CONGENITAL MALFORMATIONS SURVEY

A PRELIMINARY REPORT

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This survey is being undertaken principally to determine the incidence of congenital anomalies, both major and minor, in babies delivered at St. Luke's Hospital. We plan to examine all infants delivered over a period of one year for congenital abnormalities and we are also recording parameters such as head circumference and birth weight for gestational age to establish norms for a Maltese newborn population. The details about the baby and mother are being recorded on specially prepared sheets compiled with the help of Professor A. C. Stevenson of the M.R.C. Population Genetics Research Unit, Oxford, and the final analysis of the data will be done by computer.

The purpose of this paper is to give a preliminary report on the congenital malformations, major and minor, found in the first 1000 consecutive deliveries.

Material

All live and stillborn infants, delivered after 28 weeks of gestation were examined by at least one of us within 24 hours of delivery and the specially prepared sheets *(fig. 1)* filled in. The data about the mother were obtained from the history sheets. Between 7th July and 27th October 1971 there were 1000 pregnancies, producing 1016 infants (including 16 sets of twins).

Findings

There were 531 (53%) males and 483 (48%) females; in 2 cases the sex could not be determined. 17 were stillbirths (post-mortem examination performed on 12) and there were 14 neonatal death (9 had a post-mortem examination). In none of these cases coming to post-mortem was any additional major malformation revealed which had not been diagnosed by previous clinical examination.

The malformations discovered were classified into major and minor ones, according to the W.H.O. classification (Stevenson *et al.*, 1966), with some modifications for the minor group, and are shown

TABLE 1
Major malformations: 1016 babies
(according to W.H.O. classification)

(A)	*Down's Syndrome*	0
(B)	*Neural Tube Defects*	
	Anencephaly only *	2
	Anencephaly with Spina Bifida *	1
	Spina Bifida only	1
(C)	*Other malformations of C.N.S.*	0
(D)	*Heart and Great Vessel Defects*	1
(E)	Gut Defects:	
	Intestina Obstruction ? cause *	1
(F)	*Diaphragm Defects:*	
	Diaphragmatic Hernia *	1
(G)	*Hare-lip and Cleft Palate*	1
(H)	*Talipes*	3
(I)	*Dislocated Hips*	0
(J)	*Polydactyly*	2
(K)	*Other Skeletal Defects:*	
	Oxycephaly	2
(L)	*Urogenital Defects:*	
	Ambiguous Sex *	1
	Separate Scrotal Sacs	1
(M)	*Miscellaneous Defects:*	
	Meconium Ileus *	1
	Sirenomelia	1
(N)	*Multiple Defects* *	1
	Total	20
	Incidence about	2%

* Deaths due to malformation.

Fig. 1

Form for recording data

Mother and Child

Serial No.: | | | | | | 5

Mother's name & maiden name:

Address:

Mother's age: _____ | | 7

Date of birth: : : | | | | | 12

Date of delivery: | | | | | 17

Maternal disease (specify) — diabetes | | | | | 22
— hydramnios
— toxaemia
— other

No. of previous pregnancies: _____ | 24

Known malformation in sib (specify) Y N | 25

Gestation (weeks) _____ | 27

Birth weight (lbs) _____ | 29

1 LBA (1) LBD (2) SB (3) Sex: M (1) F (2) ? (3) | 31

2 LBA (1) LBD (2) SB (3) Sex: M (1) F(2) ? (3) | 33

3 LBA (1) LBD (2) SB (3) Sex: M (1) F(2) ? (3) | 35

Autopsy Y (1) Y (2) Y (3) N (1) N (2) N (3) | | | 38

Cause of death

1 | 40

2 | 41

3 | 43

Malformation

1 Y N (specify) | 44

2 Y N (specify) | 45

3 Y N (specify) | 46

H.C.

Note: LBA = Liveborn, left hospital alive.
LBD = Liveborn, died in hospital.
SB = Stillbirth
H.C. = Head circumference.

in *Tables 1* and *2*. A separate note was made of "mongolian blue spots", the findings on which are shown in *Table 3*. We were particularly interested in the incidence of these spots in relation to the infants' gestational age.

