A SYNDROME OF CONGENITAL DEFECTS INVOLVING THE ABDOMINAL WALL, STERNUM, DIAPHRAGM, PERICARDIUM, AND HEART

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During recent years, several patients with an unusual combination of congenital defects have been seen in The Johns Hopkins Hospital. The anomalies observed in each patient were: (1) a midline, supraumbilical abdominal wall defect; (2) a defect of the lower sternum; (3) a deficiency of the anterior diaphragm; (4) a defect in the diaphragmatic pericardium; (5) congenital intracardiac defects. The repeated association of these abnormalities suggested that this combination represented a clinical syndrome and stimulated a search for additional examples. This article was written to summarize the available clinical material, to propose a probable embryologic basis for the coexistence of these defects, and to consider the surgical treatment of patients with this syndrome.

CASE REPORTS

Case 1. C. D., a male infant, was delivered at The Johns Hopkins Hospital by Dr. W. Drummond Eaton on February 8, 1957. In the family history there were no other instances of congenital defects. This was his mother’s first pregnancy, during which there had been no recognized infections or unusual environmental exposures. Delivery was uncomplicated, birth weight was 3,170 grams. A large omphalocele was immediately apparent (Fig. 1), and it was further noted that the cardiac pulsations were abnormally prominent. The sternum was shortened and the costal arch was elevated and flared. The contour of the heart was visible beneath the skin of the unusually high epigastrium. There was a defect of the musculature of the abdominal wall above the omphalocele; the medial edges of the rectus muscles were widely separated and inserted on the costal margins at the midclavicular line. The child appeared otherwise normal. No cardiac murmurs were audible.

The child was operated upon (J.R.C.) under ethane-ether anesthesia. The amniotic sac was excised with a small margin of skin (Fig. 2). The liver, which occupied the upper portion of the omphalocele, could then be depressed to reveal a large defect in the ventral portion of the diaphragm (Fig. 3A). The diaphragm arose normally from the posterior and lateral aspects of the rib cage, but at a point in line with the insertion of the rectus muscles on the costal margins the diaphragm curved posteriorly, leaving a semilunar defect which was bounded dorsally by a small cuff of diaphragm on the liver immediately ventral to the inferior vena cava. There was a corresponding defect in the pericardium. The serous linings of the peritoneal and pericardial cavities were continuous across the edge of the diaphragmatic defect. The pleurae were intact. The heart lay somewhat further to the right than normal, and there was an anomalous anterior coronary artery. In the abdomen there was failure of attachment of the mesentery of the small bowel and right colon. The cecum lay in the epigastrum, and the appendix lay within the pericardial cavity.

Attention was first directed to closure of the dia­phragmatic defect. The junction of pericardium and peritoneum was incised along the edge of the diaphragmatic defect and the pericardium reflected from the diaphragm. The edge of the diaphragm was then sutured to the anterior rib cage (Fig. 3B). It was apparent that the rectus muscles could not be approximated. It was possible, however, to develop a layer of fascia which was continuous with the medial border of the rectus sheath and had the appearance of linea alba (Figs. 4 and 5). The edges of this fascia were brought together in the midline after a long relaxing incision had been made in each anterior rectus sheath.

The child recovered satisfactorily but during his convalescence a murmur was noted over the left second interspace. Mild congestive failure developed which required digitalization. Radiologic study
demonstrated a large right ventricle and an electrocardiogram revealed right ventricular hypertrophy. A presumptive diagnosis of tetralogy of Fallot was made although cyanosis was not apparent. He was discharged in satisfactory condition at 3 weeks of age. He was readmitted at 3 months of age because of large symptomatic indirect inguinal hernias. The pulmonary systolic murmur was again noted and cyanosis was detected on crying. The abdominal wall repair was intact (Fig. 6). His weight gain and development were approximately normal. The hernias were repaired without event. He has continued to develop normally although there is slight cyanosis on vigorous crying. Subsequent angiocardiography has substantiated the diagnosis of tetralogy of Fallot. Correction of the cardiac defects has been deferred until the child is older.

