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Intermediate Atrioventricular Septal Defect with Severe Pulmonary Arterial Hypertension Diagnosed at Age 58: A Rare Case from Central Africa

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Abstract

Background: Intermediate atrioventricular septal defect (AVSD) rarely presents in late adulthood, as most untreated patients develop irreversible pulmonary arterial hypertension (PAH) and die in early childhood. Survival beyond infancy without surgical correction is exceptional and typically associated with restrictive ventricular components that protect the pulmonary vasculature. **Case Presentation:** We report a 58-year-old mechanic from the Republic of Congo with facial dysmorphism suggestive of Down syndrome who presented with progressive dyspnea and right heart failure. Clinical examination revealed generalized cyanosis (oxygen saturation 88% on room air), New York Heart Association (NYHA) class III dyspnea, bilateral lower extremity edema, and ascites. Cardiovascular examination demonstrated a loud systolic murmur at the pulmonary area and an accentuated second heart sound. Chest radiography showed severe cardiomegaly with pulmonary artery prominence. Electrocardiography revealed right ventricular hypertrophy with ventricular ectopy. Transthoracic echocardiography confirmed intermediate AVSD with a large ostium primum atrial septal defect (34 mm) showing significant left-to-right shunting (Qp/Qs > 1.5), a restrictive inlet ventricular septal defect nearly obliterated by common atrioventricular valve attachments, severe bilateral atrioventricular valve regurgitation, and severe PAH (systolic pulmonary artery pressure 58 mmHg). N-terminal pro-B-type natriuretic peptide was markedly elevated at 340,000 pg/mL. The patient received palliative med-

ical therapy with diuretics and aldosterone antagonists, achieving symptomatic improvement from NYHA class III to II. Surgical correction was not feasible due to established PAH and limited local cardiovascular surgical resources. **Conclusion:** This case represents, to our knowledge, the first documented intermediate AVSD diagnosed in late adulthood in the Republic of Congo. The restrictive ventricular component provided relative pulmonary protection, enabling prolonged survival despite the absence of surgical intervention. This case highlights critical challenges in managing complex congenital heart disease in resource-limited settings and underscores the importance of prenatal screening and early surgical intervention programs in sub-Saharan Africa.

Keywords

Atrioventricular Septal Defect, Pulmonary Arterial Hypertension, Adult Congenital Heart Disease, Late Diagnosis, Echocardiography, Sub-Saharan Africa, Republic of Congo

1. Introduction

Atrioventricular septal defect (AVSD), also known as atrioventricular canal or endocardial cushion defect, accounts for approximately 4% - 5% of all congenital heart diseases and represents one of the most complex cardiac malformations [1] [2]. This anomaly results from the failure of the endocardial cushions to fuse properly during embryonic development, creating communications between cardiac chambers at the crux of the heart [3].

AVSD encompasses a spectrum of anatomical variants classified as complete, intermediate (also termed transitional or partial with significant ventricular component), and partial forms [4]. Complete AVSD features a large interventricular communication, primum atrial septal defect, and common atrioventricular valve with five leaflets. Intermediate AVSD is characterized by a primum atrial septal defect and a restrictive inlet ventricular septal defect, often partially or completely obliterated by attachments from the common atrioventricular valve [5]. This restrictive ventricular component critically modulates pulmonary blood flow and influences natural history.

Without surgical correction, complete AVSD typically leads to severe pulmonary arterial hypertension (PAH) within the first six months of life, with most patients dying before age two [6] [7]. Contemporary guidelines recommend surgical repair before six months of age to prevent irreversible pulmonary vascular disease [5]. However, intermediate AVSD with restrictive ventricular components may permit prolonged survival, occasionally into adulthood, particularly when the ventricular defect limits left-to-right shunting [8] [9].

