CONGENITAL HEART DISEASE IN THE BANTU: AN AUTOPSY ANALYSIS OF 123 CASES*

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Congenital heart disease is being reported with increasing frequency in the South African literature. Extensive clinical experience has been reported from Cape Town[1] and Durban.[2] Reports of autopsy studies on heart malformations are limited to the small series of Becker (13 cases)[3] and Siew (33 cases).[4] This report analyses 123 autopsy studies of congenital heart disease in Bantu patients at King Edward VIII Hospital. Cases previously reported[2] are not part of this study.

METHODS OF ANALYSIS

The records of the Departments of Paediatrics and Pathology were searched for Bantu patients in whom a cardiac malformation had been found at necropsy during the period 1 January 1961 - 31 December 1967. This was found to be the only reliable method, since many cases with pathologically proved cardiac malformations were not recorded in the medical registry. The available clinical and postmortem records of these cases were analysed. In many instances, clinical records were inadequate and neither X-rays nor electrocardiographic studies were undertaken. The results give an indication of the relative frequency of the major types of malformation; but tney are not a true reflection of the total incidence of congenital heart disease in the Bantu, for the following reasons:

(*a*) The series does not include patients admitted to the wards in whom a clinical diagnosis of congenital heart disease was made, or those who died and in whom autopsy was not permitted.

(*b*) Although 85% of all patients dying during 1961/1962 were submitted to necropsy, this figure fell to 50% in 1966/1967 as a result of increased work load and shortage of staff, the greatest reduction being postmortem examinations on infants and young children.

(*c*) The large numbers of autopsies performed by each pathologist restricted study and only the major cardiac defect may have been recorded.

(*d*) Because of the shortage of beds and the high turnover of patients in the maternity wards, at least 70% of mothers and infants are discharged from hospital within 24

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hours of delivery. Babies therefore receive only a brief clinical examination a short time after birth before discharge from hospital. Moreover, there is no follow-up. Those babies who receive a thorough clinical examination and in whom cardiac malformations might be detected, or who die and are submitted to necropsy, represent a small fraction of the infants born in hospital.

The above reasons help to explain the problem of the detection of heart diseases in the Bantu and enlarge on some of the reasons previously reported.[2] They explain why the rate and incidence of cardiac malformations have not been determined.

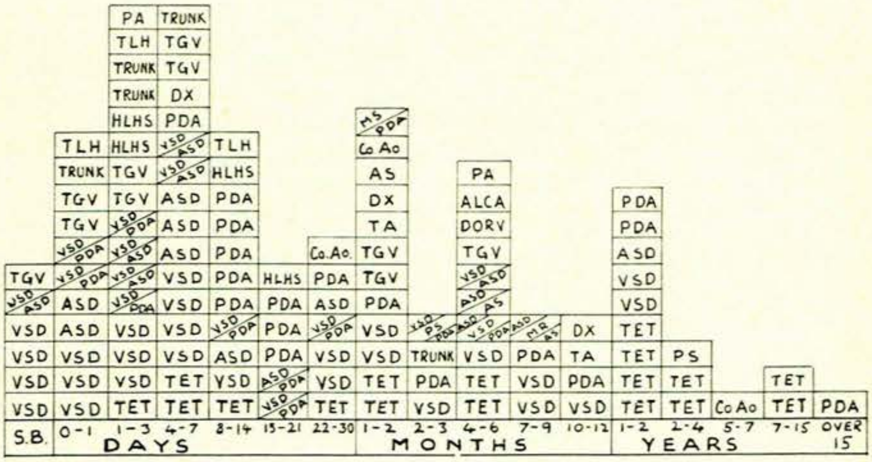

Fig. 1. The age at death of the 123 cases analysed and the type of malformations present. ASD = atrial septal defect; VSD = ventricular septal defect; PDA = patent ductus arteriosus; TET = tetralogy of Fallot; TGV = complete transposition of the great vessels; TRUNK = truncus arteriosus; HLHS = hypoplastic left heart syndrome; CoAo = coarctation of the aorta; TLH = trilocular heart; DX = dextrocardia complex; TA = tricuspid atresia; PA = pulmonary atresia; DORV = double outlet right ventricle; ALCA = anomalous left coronary artery; PS = pulmonary stenosis; AS = aortic stenosis; MS = mitral stenosis; Sub AS = subaortic stenosis; SB = stillborn.

