MALTA CONGENITAL ANOMALIES REGISTRY

Annual Congenital Anomalies Report 2000

Contents:

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IMPORTANT NOTE:

Enclosed together with this report is a short questionnaire regarding your views of this publication. I would be very grateful if you could complete and send it using the self-addressed questionnaire form. All information is confidential and will only be used for the purpose of improving future reporting.

THANK YOU

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AIMS

- To collect data about all fetal deaths and infants with a diagnosis of congenital anomalies on the islands of Malta and Gozo
- To detect any changes in occurrence of congenital anomalies
- 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.

BACKGROUND

Data on congenital anomalies diagnosed at birth at St. Luke' s Hospital (Malta) has been collected through the University of Malta since 1985. The register is a member of EUROCAT (European Registration of Congenital Anomalies and Twins). Funding by the University of Malta ran into difficulties in 1995. The Department of Health Information started co-ordinating all previous efforts of recording congenital anomalies and started a population based Malta Congenital Anomalies Register in January 1997. This register covers births from all hospitals on the Maltese Islands and includes cases diagnosed until one year of age. The register continues to be a member of EUROCAT.

COVERAGE

The register is population based and covers all births on the islands of Malta and Gozo which amount to just under 5000 births per year. The small size and population of the islands (area: 316 km^2; population: 376,335); the well defined boundaries, absence of significant ethnic minority groups and illegality of termination of pregnancy 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 echocardiography units to collect data directly and review patient notes.

The hospitals involved in data capture are St. Luke' s Hospital (SLH), Gozo General Hospital (GGH), St. Philip' s Hospital, Capua Palace Hospital, and St. James Hospital.

Present sources of data from St. Luke' s Hospital are: Doctor' s notification, Labour ward, Post-natal / Nursery ward, SCBU, Echocardiography Lab., Genetics Clinic, National Obstetric Information Systems Database, SLH Hospital Activity Analysis Database, National Mortality Register and Pathology Autopsy report and Hypothyroid screening programmes.

INTERNATIONAL RELATIONSHIPS

The Malta Congenital Anomalies Register has been a member of the European Registration of Congenital Anomalies (EUROCAT) since 1986. In September 2000 it was accepted as an Associate Member of the International Clearinghouse of Births Defects Monitoring Systems (ICBDMS).

There is more to life than increasing its speed.
- Gandhi

PRENATAL ULTRASOUND DIAGNOSIS OF FETAL ABNORMALITIES

INTRODUCTION

Prenatal diagnosis of fetal abnormalities may be achieved using ultrasound, chorionic villous sampling, amniocentesis and blood tests. In Malta only prenatal ultrasound examination is performed routinely, with ultrasonographic examination being performed at about 20 weeks and 36 weeks gestation in non-complicated pregnancies.

HISTORY OF OBSTETRIC ULTRASOUND

Ultrasonography utilises high frequency sound waves and their reflection to visualise internal structures. The acronym SONAR refers to Sound Navigation and Ranging. The earliest experiments with calculating the speed of sound through water were carried out in the waters of Lake Geneva by a Swiss physicist Daniel Colladen as early as 1822. Underwater detection systems were later developed after the Titanic sank in 1912 and for the purpose of underwater navigation by submarines in World War I. The first patent for an underwater echo ranging sonar was filed one month after the sinking of the Titanic.[1]

Probably the most significant contribution that was made in the history of ultrasound in Obstetrics and Gynaecology came from Glasgow, Scotland, where Ian Donald was Professor at the University Department of Midwifery. It was in 1955 that this Scottish doctor put into practice his idea that sonar could be used for medical diagnosis. In 1959 he noted that clear echoes could be obtained from the fetal head and began to apply this information - this was the conception of obstetric ultrasound.[1]

ULTRASOUND SCREENING AND OUTCOME

Currently, the most widely used technique for fetal evaluation is 2 dimensional real-time ultrasound. This type of ultrasound imagery is displayed on a monitor and fetal motion is observed as it occurs.

More recent advances involve the use of 3 dimensional ultrasound. These scans require special probes and software to accumulate and render the images and the rendering time has been reduced from minutes to seconds. A good 3-D image is often quite impressive and accurate. Both doctors and parents can better appreciate the presence or absence of a certain abnormality in a 3-D scan.

Real-time scanners utilise very high frequency sound waves of between 3.5-7.0 megahertz (i.e. 3.5 to 7 million cycles per second).

Ultrasound examination in the first trimester of pregnancy can determine the presence or absence of a gestational sac, fetal number, placental location and gestational age, as well as document fetal viability and evaluate the uterus and adnexae. Examination in the second or third trimester can determine fetal presentation, assess amniotic fluid volume and search for gross malformations. Targeted ultrasound examinations evaluate fetal intracranial structures (namely ventricles and cerebellum), spine, heart, bladder, kidneys, stomach, thorax, abdominal wall, long bones and the umbilical cord insertion site.

