CASE REPORT

Juvenile rheumatic mitral stenosis in association with perimembranous ventricular septal defect in a 3-year old boy

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SUMMARY

Rheumatic mitral stenosis (MS) is a well-known entity in the developing world. Though rarely seen, MS can present at a very young age with severe symptoms. We describe here an unusual association of a large perimembranous ventricular septal defect with juvenile rheumatic MS leading to severe pulmonary hypertension and heart failure in a very young child.

BACKGROUND

Rheumatic fever and rheumatic heart disease (RHD) continues to be a significant cause of morbidity and mortality in developing countries including India.¹ Rheumatic mitral stenosis (MS) is a common problem in young children and adolescents and it can often lead to severe symptoms.² Recurrent and severe episodes of rheumatic fever can lead to early development of RHD that may be confused with congenital causes of mitral valve disease in the young population. The increasing use of echocardiographic screening has led to early detection of congenital heart disease. The association of a congenital atrial septal defect with acquired rheumatic MS is a well-known entity, but the presence of a congenital ventricular septal defect (VSD) with severe rheumatic MS is very rare.

CASE PRESENTATION

A 3-year-old boy presented to us with history of recurrent lower respiratory tract infections that required hospitalisation with episodes of sore

throat and poor weight gain since 6 months of age and progressive severe dyspnoea with orthopnoea for the past 3 months. His medical records showed that he had suffered from an episode of rheumatic fever at 2 years of age that was diagnosed at a paediatric centre with clinical features of fever and carditis with evidence of raised erythrocyte sedimentation rate and antistreptolysin-O titre. The echocardiographic examination at that time showed mitral leaflets thickening with no MS and mild mitral regurgitation with large VSD. The child's parents were advised operative repair of VSD and child was discharged on penicillin prophylaxis after having recovery at the paediatric centre. The current physical examination showed tachycardia, tachypnoea and the presence of basal rales in the chest. The precordial examination revealed a grade 3/6 pansystolic murmur at the left parasternal area and mid-diastolic murmur at the apex with a loud pulmonary component of the second heart sound.

INVESTIGATIONS

ECG showed sinus tachycardia with left atrial enlargement. The chest x-ray posteroanterior view showed pulmonary plethora. The two-dimensional transthoracic echocardiogram in apical four-chamber view showed a large (18 mm) perimembranous VSD (figure 1A and video 1) and the left to right shunt across defect was confirmed on a colour Doppler (figure 1B and video 2). The apical four-chamber view showed severe thickening and

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Figure 1 (A) Two-dimensional transthoracic echocardiogram in apical four-chamber view showing a large perimembranous VSD. (B) Tilted parasternal long axis view showing left to right shunt across VSD. LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle; VSD, ventricular septal defect.

Video 1 Two-dimensional transthoracic echocardiogram (apical four-chamber view) showing large septal defect.

Video 3 Two-dimensional echocardiogram (apical four-chamber view) showing thickening of mitral leaflets.

Video 2 Two-dimensional echocardiogram colour doppler (tilted parasternal long axis view) showing flow across the ventricular septal defect.

Video 4 Two-dimensional echocardiogram colour doppler showing turbulent mitral stenotic jet.

doming of the mitral valve leaflets (figure 2A and video 3) with turbulent mitral stenotic colour jet arising at the mitral valve (figure 2B and video 4). Continuous-wave Doppler across the mitral valve showed the mean gradient of 15 mm Hg and mitral valve area of 0.7 cm² by pressure half-time method (figure 3A), which was also confirmed by planimetry. The mitral subvalvular apparatus and chordae showed severe thickening (figure 3B and video 5). The patient also had mild tricuspid regurgitation and pulmonary hypertension (estimated right ventricular systolic pressure 96 mm Hg). His clinical condition was initially

stabilised by decongestion with diuretics, bed rest and supportive therapy. Subsequently he underwent cardiac catheterisation which confirmed the large VSD with left to right shunt and showed a mean transvalvular mitral gradient of 16 mm Hg with pulmonary artery systolic pressure of 90 mm Hg and pulmonary vascular resistance of 6 wood units indexed to body surface area. There was a significant fall in pulmonary vascular resistance (from 6 to 4 wood units) on oxygen administration during cardiac catheterisation, thus decision for surgical correction was made.

Figure 2 (A) The apical four-chamber view showing severe thickening and doming of mitral valve leaflets. (B) Colour Doppler echocardiogram revealing the turbulent MS jet arising at the mitral valve. LA, left atrium; LV, left ventricle; MS jet, mitral stenotic; RA, right atrium; RV, right ventricle.

Figure 3 (A) Continuous-wave Doppler across mitral valve showing the mean gradient of 15 mm Hg and mitral valve area of 0.7 cm² by pressure half-time method. (B) The tilted parasternal long axis view showing severe thickening of the mitral subvalvular apparatus and chordae. A, aorta; LA, left atrium; LV, left ventricle; RV, right ventricle.

TREATMENT

The patient had surgery under cardiopulmonary bypass. During operative repair, inspection of the mitral valve revealed thickened, scarred, redundant leaflets with 'fish mouth' appearance of the valve, commissural fusion and thickened subvalvular apparatus. The VSD was closed with a Dacron patch and open mitral commissurotomy was performed.

OUTCOME AND FOLLOW-UP

The patient had good postoperative recovery and he was discharged on diuretics and three weekly injections of benzathine penicillin. He is asymptomatic at 2 months of follow-up.

