6. CONGENITAL HEART DISEASE (D)

It is notoriously difficult to detect many types of congenital heart disease at birth and a precise anatomical diagnosis is often impossible. In many infants, particularly if premature, the ductus arteriosus and/or the foramen ovale may be patent at birth and it is not possible to predict in which infants either will later close spontaneously. Further, before the circulatory dynamics have stabilized after birth, physical signs of malformations which later cause trouble may be minimal or absent, so that the condition is not recognized. There is no doubt that many individuals lead normal lives although they have small septal defects or valvular anomalies. In essence, malformation of the heart shades into normality, and anatomical anomalies, even if recognized and definable at birth, are uncertain pointers to the subsequent functional disability.

CONGENITAL HEART DISEASE AS THE ONLY RECORDED MALFORMATION (D)

It follows from the above-mentioned diagnostic and other difficulties that the data collected in the present study are not suitable for detailed numerical analysis. The data suggest, however, that a high proportion of cases were severe in their effects and it might be deduced that many less obvious manifestations which could be troublesome later, particularly when the child began to walk, were not identified.

Of 311 cases where congenital heart disease was the only recorded malformation, 159 were stillborn or died in hospital. In 28 cases the returns did not indicate whether or not there had been an autopsy. However, of the 131 cases where this was recorded, in 108 the diagnosis had been so confirmed.

Table 6.1 gives the basic data by centres. It does not appear justifiable to tabulate specific types of malformation recorded from all the centres. In a high proportion of cases not confirmed by autopsy the anatomical diagnosis is unlikely to be correct. However, those interested will find the specific diagnoses in the different centres in the Basic Tabulations by Centre booklet.

For what it is worth the diagnoses given in this study summed for all centres were recorded as:

Diagnosis	*Number of cases*
CHD (NFS)	132
Interventricular septal defect with or without mention of patent ductus	48
Interatrial septal defect or patent foramen ovale with or without mention of patent ductus	29
Patent ductus only mentioned	20
Transposition of great vessels	17
Aortic stenosis	11
Pulmonary stenosis	4
Fallot's tetralogy	9
Dextrocardia	11
Miscellaneous	30
Total	311

CONGENITAL HEART DISEASE OCCURRING IN INFANTS WITH DOWN'S SYNDROME (A), NEURAL TUBE DEFECTS (B1-B7) AND MULTIPLE MALFORMATIONS (N)

As is well known, congenital cardiac malformations are common in children with Down's syndrome. Such a defect was found in 28 of 347 such children in the present study (8.1%). This is likely to be a minimal estimate as only the general term descriptive of the syndrome may have been used on occasions.

Many cases of neural tube defects show multiple malformations and, in all, cardiac malformations were noted in 20 of 1079 B-group cases in this study. However, the condition was found in 13 of 14 B-group cases examined at autopsy and that is probably a better estimate of the true frequency of cardiac malformations in cases with neural tube defects.

In the 329 multiple or N-group cases a cardiac anomaly was recorded in 73 (22%) and 55 of these infants were stillborn or died. It was known whether autopsy was carried out in 48 cases and there was an autopsy in 44 of these.

Looking at the data from a different viewpoint, of 99 children with multiple malformations who were examined at autopsy, a cardiac malformation was found in 44; this is a remarkably high proportion. Over-all, the picture which emerges is that of all the 432 cases where congenital heart disease occurred, 121 (28%) were part of one of the complexes of Down's syndrome, neural tube defects or other multiple groupings of malformations. The over-all frequency of congenital heart disease alone or in any

combination was about 1 per 1000 total births, which corresponds with the data of the British Perinatal Mortality Survey (Butler & Bonham, 1963), where the figure from birth data was 1.2 per 1000 total births.

CONGENITAL HEART DISEASE IN TWINS

In one MM pair both were recorded as having congenital heart disease (NFS) and in seven MM pairs one had congenital heart disease only and the other was normal. In one MM pair one twin had patent ductus, hypospadias and inguinal hernia and the other was normal. In two FF pairs one of the twins had congenital heart disease and the other was normal, and there were two males and one female who were the only affected in MF pairs.

TABLE 6.1
ALL CONGENITAL HEART DISEASE (D) IN SINGLE BIRTHS

CENTRE		Number of cases					
		Sex			Survival		
		M	F	T	LBA	LBD & SB	T
I 1	MELBOURNE	8	4	12	7	5	12
I 2	MELBOURNE	2	4	6	6	0	6
II	SAO PAULO	6	3	9	3	6	9
III	SANTIAGO	-	3	3	2	1	3
IV 1	BOGOTA	3	6	9	6	3	9
IV 2	MEDELLIN	11	6	17	11	6	17
V	CZECHOSLOVAKIA	18	16	34	22	12	34
VI	ALEXANDRIA	-	1	1	1	-	1
VII	HONG KONG	4	5	9	-	9	9
VIII 1	BOMBAY	6	3	9	1	8	9
VIII 2	CALCUTTA	2	-	2	-	2	2
IX 1	KUALA LUMPUR	1	3	4	1	3	4
IX 2	SINGAPORE	-	-	-	-	-	-
X 1	MEXICO CITY	9	9	18	12	6	18
X 2	MEXICO CITY	4	8	12	10	2	12
XI	BELFAST	18	14	32	7	25	32
XII	PANAMA CITY	1	1	2	2	-	2
XIII	MANILA	12	9	21	16	5	21
XIV 1	CAPE TOWN	2	1	3	2	1	3
XIV 2	JOHANNESBURG	17	9	26	13	13	26
XIV 3	PRETORIA	2	-	2	1	1	2
XV	MADRID	35	29	65[a]	23	42	65
XVI 1	LJUBLJANA	2	-	2	4	5	9
XVI 2	ZAGREB	4	2	6	2	4	6
	TOTAL	170	140	311[a]	152	159	311

[a] Includes 1 sex not recorded.