MALTA CONGENITAL ANOMALIES REGISTER

http://www.health.gov.mt/ministry/dhi/mcar.htm

HALF YEARLY REPORT JANUARY – JUNE 2000

CONGENITAL ANOMALIES REPORT 1993-97

The Malta Congenital Anomalies Registry has recently published and distributed the first comprehensive Congenital Anomalies Report 1993-97 covering data for these five years. The full report will shortly be included in the Registry's website.

Due to the relatively small number of cases of congenital anomalies registered annually, interpretation of data for one particular year may be unreliable. It is therefore important to analyse aggregated data for several years for more meaningful interpretation. This minimises errors due to the fluctuation in small number statistics.

EXECUTIVE SUMMARY

The following is a summary of the results of analysis of these five years' data.

Prevalence

In Malta between 1993-1997 there were 902 babies (482 males, 418 females and 2 of indeterminate sex) registered with major congenital anomalies out of a total of 24,510 births. The overall prevalence of babies registered as having one or more major congenital anomalies from 1993-1997 was therefore 36.8 /1,000 total births.

Fetal deaths

From 1993-97, 22 out of the 902 babies registered with anomalies were fetal deaths (2.4%), whereas only 145 out of the 24,510 total births on the islands were fetal deaths (0.6%). The greater proportion of fetal deaths in babies with anomalies when compared to all babies is statistically significant.

Gender distribution

Persistently more male babies are registered as having one or more congenital anomalies. From 1993-97, 53% of babies registered with congenital anomalies were male and 46% were female.

In spite of there being more male babies registered as having an anomaly, the difference in proportions is not statistically significant.

Gender distribution of babies registered as having an anomaly 1993-97

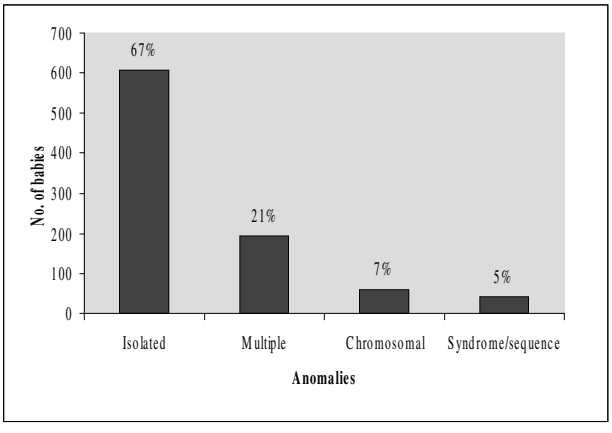
	Total births	Births registered with anomaly	Proportion of births with anomaly /1000
Male	12,731	482	37.8
Female	11,777	418	35.4
UN*	2	2	-
Total	**24,510**	**902**	**36.8**

*UN - Indeterminate sex

Isolated vs. Multiple anomalies

The majority of babies registered with congenital anomalies have isolated defects. Analysing data from 1993-97, out of 902 babies registered with congenital anomalies, 607 (67%) had isolated defects, 193 (21%) had multiple unrelated anomalies, 60 (7%) had chromosomal anomalies and 42 (5%) had multiple anomalies recognised as a syndrome or sequence.

Distribution of babies according to number of major congenital anomalies (1993-97)

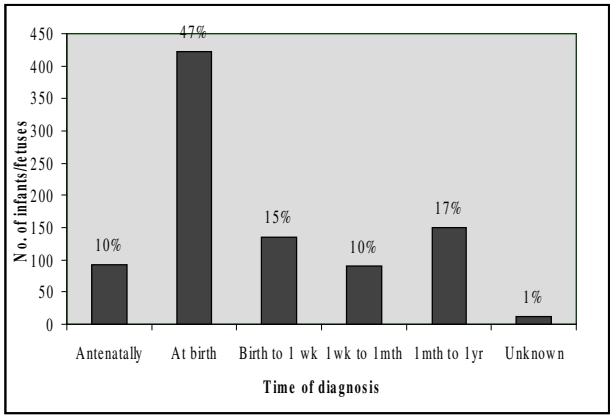


Time of diagnosis

Most babies with major congenital anomalies are diagnosed at or soon after birth. The figure below shows the distribution of babies with congenital anomalies according to the time of diagnosis. Between 1993-97, 82% of babies were diagnosed in

the perinatal period. A not insignificant proportion (17%) were, however, diagnosed after 1 month of age and before 1 year of age. This emphasises the importance of the registry to follow up babies throughout the first year of life for purposes of registration of congenital anomalies.