Adult congenital heart disease (ACHD) represents a growing challenge in developed countries, with improved surgical outcomes enabling most patients to

reach adulthood [10]. However, in sub-Saharan Africa, late presentation of congenital heart disease remains common due to limited access to prenatal diagnosis, pediatric cardiology services, and cardiovascular surgery [11] [12].

We present a case of intermediate AVSD diagnosed at age 58 in the Republic of Congo, representing one of the oldest reported patients with unrepaired AVSD. To our knowledge, this is the first documented case of intermediate AVSD presenting in late adulthood in Central Africa. This case illustrates the protective hemodynamic role of restrictive ventricular communications and highlights the diagnostic and therapeutic challenges of managing complex congenital heart disease in resource-limited settings.

2. Case Presentation

2.1. Patient Information and Clinical History

A 58-year-old male mechanic, married with four children, presented to the Department of Cardiology B at the University Hospital of Brazzaville with progressive dyspnea. The patient reported the onset of exertional dyspnea several years prior, initially occurring only with strenuous physical activity. Over time, dyspnea progressed to occur with moderate exertion and eventually at rest, prompting medical consultation when symptoms reached NYHA class III.

The patient had no history of hypertension or diabetes mellitus. He reported chronic alcohol and tobacco use. Notably, no cardiac evaluation had been performed during childhood or early adulthood, and the patient had received no specific medical follow-up for potential congenital heart disease despite visible facial dysmorphism since birth. Genetic testing had never been performed, and chromosomal analysis was not accessible in our clinical setting.

2.2. Physical Examination

Physical examination revealed distinctive facial dysmorphism consisting of flat facies, flattened nasal bridge, and upward-slanting palpebral fissures, features suggestive of Down syndrome phenotype.

Vital signs demonstrated: temperature 36.8˚C, oxygen saturation 88% on room air, respiratory rate 30 breaths/minute, heart rate 100 beats/minute (regular), and blood pressure 130/80 mmHg. The patient exhibited generalized cyanosis involving lips, tongue, and nail beds, consistent with central cyanosis.

Cardiovascular examination revealed evidence of severe right heart failure with anasarca: severe bilateral lower extremity edema with induration, moderate ascites, and scrotal edema. Jugular venous pressure was elevated. Oliguria was reported. Cardiac auscultation identified a loud grade 4/6 pansystolic murmur best heard at the left upper sternal border (pulmonary area) and markedly accentuated pulmonary component of the second heart sound (P2).

2.3. Diagnostic Assessment

Chest Radiography (Figure 1) demonstrated severe cardiomegaly with cardi-

othoracic ratio exceeding 0.6, prominent convexity of the left middle cardiac border reflecting pulmonary artery dilatation, significant right heart border enlargement, and bilateral hilar vascular congestion.

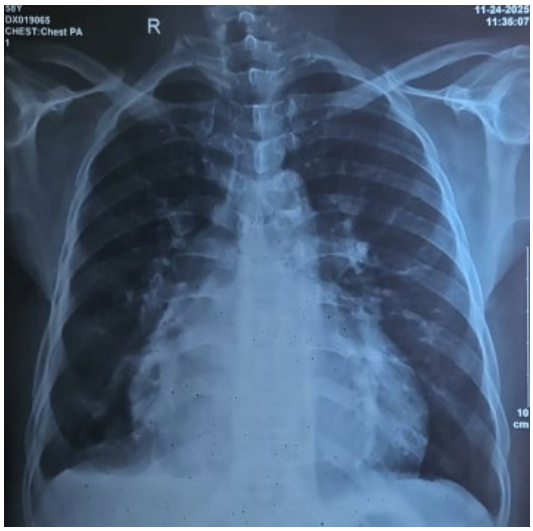

Figure 1. Chest radiograph findings. Posteroanterior chest radiograph showing severe cardiomegaly with cardiothoracic ratio >0.6, prominent left middle arch (dilated pulmonary artery trunk), right heart border enlargement, and bilateral hilar vascular congestion consistent with pulmonary arterial hypertension and significant left-to-right shunt.

