Anomalies Associated with Coarctation of Aorta
Particular Reference to Infancy

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SUMMARY

One hundred specimens from subjects with coarctation of the aorta were studied with regard to the nature and occurrence of additional cardiovascular anomalies. Seventy-seven were less than 6 months of age at the time of death (group I) and 23 were 6 months of age or older (group II).

Exclusive of cases of bicuspid aortic valve, there were one or more associated anomalies in 87 cases, that is 91% of the patients under 6 months of age and in 74% of those in the older group. The various types of additional anomalies in order of decreasing frequency were as follows: (1) tubular hypoplasia of the aortic arch (49 cases); (2) abnormal communications (mainly ventricular septal defect and patent ductus arteriosus) (49 cases); (3) left ventricular outflow obstruction (mainly subaortic stenosis) (37 cases); (4) left ventricular inflow obstruction (26 cases); and (5) positional anomalies of the great vessels (mainly transpositions) (13 cases).

More than one anomaly in addition to the coarctation was commonly present. The overall incidence of congenital bicuspid aortic valve was 46%. Among 71 cases in which the relationship to the ductus could be determined, the coarctation was proximal or opposite the ductus in 62 cases and distal to the ductus in nine cases.

Additional Indexing Words:
Hypoplasia of aortic arch  Cardiac failure in infancy  Obstruction of aorta
Congenital bicuspid aortic valve  Multiple congenital malformations

It is generally appreciated that symptomatic infants with coarctation of the aorta experience a poor prognosis. One of the major factors leading to rapid and often fatal deterioration of such patients is the occurrence of associated malformations of the heart and great vessels.

With these facts in mind, it was the purpose of this project to study at necropsy the nature and occurrence of additional cardiovascular anomalies in subjects with coarctation. Comparison was made between patients surviving less than 6 months of age with those older.

Methods

For the purposes of this study, coarctation of the aorta is defined as a localized narrowing of the aortic lumen resulting from an eccentric ridge-like thickening of the aortic media as earlier defined. This lesion is to be distinguished from that designated as "tubular hypoplasia of the aortic arch." The latter term refers to uniform narrowing of a segment of the aortic arch.

One hundred specimens of coarctation of the aorta observed at necropsy and obtained consecutively formed the basis for this study. For each case, the gross specimen of heart and great vessels was available for restudy. In many instances, continuity of the lungs with the heart had been retained. For some cases, complete clinical records were available, while in others only the age and sex of the patient were known to us.

For each specimen, gross examination was made for (1) the position of the coarctation, (2)
Results

Among the 100 patients with coarctation of the aorta, 77 were under 6 months of age (group I) at the time of death, and 23 were older (group II). In group II, only four subjects were less than 1 year old. The remaining 19 show a spread in age from 14 months to 28 years. Among these, four ranged in age from 14 months to 2 years, seven from 2 to 10 years, and the remaining eight were between 10 and 28 years.

In group I, there were 42 males and 35 females, while in group II there were 18 males and five females.

Anatomic Relationship of Coarctation to Ductus Arteriosus (Fig. 2)

In each case, the coarctation occupied the general area of junction between the arch and ascending aorta. With respect to the ductus arteriosus, the site of coarctation varied from lying proximal, opposite, or distal to the site of entry of the ductus arteriosus (or ligamentum

Figure 1

Incidence of various types of anomalies associated with coarctation of the aorta.

the nature of the ductus arteriosus, and (3) the nature of associated anomalies, if present.

A classification of the types of associated anomalies and the number of times each type was observed are given in figure 1. The types of associated anomalies were as follows: (1) bicuspid aortic valve, (2) tubular hypoplasia of aortic arch, (3) abnormal communications (including septal defects and patent ductus arteriosus), (4) left ventricular outflow obstruction, (5) left ventricular inflow obstruction, and (6) positional anomalies of the great arteries.

The cases were placed into two groups depending on age of the patients. Those patients surviving less than 6 months were placed in group I, while group II contained those who survived 6 months or longer.

After the various conditions present were identified in the specimens, the clinical records were studied with regard to the diagnoses made. The purpose of this part was to determine the prevalence not only of correct diagnosis of the coarctation but also of associated conditions.

