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Case Report
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Undiagnosed for Decades: A 48-Year-Old Woman with Primum Atrial Septal Defect, Common Atrium, and Secondary Polycythemia

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Abstract

Background: *Most congenital heart defects are diagnosed during childhood. However, some patients can reach adulthood without detection, particularly those with limited healthcare access. This case report describes a patient whose heart defect remained undiagnosed until age 48.*

Case Summary: *A 48-year-old woman from Jamaica presented to the emergency department with bilateral leg pain. She had been told as a child she had an "inoperable" heart problem and was not expected to survive past adolescence. In the emergency department, her oxygen saturation was severely low (70s-80s range). Laboratory studies demonstrated elevated hemoglobin (21 g/dL) and hematocrit (>60%). An echocardiogram revealed a large primum atrial septal defect with a common atrium, a cleft mitral valve causing severe regurgitation, and evidence of pulmonary hypertension.*

Outcome: *The patient was admitted to the intensive care unit for hypoxemia requiring supplemental oxygen and BiPAP. Cardiology consultation determined that surgical intervention was too high-risk given her advanced disease. While Columbia University Medical Center initially accepted transfer, it was subsequently denied due to lack of health insurance coverage. The patient remained in the ICU on supportive care per chart documentation.*

Conclusion: *Clinicians should maintain clinical suspicion for undiagnosed congenital heart disease in adults presenting with hypoxemia and secondary polycythemia, particularly in patients with limited healthcare access. This case further highlights how socioeconomic factors, including insurance status, can significantly impact patient outcomes.*

Keywords: *Atrial Septal Defect; Primum ASD; Common Atrium; Secondary Polycythemia; Eisenmenger Syndrome; Congenital Heart Disease; Healthcare Disparities.*

Introduction

Congenital heart disease (CHD) occurs in approximately 1% of live births worldwide [1]. While most cases are identified during childhood, some individuals with less severe defects—or those lacking adequate healthcare access—may reach adulthood without diagnosis [2]. Atrial septal defects (ASDs) are among the most common congenital cardiac anomalies. A primum ASD involves the lower portion of the atrial septum and is frequently associated with atrioventricular valve abnormalities, including cleft mitral valve leaflets [3]. Chronic left-to-right shunting can lead to pulmonary vascular remodeling, elevated pulmonary pressures, and eventual shunt reversal (Eisenmenger physiology), resulting in systemic hypoxemia and secondary erythrocytosis [4]. We present a case of a patient with a large primum ASD with common atrium who presented with bilateral leg pain, ultimately revealing severe hypoxemia and significant erythrocytosis.

Case Presentation

A 48-year-old woman of Jamaican origin presented to the emergency department (ED) with bilateral leg pain. She had immigrated to the United States in 2019 and had not sought medical care for approximately six years prior to this hospitalization. The patient provided informed consent for case publication, with identifying information removed to protect privacy.

According to chart documentation, the patient recalled being informed during childhood of a heart condition described as "inoperable," with a prognosis that she "would not live beyond her teenage years" (documented in cardiology consultation note). She reported a history of cardiac catheterization in Jamaica and believed two stents may have been placed, though details were unavailable as prior records could not be obtained. Additional medical history included rheumatic fever, meningitis, and four miscarriages. During this hospitalization, a new diagnosis of diabetes mellitus was established (HbA1c 7.6%). Surgical history included tubal ligation. Social history was negative for tobacco, alcohol, or illicit substance use per chart documentation.

The patient initially presented to an outpatient clinic with bilateral leg pain described as sharp, nocturnal, and sleep-disrupting. Pulse oximetry revealed oxygen saturation in the 70% range, prompting urgent ED referral. Upon further questioning, she endorsed progressive exertional dyspnea over several years, requiring rest after walking approximately three blocks. She denied chest pain, syncope, or palpitations. In the

ED, despite supplemental oxygen at 4 liters per minute via nasal cannula, oxygen saturation remained in the 80% range.