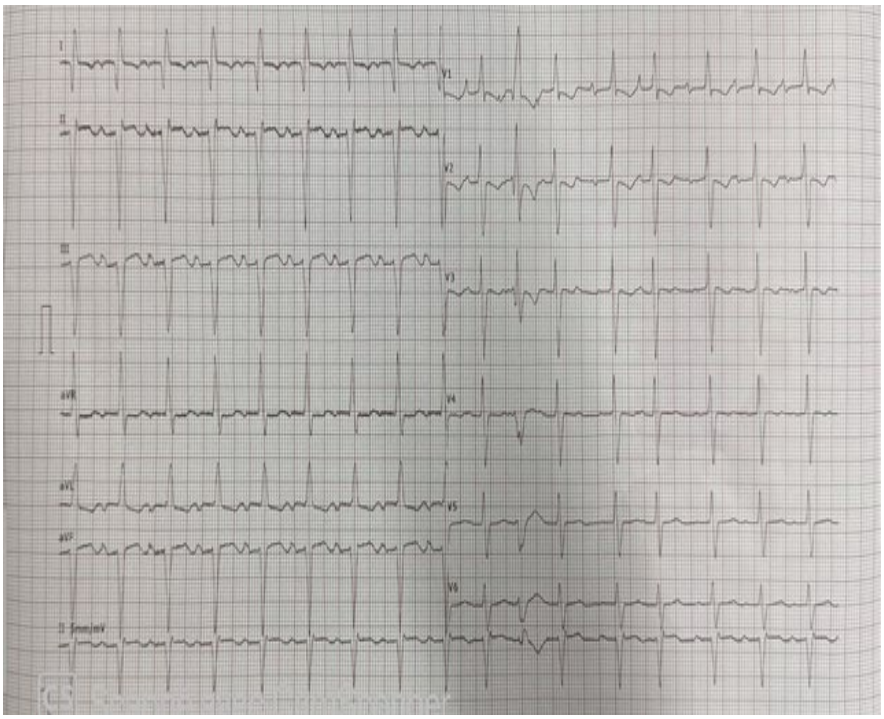

Figure 2. Electrocardiographic findings. Twelve-lead electrocardiogram demonstrating sinus rhythm with positive aVR deflection (leftward superior axis deviation, typical of AVSD), right ventricular hypertrophy pattern (R wave predominance in V1-V2, right axis deviation), and ventricular premature beats.

Electrocardiography (Figure 2) showed sinus rhythm with positive deflection in lead aVR (leftward superior QRS axis, characteristic of AVSD), right ventricular hypertrophy pattern with dominant R waves in leads V1-V2 and right axis deviation, and frequent ventricular premature complexes. Holter monitoring was not performed during the initial evaluation, which would have allowed quantification of ventricular ectopy and contributed to sudden cardiac death risk stratification.

