Aortic atresia with complete transposition

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SUMMARY Aortic atresia with ventriculoarterial discordance in a three day old neonate was studied by cross sectional echocardiography and the anatomy was confirmed at necropsy.

Aortic atresia with hypoplasia of the ascending aorta and left ventricle nearly always occurs with atrioventricular and ventriculoarterial concordant connections and normally related great arteries.^{1 2} Its association with complete transposition of great arteries has been reported only once in a six year old girl studied by cardiac catheterisation.³ We report the cross sectional echocardiographic and post-mortem findings of a neonate with this rare condition.

Case report

A three day old Chinese female baby was referred because of poor feeding and cyanosis. She was the second child of healthy non-consanguineous parents. The antenatal history was uneventful and there were no early perinatal problems. She weighed 3010 g at birth and had no external malformations. Examination revealed a blue tinge to the skin which increased when she cried and mild dyspnoea with a respiratory rate of 55 breaths per minute. The pulses were weak with a rate of 150 beats per minute and a pressure of 80/60 mm Hg in all limbs. The precordium was hyperactive with a biventricular impulse and the apex beat was displaced to the left midaxillary line. Auscultation showed a loud single second heart sound and a soft grade 2/6 ejection systolic murmur over the upper left sternal border. The lungs were clear and the liver was palpable 3 cm below the right costal margin. A chest x ray showed a cardiothoracic ratio of 0.68 and markedly plethoric lungs. The electrocardiogram showed evidence of right atrial and left ventricular hypertrophy with absence of the R wave in the right precordial leads; the mean QRS axis was 45°. Arterial oxygen partial

pressure in room air was 5.15 kPa and rose to 11.29 kPa after breathing 100% oxygen.

Cross sectional echocardiography was performed with an Advanced Technology Laboratory Mk600 Duplex system machine mounted with a 5 MHz mechanical sector transducer. Standard precordial, subcostal, and suprasternal views were taken.

There was the usual atrial arrangement and a concordant atrioventricular connection. The anterior right sided ventricle was small and had a hypoplastic though patent tricuspid valve with attachments to the interventricular septum (fig 1a). The posterior ventricle was large, smooth walled, and contained two papillary muscles with a bicuspid mitral valve. No ventricular septal defect could be identified. The pulmonary trunk arose from the posterior ventricle and was connected via a large ductus arteriosus to the descending aorta. The ascending aorta was not readily visible from the parasternal long axis view. It could be demonstrated from the apical four chamber projection by clockwise rotation of the transducer (fig 1b) and also from the high parasternal short axis view by rotating the transducer counterclockwise and tilting it towards the right shoulder; it was a small tubular structure running to the right and in front of the pulmonary artery (fig 1c). Its presumed connection with the right ventricle, however, could not be well demonstrated. The hypoplastic aorta was also clearly seen from the suprasternal notch view.

A Doppler flow study with the sample volume placed in the ascending aorta showed that during systole there was retrograde flow from the aortic arch towards the aortic root while during diastole flow was away from the ascending aorta (fig 1d).

Cardiac catheterisation was planned but was postponed because the baby developed diarrhoea after admission. On day 8 she became increasingly dyspnoeic with wet lungs and gallop rhythm. Repeat echocardiography showed progressive deterioration of left ventricular contractions while the ductus re-

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Fig 1 Cross sectional echocardiographic views. (a) Four chamber view showing the hypoplastic right ventricle and tricuspid valve. (b) Apical long axis and (c) high parasternal short axis view showing the hypoplastic ascending aorta. Its connection with the heart was not demonstrated. (d) Suprasternal notch view of the hypoplastic ascending aorta. Doppler flow pattern with the sample volume (SV) in the ascending aorta showed flow towards the aortic root during systole and away from it in diastole. RA, right atrium; RV, right ventricle; LA, left atrium; LV, left ventricle; Ao, aorta; PA, pulmonary artery.

mained widely patent. Despite active measures she finally succumbed to refractory heart failure when she was 10 days old.

NECROPSY FINDINGS

The heart was large and weighed 67 grams. There

was the usual atrial arrangement. The cardiac apex was directed to the left and the systemic and pulmonary veins drained normally to the right and left atrial chambers respectively. No definite interventricular groove was present on the surface of the heart, but the distribution of coronary arteries

Fig 2 (a) Surface anatomy of the heart showing atrial situs solitus, a small right ventricle, and large left ventricle separated by the anterior descending branch of the left coronary artery (arrows). (b) Opened view of the left ventricle showing bicuspid mitral valve and origin of the pulmonary artery from this ventricle. The main pulmonary artery continued via a large ductus arteriosus to the descending aorta after giving off the right (R) and left (L) pulmonary arteries. (c) Opened view of the right heart showing the hypoplastic thickened tricuspid valve (TV) and small right ventricular cavity. The ascending aorta was small with an atretic aortic valve and subaortic region that merged with the muscle mass of the right ventricular wall (*). The aortic arch (AA) was larger and there was no coarctation. RA, right atrium; RV, right ventricle; LA, left atrium; LV, left ventricle; Ao, aorta; PA, pulmonary artery; DA, descending aorta; PDA, ductus arteriosus.

indicated a small right and a large left ventricle (fig 2a). The internal appearance of the atria was normal and the foramen ovale was patent. The left atrium was connected by a bicuspid mitral valve to the main ventricular chamber which had the morphological characteristics of a left ventricle (fig 2b).

The pulmonary trunk originated from the left ventricle and was connected via a large ductus arteriosus to the descending aorta after giving rise to the right and left pulmonary arteries. The right atrium was connected via a stenotic and hypoplastic tricuspid valve to a small right ventricle which was present as

a tiny cavity within the thick muscle mass on the right shoulder of the left ventricle (fig 2c). No right ventricular outflow tract could be identified. The aortic valve and subaortic region were atretic and merged with the muscle mass of the right ventricle. The ascending aorta was 2 mm in diameter and there were two coronary arteries present in the usual positions. The aortic arch was larger and gave rise to normal brachiocephalic arteries.

Discussion

Aortic atresia with ventriculoarterial discordance is a rare congenital malformation of the heart. We found only six cases reported in published reports. Three were associated with double inlet univentricular atrioventricular connection with main chamber of left ventricular morphology, one with corrected transposition, and one with absent tricuspid valve and atrioventricular discordance.⁴ McGarry *et al* described the only patient with aortic atresia occurring with complete transposition of the great arteries diagnosed by angiography; this patient was alive at the age of six.³ As in our patient, the clinical picture was dominated by heart failure with mild cyanosis. Physical examination showed cardiomegaly with a single second heart and non-specific murmurs. The electrocardiogram showed characteristically right atrial and left ventricular hypertrophy rather than right ventricular hypertrophy as seen in the usual hypoplastic left heart syndrome. Definitive diagnosis required further investigations such as cardiac catheterisation, and in our case non-invasive examination by cross sectional echocardiography.

A morphologically left ventricle is considered to be more effective than a right ventricle as the supporting chamber for the systemic and pulmonary circulations and led to the long survival of McGarry's patient.³ Theoretically a morphologically left ventricle should also provide a more normal

physiological state.⁵ The early death of our patient despite the patency of the ductus arteriosus could be the result of insufficient coronary artery perfusion. Our Doppler study showed retrograde flow via the small ascending aorta that was present only during systole, while during diastole there was negative flow attributed to the low vascular resistance in the pulmonary vessels.⁶ Coronary insufficiency, together with volume overloading of the systemic ventricle, rather than ductal closure may be the major cause of death in patients with aortic atresia because the ductus arteriosus was found to be widely patent in most patients.^{1 2}

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