Ultrasound screening is advocated during pregnancy to detect congenital anomalies, multiple gestation pregnancies, fetal growth disorders, placental abnormalities and errors in the estimation of gestational age. Although ultrasound screening has for some time been an integral part of antenatal care in industrialised countries, it is not certain whether the detection of these conditions through screening leads to interventions that improve perinatal outcome. In fact much controversy exists as to whether screening low risk cases affects outcome.[2-12]

"Ultrasound scanning is a household word. Every mother knows it and many have pictures to prove it. It is painless, safe and reliable. Its success since its beginnings in the 1950's is truly astonishing. It started in Glasgow in the University Department of Midwifery under Professor Ian Donald and seemed a rather crazy experiment at the time. But Ian was no backroom boffin, but a full blown falmboyant consultant at the sharp edge of one of medicine's most acute specialities – a colourful character of Johnsonian richness for whom I am a very inadequate Boswell".

Dr. James Willocks

in Medical Ultrasound-A Glasgow Development which Swept the World. Avenue, 1996;January, no.19

DETECTION OF BIRTH DEFECTS BY ULTRASOUND

Detection of birth defects is a major reason for which screening antenatal ultrasound is advocated.[13-16] While birth defects may be apparent on the ultrasound image, the difficulty in recognising malformations by the physician or technician performing the examination is a function which is directly related to the experience of the individual performing the evaluation. In a multicentre study known as the RADIUS trial (Routine Antenatal Diagnostic Imaging with Ultrasound Study),[5] fetal ultrasound was evaluated to determine if it was useful during pregnancy. The RADIUS trial reported that identification of birth defects was related to the training of the physician performing or interpreting the examination. For example, when the ultrasound examination was performed by the obstetrician or radiologist in a community hospital, neither of which had specialised training detecting birth defects, only 13% of all major birth defects were idenitified prenatally. Therefore, 6 out of 7 fetuses with major birth defects were not identified following screening ultrasound. An additional finding of the study was that none of the serious heart defects were detected before birth. The RADIUS study pointed out, however, that if the ultrasound was performed by specialists trained in prenatal diagnosis, the detection rate of birth defects was almost three times higher (35% vs 13%).[5]

SAFETY OF PRENATAL ULTRASOUND

Ultrasound scan is currently considered to be a safe, non-invasive, accurate and cost-effective investigation of the fetus. However, certain studies have thrown some doubt on this. Concerns over a possible association between exposure to ultrasound in utero and intrauterine growth restriction, increased risk of childhood malignancies and delayed subsequent neurological development have been voiced in the past; however other more recent, larger controlled studies have not found any such associations.[17-25] Recent studies have nonetheless shown an association between fetal ultrasound and non-righthandedness in males, indicating an affect on the fetal brain.[26-28] Kieler et al, (2001)[29] conclude that "ultrasound exposure in fetal life increases the risk of left-handedness in men, suggesting that prenatal ultrasound affects the fetal brain." In view of the conflicting evidence it is therefore advisable that repeated prenatal ultrasound imaging and Doppler flow examinations should be restricted to those women in whom the information is likely to be of clinical benefit.

The issue of safety of ultrasound in pregnancy is a subject of continuing vigilance. Safety statements are regularly issued and updated by recognised bodies. Excerpts of the safety statements made by the European Committee for Medical Ultrasound (ECMUS), British Medical Ultrasound Society (BMUS) and American Institute of Ultrasound in Medicine (AIUM) are given in Annexes 1 to 3.

PSYCHOLOGICAL AND ETHICAL CONSIDERATIONS

Ultrasound scanning consitutes the first visual encounter of the parent with their baby. In a study by Whynes DK (2002),[30] investigating women's attitudes towards ultrasound scanning in pregnancy it was found that: 'Most women reported positive feelings towards scanning' and 'were satisfied with their experiences of routine ultrasound scanning.'

Since a wide range of major abnormalities may be diagnosed it is advocated that parents should be informed of the possibility of such diagnosis and in the event of an abnormality being detected, psychological support should be available.[31] Termination of pregnancy is not an option in Malta, however prenatal diagnosis can help plan the future management of the baby and also gives time for counseling and preparation of the parents.

PRENATAL ULTRASOUND DIAGNOSIS - MALTA 1993-2000

The Malta Congenital Anomalies Registry (MCAR) keeps records of all babies born with congenital anomalies and when these were first diagnosed or suspected. The Registry has recently started collaborating with the obstetric ultrasound section at St. Luke's Hospital and now all antenatal diagnoses / suspicions of fetal anomalies are being reported to the Registry and babies are being followed after birth to document the outcome.

In Malta, in the year 2000, an average of 2.6 ultrasounds were performed on every pregnant mother (National Obstetric Information Systems database – Department of Health Information).

Analysis of data from the Malta Congenital Anomalies Registry between 1993 and 2000 shows that out of the 1411 babies registered at the registry during this 8 year period, 134 (9.5%) were diagnosed antenatally. The average gestational age at diagnosis was 28 weeks. The distribution of gestational age at diagnosis is given in Table 1.

Table 1 - Gestational age at diagnosis for prenatally diagnosed defects (1993-2000).