Case 2. M. F. was first seen in the Cardiac Clinic in 1948 at the age of 16 years. He was the second of 3 siblings. Absence of the lower third of the sternum and a large defect of the abdominal musculature above the umbilicus were noted at birth. Cyanosis was first detected at 2 1/2 years and a cardiac murmur was heard at that time. He subsequently developed shortness of breath, increasing cyanosis, and marked diminution of exercise tolerance.

On physical examination, the patient was noted to be small and poorly nourished. There was an 8 by 10 centimeter defect of the musculature of the abdominal wall above the umbilicus (Fig. 7). The rectus muscles inserted into the costal margins at the midclavicular line. The lower sternum was absent and only 4 ribs joined the remaining portion. The defect was covered by hyperpigmented skin with little if any subcutaneous tissue. The contours of the heart, liver, and intestines were visible in the defect. No murmurs could be heard although cyanosis and clubbing of the extremities were marked. The physical examination was otherwise normal.

The hematocrit was 76, and an electrocardiogram revealed right axis deviation and right ventricular hypertrophy. Fluoroscopy revealed dextroversion of the heart and a left aortic arch. Cardiac catheterization and angiocardiographic studies demonstrated a functional single ventricle and pulmonic stenosis.

On May 1, 1948 an anastomosis was performed between the end of the right subclavian and the side of the right pulmonary artery by Dr. William P. Longmire. After operation a loud, continuous murmur was audible and cyanosis was strikingly diminished.

The patient returned for study 4 years later. He was at that time a college student and reported that he was no longer cyanotic except on extreme exertion. There was no visible cyanosis, and the loud continuous murmur was again noted. The abdominal wall defect was unchanged.

On May 29, 1952, repair of the ventral defect was carried out (M.M.R.). The defect was found to be covered only by skin and peritoneum. When the liver had been reflected downward, one could visualize a large semilunar defect in the anterior diaphragm with a corresponding defect in the diaphragmatic pericardium (Fig. 8). The pericardium was adherent to the heart at the margin of this defect. There was no defect in the pleurae. With dissection of the pericardium from the pleurae, diaphragm, and epicardium (Fig. 9) the heart retracted into the thorax although the apex remained visible in the epigastric defect. It was impossible to approximate the rectus muscles. Long incisions were made in the anterior rectus sheaths, and the medial fascial flaps were reflected to the midline where they were sutured (Fig. 10A). This closure was reinforced by a sheet of autogenous fascia lata (Fig. 10B). An excellent result has been noted on subsequent examinations.

Case 3. R. B., a little boy, was first seen in The Johns Hopkins Hospital at the age of 2 years. This was his mother's first pregnancy, during which there had been no recognized illnesses or unusual environmental factors. There was no history of congenital defects in the family. The child was noted at birth to have a defect of the sternum and a cardiac murmur. Subsequently, increasing cyanosis and limitation of activity had developed.

Physical examination revealed a well developed child with generalized cyanosis and marked clubbing of the fingers. The lower half of the sternum was absent. This anomaly and an associated wide separation of the rectus muscles above the umbilicus pro-
produced a large thoracoepigastric defect in which the apex of the heart was palpable beneath the skin. A harsh systolic murmur was audible over the entire precordium, maximal in the second intercostal space.

The hematocrit was 46. An electrocardiogram showed right axis deviation and right ventricular hypertrophy. Fluoroscopy revealed dextroversion of the heart and a left aortic arch. An angiogram showed marked overriding of the aorta, an interventricular septal defect, and pulmonic stenosis.

It was thought that this patient represented a typical tetralogy of Fallot, and on July 2, 1957 a right subclavian-pulmonary anastomosis was performed (J.A.H.). The clinical response was excellent
and the child was discharged from the hospital much improved (Fig. 11). He is to return for repair of the other defects.

**Case 4. R. K., a 6 weeks old white male, was admitted to The Johns Hopkins Hospital in 1952 because of an upper abdominal defect which had been present since birth. This was his mother's only pregnancy. There was no history of congenital defects in the family nor had there been any significant diseases or unusual environmental factors during the mother's pregnancy. Delivery was said to be uncomplicated but a large upper abdominal defect was immediately apparent. No other anomalies were noted. Progress was satisfactory except for a single episode of cyanosis on vigorous crying at 3 weeks of age.