Time of diagnosis of babies with major anomalies

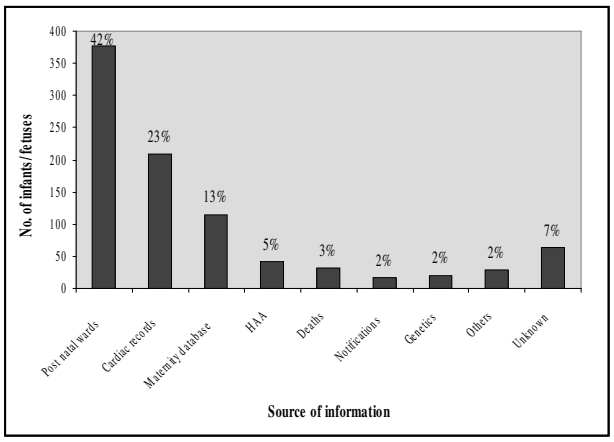


Sources of information

Multiple sources of information are employed to identify babies to be registered in the Malta Congenital Anomalies Register.

Between 1993-97, the main source of information was active data collection from maternity wards in St. Luke's Hospital (42%), followed by pick up from Cardiac records including Cardiac Lab and the Congenital Heart Diseases Register (23%). The St. Luke's Hospital Maternity Systems Database and Hospital Activity Analysis Database (HAA) were the sources of information for 13% and 5% of cases respectively. The National Mortality Register and autopsy records accounted for 3% of cases; both doctor's notifications and Genetics Clinic records accounted for 2% of cases each and 3% were from miscellaneous other sources. For the remaining 7% of babies registered, the primary source of information was unrecorded.

Main sources of information for babies diagnosed with congenital anomaly (1993-97)



Maternal ages

Analysis of 1993-97 data shows that the average maternal ages of babies with non-chromosomal anomalies were not significantly different from those of all births. However, average maternal ages of babies with chromosomal defects were significantly higher than the average for all deliveries.

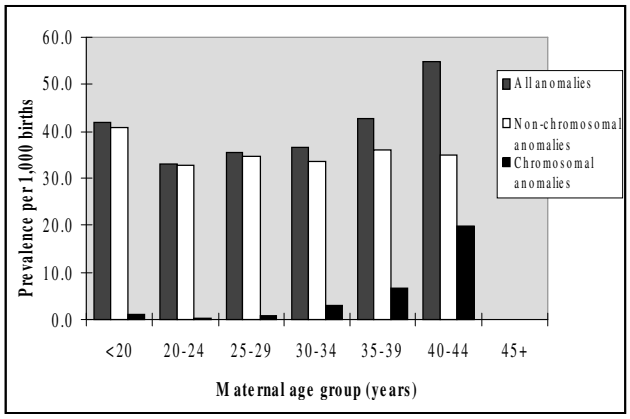
Average maternal ages for all deliveries and deliveries of babies with anomalies (1993-97)

	Avg maternal age (yrs)
Deliveries of babies with non-chromosomal congenital anomalies	29
Deliveries of babies with chromosomal anomalies	35*
All deliveries	**29**

*this is significantly higher than that for all deliveries

Prevalence of congenital anomalies increases significantly in the older maternal age groups. This is due mainly to the increase in chromosomal anomalies occurring in these age groups. In fact when considering non-chromosomal anomalies only, it is seen that there is an increased prevalence in the youngest maternal age group ie <20years.

Differences in variation of maternal age groups for all anomalies, non-chromosomal and chromosomal anomalies (1993-97)

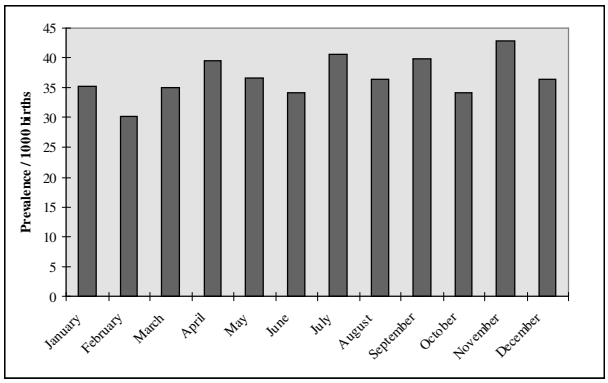


Seasonal distribution

The figure below shows the seasonal variation of prevalence of babies registered with anomalies between 1993-97.