Gestational age at diagnosis (weeks)	Babies diagnosed	%
10-14	7	5
15-19	11	8
20-24	27	20
25-29	18	14
30-34	35	26
35-39	27	20
40-45	4	3
Unknown	5	4
Total	**134**	**100%**

The most frequently antenatally diagnosed defects were renal: 47 (35%); musculoskeletal defects: 28 (21%); neural tube defects: 25 (19%); other nervous system defects: 15 (11%) and others: 19 (14%).

Of all the babies diagnosed prenatally, 68 (51%) had isolated defects; 14 (10%) had multiple defects of the same system and 52 (39%) had multiple defects affecting different systems.

Table 2 shows the percentage of babies with potentially diagnosable defects that were actually diagnosed by ultrasound in Malta from 1993 to 2000. Quoted rates for prenatally diagnosed defects vary greatly in different papers.[32-33] This variation is undoubtedly related to both the type of ultrasonographic equipment available as well as the experience of the individual carrying out the examination.[5]

Figure 1 - Distribution of antenatally diagnosed defects (1993-2000)

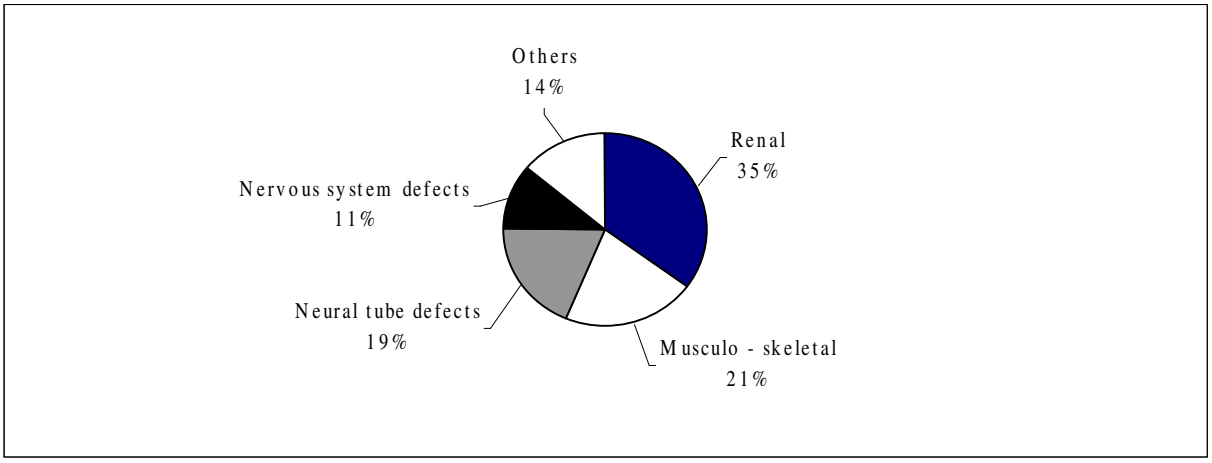

Table 2 - Proportion of specific birth defects with prenatal diagnosis (1993-2000)

Anomaly	Total no. of anomalies registered	No. diagnosed prenatally	Proportion diagnosed prenatally (%)
Anencephaly	15	15	100
Spina bifida	21	5	24
Encephalocele	7	4	57
Hypoplastic left heart	4	0	0
Cleft palate only	45	1	2
Cleft lip with or without cleft palate	30	0	0
Omphalocele	5	3	60
Gastroschisis	4	2	50
Diaphragmatic hernia	19	4	21
All Renal anomalies	64	37	58
Renal agenesis	2	2	100
Cystic kidney	10	6	60
Obstructive renal defects	36	23	64
All Musculoskeletal defects	163	2	1
Limb reduction defects: upper limb	11	0	0
lower limb	1	1	100

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ANNEX 1

European Committee for Medical Ultrasound (ECMUS)
Clinical Safety Statement for Diagnostic Ultrasound (2002)

http://www.efsumb.org/safstatement2002.htm

"Diagnostic ultrasound has been widely used in clinical medicine for many years with no proven deleterious effects. However, as the use of ultrasound increases, with the introduction of new techniques, with a broadening of the medical indications for ultrasound examinations, and with increased exposure, continuous vigilance is essential to ensure its continued safe use.
A broad range of ultrasound exposure is used in the different diagnostic modalities currently available. Doppler imaging and measurement techniques may use higher exposures than those used in B- and M-modes, with pulsed Doppler techniques having the potential for the highest levels.
The recommendations contained in this statement assume that commercial ultrasound equipment conforming to international safety standards is being used, and that it is used prudently, by competent personnel who are trained in safety matters."

"**Ultrasound exposure during pregnancy**
The embryonic period is known to be particularly sensitive to any external influences. Until further scientific information is available, investigations should be carried out with careful control of output levels and exposure times. With increasing mineralisation of the fetal bone as the fetus develops, the possibility of heating fetal bone increases. The user should prudently limit exposure of critical structures such as the fetal skull or spine during Doppler studies."

