Missed Diagnosis

Mitral valve prolapse and occult aortic coarctation

Peter Ludman, Magdi Yacoub* and Mark Dancy

Central Middlesex Hospital, Acton Lane, London NW10 7NS, UK.

Summary: A 22 year old man developed symptoms of left ventricular failure secondary to atrial fibrillation and congenital mitral regurgitation. After operation for mitral valve repair he was unable to be successfully weaned from cardiopulmonary bypass and this was ascribed to poor left ventricular function. He therefore underwent emergency cardiac transplantation but again was unable to be weaned from bypass. At post-mortem examination a previously undiagnosed aortic coarctation was revealed.

The presentation of occult aortic coarctation is discussed, and its association with congenital mitral valve abnormalities reviewed.

Introduction

The presentation of aortic coarctation may be atypical or obscured by other abnormalities. It must, however, be borne in mind in all patients with congenital heart disease particularly if there are unexpected haemodynamic developments. We report a case of occult aortic coarctation whose presence contributed to peri-operative complications during mitral valve repair.

Case report

A 22 year old man was referred to accident and emergency with a one month history of exertion associated central chest pain, shortness of breath and palpitations. As a child he had been diagnosed on clinical grounds alone as having a ventricular septal defect of no haemodynamic significance, and he was not followed up. He had been entirely well until developing his presenting symptoms.

On examination he was not dyspnoeic at rest and not clubbed or cyanosed. He had an irregular pulse, rate 120/min, blood pressure 110/70 mmHg. Jugular venous pressure was elevated at 8 cm. Left ventricular impulse was displaced to the anterior axillary line, 6th intercostal space, and was markedly hyperdynamic with a systolic thrill. There was a loud pansystolic murmur radiating to the axilla. Peripheral pulses were palpable. Res-

piratory system was normal. There was 2 cm smooth hepatomegaly.

ECG showed atrial fibrillation, right axis deviation and left ventricular hypertrophy (Figure 1). Chest radiograph showed cardiomegaly, left atrial enlargement and pulmonary venous hypertension (Figure 2). Investigations revealed no evidence of infective endocarditis.

Echocardiogram showed a dilated left ventricle (end diastolic dimension 7.5 cm – normal range 3.5 to 5.6 cm, end systolic dimension 5.0 cm – normal range 1.9 to 4.0 cm) and a dilated left atrium (5.5 cm – normal range 1.9 to 4.0). There was mild concentric left ventricular hypertrophy (posterior left ventricular wall thickness 1.5 cm – normal range 0.7 to 1.1, interventricular septal thickness 1.2 cm – normal range 0.7 to 1.2). Left ventricular function was moderately impaired and the anterior mitral valve leaflet prolapsed back into the left atrium. Marked mitral regurgitation was confirmed on colour flow Doppler. The aortic valve was bicuspid with no transvalvar gradient.

The patient's ventricular response to atrial fibrillation was reduced to 90/min with digoxin, and his symptoms were relieved. He was referred for reconstructive mitral valve surgery.

At operation the mitral valve ring was markedly dilated resulting in prolapse of the anterior cusp with elongated chordae. The valve itself was not typically floppy but appeared to be stretched secondary to the annular dilatation. Valve competence was achieved by bilateral annuloplasty and fixing part of the anterior cusp to adjoining papillary muscle and free edge of the lateral cusp.

There were immediate problems coming off cardiopulmonary bypass. Cardiac output was extremely poor and aortic balloon counter-pulsation

Correspondence and present address: P. Ludman, M.R.C.P., Cardiovascular Unit, Collier Building, Hammersmith Hospital, Ducane Road, London W12 0HS, UK.

*Present address: National Heart and Lung Institute, London, UK.

Accepted: 9 May 1990

Figure 1 Electrocardiogram at presentation.

Figure 2 Postero-anterior chest radiograph at presentation

provided no useful augmentation. Persistent hypotension unresponsive to inotropic support required re-institution of cardiopulmonary bypass which was then continued until orthotopic cardiac transplantation was performed 2 days later. The new heart was also unable to maintain an adequate cardiac output and became unstable. Attempts at resuscitation failed. Post mortem examination

revealed a preductal aortic coarctation, with the lumen diameter reduced to 6 mm.

Discussion

At presentation as a child with an asymptomatic pansystolic murmur a clinical diagnosis of ventricular septal defect was made. In the light of subsequent events it is likely that the murmur was due to congenital mitral regurgitation. Though no records are available, it must be assumed that comparative measurement of upper and lower limb blood pressure was not made.

At presentation as an adult, peripheral pulses were palpable but no specific examination for radio-femoral delay was made, nor were upper and lower limb blood pressures compared in spite of several cardiological examinations in two different hospitals. There were, however, no clinical features to suggest that coarctation might be present, apart, perhaps, from the bicuspid aortic valve. The patient was normotensive, the loud precordial pansystolic murmur and thrill obscured any other murmur, the patient had a normal body habitus with no disproportionate limb development. Chest radiograph (Figure 2) did not reveal rib notching from dilated intercostal collaterals, and the aortic arch appeared to be of normal configuration.

The undiagnosed aortic coarctation undoubtedly contributed to the patient's poor peri-operative

progress. Had the diagnosis been made, the coarctation could have been corrected first, so reducing left ventricular afterload, and hence the severity of the mitral regurgitation. The value of mitral valve repair could then have been reassessed. After repair of the leaking mitral valve the left ventricle was unable to eject the entire stroke volume through the coarctation. At the time this was attributed to poor left ventricular function (with inaccurate pre-operative assessment of left ventricular function in a patient with severe mitral regurgitation) and as a result cardiac transplantation was performed. Failure of the aortic balloon to augment cardiac output was due to the presence of a coarctation proximal to the balloon.