Physical examination on admission revealed a well nourished infant who had an obvious defect in the ventral body wall (Fig. 12). The sternum was shortened and a 9 by 5 centimeter defect was noted in the upper abdomen with wide separation of the rectus muscles. A pulsatile structure extending to the umbilicus was palpable within the defect. Its relation to the heart could not be determined. The heart was enlarged to the right, and there was a loud systolic murmur, maximal in the right fourth intercostal space. The child was not cyanotic.

Hematologic studies were unremarkable. X-ray examination and fluoroscopy of the chest revealed cardiac enlargement. An electrocardiogram revealed a balanced axis. Angiocardiography showed dextrorotation of the heart, an interventricular septal defect, and a diverticulum arising from the left ventricle.

Excision of this diverticulum was attempted but cardiac arrest occurred shortly after the skin incision was made and the patient expired.

A limited postmortem examination did not define the nature of the sternal defect. There was a dilated right atrium and hypertrophy of the right ventricle. The foramen ovale was patent. There was a 5 millimeter patent ductus arteriosus and a persistent left superior vena cava which entered the coronary sinus. A high interventricular septal defect measuring 2.5 centimeters in diameter was noted. The left ventricle was of normal size but from the apex of this ventricle arose a diverticulum which extended downward subcutaneously for a distance of 6.5 centimeters. The lumen of this diverticulum was 4 millimeters in diameter and was lined by endocardium. There were defects of the pericardium and ventral diaphragm through which the diverticulum passed. The diaphragmatic defect was semilunar in shape with the dorsal margin of the defect at the point of fusion of diaphragm and liver. The diaphragm joined the rib cage on either side at the midepigastric line.

**Case 5. A. S., a 5 months old white female, was admitted in 1952 because of cyanosis and episodes of unconsciousness. There were no congenital defects in the family history. This was the mother's first preg-

![Fig. 6](image-url) **Case 1. Photograph taken at 3 months showing the abdominal wall repair and the defect of the costal margin and sternum. Bilateral inguinal hernias have been repaired.**

nancy, during which there had been no recognizable viral infections or unusual environmental exposures. The child was delivered at another hospital in a breech presentation and was immediately noted to be cyanotic with a loud systolic murmur over the base of the heart. She was also found to have an omphalocele. Hospital records reveal that the omphalocele was repaired several hours after birth at which time a "pulsatile, bluish, pointed structure," thought to be the apex of the heart, was observed and was "pushed back into the chest cavity." Recovery from this operation was satisfactory but the child became increasingly cyanotic. At 4 months of age she began to have episodes of intense cyanosis associated with rigidity, opishotonos, and unconsciousness.

At the time of her admission to this hospital, she was malnourished and cyanotic with clubbing of the extremities. A loud systolic murmur was heard over the base of the heart, maximal in the left second and third intercostal spaces adjacent to the sternum. There was wide separation of the rectus muscles which inserted into the costal margins at the midepigastric line.
line. It was thought that the xiphoid was missing and a pulsatile mass was noted beneath the skin of the abdominal wall defect.

The hematocrit was 60. Arterial oxygen saturation was 45 per cent with the child crying. An electrocardiogram revealed slight left axis deviation. Fluoroscopy revealed enlargement of the heart with marked prominence of the right cardiac border. There was a left aortic arch.

A diagnosis of tricuspid atresia was made and, because of the severity of symptoms, the child was operated upon shortly after admission to the hospital. Cardiac arrest occurred soon after the chest was entered but resuscitation was successful and a subclavian-pulmonary arterial anastomosis was constructed. The child did poorly, however, and died several hours postoperatively.

