MALTA CONGENITAL ANOMALIES REGISTRY

Annual Congenital Anomalies Report 2002

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INTRODUCTION

MALTA CONGENITAL ANOMALIES REGISTER

Congenital Anomalies include structural defects (congenital malformations, deformations, disruptions and dysplasias), chromosomal abnormalities, inborn errors of metabolism and hereditary diseases. They are a major cause of infant mortality, childhood morbidity and long-term disability. They are also a major cause of embryonic and fetal death and are among the leading causes of years of potential life lost. Congenital anomalies carry a high burden to affected individuals, their families and the community in terms of quality of life, participation in the community and need for services. They are therefore of significant Public Health importance.

A register of congenital anomalies was first started in Malta in 1985 with data being collected from St. Luke's Hospital (SLH) as part of a research project funded by the University of Malta. In 1997, the running of the register was assumed by the Department of Health Information and a computerised database was developed with computerised data being backdated to 1993. The register presently collects data from all hospitals on the Maltese Islands and utilises multiple sources of information including information from maternity wards at SLH, Doctors' notifications, National Obstetric Information Systems database, Cardiac Lab. records, Hospital Activity Analysis database, information from the Genetics Clinic and others.

The aims of the Malta register are:

- 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 both locally and internationally;
- to issue regular reports and provide physicians and the general public with information they may need, always respecting strict confidentiality.

The Malta Congenital Anomalies Registry has a regularly updated web site: http://health.gov.mt/ministry/dhi/mcar.htm. This site contains the most recent data and summary statistics.

All data collected by the Registry is processed in accordance with the Data Protection Act, 2001.

INTERNATIONAL RELATIONSHIPS

European Registration of Congenital Anomalies (EUROCAT)
The Malta Congenital Anomalies Register has been a full member of EUROCAT (http://www.eurocat.ulst.ac.uk) since 1986, regularly transmitting anonymised case-based data to this network.

EUROCAT is a European network of population-based registries for the epidemiologic surveillance of congenital anomalies. It was started in 1979 and now more than a million births per year in Europe are surveyed by 39 registries in 19 European countries. EUROCAT Central Registry is currently located at the University of Ulster, Northern Ireland and it houses a standardised central database on more than 180,000 cases of congenital anomaly among livebirths, stillbirths and terminations of pregnancy. This database is updated every year.

The objectives of EUROCAT are: to provide essential epidemiologic information on congenital anomalies in Europe

- to facilitate the early warning of new teratogenic exposures,
- to evaluate the effectiveness of primary prevention,
- to assess the impact of developments in prenatal screening,
- to act as an information and resource center for the population, health professionals and managers regarding clusters or exposures or risk factors of concern,
- to provide a ready collaborative network and infrastructure for research related to the causes and prevention of congenital anomalies and the treatment and care of affected children and
- to act as a catalyst for the setting up of registries throughout Europe collecting comparable, standardised data.

As congenital anomalies have a relatively low prevalence and good quality exhaustive data is expensive and difficult to collect, a standard European system allows countries using data from regional registries to pool their data for studies and to exploit their differences by comparing them. European collaboration also allows for sharing of expertise and enables a joint approach to European public health questions.

International Clearinghouse of Birth Defects Monitoring Systems (ICBDMS)

The Malta Congenital Anomalies Register was accepted as a member of the ICBDMS in 2000, regularly transmitting aggregate data to the Clearinghouse Centre currently located in Rome. This data has been included in the organisation's Annual Reports and World Atlas of Birth Defects.

The ICBDMS is a non-governmental organisation in official relations with the World Health Organisation representing over 40 malformation monitoring programmes world-wide. Member programmes are actively engaged in the systematic collection and analysis of the data for the comprehensive monitoring of congenital malformations.

