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Congenital Mitral Atresia

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Mitral atresia is a rare congenital anomaly, but its true incidence is difficult to assess. In 1962, Fontana and Edwards reported 8 cases of isolated mitral atresia and found 24 others in published reports. They also described 5 cases of combined mitral and aortic atresia and found 23 other published cases. From Britain, MacMahon, McKeown, and Record (1953), in a careful survey of congenital heart disease in children born in a three-year period in Birmingham, did not mention this anomaly, and Coleman (1965) did not encounter a single case in his study of serious congenital heart disease in infants.

Mitral atresia is, however, often associated with other congenital cardiac malformations, and cases are often classified in other groups. Lev (1952) and Noonan and Nadas (1958) considered mitral atresia as part of the hypoplastic left heart syndrome, and Muir (1960), who had seen 5 cases in 19,415 necropsies in Singapore, grouped mitral atresia with cases of cor triloculare because they function similarly. The recent coincidence of finding hearts with mitral atresia in necropsies on two consecutive days prompted a review of the other hearts with this anomaly examined in this hospital.

The poor prognosis of infants with this anomaly is reflected in the relative frequency of mitral atresia at necropsy in the newborn period. An analysis of deaths from congenital heart disease in the newborn period at Johns Hopkins University showed mitral or aortic atresia or both to be the fourth most frequent group of anomalies (Mehrizi, Hirsch, and Taussig, 1964).

Of these infants, 65 per cent died between the third and seventh day of life. In Toronto, 4 per cent of neonatal deaths from congenital heart disease were due to mitral atresia or stenosis (Rowe and Cleary, 1960). More recently, Lambert, Canent, and Hohn (1966) have reported that in 165 neonates in whom congenital heart disease caused death or severe distress, the hypoplastic left heart syndrome was the commonest type of anomaly. Of 37 patients, 28 died in the first week of life; 8 had mitral atresia and 9 had combined mitral and aortic atresia. A few do survive longer, however, and a child of 15 years is included in the cases described by Fontana and Edwards (1962) and in those of Eliot and his colleagues (1965).

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Materials and Methods

Since the opening of the University Hospital of the West Indies in 1952, 9 cases of mitral atresia have been seen in 5000 necropsies. The clinical and necropsy data were reviewed. Three of the infants were examined during life by one of us (C.M) and five of the hearts have been examined at necropsy by one or more of the other authors. One case was previously reported (Watler, 1960). From the accumulated data we have attempted to classify the hearts as suggested by Eliot and his co-authors (1965) (Table I) (Fig. 1).

Anatomical Findings

All nine hearts had some features in common. They were all enlarged though the weight was not stated in every case. All had normal systemic and pulmonary venous drainage. In each case, the right atrium was larger than the left, and there was some form of interatrial communication. The tricuspid valves were normal and the ductus arteriosus persisted in each case.

In 7 hearts a dimple or depression was observed in the floor of the left atrium (Fig. 2 and 3), while in one case valve tissue was present. In all the hearts examined mitral atresia was associated with other cardiac defects. The left ventricle was hypoplastic (Fig. 4) or not identified in all cases except one in which it was not described (Table II) (Fig. 3).

We found it impossible to place two of the hearts in Eliot's classification, and therefore added a third

TABLE I

MODIFIED CLASSIFICATION OF MITRAL ATRESIA (ELIOT *et al.*, 1965)

Group I: Great vessels normally interrelated and hypoplasia of the left-sided cardiac structures
- *Type A.* Aortic valvular atresia with hypoplastic left ventricle
  1. With intact ventricular septum
  2. With ventricular septal defect
- *Type B.* Aortic valvular and left ventricular hypoplasia
  1. With intact ventricular septum
  2. With ventricular septal defect
- *Type C.* Normal-sized aorta and left ventricular hypoplasia

Group II: Great vessels transposed
- *Type A.* Common ventricle
  1. With inverted infundibulum
  2. With non-inverted infundibulum
- *Type B.* Two ventricles present

type in Group I in which the aorta is neither atretic nor hypoplastic but overrides a high interventricular septal defect (Table I). The main anatomical differences are listed in Table II. In 5 cases, the interatrial communication was a foramen ovale, and in 2 the type was not specified. In one heart there was a communication between the left atrium and the dilated coronary sinus entering the right atrium; this type of abnormality appears to be quite rare but has been described previously (Mantini *et al.*, 1966). Herniation of the valve of the foramen ovale into the right atrium, originally described by Dolgopol (1934) in mitral atresia and recently illustrated by Kanjuh, Eliot, and Edwards (1965), was not noted in any of our cases.

