Anomalies of the Aortic Arch and Ventricular Septal Defects

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SUMMARY This investigation presents additional evidence for the hemodynamic influence of intracardiac anomalies on the development of the aortic arch, based on measurements of different parts of the great vessels. Criteria are given to define the normal aortic arch and the different anomalies of the aortic arch, such as interruption, atresia, tubular hypoplasia, hypoplasia, abnormal long segment and juxta ductal coarctation. Two types of malalignment ventricular septal defects are described to illustrate how prenatal intracardiac flow disturbances can account for various aortic arch patterns.

An explanation is proposed as to how reduced blood flow through the embryonic preductal aorta may contribute to the pathogenesis of all dimensional anomalies of the aortic arch.

RECENT INVESTIGATIONS 1-3 have indicated that the size of blood vessels is directly proportional to the amount of fetal blood flow through them. To prove this concept, we examined autopsied heart specimens. Normal hearts were first analyzed to define the normal size of different segments of the aortic arch. Seventy-two heart specimens with a ventricular septal defect (VSD) present in the Laboratory of Anatomy and Embryology at Leiden University were studied to determine if any group was associated with aortic abnormalities. We found three groups (infundibular, atroventricular canal and muscular VSD types), with an anatomy such as to reduce aortic blood flow, to be associated with aortic abnormalities. Only one of them will be discussed in this paper. A fourth group, the anatomical Eisenmenger VSD type with an anatomy such as to reduce pulmonary blood flow, will also be presented.

Definition of Terms

A clear definition of terms is necessary since confusion still exists regarding the terminology of conal structures and the nomenclature of the different anomalies of the aortic arch. Until standard terminology is agreed upon we adopted an eclectic nomenclature for the different muscle masses in the outflow tract of the right ventricle based on the views of Anderson et al., 4 Van Mierop 5 and Becker et al. 6 as follows:

The conus septum is the septum which divides the embryonic conus cordis into aortic and pulmonary conuses. It is derived from the fused conal ridges. It occupies different positions in normal and abnormal specimens. In the normal heart it is the central and most prominent part of the inverted U-shaped muscular mass which separates the pulmonary and tricuspid valves. The conus septum has a parietal and a septal extension. It should be stressed that the term conus septum is not universally considered to be synonymous with the crista supraventricularis.

The parietal band, not to be confused with the parietal exponent of the conus septum, is the right limb of the inverted U. It originates in the lateral wall of the right ventricle at the level of the tricuspid ring and extends upwards to fuse with the conus septum. Embryologically it is considered to derive from the right extension of the conoventricular flange.

The septal band, again not to be confused with the septal extension of the conus septum, is the left limb of the inverted U. It extends from below the pulmonary valve along the ventricular septum to the apex. This structure does not originate from the conus. In the normal heart, the parietal band, the conus septum and the septal band are aligned and quite difficult to distinguish from each other. In some cases, they can dissociate giving a malalignment ventricular septal defect.

Twenty-nine normal heart specimens were studied to provide criteria for the dimensions of the different segments of the normal aortic arch (fig. 1). 7 Following the classification of Celoria and Patton 8 for interruption defects, the aortic isthmus is defined as the segment A lying between the left subclavian artery and the ductus arteriosus; the distal part of the aortic arch is the segment B between the left carotid artery proximally and the left subclavian distally; the proximal part of the arch is the segment C between the brachiocephalic artery and the left carotid artery.

