Congenital Absence of the Pulmonary Valve

By Lucien A. Campeau, M.D., Paul E. Ruble, M.D., and Warren B. Cooksey, M.D.

The autopsy on a 32-year-old man revealed an absence of the pulmonary valve, a ventricular septal defect, and a single coronary artery. Congenital absence of the pulmonary valve has not been previously reported. The diagnosis of the accompanying pulmonary incompetence is discussed in the light of the catheterization studies and clinical findings. The pathologic abnormalities as found at postmortem examination are described and the embryologic basis for the several congenital malformations found in this unusual case are discussed.

Congenital absence of the pulmonary valve has not been reported in the literature although cases of congenital pulmonary incompetence due to other causes have previously been described. The following case is presented because of the unexpected finding of such an anomaly in an adult who apparently tolerated it relatively well. This malformation was associated with a large ventricular septal defect and a single coronary artery.

Case Report

A 32-year-old white chef was admitted to the hospital because of dyspnea. The patient had been told that he had a heart murmur since the age of 4 years but had had no squatting or cyanosis. His growth and development were significantly retarded and he was unable to keep up with children of his own age. He was rejected by the Army at the age of 18, but was able to work as a chef in the merchant marine for the next 12 years before he began to show early signs of congestive heart failure.

The patient had been asymptomatic until about 2 years prior to admission, when he developed symptoms of increased fatigability, generalized weakness, and some exertional dyspnea. He had minimal pedal edema intermittently but no paroxysmal nocturnal dyspnea, orthopnea, or hemoptyis. These symptoms were treated intermittently with digitalis and mercurial diuretics with satisfactory improvement. One year prior to admission diagnostic studies including cardiac catheterization were performed at the National Institute of Health at Bethesda, Md. It was decided that surgical therapy could not be offered at that time and the patient was discharged without further therapy. He was admitted to our hospital because of a recent increase in symptoms.

Review of systems was negative for other significant illnesses. He had the usual childhood infectious diseases but no history of rheumatic fever. His mother died at the age of 67 with "blood poisoning."

His father died of a myocardial infarction at the age of 63. He had 4 brothers in good health.

Physical examination revealed an acutely and chronically ill man who was moderately dyspneic, orthopneic, and apprehensive. He was pale, weak, but cooperative. He appeared underdeveloped. There was moderate lip, perioral, and nailbed cyanosis. The hair was fine, sparse, and lifeless. The skin was cool and covered with diffuse perspiration. There were no petechiae of the mucous membrane or retina. The trachea was midline and no tug was present. The thyroid gland was not enlarged. There was no lymphadenopathy.

The respiratory rate was 32 per minute with some use of the accessory muscles of respiration. He had a pigeon-chest deformity, with the major bulge to the left of the midsternal line, and marked kyphoscoliosis. There were hyperresonance over the entire right side of this chest and dullness over the lower left side posteriorly. The right hemidiaphragm was depressed to the level of the eleventh rib and did not move with respiration. The breath sounds were vesicular over the entire right and upper portion of the left side of the chest. Breath sounds were diminished over the left lower lobe. There were fine crepitant rales over the right upper lobe anteriorly and over the left lower chest posteriorly and laterally. The remainder of the chest was normal to auscultation.

The radial pulses were full, equal, and regular at 130 per minute. The blood pressure was equal in each arm at 150/84. There was a turbulent movement of the precordium and a widespread systolic thrust: The left border of the cardiac dullness was in the midaxillary line and the point of maximum impulse was in the sixth and seventh interspaces in the midaxillary line. The supracardiac dullness was at the midclavicular line in the third interspace on the left and at the sternal border on the right. A rough, rumbling, grade IV systolic murmur was heard with a maximum intensity in the third and fourth interspaces at the sternal border and was widely transmitted over the entire precordium to the axilla and to the back. A less intense grade II diastolic murmur was heard over the base of the heart in the left third and fourth interspaces extending to the right of the sternum in the fourth interspace, giving

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the heart murmur a machinery-like quality. The heart sounds were obliterated by the murmur except at the apex where the mitral first sound was greater than the mitral second sound.