Preductal coarctation of the aorta was present in 2 cases and there were 4 with non-cardiac congenital abnormalities. Two of the infants had malrotation of the gut and one of these also had a diverticulum of the small intestine. One case had a unilateral cystic kidney and one had Turner's syndrome together with scoliosis and thoracic cage abnormalities.

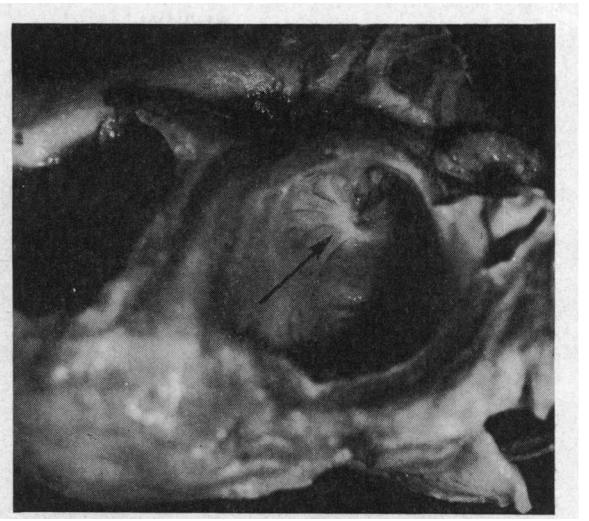

FIG. 2.—Case 4. Atretic mitral valve represented by a dimple in the floor of the left atrium (arrow).

CLINICAL FEATURES

The clinical findings (Table III) were similar in all cases, though the age of onset of symptoms, the presence or absence of cardiac murmurs, and the response to treatment varied slightly depending on the pathological type. Six cases (1, 2, 3, 4, 7, 9) presented with sudden onset of dyspnoea and cyanosis. One (Case 5), who was dyspnoeic and cyanosed at birth, improved after 4 days, but had a recurrence of cyanotic attacks and dyspnoea 6 days later. Case 6 had, in addition to shortness of breath, swelling of the face and extremities. Another (Case 8), who survived longest (8 months), experienced intermittent attacks of dyspnoea and a late onset of cyanosis. Chest *x*-ray films were taken in only 2 cases (5, 6) and showed, in both instances, a large

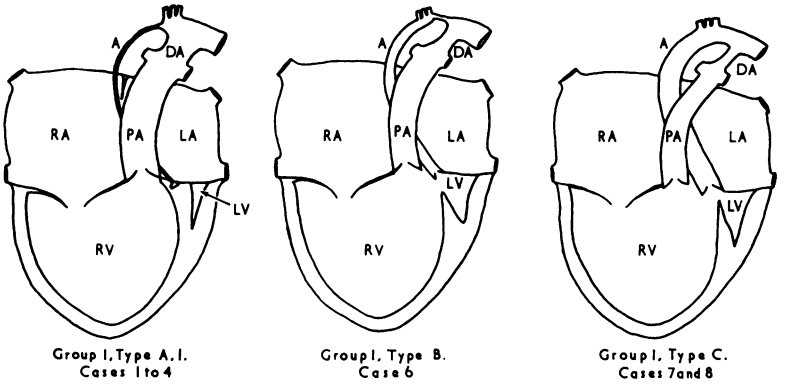

FIG. 1.—Diagrammatic representation of some of the features of Group I cases.