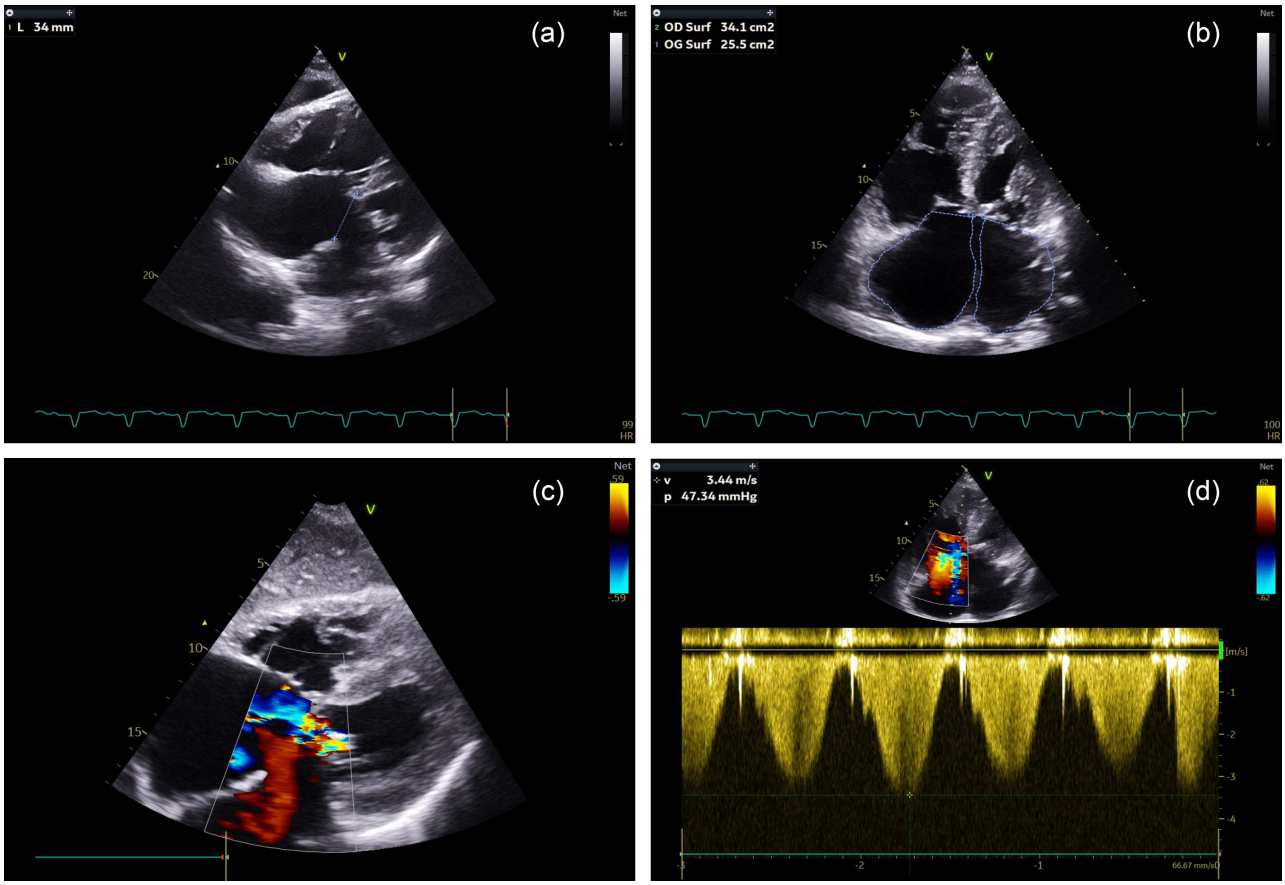

Figure 3. Comprehensive echocardiographic findings. (a) Apical four-chamber view demonstrating large ostium primum atrial septal defect measuring 34 mm (white arrow), common atrioventricular junction at the crux of the heart, and significant right atrial and right ventricular dilatation; (b) Quantification of cardiac chambers showing marked right-sided enlargement (right atrium surface 34.1 cm^2, left atrium 25.5 cm^2), confirming chronic volume overload from left-to-right shunting; (c) Color Doppler imaging in apical four-chamber view revealing significant left-to-right shunting through the ostium primum defect (blue jet) and severe regurgitation of both left and right components of the common atrioventricular valve (multicolor turbulent jets); (d) Continuous-wave Doppler interrogation of tricuspid regurgitation jet demonstrating peak velocity of 3.44 m/s, corresponding to estimated right ventricular to right atrial pressure gradient of 47 mmHg. Assuming right atrial pressure of 10 mmHg (elevated due to right heart failure), estimated systolic pulmonary artery pressure is 57 mmHg, confirming severe pulmonary arterial hypertension. Abbreviations: CIA = communication interatriale (atrial septal defect); OD = oreillette droite (right atrium); OG = oreillette gauche (left atrium); PASP = pulmonary artery systolic pressure.