At autopsy, an upper abdominal wall defect was noted with wide separation of the rectus muscles which inserted into the costal margins at the midthoracic line. A diverticulum of the left ventricle extended through a ventral defect in the diaphragm and a corresponding defect in the diaphragmatic pericardium. This diverticulum arose from the apex of the ventricle and extended about 2 centimeters below the diaphragm. A persistent left superior vena cava entered the coronary sinus. There was a large interatrial septal defect and a high interventricular septal defect. A common pulmonary vein was found to drain both lungs into the left auricle. The tricuspid valve was atretic. There was a single coronary artery.

**CLINICAL ANALYSIS**

Previously, patients with this combination of anomalies have been considered to be examples of ectopia cordis because of the abnormally exposed position of the heart. The signal articles bearing upon this subject are those of Greig, Byron, and Major. Greig summarized the earlier reports of ectopia cordis but did not analyze associated anomalies. Byron first suggested a distinction between the other categories of ectopia cordis and those associated with abdominal wall defects; he proposed that the latter be segregated as a subgroup under the term thoracoabdominal. Major has more recently presented a review of this type in which he quoted 13 cases and added a case of his own.

The clinical data available on the previously reported patients are summarized in Table I and those on the patients herein reported in Table II. Analysis of this material reveals several facts previously unrecognized: (1) Rather than a single abnormality, these patients present a remarkably consistent association of defects which includes anomalies of the abdominal wall, lower sternum, ventral diaphragm, pericardium, and heart. (2) The ventral diaphragmatic defect is a specific anomaly unrelated to the commonly recognized diaphragmatic defects. (3) Intracardiac anomalies are invariably present. (4) True ectopia of the heart is rare, occurring in but 1 patient of the entire group. Except for some degree of dextrorotation which may be present, the heart usually lies in normal relationship to the other thoracic viscera; the appearance of ectopia is created by the concomitant defects of the sternum, diaphragm, and abdominal wall which leave the heart abnormally exposed.

On the basis of these observations, the inclusion of these patients in the category of
ectopia cordis would appear to be in error. Rather, this consistent association of anomalies should be considered as a specific syndrome.

Variants of this syndrome would be anticipated in view of the postulated embryologic development of these defects. Potts has reported a patient who demonstrated cardiac, abdominal wall, and diaphragmatic defects but had a normal sternum and an intact though abnormal pericardium. Major has successfully treated a newborn infant who had an omphalocele, a defect of the sternum and lower thorax, and a ventral defect of the diaphragm without an associated pericardial defect and no clinically apparent intracardiac anomalies. These are obviously closely related conditions and may be explained on the basis of less extensive embryologic defects. While most of the component elements of this syndrome may occur as isolated anomalies, the ventral diaphragmatic defect probably will not be found in the absence of concomitant defects of the sternum and abdominal wall. It should be emphasized that this anomaly is not related to foramen of Morgagni defects but is embryologically a separate entity.

The occurrence of congenital intracardiac anomalies as a constant element of this complex has not been previously recognized. It would appear from the present study that such lesions should be considered an integral part of this association. An analysis of these
cardiac defects (Table III) reveals that an interventricular septal defect was present in every instance in which a description of the heart was available. An interatrial septal defect was present in 53 per cent of the patients. Either valvular or infundibular pulmonary stenosis occurred in 33 per cent of the patients. The only established cardiac syndrome to be noted with any frequency was the tetralogy of Fallot which was seen in 20 per cent of the patients. A left ventricular diverticulum was present in 20 per cent.

It is of clinical importance that only 4 of the entire group of patients had other significant congenital defects, and but 1 of these was incompatible with life. In only 3 patients were the total anomalies such as to preclude successful correction.

EMBRYOLOGIC DEVELOPMENT

Normal embryology. An analysis of the normal embryology of the involved structures is essential to clarification of the relationship of these associated anomalies.