Figure 2

Incidence of varying relationships between coarctation of the aorta and the ductus arteriosus as observed in 71 cases in which pertinent information was available.
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arteriosum) into the aorta (fig. 2). In 29 of the 100 cases, resection of the segment with the coarctation and insertion of the ductus had been done, and information about the position of the coarctation was not available to us. In the remaining 71 cases (57 in group I and 14 in group II), this feature could be determined. In this series, the most common relationship was the one in which the coarctation lay proximal to the ductus, being observed in 46 of 57 cases of group I (81%) and in eight of 14 cases of group II (57%). Localization of the coarctation opposite the ductus occurred in seven cases of group I (12%) and in only one case of group II (7%). The presence of the coarctation distal to the ductus was observed in four cases of group I (7%) and in five cases of group II (36%).

Among the 57 cases of group I in which the relationship between the coarctation and ductus could be determined, there were five instances without associated malformations and 52 with associated malformations (exclusive of bicuspid aortic valve).

In the absence of associated malformations, the coarctation was either proximal or opposite the ductus in four and distal to the ductus in one.

Among the 14 cases in group II in which the relationship between the coarctation and the ductus was known, there were two cases without associated malformations. In each, the coarctation lay distal to the ductus. In the remaining 12 cases, each with one or more associated anomalies, the coarctation was proximal to or opposite the ductus in nine specimens, while in three, the coarctation lay distal to the ductus.

Associated Anomalies (Fig. 1)

Whether occurring alone or in combination with other anomalies, the following conditions were observed among the 100 specimens with coarctation.

Bicuspid Aortic Valve

Bicuspid aortic valve was found in 31 of the 77 cases of group I (40%) and in 15 of the 23 cases of group II (65%), an overall incidence of 46%. In nine cases, this was the only associated anomaly, while in 37 cases of bicuspid aortic valve, additional malformations were found.

Exclusive of nine cases having only bicuspid aortic valve, there were 87 cases with one or more associated anomalies. Seventy (91%) of the 77 cases of group I exhibited such malformations, while 17 (74%) of group II were so involved.

Among the 87 cases with associated anomalies, there were 76 cases with two or more associated malformations (exclusive of bicuspid aortic valve). The distribution of cases with two or more associated anomalies according to groups was as follows: 63 (82%) of 77 cases of group I and 13 (56%) of 23 cases of group II.

Tubular Hypoplasia of Aortic Arch

Exclusive of the seven cases with aortic atresia, tubular hypoplasia was found in 49 cases of the remaining 93 cases of the entire series. These specimens were from 44 of the 70 patients (63%) under 6 months of age and five of 23 patients (22%) in the older group. Thus, over half of the patients who died with coarctation of the aorta in the first 6 months presented with tubular hypoplasia. This fact seems especially important, since there is a distinct tendency for tubular hypoplasia to
occur in combination with additional malformations. In fact, none of the cases with tubular hypoplasia was devoid of an additional intracardiac anomaly. Among the 44 cases in group I with tubular hypoplasia, the most common type of additional anomaly was an “abnormal communication.” Such defects were observed in 28 cases, ventricular septal defect being the most common (14 cases). A second type of anomaly frequently encountered in combination with coarctation of the aorta and tubular hypoplasia was “left ventricular outflow obstruction.” Exclusive of aortic atresia, this occurred in 16 cases, of which subaortic stenosis was the most frequent type (14 of 16 cases).

Abnormal Communications (Fig. 3)

Abnormal communications allowing shunts were common and second only to tubular hypoplasia as a type of anomaly associated with coarctation of the aorta. Exclusive of cases with positional anomalies of the great vessels, 37 cases (48%) were encountered among the 77 cases in group I and 12 cases (52%) of 23 in group II. These anomalies included (1) patent ductus arteriosus, (2) ventricular septal defect, (3) atrial septal defect, (4) persistent common atriovenous canal, and (5) single ventricle.

Patent Ductus Arteriosus. Patency of the ductus arteriosus was found in 77 of the total series of 100 specimens. It is generally accepted that under normal circumstances the ductus is closed by 3 weeks of age, while a ductus still patent at 2 months of age usually remains patent. Therefore, in this study, patency of the ductus as an abnormality was recorded only when this condition was found in patients over 2 months of age. In the 100 cases of our series, 34 patients were over 2 months of age (11 in group I and the 23 in group II); 13 of them (38%) showed persistent patency of the ductus arteriosus (five of the 11 in group I and eight of the 23 in group II).

Ventricular Septal Defect. In 26 cases of the total series, represented by 22 cases (28%) of the 77 in group I and four (17%) of the 23 in group II, ventricular septal defect was associated with coarctation. In the majority of cases, the defect was located inferior to the supraventricular crest, i.e., the predominant location for ventricular septal defects.

