MORPHOLOGICAL STUDY OF VENTRICULAR SEPTAL DEFECT ASSOCIATED WITH OBSTRUCTION OF AORTIC ARCH AMONG JAPANESE

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This report describes racial differences in the morphology of ventricular septal defects associated with coarctation or interruption of the aortic arch. Twenty-three heart specimens with obstruction of the aortic arch associated with ventricular septal defects collected in southern Japan were studied. Six specimens had an interrupted aortic arch and the remaining 17 cases had coarctation of the aorta. Fifteen cases (65.2% of the total) had malalignment defects with posterior deviation of the conal and/or truncal septum. In contrast to Anderson's report, only one case of perimembranous defect with aortic overriding was seen in our series. These anatomical findings suggest that the majority of ventricular septal defects associated with coarctation or interruption among Japanese is a malalignment defect with left ventricular outflow tract obstruction.

THE morphology of ventricular septal defects associated with obstruction of the aortic arch has previously been described¹⁻⁵. An experimental study has been made by Kawamura et al., using neutron irradiation as a teratogen to produce a cardiovascular anomaly complex similar to the coarctation complex found in man⁶.

More recently, a detailed summary of the morphology of ventricular septal defects associated with coarctation of the aorta was presented by Anderson et al.⁷ in 1983. There are definite differences between their data and the present data. This report points out racial differences in the morphology of ventricular septal defects associated with coarctation or interruption of the aortic arch.

Key Words:

Co/Ao type VSD
Malalignment VSD
Posterior deviation of septum
2D-echocardiography

MATERIALS AND METHODS

Among 150 heart specimens of congenital heart disease collected from the southern Japan (Miyazaki, Kagoshima, Kumamoto, Okinawa, and Oita prefectures) April 1981 to March 1984, 23 specimens with obstruction of the aortic arch associated with simple ventricular septal defects were studied.

The combination of these 23 hearts was viscero-atrial situs solitus, atrioventricular concordance, and ventriculo-arterial concordance. An association of patent ductus arteriosus was seen in twenty of these cases. In the 23 hearts, morphology of ventricular septal defect, left ventricular outflow tract obstruction, mitral valve malformation, and aortic malformation was examined. Furthermore, cross sectional echocardiography was performed in a part of our cases using Toshiba model SSH-11A.

RESULTS

Among the collection of 150 malformed heart

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TABLE I OBSTRUCTION OF AORTIC ARCH
IN 47 CASES

<i>Isolated coarctation</i>	7 (14.9%)
<i>Coarctation or Interruption complex</i>	40 (85.1%)
<i>Simple VSD & Co/Ao</i>	17
<i>Simple VSD & Int/Ao</i>	5 23 (48.9%)
<i>Simple VSD & Int/Ao with atretic strand</i>	1
<i>Hypoplastic left heart & Co/Ao</i>	10 (21.2%)
<i>ECD & Co/Ao</i>	2 (4.2%)
<i>TA & d-TGA & Co/Ao</i>	1 (2.2%)
<i>SRV & d-DORV & Co/Ao</i>	1 (2.2%)
<i>DORV & Int/Ao with atretic strand</i>	1 3 (6.4%)
<i>DORV & Co/Ao</i>	2

Abbreviations: VSD = ventricular septal defect; Co/Ao = coarctation of aorta; Int/Ao = interruption of aorta; ECD = endocardial cushion defect; TA = tricuspid valve atresia; TGA = transposition of the great arteries; SRV = single right ventricle; DORV = double outlet right ventricle.

TABLE II VENTRICULAR SEPTAL DEFECT
AND OBSTRUCTION OF AORTIC
ARCH IN 23 CASES

<i>VSD</i>	
<i>Muscular outlet with deviated septum</i>	8
<i>Perimembranous with deviated outlet septum</i>	7
<i>Perimembranous with aortic override and closure</i>	1
<i>Simple perimembranous</i>	7
<i>Additional left ventricular outflow obstruction</i>	
<i>Hypertrophic anterior lateral muscle bundle</i>	13
<i>Anomalous insertion of mitral valve</i>	2
<i>Septal bulge</i>	1
<i>Mitral valve malformation</i>	
<i>Hypoplasia</i>	2
<i>Aortic malformations</i>	
<i>Hypoplasia of aortic valve</i>	12
<i>Hypoplasia of ascending aorta</i>	11
<i>Bicuspid aortic valve</i>	6
<i>High take-off of right coronary ostium</i>	4
<i>Aberrant right subclavian artery from descending aorta</i>	2

specimens, association of obstruction of the aortic arch was found in 47 hearts (31.3% of the total). Associated cardiac anomalies in 47 cases

are listed in Table I. Seven cases were isolated coarctation of the aorta and the remaining 40 cases were coarctation or interruption complex.