The organisation was established in 1974, at a meeting in Helsinki, Finland, where representatives of malformation monitoring systems in ten countries were present. The mission of the ICBDMS is to help local registries of congenital malformations to identify and to prevent birth defects and to serve as an early warning system to avoid the spread of an epidemic of congenital malformations. To accomplish this the ICBDMS has three main objectives:

- exchange of routine information in the prevalence of congenital malformations
- collaborative epidemiologic research
- expert consultation and assistance for existing monitoring systems to investigate outbreaks and for helping new monitoring systems get established.

THE STUDY POPULATION

The Malta Congenital Anomalies Register is population based and covers all births on the islands of Malta and Gozo. All infants/fetuses who are diagnosed or suspected of having a congenital anomaly until one year of age are included.

The small size and population of the islands (Area: 316 km^2, Population: 386,938 in 2002); the geographically well defined boundaries, absence of significant ethnic minority groups and illegality of termination of pregnancy make the islands ideal for epidemiological studies.

This report gives a detailed breakdown of all births diagnosed and registered with congenital anomaly in 2002. Denominator data of total livebirths and stillbirths have been taken from the 2002 Demographic Review of the Maltese Islands.

ASCERTAINMENT METHODS

To ensure as complete an ascertainment as possible, the Malta Congenital Anomalies Register makes use of active case finding and multiple sources of information. Cases of congenital anomalies diagnosed in the first few days of life in babies born at St. Luke's Hospital are recorded by staff from the registry who visit the postnatal wards daily. Gozo General Hospital and private hospitals (St. Philip's Hospital, St. James Sliema and Zabbar) can notify any cases of congenital anomalies using a standardised report form (Annex 1) or through the National Obstetric Systems Database. All paediatricians, paediatric surgeons and other medical doctors are encouraged to report any infants with congenital anomalies under their care to the registry. Other sources of information used in this report include: Echocardiography Lab., Genetics Clinic, St. Luke's Hospital Activity Analysis Register, National Mortality Register, Pathology Autopsy reports, National Cancer Register and the Hypothyroid screening programme. The process of data collection and processing is outlined in the following flowchart.

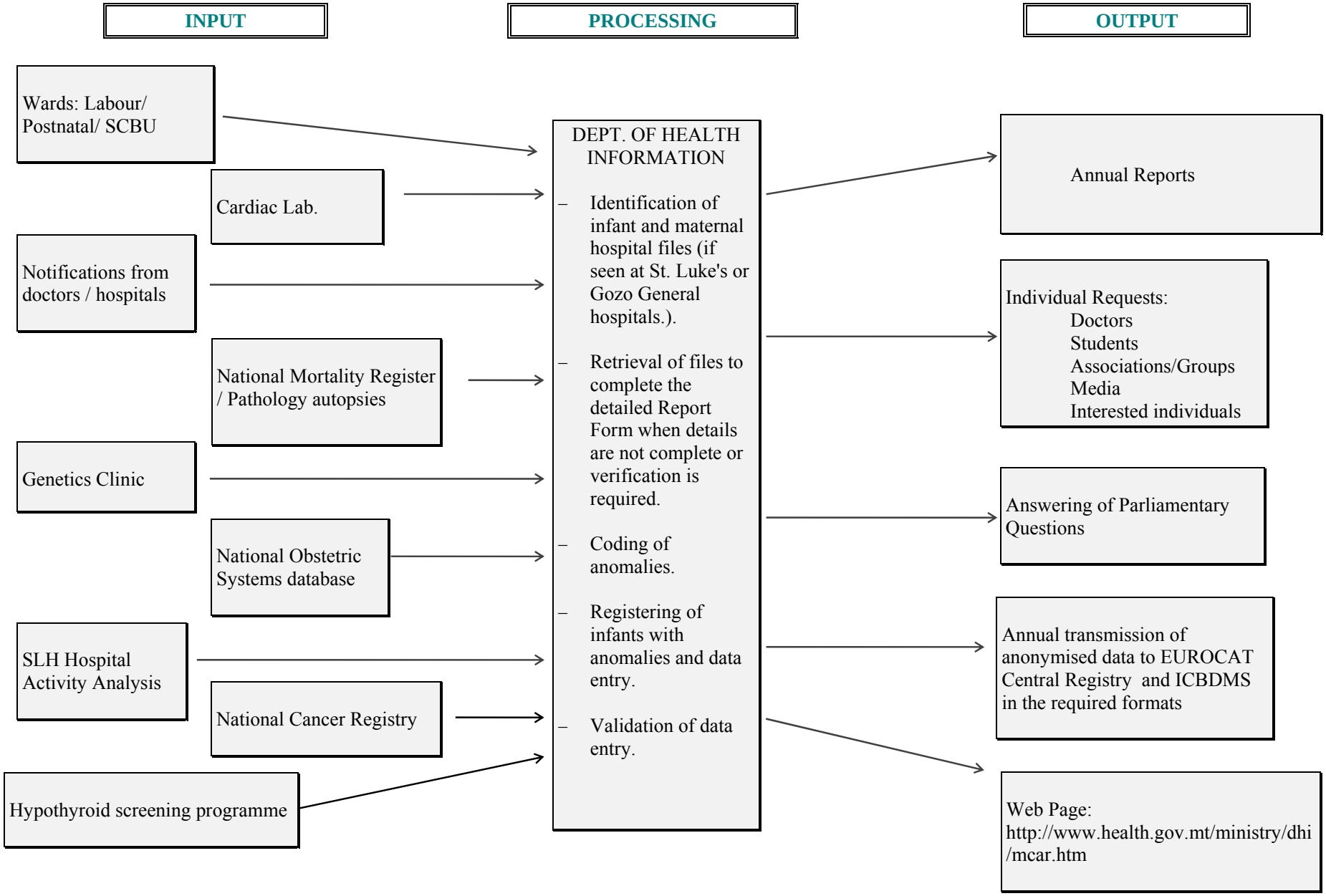
Every effort is made to record all cases of congenital anomalies and the database is continually updated as new information reaches the registry. It is however extremely difficult, if not impossible, for any registration system to claim complete case ascertainment or complete accuracy of information. First of all, neither ascertainment nor accuracy are absolute in a world of changing diagnostic methods and diagnostic definitions. Furthermore, depending on the malformation, some cases may escape the system.