Origin of the mesoderm.—During the eighth day of embryonic development, a layer of cells differentiates from the inner surface of the inner cell mass and pushes out into the blastomere; this is the primitive endoderm which will eventually form the lining of the gut. Immediately thereafter, a localized proliferation of cells is noted on the inner surface of the embryonic disc and migrates laterally between the trophoblast and endoderm; this is the primitive mesoderm. These cells migrate rapidly in a peripheral fashion so that the mesodermal layer soon extends not only beneath the embryo but also lines the entire blastodermic vesicle. At about 16 days the mesoderm splits laterally into an outer somatic layer and an inner splanchnic layer (Fig. 13). The cavity between these layers is the primitive coelom. This somatic-splanchnic differentiation does not occur everywhere simultaneously; it is significant that it appears first in the mesodermal tissue from which the heart and pericardium will develop.

Development of the heart and pericardium.—The heart arises far cephalad from paired primordia which are brought together beneath the embryo at about the
The seventeenth day by the downfolding of the lateral splanchnopleure and ventral flexion of the embryo. This ventral folding appears first in the cephalic and caudal portions of the embryo so that the heart reaches its definitive position and begins its differentiation very early in embryonic life (Fig. 13). The myocardium and epicardium are derived from the splanchnic layer of mesoderm while the pericardium arises from the corresponding somatic mesoderm. The intimate developmental relationship of these structures is apparent.

Development of the diaphragm.—From the mesoderm adjacent and immediately anterior to the cardiac primordia arises the transverse septum. This tissue comes to lie in the midline just caudal to the heart by the same process of flexion and ventral folding and is the anlage of the ventral portion of the diaphragm (Fig. 14). The transverse septum develops contemporaneously with and in close proximity to the heart and pericardium and is derived from the same primitive layer. The dorsal and lateral portions of the diaphragm are derived from the pleuropericardial and pleuropitoneal folds which coalesce with the transverse septum.

Development of the abdominal wall.—It is thought that the somatic mesoderm contributes primarily to the subcutaneous connective tissue of the anterior abdominal wall. The muscular components are derived from the dorsal mesoderm of the somites. The dorsomesial portion of each somite proliferates and extends around toward the ventral midline; termed the myotome, this tissue splits to form the various muscle layers. The skin of the ventral body wall is derived from the embryonic ectoderm which is continuous with the amnion. The development of the skin is not closely related to that of the muscles.

Development of the sternum.—The sternum is formed from paired lateral concentrations of mesenchymal cells in the thoracic region of the embryo (Fig. 15). These cannot be clearly identified prior to the fifth week but the primitive mesoderm from which they are derived is obviously laid down at a much earlier date. They are converted into precartilage and advance toward the midline where they fuse into a single structure. The process of fusion begins at the cephalic end and progresses caudally. Simultaneously, rib cartilages are growing ventrally from the vertebrae and by the ninth week the sternum is complete and the anterior rib ends have become attached.

Development of the specific defects. The anomalies seen in this syndrome may be classified into two groups based on the mechanism of embryologic development. The first group, comprising 3 of the defects, would appear to arise as the result of developmental failure of a segment of the mesoderm.
The diaphragmatic defect results from total or partial failure of the transverse septum to develop. When complete, the defect corresponds exactly to that portion of the definitive diaphragm which is derived from the transverse septum; it is a ventral defect extending laterally to the region of the pleuropertitoneal folds and dorsally to the point of attachment of liver and diaphragm (Fig. 14). Less extensive defects represent partial rather than total loss of the transverse septum. This anomaly is not related to defects of the foramen of Morgagni.

The pericardial defect is closely related to faulty development of the transverse septum. That portion of the pericardium which is to lie on the diaphragm arises from the somatic mesoderm immediately adjacent to that region of the same layer from which the transverse septum is derived. Defective development of one of these structures without a corresponding defect in the other requires a highly specific loss of somatic mesoderm and is seen only in the uncommon variants in which the diaphragmatic defect is incomplete. Coexisting defects are much more probable and are seen in the great majority of patients.

