Research Article

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Laubry-Pezzi Syndrome: A Rare Congenital Heart Disease in Sub-Saharan Africa

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Abstract

Introduction: Laubry-Pezzi syndrome was first described by Charles Laubry and Cesare Pezzi in 1921. It is a rare congenital heart disease in clinical practice, characterized anatomopathologically by aortic regurgitation due to prolapse of an aortic valve cusp, most commonly the right cusp, and a ventricular septal defect.

Case presentation: The authors report a medical observation of a 29-year-old young man followed in the cardiology department B of Brazzaville University Hospital for dyspnea, intermittent constrictive chest pain, and palpitations. The onset of symptoms dates back more than 7 years with dyspnea during significant exertion, which had not received any medical attention. Due to the appearance of frequent anginal pain, mostly on exertion, and stage III NYHA dyspnea, the patient consulted at Brazzaville University Hospital.

Clinical examination revealed a particular morphotype with distinct facial features and short stature. The patient was afebrile. Cardiovascular examination showed arterial hyperpulsatility, an intense steam-jet systolic regurgitation murmur in the precordial area radiating in a wheel-spoke pattern, and a 4/6 soft, high-pitched diastolic murmur of aortic insufficiency. There were no signs of heart failure.

Paraclinical investigations revealed left ventricular hypertrophy on electrocardiogram. Transthoracic echocardiography coupled with Doppler showed a perimembranous ventricular septal defect associated with grade 4 aortic regurgitation. The aortic regurgitation was primarily due to prolapse of the right cusp of the aortic valve, and left ventricular dilatation with preserved left ventricular ejection fraction at 62%. Pending corrective surgery, symptomatic medical treatment based on angiotensin-converting enzyme inhibitors and nitrate derivatives had been instituted. The patient's hemodynamic status is stable except for dyspnea during significant physical exertion. Cardiovascular surgery has not yet been performed to date.

Conclusion: Laubry-Pezzi syndrome is a rare congenital heart disease; its diagnosis is made by echocardiography and its treatment is surgical.

Keywords

Congenital heart disease, Laubry-Pezzi syndrome, Doppler echocardiography, Congo.

INTRODUCTION

Laubry-Pezzi syndrome was first described by Charles Laubry and Cesare Pezzi in 1921 [1]. It is a rare condition worldwide; its incidence is poorly defined and remains disparate across populations. Although considered a rare condition, the incidence of aortic cusp prolapse in patients with perimembranous VSD is estimated to be between 5% and 8% [2,3]. Laubry-Pezzi syndrome occurs in the context of a genetic disorder due to a TGFBR1 gene mutation, which participates in the development of connective tissue and cardiovascular structures [4,5]. However, being a genetic abnormality, patients typically present with particular morphotypes and short stature [4,5]. The anomalies of Laubry-Pezzi syndrome associate congenital cardiac malformations characterized by a VSD and aortic valve prolapse with coaptation defect of the latter, resulting in the Venturi effect [6]: a significant pressure drop when blood flows through the VSD beneath the aortic valve, thus aspirating the non-coronary or right leaflet of the aortic valve, causing aortic regurgitation [7,8]. It is most commonly found in infundibular VSDs but can also be encountered in

perimembranous VSDs. Diagnosis relies largely on echocardiography, which allows detailed assessment of the ventricular septal communication, valvular morphology, and degree of regurgitation.

In sub-Saharan Africa, a few rare cases have been noted [8]. In Africa, diagnostic errors of this condition remain frequent, as clinical manifestations can mimic those of other endemic conditions, often leading to delayed recognition and inappropriate treatment [9]. Moreover, access to advanced diagnostic modalities, particularly high-quality echocardiography, is not always available, further limiting rapid detection [10]. Therefore, we deemed it appropriate to describe a medical observation of the natural clinical evolution of Laubry-Pezzi syndrome in a young adult, an extremely rare clinical situation whose management in our practice context remains delicate due to the absence of cardiovascular surgery facilities.