The results of the measurements of the external diameters and the lengths of the various aortic arch segments of the normals were reported earlier. 7 As a rule the brachiocephalic artery, left carotid artery and left subclavian artery arise fairly close together on the aortic arch. If one of the arch segments in infants is longer than 5 mm it is considered abnormally long. The proximal arch segment is defined as hypoplastic when the external diameter of that segment amounts to 60% or less of that of the ascending aorta. The corresponding limit for hypoplasia in the distal arch is 50%, and for the isthmus 40%. Tubular hypoplasia represents a combination of abnormal great length and small diameter of a segment. 7 According to our criteria, tubular hypoplasia of the aortic isthmus is present when this segment is longer than 5 mm and its external diameter is 40% or less than that of the ascending aorta. Juxta ductal coarctation is a sharply localized narrowing of the aortic lumen resulting from an eccentric ridge-like thickening of the aortic wall. This fold is found characteristically at the junction of the preductal and the descending aorta. 9

Interrupt of a segment occurs when there is a discontinuity between two parts of the aortic arch. Atresia in-
dicates that a ligamentous connection persists between the parts.

According to these criteria, a normal aortic arch has no segment exceeding 5 mm in length and the external diameters of the proximal, distal and isthmus segment amount to at least 60%, 50% and 40% of the diameter of the ascending aorta, respectively. Within these limits all our normal hearts except one possessed a normal aortic arch.

Results

1. Ventricular Septal Defects Which Reduce Aortic Blood Flow (fig. 2a–b)

Of our group of 72 heart specimens with VSD, eight had an anatomy predisposing to an early intracardiac left-to-right shunt. In these hearts the VSD is located above the level of the papillary muscle of the conus and appears to involve the inferior part of the conus septum (fig. 2a). Within the classification of Goor et al.16 this defect would be considered infundibular VSD type I. Our observations suggest that this is a malalignment defect between the conus septum and the septal band. Indeed, its characteristic feature is the extension of the septal insertion of the conus septum along the cranioposterior edge of the defect into the left ventricle (LV) where it obstructs the anterior portion of the LV outflow tract (fig. 2b). Consequently the aortic orifice is narrowed and displaced posteriorly in the outflow tract. The degree of narrowing of the aortic orifice depends on the thickness of the muscular extension. The subaortic stenosis is sometimes aggravated by the presence of the anterolateral muscle bundle of the LV, a structure described in greater detail in earlier publications.7–14 This muscle bundle is a remnant of the left margin of the conoventricular flange. Due to the crossing over of the conus septum into the LV, the opening of the VSD and the aortic orifice lie adjacent in the same horizontal plane in the LV outflow tract, where they compete for the blood flow from the LV inflow tract. A preferential flow to the RV and the pulmonary artery apparently existed from early embryonic life at the expense of the flow to the preductal aorta, since in this particular type of malalignment VSD the defect is large and the aortic orifice fairly small.

This intracardiac hemodynamic situation was clearly reflected in the severe abnormalities observed in the aortic arches of seven of the eight specimens (fig. 3). The discrepancy in division of the flow between both great arteries could be evaluated by using the mean ratio between the external diameter of the ascending aorta and the pulmonary

Figure 1. Diagram of the great arteries. Arrows indicate the points at which measurements were taken. AoA = ascending aorta, AoD = descending aorta, PA = pulmonary artery, A = aortic isthmus, B = distal arch segment, C = proximal arch segment.

Figure 2. Ventricular septal defect with muscular subaortic stenosis due to crossing over of the septal insertion of the conus septum into the left ventricular outflow tract. a) Interior of the right ventricle. The pulmonary orifice (PO) is separated from the tricuspid orifice (TO) by the parietal band (P) and the conus septum (CS). The large ventricular septal defect (VSD) is due to a malalignment between the conus septum and the septal band (S). b) Interior of the left ventricle. The aortic orifice (AoO) is displaced posteriorly in the left ventricular outflow tract by the septal insertion of the conus septum (CS) extending through the ventricular septal defect (VSD) into the anterior part of the left ventricular outflow tract. The anterolateral muscle bundle of the left ventricle (Mu) also contributes in reducing the space for the aortic orifice. Ventricular septal defect and aortic orifice lie in nearly the same horizontal plane.
trunk, which was 56%. The group of normal heart specimens had a mean ratio of 96%.1

The aortic arch anomalies in the eight specimens included four interruptions of the distal B segment, one interruption of the aortic isthmus, one tubular hypoplasia of the isthmus with hypoplastic distal B segment and one juxtafunctional coarctation. Only one specimen (3928) had a normal aortic arch.

