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Navigating Challenges And Outcomes In A 25-Year-Old Male With Adult Congenital Heart Disease: A Case Study

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Abstract
This case report presents a 25-year-old male mason diagnosed with adult congenital heart disease (Complete atrioventricular canal (CAVC) defect), who had an uneventful medical history until six months ago when he presented with symptoms of easy fatigue, nocturnal coughing, shortness of breath, orthopnea, and lower limb swelling. On physical examination, he was desaturating, with mild pallor but not cyanotic, and had raised JVP with bilateral basal crackles. Laboratory investigations revealed mild anemia, normal renal function, and electrolytes. Echocardiography revealed a CAVC defect. This report explores the unique clinical presentation and challenges faced in treating congenital heart disease in adulthood in a resource-limited area, and discusses the patient's eligibility for surgery in the absence of Eisenmenger syndrome.

Keywords*: Congenital heart disease, Atrial-ventricular septal defect, Heart failure*

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I. Introduction

This case report details the clinical course of a 25-year-old male patient who presented with signs and symptoms of heart failure. Notably, the patient had no documented history of previous admissions, treatment, or any known chronic illness. The unique clinical presentation of this patient underscores the challenges and complexities encountered in managing adult congenital heart disease (ACHD) in a resource-limited area. The absence of prior clinical signs and symptoms, and the evolution to heart failure later in life, make it challenging to diagnose this kind of disease at its earlier stage for prompt surgical treatment. This report aims to highlight the unique challenges faced in treating congenital heart disease in adulthood in an area with no capabilities for cardiac surgeries. It also discusses whether the patient is still a candidate for surgery if supported, in case he has not developed Eisenmenger syndrome. This introduction provides context and sets the stage for a detailed exploration of the challenges and implications associated with managing adult congenital heart disease in resource-constrained environments.

II. Patient Information And Observation

Patient Information: A 25-year-old male, secondary level education, non-insured, non-alcoholic, non-smoker, married with no children, employed as a mason, born through spontaneous normal delivery without complications. The patient led a normal life with no symptoms until six months prior to presentation.
Timeline: The patient has no prior history of admissions or being treated for any chest or cardiac infections. He has no family history of any chronic illnesses, and no history of surgeries or blood transfusions.
Clinical Findings: Six months ago, the patient started experiencing easy fatigue, which progressively worsened. This was associated with nocturnal coughing, shortness of breath, inability to lie flat (orthopnea), lower limb swelling, and a sensation of air hunger at night. These symptoms prompted the patient to seek medical attention. On arrival at the emergency department:

Physical Examination:

- General Appearance: Alert, mild pallor, not cyanotic, no finger clubbing, desaturating on room air to 80% and above 90% on 8 liters.
- Edema: Bilateral lower limb pitting edema.
- Cardiovascular System:
  - Raised jugular venous pressure.
  - Positive hepatojugular reflux.
  - Tachycardia (Pulse: 100 bpm, regular rhythm, good volume).

- Heart sounds: S1 and S2 were heard, with a systolic murmur at the mitral region.
- Respiratory System: Bilateral basal crackles.

Investigations: Due to limited resources, initial laboratory investigations included a complete blood count (CBC), electrolytes, renal function tests, and urine analysis. These tests revealed:
- CBC: Low hemoglobin level of 10 g/dL.
- Electrolytes: Normal.
- Renal Function Tests: Normal.
- Urine Analysis: Normal.

Further diagnostic tests included:
- Electrocardiogram (ECG): Showed left ventricular hypertrophy.
- Chest X-ray: Revealed cardiomegaly.
- Echocardiography: Identified a complete atrioventricular septal defect.