The intracardiac lesions noted are the result of faulty development of the epimyocardium which is derived from the splanchnic mesoderm corresponding to that portion of the somatic mesoderm from which the pericardium is derived. The cardiac anomalies

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<th>Patient</th>
<th>Sternal</th>
<th>Abdominal wall</th>
<th>Diaphragmatic</th>
<th>Cardiac</th>
<th>Pericardial</th>
<th>Other</th>
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<tr>
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<td>Malrotation intestines</td>
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</tr>
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<td>Ventral defect</td>
<td>Tetralogy of Fallot</td>
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<td>Malrotation intestines</td>
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<td>Ventral defect</td>
<td>IASD, IVSD</td>
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<tr>
<td>4. R.K.</td>
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<td>Ventral defect</td>
<td>IASD, IVSD</td>
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<td>5. A.S.</td>
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<td>Ventral defect</td>
<td>IASD, IVSD</td>
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demonstrated in these patients are varied but it is striking that in all patients adequately described, defects of the cardiac septa were present. The exact origin of the individual cardiac structures from the primordial tissues is not defined but the consistency of septal defects in these patients might provide a clue to the derivation of these structures.

The second group includes the sternal and abdominal wall defects. These also appear to arise as the result of failure of the normal development of the mesodermal components but would seem to evolve by a somewhat different mechanism than that described for the diaphragmatic, pericardial and intracardiac anomalies.

The sternal defect results from failure of complete ventral migration of the paired primordial structures. It is important to realize that all of the elements of the sternum are present but lie in a position lateral to their normal midline location. The costal cartilages connect with cartilaginous plates which represent the paired sternal anlagen and, in most instances, variable degrees of fusion occur in the more cephalic portion of the thorax. The defect, therefore, does not represent absence of the sternal primordia but rather failure of the paired primitive structures to complete migration.

The abdominal wall defect arises in similar fashion. Here, also, the defect is due not to absence of the normal elements of the ventral abdominal wall but to a failure of complete ventral migration of the myotomes in this region so that normal differentiation into the various muscle layers occurs at an abnormally lateral point. This is illustrated in these patients by the structurally normal rectus muscles which attach correctly to the pubis but deviate laterally as they run cephalad so that they insert into the costal margin at the midclavicular line.

At least two theories may be proposed to explain the failure of ventral migration of the sternal anlagen and of the myotomes: (1) The existence of a primary defect in the growth potential of the migrating tissues. (2) An absence or deficiency of ventral paramidline mesenchymal tissue into which the migrating structures would normally grow. The first possibility seems unlikely in view of the otherwise normal development and differentiation of these tissues. The second alternative is the more attractive since it postulates a developmental defect which is basically similar to that proposed for the associated anomalies of the diaphragm, pericardium, and heart, i.e., absence or
Immediately after the differentiation of the primitive intraembryonic mesoderm into its splanchnic and somatic layers since derivatives of both these layers are involved. This localizes the time of origin at about 14 to 18 days of embryonic life. The basic etiology of this mesodermal maldevelopment remains obscure.

**TREATMENT**

In view of the multiplicity of problems posed by these patients, it is not practical to formulate a specific plan of attack which will be optimal for all patients but, in general, treatment should consist of immediate surgical correction of as many of the defects as possible. Repair of the cardiac lesions should be deferred, however, since diagnosis in the neonatal period is difficult and inexact. Furthermore, total correction will require an open heart procedure in most or all of these patients and this technique is at present unsatisfactory in the newborn. The cardiac lesions noted in the reported patients were not so severe as to require immediate correction. A possible exception to this generalization is the repair of ventricular diverticula which may be amenable to immediate correction.

Some of the sternal defects will not require correction. Minor abnormalities of the lower sternum are not cosmetically or physiologically serious. Should there be an extensive defect, however, immediate repair must be strongly considered since the correction of large defects becomes progressively more difficult with advancing age.

In those cases in which an omphalocele is present, immediate surgical intervention is, of course, mandatory and it would seem logical to attack any other reparable defects at that time. Certainly in such cases there can be no justification for deferring repair of the diaphragmatic defect. Should the omphalocele be sufficiently large so that primary repair is difficult or impossible, necessitating simple closure of the skin over the viscera, it would seem doubly important to close the diaphragmatic defect to avoid...
the exertion of undue pressure on the intrathoracic contents and herniation of abdominal viscera through this defect into the pericardium. Repair of even the largest diaphragmatic defects of the type seen in these patients can be accomplished in the newborn (as in our Case 1) by bringing the remaining diaphragm anteriorly and attaching it to the anterior thoracic wall and costal margin. This is easily done since the remaining diaphragm is structurally normal and permits the necessary anterior displacement without undue tension. This maneuver simultaneously obliterates the pericardial defect.

