Case Report

# Interventional Management of a Rare Case of Complex Congenital Heart Disease in an Adult Patient: A Case Report

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## Abstract

*Congenital heart disease (CHD), which accounts for about one-third of all congenital birth defects and affects about 1% of all live births worldwide, has had a stable incidence rate and decreased mortality rate since 1990. Despite advances in fetal cardiac ultrasound examinations and routine pulse oximetry screening of newborns, a considerable proportion of patients with CHD may still be missed until adulthood, leading to major morbidity and mortality due to physical limitations and reduced quality of life.*

*We herein describe a 29-year-old woman who was mistaken as an inoperable case of CHD and referred to our center with palpitation and exertional dyspnea. During workups, she was diagnosed with severe pulmonary hypertension associated with patent ductus arteriosus (PDA), pre-ductal aortic coarctation, ventricular septal defect (VSD), and bicuspid aortic valve, all of which had been missed from infancy. After initial medical treatment for pulmonary hypertension, a simultaneous transcatheter approach was selected, whereby the PDA was closed with an occluder device, and the coarctation was repaired simultaneously with a self-expanding stent. Eight months later, her VSD was closed successfully via an interventional technique using a muscular VSD occluder device.*

*This case highlights the significance of adult CHD fellowship training. A cardiologist specialized in this field was able to properly diagnose and treat an adult with complex CHD, which had been overlooked since infancy. As a result, the patient experienced complete relief from her symptoms and was saved from developing Eisenmenger syndrome.*

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## Introduction

Congenital heart disease (CHD) accounts for about 28% of all congenital defects. CHD had a global prevalence of 0.8% to 1.2% and an incidence rate of 17.9/1000 in 2017, with the rate having remained stable since 1990.[1, 2] Although CHD is predominantly diagnosed through antenatal ultrasound assessments or neonatal pulse oximetry screening, a significant proportion of patients are missed during this early screening and are diagnosed with CHD after discharge from the hospital, during their childhood, or even adulthood.[2, 3] Delayed diagnosis of CHD is associated with major morbidity and mortality caused by physical limitations, which can reduce their quality of life.[4]

This case report presents a rare case of a 29-year-old woman with a late-onset presentation of ventricular septal defect (VSD), preductal aortic coarctation (CoA), and patent ductus arteriosus (PDA), accompanied by a bicuspid aortic valve and severe pulmonary hypertension.

## Case Report

A 29-year-old woman was admitted to our hospital with new-onset palpitations and classified as New York Heart Association (NYHA) functional class I. Her mother reported episodes of cyanosis during breastfeeding and palpitations in childhood, at which time she was diagnosed with CHD. Nonetheless, there was no available documentation regarding the specific type of CHD, and her parents did not pursue proper treatment. In adulthood, she was mistakenly considered an inoperable case of CHD and was referred to our center due to severe pulmonary hypertension.

On physical examination, the patient had relatively bounding pulses in the upper extremities with a rate of 70 beats per minute, a respiratory rate of 16 breaths per minute, a body temperature of 36.3 °C, a blood pressure reading of 180/100 mm Hg in the upper extremities and 130/90 mm Hg in the lower extremities, and an $O_2$ saturation level of 93% in the upper and lower extremities. On cardiac examination, an enhanced $S_2$ sound was auscultated in addition to a grade 4/6 holosystolic murmur with the maximum intensity in the pulmonic area. The rest of the examinations were unremarkable. A chest X-ray revealed cardiomegaly, pulmonary artery enlargement, and increased pulmonary vascular marking. Her laboratory findings were all unremarkable.

On echocardiography, a large subaortic VSD (1.5 cm) with a left-to-right shunt and a peak gradient (PG) of 30 mm Hg was seen. The high gradient (PG=30 mm Hg) systolic turbulent flow and a diastolic tail in the descending aorta favored CoA (Figure 1). Additionally, the continuous turbulent flow between the left pulmonary artery and the descending aorta with a PG of 40 mm Hg suggested a large PDA after the CoA. Other echocardiographic findings included mild left atrial enlargement, moderate to severe left ventricular enlargement with normal systolic function (ejection fraction =55%), no left ventricular hypertrophy, and a right ventricle of a top normal size with mild systolic dysfunction and moderate hypertrophy. Valvular involvements included mild-to-moderate mitral regurgitation, pulmonary insufficiency, and moderate tricuspid regurgitation (tricuspid regurgitation gradient =115 mm Hg). Moreover, a bicuspid aortic valve formed by the fusion of the left and right coronary cusps was observed. Our patient had severe pulmonary hypertension with a systolic pulmonary artery pressure of 120 mm Hg.

