CONGENITAL HEART DISEASE *

A Persistent Ostium Atrioventriculare Commune with Septal Defects in a Mongolian Idiot

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A defect of the adjoining portions of the interauricular and interventricular septa with a common auriculoventricular orifice is an unusual cardiac anomaly. In 1927 Gunn and Dieckmann [1] reported two such cases, both occurring in Mongolian idiots. They were able to find six cases previously described in the literature and quote Keith as stating that he had seen fourteen and as citing two more, making a total of twenty-two such cases which they found described or referred to in the literature prior to their report. However, Keith [2] did not describe his cases, but simply stated that he had seen fourteen hearts with common auriculoventricular orifices and that this condition was always associated with other grave defects, such as pulmonary stenosis or transposition of the great vessels. Neither of Gunn and Dieckmann's cases had any associated major defect (one had a patent foramen ovale and a patent ductus arteriosus), and of the six cases which they found described in the literature only two showed associated defects, namely a bicuspid pulmonic valve in one and a partial transposition of the aorta in another. Mönckeberg [3] also described a case, not included in those found by Gunn and Dieckmann, which showed no associated defect other than a small patent foramen ovale. From this it is evident that a common auriculoventricular orifice and septal defects, uncomplicated by other grave anomalies, is of rare occurrence.

Report of Case

Clinical History: The patient was a white female who died of scarlet fever at the age of 4 years and 9 months. The three older children of the family were normal. This child was extremely cyanotic at birth, although it was a full-term baby with a normal labor and easy low forceps delivery. At birth a loud systolic

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murmur was noted over the base of the heart. The heart seemed to be enlarged, and a thoracic X-ray taken the day after birth revealed an enlarged thymus. The thymus decreased in size under X-ray therapy and the cyanosis gradually subsided. However, during the early months of life great difficulty in feeding was experienced. Within two months after birth the child was recognized as a Mongolian idiot, the characteristic stigmata of this condition gradually developing.

Starting at the age of 6 months with an attack of bronchitis, the patient suffered from recurring pulmonary infections associated with severe cyanosis. Between these attacks cyanosis was absent. Death finally occurred in the course of a mild attack of scarlet fever, the patient, while seemingly in little danger, suddenly became extremely cyanotic and died within a few hours.

The cardiac condition was repeatedly studied during the life of the patient. The heart was definitely enlarged, especially to the right, so that it seemed more centrally located than usual. Over the base there was a loud blowing systolic murmur that was not transmitted to the great vessels. During the attacks of cyanosis which accompanied respiratory infections, this murmur increased in magnitude. Exertion never was a factor of importance because the child was inactive. At the age of 2 years she learned to stand, while holding to something, but never was able to walk independently.

Description of Heart

The heart, while normal in shape, is greatly hypertrophied, weighing 183 gm. (normal for age about 80 gm.),[4] and shows a slight dilatation of all chambers. The right ventricular wall approximates the left in thickness, the right measuring 13 mm. and the left 14 mm. in width. Both auricular walls are moderately thickened. The interauricular septum is complete above and shows a normal fossa ovalis (Fig. 2). The septum is incomplete below, its crescentic free border arching over a large common auriculoventricular orifice to join its anterior and posterior margins at the base of the auricles, thus forming a free communication between the two auricles (Fig. 2). This opening measures 1.6 cm. in anteroposterior diameter and 0.6 cm. in height. Above, it is bounded by the arching free border of the interauricular septum. Below, it is separated partially from the large defect of the interventricular septum by the incomplete diaphragm formed by the central segments of the common auriculoventricular valve. The free margin of the interventricular septum arches downward from the anterior and posterior margins of the auriculoventricular orifice to form an opening through which the two ventricles directly communicate (Figs. 3 and 4).