Congenital mitral regurgitation is an unusual disorder and often associated with other congenital cardiac abnormalities. For this reason pre-operative assessment by cardiac catheter may have been advisable. Nevertheless, as the cardiac catheter might well have been performed from the brachial rather than femoral artery, the aortic coarctation would have remained undiagnosed unless specifically sought.

This case demonstrates how aortic coarctation can be present in the absence of many of the usual associated features. Peripheral pulses are frequently palpable in patients with aortic coarctation. In a review of 65 consecutive referrals¹ in whom a diagnosis of coarctation was later confirmed, 44% had palpable femoral pulses, 16% of which were described as of normal character. In another review,² 25% of patients over 1 year of age had palpable femoral pulses. Twenty per cent of patients under the age of 19 years and 16% of over 19 year olds are normotensive.³ A retrospective analysis of the chest X-ray appearance in 48 patients with aortic coarctation revealed it to be abnormal in 92% of cases. The aortic contour was abnormal in 90% but rib notching obvious in only 23%.⁴

Impaired left ventricular function is common in patients with coarctation. In a study of 11 uncomplicated patients, all had abnormal left ventricular

function.⁵ This was attributed to elevated end systolic wall stress with a resultant increase in myocardial oxygen demand and impaired early diastolic function.

Congenital mitral valve anomalies are rare, and if present are rarely isolated. Coexistent congenital heart disease is present in 50% to 74%.^{6,7} There is a well documented association between stenotic left ventricular inflow abnormalities and aortic coarctation. In post-mortem studies Becker,⁸ and later Rosenquist,⁹ showed a high incidence of (predominantly stenotic) mitral valve malformations (26% and 58% respectively) in patients with coarctation.

The association between mitral regurgitation and coarctation is less firmly based. Freed¹⁰ noted mitral regurgitation in 18 of 861 patients with aortic coarctation (2.1%). Many had abnormalities of leaflets of chordae tendinae. In 15 of the 18 patients the aortic coarctation was repaired: 4 developed less severe mitral regurgitation, in 7 there was no change and 4 deteriorated further. Celano¹¹ found echocardiographic evidence of mitral valve prolapse in 11.3% of a series of 56 patients with aortic coarctation, and 7.5% had mitral regurgitation (all of these showing echocardiographic features of mitral valve prolapse).

The structural abnormalities of the mitral valve leaflets and chordae found in patients with mitral valve prolapse are broadly similar to those described in patients with mitral valve abnormalities and aortic coarctation and as yet there is no evidence for more than a casual association between mitral regurgitation and aortic coarctation. It would seem reasonable to propose that in the presence of coarctation, any tendency for the mitral valve to prolapse may be exaggerated by the high left ventricular afterload.

The diagnosis of coarctation of the aorta must be borne in mind in all patients with congenital heart disease. Although the presentation of aortic coarctation may be atypical or obscured by other conditions, it can always be diagnosed if upper and lower limb blood pressures are compared.

References

- Stafford, M.A., Griffiths, S.P. & Gersony, W.M. Coarctation of the aorta: a study in delayed detection. *Pediatrics* 1982, **69**: 159–163.
- Keith, J.D. Coarctation of the aorta. In: Keith, J.D., Rowe, R.D. & Vlad, P. (eds) *Heart Disease in Infancy and Childhood*. Macmillan, New York, 1978, pp. 736–760.
- Borow, K.M. & Braunwald, E. Congenital heart disease in the adult. In: Braunwald, E. (ed.) *Heart Disease, a Textbook of Cardiovascular Medicine*. W.B. Saunders, Philadelphia, 1988, pp. 976–1008.
- Martin, E.C., Stafford, M.A. & Gersony, W.M. Initial detection of coarctation of the aorta: an opportunity for the radiologist. *Am J Roentgenol* 1981, **137**: 1015–1017.
- Borow, K.M., Colan, S.D. & Neumann, A. Altered left ventricular mechanics in patients with valvular aortic stenosis and coarctation of the aorta. *Circulation* 1985, **72**: 515–522.
- Bloom, K.R. Mitral valve and tricuspid valve stenosis. In: Keith, J.D., Rowe, R.D. & Vlad, P. (eds) *Heart Disease in Infancy and Childhood*. Macmillan, New York, 1978, pp. 802–809.
- Collins-Nakai, R.L., Rosenthal, A., Castenada, A.R., Bernhard, W.F. & Nadas, A.S. Congenital mitral stenosis. A review of 20 years experience. *Circulation* 1977, **56**: 1039–1046.

8. Becker, A.E., Becker, M.J. & Edwards, J.E. Anomalies associated with coarctation of the aorta with particular reference to infancy. *Circulation* 1970, **41**: 1067–1075.
9. Rosenquist, G.C. Congenital mitral valve disease associated with coarctation of the aorta: a spectrum that includes parachute deformity of the mitral valve. *Circulation* 1974, **49**: 985–993.
10. Freed, M.D., Keane, J.F., Van Praagh, R., Castenada, A.R., Bernhard, W.F. & Nadas, A.S. Coarctation of the aorta with congenital mitral regurgitation. *Circulation* 1974, **49**: 1175–1184.
11. Celano, V., Pieroni, D.R., Morera, J.A., Roland, J.-M.A. & Gingel, R.L. Two dimensional echocardiographic examination of mitral valve anomalies associated with coarctation of the aorta. *Circulation* 1984, **69**: 924–932.