Transthoracic Echocardiography (Figure 3) provided a definitive diagnosis: situs solitus with concordant ventriculo-arterial connections. A large ostium primum atrial septal defect measuring 34 mm was identified, associated with a sig-

nificant left-to-right shunt (Qp/Qs > 1.5). At the ventricular level, a restrictive inlet ventricular septal defect was observed, almost completely obliterated by chordal attachments from the common atrioventricular valve, resulting in minimal residual interventricular communication. The significant volume load was primarily attributable to the large atrial septal defect.

The atrioventricular junction was common, with severe regurgitation of both left and right atrioventricular valve components. There was marked dilatation of the right atrium and right ventricle, associated with leftward displacement of the interventricular septum. Left ventricular systolic function was preserved, with an estimated ejection fraction of 74%. Finally, echocardiography suggested severe pulmonary arterial hypertension, with an estimated systolic pulmonary artery pressure of 58 mmHg, derived from continuous-wave Doppler analysis of tricuspid regurgitation.

2.4. Therapeutic Intervention

Given the presence of severe, likely fixed PAH with central cyanosis and the absence of cardiovascular surgical facilities in the Republic of Congo, surgical repair was deemed contraindicated and unfeasible. The patient received palliative medical management consisting of:

- Sodium restriction (<2 g/day)
- Loop diuretic (furosemide 80 mg twice daily)
- Aldosterone antagonist (spironolactone 50 mg daily)
- Prophylactic-dose low-molecular-weight heparin for immobilization-related thromboprophylaxis during hospitalization
- Long-term anticoagulation was not initiated, given the predominantly left-to-right shunting pattern and absence of documented atrial arrhythmias, though thrombotic risks associated with cyanosis and potential erythrocytosis were acknowledged and will be monitored during follow-up.

2.5. Follow-Up and Outcomes

Clinical improvement was observed during the 10-day hospitalization. Dyspnea improved from NYHA class III to class II, and congestive symptoms markedly decreased with 10 kg weight loss attributed to diuresis. The patient was discharged with maintenance diuretic therapy and scheduled for monthly cardiology follow-up.

Long-term curative treatment remains unavailable. The patient is registered for potential medical evacuation to an international center for comprehensive genetic evaluation and assessment of candidacy for advanced PAH therapies or, if PAH is not fixed, potential surgical correction.

3. Discussion

3.1. Epidemiology and Natural History of AVSD

Congenital heart diseases affect approximately 1% of live births worldwide, rep-

resenting around 1.5 million cases annually [12] [13]. AVSD accounts for 4% - 5% of congenital cardiac malformations, with complete forms more common than intermediate or partial variants [1] [2]. The prevalence of AVSD in adults with congenital heart disease has been estimated at 14 per 10,000 in population-based studies from North America, with female predominance [14].

AVSD is strongly associated with Down syndrome (trisomy 21), occurring in 40% - 45% of AVSD cases, while approximately 40% of children with Down syndrome have congenital heart disease, most commonly AVSD [15] [16]. However, AVSD can occur as an isolated malformation or in association with other genetic syndromes. Our patient exhibited phenotypic features suggestive of Down syndrome, though chromosomal confirmation was not accessible in our resource-limited setting.

Patients with Down syndrome and AVSD typically develop pulmonary arterial hypertension earlier and more rapidly than non-syndromic patients with AVSD [3]. The increased pulmonary vascular reactivity observed in trisomy 21 has been attributed to overexpression of anti-angiogenic factors encoded on chromosome 21, leading to impaired pulmonary vascular development and signaling [17]. This heightened susceptibility leads to accelerated pulmonary vascular remodeling, with irreversible changes often occurring by 6 - 12 months of age in complete AVSD [18]. This makes our patient's survival to age 58 particularly remarkable and underscores the protective role of the restrictive ventricular component in limiting pulmonary blood flow throughout life.