The abdomen was negative except for an enlarged, tender, smooth liver extending 5 cm. below the right costal margin in the right midclavicular line. There was no clubbing of the fingers. There was 1 plus pedal edema.

Laboratory data on admission revealed a hemoglobin of 15.6 Gm. per cent, a red blood cell count of 5.69 million, and a white blood cell count of 9,100, with 69 per cent neutrophils, 29 per cent lymphocytes, and 2 per cent basophils. Urinalysis revealed 4 plus albumin and a specific gravity of 1.008. There were 2 to 3 granular casts and occasional pus cells per low-power field on microscopic examination.

The admission chest roentgenogram (fig. 1) revealed a grossly enlarged heart apparently due mainly to right ventricular enlargement. There was gross dilatation of the main pulmonary artery, and the intrapulmonary vessels were prominent. At fluoroscopy the pulmonary arteries were markedly pulsatile and the aorta was hypoplastic. Clouding the left costophrenic sulcus suggested the presence of a minimal pleural effusion.

The patient was treated with oxygen, digitalis, diuretics, and analgesics. His temperature rose to 102 F. rectally and the pulse to 160 per minute 5 hours after admission and he had a marked increase in diaphoresis. He died quietly 9 hours after admission.

The following information was obtained from the National Institute of Health, Bethesda, Md. where the patient had been intensively studied 1 year ago. The electrocardiogram was reported to show right bundle-branch block, with right ventricular and atrial hypertrophy. Cardiac catheterization revealed a pulmonary artery pressure of 180/23–28 and a right ventricular pressure of 180/0. The pressure curve obtained from the pulmonary artery is presented in figure 2. A gradient of 3 to 4 volumes per cent oxygen content was found between the right ventricle and the right atrium.

It was thought that this specimen of highly oxygenated blood obtained at the right ventricular level had been drawn from the left ventricle through a ventricular septal defect. The total systemic blood flow was calculated at 15 L. per minute and the pulmonary blood flow at 2.72 L. per minute, giving a right-to-left shunt of 12.3 L. per minute. These figures were considered to be unreliable because the patient, who was questionably cyanotic before catheterization, became frankly so during the procedure, indicating a possible reversal of shunt. The cyanosis, however, was also partially due to pulmonary factors, as evidenced by the improvement in the arterial oxygen saturation after oxygen was administered by mask. The angiocardiogram was of no diagnostic aid because of the great dilution of the dye in the dilated right ventricle and pulmonary artery. The data from the cardiac catheterization suggested the presence of an interventricular septal defect and pulmonary hypertension.

Necropsy Findings. An autopsy was performed by Dr. W. L. Brosius.* The body was poorly developed, with the chest deformity previously described. Approximately 200 ml. of clear yellow fluid were found in each pleural space. The right and left lungs weighed 730 and 444 Gm. respectively. There were dependent congestion and emphysematous changes in both lungs. The left upper lobe contained large emphysematous bullae. There were no pulmonary

* We are indebted to Dr. P. F. Morse, and Dr. W. L. Brosius, Dept. of Pathology, Harper Hospital for the description of the postmortem findings.
emboli or infarctions and the tracheobronchial tree was free of obstruction.

The heart weighed 1160 Gm. and occupied most of the left thoracic cavity. The pericardial sac contained approximately 400 ml. of clear yellow fluid. The venae cavae and the right atrium were markedly dilated. The tricuspid valve appeared normal. The measurements of the heart structures are listed in Table 1. The right ventricle was greatly enlarged and its walls were markedly hypertrophied. There was no infundibular stenosis. There was complete absence of the pulmonary valve; however, a small fibrous band 2 cm. in length at the pulmonic ring could possibly represent a rudimentary single valve cusp (Fig. 3). There was no evidence of scarring or previous inflammatory lesions at the pulmonic ring area. The pulmonary arterial tree was grossly dilated; the walls were extremely thin and several arteriosclerotic plaques were noted at the main bifurcations.