On initial examination, the patient was alert and oriented. Vital signs were: blood pressure 130/66 mmHg, heart rate 88 beats per minute and regular, respiratory rate 20 breaths per minute, and oxygen saturation 84% on supplemental oxygen. Cardiovascular examination demonstrated a displaced and diffuse point of maximal impulse. The second heart sound was loud and widely split. A holosystolic murmur, grade III/VI, was auscultated along the left sternal border with radiation to the apex, accompanied by a palpable thrill. Pulmonary auscultation was clear. Abdominal examination revealed hepatomegaly without tenderness. Digital clubbing was noted, though there was no peripheral edema or cyanosis. Neurological examination was non-focal.

Laboratory findings demonstrated significant erythrocytosis (hemoglobin 21 g/dL [N=12-16 g/dL], hematocrit >60% [N=36-48%]). White blood cell count was normal at 4.7 × 10^3/µL. Troponin was within normal limits, and D-dimer was <200 ng/mL. Chest radiography showed cardiomegaly with bilateral pulmonary nodular opacities. A CT angiogram of the chest excluded pulmonary embolism but revealed a dilated pulmonary trunk and branches, as well as a left-sided inferior vena cava with hemiazygos continuation to a left brachiocephalic vein—a congenital venous anomaly. Hepatosplenomegaly was also noted.

Figure 1 shows the echocardiogram, which demonstrated preserved left ventricular ejection fraction of 54.6% with mild concentric left ventricular hypertrophy. A large primum atrial septal defect with a common atrium was identified, along with septal dyskinesis and flattening consistent with right ventricular volume and probable pressure overload. A cleft anterior mitral valve leaflet with severe mitral regurgitation was observed. Estimated right ventricular systolic pressure was 24 mmHg, though this was likely underestimated. Findings were consistent with probable pulmonary hypertension.

