CONGENITAL INTERRUPTION OF THE AORTIC ARCH

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Complete interruption of the aortic arch is rare, and so far only seven cases appear to have been recorded (Maud Abbott, five cases, 1927; Evans, one case, 1933; Sewart, one case, 1948).

Apart from the rarity of this condition, the present case is recorded because it emphasizes that the clinical recognition of the condition may be complicated where there is persistent patency of the ductus arteriosus, and when an interventricular shunt allows better oxygenated blood to reach the lower part of the body. The case also illustrates, by the coincidence of abnormalities representing disturbances at the same period of development and by the occurrence of a maternal illness at, or about, the same time, the theory that many congenital abnormalities are the result of arrests of development at critical periods of embryonic development.

In infantile coarctation the aorta narrows proximal to the entrance of the ductus arteriosus, and here the diameter of the vessel is less, and usually much less, than that of the subclavian artery. In consequence, the lower part of the body is almost entirely supplied during foetal life by blood from the right ventricle which reaches it *via* the ductus arteriosus. There is no stimulus to develop anastomoses between branches from the aorta above and those below the narrowing. After birth and with the closure of the ductus, the circulation in the lower half of the body is dependent on the flow through the coarctated part of the arch and becomes insufficient. In complete interruption of the aortic arch the lower part of the body is at all times completely isolated from the left ventricle and is supplied by blood from the right ventricle through the patent ductus arteriosus. After birth this is venous blood and even that is only available for the short period that the ductus remains patent.

Clinical History

The mother was 24 years old and her blood group was Rh positive. After a 39-week pregnancy, a baby boy was delivered normally, on December 9, 1956, weighing 2,700 g. He was the second child, the elder,

being premature, weighed 1,050 g. at birth, but is at present alive and well.

On the fourth day the infant fed poorly from the breast and supplementary feeding was started. He was then slightly jaundiced. On the sixth day complete artificial feeding was necessary and the baby was tube fed. Later on the same day he appeared ill, was pale and shocked and had a slightly jaundiced appearance. There was no pyrexia or cyanosis. The respirations were grunting and laboured with marked abdominal breathing. The liver and spleen were both palpable. The umbilicus was healing satisfactorily and the fontanelle was not depressed. The heart sounds were normal and no murmurs were heard. There were widespread crepitations over the right lung. The haemoglobin was 80% (Sahli). A radiograph showed a large heart with a shadow on its right border. It was considered that there was some central consolidation of the lung and probably an aspiration pneumonia. Oxygen, 'alevaire' and 'crystamycin' (0.5 ml. twice daily) were administered. In spite of the fact that the infant had never fed satisfactorily the birth weight had not decreased and he now weighed 2,800 g. At 9.30 p.m. on the same day the infant's condition suddenly deteriorated and he died in a few minutes.

Necropsy

The body was that of a well-developed male infant. The skin and conjunctivae had an icteric tinge. There was no pitting oedema. On opening the chest a large heart, with a big right ventricle, occupied the major part of the front of the chest, compressing the left lung posteriorly. The venous drainage was normal. The right auricle was not dilated and opened by a normal tricuspid valve into the right ventricle. This valve showed several cherry red, pin-head-sized cysts on its auricular aspect, the so-called 'congenital cysts'. The right ventricle was large with well-developed muscular walls. There was a patent interventricular septal defect high up in the membranous portion, measuring 3 mm. by 4 mm. The pulmonary outflow tract was prominent and a large pulmonary artery, normal in position and direction, proceeded from it, giving off a branch to each lung and then, without change in its diameter, continued through the widely dilated patent ductus, to become the descending aorta. This gave origin to the usual thoracic and abdominal branches and divided into the common iliac arteries. The left auricle was not dilated.

There was a defective interauricular septum due to an inadequate and fenestrated septum secundum. The persistently patent slit-like opening measured 9 mm. by 2 mm. The left auricle opened through a normal mitral valve into the left ventricle which was smaller than the right. The aorta originated from the left ventricle. It was a smaller vessel than the pulmonary artery and was guarded by three semilunar cusps normally placed. The coronary vessels arose normally. A normal innominate and left carotid vessel were present and the aorta terminated as the left subclavian artery. There was no communication or strand of fibrous tissue between it and the distal aorta (Figs. 1-2).

The left lung was compressed by the enlarged heart, but the right lung filled the pleural cavity well. The other organs were congested, but were not otherwise abnormal.

