CARDIAC MALFORMATIONS DETECTED AT BIRTH

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Diagnosis of congenital heart disease in the neonatal period is difficult and dependant on the availability of echocardiogaphy and experienced pediatric cardiologist in this type of malformations. Cardiac malformation account for 2-5.5 per 1000 live births reported in large epidemiological studies [1, 2, 3, and 4]. One of these studies included still born and elective terminations [4]. Cardiac malformations detected at birth do not represent all the malformation for different reasons. In our situation detection was based upon clinical examination before discharge and verification of suspected cases by echocardiography. A full time pediatric cardiologist has joined the team in late 92. Another important limitation in detecting all newborns with cardiac malformation was the very early discharge of most babies who were considered clinically healthy within 36 hours of delivery. Some of those who will develop cardiac manifestation after discharge will be followed in other facility in a system where coordination was inexistent.

RESULTS

Transient malformations like PDA and PDA of prematurity were not considered. We have divided cardiac malformations in 2 groups:

I- Isolated cardiac malformations

Newborns included in this group did not have other malformative pathology than cardiac, 67 (1.97 per 1000 total births) patients were born with a major cardiopathy that could be detected and diagnosed before discharge. Their distribution is shown in Table I. We do not consider it useful at this stage of data reporting to enter in the causative details of these malformations.

In fact a lot of important informations that might help in the study of risk factors were not documented on the new born data information sheet as epilepsy, diabetes, other familial cardiopathies and exposure to suspected risk factors as the present trends in cardiac malformations researches [5]. The classification of these cardiopathies was descriptive as given by echocardiography and in some cases cardiac catheterization. We do not use the mechanistic classification that is now used in some large epidemiological studies.

II-Cardiac malformations detected in newborns with a major other malformation.

Another 35 (34% of all cardiac malformations) patients were encountered in this group. Congenital cardiac malformations are frequently associated with non-cardiac malformations and chromosomal anomalies. Management is consequently influenced by the presence of extra- cardiac anomalies. A study from Malta, [6] estimated that during 1990-1994, the birth prevalence of congenital heart disease was 8.8/1000 live births; 231 patients among them 39 (17%) had recognized chromosomal (mainly Trisomy 21) or non-chromosomal syndromes. As in our series, the commonest lesion found in association with Down syndrome was isolated ventricular septal defect, not atrio-ventricular septal defect.

The total number of babies with a cardiac malformation detected at birth in this series was 102. The overall prevalence of Cardiac Malformations in this series was 3/1000. This figure is within the scope of results reported worldwide but from our every day experience we are overwhelmed by infants who have cardiac malformations in need of surgery. Most of them are discovered after the first few weeks of life up to 3-5 years. This fact confirms our impression that cardiac malformation detection was not optimal in this study.

Furthermore we think that this rate is quite high and the population by its high consanguinity and large families and clusters of malformation can constitute a model to study the influence of genetic and non genetic factors in cardiac malformations in general. Further longitudinal studies will confirm or correct these impressions.

Table I
ISOLATED CARDIAC MALFORMATIONS

MALFORMATION	M	F	TX	SB	ND	M/1000D	M/N DEL
CYANOTIC HD	**10**	**7**	**17**	**1**	**11**	**0.50**	**1998**
-Single ventricle	3	1	4	0	1		
-Truncus arteriosus	0	2	2	0	1		
-Tricuspid atresia	1	0	1	0	1		
-Pulmonary atresia	1	0	1	0	1		
-Hypoplastic right heart	0	1	1	0	1		
-Fallot	0	1	1	0	0		
-Ebstein	1	0	1	0	1		
-Double outlet ventricles	1	0	1	0	1		
-Other complex	3	2	5	1	4		
Acyanotic CHD	**15**	**12**	**27**	**0**	**6**	**0.80**	**1258**
-Coarctation	2	1	3	0	0		
-Coarctation hypoplastic arch	1	1	2	0	1		
-Interrupted aortic arch	1	1	2	0	1		
-Pulmonary stenosis	2	0	2	0	0		
-Hypoplastic Left heart	2	3	5	0	4		
-PDA+other anomalies	1	3	4	0	0		
-VSD	2	2	4	0	0		
-VSD+aortic stenosis	1	0	1	0	0		
-others	3	1	4	0	0		
Myocardiopathies	**6**	**7**	**13**	**0**	**3**	**0.38**	**2613**
-dilated	3	2	5	0	3		
-hypertrophic	2	4	6	0	0		
-obstructive	1	1	2	0	0		
Dextrocardia	**6**	**4**	**10**	**0**	**1**	**0.29**	**3400**
-simple	1	2	3	0	0		
-Situs inversus	1	1	2	0	0		
-AVC	1	0	1	0	0		
-Complex cyanotic HD	3	0	3	0	1		
-Klippel-Feil	0	1	1	0	0		
Total cardiac	**37**	**30**	**67**	**1**	**21**	**1.97**	**508**

Table II
CARDIAC MALFORMATION ENCOUNTERED IN OTHER CATEGORIES

MALFORMATION	M	F	TX	SB	ND	M /1000D	M /N DEL
Trisomy 21 -AVC -AVC+duodenal atresia -Tetralogy of Fallot -PDA -VSD	9 1 0 0 0 8	6 1 1 1 1 2	15 2 1 1 1 10	1 1 0 0 0 0	0		
TRISOMY 13 VSD+	3	4	7	0	7		
TRISOMY 18 VSD+	2	5	7	0	7		
TURNER VSD+	0	2	2	2	0		
NOONAN -Pulmonary stenosis +	1	0	1	0	2		
Hydrocephaly -Obstructive myocardiopathy	1	0	1	0	0		
Imperforated anus -VSD	0	1	1	0	0		
Arthrogryposis -VSD	1	0	1	1	0		
Total associated	**17**	**18**	**35**	**4**	**16**		
All cardiac malformations	**54**	**48**	**102**	**5**	**37**	**3**	**333**
TTT	**449**	**363**	**812**	**156**	**306**	**23.90**	**42**

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