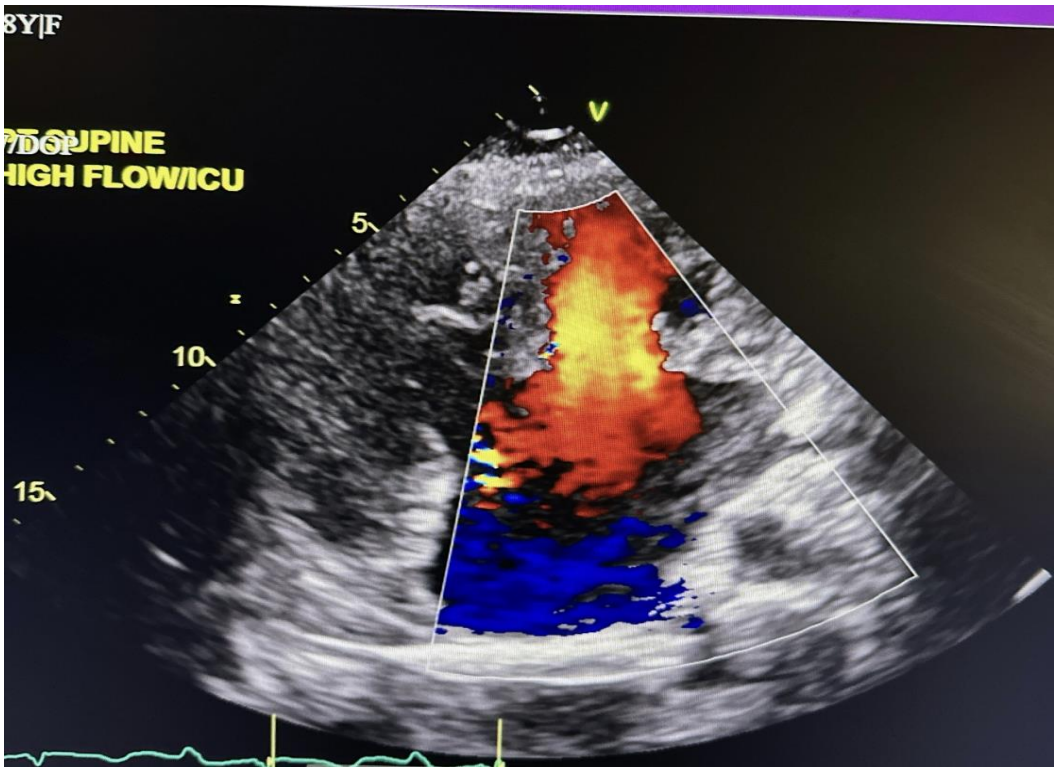

Figure 1: Transthoracic echocardiogram (apical four-chamber view) demonstrating a large primum atrial septal defect with near-complete absence of the interatrial septum (common atrium). Color Doppler revealed severe mitral regurgitation due to a cleft anterior mitral valve leaflet (not shown). RA = right atrium; LA = left atrium; RV = right ventricle; LV = left ventricle.

Given the patient's leg pain, lumbar spine CT was performed, revealing L4-L5 disc disease with neuroforaminal stenosis—likely the primary etiology of her lower extremity symptoms. Hematology consultation was obtained for evaluation of erythrocytosis. BCR-ABL testing was negative, ruling out polycythemia vera. Antiphospholipid antibody panel (anticardiolipin, lupus anticoagulant, anti-beta-2 glycoprotein) was negative, reducing concern for antiphospholipid syndrome as an etiology for her prior miscarriages.

Alternative etiologies for the patient's hypoxemia were considered, including pulmonary arteriovenous malformations (excluded by CT angiography), platypnea-orthodeoxia syndrome (not formally evaluated), primary pulmonary hypertension without intracardiac shunt (excluded by echocardiographic findings), and polycythemia vera (excluded by negative BCR-ABL testing).

The patient was admitted to the intensive care unit for close monitoring. She required supplemental oxygen via mask and intermittent BiPAP support. Medical management included aspirin 81 mg, atorvastatin 40 mg, enoxaparin 40 mg subcutaneous daily for thromboprophylaxis, and nitroglycerin patch. Insulin sliding scale was initiated for hyperglycemia (blood glucose up to 235 mg/dL). Phlebotomy was considered to reduce hematocrit below 53%; however, this intervention was

deferred due to concerns that acutely lowering blood viscosity might paradoxically impair oxygen delivery in the setting of cyanotic heart disease [5].

Cardiology consultation recommended right heart catheterization for accurate hemodynamic assessment. However, surgical repair was deemed prohibitively high-risk given the patient's advanced disease burden (large ASD, severe mitral regurgitation, long-standing hypoxemia). The decision was made to proceed with supportive care.

The patient remained hemodynamically stable but persistently hypoxemic throughout her hospitalization—a finding attributed to her chronic cardiopulmonary disease. No new infections or thromboembolic events occurred. Significantly, transfer to Columbia University Medical Center, a tertiary referral center specializing in adult congenital heart disease, was initially accepted but subsequently denied due to lack of health insurance coverage (documented in progress note, page 26). Alternative transfer to Newark Beth Israel Medical Center was considered. At the time of last chart review, the patient remained hospitalized in the ICU on supportive care.

Discussion

This case demonstrates a patient who survived to age 48 with an undiagnosed primum ASD and common atrium, despite being informed during childhood of a life-limiting cardiac condition. Literature review suggests that such defects are typically identified in childhood due to early murmurs or symptoms [3]. In this patient, limited healthcare access following immigration to the United States likely contributed to diagnostic delay.

In large ASDs, chronic left-to-right shunting increases pulmonary blood flow. Over decades, this volume overload induces pulmonary vascular remodeling—medial hypertrophy, intimal fibrosis, and plexiform lesions—leading to progressive pulmonary hypertension. When pulmonary pressures approach systemic levels, shunt reversal (right-to-left) occurs, resulting in cyanosis and secondary erythrocytosis [5]. The patient's erythrocytosis (hemoglobin 21 g/dL) reflects a physiological compensatory response to chronic hypoxemia, increasing oxygen-carrying capacity. However, this adaptation carries risks of hyperviscosity, thrombosis, and stroke [5].