Diagnostic Assessment: Upon presentation to the internal medicine department, the patient exhibited classic signs and symptoms of heart failure. The diagnosis of heart failure with reduced ejection fraction, estimated at 38.39%, was confirmed by echocardiogram (Figure 1), further more there was right ventricular hypertrophy (RVH), primum atrial septal defect, an inlet ventricular septal defect and a common atrioventricular valve(Figure 2), and a left to right shunt (figure 3). There was hypertrophy and enlargement of the left ventricle (Figure 2). The patient's asymptomatology in early life highlights the difficulties in identifying ACHD in settings with limited access to advanced diagnostic tools as routine check-ups. The absence of a prior diagnosis of ACHD coupled with uneventful early life further complicates the early detection of underlying cardiac pathology.

Therapeutic Interventions and Outcomes: The management of this case was particularly challenging due to limited resources and the lack of guideline-directed medical therapy for heart failure in the remote setting. Despite these challenges, the patient responded positively to treatment, including oxygen supplementation, loop diuretics, mineralocorticoid receptor antagonist (spironolactone), and angiotensin receptor blockers (losartan). The recovery of the patient underscores the critical role of stabilizing a patient with ACHD even in resource-limited environments.

This case highlights the challenges faced in resource-limited areas to manage ACHD, which requires repair before ending up in Eisenmenger syndrome. Challenges in stabilizing such patients where reliance on guideline-directed therapies may be limited. Furthermore, it underscores the necessity of raising awareness about advanced cardiac conditions and equipping healthcare providers in remote areas with effective tools and strategies for early diagnosis and intervention.

Follow-Up and Outcomes/Prognosis: The patient returned for follow-up after one month and was in HF NYHA Class II. He was still on anti-failure medications. The prognosis of the patient is poor due to various factors surrounding him, including his low economic status, which hinders him from traveling for the surgical procedure, and his impending risk of developing Eisenmenger syndrome.

Patient Perspective: Our patient reports that he is satisfied with the care and treatment he received at Village Health Works, Women Health Pavilion, Kigutu Hospital.

Ethics Declaration and Consent for Publication: Ethical approval was not applicable. Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

III. Discussion

Congenital heart disease (CHD) is traditionally considered a pediatric condition, as many severe congenital defects are diagnosed and treated in early childhood [1]. However, with advancements in medical and surgical management, an increasing number of children with CHD are surviving into adulthood [2]. Consequently, adult congenital heart disease (ACHD) has emerged as a distinct field within cardiology .

The late presentation of CHD in adulthood, as demonstrated in this case, poses significant diagnostic challenges. Adults with undiagnosed CHD often present with symptoms that mimic more common acquired cardiac conditions, such as heart failure [3], which can lead to delays in appropriate diagnosis and management . The patient's symptoms, including progressive fatigue, nocturnal coughing, orthopnea, and lower limb swelling, are classic signs of heart failure but are not typically associated with congenital defects in adults. In resource-limited settings, where access to advanced diagnostic tools like echocardiography is limited, these challenges are exacerbated .

Resource limitations significantly affect the management of CHD in adults. In many low- and middle-income countries (LMICs), there is a lack of specialized centers for ACHD [4], and patients often have to travel long distances to receive appropriate care . This case highlights the importance of primary care providers and general practitioners being aware of ACHD and considering it in differential diagnoses, despite the limited resources. Basic diagnostic tools such as ECG and chest X-rays, which are more readily available, can provide critical initial information to suspect CHD .

The mainstay of treatment for many congenital heart defects is surgical correction [5]. However, access to cardiac surgery is often limited in resource-poor settings [6]. For the patient described, surgical intervention for the complete atrioventricular septal defect was not immediately available, emphasizing the need for improved healthcare infrastructure and access to specialized cardiac care [7]. The risk of developing complications such as Eisenmenger syndrome, where increased pulmonary vascular resistance leads to cyanosis and inoperability [8], further complicates the clinical picture and underscores the urgency of timely intervention.

In the absence of surgical options, medical management focuses on symptom relief and preventing complications. This includes the use of diuretics, ACE inhibitors or angiotensin receptor blockers, beta-blockers, and sometimes anticoagulants [5]. The patient in this case was managed with oxygen supplementation, loop diuretics, spironolactone, and losartan, which provided symptomatic relief and stabilized his condition. However, long-term outcomes are often suboptimal without definitive surgical repair .

