MALTA CONGENITAL ANOMALIES REGISTER

HALF YEARLY REPORT JANUARY - JUNE 1997

This is the first of a series of biannual reports to be published by the Malta Congenital Anomalies Registry. It is appropriate here to introduce the register:

AIMS

- To collect data about all fetal deaths and infants with a diagnosis of congenital anomalies on the islands of Malta and Gozo
- To keep a register of all cases of congenital anomalies diagnosed until one year of age
- To provide data which may be required for epidemiological studies
- To issue regular reports and provide physicians and the general public with information they may need, always respecting strict confidentiality
- To detect any changes in occurrence of specific congenital anomalies

BACKGROUND

Data on congenital anomalies diagnosed at birth at St. Luke's Hospital (Malta) has been collected since 1983. In 1986, through the University of Malta, the register became a member of EUROCAT (European Registration of Congenital Anomalies and Twins). Funding by the University of Malta ran into financial difficulties in 1995. The Department of Health Information in collaboration with Prof. A. Cuschieri, geneticist, is now co-ordinating all previous efforts of recording congenital anomalies and starting a Malta Congenital Anomalies Register as from January 1997. This register covers births from all hospitals on the Maltese Islands and includes cases diagnosed until one year of age.

COVERAGE

The register is population based and covers all births on the islands of Malta and Gozo which amounted to 4,944 births in 1996.

The small size and population of the islands (Area: 316km^2 and Population: 373,958 in 1996), the well defined boundaries and absence of significant ethnic minority groups make the islands ideal for epidemiological studies.

DATA COLLECTION AND SOURCES OF INFORMATION

Congenital Anomalies for the purposes of the register, are defined as *'structural, functional, metabolic, behavioural and hereditary defects present at birth'.*

Data collection occurs on both a passive and active basis. On a passive basis, paediatricians and private hospitals are asked to report any newly diagnosed cases of congenital anomalies. On an active basis, members of the staff of the Department of Health Information visit St. Luke's Hospital maternity, paediatric and laboratory units to collect data directly and review patient notes.

The hospitals presently involved in data capture are St. Luke's Hospital (Malta), Gozo General Hospital (Gozo), St. Philip's Hospital, Capua Palace Hospital and St. James Hospital. Present sources of data from St. Luke's Hospital are: doctors' notifications, records at labour ward, post-natal/nursery ward, SCBU, Echocardiography lab., Genetics clinic, SLH Maternity Information Systems database, SLH Hospital Activity Analysis database, National Death Register, pathology autopsy reports, Congenital Heart Defects Register, hypothyroid and thalassemia screening programmes. It is hoped that these sources will increase in the future.

INTERNATIONAL RELATIONSHIPS

The Malta Congenital Anomalies Register is a member of EUROCAT through Prof. A. Cuschieri.

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OMPHALOCELE AND GASTROSCHISIS

During the first half of this year there was one fetal death due to omphalocele (exomphalos). This was diagnosed prenatally by ultrasound.

Omphalocele and gastroschisis are both anomalies of the abdominal wall and are distinct from umbilical hernia. Omphalocele and gastroschisis are separate conditions having different epidemiology. Both can be diagnosed by prenatal ultrasound.

Omphalocele is a malformation of the umbilical cord with various portions of the intestinal tract extruded through the umbilical ring with a covering sac or its remnants. Gastroschisis is generally a small defect in the abdominal wall through which viscera are exposed, usually to the right and lateral to the normally closed umbilical ring.

The aetiology of both omphalocele and gastroschisis is largely unknown. Familial recurrence has been reported for both. Although a male excess of omphalocele is reported, there are no statistically significant gender differences for either condition.[1,2]

In a survey of three million births carried out by EUROCAT[1] overall prevalence rates were calculated as 2.52 per 10,000 births for omphalocele (including induced abortions) and 0.94 per 10,000 births for gastroschisis. In the case of omphalocele, persistently high rates were found in United Kingdom and Ireland. Prevalence rates of omphalocele are decreasing while those of gastroschisis are increasing in England and Wales.[2]

For omphalocele, 17.5% of cases were fetal deaths of gestational age of at least 20 weeks, while 14.6% of gastroschisis were recorded as fetal deaths.[1] Of those that survive, surgical correction of both conditions leads to favourable survival rates in the abscence of lethal or co-existing major congenital anomalies. Quality of life in survivors is also described as ' good' in 80% of patients.[3]

Both conditions can be found as isolated defects, as part of a recognised syndrome or sequence or in combination with other defects, namely neural tube and limb defects.