There is a single large auriculoventricular orifice which is slightly constricted in the middle (Fig. 1). The right half of this functions

as the right, and the left half as the left auriculoventricular orifice. This large orifice is guarded by a valve composed of five segments. There are two large mesial segments, one anterior and the other posterior, each lying half in the right and half in the left heart. The right halves of these two segments represent the septal segment of the tricuspid valve, while the left halves correspond to the aortic segment of the mitral valve. On the right there are two additional divisions — the normal anterior and posterior tricuspid segments. On the left a normal posterior mitral segment is present. The arrangement of the valve segments is illustrated in Fig. 1. The large posterior central segment is attached closely to the margin of the underlying interventricular septum by a group of partially fused, short, cord-like strands of connective tissue; thus there is very little communication between the ventricles in this location. Under the anterior central segment the defect of the interventricular septum is quite deep, this segment being attached to the margin of the defect by only one large branched chorda tendinea (Fig. 4).

The defect of the interventricular septum is continuous with the auricular defect, the two being separated only by the incomplete diaphragm formed by the large central segments of the auriculoventricular valve. Its anteroposterior diameter is the same as that of the defect of the interauricular septum (1.6 cm.). Its greatest depth is 0.6 cm. and this lies under the anterior central valve segment. The defect extends well forward under the aortic orfice on the left (Fig. 4). The opening into the right ventricle lies under the central valve segments, especially under the anterior one and behind the conus, which is quite thick-walled (Fig.3). The endocardium in this region is thickened and hyalinized. Several thickened chordae tendineae arise from this site, some of which pass into the left heart to be attached to the valve there. An especially large branching chorda arises from the free margin of the defect (Fig. 4). The remaining chordae are normal.

The orifice of the systemic aorta, which lies immediately above the anterior portion of the defect in the interventricular septum, is guarded by three normal cusps. It measures 4.5 cm. in circumference. The pulmonic orifice measures 5 cm. in circumference and also has three normal cusps. The systemic and pulmonic aortae are free from anomalies. The ductus arteriosus is not patent. It is represented by a fibrous cord connecting the two with a dimple-like

depression in the systemic aorta at the site of its attachment. The pulmonary artery and its branches are very large. The systemic aorta is 4 cm. in circumference, while the pulmonary artery measures 6.5 cm. Its branches are also large, the left 3 cm. and the right 4.5 cm. in circumference. The pulmonary artery shows several very small yellowish patches of atherosclerosis. None are present in the systemic aorta. The cavae, coronary sinus, and pulmonary veins are all normal and enter the auricles in the usual manner. The Eustachian valve is well formed.

Other positive findings at autopsy were a terminal bronchopneumonia and an interesting anomalous condition of the ovaries. Two small bodies, each about 6 mm. in diameter, were found attached to the posterior surfaces of the broad ligaments in the position normally occupied by the ovaries. Except for their small size they appeared to be normal ovaries. However, on microscopic examination they were found to be composed of a vascular and fibrous stroma containing a large number of tubular structures lined by high columnar epithelium, evidently vestigial wolffian tubules. A careful search of serial sections through each of these bodies revealed a complete absence of any ovarian follicles or germinal epithelium.

Discussion

The interpretation of this type of cardiac anomaly is extremely interesting, but is complicated by the uncertainty regarding the exact process by which the final complete separation of the two sides of the heart is accomplished normally. Particularly is this true in regard to the relative importance of the parts played by the septa and the endocardial cushions. Mönckeberg [5] believes that the septa play the larger part, the endocardial cushions growing out along the margins of the interauricular and interventricular septa when the septa reach the level of the auricular canal. He states that the presence of the margin of one or the other of these septa at the level of the auricular canal is a prerequisite for the fusion of the endocardial cushions, and that the absence of the margins of both septa at this level necessitates the persistence of the primitive single auriculoventricular orifice. From this it would follow that the primary fault in these cases is a growth deficiency in both the interauricular and the interventricular septa. Thus the common auriculo-

ventricular orifice is not due to any defective development inherent in the endocardial cushions, but results from the absence of a septal margin at the level of the auricular canal. On the other hand, Gunn and Dieckmann, following Mall, believe that the final closure of the ostium primum (primary interauricular foramen) and probably of the interventricular foramen is brought about by the fusion and growth of the endocardial cushions. They conclude, therefore, that the primary fault is a growth deficiency on the part of these structures. For a detailed discussion of this problem the reader is referred to the article by Gunn and Dieckmann.[1]

