BRIEF COMMUNICATIONS

AORTOPULMONARY WINDOW ASSOCIATED WITH COMPLETE ATRIOVENTRICULAR SEPTAL DEFECT

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Aortopulmonary (AP) window and atrioventricular (AV) septal defect are congenital cardiovascular defects that typically occur without major associated cardiac anomalies but in a minority of cases are found in conjunction with a variety of other lesions. We describe the case history of an infant with coexisting AP window and complete AV septal defect who underwent complete primary repair at 3 months of age. To our knowledge, this combination of congenital cardiovascular defects has not previously been described.

Clinical summary. The patient was born at 37 weeks' estimated gestational age by spontaneous vaginal delivery, with a birth weight of 3.0 kg and Apgar scores of 8 at 1 minute and 9 at 5 minutes. At 2 months of age, she was referred for cardiac evaluation because of a cardiac murmur and failure to thrive. She weighed 3.4 kg and was feeding poorly. On evaluation, she was found to be nondysmorphic. Cardiac examination disclosed a grade 3/6 holosystolic murmur, a right ventricular heave, a diastolic rumble over the apex of the heart, and a gallop. The oxyhemoglobin saturation was 97% on room air, and the heart rate and blood pressure were within the normal ranges for age. The electrocardiogram showed a normal sinus rhythm, left axis deviation, right atrial enlargement, biventricular hypertrophy, and nonspecific changes in the ST segment. The echocardiogram demonstrated a Rastelli type C complete AV septal defect with large atrial and ventricular components and moderate regurgitation of the left-sided component of the common AV valve, along with a left-sided superior caval vein draining to the right atrium via an enlarged coronary sinus. The patient was initially managed medically with digoxin and furosemide, but there was little improvement in her growth. At the age of 3 months, a repeat echocardiogram confirmed the prior diagnosis but also revealed a large AP window between the proximal pulmonary trunk and ascending aorta, with continuous left-to-right shunting. Cardiac catheterization was performed to assess her hemodynamic

status. Angiography confirmed the diagnosis, and hemodynamic and oximetric measurements suggested normal pulmonary vascular resistance and a pulmonary/systemic blood flow ratio of more than 3:1.

The following day, surgical repair was performed with the use of cardiopulmonary bypass and a period of deep hypothermic circulatory arrest. After aortic and right atrial cannulation, the patient was cooled to a core temperature of 18°C, after which the cannulas were removed and circulatory arrest instituted. The AP window was incised anteriorly and closed with a patch of pulmonary allograft tissue, which was incorporated anteriorly into the arteriotomy closure. The AV septal defect was then approached through the right atrium. The diagnosis of a Rastelli type C defect was confirmed, and it was noted that the posterior bridging leaflet was bound to the crest of the ventricular septum, leaving almost no inter-ventricular communication beneath. The AV septal defect was repaired by means of a double-patch technique. Continuous 6-0 polypropylene suture was used to sew a Dacron patch to the crest of the ventricular septum to close the large anterosuperior interventricular communication. The patch was then attached to the ventricular surfaces of the posterior and anterior bridging leaflets of the common AV valve, and the left- and right-sided components of the respective bridging leaflets were brought into apposition. The septal commissure was then closed with interrupted sutures. The interatrial communication was closed with a patch of autologous pericardium, in such a fashion that the coronary sinus, which carried left superior caval flow, drained to the right atrium.

On the second day after the operation, the patient exhibited movements that were suggestive of seizure activity. An electroencephalogram, however, showed no epileptiform activity. At follow-up of 2 years, the patient is alive and well, has undergone no subsequent cardiovascular interventions, and has had no subsequent seizure-like activity. Echocardiography demonstrates normal ventricular inflow and outflow, trivial AV valve regurgitation, and no evidence of flow disturbance in the area of the AP window repair.

Comment. AP window is an uncommon congenital anomaly that is typically found without associated defects. When it does occur with other significant lesions, the associated defect is usually either a simple ventricular septal defect or some form of obstruction to systemic or pulmonary outflow, such as interruption of the aortic arch, tetralogy of Fallot, or aortic or pulmonary atresia.¹ Several cases of AP window in conjunction with transposition of the great arteries have also been reported.¹ Complete AV septal defect is a relatively common

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form of congenital heart disease that also tends to occur without associated anomalies, except when it is found in patients with abnormalities of viscerocardiac situs. In patients without heterotaxy, major associated cardiovascular defects include "conotruncal anomalies," coarctation of the aorta, and subaortic obstruction.² In this report, we describe a patient with both AP window and complete AV septal defect, a combination that has not been reported before. Repair was not complicated by the combination of these two lesions.

The tendency for AV septal defect to occur with anomalies of the outflow tracts has led some investigators to hypothesize that there may be a developmental relationship between such anomalies.^{3,4} In the mature heart, the AV junction is in direct apposition to the so-called "conotruncal" region, and it is conceivable that developmental anomalies of one area may affect the other. In the typical form of complete AV septal defect, the failure of septation of the AV junction results in incomplete absorption of the systemic ventriculoarterial junction, producing a "goose-neck deformity" of the left ventricular outflow tract. Although this may cause subaortic obstruction and may be related in approximately 10% of cases to the development of associated tetralogy of Fallot, the valvular and supravalvular portions of the systemic outflow tract are infrequently affected.² This is true in cases of heterotaxy/atrial isomerism as well, in which the most common anomalies of the outflow tracts are double-outlet ventricle and pulmonary atresia, often found together.⁵ In patients with abnormalities of ventriculoarterial concordance, such as double-outlet right ventricle and transposition of the great arteries, there is a higher rate of AV valve abnormalities, if not AV septal defect, than in patients with normally related great arteries.⁶ These factors, taken together, suggest that any association between anomalies of the AV junction and the outflow tracts is most likely to involve impaired rotation/alignment of the "conotruncal" region.

The nature of any potential relationship between anomalies of the AV junction and outflow tracts is not clear. Abnormalities of the cardiac neural crest axis have been implicated in many "conotruncal" anomalies, based on the association of such lesions with deletions of chromosome 22q11.⁷ However, neural crest ablation in embryonic avian models almost never results in anomalies of the AV junction, suggesting that the neural crest axis is not central to the development of such lesions.⁷ Similarly, other genetic models of congenital heart disease have not found a significant association between anomalies of the AV canal and the outflow tracts.⁸

Thus, there is little evidence to suggest that specific genetic causes are responsible for the common occurrence of AV septal defect and anomalies of the outflow tracts. Whether there is some local mechanical, paracrine, or growth factor-related effect is not known. The proximity of these regions in the developing heart is consistent with such a mechanism, and anomalies of both the AV canal and the outflow tracts have been produced in a model of altered blood flow patterns in the developing avian embryo.⁹ Ultimately, however, there is little direct evidence to support genetic, flow-related, or other mechanisms that might explain the combination of AV septal defect and anomalies of the outflow tracts, including AP window.

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