



## Isolated Congenital Mitral Stenosis

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Congenital MS, a rare entity, takes several forms. These include hypoplasia of the mitral valve annulus, mitral valve commissural fusion, double orifice mitral valve, shortened or thickened chordae tendinae and parachute mitral valve. The most common associated malformations are coarctation of the aorta, valvular aortic stenosis and subvalvular aortic stenosis. The association of multiple levels of left-sided inflow and outflow tract obstruction is termed the “Shone’s Complex”(1-3). Here, we describe a neonate with isolated congenital mitral stenosis.

A 25 day-old female was admitted to a tertiary care NICU with congestive heart failure. No history of cyanosis was present. The patient was full term, born by vaginal delivery and did not require any resuscitation. The Apgar scores were 9, 9 and 10 at 1min, 5min and 10 minutes respectively. Antenatal history was uneventful and there was no family history of diabetes, hypertension, seizure disorders, mental retardation or congenital heart disease.

On physical examination, the neonate was found to have dyspnea and pallor. She weighted 3500gm. Her respiratory rate was 50/min. Her cardiac rhythm was constant at 160 bpm. Her pulse was palpable in all the 4 limbs with systemic blood pressure of 65 / 40mmHg. Examination of the cardiovascular system revealed a grade IV/VI pansystolic murmur in the mitral region and conducted widely to all areas of the precordium. Oxygen saturation was normal. The second heart sound had a loud pulmonary component. The liver was palpable 3 cm below the right costal margin. The rest of the systemic examination was normal.

The chest X-ray showed a cardiomegaly and accentuated pulmonary plethora. The electrocardiogram revealed sinus rhythm and was suggestive of both left atrium and right ventricular hypertrophy. The echocardiogram was diagnostic and showed dilatation of left atrium and right atrium. Anterior mitral valve had a

hockey-stick appearance and mitral valve area was considerably reduced and measured 0.7cm/m<sup>2</sup> on the parasternal short axis view. Severe mitral regurgitation with moderate tricuspid regurgitation was noted. Severe pulmonary hypertension was present with a doppler gradient of 70mmHg. A patent foramen ovale of 3mm size was noted with a left to right shunt. The final diagnosis was isolated Congenital Mitral Stenosis.

Congenital mitral stenosis occurs as an isolated malformation or in combination with additional cardiac lesions. Isolated congenital mitral stenosis is a rare malformation. In combination with additional obstructive, left-sided cardiac malformations, mitral stenosis occurs more frequently (4). Congenital mitral valvular lesions are diverse and rare, found in 0.6% of autopsied patients with congenital heart disease and 0.21%-0.42% of clinical series (1-3). Cardiac anomalies frequently coexist with this malformation. Coexisting intracardiac lesions may mask or unmask mitral valve disease (MVD) and may frequently increase the complexity of surgical repair. The etiology of congenital MS remains unknown. However, prevalence of MS in offspring of family members (especially the mother) with left ventricular outflow tract obstruction is relatively high.

Patients with severe MS may present with respiratory distress from pulmonary edema shortly after birth if a significant atrial septal communication does not exist. Patients with mild-to-moderate MS present after the neonatal period with signs of low cardiac output and RV failure such as pulmonary infections, failure to gain weight, exhaustion and diaphoresis with feeding, tachypnea and chronic cough (5). Physical examination in severe MS reveals diminished peripheral perfusion and pulses, palpation of a RV impulse (when pulmonary hypertension is present), soft S1 in the presence of heart failure and diminished left ventricular filling. Holodiastolic murmur with presystolic accentuation is best heard at the apex

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with severe pulmonary hypertension (6). Ito T *et al* concluded that the combination of the accessory mitral valve and left-to-right shunt due to ventricular septal defect or patent ductus arteriosus might have led to a critical hemodynamic condition due to relative mitral stenosis in the neonatal period with the decrease in pulmonary vascular resistance (7).