The left atrium was of normal size. There was no interatrial septal defect and the foramen ovale was not patent. The mitral valve appeared normal. The left ventricle was slightly enlarged and hypertro-

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<th>Table 1.—Measurements of Heart at Autopsy</th>
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<tr>
<td>Weight</td>
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<tr>
<td>Left ventricular wall (thickness)</td>
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<tr>
<td>Right ventricular wall (thickness)</td>
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<tr>
<td>Tricuspid valve (circumference)</td>
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<td>Conus arteriosus (circumference)</td>
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<td>Pulmonic ring (circumference)</td>
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<td>Pulmonary artery (at its greatest circumference, 4 cm. above the pulmonic ring)</td>
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<tr>
<td>Mitral valve (circumference)</td>
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<td>Aortic valve (circumference)</td>
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<td>Interventricular septal defect (diameter)</td>
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Fig. 3. Photograph of the heart illustrating the absence of the pulmonary valve and the dilated pulmonary artery.
The interventricular septal defect as seen from the opened left ventricle.

A single coronary ostium (fig. 5), situated 0.5 cm. above the right posterior aortic cusp gave origin to a large trunk from which immediately arose 3 vessels. The largest followed the distribution of a right coronary artery. The smallest continued anteriorly over the right ventricle. The third, coursing in front of the conus arteriosus to the left ventricle, assumed a pattern grossly similar to the normal left coronary artery with short circumflex and hypoplastic anterior descending branches. No occlusive lesions or pathologic narrowing was seen in the underdeveloped left coronary artery. The intimal surface of the coronary vessels was normal.

The liver weighed 1435 Gm. and was nutmeg in appearance. The spleen weighed 275 Gm. The gastrointestinal and biliary tracts, pancreas, adrenal glands, and kidneys were free of significant pathology, except for generalized passive congestion that involved all the abdominal viscera.
On microscopic examination no specific inflammatory changes were observed in the pulmonary arterial walls and pulmonic ring area (fig. 6). A marked degree of medial hyperplasia and subintimal thickening was present in medium-sized and small arteries (fig. 7). Many terminal arterioles were occluded and microscopic infarctions were noted. Extensive changes characteristic of an acute myocardial infarction were seen on the sections taken from the apical segment of the left ventricle and the lower portion of the septum. The entire thickness of the ventricular wall was involved in some areas.

**DISCUSSION**

It appears that the pulmonary valve anomaly is a congenital malformation. The disappearance of aortic valve cusps has been reported to follow acute endocarditis with resulting ulceration and almost complete destruction of the valve leaflets. Acquired lesions of the pulmonary valve are extremely rare and such a complete destruction of pulmonary valve cusps has not been described. The only case of reduction in the number of pulmonary valves secondary to arteritis, a bicuspid valve in that instance, was reported by Grawitz. In our case, the gross and histologic examinations of the pulmonary valve showed no evidence of scarring or previous inflammatory diseases. The association of other developmental anomalies further strengthens the probability of a congenital origin.

This malformation was necessarily associated with pulmonary incompetence. Few cases of congenital pulmonary insufficiency have been reported in the literature. Abbott described 8 cases; 6 were due to congenital dilatation of the pulmonary artery and the other 2 resulted from supernumerary cusps. Oppenheimer reported 8 cases of idiopathic dilatation of the pulmonary artery and its branches, in 4 of which there was evidence of pulmonary insufficiency. Ford and co-workers recently described a case of bicuspid pulmonary valve with pulmonary insufficiency and, in an excellent review of the literature on bicuspid valves, concluded that 4 other cases may have had pulmonary incompetence. Functional pulmonary insufficiency associated with other congenital heart defects is, on the other hand, quite common.