Table 1- Percentage of isolated defects, recognised conditions and multiple anomalies

	Single	Recognised condition	Multiple anomalies
Omphalocele	23.9%	21.9%	54.2%
Gastroschisis	71.9%	7.3%	20.8%

Taken from a EUROCAT Survey [1]

In the EUROCAT Survey[1], analysis of maternal age distribution showed that (excluding chromosomal cases) there was an increased risk for both omphalocele and gastroschisis for young mothers. This was more marked for gastroschisis (Table 2). Young maternal age therefore appears to be a strong risk factor.[1,2]

Table 2 - Effect of maternal age on risk of omphalocele and gastroschisis

Maternal age	Odds Ratio - Risk estimate	
	Omphalocele	Gastroschisis
< 20	2.08	11.24
20-24	0.83	1.90
25-29	1.00	1.00
30-34	0.83	0.63
> 34	1.24	0.85

Taken from a EUROCAT Survey [1]

In Malta and Gozo during the years 1993-1995, there were 3 cases of omphalocele (two of which were prenatally diagnosed by ultrasound) and 2 cases of gastroschisis reported (one of which was prenatally diagnosed) in a total of 14,586 births.

Of the three cases of omphalocele, one case was a single defect and the baby suffered fetal death, another case was part of a syndrome (trisomy 21); this child survived its first year of life. A third case was part of a multiple malformation picture with no identifiable syndrome, and this baby was also a fetal death. Both cases of gastroschisis were isolated anomalies; one died within 24 hours while the other was successfully operated for and survived the first year of life.

In Malta and Gozo (1993-1995), the average maternal age of mothers having children with omphalocele was 27.0 years while that of mothers having children with gastroschisis was 20.5 years (average maternal age for all deliveries during this period was 29.1 years[4]). These observations are inkeeping with young maternal age being a risk factor for abdominal wall defects.

References:

1 Calzolari E. et al. *Omphalocele and Gastroschisis in Europe: A survey of 3 Million Births 1980-1990.* Am. J. Medical Genetics, 1995; 58: 187-194

2 Tan KH et al. *Congenital anterior abdominal wall defects in England and Wales 1987-93: retrospective analysis of OPCS data.* BMJ, 1996; 313: 903-906

3 Tunell WP et al. *Abdominal Wall Defects in Infant Survival and Implications for Adult Life.* Ann. Surgery, 1995; 221: 525-530

4 Demographic reviews 1993-1995. Published by The Central Office of Statistics, Malta (Annual publications).

JANUARY - JUNE 1997

This report includes all reported and confirmed congenital anomalies diagnosed during the period January to June 1997 in Malta and Gozo. Anomalies included in this report are those classified under ICD-9 chapter XVII entitled "Congenital Anomalies".

Total number of infants/fetuses diagnosed with congenital anomaly

Hospital		Total births		With Congenital anomaly	
		Livebirths	Fetal Deaths	Livebirths	Fetal Deaths
St. Luke' s Hosp.	M	1034	5	29	0
	F	879	7	27	0
Gozo Gen. Hosp.	M	96	2	2	0
	F	85	0	2	0
Private Hosp.**	M	127	0	1	0
	F	87	0	0	0
Total		2308	15*	61	1*

*Includes one baby of indeterminate sex delivered at St. Luke' s Hospital

** Private Hospitals include: Capua Palace Hospital, St. James' Hospital and St. Philip' s Hospital.

Distribution of infants/fetuses according to number of major anomalies

Anomalies	Number of Infants/fetuses
Single major anomaly	44
Two or more major anomalies of same system	7
Major anomalies involving two systems	4
Major anomalies involving three or more systems	1
Chromosomal anomalies	4
Multiple anomalies and other syndromes not elsewhere classified	2
Total	**62**

Distribution of infants/fetuses with congenital anomalies according to system/s involved