The author is inclined to believe that their position is the correct one. The careful work of Mall,[6] which is stressed by these authors, is especially convincing. Mall observed in a human embryo of 8 mm. the upward growth of the anterior and posterior endocardial cushions encroaching upon the ostium primum and uniting with the interauricular septum well above the auricular canal. This condition is clearly shown in Mall's illustrations. In an embryo of 9 mm. he found the endocardial cushions fused within the auricular canal, while the interventricular foramen was still open. In the case of this embryo he does not state whether or not the ostium primum was closed. This is strong evidence that the endocardial cushions play a very large part in the closure of the ostium primum and that they do fuse before the interauricular and interventricular septa reach the level of the auricular canal. In support of the opinion that a primary deficiency of growth on the part of the endocardial cushions is responsible, at least for the valvular anomaly and the defect in the lower part of the interauricular septum, is the fact that in cases of persistent ostium primum, uncomplicated by a defect at the base of the interventricular septum, there is commonly an associated anomaly of the valve segments, the aortic leaflet of the mitral valve being cleft from its free border to its insertion (Abbott [7]). This suggests that in such cases the fault may be in the endocardial cushions rather than in the development of the interauricular septum.

Gunn and Dieckmann also discuss in detail the defect of the base of the interventricular septum, concluding that there is little evidence in their cases that the interventricular septum was deficient; therefore the defect was due probably to a failure of downward growth of the endocardial cushions. Their reasoning applies with equal force to our case. In this connection, a case reported by

Abbott [8] seems to fill partly the gap between the cases of persistent ostium primum with deformed auriculoventricular valve segments, normal auriculoventricular orifices and intact interventricular septum, and the type of case we have reported. In Abbott's case the aortic segment of the mitral valve was not only cleft, but was completely divided and the upper part of the interventricular septum "appeared to be slightly defective below." As Gunn and Dieckmann point out, while embryologists are not agreed as to the exact mechanisms of the process, it is safe to assert that the final closure of the interventricular foramen is brought about by the fusion and growth of three structures, namely the bulbar septum, the interventricular septum, and the endocardial cushions. A deficiency in any one of these three could lead, therefore, to a defect in the base of the interventricular septum. We believe Abbott's case, mentioned above, to represent a slight defect of the interventricular wall brought about by a failure of downward growth of the endocardial cushions, and our case to represent a more severe defect originating in a similar manner.

To summarize this discussion, it is suggested that a persistent ostium primum with deformed valve segments is due to a failure on the part of the endocardial cushions to grow up and unite with the interauricular septum, and not to a failure of the downward growth of that septum. In these cases fusion of the endocardial cushions occurs in the auricular canal, but even there it is not complete or normal, as shown by the cleft valve segments. If the endocardial cushions are further arrested in their growth the segments are not only cleft, but completely divided with a smaller (Abbott's cases) or larger (our case) defect of the base of the interventricular septum. The plausibility of this explanation is strongly supported by Mall's observations and it has the practical advantage of explaining the defects observed in our case on the basis of a single primary growth deficiency of one structure (the endocardial cushions) rather than by the coincidental failure of two or more structures. However, it should be admitted in passing that defects of the base of the interventricular septum do occur without the slightest evidence of faulty development of the endocardial cushions. Since, as stated above, the processes of fusion and growth of the bulbar and interventricular septa take part in the closure of the interventricular foramen, these defects, as is generally accepted, are due to a deficiency in the septum

and belong in a different category from the defect associated with a persistent ostium primum and a common auriculoventricular orifice.

The frequency of congenital heart disease in Mongolian idiots is well recognized, Cassel [9] finding it in eight of sixty cases and von Hofe [10] in fourteen of one hundred and fifty cases. Abbott,[8] reporting the case of persistent ostium primum referred to above, emphasizes this fact and states that in her experience the cardiac defect not infrequently is a persistent ostium primum. Of the nine cases of persistent ostium atrioventriculare commune with septal defects, which we have found in the literature, four have ocurred in Mongolian idiots — both of Gunn and Dieckmann's cases,[1] one of the six which they cited from the literature, and the one reported by Mönckeberg.[3] With our cases added, five of the ten reported cases have occurred in Mongolian idiots.