The pattern shows no definite seasonal variation in prevalence of babies with congenital anomalies when considering all anomalies together. The pattern may change if babies with one particular anomaly only are considered.

Monthly distribution of babies registered with congenital anomalies (1993-97)



Geographic distribution

Malta and Gozo may be divided into eleven regions. The distribution of the prevalence of all babies registered with congenital anomalies between 1993-97 is shown below.

The highest prevalence of livebirths with congenital anomalies was found in the Central East region of Malta (44.9/1,000 births), and the lowest in the Central West region (30.3/1,000 births). There are no statistically significant differences between the various regions.

Geographical distribution of all babies registered with congenital anomalies in Malta and Gozo 1993-97

Prevalence per 1,000 births		
Central East	(CE)	**44.9**
South	(S)	42.8
Central	(C)	42.1
West	(W)	38.7
North	(N)	38.3
Gozo	(G)	37.9
Central North	(CN)	34.5
Harbour	(GH)	34.5
Central South	(CS)	33.9
East	(E)	32.5
Central West	(CW)	**30.3**

Distribution of anomalies

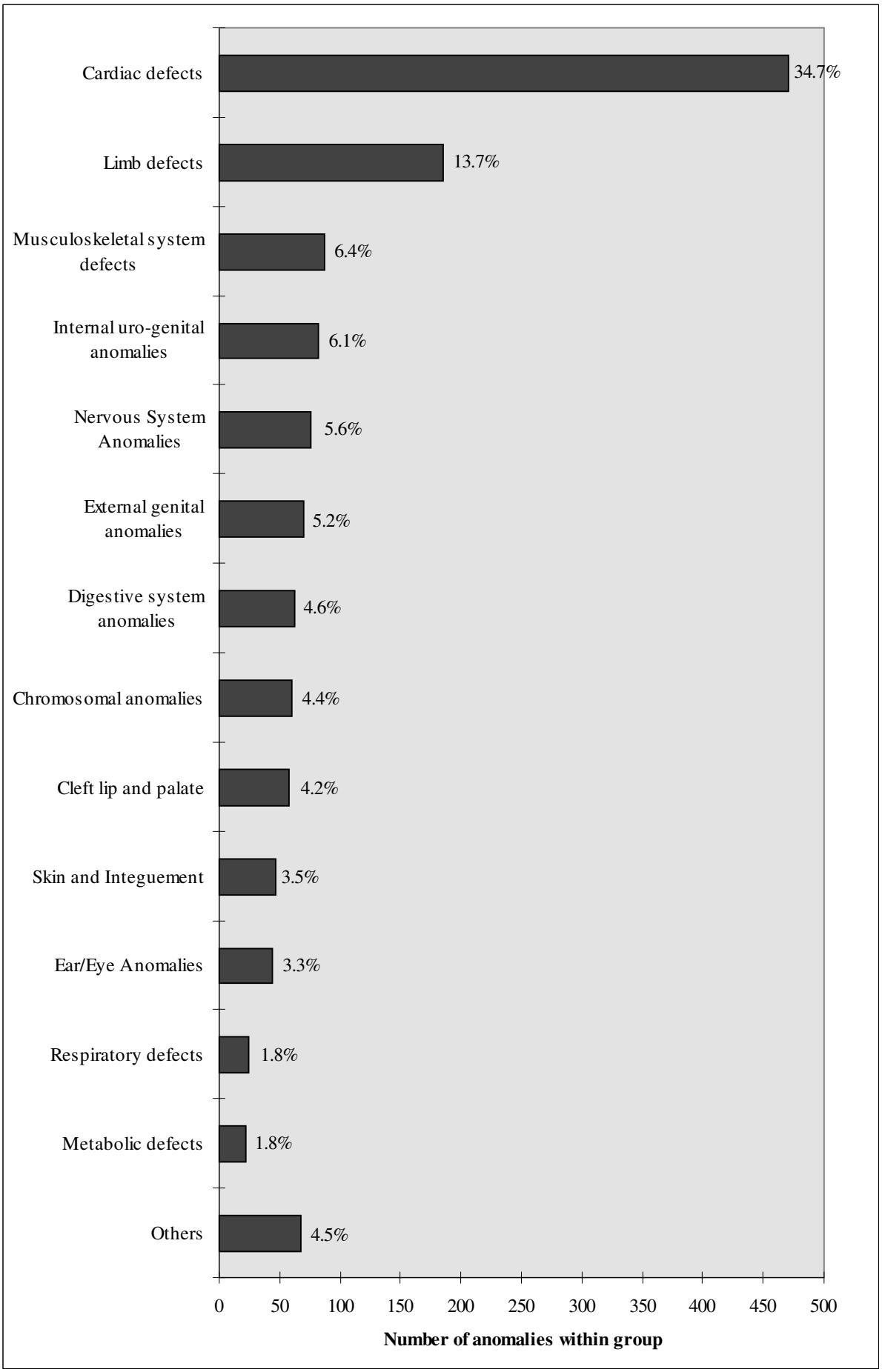
One baby may have one or more different anomalies affecting the same or different systems. The total number of anomalies registered, therefore, does not add up to the same number of babies with anomalies. The table below gives a breakdown of infants/fetuses according to anomaly groups. In this tabulation, a baby having more than one anomaly classified within the same classification subgroup was counted only once within this group. However, a baby/fetus with anomalies in different subgroups was counted once within each subgroup. The distribution of anomalies in decreasing order of frequency is depicted in the figure below. The subgroup classification is that used by EUROCAT (European Registration of Congenital anomalies) of which the Malta Register is a member.