RESULTS

Fig. 1 shows the age at death and the malformations present in the 123 cases analysed. Seventy-three (including 6 stillbirths), or 59·3%, of this series died before 1 month and 81% before 1 year. Four were over 5 years, only one being an adult aged 23 years. The relative frequency of the malformations is shown in Table I, indicating that septal defects, patent ductus arteriosus, tetralogy of Fallot and transposition of the great vessels account for 74% of all anomalies. Isolated patent ductus arteriosus was included only where it appeared pathologically abnormal in infants over 1 week old.

TABLE I. FREQUENCY OF CARDIAC MALFORMATIONS IN THIS SERIES

Type of malformation	*No. of cases*
Ventricular septal defect	26
Patent ductus arteriosus	17
Tetralogy of Fallot	17
Transposition of the great vessels	10
Atrial septal defect	8
Ventricular septal defect/patent ductus arteriosus	7
Atrial septal defect/ventricular septal defect	6
Truncus arteriosus	5
Hypoplastic left heart syndrome	4
Coarctation aortae	3
Trilocular heart (single ventricle or single atrium)	3
Dextrocardia complex	3
Tricuspid atresia complex	2
Pulmonary atresia complex	2
Miscellaneous—one of each of: Double outlet right ventricle; atrial septal defect / ventricular septal defect / patent ductus arteriosus; atrial septal defect/ patent ductus arteriosus; anomalous left coronary artery;* isolated pulmonary stenosis; isolated aortic stenosis; mitral stenosis plus patent ductus arteriosus; aortic stenosis plus atrial septal defect; pulmonary stenosis plus ventricular septal defect plus patent ductus arteriosus; subaortic stenosis, atrial septal defect and mitral regurgitation	10
Total	123

* Reported previously.[5]

Included in the group classified under hypoplastic left heart syndrome are the following cases:

Case 1. A 10-day-old infant had hypoplasia of the left atrium, left ventricle and aortic arch as far as the ductal area, associated with a large patent ductus and large atrial septal defect.

Case 2. A 2-day-old infant had hypoplasia of the left heart and aorta, a large right ventricle, large patent ductus arteriosus and a bicuspid pulmonary valve.

Case 3. A 39-hour-old infant had mitral atresia, single atrium, hypoplastic left ventricle and first part of the aorta, ventricular septal defect and patent ductus arteriosus.

Included in the coarctation aortae group are the following:

Case 4. A 2-month-old infant with a patent ductus arteriosus distal to the coarctation, with also pre-ductal coarctation of the aorta, aortic stenosis and an atrial septal defect.

Case 5. A 5-year-old child had a complete interruption of the aortic arch, a patent ductus supplying the descending aorta and a ventricular septal defect.

Case 6. A coarctation of the aorta was associated with a patent ductus arteriosus and ventricular septal defect. (No further details are available.)

The 2 cases with pulmonary atresia complex had complex malformations:

Case 7. Pulmonary atresia plus tricuspid stenosis was associated with a small, high atrial septal defect plus a patent foramen ovale, and associated patent ductus arteriosus, in a 6-month-old infant.

Case 8. Cor biloculare with a single atrium and single ventricle, pulmonary atresia, total anomalous pulmonary venous drainage into the superior vena cava and arterial supply to the lungs direct from the aorta, associated with asplenia, were found.

The 3 cases of dextrocardia can be further described as follows:

Case 9. A 4-day-old infant had polysplenia, endocardial cushion defect with an atrial and ventricular septal defect plus a patent ductus, with drainage of the left superior vena cava to the left atrium.

Case 10. Complete situs inversus was found, together with

tetralogy of Fallot, with a bicuspid pulmonary valve and a high ventricular septal defect.

Case 11. Dextrocardia without situs inversus, a large ventricular septal defect and severe valvular pulmonary stenosis.

Although mongolism is thought to be rare in the Bantu, 10 patients were considered to show this syndrome, of whom 2 had both atrial and ventricular septal defects and 5 had isolated ventricular septal defects, while the remaining 3 had a patent ductus arteriosus (2 years), atrial septal defect and aortic stenosis (6 months) and tricuspid atresia complex (2 months). The 8 mongols with septal defects are suspected to have had unrecognized endocardial cushion defects.

A further 6 infants with multiple system malformations were suspected of having other chromosomal anomalies. Three of these had isolated atrial septal defects, and 2 had isolated ventricular septal defects—one with combined ventricular and atrial septal defect and one with a ventricular septal defect and patent ductus arteriosus.

Table II illustrates the associated non-cardiac malformations in addition to the above. There were 28 infants with malformations of other systems, some of which were major and were contributory factors leading to death.