In group I, there was a frequent association of tubular hypoplasia with ventricular septal defect, 14 of 22 cases with ventricular septal defect also exhibiting tubular hypoplasia. In seven of the cases in group I with ventricular septal defect, there was an additional abnormal communication. In four cases, the additional defect was an atrial septal defect and in three, a patent ductus arteriosus. Five of 22 cases with ventricular septal defect presented subaortic stenosis.

Atrial Septal Defect. In 13 cases of the total series (nine cases [12%] in group I and four [17%] in group II), atrial septal defect exclusive of valvular competent patent foramen ovale was present. With only two exceptions, those defects were of the fossa ovalis (secundum) type. One case with polysplenia exhibited an atrial septal defect of the “coronary sinus type,” as well as persistent left superior caval vein entering the left atrium as part of the developmental complex described by Raghib and associates. In the other case, a sinus venosus type of atrial septal defect was found in combination with partial anomalous pulmonary venous connection to the right atrium.

Persistent Common Atriovenous Canal. This was recognized as an entity separate from atrial septal defect and ventricular septal defect. Ten cases were seen in the total series (eight cases [10%] of the 77 in group I and two [9%] of the 23 in group II). Among the 10 cases of persistent common atriovenous canal, the “complete” type was observed in eight instances and the partial type in two.

Single or Common Ventricle. This condition was rare in this series. It occurred in four cases; three (4%) of group I and one case (4%) of group II. In each case, the great vessels were transposed.

Left Ventricular Outflow Obstruction (Fig. 4)

In this study, conditions causing obstruction to left ventricular outflow were categorized as follows: (1) subaortic stenosis, (2) unicom-
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Incidence and distribution of conditions characterized by left ventricular outflow obstruction in association with coarctation of the aorta.

Figure 4

missural aortic valvular stenosis, and (3) aortic atresia. Of the total series of 100 cases with coarctation of the aorta, 37 showed one or more of these anomalies. These 37 cases were represented by 31 cases (40%) of group I and six (26%) of group II.

Subaortic Stenosis. This condition was found to be the most common anomaly in the class of left ventricular outflow obstruction. It was observed in 20 cases (26%) of group I and in five cases (22%) of group II. Three anatomic varieties of subaortic stenosis were encountered. Muscular (hypertrophic) subaortic stenosis was present in 19 cases, membranous subaortic stenosis in two. The third type was seen in each of four cases with persistent common atroventricular canal, the basis for subaortic stenosis being adhesion of the anterior mitral leaflet to the edge of the deficient ventricular septum.

Three of the cases with muscular subaortic stenosis were examples of the Shone syndrome being associated with parachute mitral valve and supravalvular ring of the left atrium (see Left Ventricular Inflow Obstruction).

Tubular hypoplasia or abnormal communications, or both, commonly occurred in those cases with subaortic stenosis, this association being seen 16 and 12 times, respectively.

Congenital Unicommissural Aortic Valvular Stenosis. This was observed in six cases of the total series, five in group I and one in group II. Associated hypoplasia of the left ventricle and of the mitral valve was seen in four of these cases.

Aortic Atresia. This was encountered in seven cases, each in group I. The incidence of aortic atresia in this particular age group was 9%. In each of these cases, a patent ductus was present but it was not listed as an anomaly.

Left Ventricular Inflow Obstruction (Fig. 5)

Conditions encountered which were responsible for obstruction to left ventricular inflow included (1) left ventricular and mitral valvular hypoplasia associated with fibroelastosis of the left ventricular endocardium, (2) parachute deformity of the mitral valve, (3) stenosing supravalvular ring of the left atrium, and (4) mitral valvular atresia. In 26 cases of the total series at least one of these anomalies was present and, in some of the affected, more than one of these conditions were present. Of the 26 cases, 19 were in group I (25%), and seven were in group II (30%).

Hypoplasia of the Left Ventricle and Mitral Valve. This occurred with left ventricular endocardial fibroelastosis in 10 cases of the total series (eight cases [10%] in group I and two [9%] in group II). The latter two patients

Figure 5

Distribution and incidence of conditions characterized by left ventricular inflow obstruction found in association with coarctation of the aorta.
were seven and 12 months old, respectively, and each had associated congenital aortic stenosis.

In group I, the eight cases classified as showing left ventricular inflow obstruction characterized by underdevelopment of the left ventricle were uniformly associated with left ventricular outflow obstruction. Five of these cases manifested aortic atresia and three unicommissural aortic valvular stenosis. (The remaining two cases of aortic atresia present in the total series were associated with mitral atresia and so classified.)