ANNEX 2

Safety Group of the British Medical Ultrasound Society (BMUS)
Statement on the safe use, and potential hazards, of diagnostic ultrasound (2000)

http://www.bmus.org/safety_of_ultrasound.htm#state

"Ultrasound is now accepted as being of considerable diagnostic value. There is no evidence that diagnostic ultrasound has produced any harm to patients in the four decades that it has been in use. However, the acoustic output of modern equipment is generally much greater than that of the early equipment and, in view of the continuing progress in equipment design and applications, outputs may be expected to continue to be subject to change. Also, investigations into the possibility of subtle or transient effects are still at an early stage. Consequently diagnostic ultrasound can only be considered safe if used prudently."

ANNEX 3

American Institute of Ultrasound in Medicine (AIUM)
Clinical Safety (1997)

http://www.aium.org/consumer/statement_selected.asp?statement=11

"Diagnostic ultrasound has been in use since the late 1950s. Given its known benefits and recognized efficacy for medical diagnosis, including use during human pregnancy, the American Institute of Ultrasound in Medicine herein addresses the clinical safety of such use:
There are no confirmed biological effects on patients or instrument operators caused by exposures from present diagnostic ultrasound instruments. Although the possibility exists that such biological effects may be identified in the future, current data indicate that the benefits to patients of the prudent use of diagnostic ultrasound outweigh the risks, if any, that may be present."

ANNUAL REPORT - 2000

This report includes congenital anomalies diagnosed and confirmed in infants/fetuses born during the period January to December 2000 in Malta and Gozo and which were registered at the Malta Congenital Anomalies Registry by March 2002. All major anomalies as defined by EUROCAT guidelines (European Registration of Congenital Anomalies) are registered. Minor anomalies are only included when they occur in combination with major defects. Infants are registered if diagnosis of the defect(s) is made until one year of age.

Since a baby may have more than one defect, this report makes a clear distinction between the analyses of the *numbers of infants* having one or more congenital anomalies and the analyses of the *number of anomalies;* the latter do not add up to the number of infants.

A description of the functioning of the Malta Congenital Anomalies Registry may be found on our web page: http://www.health.gov.mt/ministry/dhi/mcar.htm

INFANTS / FETUSES

The total number of births (live and stillbirths) in Malta and Gozo during the year 2000 was 4272 (4255 livebirths and 17 fetal deaths). Of these, 135 were registered as having one or more major anomalies, giving an overall birth prevalence rate of 31.6/1000 total births.

Of the 135 registered infants/fetuses with congenital anomalies, 69 (51%) were male and 66 (49%) were female giving a male:female ratio of 1:0.9. A detailed breakdown is given in Table 1.

Table 1 - Infants / fetuses diagnosed with major congenital anomaly in 2000

	Sex	Total Births		With Congenital Anomalies	
		Live births*	Fetal Deaths**	Live births	Fetal Deaths
Malta	M	1967	10	66	1
	F	1937	5	61	1
Gozo	M	159	0	2	0
	F	192	2	4	0
Total		**4255**	**17**	**133**	**2**

* Source: Demographic Review of the Maltese Islands - 2000
** Source: National Mortality Register – Dept. of Health Information

SOURCE OF INFORMATION

The Malta Congenital Anomalies Register mainly utilises active data collection from several sources including: St. Luke's Hospital (SLH) Labour, Post-natal, Nursery and SCBU wards, SLH Cardiac Lab., Genetics Clinic, SLH Hospital Activity Analysis Register (HAA), National Obstetric Information Systems database (NOIS), National Mortality Register and Autopsy reports and Hypothyroid screening programme.

The majority of cases 108 (80%) came from the SLH obstetric and paediatric wards and cardiac lab., where staff from the registry regularly go to collect information on neonates and echocardiography records respectively. The distribution of the major sources of information is given below.

Table 2 - Main Sources of information for babies diagnosed with major congenital anomalies (2000)

First Source of information	Number	(%)
Active collection from SLH Post natal wards	51	38
Cardiac lab records	57	42
NOIS (National Obstetric Inforamtion Systems database)	5	4
HAA (SLH Hospital Activity Analysis database)	6	4
Genetic Clinic Records	5	4
Death Register / Autopsy records	4	3
Doctor / Midwife notification	3	2
Others	4	3

ISOLATED vs MULTIPLE ANOMALIES

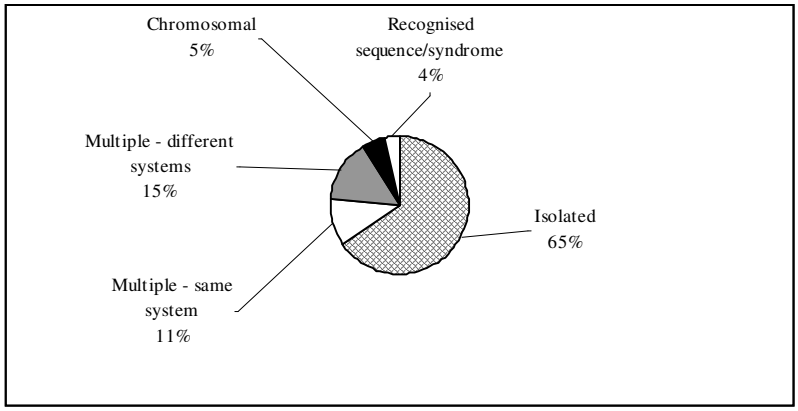
Table 3 - Distribution of infants / fetuses according to number of major anomalies (2000)

Anomalies	Number	(%)
Isolated	88	65
Multiple anomalies of same system	15	11
Multiple anomalies of different systems	20	15
Chromosomal Anomalies	7	5
Recognised sequences / syndromes	5	4

The majority of babies registered in 2000 had isolated defects (65%). Multiple anomalies of the same system and multiple anomalies of different systems occurred with similar frequencies.