TABLE II. NON-CARDIAC ANOMALIES FOUND IN ASSOCIATION WITH CARDIAC MALFORMATIONS

*Cardiac condition**	*Type of abnormality*	*Age at death*
TET	Absence of gallbladder	5 days
TET	Biliary atresia	1 month
TET	Micromelia of left arm. Polycystic kidneys	7 weeks
TET	Accessory digits	7 years
TET	Meningomyelocele	7 days
TET	Fused kidneys both in right abdomen	4 months
ASD	Imperforate anus	7 days
ASD	Lower thoracic meningomyelocele. Spina bifida. Polycystic kidneys	6 hours
ASD	Exomphalos. Fused kidneys	15 min.
ASD	Micrognathia. Cleft palate. Clubbed feet	4 days
ASD+VSD	Rectal and vaginal atresia. Absent urethra. Hydro-ureter and hydronephrosis	Stillborn
ASD+VSD	Hirschsprung's disease (mongol)	4 days
ASD+VSD	Exomphalos. Accessory digits both hands and feet	1 day
ASD+VSD +LSVC	Absent gallbladder	6 months
VSD	Hare-lip and cleft palate. Bilateral absent fibulae. Hydrocephalus	1 day
VSD	Polydactyly. Six fingers and toes both hands and feet	Stillborn
VSD+PDA	Lumbosacral meningocele and spina bifida	9 days
VSD+PDA	Urethral stenosis on left with hydro-ureter and hydronephrosis	3 days prem.
VSD+PDA	Horseshoe kidneys	3 weeks
VSD+PDA +PS	Double ureter. Hare-lip and cleft palate. Hypospadias	3 months
VSD+PDA	Polycystic kidneys. Left renal hypoplasia	3 hours
PDA	Cleft lip. Lumbosacral meningomyelocele and spina bifida	15 days
PDA	Lumbosacral meningocele and spina bifida	10 days
PDA	Hare-lip and cleft palate	12 days
PDA+MS	Bicornate uterus. Webbed neck. Epicanthus	42 days
TRUNK	Hare-lip and cleft palate	4 days
TRUNK	Polydactyly. Cleft palate. Ectopicanus. Fused urethra and vagina	2 days
DX	Malrotation of bowel	4 days

* For explanation of abbreviations see legend to Fig. 1.

Table III shows associated clinicopathological features which appeared to be a contributory cause of death in 36 instances. A few had two such features. The importance of prematurity could not be assessed from available data.

TABLE III. ASSOCIATED CLINICOPATHOLOGICAL FEATURES

Description	*No. of cases*
Surgery on organ malformations other than cardiac	8
Infections: Gastro-enteritis (6), pulmonary abscesses (3), meningitis (3), congenital syphilis (1), neonatal hepatitis (1), tuberculosis (1), empyema and fibrinous pericarditis (1), tetanus neonatorum (1), brain abscess (3), bilharziasis and endocarditis (1)	21
Haemorrhage: Intracranial (6), renal (1), pulmonary (1)	8
Other: Pulmonary syndrome of newborn (4), cavernous sinus thrombosis (1), kwashiorkor, rickets and convulsions (1) and probable rubella syndrome (1)	7
Total	44

Eight infants died following non-cardiac surgery for meningocele (5), exomphalos (1), Hirschsprung's disease (1) and imperforate anus (1), and a further child died from bronchopneumonia following surgery for patent ductus arteriosus. Infection accounted for a number of deaths. Six infants had severe gastro-enteritis, two of whom had concomitant bronchopneumonia and one meningitis. There were 2 other cases of meningitis, one following surgery for meningomyelocele. Three children had brain abscess, in one instance possibly as a result of paradoxical embolism in a 2-year-old child with a large atrial septal defect. The other 2 cases were a 3½-year-old with tetralogy of Fallot and a 5-year-old with complete interruption of the aortic arch. An infant with double outlet right ventricle had miliary pulmonary tuberculosis with terminal cavernous sinus thrombosis. Numerous infants were labelled as having bronchopneumonia at autopsy; but pulmonary infection is frequently found in infants dying in heart failure, and the importance of this is difficult to interpret. Three infants did, however, have pulmonary abscesses.

Intracranial haemorrhage caused death in a group of 6 infants with a ventricular septal defect. Four other infants dying within 24 hours of birth showed pulmonary syndrome of the newborn. Of the 6 stillborn infants, one died from intraperitoneal haemorrhage due to rupture of a subcapsular splenic haematoma, another from subdural haemorrhage following a tentorial tear and a third with intra-alveolar pulmonary haemorrhage. A further stillborn infant was hydropic; and another had rectal and vaginal atresia, and absent urethra with hydro-ureter and hydronephrosis.