Obstruction of the aortic arch associated with simple ventricular septal defect was seen in 23 of cases studied in this series. Five of these had an interrupted aortic arch, and remaining 17 cases had coarctation of the aorta. Interruption of the aortic arch with atretic strand was found in only one case (Fig. 1). A summary of the findings in the 23 autopsy cases is shown in Table II.

In regard to the morphology of the ventricular septal defects, fifteen cases (65.2%) had a malalignment type of defect. In eight cases (34.8%), the ventricular septal defect was located at the outlet muscular septum. In these hearts, the conal and/or truncal septum was deviated posteriorly, and caused left ventricular outflow tract obstruction (Fig. 1). In seven cases (30.4%), the ventricular septal defect was the perimembranous type with posterior deviation of the conal septum (Fig. 2).

In 6 cases with interrupted aortic arch, the morphology of the ventricular septal defect was muscular outlet with deviated septum in 3 cases (Fig. 1), and perimembranous with deviated conal septum in 3 cases (Fig. 2). When we excluded these cases with interruption complex, the malalignment type of ventricular septal defect was found in 9 of 17 cases (52.9%).

On the other hand, only one case (4.3%) had a perimembranous ventricular septal defect with overriding of aorta to the right ventricle (Fig. 3). In this heart, the size of the ascending aorta was large, and the ventricular septal defect was almost occluded by the septal leaflet of the tricuspid valve (Fig. 3).

The remaining seven hearts had a simple perimembranous ventricular septal defect without deviation of the septum. In these cases, the left ventricular outflow tract obstruction was caused by an anomalous insertion of the mitral valve in two cases; by hypertrophic anterolateral muscle bundle in three cases; and by a subaortic muscle bundle and a septal bulge in one case. We found no cause of decrease in blood flow to the ascending aorta in 3 of 7 cases of simple perimembranous ventricular septal defect.

As mentioned above, in most of cases of our series, blood flow to the ascending aorta from the left ventricle was decreased by posterior deviation of the conal and/or truncal septum.

In addition, other obstructive lesions of the

Fig.1. Right ventricle (RV) in a patient with interruption of aortic arch (Int/Ao) and isthmic atresia (arrow). A large subpulmonary ventricular septal defect (VSD) is present. Although the conal septum is almost absent, the left ventricular outflow tract is narrowed due to posterior deviation of the truncal septum. This type of VSD was also seen in 5 cases of coarctation complex.

PA = pulmonary artery; BCA = brachiocephalic artery; LCA = left carotid artery; LSA = left subclavian artery; d-Ao = descending aorta

Fig.2. Right ventricle (A) and left ventricle (B) in a patient with perimembranous VSD and deviated conal septum.

PA = pulmonary artery; VSD = ventricular septal defect

Fig.3. Right ventricle (A) and left ventricle (B) in a patient with perimembranous VSD, aortic overriding and preductal coarctation of aorta. VSD is almost occluded by the tricuspid valve (TV).
MV = mitral valve; LV = left ventricle; PA = pulmonary artery;
AO = aorta; VSD = ventricular septal defect

Fig.4. Left ventricular outflow tract is obstructed by a hypertrophic anterolateral muscle bundle. Similar abnormality were seen in thirteen cases.
LV = left ventricle; VSD = ventricular septal defect

Fig.5. Left ventricular outflow tract is narrowed by anomalous insertion of the mitral valve (arrow) and a hypertrophic anterolateral muscle bundle (star).
Ao = aorta; LV = left ventricle; VSD = ventricular septal defect

left ventricular outflow tract were found in 16 of 23 cases (69.6%). Such obstructions were formed by hypertrophic anterolateral muscle bundle of the left ventricular outflow tract in 13 cases (Fig. 4); by anomalous insertion of the

mitral valve in 2 cases (Fig. 5); and by a subaortic muscle bundle and a septal bulge in one case (Fig. 6).