Summary

1. A case is reported in which a persistent ostium atrioventriculare commune is associated with a defect in the base of the interventricular septum and a persistent ostium primum. This occurred in the heart of a Mongolian idiot who showed also complete absence of true ovarian tissue.

2. The cardiac defect is believed to be due to faulty development of the endocardial cushions.

3. Four of the nine similar cases found reported in the literature occurred in Mongolian idiots.

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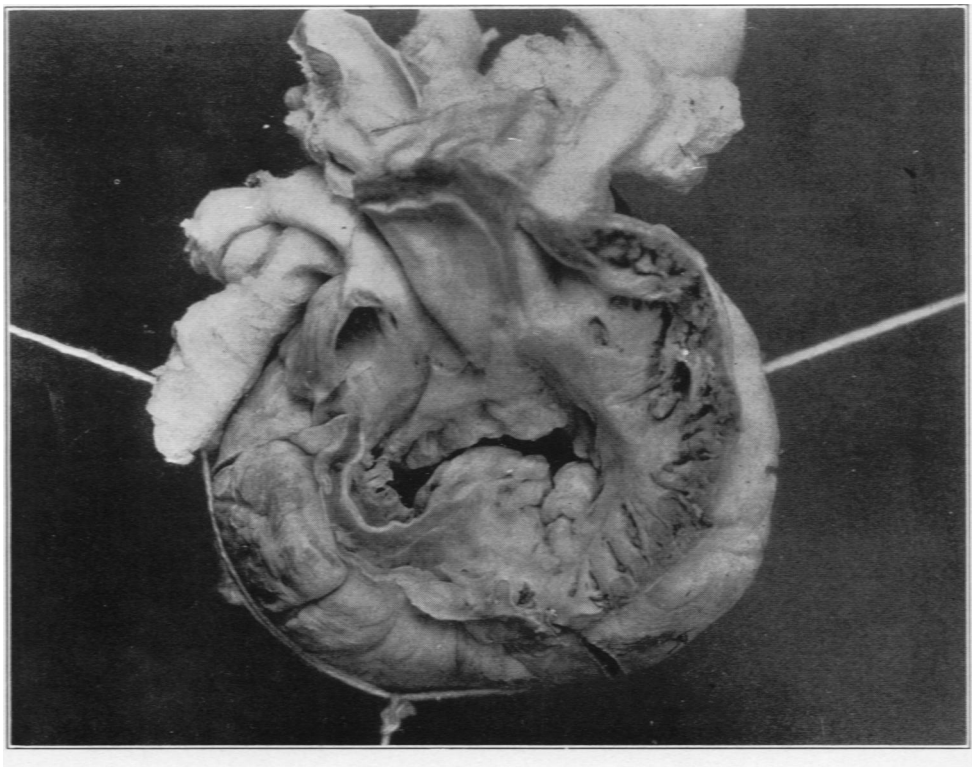
DESCRIPTION OF PLATES

PLATE 44

FIG. 1. The auricles have been partly cut away and the defective interauricular septum reflected exposing the common auriculoventricular orifice with its five valve segments. The two large mesial segments are clearly seen, the anterior above and the posterior below. The right halves of these two segments represent the septal segment of the tricuspid valve, the left halves the aortic segment of the mitral valve. To the right two small segments represent the anterior and posterior tricuspid segments. To the left a single posterior segment is present, the normal posterior mitral segment.

FIG. 2. The right auricle and ventricle are laid open. A large defect of the lower portion of the interauricular septum (persistent ostium primum) is present with the free margin of the septum arching over it. Below this defect the right halves of the mesial segments of the valve guarding the common auriculoventricular orifice are seen (*cf.* Fig. 1). Between these segments there is a deep notch representing the defect in the base of the interventricular septum. The fossa ovale is present in the upper portion of the interauricular septum. The wall of the right ventricle is greatly hypertrophied.