5% of infants / fetuses registered in 2000 had chromosomal anomalies and 4% had recognised syndromes or sequences.

Figure 1 - Distribution according to number of anomalies (2000)



MATERNAL AGE DISTRIBUTION

Table 4 - Prevalence of infants/fetuses with anomaly according to maternal age (2000)

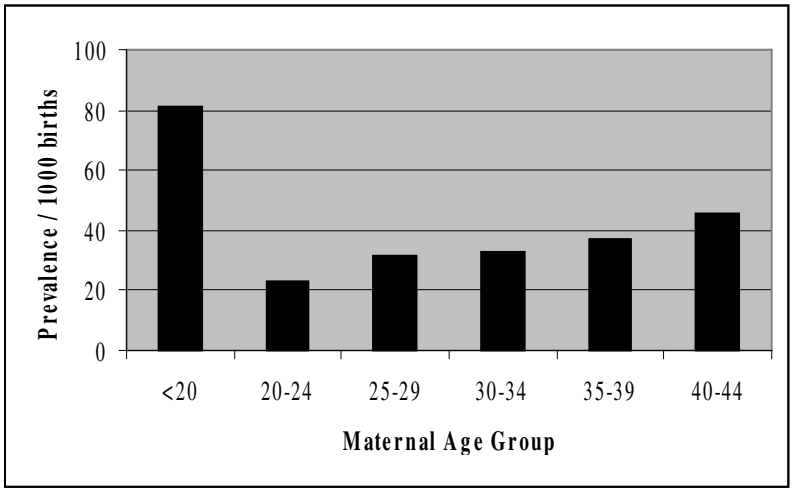
Maternal age	Total Deliveries	Deliveries with anomaly	Prevalence /1000 births
<20	239	10	81.3
20-24	873	20	22.9
25-29	1545	49	31.7
30-34	1056	35	33.1
35-39	428	16	37.4
40-44	110	5	45.5
45 and over	4	0	na

The highest prevalence of babies with birth defects in 2000 was registered in the youngest maternal age group (81.3 / 1000 births), while the lowest prevalence was recorded in the 20-24 year age group. A steady increase of prevalence with rising age was recorded from 25 years on.

An overall prevalence of 39.0/1000 births was recorded in deliveries to mothers 35 years and over.

Higher prevalence of birth defects in the younger and older maternal age groups has been documented in scientific literature and is an area of ongoing research.

Figure 2 – Maternal age distribution (2000)



GEOGRAPHICAL DISTRIBUTION

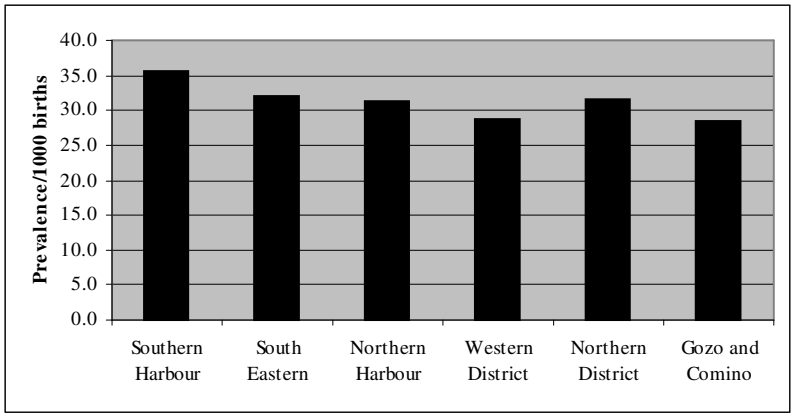
Table 5 - Prevalence of infants/fetuses with anomaly according to geographical distribution (2000)

Locality	Total Deliveries	Deliveries with anomaly	Prevalence /1000
Southern Harbour	811	29	35.8
South Eastern	718	23	32.0
Northern Harbour	1084	34	31.4
Western District	592	17	28.7
Northern District	699	22	31.5
Gozo and Comino	351	10	28.5

The geographical break-down used in this analysis is that used by the National Statistics Office in the Demographic Review of the Maltese Islands - 2000. Details of this breakdown may be found in the latter publication.

The highest prevalence of babies with birth defects in 2000 was registered for mothers residing in the Southern Harbour region (35.8 / 1000 births), while the lowest prevalence was recorded for Gozo and the Western regions- 28.5 and 28.7/1000 births respectively.