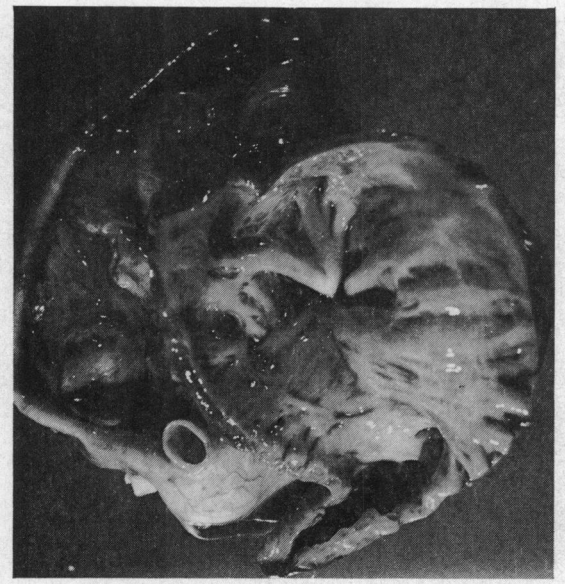

FIG. 3.—Case 6. The floor of the left atrium showing the atretic mitral valve.

globular heart (Fig. 5). Similarly, electrocardiograms were taken in only 2 patients (Cases 4, 6). In Case 4, there was evidence of intermittent heart block and a widened QRS complex. Case 6 had right axis deviation.

DISCUSSION

Patients with mitral atresia and associated aortic valve atresia (Group I, type A) are clinically indistinguishable from patients with isolated aortic atresia. They often appear normal at birth and may remain well until they suddenly become critically ill within the neonatal period. Symptoms of acute heart failure are often precipitated by feeding and the diagnosis of aspiration may consequently be entertained especially as murmurs are usually absent or insignificant. The clinical picture may also be mistaken for septicaemia. However, if congenital heart disease is suspected, careful examination will usually reveal cardiomegaly, signs of left- and right-sided heart failure, and a single accentuated second heart sound. Chest radiographs show a large globular heart with associated pulmonary congestion in most cases, and electrocar diograms

TABLE II

ANATOMICAL FINDINGS

	Type	Case No.	Atrial septal defect	Left ventricle	Aortic coarctation	Non-cardiac anomalies
Group I: no transposition of great vessels	Type A.1 (aortic valve atresia)	1	Coronary sinus	Not identified	—	—
		2	Probably ostium primum	Not identified	—	—
		3	Foramen ovale	Microscopical identification	—	Malrotation of gut; Meckel's diverticulum
		4	Foramen ovale	Not identified	—	Malrotation of gut
	Type A.2 (with VSD)	5	Present but not described	Identified but not described	—	Turner's syndrome; skeletal deformities
	Type B.2 (aortic hypoplasia)	6	Small foramen ovale	Hypoplastic; high VSD	Preductal	Cystic kidney
	Type C (normal-sized aorta)	7	Large foramen ovale	Hypoplastic; high VSD	—	—
		8	Present but not described	Hypoplastic; high VSD	—	—
Group II: transposed great vessels	Type A (with common ventricle)	9	Foramen ovale	Rudimentary interventricular septum only	Preductal	—

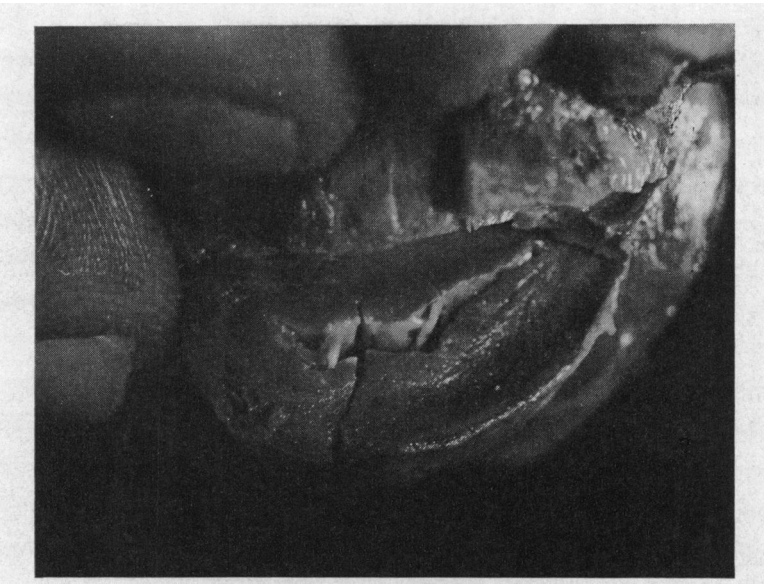

FIG. 4.—Case 6. A view of the slit-like cavity of the left ventricle. Note the hypertophied muscle wall.