DISCUSSION

Acute rheumatic fever is the result of an autoimmune response that occurs after few weeks of throat infection caused by group A β -haemolytic streptococci.³ RHD is a late sequelae of rheumatic fever due to immune-mediated damage to the cardiac valves, and the commonest valve affected is mitral valve.⁴ Congenital heart disease is usually the result of developmental defects at the early embryonic stage. It is not clear whether the occurrence of RHD in congenital heart disease is merely a coincidence or if congenital heart disease predisposes to RHD.^{5, 6} The association of both the entities has been described in the literature.

Video 5 Two-dimensional echocardiogram (parasternal long axis view) showing severe thickening of mitral subvalvular apparatus and chordae.

Severe rheumatic MS is rampant in children and young adults in developing countries and around 25% of patients are younger than 20 years of age.^{7, 8} This entity commonly known as juvenile MS⁸ is associated with a high incidence of severe pulmonary arterial hypertension with congestive heart failure. In juvenile MS, there is severe involvement of subvalvular apparatus with more critical stenosis of the mitral valve. The probable aetiologies of this condition are continued smouldering rheumatic activity, tissue response to multiple attacks of rheumatic fever and hypersensitive reaction of the pulmonary vasculature to the fulminating rheumatic process.⁸ Approximately 10% of the patients with juvenile rheumatic MS present before 12 years of age and associated with rapid haemodynamic progression with severe symptoms.⁹ The age is also an important factor, as occurrence of rheumatic fever below 5 years of age increases the risk of severe cardiac involvement and chronic RHD due to increased severity of the valvulitis during acute episodes of rheumatic fever.¹⁰

There are reported series of juvenile rheumatic MS in less than 10 years of age group and its successful treatment with balloon mitral commissurotomy.^{11, 12} The rheumatic fever below 5 years of age is uncommon because of incomplete maturation of the immune system and approximately 5% of patients with rheumatic fever belong to this age group. However, rare cases occurring at as young as 2 years of age are reported.¹³ There are also sporadic case reports of severe rheumatic MS and its interventional treatment below 5 years of age.¹⁴ Our case is probably the youngest case of juvenile rheumatic MS with VSD, however unavailability of histopathological evidence to confirm rheumatic aetiology is the limitation of our case.

Learning points

- ▶ Rheumatic mitral stenosis is a common problem in developing countries but rarely may it present as juvenile rheumatic mitral stenosis at a very young age with heart failure.
- ▶ Although congenital mitral stenosis is common in association with other congenital cardiac defects, but the possibility of rheumatic mitral stenosis should always be ruled out in these patients.
- ▶ Transthoracic echocardiography is an important modality to differentiate rheumatic from congenital causes of mitral stenosis, thus careful evaluation is mandatory.

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Competing interests None.

Patient consent Obtained.

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REFERENCES

- 1 Kumar RK, Tandon R. Rheumatic fever & rheumatic heart disease: the last 50 years. *Indian J Med Res* 2013;137:643–58.
- 2 Rheumatic fever and rheumatic heart disease. *World Health Organ Tech Rep Ser* 2004;923:1–122, back cover.
- 3 Bisno AL. Group A streptococcal infections and acute rheumatic fever. *N Engl J Med* 1991;325:783–93.
- 4 Otto CM, Bonow RO. Braunwald's heart disease: A textbook of cardiovascular medicine. 9th edn. Philadelphia: Saunders Elsevier, 2012.
- 5 Bokhandi SS, Tullu MS, Shaharao VB, *et al.* Congenital heart disease with rheumatic fever and rheumatic heart disease: a coincidence or an association? *J Postgrad Med* 2002;48:238–8.
- 6 Hollman A, Hamed M. Mitral valve disease with ventricular septal defect. *Br Heart J* 1965;27:274–85.
- 7 Shrivastava S, Tandon R. Severity of rheumatic mitral stenosis in children. *Int J Cardiol* 1991;30:163–7.
- 8 Roy SB, Bhatia ML, Lazaro EJ, *et al.* Juvenile mitral stenosis in India. *Lancet* 1963;2:1193–5.
- 9 Gotsman MS, Van der Horst RL. Surgical management of severe mitral valve disease in childhood. *Am Heart J* 1975;90:685–7.
- 10 Ullah M, Sultan M, Akbar H, *et al.* Percutaneous transluminal mitral commissurotomy for rheumatic mitral stenosis in a 5-year-old child. *Pediatr Cardiol* 2012;33:814–17.
- 11 Kothari SS, Ramakrishnan S, Kumar CK, *et al.* Intermediate-term results of percutaneous transvenous mitral commissurotomy in children less than 12 years of age. *Catheter Cardiovasc Interv* 2005;64:487–90.
- 12 Krishnamoorthy KM, Tharakan JA. Balloon mitral valvulotomy in children aged < or = 12 years. *J Heart Valve Dis* 2003;12:461–8.
- 13 Nirmal H, Vani P, Chhabra M, *et al.* Rheumatic fever in a 2-year-old child. *Pediatr Cardiol* 2008;29:160–2.
- 14 Kapoor A, Moorthy N, Kumar S. Inoue balloon mitral valvotomy in a 4-year-old boy: to treat fulminant rheumatic mitral stenosis. *Tex Heart Inst J* 2012;39:108–11.

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