Figure 3 – Geographical distribution (2000)



DISTRIBUTION BY MONTH OF BIRTH

Table 6 - Prevalence of infants/fetuses with anomaly according to month of birth (2000)

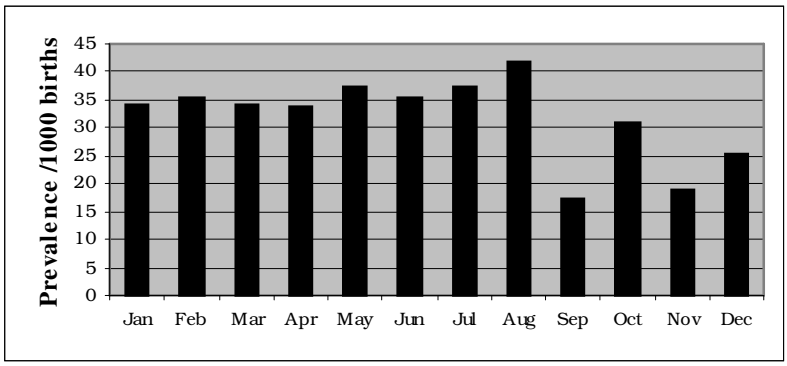
Month of birth	Total Deliveries	Deliveries with anomaly	Prevalence / 1000 births
January	381	13	34.1
February	338	12	35.5
March	350	12	34.3
April	327	11	33.6
May	320	12	37.5
June	312	11	35.3
July	374	14	37.4
August	385	16	41.6
September	350	6	17.1
October	356	11	30.9
November	369	7	19.0
December	393	10	25.4

The highest prevalence of babies with birth defects in 2000 was registered in the month of August: (41.6/1000 births).

The lowest prevalence was recorded in September: (17.1/1000 births).

No significant variation in seasonal prevalence is observed.

Figure 5 – Distribution by month of birth (2000)



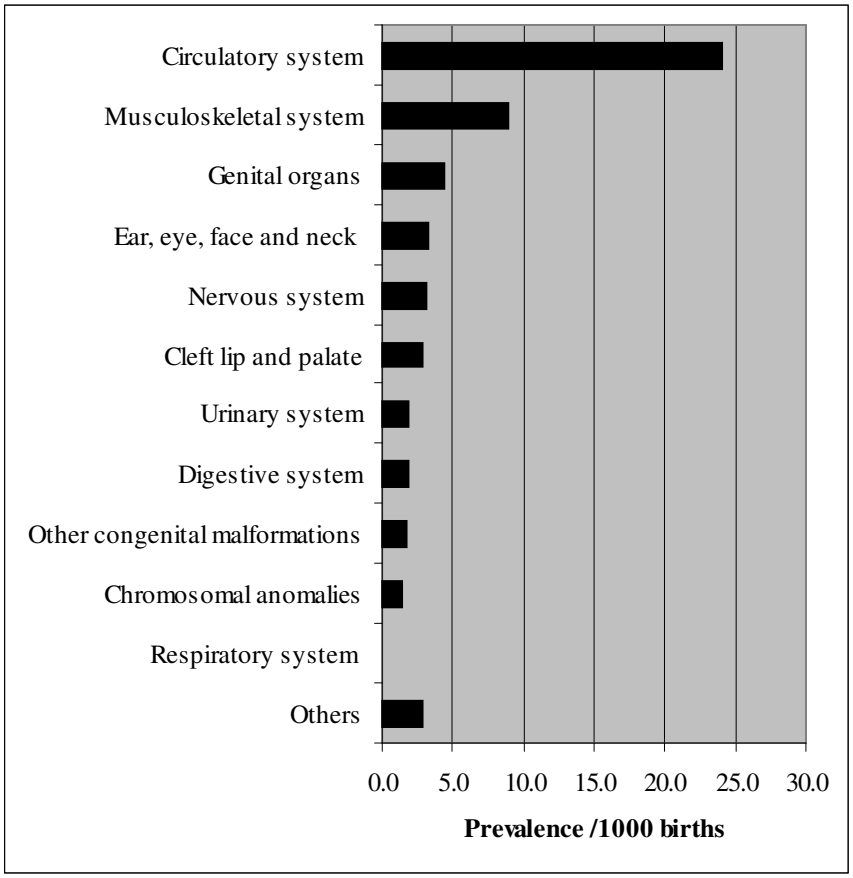
A total of 241 anomalies were recorded in the 135 babies registered at the registry; giving a prevalence rate of 56.4/1000 births.

The distribution of anomalies by system affected is given in Table 7 and represented graphically in Figure 6.