ICD code	System	Number of infants/fetuses			Fetal deaths
		Total	Males	Females	
7400-20	Neural tube defects	1	0	1	-
7421-29	Other nervous system defects	0	0	0	-
7430-49	Eyes, ears, face and neck	1	1	0	-
7450-79	Cardiovascular	17	11	6	-
7480-89	Respiratory	0	0	0	-
7490-93	Cleft palate and lip	3	2	1	-
7500-19	Upper alimentary tract	3	2	1	-
7520-29	Genital organs	7	6	1	-
7530-39	Urinary	2	0	2	-
7540-59	Musculoskeletal limbs & skull	9	5	4	-
7560-69	Other musculoskeletal defects	4	3	1	-
7570-79	Skin	4	1	3	-
7580-89	Chromosomal anomalies	4	1	3	-
7590-92	Endocrine glands	0	0	0	-
7593-96	Other anomalies	0	0	0	-
7597-99	Multiple anomalies/ Syndromes	2	0	2	-
	Two systems	4*	0	3	1
	Three or more systems	1	0	1	-
	Total Infants with anomalies	**62***	**32**	**29**	**1**

*Includes one baby of indeterminate sex

All anomalies reported during the period January - June '97

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	Anomalies grouped according to system involved	Total	Male	Female
7400-7420	**Neural Tube defects**	**1**	**0**	**1**
7420	Encephalocele	1	0	1
7421-7429	**Other anomalies of nervous system**	**0**	**0**	**0**
7430-7449	**Anomalies of the eye, ear, face and neck**	**1**	**1**	**0**
7442	Abnormally shaped ears	1	1	0
7450-7479	**Anomalies of cardiovascular system**	**29**	**14**	**15**
7451	Discordant ventriculoarterial connection	1	1	0
7452	Tetralogy of Fallot	1	1	0
7454	Ventricular septal defect	8	4	4
7455	Atrial septal defect	12	4	8
7460	Anomalies of pulmonary valve	3	1	2
7462	Ebstein' s anomaly	1	1	0
7463	Aortic valve atresia / stenosis	1	1	0
7471	Coarctation of the aorta	1	0	1
7473	Anomalies of pulmonary artery	1	1	0
7480-7489	**Anomalies of respiratory system**	**2**	**0**	**2**
7485	Agenesis/ hypoplasia/ dysplasia of lung	2	0	2
7490-7493	**Cleft palate and cleft lip**	**4**	**2**	**2**
7490	Cleft of secondary palate	3	1	2
7491	Cleft of primary palate	1	1	0
7500-7509	**Anomalies of upper alimentary tract**	**1**	**1**	**0**
7503	Tracheo-oesophageal fistula, oesophageal atresia	1	1	0
7510-7519	**Other anomalies of the digestive system**	**2**	**1**	**1**
7511	Atresia and stenosis of small intestine	1	0	1
7513	Hirschprung' s disease	1	1	0
7520-7529	**Anomalies of the genital organs system**	**8***	**6**	**1**
7524	Anomalies of cervix, vagina & external genitalia	1	0	1
7526	Anomalies of penis	6	6	0
7527	Indeterminate sex	1*	0	0
7530-7539	**Anomalies of the urinary system**	**4**	**0**	**4**
7530	Renal agenesis and dysgenesis	2	0	2
7532	Obstructive defects of renal pelvis/ureter	2	0	2
7540-7559	**Deformities of the musculoskeletal system**	**11**	**6**	**5**
7545	Varus deformities of feet	2	1	1
7547	Other deformities of feet	2	1	1
7550	Polydactyly/ polysyndactyly, hand & foot	3	2	1
7551	Syndactyly hand and foot	3	1	2
7552	Reduction deformities of upper limb	1	1	0
7560-7569	**Other musculoskeletal anomalies**	**6***	**3**	**2**
7565	Osteodystrophies	1	0	1
7566	Congenital diaphragmatic hernia	4	3	1
7567	Anomalies of abdominal wall	1*	0	0
7571-7579	**Congenital anomalies of the integuement**	**5**	**1**	**4**
7573	Other specified anomalies of skin	5	1	4
7580-7589	**Chromosomal anomalies**	**5**	**1**	**4**
7580	Down syndrome	4	1	3
7582	Edward' s syndrome	1	0	1
7593-7599	**Other congenital anomalies**	**1**	**0**	**1**
7598	Other syndromes or associations	1	0	1

* Includes one baby with indeterminate sex

NB - Spina bifida occulta is only registered when there are complications or when it occurs in combination with other major anomalies

- Hydrocephaly occuring with spina bifida not included
- Glandular hypospadias is not recorded, unless in combination with other congenital anomalies