

Case Report

Lutembacher syndrome variant: Rheumatic heart disease involving all four valves and associated with an atrial septal defect in a child

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ABSTRACT

Rheumatic heart disease (RHD) is the most common cause of acquired heart disease in children and young adults in developing countries. It results from throat infection with group A beta hemolytic streptococcus that proceeds to acute rheumatic fever (ARF). We report a 13 years old girl from Darfur presenting with recurrent acute rheumatic fever for 4 years that led to affection of all her heart valves with severe mitral and tricuspid regurgitation together with moderate pulmonary and mild aortic valve regurgitation. There was an associated atrial septal defect (Lutembacher syndrome variant). The disease was severe and led to cardiogenic shock and death while awaiting surgery. The case highlights the impact of RHD on young people and the need to implement control programs for RHD in Sudan.

Keywords:

Rheumatic Heart Disease, Quadrivalvular, Atrial Septal Defect, Lutembacher syndrome variant.

INTRODUCTION

Rheumatic heart disease (RHD) is a non-suppurative complication of throat infection with group A beta hemolytic streptococci. It is considered the commonest cause of acquired heart disease in young people worldwide, with a high burden in Sub Saharan Africa as well as in Sudan [1,2]. The most common valves affected are the mitral and aortic valves; tricuspid valve is less commonly affected while pulmonary valve affection is considered very rare [3,4].

Presentation with severe and multiple valve affection is common in Sudanese children leading to significant morbidity and mortality. Although open heart surgery is available, only about 7% of patients reach the surgeon due to financial and technical constrains [5].

We are presenting a child with all 4 valves affected with RHD associated with an atrial septal defect (ASD), a variant of Lutembacher's syndrome (LS). This case illustrates the need to implement prevention programs to control RHD especially in remote areas and the fatal outcome emphasizes the importance of having prompt surgical treatment for such cases.

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CASE REPORT

A 13 years old girl from Western Darfur presented with shortness of breathing on mild exertion (New York Heart Class III) and migratory knee joint pain which started 4 years ago and recurred at the time of presentation. She was managed by traditional healers. Physical examination revealed a weight of 22 kilograms (4 standard deviations below the mean for age), respiratory rate of 36/min and heart rate of 90 b/min. She was in congestive heart failure, the liver was 8 centimetres below the right costal margin with a span of 13 centimetres, the jugular venous pressure was 12 centimetres. There were pansystolic and mid diastolic

murmurs both at the apex and the tricuspid areas. Blood Pressure was 93/55 mm Hg. Joint examination was normal.

Complete blood count showed a total white blood cells of 7800/cubic millimetre, haemoglobin 9.2 gram/deciliter with normal other indices. The anti-streptolysin O titer was 323 IU/ml and C-reactive protein was positive. Chest X ray showed biventricular and left atrium enlargement. Electrocardiogram showed sinus rhythm, prolonged PR interval of 0.20 seconds, biatrial and biventricular enlargement (Figure 1).

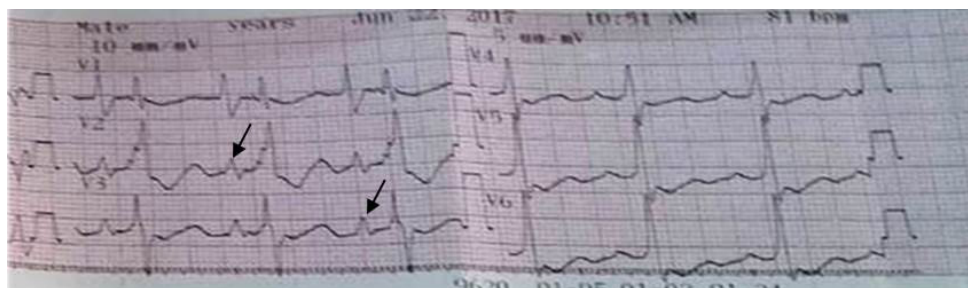


Figure 1- ECG showing prolonged PR interval and marked biatrial and biventricular enlargement (arrows point to P waves) and biventricular enlargement (ECG voltage) is half standard.

Echocardiogram showed that all the 4 heart valves have nodular thickening of their leaflets. The mitral valve posterior leaflet has restricted motion and there is severe mitral regurgitation with a jet vena contracta of 9 mm, the jet is filling 80% of the left atrium with flow reversal in pulmonary veins. The tricuspid leaflets were malcoapting with severe regurgitation, the jet vena contracta was 8 mm and it was filling 70% of the right atrium (Figure 2).

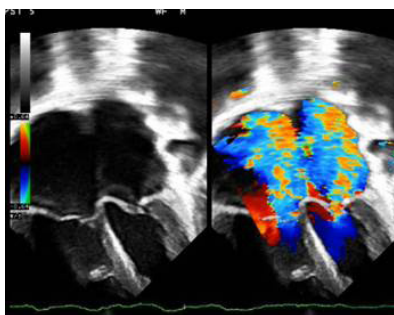


Figure 2- Four-chamber view showing thickened mitral and tricuspid valves (left) and severe mitral and tricuspid regurgitation (Color Doppler, right).

Both left and right atria were dilated. The pulmonary valve leaflets showed abnormal coaptation with moderate pulmonary regurgitation (Figure 3).