Table 7 - Distribution of anomalies by system affected (2000)

Systems affected	ICD-10 Code	Number of anomalies	Prevalence / 1000 births	Relative frequency (%)
Nervous system anomalies	Q00-Q07	13	3.0	5.4
Ear, eye, face and neck anomalies	Q10-Q18	14	3.3	5.8
Circulatory system defects	Q20-Q28	103	24.1	42.9
Respiratory system defects	Q30-Q34	0	0.0	0.0
Cleft lip and palate	Q35-Q37	12	2.8	5.0
Malformations of the digestive system	Q38-Q45	8	1.9	3.3
Malformations of genital organs	Q50-Q56	19	4.4	7.9
Malformations of urinary system	Q60-Q64	8	1.9	3.3
Malformations of musculoskeletal system	Q65-Q79	38	8.9	15.8
Other congenital malformations	Q80-Q89	8	1.9	2.9
Chromosomal anomalies	Q90-Q99	6	1.4	2.5
Others		12	2.8	5.0
TOTAL		241	56.2	100.0

Figure 6 - Distribution of anomalies by system and in order of frequency (2000)



The most commonly encountered group of anomalies in the year 2000 were defects of the Circulatory system which include cardiac defects. These accounted for 42.9% of all anomalies registered.

The next most frequently encountered group of anomalies was musculo-skeletal defects, which accounted for 15% of all anomalies registered in 2000.

DETAILED BREAKDOWN OF ALL ANOMALIES – 2000

Table 8 - All Anomalies registered for infants/fetuses born from January – December 2000 (as registered by March 2002)*

Code	Malformations	Males	Females	Unspecified	Total
	Total anomalies registered	**143**	**98**	**0**	**241**
Q00-Q01	**Neural tube defects**	**2**	**0**	**0**	**2**
Q01.2	Occipital encephalocele	2	0	0	2
Q02-Q07	**Other anomalies of nervous system**	**7**	**4**	**0**	**11**
Q02.	Microcephaly	1	0	0	1
Q03.1	Dandy walker syndrome	1	0	0	1
Q03.8	Other congenital hydroephalus	0	1	0	1
Q03.9	Congenital hydrocephalus, unspecified	0	1	0	1
Q04.0	Congenital malformations of corpus callosum	1	0	0	1
Q04.3	Other reduction deformities of brain	1	0	0	1
Q04.5	Megalencephaly	1	0	0	1
Q04.6	Congenital cerebral cysts	1	0	0	1
Q05.2	Lumbosacral spinabifida with hydrocephalus	1	0	0	1
Q05.7	Lumbosacral spina bifida nos	0	1	0	1
Q07.8	Other specified congenital malformations of nervous system	0	1	0	1
Q10-Q18	**Anomalies of eye, ear, face and neck**	**10**	**4**	**0**	**14**
Q11.2	Microphthalmos	1	0	0	1
Q13.3	Congenital corneal opacity	0	1	0	1
Q13.4	Other congenital corneal malformations	1	0	0	1
Q14.1	Congenital malformation of retina	1	0	0	1
Q17.3	Other misshapen ear	1	0	0	1
Q17.4	Misplaced/low set ears	3	2	0	5
Q18.1	Preauricular sinus and cyst	0	1	0	1
Q18.4	Macrostomia	1	0	0	1
Q18.9	Congenital anomaly of face and neck nos	2	0	0	2
Q20-Q28	**Anomalies of circulatory system**	**48**	**55**	**0**	**103**
Q20.3	Transposition of great vessels	2	1	0	3
Q20.5	Transposition of great arteries	0	1	0	1
Q21.0	Ventricular septal defect	17	19	0	36
Q21.1	Atrial septal defect	12	19	0	31
Q21.2	Atrioventricular septal defect	2	1	0	3
Q21.3	Tetralogy of fallot	1	2	0	3
Q22.1	Congenital pulmonary valve stenosis	4	3	0	7
Q22.4	Congenital tricuspid stenosis	1	0	0	1
Q23.0	Congenital stenosis of aortic and mitral valves	2	1	0	3
Q25.0	Patent ductus arteriosus	2	6	0	8
Q25.1	Coarctation of aorta	2	1	0	3
Q25.4	Other congenital malformations of aorta	1	0	0	1
Q25.6	Stenosis of pulmonary artery	1	1	0	2
Q26.2	Total anomalous pulmonary venous drainage	1	0	0	1
Q30-Q34	**Anomalies of respiratory system**	**0**	**0**	**0**	**0**