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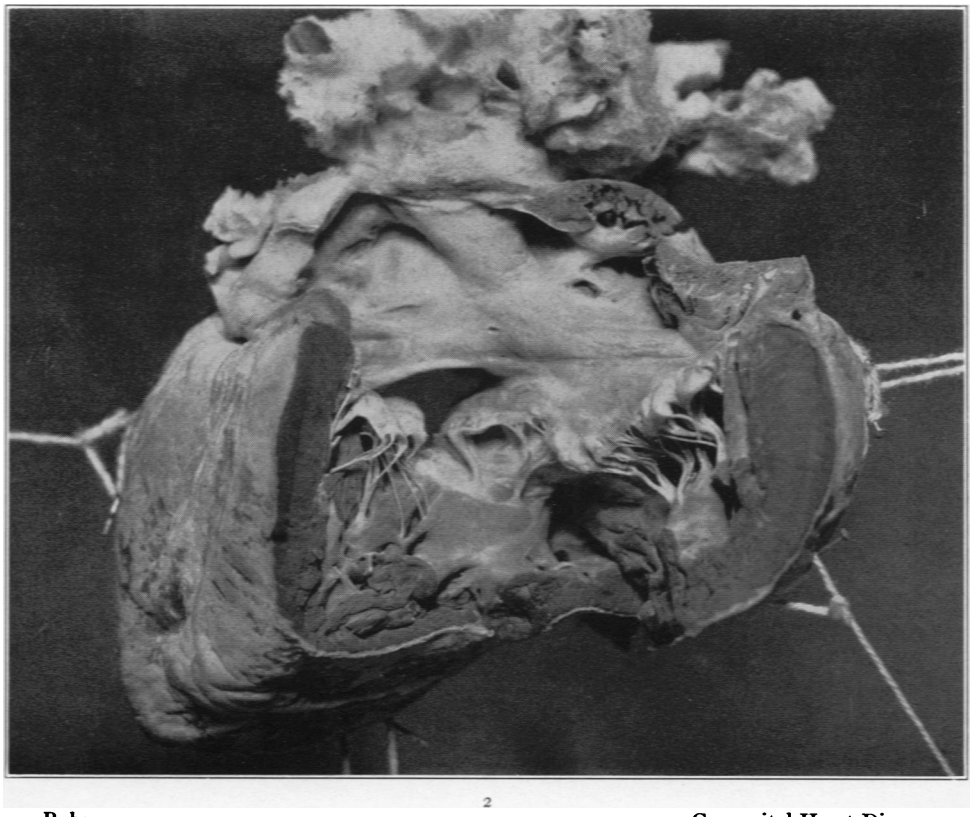

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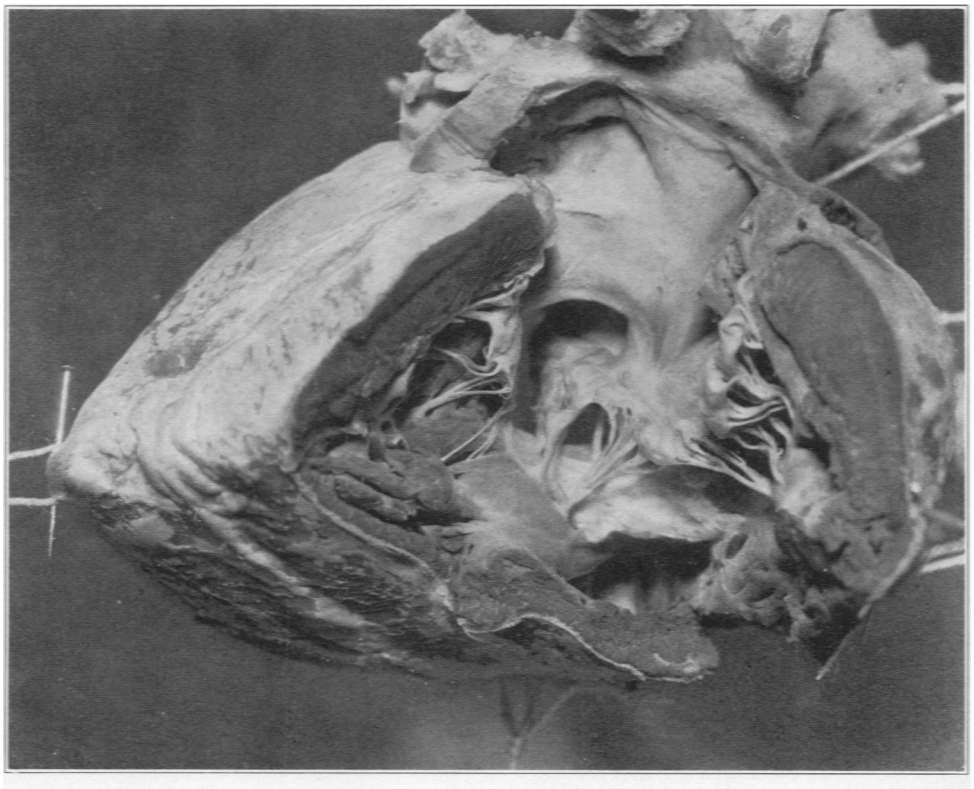
PLATE 45