The natural history of untreated AVSD is dismal. Complete AVSD without surgical intervention typically results in severe PAH developing within the first 6 - 12 months of life, with 80% - 90% mortality by age two years [6] [7]. Progressive pulmonary vascular disease leads to Eisenmenger syndrome—reversal of the shunt from left-to-right to right-to-left with resultant central cyanosis and multi-organ complications [19].

3.2. Exceptional Survival in Intermediate AVSD: The Protective Role of Restrictive VSD

Our patient's survival to age 58 without surgical intervention is exceptional and directly attributable to the intermediate anatomical variant with a restrictive ventricular component. Several case reports have documented late presentation of AVSD in adulthood, but typically in younger patients (third to fourth decade) and predominantly with partial forms lacking significant ventricular communication [8] [9] [19].

The restrictive inlet ventricular septal defect in intermediate AVSD, particularly when significantly narrowed or nearly obliterated by valve tissue attachments, functions as a natural "protective barrier" limiting left-to-right shunting at the ventricular level [20]. This hemodynamic protection reduces pulmonary blood flow and attenuates the rate of pulmonary vascular remodeling compared to complete AVSD with unrestricted interventricular communication.

Rahmoun and colleagues described a 34-year-old patient with intermediate AVSD featuring a small subaortic VSD who remained relatively asymptomatic until adulthood [21]. Similarly, Rabarijaona *et al.* reported late discovery of complete AVSD in an adult with severe PAH [20] [22]. However, survival beyond the fifth decade remains extraordinarily rare in the literature, making our case among the oldest reported unrepaired AVSD patients.

The prolonged clinical tolerance observed in our patient reflects several hemodynamic features:

1) **Restrictive VSD** minimizing interventricular left-to-right shunting.

2) **Large primum ASD** allowing preferential left-to-right shunting at the atrial level with a lower pressure gradient.

3) **Gradual progression** of PAH rather than acute onset, permitting right ventricular adaptation.

4) **Preserved left ventricular systolic function** (ejection fraction 74%) despite chronic volume overload.

3.3. Pulmonary Arterial Hypertension and Eisenmenger Physiology

Despite the protective restrictive VSD, our patient developed severe PAH (systolic pressure 58 mmHg) by late adulthood. The estimated systolic PAH of 58 mmHg represents approximately 45% of systemic systolic pressure (assuming systolic BP ~130 mmHg), indicating severe but potentially not yet "fixed" or suprasystemic PAH.

Pulmonary arterial hypertension in AVSD results from chronic pulmonary overcirculation with increased pulmonary blood flow (high Qp/Qs ratio) causing endothelial dysfunction, pulmonary vascular remodeling, and progressive increase in pulmonary vascular resistance [23]. When pulmonary vascular resistance exceeds systemic vascular resistance, shunt reversal occurs, creating Eisenmenger syndrome with irreversible PAH [24].

Clinical features in our patient consistent with advanced PAH and evolving Eisenmenger physiology included:

- Central cyanosis with resting oxygen saturation 88%.
- Accentuated pulmonary component of second heart sound.
- Right ventricular hypertrophy on ECG and echocardiography.
- Severe right heart failure with anasarca.
- Markedly elevated NT-proBNP (340,000 pg/mL).

The presence of central cyanosis (SpO2 88%) despite predominantly left-to-right shunting (Qp/Qs > 1.5) represents an apparent hemodynamic contradiction. The cyanosis is likely explained by ventilation-perfusion mismatch secondary to severe pulmonary vascular disease and chronically elevated pulmonary pressures, rather than established shunt reversal to Eisenmenger physiology. True bidirectional or right-to-left shunting at the atrial level may occur during exertion or with elevated right-sided pressures, though resting echocardiography demonstrated

net left-to-right flow.

Definitive determination of whether PAH is reversible or fixed would require right heart catheterization with vasodilator testing, which is the gold standard for assessing pulmonary vascular reactivity and guiding surgical candidacy [25]. This procedure was not available in our setting.