The main anomalies encountered in babies/fetuses were cardiovascular (34.7%) followed by limb and musculoskeletal defects (13.7% and 6.4% respectively). This pattern is similar to that encountered in all years.

Distribution of babies/fetuses with major anomalies registered in Malta from 1993-1997 according to the group/system(s) affected

Subgroup Classification / System(s) affected	1993	1994	1995	1996	1997	1993-97		
						No.	Prevalence / 1000 births	Relative Freq. %
Nervous system anomalies	14	20	16	14	12	76	31.0	5.6
Ear and Eye anomalies	6	9	11	9	9	44	18.0	3.3
Cardiac defects	69	102	81	111	107	470	191.8	34.7
Respiratory system defects	3	9	4	3	5	24	9.8	1.8
Cleft lip and palate	13	12	12	9	11	57	23.3	4.2
Digestive system anomalies	8	13	12	11	18	62	25.3	4.6
External genital anomalies	19	11	13	11	16	70	33.5	5.2
Internal urogenital anomalies	15	23	14	15	15	82	28.6	6.1
Limb defects	33	40	53	24	35	185	75.5	13.7
Musculoskeletal defects	17	21	11	17	21	87	35.5	6.4
Skin and Integuement	13	10	9	6	9	47	19.2	3.5
Chromosomal anomalies	17	8	14	12	9	60	24.5	4.4
Metabolic defects	5	4	7	3	6	25	10.2	1.8
Others	12	13	12	15	9	61	24.9	4.5
Total	244	295	269	260	282	1350	552.0	100

Distribution (%) of infants/fetuses registered in Malta from 1993-1997 according to the group/system affected.



Specific Anomaly Groups

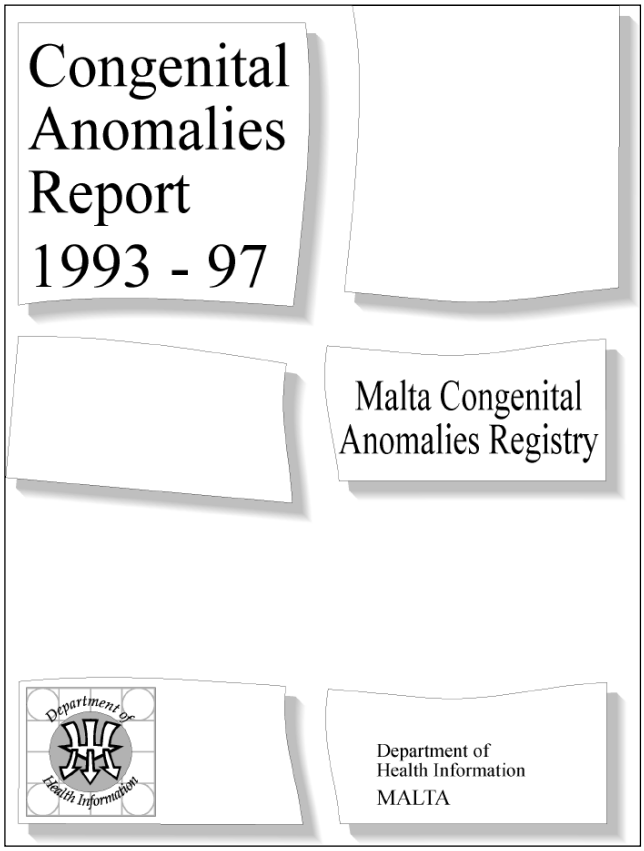
After giving a detailed breakdown of the occurrence of all anomalies by gender and year of birth, the last section of the 1993-97 Report describes certain specific anomaly groups in more detail. The anomaly groups chosen are those occurring with a relatively high prevalence and which contribute to significant morbidity and/or mortality in infants.