Parachute Mitral Valve. This condition was encountered in 10 cases, represented by six cases (8%) of group I and by four cases (17%) of group II. Included in this category is the phenomenon in which the papillary musculature is localized to one area, either as only one left ventricular papillary muscle or as both the posteromedial and anterolateral papillary muscles being present but very closely allied. The chordae of both mitral leaflets converge to insert into the abnormal papillary musculature. Such valves are usually stenotic.

Stenosing Supravalvular Ring of the Left Atrium. This was seen in seven cases of the entire series, three among the 77 cases of group I (4%) and four among the 23 cases of group II (17%). In three cases, this lesion was associated with a parachute mitral valve, each presenting the complete picture of a developmental complex described by Shone and associates.5

Mitral Valvular Atresia. This was observed in five cases, each in group I. In four of these cases left ventricular hypoplasia was associated and, in two of these four, aortic atresia was also present. The fifth case presented with a single ventricle.

Positional Anomalies of the Great Arteries (Fig. 6)

Exclusive of cases with single ventricle, three types of positional anomalies of the great vessels were observed among 13 cases of this series with the following types represented: (1) complete transposition of the great vessels, (2) double outlet right ventricle, and (3) biventricular origin of pulmonary trunk with subaortic stenosis. Twelve of the 13 patients were under 6 months of age (16% of group I), and only one case belonged to group II (4%).

Complete Transposition. The most common type of positional anomaly of the great vessels in this series, represented by eight cases (seven [9%] in group I and one [4%] in group II) was complete transposition. A ventricular septal defect was present in six of the seven cases of group I with complete transposition. The one case in group II was associated with a ventricular septal defect, the patient being 7 months old at the time of death.

Double Outlet Right Ventricle. This condition was observed in four of the 100 cases, each in group I (5%) and each without pulmonary stenosis. In two of the cases, the malformation was of type II according to the classification of Lucas and associates (Tausig-Bing heart) and two cases, type I.

Biventricular Origin of the Pulmonary Trunk. This condition, with subaortic stenosis, a complex described by Becu and associates,7 was seen once; this was in a 4-month-old infant.

Accuracy of Clinical Diagnosis

The records, when adequate, were studied to determine whether or not the diagnosis of coarctation and of associated anomalies, when present, had been made clinically.

In the total series of 100 consecutive specimens with coarctation of the aorta, 13 were devoid of associated malformations. In these, the clinical records available to us were considered to be adequate in 12 instances. In 10, the diagnosis of coarctation had been made.

Associated malformations, exclusive of bicuspid aortic valve, were observed in the remaining 87 specimens. In 10 of these, there was lack of sufficient data to ascertain whether or not the diagnosis of coarctation had been established during life, leaving for analysis 77 cases with associated anomalies and with adequate clinical records. In 38 (49%) of the 77 cases, the diagnosis of coarctation had been made correctly, while this diagnosis had not been made in 39 (51%). In nine (12%) of the
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Distribution and incidence of conditions characterized by the occurrence of positional anomalies of the great vessels in association with coarctation of aorta. This grouping does not include cases of single ventricle with transposition of the great vessels.

38 cases with a correct clinical diagnosis of coarctation, an associated anomaly had been correctly diagnosed. In 29 cases (38%), the additional anomaly had not been indicated.

Among the 39 cases with an associated anomaly and in which the diagnosis of coarctation had not been made, there were 20 cases (26%) in which the associated anomaly had been correctly diagnosed. In the remaining 19 cases (25%), neither the coarctation nor the associated malformation had been identified clinically.

Discussion

Associated malformations were found in 87% of this series of 100 consecutive necropsy specimens exhibiting coarctation of the aorta. It should be emphasized, however, that this percentage may give a wrong impression as to the true incidence of associated malformations among patients with coarctation of the aorta, since the material was selected in the sense that each case represented a fatality. Several clinical series (Glass and associates8; Watersston and Aberdeen9; Sinha and associates10) present an overall incidence of associated malformations of approximately 70% among infants presenting with coarctation and congestive heart failure. These reports indicate a trend for a direct relationship between the presence of associated malformations and early onset of symptoms.

This trend is supported by our material which showed that, in the patients dying before 6 months of age, the incidence of associated malformations was greater than in those over 6 months of age, the incidences being 91 and 74%, respectively. These figures do not include bicuspid aortic valve, primarily since this condition only rarely is of importance in the young. Nevertheless, the high overall incidence of bicuspid aortic valve (46%) should be kept in mind, since late complications may arise from it. There were two young adult patients in our series, in each of whom it was necessary to replace the aortic valve because of aortic stenosis, resulting from secondary changes in a congenital bicuspid valve.