Histology

The left lung showed extensive areas of primary non-aeration with some vernix 'squames' and mononuclear cells with foamy cytoplasm. A similar reaction was present in the right lung and the changes in both were consistent with the clinical diagnosis of an 'aspiration pneumonia'. The liver showed focal areas of necrosis with some prominence of myeloid haemopoiesis in relation to the portal tracts. The adrenal had a nest of primitive neuroblasts which measured 1.6 mm. by

0.8 mm. Sections of spleen, thymus, pancreas, kidney and brain showed the changes of acute congestion.

Discussion

To understand the pathology of this condition the embryology should be considered. During the course of development six pairs of aortic arches develop but not all are present at any one time. The period of development of the aortic arches extends throughout the fourth week of intra-uterine life and their transformation mainly occupies the fifth to the seventh week. The first arch appears at the beginning of the fourth week. Second, third, fourth, fifth and sixth arches appear in rapid succession but by the sixth week, when the sixth arch has appeared, the first and second arches are beginning to atrophy. Only towards the end of the fifth week are all the arches open and even the first is atrophic when the sixth is developing. Normally the first, second and fifth arches disappear entirely. The common carotid vessels are formed from the third arches. The fourth left arch forms that part of the arch of the aorta between the origin of the left common carotid artery and the aortic end of the ductus. This is the site of the defect in the present

FIG. 1.—The heart showing the continuity of the descending aorta with the pulmonary artery and the interventricular septal defect. DUCT. ART., ductus arteriosus; DESC. AORTA, descending aorta; Rt PUL. A., right pulmonary artery; I-V.S.D., interventricular septal defect; Rt V., right ventricle; Lt V., left ventricle.

FIG. 2.—The left ventricle showing the aorta giving rise to the vessels of the head and upper extremities. D.A., ductus arteriosus; Lt PUL. A., left pulmonary artery; I-V.S.D., interventricular septal defect; Lt VENT., left ventricle.

case, which essentially represents an involution of this portion of the fourth arch similar to the process normally affecting the first, second and fifth arches. Normally the portion of the same left arch distal to the origin of the subclavian artery joins the sixth arch to form the descending part of the arch of the aorta. The sixth arch forms both the pulmonary arteries and the ductus arteriosus, the latter closing in the neonatal period and its remains being identified as a fibrous band.

The interauricular septal formation begins in the fourth or fifth week of intra-uterine life and this is complete, apart from the post-natal closure of the foramen ovale, by the eighth week. In the first two weeks of neonatal life the foramen ovale closes in approximately 80% of infants. The interventricular septal formation begins in the fifth week of gestation and is completed by the eighth week.

In interruption of the aortic arch there is a complete disappearance of a portion of the fourth arch as well as the normal involution of the first, second and fifth arches. In infantile coarctation there is a failure of the structures derived from the arch in the region proximal to the ductus arteriosus to grow or to develop at the same rate as the rest of the vascular system at some period in foetal life.

The cause of any congenital defect is usually unknown, but at the present time environmental influences resulting from maternal illness or metabolic upsets during the first three months of pregnancy must receive most careful consideration. The period from the fourth to the eighth week is most critical with regard to heart development. Many of the known virus infections and obscure febrile illnesses have been implicated from time to time.

In the present case the defects would suggest some upset during the fifth to the sixth week of prenatal life. Careful investigation of the mother's health in the early months of her pregnancy was significant. Conception date could be more or less

restricted to March 28, 1956. On April 28 and 29 the patient was ill with headache, vomiting, general malaise and slight pyrexia. The diagnosis of 'sinusitis' was made and only sedatives and analgesics were prescribed. The age of the foetus at this time was four and a half weeks. After a rapid recovery the mother's health was good until May 25, 1956, when she was confined to bed for a few days with a typical influenzal syndrome of headache, prostration, generalized muscle pain and vomiting. The foetal age at this time was eight weeks.

During the fourth-fifth week of prenatal life at a critical stage in the development of the aortic arches and the interauricular septum and just before the formation of the interventricular septum this foetus could feasibly have been subjected to some upset secondary to the maternal illness which resulted in the defects already described. The illness at the eighth week is probably of secondary importance as the cardiac structures are more or less established at this date.

Summary

A case is described of complete interruption of the arch of the aorta in a male infant dying aged 6 days. A patent interauricular and interventricular septum resulted in the absence of clinical cyanosis. The mother had a febrile illness at a critical period in the development of the aortic arch and cardiac septa in the foetus.

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