Echocardiography is the most important diagnostic tool to evaluate patients with MS. Cardiac catheterization may be used to obtain direct intracardiac pressure measurements, the mitral valve gradient, pulmonary vascular resistance and systemic cardiac output (8). Asymptomatic patients with mild MS require no significant therapy. Congestive heart failure is treated with loop diuretics plus potassium-sparing diuretics. Digoxin may improve right ventricular function in the setting of pulmonary hypertension. Surgical options depend upon specific mitral valve pathology; mitral valve repair and mitral valve replacement with mechanical valve or bioprosthesis.

Mitral valve replacement is best avoided in infants and small children because of frequent size mismatch between the smallest mechanical valves and the hypoplastic mitral valve annulus. In addition, somatic growth in children leads to the need for subsequent mitral prosthesis replacement (9). Morre *et al* (10) reported that infants with severe CMS have 2-year mortality rates approaching 40%, regardless of the treatment modality. Balloon dilation significantly reduces the transmitral gradient in majority, but symptomatic improvement persists in only 40%. Whereas, Duncan *et al* (11) believe that prosthetic valve replacement could become a viable therapy for the neonate with severe mitral valve abnormalities. Untreated newborns with severe MS have a grim prognosis. Surgical intervention is ideally avoided for as

long as possible. Mechanical mitral valve replacement in a small infant or child is a high-risk procedure and carries a guarded prognosis. Operative results and long-term outcome are extremely variable and highly dependent on coexisting abnormalities (6,12).

#### References

1. Collins-Nakai RL, Rosenthal A, Castaneda AR, Bernhard WF, Nadas AS. Congenital mitral stenosis. A review of 20 years' experience. *Circulation* 1977; 56: 1039-47.
2. Carpentier A, Branchini B, Cour JC, *et al*. Congenital malformations of the mitral valve in children. Pathology and surgical treatment. *J Thorac Cardiovasc Surg* 1976; 72: 854-66.
3. Gersony WM. Ventricular septal defect and left sided obstructive lesions in infants. *Curr Opin Pediatr* 1994; 6: 596-99 .
4. Vogt J, Eckhardt R, Beuren AJ. Congenital mitral stenosis. Report on 33 patients. *Z Kardiol.* 1975 ;64:516-35.
5. Bruce CJ, Nishimura RA. Clinical assessment and management of mitral stenosis. *Cardiol Clin* 1998 ; 16: 375-403.
6. Lamberti JJ, Mainwaring RD. Congenital anomalies of the mitral valve. In: *Pediatric Cardiac Surgery*. St. Louis, Mo: *Mosby-Year Book*; 1994.pp. 459-73.
7. Ito T, Okubo T. A neonate with mitral stenosis due to accessory mitral valve, ventricular septal defect, and patent ductus arteriosus: changes in echocardiographical findings during the neonatal period. *Heart Vessels* 2002 ;17:83-85.
8. Bruce CJ, Nishimura RA. Newer advances in the diagnosis and treatment of mitral stenosis. *Curr Probl Cardiol* 1998 ; 23: 125-92.
9. Castañeda AR, Jonas RA, Mayer JE. Pediatric Valve Replacement. In: *Cardiac Surgery of the Neonate and Infant*. Philadelphia, Pa: *WB Saunders*; 1994.pp. 387-95.
10. Moore P, Adatia I, Spevak PJ, Keane JF, Perry SB, Castaneda AR, Lock JE. Severe congenital mitral stenosis in infants. *Circulation* 1994 ;89:2099-106.
11. Duncan WJ, Bharadwaj B, Tyrrell MJ. Mitral valve replacement in the neonate: a report of two cases. *Pediatr Cardiol* 1984;5:307-11.
12. Uva MS, Galletti L, Gayet FL: Surgery for congenital mitral valve disease in the first year of life. *J Thorac Cardiovasc Surg* 1995 ; 109: 164-74.

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