Exclusive of bicuspid aortic valve, the most common type of associated malformation was tubular hypoplasia of the aortic arch. The presence of this anomaly was more common in patients under 6 months of age (65%) than in the older age group (22%). Sinha and associates10 present a similar percentage (78%) of tubular hypoplasia in their series of infants under 6 months of age with coarctation and signs of congestive heart failure. The early recognition of the association of tubular hypoplasia and coarctation could be of significance in suggesting the presence of other anomalies, since in our series each case with this condition presented an additional malformation, as well. Among these, ventricular septal defect and muscular subaortic stenosis were the most common "third" lesions, each being present in 14 of the 44 patients with tubular hypoplasia. Either ventricular septal defect or subaortic stenosis may play an important role in altering cardiac dynamics in addition to the coarctation and the tubular hypoplasia.

Abnormal communications were present in approximately equal percentages in both age groups (48% in group I and 52% in group II),
but persistent patent ductus arteriosus and ventricular septal defect were more common in infants under 6 months of age than in older infants. It will be recalled that a patent ductus was considered an abnormality only when it was seen in a subject more than 2 months of age. With this stipulation in mind, it was observed that in group I 45% of the patients over 2 months of age showed a patent ductus. In group II, the percentage was 35.

The most common intracardiac malformations associated with coarctation were ventricular septal defect and conditions peculiar to the left side of the heart.

Ventricular septal defect was the leading type of intracardiac abnormal communication. Several clinical studies reported a similar frequency to that found with this study.

Lucas (Lucas RV Jr: Personal Communication to the authors) observed that patients with coarctation and ventricular septal defect may improve by surgical removal of the coarctation as a consequence of reducing systemic vascular resistance with corresponding decrease in the magnitude of the left-to-right shunt. A similar relationship exists in persistent common atrioventricular canal and in single ventricle. The low incidence of atrial septal defect (slightly higher in the group over 6 months of age) indicated the relatively insignificant role of this defect.

Among the anomalies peculiar to the left side of the heart were those responsible for left ventricular outflow and inflow obstruction, the former being more common, particularly in the younger age group. The most common type of anomaly causing left ventricular outflow obstruction was subaortic stenosis of the muscular type. Aortic valvular stenosis accounted only for a minority of cases but was of major degree when present.

Left ventricular inflow obstruction occurred in about the same frequency in two age groups, although the conditions causing this disturbance tended to be different in the two groups. In the younger, mitral and left ventricular hypoplasia were the most common causes, while in the older group, parachute mitral valve and supravalvular ring of the left atrium were the most common causes.

Positional anomalies of the great vessels encountered in 13 cases were regarded as a separate entity in which the serious prognosis is independent of the presence or absence of coarctation of the aorta.

From a view of the entire material studied, it will be recalled that the relationship between the site of coarctation and the aortic entrance of the ductus was most commonly the one in which the coarctation lay either proximal to or opposite the ductus. This relationship is characteristic in those patients with coarctation in whom symptoms appear at a young age and of itself may have a bearing in causing left ventricular failure in infancy.

This is in contrast to those patients who survive to adult life in whom a reverse relationship usually applies.

It had been hoped to determine the incidence of correct clinical diagnosis of coarctation and of associated anomalies, when present, by reviewing the clinical records. This goal was not adequately fulfilled because the specimens had come from a relatively large number of different institutions and there was a wide range in quality of clinical data available. Recognizing these shortcomings, it was apparent that even with "adequate" clinical records the clinical diagnosis not infrequently failed to identify the entire picture when coarctation was associated with an additional anomaly of significance.

In instances of incomplete diagnosis, the cases in which the coarctation was diagnosed but the associated conditions went undetected about equaled those in which the associated condition was identified while the coarctation was unrecognized.

We are left with the conclusion that, although the seriously ill infant or child with coarctation may have no other malformations, there is a highly likely chance of an additional cardiac malformation of functional significance being present.

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Circulation. 1970;41:1067-1075
doi: 10.1161/01.CIR.41.6.1067

Circulation is published by the American Heart Association, 7272 Greenville Avenue, Dallas, TX 75231
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Print ISSN: 0009-7322. Online ISSN: 1524-4539

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located on the World Wide Web at:
http://circ.ahajournals.org/content/41/6/1067

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