In our patient, the pulmonary insufficiency suspected before death was considered to be functional. In retrospect, the absence of the
pulmonic second sound in the presence of pulmonary hypertension and the abnormal pressure pattern obtained from the pulmonary artery may have suggested an organic insufficiency. When regurgitation is due to pulmonary hypertension a very loud second heart sound is heard over the pulmonic area. The complete absence of an incisura on the descending limb of the systolic component of the pressure curve (fig. 2), which normally indicates the closure of the valves, correlates well with the absence of the pulmonary valve cusps and with the probable lack of a valvular mechanism during life. The shape of the pressure tracing with its rapid and steep fall to a relatively low diastolic level is quite similar to a ventricular pattern and may suggest a valvular incompetence. Several authors\(^{3,4}\) have described pressure patterns suggestive of pulmonary regurgitation: the most significant criterion appears to be a drop of the arterial diastolic pressure to, or very close to, the level of the ventricular end-diastolic pressure, producing a wide pulse pressure. In our case, the diastolic pressure was relatively low but yet it did not approach the diastolic ventricular pressure. On the other hand, there was considerable widening of the pulse pressure. If the ventricular diastolic pressure of 0 was correct, the persistence of a diastolic pressure in the presence of such a complete anatomic incompetence may be explained by the tachycardia and the marked pulmonary systolic hypertension.

The degree and significance of the pulmonary insufficiency are difficult to evaluate clinically in the presence of the associated interventricular septal defect. If the functional disturbance was comparable to the anatomic malformation, the regurgitation must have been extremely severe and most likely of hemodynamic significance. However, pulmonary incompetence, on theoretic grounds at least, may be relatively well tolerated, since the pulmonary diastolic gradient is normally quite small. This hypothesis appears to be corroborated by the experimental studies of Barger and associates\(^5\) who produced pulmonary insufficiency in dogs but were unable to precipitate congestive failure. Ford and co-workers\(^4\) concluded that a bicuspid pulmonary valve, even if anatomically incompetent, does not induce failure unless additional strain is superimposed on the heart by other cardiac or pulmonary diseases. Similarly, the large ventricular septal defect and pulmonary hypertension in our patient may have grossly exaggerated the pulmonary incompetence, which might have been relatively asymptomatic as an isolated defect.

Although we are unable to clearly define the role played by the pulmonary valve anomaly, we believe that the ventricular septal defect was largely responsible for the altered circulatory dynamics and the resultant clinical picture. The small aorta and aneurysmal dilatation of the pulmonary arterial tree suggest a previous left-to-right shunt at the level of the ventricles. The pulmonary arterioles showed a marked medial hyperplasia (fig. 7) and endothelial thickening, as described in cases of Eisenmenger’s complex and large interventricular septal defect with pulmonary hypertension. With the development of the secondary hypertension, a right-to-left shunt ensued, which was manifested clinically by the late appearance of cyanosis.

The clinical course and catheterization data suggested an Eisenmenger’s complex or a large ventricular septal defect with pulmonary hypertension. The absence of the pulmonary valve and the single coronary artery were unexpected postmortem discoveries.

Death followed an acute myocardial infarction. Such an occurrence is not unusual in the presence of congenital anomalies of the coronary vessels. In a series of 27 adults with single coronary artery, 4 deaths resulted from this complication.\(^8\) Our patient presented a single coronary ostium with hypoplastic left branch arising essentially from a large right coronary artery (fig. 5). A true single coronary artery acceptable to Hyrtl\(^9\) is one that supplies the entire heart and from which arises no significant anomalous branches.\(^8,10\) Others have defined a heart with a single coronary artery as one in which the entire myocardium is supplied by an artery that, regardless of distribution, arises from a single ostium. Associations of a single coronary artery,
taken in its broad sense, and other congenital heart malformations have not been reported in adults. However, Smith claimed that autopsy examination of infants with a single coronary artery frequently reveals other anomalies of the heart and great vessels.