2. Ventricular Septal Defects Which Increase Ventricular Blood Flow (fig. 4)

Seven cases of anatomical Eisenmenger VSD were found.12 The site of this VSD usually resembles that of tetralogy of Fallot, but the Eisenmenger VSD should not be confused with the functional concept “Eisenmenger syndrome,” which predisposes to an early right-to-left shunt. Eisenmenger VSD is below the conus septum, which protrudes anteriorly in the infundibulum. On the right side of the defect, placed between the VSD and the tricuspid valve, another distinct muscular band is always present which resembles a second conus septum or a proximal crista supraventricularis.13 14 This bundle is the parietal band derived from the conoventricular flange. In the area of the defect, the aorta overrides the ventricular septum with significant dextroposition. The papillary muscle of the conus is usually abnormal or absent. When observed from the left ventricle the aortic orifice is deviated to the right but fibrous mitral-aortic continuity is still present. Embryologically this VSD may be regarded as a malalignment defect with aortic overriding caused by lack of embryonic conal inversion or conal rotation. This event is responsible for the anterior deviation of the parietal insertion of the conus septum in the infundibulum and the divorce between the parietal band and the conus septum.13 14 As a result of the anterior displacement of the prominent conus septum and the dextro-position

![Figure 3. Diagram of the group ventricular septal defect with muscular subaortic stenosis due to crossing over of the septal insertion of the conus septum into the left ventricular outflow tract. Top left: interior of the right ventricle (RV). The large left-to-right shunt is reflected in the jet dilation of the infundibulum. Right: interior of the left ventricle (LV). The associated aortic arch pattern of the eight specimens of this group are given in scale drawings. Below the diagrams a few essential data of the heart specimens are summarized: specimen number, age at death in days, diameter of ventricular septal defect, diameters of pulmonary and aortic orifice in mm, muscular extension of the conus septum (CS) and anterolateral muscle bundle of the left ventricle (Mu). 4055, 3613 and 3454: interruption of the distal aortic B segment with the right subclavian artery arising from the descending aorta; 4908: interruption of the distal B segment; 3821: interruption of the isthmus; 3607: hypoplastic distal B segment with tubular hypoplasia of the isthmus; 2573: juxtafunctional coarctation; 3928: normal aortic arch.](image)

![Figure 4. Ventricular septal defect of the Eisenmenger type. Interior of the right ventricle. The pulmonary orifice (PO) is separated from the dextroposed aortic orifice (AoO) by a prominent, anteriorly displaced conus septum (CS). This ventricular septal defect (VSD) is due to a malalignment between the conus septum (CS) and the parietal band (P), which derives from the conoventricular flange. The malalignment defect allows the aortic overriding. Note the absence of the papillary muscle of the conus.](image)
of the aorta, the blood from the RV inflow tract is diverted from the pulmonary artery to the aorta. Greater degrees of this anterior deviation would result in significant sub pul monary stenosis and produce a tetralogy of Fallot.

With the exception of specimen 3784 with its abnormally long distal B segment, all the aortic arches from these hearts were normal (fig. 5). The mean ratio between the external diameters of the ascending aorta and the pulmonary trunk was 87%, which is within the normal limits. In nearly every case the aortic isthmus was as wide as the descending aorta. In four cases the aortic isthmus could not be recognized as a discrete entity, since there was no interval between the left subclavian artery and the ductus. Moreover in three specimens the brachiocephalic artery and the left carotid artery arose from a common trunk on the aortic arch, another indication of additional shortening with widening caused by supranormal aortic flow.

Discussion

During recent years circumstantial evidence has been presented in support of a hemodynamic pathogenesis for congenital anomalies producing altered dimensions of the aortic arch. In this connection it appears appropriate to focus attention on the presently existing relationship between aortic arch anomalies and certain types of VSD.

