe pulmonary hypertension with marked right-sided chamber dilatation, depressed right ventricular systolic function, and interventricular septal flattening producing a D-shaped left ventricle (**Figures 2-4**). Pulmonary computed tomography angiography (CTA) confirmed pulmonary valve regurgitation, enlargement of the pulmonary artery, and absence of obstructive chronic thromboembolic disease (**Figure 5**). Three-dimensional reconstruction highlighted massive dilatation of the pulmonary trunk and right-heart chambers (**Figure 6**).

Comprehensive pulmonary-function testing performed later the same year reaffirmed preserved ventilatory mechanics: forced vital capacity (FVC) 2.67 L (85% of predicted), forced expiratory volume in 1 second (FEV1) 2.08 L (81% of predicted), FEV1/FVC 0.78, and peak expiratory flow 118% of predicted. However, diffusing capacity remained near normal at 92% of predicted, although hemoglobin-adjusted diffusing capacity of the lung for carbon monoxide (DLCO) declined to 74% of predicted, reflecting impaired alveolar-capillary conductance. Moreover, repeat right-heart catheterization corroborated prior suprasystemic pressures (mean pulmonary artery pressure 60 mm Hg), extreme pulmonary vascular resistance of 23 Wood units, and a depressed cardiac index of 1.4 L min^{-1} m^{-2}. Finally, selective bilateral pulmonary angiography revealed webs and bands compatible with chronic thromboembolic sequelae superimposed on the Eisenmenger physiology, but no flow-limiting obstructions amenable to mechanical intervention.

A multidisciplinary PAH team commenced dual oral vasodilator therapy with bosentan 125 mg twice daily and tadalafil 10 mg once daily, intensified diuretic management with nifedipine 30 mg daily and spironolactone 25 mg daily, reduced rivaroxaban to 10 mg daily, and titrated continuous oxygen therapy from 4 L min^{-1} to 2 L min^{-1} as tolerated. Consequently, after 3 days of aggressive optimization, she achieved negative fluid balance, resolution of orthopnea, and improved resting saturation, permitting discharge with close outpatient follow-up and referral for consideration of heart-lung transplantation.

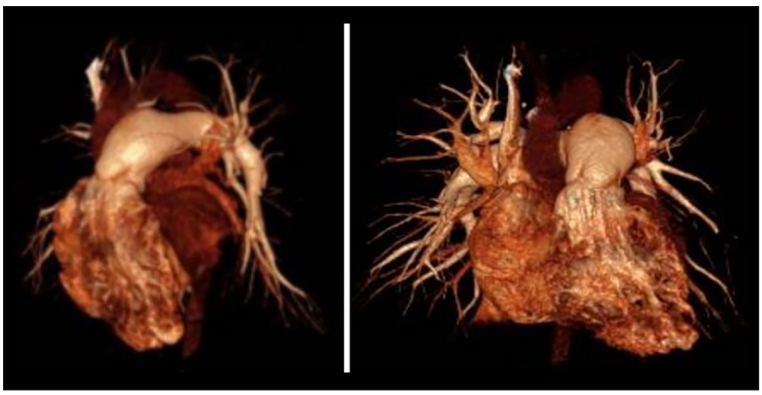

Figure 6. Three-dimensional (3D) volumetric cardiac computed tomography angiography, shown in the left anterior oblique and right anterior oblique views, depicts pronounced ectasia of the pulmonary trunk and branches with smooth-contoured right atrial enlargement, indicating chronic right-sided volume overload.

Indexed in: [PMC] [PubMed] [Emerging Sources Citation Index (ESCI)] [Web of Science by Clarivate]

Discussion

Eisenmenger syndrome develops when a longstanding left-to-right shunt reverses after irreversible pulmonary-vascular remodeling [1]. In our patient, late closure of an atrial septal defect at age 24 allowed pulmonary-vascular disease to progress, resulting in chronic hypoxemia, cyanosis, erythrocytosis, and right-sided heart failure. Her presentation with severe dyspnea, exercise intolerance, clubbing, and marked hypoxemia was therefore consistent with advanced Eisenmenger syndrome.

Current guidelines emphasize that congenital shunt closure is contraindicated once pulmonary-vascular resistance reaches ≥5 Wood units, because closure at that stage may precipitate acute right ventricular failure [4]. In our patient, right ventricular systolic pressure had already risen to 130 mm Hg before repair, making Eisenmenger physiology irreversible. Although targeted therapy was initiated promptly after diagnosis, her prognosis remains guarded. Contemporary series report 5-year survival of approximately 70% to 75% in Eisenmenger syndrome [6], and severe right ventricular dysfunction and advanced functional limitation further worsen expected outcomes.