It is quite possible that these congenital malformations resulted primarily from the same embryologic error, a misplacement of the aortic septum at the time of the division of the bulbous arteriosus into pulmonary artery and aorta. Interventricular septal defects may result from an abnormal position or untimely development of the aortic septum, which then fails to unite with the interventricular septum to form the pars membranacea. Such defects are frequently associated with bicuspid pulmonary valves. Simmons explained the development of reductions of pulmonary valve cusps, particularly bicuspid valves, by a similar maldirection of the aortic septum, which then improperly divides the endocardial cushions, the precursors of the cusps. A single coronary ostium has also been attributed by Robert and Loubé to a misdirection of the aortic septum with crowding together of the coronary anlages.

**Summary**

A clinical and pathologic study of a case of congenital absence of the pulmonary valve associated with a large ventricular septal defect and a single coronary artery in an adult is presented. It is of interest that a review of the literature failed to discover other cases of complete absence of pulmonary valve cusps. However, congenital pulmonary incompetence due to other causes has been reported.

The diagnosis of the pulmonary incompetence is briefly discussed. It might have been suggested by the pressure tracings obtained in the pulmonary artery, particularly by the relatively low diastolic pressure and absence of the incisura on the descending limb of the systolic component. It is believed that the pulmonary incompetence was of hemodynamic significance but that the ventricular septal defect was largely responsible for the altered hemodynamics and resultant clinical picture. Death followed an acute myocardial infarction that was most likely related to the congenital anomaly of coronary vessels.

It is suggested that the same embryologic error may have induced the several congenital malformations found in this unusual case.

**Summario in Interlingua**

Es presentate studios clinic e pathologic de un caso de congenite absentia del valvula pulmonary in un adulto, associate con un grande defecto del septo ventricular e un sol arteria coronari. Es interessante notar que un revista del literatura resultava in le discoperta de nulli altere caso de complete absentia del cuspides pulmono-valvular. Tamen, reportos de casos de congenite incompetentia pulmonary per altere causas non es absentia in le litteratura.

Le diagnose del incompetentia pulmonar in le caso hic presentate es discutite brevemente. Il haberea essite possibile susciper le diagnose super le base del registrationes de pression obtenite in le arteria pulmonary, e specialmente super le base del relativamente basse pression diastolico e le absentia del incisura in le portion descendente del componento systolico. Nos opinia que le incompetentia pulmonar eseva de significacion hemodynamic sed que le defecto del septo ventricular eseva in grande mensura responsabile pro le alterate hemodynamica e le resultante tableau clinica. Le morte del patiente occurreva post aun acute infarction myocardial. Isto eseva probablemente relationate al normalitate congenite del vasos coronari.

Es stipulate le these que le mesme error embryologic habeva inducite le varie malformations congenite que eseva trovate in iste caso unusual.

**References**

4. Ford, A. B., Hellerstein, H. K., Wood, C., and


Contradictory reports have appeared concerning the effect of sitosterol upon blood cholesterol and lipids in human subjects. In the present study the serum cholesterol level and lipoprotein pattern were measured to observe the effect of sitosterol on patients on an unrestricted diet and on a low-fat diet using various doses and forms of the agent. It was found that $\beta$-sitosterol produced a reduction in serum cholesterol concentration on unrestricted diets regardless of the initial blood cholesterol level. No untoward side effects were encountered, nor was there a significant weight change. The minimum effective dose was found to be above 10 Gm. daily for a 4-week course of treatment. Two patients maintained on a low-fat diet evidenced a further lowering of serum cholesterol upon the addition of sitosterol. The electrophoretic lipoprotein patterns during the treatment program revealed a decrease in the large $\beta$-lipid zone including the tall beta peak and extending to the cathode side of the point of application. There was no significant alteration in the $\alpha$-lipid fraction or in the protein pattern. The authors state that a reduction of lipid in the $\beta$-globulin fraction and of serum cholesterol may be effected with sitosterol without dietary fat restriction.

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