The patient's history of four miscarriages is notable. Pregnancy imposes significant hemodynamic demands, and women with cyanotic CHD face elevated risks of maternal and fetal complications, including miscarriage, due to hypoxemia, erythrocytosis, and increased thrombotic risk [6]. Antiphospholipid antibody testing was negative, supporting the likelihood that her miscarriages were secondary to underlying cardiac disease.

Surgical intervention in patients with Eisenmenger physiology is controversial. Closure of an intracardiac defect after development of severe pulmonary hypertension can precipitate right ventricular failure, as the right ventricle has become accustomed to pumping against elevated pulmonary vascular resistance [7]. Given this patient's long-standing hypoxemia and erythrocytosis, the clinical team appropriately judged surgical risk to be prohibitive without invasive hemodynamic data from cardiac catheterization.

This case highlights a critical healthcare disparity: although a specialized tertiary center (Columbia University Medical Center) accepted transfer, the patient's lack of health insurance ultimately precluded this potentially life-saving referral. This outcome underscores how socioeconomic factors profoundly influence clinical outcomes. Such barriers are particularly consequential for patients with complex, chronic conditions requiring multidisciplinary specialty care.

Several limitations of this case report warrant acknowledgment: inability to obtain prior medical records from Jamaica, absence of invasive hemodynamic data (cardiac catheterization was not performed), and incomplete longitudinal follow-up (the patient remained hospitalized at the time of chart review).

Conclusion

Clinicians should maintain a high index of suspicion for undiagnosed congenital heart disease in adults presenting with hypoxemia and secondary erythrocytosis, particularly in patients with limited healthcare access. This case further demonstrates how socioeconomic factors, including insurance status, can significantly impact access to specialized care. Further research is needed to identify strategies for earlier detection of CHD in underserved populations and to address systemic barriers to equitable healthcare delivery.

Informed Consent: Written informed consent was obtained from the patient for publication of this case report and any accompanying images. All identifying information has been removed to protect patient privacy.

Conflicts of Interest: The authors declare no conflicts of interest relevant to the content of this manuscript.

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Ethics Approval: Institutional review board (IRB) approval was not required for this case report, as it involved a single patient and did not constitute human subjects research per institutional policy. All procedures were performed in accordance with the ethical standards of the responsible institutional committee and with the 1964 Helsinki Declaration and its later amendments.

AI Use Disclosure: The authors used AI-based language tools to assist with literature review, drafting, and reference formatting during manuscript preparation. All content was reviewed, verified, and approved by the authors, who take full responsibility for the accuracy and integrity of the manuscript.

Works Cited

1. Liu Y, Chen S, Zühlke L, et al. Global birth prevalence of congenital heart defects 1970-2017: updated systematic review and meta-analysis of 260 studies. *J Am Heart Assoc.* 2019;8(16):e010784.
2. Stout KK, Daniels CJ, Aboulhosn JA, et al. 2018 AHA/ACC guideline for the management of adults with congenital heart disease. *Circulation.* 2019;139(14):e698-e800.
3. Craig B. Atrioventricular septal defect: from fetus to adult. *Heart.* 2006;92(12):1879-1885.

4. Galie N, Humbert M, Vachiery JL, et al. 2015 ESC/ERS guidelines for the diagnosis and treatment of pulmonary hypertension. *Eur Heart J.* 2016;37(1):67-119.
5. Kumar A, et al. Hemodynamic rounds and clinical pathology correlation: Evaluation of a polycythemic patient guided by imaging, hemodynamics, and endomyocardial biopsy. *Ann Pediatr Cardiol.* 2022;14(4):516-520.
6. Canobbio MM, Warnes CA, Aboulhosn J, et al. Management of pregnancy in patients with complex congenital heart disease. *Circulation.* 2017;135(8):e50-e87.
7. Brida M, Chessa M, Celermajer D, et al. Atrial septal defect in adulthood: a new paradigm for congenital cardiology. *Eur Heart J.* 2022;43(28):2660-2671.

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