Although coexistent malformations and other pathology may have contributed to death in many instances, 53 of the 123 cases were considered to have been in heart failure. At least 14 with cyanotic heart disease died during a hypoxic spell, and general hypoxia precipitated death in others. Many infants were admitted to the wards with respiratory difficulties and pneumonia was diagnosed, although crepitations in the lungs may have been an indication of pulmonary oedema in some.

DISCUSSION

Basic knowledge of cardiac malformations by the pathologist is desirable so that recorded data conform with accepted terminology. Meticulous attention to detail is necessary. Not only should the pathologist consider recognition of heart disease, note the major defect and give attention to anatomical detail, but other associated cardiac malformations should be carefully sought. Both failure to recognize and failure to note defects contribute to error in the true assessment of a case.

The practice of sectioning the heart from the lungs is not satisfactory and possibly accounts for the paucity of cases of anomalous pulmonary venous drainage in this series. Careful observation of the presence and morphology of each valve, chamber and vessel and also microscopic study of the pulmonary vascular bed are essential in all cases. Many of these basic principles, discussed in more detail by various authors,[6-11] were not applied during the period under review because of work load and inexperience of the pathologists, and this has led to difficulties in analysis of this series.

Of course, analysis of a postmortem series does not reflect the wide spectrum of clinical manifestations of congenital heart disease. For example, the finding of a ventricular septal defect at autopsy does not give information as to the haemodynamics during life, such as the presence or absence of pulmonary hypertension, the direction of shunting via the defect or the volume of the shunt.

Early clinical recognition and treatment of heart disease are vital to survival in many instances. Although pulmonary oedema and pneumonia may coexist in the same infant, respiratory distress with or without lung crepitations should always suggest a cardiac disorder, whether a murmur is heard or not. Because of the frequent coexistence of malformations, it is wise to examine carefully the heart in any infant with a congenital malformation of other systems, and an electrocardiogram and X-ray of the chest should be mandatory whether surgery of other organs is contemplated or not.

Conversely, in infants with cardiac malformations, other systems should be examined in detail. Non-cardiac malformations were found in 28 cases, excluding 10 mongols and 6 with suspected chromosomal disorder. This incidence is similar to that found by Wiland[12] and Mehrizi *et al.*[13] Transposition of the great vessels was not associated with any extracardiac malformation, confirming the views of the last-mentioned workers and suggesting that death is a direct result of the cardiac defect. It is noteworthy that none of the cases had tracheo-oesophageal fistulae, which are not uncommonly associated with congenital heart disease.[14,15]

The longer a baby remains in hospital after birth, the less likely it is that a pathological lesion will remain undetected before discharge. However, the difficulties in the clinical diagnosis of congenital heart disease in the early postnatal period, and especially that of recognizing cyanosis with the pigmented skin, make it impossible to assess the exact effect of early discharge on heart disease detection. With approximately 16,000 Bantu (and 4,000 Indian) deliveries at King Edward VIII Hospital *per annum*,[16] it is estimated that 140 babies are born in this institution each year with congenital heart disease and that 84 will die within 1 year of birth. On this basis, therefore, one concludes that many more babies born in this hospital are dying elsewhere with congenital heart disease than are being seen at autopsy.

It is of interest to compare the data in this series with those previously reported.[2] Whereas in the preceding paper there were 15 Bantu infants under 1 year in a series of 117 patients with congenital heart disease, 107 of the present series of 123 cases died under 1 year of age. A combination of data in both reports concerning Bantu with congenital heart disease reflects more accurately the spectrum of heart malformations in this race, supports the view that congenital heart disease is common and indicates that the age incidence of malformations does not differ between Bantu and other racial groups.

SUMMARY

Analysis of the autopsy records of King Edward VIII Hospital between 1961 and 1967 showed that the most common cardiac malformations among 123 cases with congenital heart disease were ventricular septal defect, patent ductus arteriosus, tetralogy of Fallot and transposition of the great vessels. More than half the deaths occurred in the neonatal period, 107 died before 1 year of age and 116 before 3 years of age. Four were older than 5 years and only 1 was an adult. Non-cardiac malformations were present in 28, 10 others were mongols and an additional 6 had suspected chromosomal disorders.

The present report of 123 cases and a previous report of 117 cases[2] reflect more accurately the spectrum of congenital heart disease in the Bantu. It is likely that the frequency and type of cardiac malformations are similar in Bantu and Whites. Difficulties encountered in assessing the incidence of congenital heart disease in the Bantu are indicated.

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