CASE PRESENTATION

A 29-year-old young adult male, general practitioner, residing in Brazzaville, single without children, was admitted to the cardiology department B of Brazzaville University Hospital on April 20, 2025, for dyspnea and constrictive chest pain. The onset of symptoms dates back more than 5 years with dyspnea during significant exertion, which had not received any medical attention. Due to persistent dyspnea progressing to NYHA stage III and the appearance of frequent anginal pain, mostly on exertion, the patient decided to consult at Brazzaville University Hospital, leading to his admission to the department.

Clinical examination revealed a particular morphotype with distinct facial features and short stature. The patient was afebrile, with a heart rate of 88 bpm, respiratory rate of 16 breaths per minute, and pulse oximetry saturation in ambient air of 99%. Body mass index was 26.6 kg/m^2. Additionally, there was widening of the arterial pulse pressure with blood pressure at 130/55 mmHg, and palpable thrill in the left parasternal area. Heart sounds were regular, with an intense steam-jet systolic murmur in the mesocardiac area radiating in a wheel-spoke pattern, and a 3/6 soft, high-pitched diastolic murmur of aortic insufficiency. There were no signs of heart failure. Examination of other systems was unremarkable.

PARACLINICAL FINDINGS

Chest radiography showed cardiomegaly with a cardiothoracic ratio of 0.58.

Transthoracic echocardiography (TTE) (Figure 2): demonstrated a restrictive perimembranous VSD (5 mm on the left ventricular side and 4 mm on the right ventricular side) with left-to-right shunt (peak pressure gradient: 101 mmHg). The left ventricle was dilated at 64 mm; left atrium at 15.34 cm^2, left ventricular ejection fraction at 68%. Color Doppler showed systolic flow through the VSD. Moderate grade III aortic insufficiency (AI) was present.

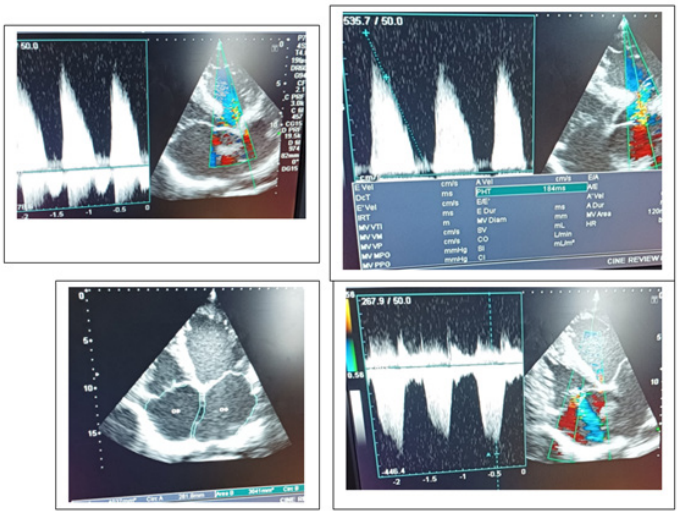

Figure 2: Demonstrated a restrictive perimembranous VSD (5 mm on the left ventricular side and 4 mm on the right ventricular side) with left-to-right shunt (peak pressure gradient: 101 mmHg). The left ventricle was dilated at 64 mm; left atrium at 15.34 cm^2, left ventricular ejection fraction at 68%. Color Doppler showed systolic flow through the VSD. Moderate grade III aortic insufficiency (AI) was present.

Therapeutic management

Symptomatic medical treatment based on angiotensin-converting enzyme inhibitors and nitrate derivatives was administered pending indicated surgery, which would consist of surgical repair of the VSD and aortic valve repair or aortic valve replacement. However, this management cannot be performed in our region due to lack of cardiovascular surgery facilities. Nevertheless, the patient is regularly followed up with a fairly stable hemodynamic status and is awaiting medical evacuation for surgery.