Ascertainment of livebirths and fetal deaths

Livebirths with congenital anomalies are not always diagnosed at birth or in the early neonatal period, particularly certain cardiac anomalies, internal urogenital system anomalies and central nervous system anomalies among others. For this reason all anomalies diagnosed until one year of age are included in the register; this is in-keeping with EUROCAT guidelines.

Fetal deaths include all registered malformed cases of spontaneous fetal deaths or stillbirths of 20 weeks gestational age or more.

Flowchart showing data collection and management by the Malta Congenital Anomalies Registry



DEFINITIONS AND EXCLUSIONS

In this report the term 'congenital anomalies' refers to structural defects (congenital malformations, deformations, disruptions and dysplasias), chromosomal abnormalities, inborn errors of metabolism and hereditary diseases. This is the definition used also by EUROCAT for the registration of congenital anomalies.

Minor congenital anomalies are not normally included in congenital anomalies registers as they would markedly swell the incidence figures and dilute the significance of major anomalies. The precise line of demarcation between anomalies to be included and those to be excluded is not clear cut and may be influenced by subjective differences in interpretation. Minor anomalies do not in themselves have serious medical or cosmetic consequences for the child. Some may nevertheless be of certain significance, since they can be predictive of major underlying pathologies. There is increasing awareness of dysmorphic syndromes which present great diagnostic difficulties, not only in their precise identification, but also in their recognition which may be delayed for many months before they are detected.

EUROCAT applies a standard list of minor and commonly occurring anomalies for exclusion; this list is given in Annex 2. These conditions are not registered unless occurring in combination with other major anomalies.

It is necessary to exercise great care and attention to detail in distinguishing some of the anomalies listed for exclusion from those which are to be registered. Thus glandular and coronal hypospadias are excluded wheras penile and perineo-scrotal hypospadias are to be registered. Similarly, structural talipes has to be distinguished from postural talipes which is excluded. Birthmarks are registered as skin anomalies only when they exceed an area of $4cm^2$.

