Association between congenitally quadricuspid aortic valve and mitral valve prolapse

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We describe transthoracic echocardiograms in three patients with combined quadricuspid aortic valve and prolapsing mitral valve. None had symptoms of cardiac dysfunction. Two patients had precordial murmurs. A third patient was referred for evaluation of infective endocarditis. The fact that a quadricuspid aortic valve is clearly a congenital anomaly supports the view that mitral valve prolapse is a congenital anomaly that may be more strongly associated with quadricuspid aortic valves than once thought.

The quadricuspid aortic valve (QAV), although uncommon, is clearly recognized as a congenital defect. Mitral valve prolapse (MVP), in contrast, is not often thought of as a congenital anomaly. The occurrence of both in a series of patients supports the view that MVP and QAV may be associated congenital anomalies. A description of findings in these three patients and a summary of previously reported such cases are the purposes of this report.

DESCRIPTION OF PATIENTS

Pertinent clinical features in each of the three patients are summarized in *Table 1*. Echocardiograms were performed in cases 1 and 2 to evaluate asymptomatic precordial murmurs, and in case 3, to evaluate infective endocarditis. These studies disclosed a QAV with mild aortic regurgitation (*Figures 1a, 1b*). All patients had normal left ventricular systolic function and normal chamber sizes. The three patients also had MVP but with varying degrees of mitral regurgitation (MR) (*Figure 1c, 1d, 2d*). There was no evidence of vegetation on any of the valves. No patient required surgery for their dysfunctional valves. None had aortic stenosis, coronary anomalies (*Figure 2c*), or a family history of congenital heart or connective tissue diseases.

DISCUSSION

Each of these asymptomatic patients had both a QAV and MVP. The occurrence of both these valvular anomalies in the same patient supports the observation that they are of congenital origin and may be associated. Five other cases of patients with QAV and MVP have been reported (1–5). To our knowledge, such a strong association between QAV and MVP in a case series has not been described.

Table 1. Features of three patients with quadricuspid aortic valve and mitral valve prolapse

Variable	Case 1	Case 2	Case 3
Age (years)	41	18	54
Gender	Male	Female	Male
Type of QAV	4E	4E	3E1U
Type of murmur	MR	AR	None
Aortic regurgitation (0–4+)*	1+	1+	1+
Mitral regurgitation (0–4+)*	4+	1+	1+
Blood pressure (mm Hg)	120/80	140/80	185/90
Heart rate (beats/minute)	60	70	90
Body mass index (kg/m ²)	21	23	20
Left ventricular end-systolic diameter (cm)	3.9	3.0	3.7

*Valvular dysfunction scale: 0, none; 1–2+, trace/mild; 3+, moderate; 4+, severe. 4E indicates four equally sized cusps; 3E1U, three larger equal cusps and one smaller cusp; MR, mitral regurgitation; AR, aortic regurgitation.

Table 2 summarizes the clinical findings in five previously reported cases. This review reveals a slight male predominance. The two most common types of QAV were Hurwitz and Roberts type A (four equally sized cusps) and type B (three larger equal cusps and one smaller cusp) (6). Advanced age did not correlate with worsened aortic regurgitation (AR) but did correlate with worsened MR. Only one patient had aortic stenosis and severe AR requiring surgery. Two cases had severe MR requiring surgery.

The occurrence of MVP in patients with the more common congenital aortic valvular anomaly, the bicuspid valve, has been described previously. In a recent study by Roberts

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Table 2. Summary of reported cases of combined quadricuspid aortic valve and mitral valve prolapse

Reported cases	Year of publication	Age of diagnosis (yrs)	Gender	QAV type	AR*	AS*	MR*	Method of diagnosis	Valve replaced
Yildirim et al (1)	2008	10	M	3E1U	4+	4+	0	OR	AV
Di Pino et al (2)	2008	11	F	3E1U	0	0	1+	TTE	None
Ozlu et al (3)	2008	72	M	4E	3+	0	4+	TTE	MV
Cooke et al (4)	2000	75	M	4E	1+	0	4+	TEE	MV
Jagannath et al (5)	2011	75	F	2E2e	3+	0	3+	TTE	None

*Valvular dysfunction scale: 0, none; 1–2+, trace/mild; 3+, moderate; 4+, severe.

2E2e indicates two equal larger cusps and two equal smaller cusps; 3E1U, three larger equal cusps and one smaller cusp; 4E, four equally sized cusps; AR, aortic regurgitation; AS, aortic stenosis; AV, aortic valve; MR, mitral regurgitation; MV, mitral valve; OR, operating room/at surgery; TEE, transesophageal echocardiogram; TTE, transthoracic echocardiogram.

Figure 1. Case 1 transesophageal echocardiogram. Short-axis view of the type A quadricuspid aortic valve (a) closed and (b) open. (c and d) Midesophageal view of the mitral valve prolapse (arrow).

et al (7), four patients who had a congenitally bicuspid aortic valve and MVP required replacement of both valves. Our review, however, demonstrated that no patients with a combined QAV and MVP required surgery of both valves.

In patients with QAV, the attention is usually focused on that valve in particular, and so the association with MVP may not have been as strongly linked. Due to the rarity of both anomalies, it is difficult to judge the exact level of association, but it may be stronger than previously noted.

Supplementary material

Two videos are available online. The first, available at www.BaylorHealth.edu/Proceedings/26_3_Schussler. Video1, shows a myxomatous mitral valve in a patient with a quadricuspid aortic valve. The second, available at www.BaylorHealth.edu/Proceedings/26_3_Schussler. Video2, shows a quadricuspid aortic valve with normal function, discovered as an incidental finding in a patient with degenerative myxomatous disease of the mitral valve.

Figure 2. Case 2. **(a, b, c)** Cardiac computed tomography scan: **(a)** short-axis view of the type A quadricuspid aortic valve; **(b)** view of the quadricuspid aortic valve with symmetrical cusps (RCC, right coronary cusp; LCC, left coronary cusp) and coronary ostia (arrows); **(c)** view of the coronary arteries revealing no coronary anomalies. **(d)** Transesophageal echocardiogram of the mitral valve prolapse at the midesophageal level.

1. Yildirim SV, Gümüş A, Coşkun I, Türköz R. Quadricuspid aortic valve: a rare cause of aortic regurgitation and stenosis. *Turk J Pediatr* 2008;50(5):500–502.
2. Di Pino A, Gitto P, Silvia A, Bianca I. Congenital quadricuspid aortic valve in children. *Cardiol Young* 2008;18(3):324–327.
3. Ozlü MF, Özcan F, Çağlı K, Gölbaşı Z. Quadricuspid aortic valve in a patient with mitral valve prolapse. *Can J Cardiol* 2008;24(4):e27.
4. Cooke JC, Dupuche DR, Gay TJ. Quadricuspid aortic valve. *Echocardiography* 2000;17(7):699.
5. Jagannath AD, Johri AM, Liberthson R, Larobina M, Passeri J, Tighe D, Agnihotri AK. Quadricuspid aortic valve: a report of 12 cases and a review of the literature. *Echocardiography* 2011;28(9):1035–1040.
6. Hurwitz LE, Roberts WC. Quadricuspid semilunar valve. *Am J Cardiol* 1973;31(5):623–626.
7. Roberts WC, Janning KG, Vowels TJ, Ko JM, Hamman BL, Hebel RF Jr. Presence of a congenitally bicuspid aortic valve among patients having combined mitral and aortic valve replacement. *Am J Cardiol* 2012;109(2):263–271.