A diminished blood flow through the embryonic preductal aorta may be considered the common factor responsible for the different anomalies of the aortic arch such as interruption, atresia, tubular hypoplasia, hypoplasia, abnormal long segment and juxtaductal coarctation.

Tubular hypoplasia can be considered as a point of departure for this concept, because an abnormally long and narrow segment is an important feature of the general pattern of the branchial arterial system which is present early in the sixth week of embryonic development. By the end of the sixth week, however, a widening with shortening of the developing aortic arch has taken place, explained by the rapid and considerable increase in circulation during this sixth week. Should there be an insufficient increase of aortic flow, then the embryonic pattern could persist as a tubular hypoplasia. This would represent a standstill of part of normal developmental processes. From this point of view, therefore, the tubular hypoplasia can be considered to represent, embryologically, the simplest type of aortic arch anomaly. A further decrease in flow through the embryonic ascending aorta could result in cessation of flow across a particular aortic arch segment in the same fashion as is described for several parts of the branchial arterial system during normal development. In the circumstances envisaged, however, it would lead to a developmental abnormality. On the other hand it is conceivable that early hemodynamic disturbances may disappear in a later stage of development. Following subsequent normalization of flow through the preductal aorta, a tubular hypoplasia could be gradually converted into an abnormally long arch segment as the only reflection of a temporary flow disturbance. In cases of an isthmal hypoplasia, another possibility is transformation into juxtaductal coarctation.

It has been shown previously that a number of intracardiac anomalies can cause disturbances in fetal flow patterns, which in turn can influence dimensions of the great arteries. On the one hand there are the heart malformations with a functioning prenatal right-to-left shunt. In these circumstances the pulmonary artery is decreased in size, the ductus arteriosus at birth is narrow and long, or even absent, and the preductal aorta is well developed, with a short isthmus usually as wide as the descending aorta. The prototype of this group is the tetralogy of Fallot. On the other hand there are heart malformations with a prenatal left-to-right shunt, as a result of which the pulmonary artery

![Diagram of the group ventricular septal defect of the Eisenmenger type. Same presentation and data as in figure 3. Heart specimen 3784 has an abnormally long distal aortic arch B segment. The other cases of this group all show a normal aortic arch pattern. 3784, 1322, 4071 and 4574 have no interval between the left subclavian and the ductus. Note also the common trunk for the brachiocephalic artery and the left carotid artery on the aortic arches of specimens 3784, 4071 and 4574.](http://circ.ahajournals.org/doi/10.1161/01.CIR.53.6.1014)
and the ductus are increased in size and the preductal aorta is underdeveloped. Depending on the severity and also on the time of onset of the flow disturbances, interruption, atresia, tubular hypoplasia or juxtaductal coarctation may occur.

An important potential factor for producing fetal flow disturbances is the association of ventricular septal defects with hypertrophic muscle bundles in one of the ventricular outflow tracts. Site and size of the VSD, in combination with the exact localization of the flow-diverting muscular bands, are points of major importance in this hemodynamic process. The group of defects with the muscular extension of the conus septum in the LV producing aortic stenosis clearly illustrates that defects which reduce aortic flow produce severe obstructive lesions to the aortic arch. In contrast, the group of Eisenmenger VSDs with the distinctly anteriorly displaced conus septum shows that an increased aortic flow produces normal or even widened arches.

More evidence concerning the relation between intracardiac flow disturbances and the origin of aortic arch anomalies can be obtained from microreconstructions of hearts and aortic arches from certain breeds of minipigs which are prone to develop congenital heart disease, in particular the combination of VSD with outflow obstruction of the LV and anomalies of the aortic arch. Studies of this process are in progress, and will be reported in due course.

In conclusion, we feel that it is important for clinicians dealing with congenital heart disease to be aware of the frequent association of intracardiac malformations with flow pattern disturbances and dimensional changes of the great arteries. Hence particular attention should always be given to the angiograms of the aortic arch and the pulmonary artery as they may frequently mirror the intracardiac anatomy.

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