FIG. 3. The right auricle and ventricle are laid open. The defect of the lower portion of interauricular septum and of the base of the interventricular septum are shown with the anterior mesial segment of the auriculoventricular valve extending through this opening (the left-hand portion in shadow). The endocardium of the interventricular septum near the anterior margin of the defect and of the posterior wall of the conus is much thickened, having a white hyaline appearance. From this region spring a number of chordae tendineae which are attached to the anterior mesial segment and to the anterior tricuspid segment (*cf.* Fig. 1). Some of these chordae are seen extending into the left heart through the defect. Just below the area of thickened endocardium the roomy, thick-walled conus is seen.

FIG. 4. The aorta and left ventricle have been laid open. The aorta is normal. Below its orifice the defect of the base of the interventricular septum is seen. From the free margin of the septal defect arises one large branched chorda tendinea which is attached to the anterior mesial segment (*cf.* Fig. 1).



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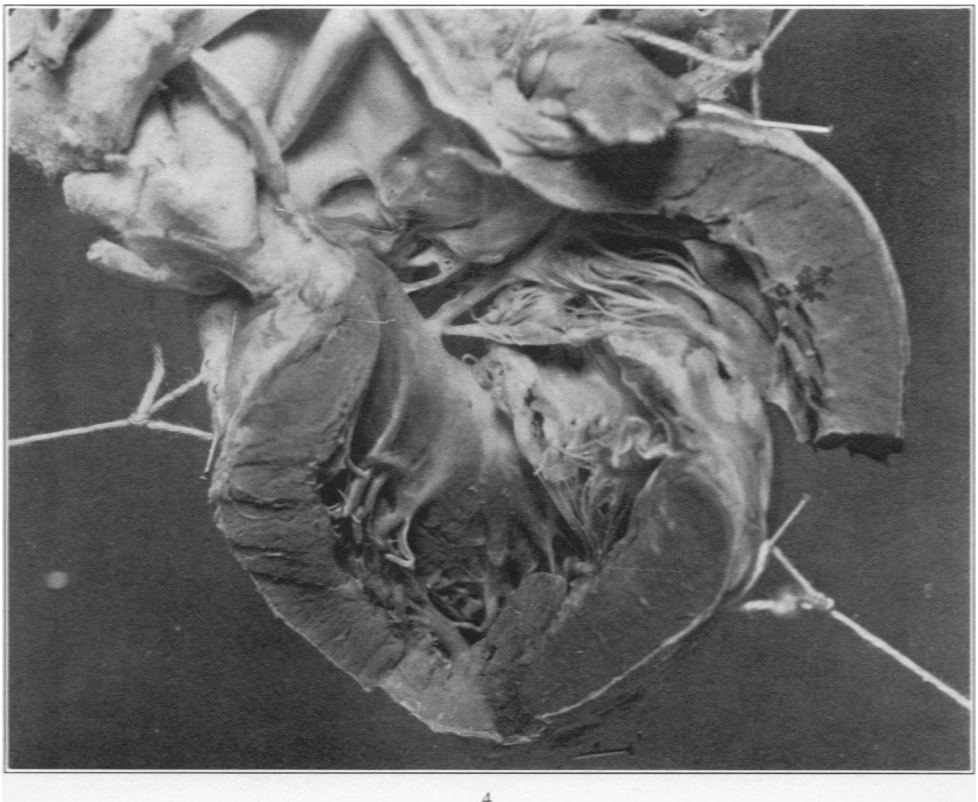

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