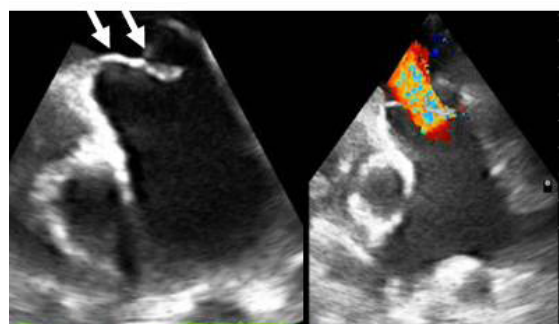


Figure 3- Parasternal short axis view of the pulmonary valve showing thick deformed leaflets (left) and moderate pulmonary regurgitation (Color Doppler, right).

The estimated pulmonary artery pressure from tricuspid and pulmonary regurgitations was 40/25 mm Hg. There was mild aortic regurgitation. (Figure 4).

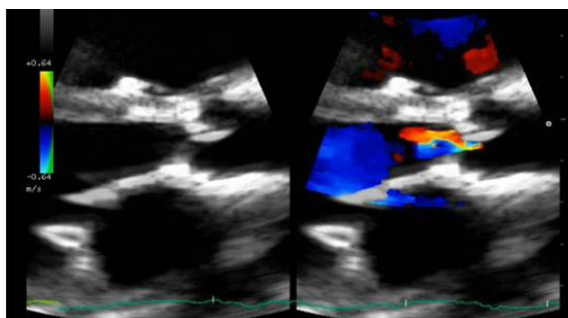


Figure 4- Parasternal long axis view showing thick malcoapting aortic valve (left) and mild aortic regurgitation shown with color Doppler (right).

The left ventricle end diastolic dimension was 41.4 mm and ejection fraction was 66%. There is a secundum atrial septal defect measuring 10 mm with left to right shunt (Figure 5).

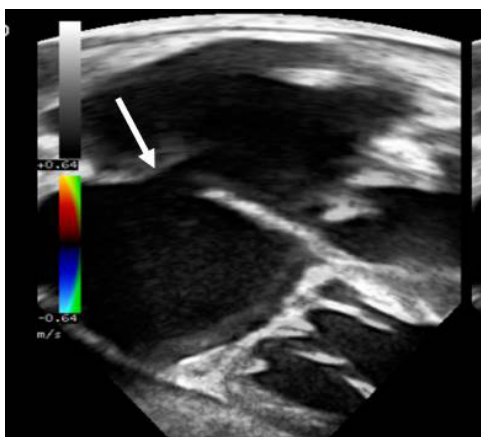


Figure 5- Subcostal coronal view showing an atrial septal defect with a flap valve of fossa ovalis (arrow).

The patient was diagnosed as acute rheumatic fever (ARF) recurrence as she was fulfilling the Jones Criteria with 2 major criteria (arthralgia and carditis) and minor criteria of positive C-reactive protein together with a positive ASO titre. She was started on prednisolone 2 milligrams per kilogram per day, intramuscular benzathine penicillin, furosemide, spironolactone, digoxin and captopril and a surgical consultation was made. On the next day, she developed hypotension necessitating the use of dopamine and dobutamine infusions at a dose of 10 micrograms per kilogram per minute each. She continued to deteriorate over the next 2 days and ultimately developed cardiac arrest and did not survive resuscitation.

DISCUSSION

The late presentation of this patient, 4 years after initial episode of ARF, led to a severe form of RHD affecting all 4 valves. It is likely that she had multiple recurrences of ARF leading to progressive involvement of all her valves. This is a rare presentation of RHD that has been reported in adults but to our knowledge, not in pediatric age [6,7,8]. It reflects the weakness of primary health care system in rural Sudan as well as the lack of public awareness about the early manifestations of the disease. Such cases highlight the need to implement programs for primary and secondary prevention in these areas.

The classical LS is a combination of an ASD with mitral stenosis, typically of rheumatic aetiology [9]. Currently, any combination of mitral valve disease (stenosis or regurgitation) and ASD is referred to as LS [10]. The ASD is thought to be secondary to the high left atrial pressure; in patients who underwent balloon mitral commissurotomy, it is usually iatrogenic. In this child with quadrivalve RHD, the hemodynamic effect of ASD may not be of major significance, however, it needs to be closed at the time of surgery.

Management of such patients is challenging when surgical treatment is not immediately available. She presented in a late stage and developed cardiogenic shock at a stage where her cardiac reserves were not expected to compensate. Successful surgical replacement of mitral valve and aortic valves together with repair of tricuspid valve had been reported, however, the severe nature of the disease as well as the social constraints may compromise post-operative follow up in patients coming from rural areas [11]. Such patients will need lifelong anticoagulation, which requires a high level of compliance with medications as well as with international normalization ratio (INR) monitoring. Compliance with penicillin prophylaxis is crucial especially in patients who still have an unaffected valve. In Sudan, compliance with penicillin prophylaxis was found to be only 51% [5]. This lack of compliance needs to be addressed when we implement programs for improving public awareness about this disease. All these factors contribute to the guarded prognosis of such patients

even when surgery is timely done. Therefore, all efforts should be directed to early diagnosis and treatment of bacterial pharyngitis (primary prevention) and ARF (secondary prevention) as well as strengthening surgical facilities (tertiary prevention) in order to prevent premature cardiac death from RHD.

CONCLUSION

This case represents an advanced, fatal pattern of RHD in a child from Western Sudan associated with LS. An adverse outcome is the result of late presentation and the unavoidable delay in surgical treatment.

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