Should the patient have an intact skin covering over the defect in the muscular portion of the abdominal wall, repair might be deferred. There is little to be gained by such delay, however, since the ability of the newborn to withstand operation is well known and will not be significantly augmented until well past the neonatal period. It is generally conceded that most congenital defects do not become more amenable to repair with increasing age but rather that correction becomes more difficult.

The technique of abdominal wall closure in such patients is worthy of specific comment. The lateral displacement of the rectus muscles precludes their approximation. In our Case 1, it was possible to develop a fascial structure which extended ventrally from the medial border of the rectus muscles and resembled linea alba. A strong closure of the abdominal wall defect was thus obtained using this local tissue. Linear relaxing incisions in the anterior rectus sheath are important in relieving tension on this closure.

The increasing difficulty of repair attendant upon delay in correction is illustrated by Case 2. This patient was operated upon at 20 years of age, at which time it was impossible to isolate the fascial structure described in Case 1 and closure of the abdominal wall was accomplished only with difficulty by swinging flaps of anterior rectus sheath medially and the insertion of autogenous fascia lata as reinforcement. Extensive dissection of the pericardium from the heart and surrounding structures was necessary to accomplish the repair. Closure of the diaphragmatic defect was not considered feasible. In all probability, a much simpler procedure would have been possible in infancy.

**TABLE III.—ANALYSIS OF CONGENITAL CARDIAC LESIONS IN 15 ADEQUATELY DESCRIBED PATIENTS**

<table>
<thead>
<tr>
<th>Lesion</th>
<th>No. of cases</th>
</tr>
</thead>
<tbody>
<tr>
<td>Interventricular septal defect</td>
<td>15</td>
</tr>
<tr>
<td>Interatrial septal defect</td>
<td>8</td>
</tr>
<tr>
<td>Pulmonary stenosis</td>
<td>5</td>
</tr>
<tr>
<td>Tetralogy of Fallot</td>
<td>3</td>
</tr>
<tr>
<td>Left ventricular diverticulum</td>
<td>3</td>
</tr>
<tr>
<td>Patent ductus arteriosus</td>
<td>3</td>
</tr>
<tr>
<td>Anomalous venous return (caval)</td>
<td>3</td>
</tr>
<tr>
<td>Tricuspid atresia</td>
<td>1</td>
</tr>
<tr>
<td>Truncus arteriosus</td>
<td>1</td>
</tr>
<tr>
<td>Anomalous venous return (pulmonary)</td>
<td>1</td>
</tr>
</tbody>
</table>

**SUMMARY**

The consistent association of a group of congenital anomalies involving the abdominal wall, lower sternum, ventral diaphragm, pericardium, and heart has been studied in 21 patients, 5 of whom are herein reported. This complex should be considered as a specific syndrome. It is inadequate and inaccurate to consider such patients as examples of ectopia cordis.
The entire group of anomalies would appear to be closely related in embryologic development, arising as the result of defective formation and differentiation of the ventral mesoderm at about 14 to 18 days of embryonic life.

The unusual ventral defect of the diaphragm seen in these patients results from failure of the transverse septum to develop. This specific anomaly has failed to gain proper recognition in the classification of congenital diaphragmatic defects. Case 1 is the first recorded instance in which repair of this defect has been accomplished.

The consistent association of congenital intracardiac lesions has not been previously noted. An interventricular septal defect would appear to be invariably present.

The coexistence of serious unrelated anomalies appears to be rare.

Treatment should consist of immediate surgical repair except for the intracardiac abnormalities. Correction of the latter is probably better deferred until more exact diagnosis and safer surgical intervention are possible.

REFERENCES

4. Ehrenhaft, J. L. Personal communication.
17. Sandifort, E. Quoted by Greig (8).