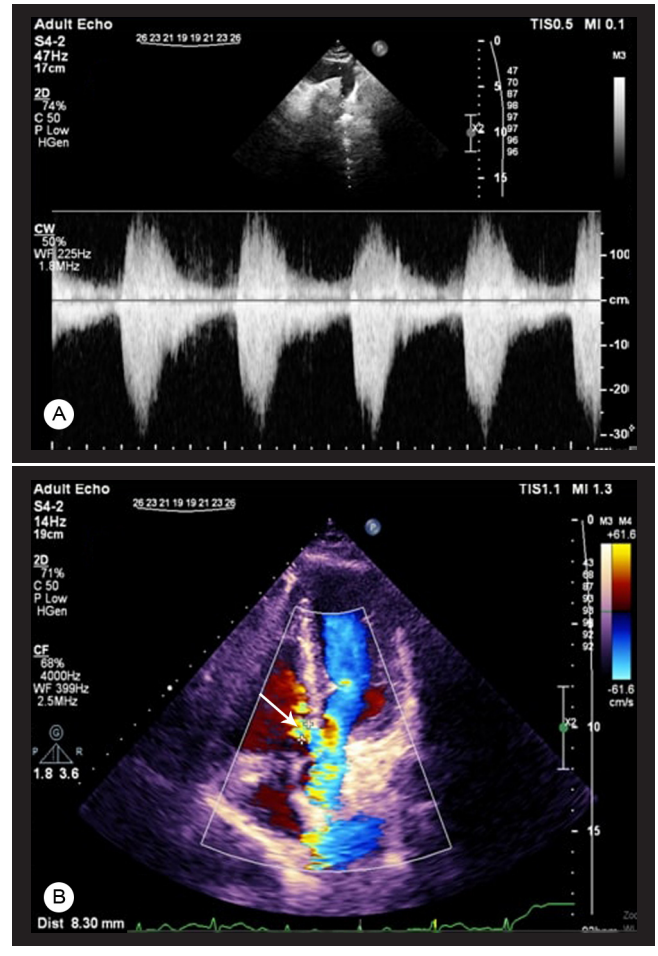


Figure 1. A) Transthoracic echocardiography in the suprasternal view demonstrates diastolic tail and gradient, favoring aortic coarctation. B) Transthoracic echocardiography in the 4-chamber shows a large subaortic ventricular septal defect (the white arrow).

The patient underwent cardiac catheterization, demonstrating a left-to-right shunt, a $Q_P/Q_s$ ratio above 1.5, a pulmonary vascular resistance index of less than 8, and a significant step-up between the main and left pulmonary arteries. Supplemental oxygen therapy led to an improvement in her condition, with $O_2$ saturation rising to 93% in both the ascending and descending aorta after the PDA. Pulmonary artery pressures were as follows: systolic, 120 mm Hg;

diastolic, 40 mm Hg; and mean, 70 mm Hg. The remaining results aligned with echocardiographic findings, confirming that the patient had not yet progressed to Eisenmenger syndrome.

The patient was, therefore, discharged with a prescription of sildenafil (50 mg TDS), macitentan (10 mg daily), and bisoprolol (2.5 mg BD) for 2.5 months to control her pulmonary hypertension before the repair of her congenital cardiac anomalies.

Three months later, she was readmitted to the hospital for simultaneous PDA device closure and CoA repair. Cardiac catheterization was performed through the right femoral access, and the PDA was wired. Successful closure of the PDA was achieved using an Occlutech PDA occluder measuring 12×15 mm. Concurrent coarctoplasty was also performed using a self-expanding Sinus-XL stent

measuring 22×60 mm with positive outcomes, resulting in no residual gradient. Postoperative echocardiography confirmed the appropriate positioning of the PDA occluder device with no compressive effect or residual PDA. The stent was also seen in the proper place with no remaining recoarctation. Figure 2 displays chest X-rays obtained before and after the procedure. The patient was discharged with a prescription of ASA (80 mg daily) and clopidogrel (75 mg daily) for 6 months, while the remaining medications were continued with the same instructions.

Eight months post-discharge, the patient was readmitted for VSD device closure. A large perimembranous VSD with a $Q_p/Q_s$ ratio of 1.7 was successfully repaired using a 16 mm Occlutech muscular VSD occluder via cardiac catheterization through the right femoral vein and artery access. Intraprocedural transesophageal echocardiography confirmed the proper deployment of the occluder

device, demonstrating good alignment with the interventricular septum. No residual shunt or clot formation on the device was observed. Figure 3 presents the primary and final images from cardiac catheterization. The patient was discharged with

a prescription of ASA (80 mg daily), clopidogrel (75 mg daily) for 3 months, sildenafil (50 mg TDS), macitentan (10 mg daily), and bisoprolol (2.5 mg BD). In the follow-up visit, 2 months after the final procedure, the patient reported relief from her symptoms of palpitations and dyspnea, and her vital signs were stable. An echocardiogram indicated improvement in her pulmonary hypertension (systolic pulmonary artery pressure = 45 mm Hg). Furthermore, the VSD and PDA occluder devices, as well as the aortic stent, were all appropriately positioned with no evidence of compression or residual defects.