Code	Malformation	Males	Females	Unspecified	Total
Q35-Q37	**Cleft lip and cleft palate**	**9**	**3**	**0**	**12**
Q35.3	Cleft soft palate	1	1	0	2
Q35.7	Cleft uvula	1	0	0	1
Q35.9	Cleft palate, unspecified	2	0	0	2
Q36.0	Cleft lip, bilateral	0	1	0	1
Q36.9	Cleft lip nos	1	0	0	1
Q37.0	Cleft hard palate with cleft lip, bilateral	2	0	0	2
Q37.4	Cleft hard and soft palate with cleft lip, bilateral	0	1	0	1
Q37.9	Cleft palate with cleft lip	2	0	0	2
Q38-Q45	**Anomalies of digestive system**	**5**	**3**	**0**	**8**
Q39.0	Atresia of oesophagus without fistula or nos	0	1	0	1
Q39.1	Atresia of oesophagus with tracheo-oesophageal fistula	0	1	0	1
Q40.0	Congenital hypertrophic pyloric stenosis	3	0	0	3
Q43.0	Meckel's diverticulum	0	1	0	1
Q43.1	Hirschsprung's disease	2	0	0	2
Q50-Q56	**Anomalies of genital organs**	**18**	**1**	**0**	**19**
Q52.6	Congenital malformation of clitoris	0	1	0	1
Q53.2	Undescended testicle bilateral	2	0	0	2
Q54.1	Hypospadias penile	4	0	0	4
Q54.3	Hypospadias, perineal	3	0	0	3
Q54.4	Congenital chordee	4	0	0	4
Q55.2	Other congenital malformations of testis and scrotum	2	0	0	2
Q55.6	Other congenital malformations of penis	2	0	0	2
Q56.4	Indeterminate sex, ambiguous genitalia	1	0	0	1
Q60-Q64	**Anomalies of urinary system**	**7**	**1**	**0**	**8**
Q60.0	Renal agenesis, unilateral	1	0	0	1
Q61.3	Polycystic kidney, unspecified	1	0	0	1
Q61.4	Renal dysplasia	1	0	0	1
Q61.9	Cystic kidney disease, unspecified	1	0	0	1
Q62.0	Congenital hydronephrosis	1	1	0	2
Q62.7	Congenital vesico-uretero-renal reflux	1	0	0	1
Q64.2	Congenital urethral valves	1	0	0	1
Q65-Q79	**Deformities of musculoskeletal system**	**21**	**17**	**0**	**38**
Q66.0	Talipes equinovarus	3	0	0	3
Q66.4	Talipes calcaneovalgus	1	0	0	1
Q66.6	Other congenital valgus deformities of feet	1	0	0	1
Q66.8	Other congenital deformities of feet	3	1	0	4
Q67.5	Congenital deformity of spine	0	1	0	1
Q68.1	Congenital deformity of hand	1	0	0	1
Q68.8	Other specified congenital musculoskeletal deformities	0	2	0	2
Q69.0	Accessory finger(s)	0	4	0	4
Q69.1	Accessory thumb(s)	0	1	0	1
Q69.2	Accessory toes	0	2	0	2
Q70.2	Fused toes	1	1	0	2
Q70.3	Webbed toes	1	0	0	1
Q71.3	Congenital absence of hand and fingers	2	1	0	3
Q72.3	Congenital absence of foot and toes	1	0	0	1
Q72.9	Reduction defect of lower limb, unspecified	1	0	0	1

Code	Malformation	Males	Females	Unspecified	Total
Q65-Q79	**Deformities of musculoskeletal system (cont.)**				
Q74.9	Unspecified congenital malformation of limb(s)	1	1	0	2
Q75.2	Hypertelorism	0	1	0	1
Q77.4	Achondroplasia	1	1	0	2
Q79.0	Congenital diaphragmatic hernia	2	1	0	3
Q79.1	Other congenital malformations of diaphragm	1	0	0	1
Q79.3	Gastroschisis	1	0	0	1
Q80-Q85	**Congenital anomalies of skin and integument**	**6**	**0**	**0**	**6**
Q82.5	Congenital non-neoplastic naevus	2	0	0	2
Q82.8	Other specified congenital malformations of skin	1	0	0	1
Q82.9	Congenital malformation of skin, unspecified	1	0	0	1
Q84.6	Other congenital malformations of nails	1	0	0	1
Q84.8	Other specified congenital malformations of integument	1	0	0	1
Q86-Q89	**Other congenital anomalies**	**0**	**2**	**0**	**2**
Q87.1	Congenital malformation syndromes associated with short stature	0	1	0	1
Q89.4	Conjoined twins	0	1	0	1
Q90-Q99	**Chromosomal anomalies**	**4**	**2**	**0**	**6**
Q90.0	Trisomy 21, meiotic nondisjunction	2	0	0	2
Q90.9	Trisomy 21, NOS	1	1	0	2
Q91.3	Edward' s syndrome, unspecified	0	1	0	1
Q91.4	Trisomy 13, meiotic non disjunction	1	0	0	1
Other anomalies registered		**6**	**6**	**0**	**12**
C41.4	Malignant neoplasm of pelvic bones, sacrum and coccyx	0	1	0	1
D10.3	Benign neoplasm of other and unspecified parts of the mouth	0	1	0	1
D17.7	Benign lipomatous neoplasm of other sites	0	1	0	1
D18.0	Haemangioma any site	0	1	0	1
G24.8	Dystonia	1	0	0	1
H55	Nystagmus and other irregular eye movements	1	0	0	1
I42.4	Congenital cardiomyopathy	1	0	0	1
K07.0	Macrognathism	2	0	0	2
N04	Congenital nephrotic syndrome	0	1	0	1
P55.1	ABO isoimmunisation of fetus and newborn	0	1	0	1
P83.5	Congenital hydrocele	1	0	0	1

* The table gives a breakdown of all the major anomalies registered in babies born between January and December 2000 as registered by March 2002. 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.
- Skin tags, naevi, angiomas, hemangiomas, glomus tumors, lymphangiomas and birthmarks less than 4 cms^2 are 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.