AN AUTOPSY CASE OF CONGENITAL INTERRUPTION OF THE AORTIC ARCH

Yumiko Yoshie, Yūji Hamamoto, and Kei Okuda

CONGENITAL Absence of the aortic arch is a very rare malformation of unknown etiology. Since the nineteenth century, this abnormality has been reported repeatedly in the literature of several countries, but in Japan a few autopsy cases have been reported up to the present.

Presented here is an autopsy case of congenital absence of the aortic arch surviving for four months with death from congestive heart failure.

The purpose of this report is to clarify the type to which this case belongs and to discuss its pathogenesis.

CLINICAL FINDINGS

This male baby, approximately 37 gestatory weeks of age, was delivered on Aug. 18, 1973. The delivery was normal and the infant weighed 2900 g at birth. There was no cyanosis and no other abnormality was noted.

He was transferred to Yodogawa Christian Hospital on Sept. 18, 1973, because of poor feeding and a heart murmur. On admission his general condition was not good. He was pale, but there was no cyanosis. Respiration was shallow and he had rough breath sounds. Heart systolic murmur, especially at the left 3rd intercostal region was Grade IV and a low grade thrill was palpable. The liver was palpable three fingerbreadth below the costal margin. After admission he had frequently bouts of pneumonia and was treated with antibiotics and digitalis etc.

Laboratory examinations showed red cell counts on the average 3,790,000 cells per cmm, with 10.3 g/dl of hemoglobin, and 35% hematocrit. There were 6000 leucocytes per cmm. Serum electrolytes of Na, K and Cl were respectively 137 mEq/L, 5.5 mEq/L and 97 mEq/L. Urinalysis was normal.

On the evening of Dec. 18, he had a severe attack of dyspnea after vomiting and no treatment could help him, and he expired.

The pregnancy and delivery were uneventful. There was no history of infection in the mother during pregnancy.

There was no family history of congenital heart diseases. The mother, 27 years old, had one living and normal child.

AUTOPSY FINDINGS (Autopsy No. 2920)

The body was that of a well developed, slightly emaciated, male infant measuring 57 cm in length and weighing about 4 Kg. Slight edema was noted in both legs.

The heart, weighing 100 g (normal, 31 g), was thrice as large as the baby’s fist. The wall of the left and right ventricles measured respectively up to 1.2 cm and 1.0 cm in thickness. An interventricular septal defect of the membranous portion measured 0.8 cm in diameter and the foramen ovale was closed membranously. The left ventricle was extremely small and the right ventricle was hugely dilated, primarily due to enlargement of the pulmonary conus. Pulmonary, aortic, tricuspid and mitral valves were grossly normal.

The endocardia of atria and ventricles were

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smooth. The trabeculae and papillary muscles of right and left ventricle were well developed, but papillary muscles of the right ventricle were thicker than those of the left ventricle.

Fig.1. Chest X-ray.

The pulmonary artery passed over the ascending aorta, and ran through its normal course. The innominate artery arising from the aorta divided into the right subclavian, the right common carotid artery and the left common carotid artery. Absence of the aortic arch was noted. The pulmonary artery was dilated and measured 5 cm in diameter. From this artery arose the right and branches to the lungs, and the pulmonary artery continued as the descending aorta through the ductus arteriosus. The left subclavian artery was derived from the descending aorta. Vena cava superior entered the right atrium in the normal position. The pulmonary vein entered the left atrium in the normal position.

On microscopic examination, the heart muscle fibers of both ventricles were slender and thin in general, but muscular cross-striation was not clearly visible. In the myocardium of the right ventricle, muscle fibers showed slight hypertrophy and edema appeared in the interstitial connective tissue. Moderate fibrosis of the subendocardium was noted here and there.

The left lung weighed 50 g; the right one, 75 g (normal, l:17 g; r:24 g). The lungs were fairly well aerated and dark red in color. Microscopically catarrhal pneumonia was seen in both lungs, that is the alveolar septum was thickened with congestion, and desquamative epithelial cells, fibrinous exudate and leucocytes were seen in the alveoli. There were a slight proliferation of endothelial cells and thickening of the wall of some vessels.

Fig. 3. External appearance of the heart. Hypertrophied right ventricle and the descending aorta continuing pulmonary artery.

The left kidney weighed 22 g; the right one, 25 g (normal, l. or r.: 20 g). The surface appearance of the kidneys was smooth and fetal. Most of the glomeruli were histologically immature and some of them revealed severe damage and fibrosis.

The thymus weighed 3 g (normal, 32 g) and the cortex was poorly developed histologically and only a few Hassall's corpuscles were observed in the medulla.

No significant changes were found in any other organs or tissues. Pathological diagnosis: 1) Steidele's complex (Type B) a) Congenital absence of aortic arch. b) PDA. c) VSD. d) Hypertrophy of the heart. e) Hypoplasia of ascending aorta. 2) Catarhal pneumonia. 3) Hemorrhagic diathesis. 4) Moderate congestion of the abdominal viscera. 5) Atrophy of the thymus.

DISCUSSION

The first case recorded with this anomalies appears to be by Steidele in 1818. Thereafter there were many reports. In 1967, Lie collected 88 cases from the literature, added 2 cases of his own and suggested the term “Steidele's complex” for this anomaly.