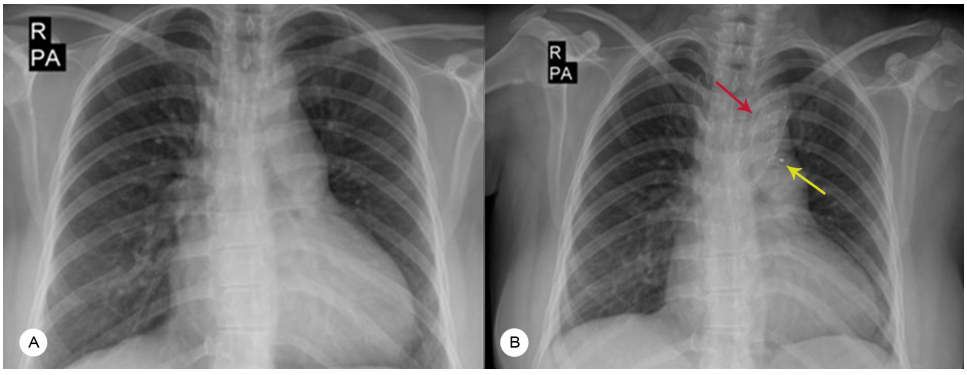


Figure 2. The images present the patient's chest X-rays A) before the procedures and B) after patent ductus arteriosus device closure (the yellow arrow) and aortic coarctation repair (the red arrow).

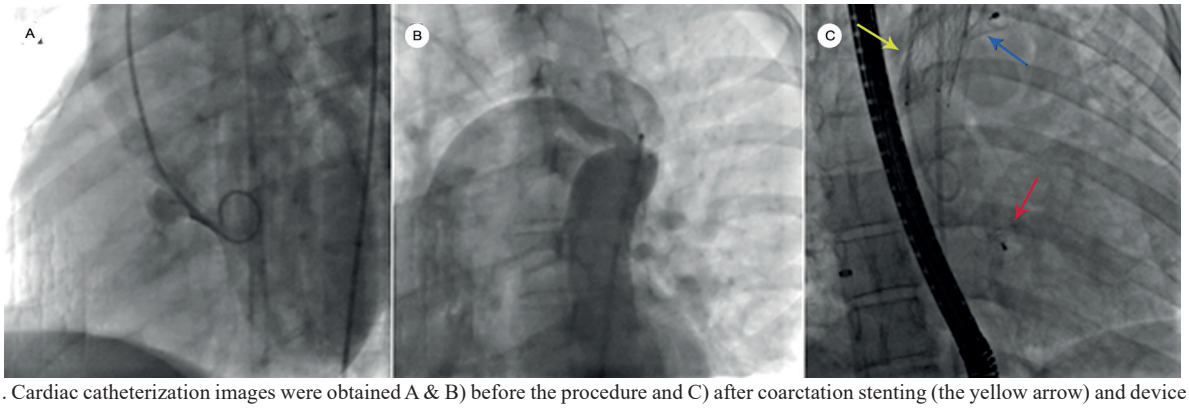


Figure 3. Cardiac catheterization images were obtained A & B) before the procedure and C) after coarctation stenting (the yellow arrow) and device closure of the patent ductus arteriosus (the blue arrow) and ventricular septal defect (the red arrow).

## Discussion

CHD, accounting for about one-third of all congenital birth defects, affects approximately 1 in every 100 live births worldwide.[2] The global incidence rate of CHD has remained stable since 1990, with the highest rates in developing African and Asian countries. There was also a 38.1% decline in CHD mortality rates, from 6.3/100000 in 1990 to 3.9/100000 in 2017, positively correlated with the regional sociodemographic index.[1]

Although advances in fetal cardiac ultrasound examinations and routine pulse oximetry screening of newborns have led to early detection of CHD, a major proportion of these patients may still be missed even until adulthood.[2, 3] The primary causes of delayed diagnosis of CHD include an inadequately trained healthcare system, which results in delayed diagnoses by physicians or midwives, subsequently leading to postponed referrals to tertiary care centers, as well as socioeconomic barriers.[4, 5] Delayed diagnosis of CHD is associated with a significant socioeconomic burden and major morbidity and mortality caused by failure to thrive, recurrent infections, poor nutritional status, arrhythmia, thromboembolism, cardiac arrest, pulmonary hypertension, and heart failure, which can adversely affect the long-term survival of these patients and cause a major restraint in children's daily activities and social development.[4, 6]