DISCUSSION

In the literature, it is commonly accepted that subcrystalline VSDs are complicated by AI five times more often than in patients with perimembranous VSD [11]. Laubry-Pezzi syndrome represents a progressive pathological process, often developing in adolescence or early adulthood [6,12]. Although many patients are asymptomatic in childhood, progressive exertional dyspnea, palpitations, and heart murmurs may develop as AI worsens [2,12]. The insidious nature of its progression often delays diagnosis and worsens prognosis [13,14].

Epidemiologically, a well-documented male predominance is noted, consistent with our observation [15-17]. Additionally, the literature

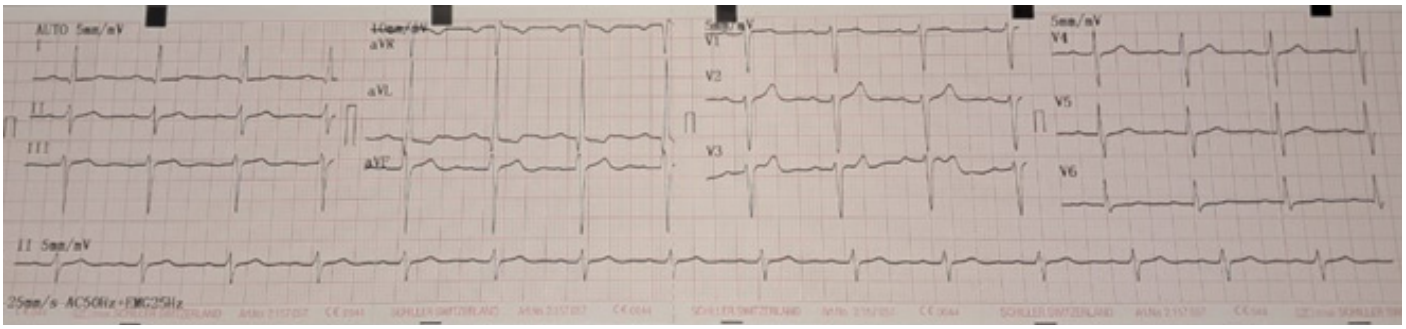

Figure 1: Electrocardiogram showed sinus rhythm with left ventricular hypertrophy.

reports that older patients presented more severe AI. Diagnosis relies primarily on TTE, which allows visualization of the VSD, the prolapsing cusp involved, and the severity of aortic regurgitation [18]. However, images are clearer with transesophageal echocardiography.

Although considered a rare condition, aortic valve prolapse and its corollary, aortic insufficiency, are more severe when the disease is discovered late in adulthood. The most frequently affected cusp is the right coronary cusp, although the non-coronary cusp may also be involved [6]. In our observation, the VSD on echocardiographic imaging was of perimembranous type with significant valvular insufficiency, resulting in left ventricular volume overload, progressive left ventricular dilatation, and no systolic dysfunction. Computed tomography angiography to determine the extent of the defect and plan surgical intervention was not performed. Furthermore, AI can eventually progress to severe systolic dysfunction, threatening the patient's vital prognosis. Failure to rapidly close the VSD can lead to irreversible valvular lesions, requiring cardiovascular surgery, which is difficult in our practice context.

Indeed, early closure of the VSD in childhood is paramount to prevent the onset or worsening of aortic insufficiency. Hence the need for diagnosis and surgical intervention that must address both surgical repair of the VSD and the aortic valve. This may therefore involve either surgical repair or aortic valve replacement. Note that there is no consensus on the optimal timing of surgical intervention in Laubry-Pezzi syndrome. However, early closure of the VSD, before the appearance of significant AI, is generally recommended [21]. In young patients, valve repair or radical repair with valve preservation is preferred to preserve the native valve and avoid lifelong anticoagulation [19-21]. Surgical treatment of Laubry-Pezzi syndrome is generally satisfactory [21]. In the absence of early surgery, dreaded complications may appear, such as progressive aortic valve dysfunction and rarely infective endocarditis. Furthermore, at an advanced stage of the disease, due to structural abnormalities of the sinus and ascending aorta, aneurysmal dilatation of the aortic root and ascending aorta is possible [19].

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