In Japan, Ino reported the first case in 1918 and lately Imamura et al. described 18 cases from the literature and his own in detail.

In 1959, Celoria described three types of absence of the aortic arch, based on the sites of origin of the branches of the aortic arch. Thereafter this pathologic classification has received considerable interest.

The type A interruption, which occurs immediately distal to the left subclavian artery, appears to result from a regression or atrophy of the segment of left dorsal aorta between the ductus arteriosus and the left subclavian artery.

In the type B deformity, the interruption is just distal to the origin of the left carotid artery and represents a failure of formation of the left fourth arch, since the left subclavian artery arises from the descending aorta.

In the type C malformation, only the right carotid artery arises from the ascending aorta. This anomaly was included in “the...
left heart syndrome" (Noonan et al.).

This report is type B anomaly.

Ventricular septal defect (VSD) and patent ductus arteriosus (PDA) were seen in most cases. The constant association of the absence of the arch of the aorta with PDA and VSD has led Everts-Suárez⁴ to suggest that the anomalies should be known as a cardiovascular triology. Atrial septal defect (ASD), patent foramen ovale, hypoplasia of ascending aorta, bicuspid aortic valve etc. have been observed frequently.

The symptoms of interruption of the aortic arch are non-specific and the physical signs are of only limited value. It would seem that differential cyanosis would always be present, but it is apparently the exception rather than the rule, because VSD, ASD, and patent foramen ovale are present in most cases, permitting a shunt of oxygenated blood to the descending aorta.

According to the Annual of Pathological Autopsy Cases in Japan¹⁰ 40 cases of absence of the aortic arch were autopsied in the last 5 years (1968 - 1972). In 41 cases including our own case, 20 cases were male and 21 cases were female. Thirtyseven of the 41 patients (90%) died in the first year of life and 26 died in the first month. Only 4 survived for longer than one year (Table). Dische¹¹ suggests that solitary interruption of the aortic arch without PDA or other cardiac anomalies is usual favorable with prolonged survival.

The triad of Everts-Suárez and Carson⁴ was seen in 17 cases (41 per cent). A VSD was also present in 34 of the 41 cases (83 per cent). An ASD or a patent foramen ovale was seen in 16 cases (39 per cent). Other additional congenital cardiac anomalies included hypoplasia of ascending aorta, bicuspid aortic valve, and hypertrophy of the heart. Transposition of the great arteries, hare lip, atresia of esophagus and Di-George's syndrome¹⁰ have been observed as associated malformations. The site of the interruption of the arch of the aorta was clearly described in 3 cases (Type B anomaly).

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### TABLE 1 AUTOPSIED CASES OF ABSENCE OF THE AORTIC ARCH FROM ANNUAL OF THE PATHOLOGICAL AUTOPSY CASES IN JAPAN (1968–1972)

<table>
<thead>
<tr>
<th>Year</th>
<th>Number of cases</th>
<th>Sex</th>
<th>Age when died</th>
<th>VSD</th>
<th>PDA</th>
<th>ASD or patent</th>
<th>For. orale</th>
</tr>
</thead>
<tbody>
<tr>
<td>1968</td>
<td>6 M 3 F 3</td>
<td></td>
<td>2 died in first year</td>
<td>5</td>
<td>3</td>
<td>5</td>
<td>Hare lip, RVH Atresia of esophagus</td>
</tr>
<tr>
<td>1969</td>
<td>7 M 4 F 3</td>
<td></td>
<td>5 died in first months</td>
<td>6</td>
<td>1</td>
<td>2</td>
<td>Transposit. of great arteries Bicuspid aortic valve</td>
</tr>
<tr>
<td>1970</td>
<td>9 M 5 F 4</td>
<td></td>
<td>6 died in first months 1 lived over 1 year</td>
<td>8</td>
<td>6</td>
<td>4</td>
<td>Transposit. of great arteries RVH, LVH</td>
</tr>
<tr>
<td>1971</td>
<td>6 M 2 F 4</td>
<td></td>
<td>4 died in first months</td>
<td>5</td>
<td>2</td>
<td>3</td>
<td>Bicuspid aortic valve Mitral stenosis, Type B (1)</td>
</tr>
<tr>
<td>1972</td>
<td>12 M 5 F 7</td>
<td></td>
<td>9 died in first months 2 lived over 1 year</td>
<td>9</td>
<td>4</td>
<td>2</td>
<td>Transposit. of great arteries Hypoplasia of ascending aorta DiGeorge’s syndrome Type B (1)</td>
</tr>
<tr>
<td>1973</td>
<td>1 M 1</td>
<td></td>
<td>4 months</td>
<td>1</td>
<td>1</td>
<td>0</td>
<td>Hypoplasia of ascending aorta Hypertrophy of the heart Type B</td>
</tr>
<tr>
<td></td>
<td>41 M 20 F 21</td>
<td></td>
<td></td>
<td>34</td>
<td>17</td>
<td>16</td>
<td></td>
</tr>
</tbody>
</table>

On microscopic descriptions in a few cases of previous reports, there was a slight proliferation of endothelial cells and a thickening of the wall of the pulmonary artery and renal glomerular damage. The histologic findings in our case confirm almost to these reports.

**Summary**

An autopsy case of congenital interruption of the aortic arch in a four months old male with PDA and VSD is reported and a review of the pertinent literature is presented.

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**References**