CoA is the fifth most common CHD, with an incidence rate of 3 to 4 in 10,000 live births and a male-to-female ratio of 2:1.[4, 5] Preductal CoA, typically located at the insertion of the ductus arteriosus and distal to the left subclavian artery, is usually diagnosed during infancy or early childhood and is typically associated with a large PDA and pulmonary hypertension.[7] Nevertheless, in our case, it was diagnosed in the third decade of life. The most common cardiac anomalies associated with CoA include bicuspid aortic valve (60%), aortic arch anomalies (18%), VSD (13%), and mitral valve abnormalities (8%).[8]

PDA, the second most common CHD with an incidence rate of approximately 1 in 2000 live births, refers to a term infant whose ductus arteriosus remains open after 3 months of life.[9] In patients with moderate or large PDAs, continuous left-to-right shunts can lead to progressive arterial changes in pulmonary arteries, including arteriolar medial hypertrophy, intimal proliferation, and obliteration of pulmonary arteries, eventually leading to pulmonary hypertension. When pulmonary vascular resistance exceeds systemic vascular resistance, the direction of ductal shunting reverses from right to left, leading to the development of Eisenmenger syndrome.[9, 10]

The combination of CoA and PDA can be treated through surgical intervention, which is recommended for infants and children, or through interventional techniques, which is a safe and effective approach for adolescents and adults, offering fewer risks and shorter hospitalization periods than thoracic surgery. Selecting the appropriate treatment option and determining whether to administer it simultaneously or sequentially depends on the patient's age, CoA anatomy, and PDA size. In cases of isthmic hypoplasia, CoA stenting is essential. If there is no dilatation in the aorta beyond the CoA, a single covered stent that excludes the PDA while relieving the CoA gradients is the preferred option. However, if dilation is present post-CoA, PDA closure should be accomplished using an occluder device, along with simultaneous stent angioplasty of the CoA. In cases of discrete membranous CoA with a proper-sized isthmus, the method of choice would be balloon aortoplasty and PDA device closure.[11]

The adult CHD fellowship is a unique and essential subspecialty of cardiology that provides comprehensive and multidisciplinary care to the growing population of patients with CHD. In addition to its capacity to diagnose and manage a variety of complex congenital cardiac defects and their associated complications, such as residual lesions from previous surgeries, arrhythmias, pulmonary hypertension, and heart failure, it also enhances our understanding of the natural history and outcomes of CHD. It can also effectively identify and diagnose adult patients with CHD who were missed during childhood, ensuring they receive appropriate treatment to prevent further complications.

In this case report, we described a 29-year-old woman presenting with palpitations and exertional dyspnea. She was referred to our center as an inoperable case of CHD due to severe pulmonary hypertension. During our workups, we detected PDA, preductal CoA, VSD, bicuspid aortic valve, and severe pulmonary hypertension, conditions that had been overlooked since infancy and childhood. These diagnoses were made at our center by a cardiologist specializing in adult CHD. After confirming the absence of Eisenmenger syndrome, we initiated medical treatment for pulmonary hypertension with sildenafil and macitentan. Subsequently, we chose a simultaneous transcatheter approach, during which the PDA was closed with an occluder device, and the CoA was repaired simultaneously with a self-expanding stent. Given the patient's large PDA and the increased likelihood of achieving complete closure of PDA flow through the use of a specific PDA occluder device along with stent coarctoplasty, this approach was chosen for the present case instead of utilizing a single covered stent. Following the successful closure of the PDA, elimination of the left-to-right shunt, and restoration of aortic patency, an increase in the $Q_p/Q_s$ ratio was observed. This change may be related to the hemodynamic alterations after the PDA closure and aortic coarctoplasty. Eight months later, the patient's VSD was successfully closed using an interventional technique with a muscular VSD occluder device, resulting in complete relief of her symptoms and improvement in her pulmonary hypertension.

## Conclusion

This case underscores the significance of adult CHD fellowship training, as it enabled a cardiologist specializing in this field to accurately diagnose a complex case of CHD in an adult who had been overlooked since infancy and early childhood and was referred to our center as an inoperable case. Following a thorough evaluation and confirmation of a left-to-right shunt, she received appropriate medical treatment for her severe pulmonary hypertension. Her defects were subsequently corrected through a 2-step interventional approach, ultimately leading to complete relief of her symptoms and preventing the progression to Eisenmenger syndrome.

### Ethics Approval and Consent to Participate

This study was conducted following the Declaration of Helsinki and received approval from the Ethics Committee of Rajaie Cardiovascular Medical and Research Center. Patient participation was secured through both written and verbal consent.

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