3.4. Arrhythmic Complications

Our patient exhibited ventricular ectopy on ECG. Approximately 50% of adults with AVSD develop tachyarrhythmias during their lifetime, including atrial fibrillation, atrial flutter, and ventricular arrhythmias [26]. Ventricular arrhythmias represent a major cause of sudden cardiac death in ACHD, particularly in patients with ventricular volume overload, ventricular hypertrophy, and myocardial fibrosis [27]. Long-term monitoring and consideration of antiarrhythmic therapy or implantable cardioverter-defibrillator may be warranted.

3.5. Diagnostic Challenges in Sub-Saharan Africa

This case highlights critical gaps in congenital heart disease diagnosis and management in sub-Saharan Africa:

Prenatal and Neonatal Screening: Fetal echocardiography, which enables prenatal AVSD diagnosis in developed countries, remains largely unavailable in Central Africa [28] [29]. Routine neonatal cardiac screening is not standard practice.

Pediatric Cardiology Services: Limited access to pediatric cardiologists and diagnostic echocardiography results in delayed or missed diagnoses during critical early childhood period when surgical intervention is optimal [11].

Cardiovascular Surgery: The Republic of Congo lacks cardiac surgical facilities for complex congenital repairs. Patients requiring surgery must be evacuated internationally, which is financially prohibitive for most families [30].

Genetic Testing: Chromosomal analysis and genetic counseling are not readily accessible, preventing confirmation of associated genetic syndromes and family screening.

These systemic barriers contribute to late presentations like our case, where patients survive to adulthood with undiagnosed complex congenital heart disease, often presenting only when severe complications develop.

3.6. Management Considerations

Medical Therapy: Our patient received standard heart failure management with diuretics and aldosterone antagonists, achieving symptomatic improvement. However, definitive therapy for AVSD is surgical [31].

Surgical Candidacy: Surgical repair of AVSD involves closure of atrial and ventricular septal defects with patchwork and reconstruction of the atrioventricular valves [32]. Modern techniques achieve excellent outcomes when performed in childhood before development of severe PAH [5]. However, surgery is generally contraindicated when PAH becomes fixed with pulmonary vascular resistance ex-

ceeding 8 - 10 Wood units or Eisenmenger syndrome develops [33].

Determining whether PAH in our patient is reversible or fixed would require right heart catheterization with vasodilator testing, which was not available. The presence of severe cyanosis and clinical features of Eisenmenger physiology suggest advanced, likely irreversible pulmonary vascular disease, making surgical repair extremely high-risk or contraindicated [34].

Advanced PAH Therapies: Pulmonary vasodilator therapies (phosphodiesterase-5 inhibitors, endothelin receptor antagonists, prostacyclin analogs) may improve functional capacity and quality of life in Eisenmenger syndrome, though evidence specifically for AVSD-related Eisenmenger syndrome is limited [35] [36]. These medications are not currently available in the Republic of Congo.

Prognosis: Without surgical correction and with established severe PAH, prognosis is poor. Eisenmenger syndrome has a median survival of 30 - 50 years from birth, with progressive deterioration [37]. Our patient, at age 58, has already exceeded typical life expectancy for unrepaired complete AVSD, though intermediate forms may have a better prognosis.

4. Conclusions

We report an exceptionally rare case of intermediate AVSD diagnosed at age 58 in the Republic of Congo, representing, to our knowledge, the oldest documented unrepaired AVSD patient from Central Africa. Survival to late adulthood was enabled by the restrictive ventricular septal component, which provided relative pulmonary protection by limiting interventricular left-to-right shunting. However, the patient ultimately developed severe PAH with evolving Eisenmenger physiology, manifesting as right heart failure and central cyanosis.

This case illustrates critical challenges in managing complex congenital heart disease in resource-limited settings, where delayed diagnosis, absent prenatal screening, limited pediatric cardiology infrastructure, and lack of cardiovascular surgical facilities prevent timely intervention during the optimal therapeutic window.

Patient Consent

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

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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