TABLE 2
Minor malformations: 1016 babies

(A)	Hypospadias	6
(B)	Undescended Testicles	8
(C)	Hydrocoele	15
(D)	Malformed pinnae	9
(E)	Minor skin blemishes e.g.: naevi	11
(F)	Asseccory auricles/ preauricular sinuses	3
(G)	Single transverse palmar crease	4
(H)	Accessory nipples	1
(I)	Skin tag (chest)	1
(J)	Heart murmur	12
(K)	"Clicking Hip"	36
	Total	106

Comments

A congenital malformation may be defined as any developmental defect which is present on clinical examination at the time of birth or within the first few months of life. Most major anomalies will be apparent at birth or soon after, but some may not become manifest for several weeks or months, e.g. Hirschsprung's disease, the metabolic disorders and some congenital heart defects. As regards minor anomalies, the difficulty is often not only to determine whether an anomaly is present, but also whether it is a normal variant e.g. malformed ears, receding chin, partial syndactyly. Nonetheless, even minor external defects may be an indicator of a serious internal anomaly, e.g. facial haemangioma in Sturge-Weber syndrome.

The ideal criteria for the determination of the true incidence of congenital malformations include the following:

(1) Rigid terms of definition of at least the major malformations.
(2) Examination by a trained observer.
(3) A prospective rather than a retrospective series.
(4) Inclusion of stillbirths.
(5) Long term follow up.

This preliminary survey satisfies four of the above criteria as does the comparable recent study from Edinburgh reported by Stewart *et al.* (1969) on 2500 consecutive live births. *Table 4* compares our findings with similarly conducted studies from other centres.

In 1970 there were 5384 total births in Malta and Gozo and of these, 2346 (about half) were delivered at St. Luke's Hospital. We therefore consider the number of deliveries at the hospital to be fairly

TABLE 3

"Mongolian Blue Spots"

Number	131 out of 1016 (13%)
Sex	Males 72; Females 59
Site	Always sacral region and buttocks
	Additional sites involved as follows:

1 case: left thigh
1 case: right thigh
1 case: left hand
1 case: left leg
1 case: left foot

— not seen on face, palms, soles and abdomen.

Gestation (weeks):	<35	36	37	38	39	40	>40
No. of infants	0	2	1	20	26	40	42
% age of total:	—	6%	3%	13%	15%	12%	16%

TABLE 4

Authors	*Locality*	*Period*	*No. of Births*	*No. Affected*	*% Affected*	*Remarks*
Stevenson and Warcock	Belfast	1957–	8519	120	1.4	LB + SB Gross anomalies only
Nelson	Edinburgh	—	8648	496	5.74	total
					1.50	major
Villumsen and Zachau-Christiansen	Copenhagen Denmark	1959–61	1707	61	3.51	—
Greco *et al.*	Gargano Italy	1957–61	1435	48	3.35	—
Kolbas	Jugoslavia Croatia	—	1706	119	6.97	all babies at birth
Mestwerdt	Hamburg-Barmbek	1960–61	3202	76	2.37	—
Schubert	Berlin Moabit	1950–57	5314	112	2.10	—
Marden *et al.*	Madison-Wisconsin	1960–62	4412	609	13.8	minor
				91	2.06	major
Tabuchi *et al.*	Hiroshima Japan	1961–62	1344	10	0.74	major
				13	0.97	minor
Khan	Nairobi Kenya	1963–64	3016	54	1.79	major only
Stewart *et al.*	Edinburgh	1965–67	2500	184	7.4	major and minor liveborn only
Data extracted from Kennedy (1967)						
Present series	SLH (Malta)	1971	1016	20	2.0	major
				106	10.0	minor

representative of the newborn population as a whole.

The overall incidence of major malformations was found in this study to be about 2% (20 out of 1016 total births). The incidence of minor anomalies was about 10% (106 out of 1016 total births). This latter figure includes 12 cases of heart murmur and 36 cases of "clicking hip". It is notoriously difficult to evaluate the significance of these findings at 24 hours of age and only long term follow up will determine which cases have in fact true congenital heart disease or dislocated hips respectively. It was therefore decided to include these conditions with the minor rather than the major group.

Finally, it is important to mention that the small number of births in this preliminary report, does not allow us to evaluate the possible aetiological rôle of various factors like maternal diabetes mellitus, toxaema of pregnancy, etc.

Conclusion

This preliminary survey indicates that the incidence of major and minor congenital malformations in babies born at St. Luke's Hospital is in the region of 2% and 10% respectively. This is similar to that reported from other countries. It is concluded that in spite of alleged inbreeding in a relatively small island population, there is probably no greater incidence of major and minor anomalies in the Maltese islands.

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