The groups discussed are: Neural tube defects, Cleft lip with or without cleft palate, Cleft palate, Congenital heart disease, Anomalies of the digestive system, Pyloric stenosis, Ano-rectal atresia and stenosis, Anomalies of the internal urogenital system, Cystic kidney disease, Limb reduction defects, Polydactyly, Syndactyly, Diaphragmatic hernia and Down syndrome.

For each of these anomaly groups, the distribution of prevalence between Gozo, North West Malta and South East Malta are described. The time trends over the 5 year period 1993-97 are depicted and compared to time trends of European average prevalence rates. The overall prevalence with confidence intervals is also given for each anomaly group and compared to that reported by other European Registries.

For most of the anomalies analysed, there were no statistically significant differences between the prevalence rates found in Malta and those found in other European Registries. A notable exception was in the prevalence of Cleft Palate.

Analysis of data showed that the prevalence of cleft palate in Malta over the period 1993-1997 has been persitently higher than that reported by other European Registries; this higher prevalence is statistically significant. The reasons for this are unknown and require further investigation.



The Malta Congenital Anomalies Report 1993-97 is available at the Department of Health Information and on the Department's web site:

http://www.health.gov.mt/ministry/dhi/mcar.htm

JANUARY - JUNE 2000

This report includes congenital anomalies diagnosed and confirmed in infants/fetuses born during the period January to June 2000 in Malta and Gozo and having been registered at the registry by December, 2000. All major anomalies as defined by EUROCAT (European Registration of Congenital Anomalies) guidelines are included. Minor anomalies are reported only when they occur in combination with major defects.

Total number of infants/fetuses diagnosed with major congenital anomaly

Hospital		Total births		With Congenital anomaly	
		Livebirths*	Fetal Deaths**	Livebirths	Fetal Deaths
St. Luke' s Hosp.	M	855	1	28	1
	F	834	3	22	0
Gozo Gen. Hosp.	M	70	1	1	0
	F	116	0	1	0
Private Hosp.***	M	101	0	1	0
	F	100	0	2	0
Total		**2931**	**5**	**55**	**1**

* Data from National Obstetric Information System (NOIS), WHO-OBSQID project, Malta, GGH & private hosp. records

** Data from National Mortality Register

*** 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
Isolated major anomaly	40
Two or more major anomalies of same system	7
Major anomalies involving two different systems	4
Major anomalies involving three or more different systems	2
Chromosomal anomalies	3
Other congenital malformation syndromes and malformations NEC	0
Total	**56**

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

ICD code	System	Number of infants/fetuses			Fetal
		Total	Males	Females	deaths
Q00-Q99 Congenital malformations, deformations and chromosomal anomalies					
Q00-Q01	Neural tube defects	0	0	0	-
Q02-Q07	Other nervous system defects	1	1	0	-
Q10-Q18	Eyes, ears, face and neck	0	0	0	-
Q20-Q28	Cardiovascular	31	12	19	-
Q30-Q34	Respiratory	0	0	0	-
Q35-Q37	Cleft palate and lip	2	1	1	-
Q38-Q45	Digestive system	1	1	0	-
Q50-Q56	Genital organs	1	1	0	-
Q60-Q64	Urinary	1	1	0	-
Q65-Q79	Musculoskeletal deformities	8	5	3	1
Q80-Q85	Skin & Integument	0	0	0	-
Q86-Q89	Other congenital malformation syndromes and malformations NEC	0	0	0	-
Q90-Q99	Chromosomal anomalies	3	3	0	-
	Two different systems	4	3	1	-
	Three or more different systems (not recognised as a syndrome or sequence)	2	2	0	-
Other Anomalies*					-
C41.4	Congenital sacrococcygeal teratoma	1	0	1	-
G24.8	Levodopa-responsive diurnal dystonia	1	1	0	-
	Total Infants with anomalies	**56**	**31**	**25**	**1**

* These anomalies are not tabulated in EUROCAT reports

The table overleaf gives a breakdown of all the major anomalies registered in babies born between January and June 2000.
In this table:- Hydrocephaly occurring with spina bifida is not included

- Spina bifida occulta is reported only if there are complications or when it occurs in combination with major defects
- Confirmed glandular / coronal / 1st degree hypospadias is not registered unless in combination with major defects.