Long-term follow-up is crucial for adults with CHD to monitor for complications, manage symptoms, and provide timely interventions as needed. In LMICs, establishing a structured follow-up system is challenging due to resource constraints and logistical issues . However, creating such systems is essential to improve outcomes for patients with ACHD [7]. This case illustrates the need for regular follow-up and the provision of ongoing medical care to manage heart failure symptoms and monitor for the development of complications like Eisenmenger syndrome .

Raising awareness among healthcare providers about the potential for CHD in adults and providing education on recognizing and managing these conditions is vital. Training programs and continuing medical education can help equip healthcare providers with the necessary skills and knowledge to identify and manage ACHD in resource-limited settings . Public health initiatives to raise awareness about CHD and the importance of early diagnosis and treatment can also play a significant role in improving patient outcomes .

IV. Conclusion

This case underscores the challenges of diagnosing and managing congenital heart disease in adulthood, particularly in low-resource settings. The patient's late presentation and the limited availability of diagnostic and therapeutic resources highlight the importance of awareness, early recognition, and adaptable management strategies. Improving access to specialized care, enhancing diagnostic capabilities, and providing ongoing education for healthcare providers are critical steps toward better outcomes for adults with congenital heart disease in resource-limited environments.

Competing interests

The authors declare no competing interests.

Authors' contributions

Author´s contribution MPM, LH, MN, GK and PM admitted the patient and were his attending physicians in the ward. KK and HM provided their expert opinion in the diagnosis of this patient. MPM guided the preparation of the manuscript. All authors read and approved the manuscript.

Acknowledgments

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Figures

Figure 1: - Echocardiogram, long axis view, showing the measurement of ejection fraction.

Figure 2: - Echocardiogram, four chamber view, showing CAVC defect, RVH, primum atrial septal defect, an inlet ventricular septal defect and a common atrioventricular valve

Figure 3: - Echocardiogram, four chamber view, showing a left to right shunt

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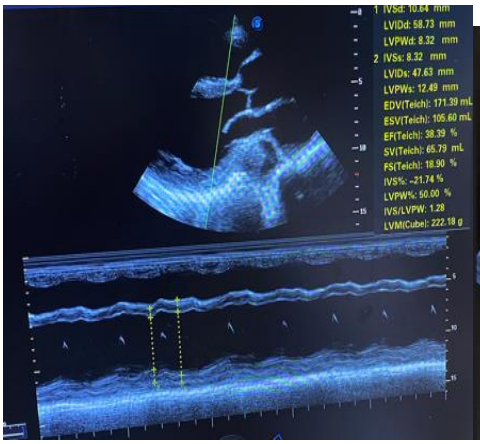

Figure 1

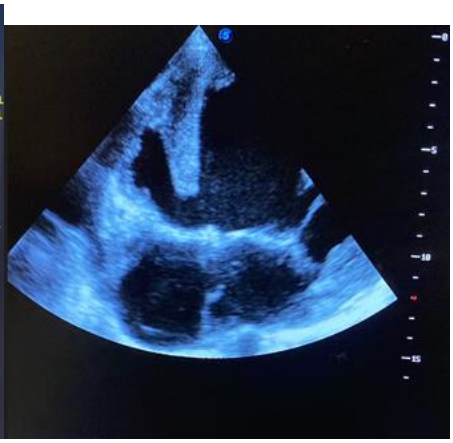

Figure 2

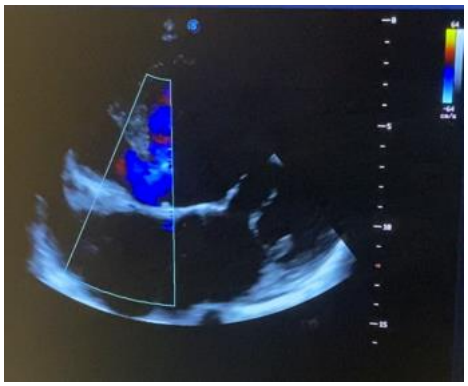

Figure 3