High-altitude residence likely aggravated her condition. Chronic hypobaric hypoxia at approximately 2800 m (9186 ft) above sea level can intensify pulmonary vasoconstriction and erythrocytosis, thereby increasing pulmonary arterial pressure and right ventricular load. In this case, altitude was not considered the sole cause of Eisenmenger syndrome, but it likely acted as an additional hemodynamic stressor after delayed atrial septal defect repair by worsening hypoxemia and increasing right ventricular afterload. Her echocardiographic findings of marked right ventricular dilatation, reduced systolic function, and septal flattening, together with imaging evidence of a dilated pulmonary artery and right-to-left shunting, were compatible with advanced Eisenmenger syndrome [1].

Treatment of established Eisenmenger syndrome is primarily palliative and focuses on symptom control and reduction of pulmonary-vascular resistance [1]. Our patient was started on dual therapy with bosentan and tadalafil, consistent with current practice [1]. This approach is supported by the BREATHE-5 trial, in which bosentan improved exercise capacity and reduced pulmonary-vascular resistance without lowering systemic oxygen saturation [5]. Supportive measures, including diuretics, oxygen supplementation, and anticoagulation in selected patients, may also be considered, although their effect is limited in fixed shunts.

The present case also highlights the impact of limited access to specialized care in low-resource settings. Delayed referral, intermittent availability of advanced pulmonary vasodilators, and restricted access to adult congenital heart disease expertise likely contributed to the unfavorable course. Heart-lung transplantation remains the only potentially definitive option in end-stage disease, but it is rarely feasible because of donor and infrastructure constraints [7,8].

In conclusion, late atrial septal defect closure can culminate in Eisenmenger syndrome, particularly when diagnosis and intervention are delayed. In this patient, chronic high-altitude hypoxia and limited therapeutic resources compounded the disease burden. The case underscores the importance of early shunt detection and repair, systematic congenital heart screening, and equitable access to PAH therapies to prevent irreversible pulmonary-vascular remodeling and improve outcomes.

Conclusions

Eisenmenger syndrome after delayed atrial septal defect closure highlights the consequences of late diagnosis and chronic hypoxic exposure at high altitude, which can lead to irreversible pulmonary-vascular disease that treatment can only palliate. This altitude was clinically important because chronic hypobaric hypoxia can intensify pulmonary vasoconstriction, erythrocytosis, pulmonary arterial pressure, and right ventricular workload. Early defect detection, closure before pulmonary-vascular resistance reaches guideline thresholds, and timely initiation of pulmonary vasodilator therapy are essential to prevent this outcome. In addition, equitable access to specialized care and targeted therapies should be strengthened in settings where congenital heart disease is often diagnosed late.

Department and Institution Where Work Was Done

Hospital Axxis, Quito, Ecuador.

Data Availability

All data supporting the conclusions are contained within the article.

Patient Consent

Written informed consent for publication was obtained from the patient.

Artificial Intelligence (AI) Disclosure

An AI-based language model was used only for English language editing, formatting, style, readability, and revision wording support. It was not used to generate or analyze patient data, modify figures, interpret results, or draw scientific conclusions. All content was reviewed, verified, and approved by the authors, who take full responsibility for the manuscript.

Indexed in: [PMC] [PubMed] [Emerging Sources Citation Index (ESCI)] [Web of Science by Clarivate]

Declaration of Figures' Authenticity

All figures submitted have been created by the authors who confirm that the images are original with no duplication and have not been previously published in whole or in part.

References:

1. Arvanitaki A, Giannakoulas G, Baumgartner H, Lammers AE. Eisenmenger syndrome: Diagnosis, prognosis and clinical management. Heart. 2020;106:1638-45
2. Arvanitaki A, Gatzoulis MA, Opotowsky AR, et al. Eisenmenger syndrome: JACC state-of-the-art review. J Am Coll Cardiol. 2022;79:1183-98
3. Gómez Tejada R, Orozco-Levi M. Hipertensión pulmonar en Latinoamérica: Iniciativa de convergencia para las bases de datos RELAHP II y HAPred.co. Respirar. 2022;14(1):1-7 [in Spanish]
4. Baumgartner H, De Backer J, Babu-Narayan SV, et al. 2020 ESC Guidelines for the management of adult congenital heart disease. Eur Heart J. 2021;42:563-645
5. Galiè N, Beghetti M, Gatzoulis MA, et al. Bosentan therapy in patients with Eisenmenger syndrome: A multicenter, double-blind, randomized study. Circulation. 2006;114:48-54
6. Diller GP, Körten MA, Bauer UMM, et al. Current therapy and outcome of Eisenmenger syndrome: Data of the German National Register for congenital heart defects. Eur Heart J. 2016;37:1449-55
7. Clavé MM, Maeda NY, Castro CRP, et al. Factors influencing outcomes in patients with Eisenmenger syndrome: A nine-year follow-up study. Pulm Circ. 2017;7(3):635-42
8. Valverde AB, Soares JM, Viana KP, et al. Pulmonary arterial hypertension in Latin America: epidemiological data from local studies. BMC Pulm Med. 2018;18:202

Indexed in: [PMC] [PubMed] [Emerging Sources Citation Index (ESCI)] [Web of Science by Clarivate]