Other minor anomalies as defined by EUROCAT guidelines are also not registered unless in combination with other major defects.

Anomalies registered for infants born from Jan – June '00 (as registered by 31 December 2000)

Code	Anomaly	Total	Male	Female
Q00-Q01	**Neural Tube defects**	**0**	**0**	**0**
Q02-Q07	**Other anomalies of nervous system**	**0**	**0**	**0**
Q10-Q18	**Anomalies of the eye, ear, face and neck**	**3**	**3**	**0**
Q11.2	Microphthalmos	1	1	0
Q17.3	Other misshapen ear	1	1	0
Q17.4	Misplaced / Low set ears	1	1	0
Q18.9	Congenital anomaly of face and neck, NOS	2	2	0
Q20-Q28	**Anomalies of cardiovascular system**	**47**	**25**	**22**
Q20.5	Transposition of great arteries	1	0	1
Q21.0	Ventricular septal defect	16	9	7
Q21.1	Atrial septal defect	20	9	11
Q21.2	Atrioventricular septal defect	2	1	1
Q21.3	Tetralogy of Fallot	1	0	1
Q22.1	Congenital pulmonary valve stenosis	3	3	0
Q23.0	Congenital stenosis of aortic and mitral valves	1	1	0
Q25.0	Patent ductus arteriosus	3	2	1
Q30-Q34	**Anomalies of respiratory system**	**0**	**0**	**0**
Q35-Q37	**Cleft lip and cleft palate**	**3**	**2**	**1**
Q35.3	Cleft soft palate	1	0	1
Q35.9	Cleft palate, unspecified	1	1	0
Q36.9	Cleft lip, NOS	1	1	0
Q38-Q45	**Anomalies of digestive system**	**2**	**1**	**1**
Q39.1	Atresia oesophagus with tracheo-oesophageal fistula	1	0	1
Q43.1	Hirschsprung's disease	1	1	0
Q50-Q56	**Anomalies of the genital organs system**	**6**	**6**	**0**
Q54.1	Hypospadias, penile	3	3	0
Q54.3	Hypospadias, perineal	1	1	0
Q54.4	Congenital chordae	1	1	0
Q55.2	Congenital malformation of testis or scrotum, NOS	1	1	0
Q60-Q64	**Anomalies of the urinary system**	**4**	**4**	**0**
Q61.4	Renal dysplasia	1	1	0
Q62.0	Congenital hydronephrosis	1	1	0
Q62.7	Congenital vesico-uretero-renal reflux	1	1	0
Q64.2	Congenital urethral valves	1	1	0
Q65-Q79	**Deformities of the musculoskeletal system**	**14**	**10**	**4**
Q66.0	Talipes equinovarus	1	1	0
Q66.4	Talipes calcaneovalgus	1	1	0
Q66.8	Other congenital deformities of feet	1	1	0
Q69.0	Accessory finger(s)	2	0	2
Q69.2	Accessory toes	1	0	1
Q70.2	Fused toes	1	1	0
Q70.3	Webbed toes	1	1	0
Q71.2	Congenital absence of both forearm and hand	1	1	0
Q71.3	Congenital absence of hand and fingers	1	1	0
Q72.3	Congenital absence of foot and toes	1	1	0
Q72.9	Constriction ring syndrome of lower limb, NOS	1	1	0
Q77.4	Achondroplasia	1	0	1
Q79.3	Gastroschisis	1	1	0
Q80-Q85	**Congenital anomalies of the skin & integument**	**3**	**3**	**0**
Q82.5	Congenital non neoplastic naevus (>4cm^2)	1	1	0
Q82.8	Other specified congenital malformations of skin	1	1	0
Q84.6	Other congenital malformations of nails	1	1	0
Q86-Q89	**Other congenital anomalies / multiple anomalies**	**0**	**0**	**0**
Q90-Q99	**Chromosomal anomalies**	**2**	**2**	**0**
Q90.0	Down syndrome /Trisomy 21, meiotic non	1	1	0
Q90.9	Down syndrome / Trisomy 21 NOS	1	1	0
Other major anomalies registered		**3**	**2**	**1**
C41.4	Congenital sacrococcygeal teratoma	1	0	1
G24.8	Levodopa-responsive diurnal dystonia	1	1	0
K07.0	Macrognathism	1	1	0