As an abnormality of the mitral valve, hypoplasia was present in two cases, but there were no

Fig.6. Left ventricle (LV) in a patient with perimembranous VSD and coarctation of the aorta. Left ventricular outflow tract is narrowed by a subaortic muscle bundle and a septal bulge (star). High take-off of the right coronary ostium (RCO) is present. Ao = aorta

parachute malformation or other deformities. Examination of aortic malformation revealed the following: hypoplasia of the aortic valve was seen in 12 cases (52.2%), hypoplasia of the ascending aorta in 11 cases (47.8%), and bicuspid aortic valve in 6 cases (26.1%). In addition, there was high take-off of the right coronary ostium in 4 hearts (Fig. 6), and aberrant right subclavian artery from the descending aorta in two cases (one in interruption and another in coarctation

of aorta). As other cardiac anomalies, there were patent ductus arteriosus in 20 cases, bicuspid pulmonary valve in 3, and persistent left superior vena cava into coronary sinus in one.

In our clinical study, the malalignment type of ventricular septal defect due to posterior deviation of the conal and/or truncal septum was visualized in the parasternal left ventricular long axis view (Fig. 7).

DISCUSSION

The morphology of ventricular septal defects associated with obstruction of aortic arch, especially in interrupted aortic arch, has been described.¹⁻⁵ A recent detailed study on the morphology of ventricular septal defects with coarctation of the aorta was presented by Anderson et al.⁷ in 1983.

The incidence of malalignment defects with posterior deviation of the conal and/or truncal septum was high in our series (65.2%), but low in Anderson's report⁷ (16.0%). On the other hand, perimembranous defects with aortic override were found in most of Anderson's cases⁷ (80.0%), while only one case was found in our series (4.3%). These two findings suggest definite racial differences between Caucasians and Japanese.

One of the causes of it to be thought that the occurrence of infundibular (conal) ventricular septal defect in Japanese specimens is much more frequent than in Caucasian specimens. Such hearts were found in 51.2% of our 43 simple ventricular septal defects. As shown in Ando's report⁸ this type of defect is about twice as frequent among Japanese as among Caucasians.

Fig.7. Two-dimensional echocardiogram in a patient with interruption or coarctation of aorta. Parasternal LV long axis view shows a malalignment type of VSD due to posterior deviation of the conal septum (arrow).

LA = left atrium; RV = right ventricle; Ao = aorta; LV = left ventricle

It seems that interrupted aortic arch and coarctation of the aorta are found at opposite ends of the same spectrum. Between these two extremes, there is interruption of the aortic arch with isthmus atresia as shown in Fig. 1. In our series, moreover, we found no morphological differences in ventricular septal defects between the coarctation group and the interruption group.

These findings suggest that the hemodynamic effect of reduced blood flow to the ascending aorta is one of the causes of coarctation or interruption of the aortic arch, but this is not the only cause. In fact, no prominent findings of left ventricular outflow tract obstruction were found in 3 of our cases.

Craigie⁹ and Skoda¹⁰ proposed that aortic coarctation could be produced postnatally by extension of contractile ductus tissue into the aortic wall.⁵ According to the angiocardigraphic study by Momma et al.,¹¹ ischemic hypoplasia results from prenatal decrease of blood flow to the ascending aorta, and localized stenosis is related to abnormal distribution of ductal tissue to the descending aorta and post-natal constriction of the ductus arteriosus.

Coarctation or interruption of the aortic arch associated ventricular septal defect is one of the congenital heart diseases which is critical in the neonatal period and early infancy, so we should confirmed the diagnosis early and try surgical correction without delay.

Two-dimensional echocardiography is the only accurate method of non-invasive diagnosis of this condition. Two-dimensional echocardiographic findings of ventricular septal defects associated with coarctation of aorta were described in detail by Smallhorn et al.¹² In our study, the malalignment type of ventricular septal defect due to posterior deviation of the conal septum was visualized in a parasternal left ventricular long axis view.

In conclusion, there are definite racial differences between Japanese and Caucasians in the morphology of ventricular septal defects associated with coarctation or interruption of the aortic arch. The characteristic type of ventricular septal defects associated with coarctation or interruption of the aortic arch among Japanese seems to be a malalignment type of defect with posterior deviation of the conal and/or truncal septum. Therefore, this type of ventricular septal defect may be said to be an oriental disease.

Our data suggest that it is important to investigate the aortic arch in Japanese patients whenever we find this malalignment type of the ventricular septal defect by two-dimensional echocardiography.

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