TABLE III

SUMMARY OF CLINICAL FINDINGS IN 9 CASES OF MITRAL ATRESIA

	Type	Case No.	Sex	Birth-weight (g.)	Age at onset of symptoms	Age at death	Clinical signs	Clinical diagnosis
Group I	A.1	1	F	2840	21 dy.	21 dy.	Dyspnoeic and cyanosed; no cardiac murmur	Septicaemia
		2	M	3111	9 dy.	9½ dy.	Dyspnoeic and cyanosed; LV failure with gallop rhythm and cardiomegaly; no murmurs	Congenital heart disease
		3	M	3523	45 hr.	52 hr.	Dyspnoeic and cyanosed; pulmonary râles; no murmurs	Aspiration of food
		4	F	3551	39 hr.	51 hr.	Shocked, dyspnoeic, and cyanosed with weak pulses, cardiomegaly, congestive cardiac failure, hepatomegaly; no murmurs	Septicaemia and heart failure
	A.2	5	F	2244	Birth	22 dy.	Short webbed neck; deformed ears; bulging praecordium; loud heart sounds but no murmurs	Turner's syndrome and congenital heart disease; ? single ventricle
	B.2	6	F	2500	7 wk.	9 wk.	Dyspnoeic; cyanosed on crying; congestive cardiac failure with cardiomegaly; gallop rhythm; loud pansystolic murmur maximal at left sternal border; weak pulses	Ventricular septal defect; ? hypoplastic left heart syndrome
	C.	7	M	1886	58 hr.	58 hr. 40 min.	Not examined after onset of symptoms; apparently normal at birth	Aspiration of food
		8	F	?	Soon after birth	8 mth.	Dyspnoeic and cyanosed; clubbing of fingers; cardiomegaly; soft systolic murmur at left sternal border; congestive cardiac failure	Congenital heart disease
Group II		9	M	3295	5 dy.	5½ dy.	Dyspnoeic and cyanosed; weak radial and absent femoral pulses, cardiomegaly; soft systolic murmur at left sternal border	Congenital heart disease

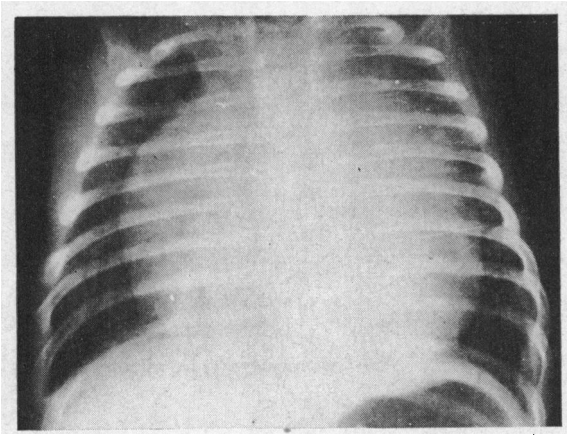

FIG. 5.—Case 4. Chest *x*-ray showing a large globular heart.

demonstrate right ventricular hypertrophy and P pulmonale. The latter finding will differentiate mitral atresia and hypoplasia of the left ventricle from severe isolated aortic stenosis, which in the neonate may have a closely similar clinical picture but in which electrocardiograms usually show left ventricular hypertrophy (Nadas, 1963).

The unusual pattern seen in Case 4 may have been due to severe anoxia, and she died almost immediately after the tracing was taken. Kanjuh *et al.* (1965) have stated that few patients with combined mitral and aortic atresia survive beyond the first week of life and the length of survival depends upon the type of interatrial communication. In their series of 14 patients, those with atrial septal defects (or absence of the interatrial septum) lived longer than those with secondary interatrial communications caused by herniation and prolapse of the valve of the foramen ovale into the right atrium. Our findings do not support this as only one of our four cases had a true atrial septal defect (Case 3) and survived for only 52 hours.

The clinical picture seen in patients without aortic atresia is more variable. In Group I, patients with a type B anomaly (see Table I) have a similar clinical picture to those with type A, except that those with an associated ventricular septal defect have, in addition, a systolic murmur heard maximally along the left sternal edge (Eliot *et al.*, 1965). Our patient (Case 6), as in those described by Noonan and Nadas (1958), presented later than those with aortic atresia and the response to medical therapy, though not dramatic, was at least initially encouraging.

Two of our patients in Group I (Cases 7 and 8) had normal aortic valves and aortas. Relatively prolonged survival then seems possible provided that there are atrial and ventricular septal defects. Evidence that this may be so is the fact that Case 8 survived 8 months without therapy; in addition Watson *et al.* (1960), in a review of 52 cases of mitral atresia with normal aortic valves, found an average survival of 6 months. Pulmonary stenosis was a common associated anomaly in their cases. Our Case 7 was born prematurely and this may have contributed to his early demise, especially as there was a large atrial septal defect and no evidence of pulmonary stenosis. Anomalous pulmonary venous drainage, a common associated abnormality in mitral atresia (Watson *et al.*, 1960; Shone and Edwards, 1964), was not present in any of our cases.

In our series there was only one case with transposition of the great vessels (Group II), and this infant (Case 8) has been previously discussed in detail (Watler, 1960). Clinically, this case was indistinguishable from those in Group I. Eliot *et al.* (1965) noted great variability in the presentation and prognosis of their cases, depending on the presence of coexisting cardiac anomalies; although 6 of them died within 4½ months of birth, the seventh survived until 15 years of age.

Mitral atresia is difficult to diagnose not only during life but also at necropsy, and a knowledge of its anatomical features is necessary if the true nature of the lesion is to be recognized. In hearts with combined mitral and aortic atresia the large pulmonary artery may lead to a diagnosis of truncus arteriosus arising from a single ventricle, and even the discovery of the coronary arteries arising posteriorly from the atretic aorta may not prevent an erroneous diagnosis of high origin of the coronary arteries from the truncus. Such an error was made in one of our cases. Histological examination should differentiate between the elastic aorta and the muscular coronary arteries (Kanjuh *et al.*, 1965). The hypoplastic left ventricle may also be difficult to identify, and blind incisions in the muscle mass on the left side of the heart may only add to the confusion. Because the coronary artery distribution is normal, the site of the left ventricle may be identified between the left anterior descending and posterior coronary arteries. If an incision is made in this area the minute blind cavity should be found, though histological examination of a section from this site may sometimes be necessary to confirm its presence. In only one of our 4 cases was this done; hence the left ventricle was not identified in the other 3, and 2 of them were described as cor triloculare. Keith, Rowe, and Vlad (1958) maintain that the term cor triloculare should be reserved for cases in which both atria enter a common ventricle separately or by a common orifice. In mitral atresia there is no direct communication between the left atrium and the functioning ventricle, which cannot therefore be described as a common ventricle.

Another error that was made in two of our cases was the impression that both the aorta and the pulmonary artery arose from a common ventricle. Re-examination of one of these disclosed a high ventricular septal defect just beneath the aortic valves leading into a minute left ventricle. Description of the other case suggests that it was identical and these two cases are classified as Group I, type C. Friedman, Murphy, and Ash (1955) described two such cases in their series.

Summary

The clinical and pathological features of 9 cases of congenital mitral atresia are described. Six cases were associated with aortic atresia or hypoplasia and one with transposition of the great vessels. The usual clinical presentation was sudden onset of dyspnoea and cyanosis in the neonatal period followed by rapid deterioration, a picture producing diagnostic difficulties that are often increased because of coexisting cardiac anomalies. A correct anatomical diagnosis can be made at necropsy by careful inspection of the heart and great vessels, aided in some cases by microscopical examination.

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