Minor anomalies such as single palmar crease, low set or dysmorphic ears, slanting palpebral fissures, high arched palate, micrognathia, and other dysmorphic features of the face and lips are not included in the register unless associated with other major defects. These anomalies are often difficult to define accurately and interpretation may be highly subjective especially for those who are not experienced in dysmorphology; nevertheless they are highly significant in syndrome identification. Also excluded from the register are hernias, varicocele, undescended testes, spina bifida occulta, small birthmarks and pre-auricular pits or tags.

CASE INFORMATION

For each baby with an anomaly, information about the child, the diagnosis, the pregnancy, the parents, their occupation and risk factors (assisted conception, illness before pregnancy, habitual and unusual exposures, drugs), diagnosis of malformations, and family history is recorded. Annex 3 shows the details recorded on the EUROCAT Report Form. Case records are updated when necessary (e.g. diagnosis of later discovered anomalies, change or precision in diagnosis, knowledge of an additional risk factor).

CODING OF CONGENITAL ANOMALIES

Prior to 1998, the Malta Congenital Anomalies Register coded anomalies using a six-digit EUROCAT code, which was a compatible expansion of the coding system of the British Paediatric Association Classification of diseases (BPA-9). The BPA-9 is itself a five-digit extension of the 9th revision of the International Classification of Diseases. Since 1998, anomalies have been coded using the Royal College of Paediatrics and Child Health (RCPCH) Classification of Diseases which is a paediatric adaptation of ICD-10 (International Classification of Diseases - 10th Edition).

Each anomaly is precisely and individually coded by a principal medical officer at the registry. As far as possible all case histories are reviewed to ensure that all details are included in the registration form. Where necessary babies' files are followed up to ensure completeness and accuracy of the database.

For reporting purposes, anomaly subgroups are defined. These subgroups make the interpretation of results more meaningful for the clinician and facilitate comparisons of prevalence rates between different centres. A list of anomaly subgroups based on EUROCAT guidelines is given in Annex 4.

CALCULATION OF PREVALENCE RATES AND STATISTICAL METHODS

Babies may be born with one or more anomalies. As some infants may have more than one anomaly, the number of anomalies do not add up to the number of infants.

In prevalence rate calculations, a baby/fetus with several anomalies is counted once within each class of anomaly. The number in different classes cannot be added to reach the total number of babies/fetuses. A baby is counted once only in any given prevalence rate for a specific anomaly or anomaly group.

$$\text{Total Birth Prevalence } * = \frac{\text{Number of live and still births registered with anomalies}}{\text{Total number of live and still births in that period}}$$

When counting anomalies, babies with more than one anomaly within a specific subgroup/ICD classification group are counted only once within that class. Babies with anomalies affecting more than one specific subgroup/classification group are counted once within each class. This method of counting anomalies is that adopted by EUROCAT Central Registry for reporting purposes.

The *total prevalence rate of a congenital anomaly or congenital anomaly subgroup* is expressed as the number of cases (babies/fetal deaths registered with that anomaly) divided by the total number of livebirths and fetal deaths in that period.

$$\text{Total prevalence rate of congenital anomalies} * = \frac{\text{Total number of cases registered in livebirths/fetal deaths with anomaly}}{\text{Total number of livebirths/fetal deaths in that period}}$$

The *livebirth prevalence rate of congenital anomalies* is expressed as the number of liveborn affected babies divided by the number of livebirths in that period.

$$\text{Livebirth prevalence rate } * = \frac{\text{Total number of anomalies registered in livebirths}}{\text{Total livebirths in that period}}$$

* Expressed per 10,000 births

Whenever comparison with prevalence rates of other countries is performed, the number of induced abortions due to prenatal diagnosis of congenital anomaly were included with the number of fetal deaths due to anomaly. This is not applicable to Malta as termination of pregnancy is illegal.

ANNUAL REPORT - 2002

This report includes congenital anomalies diagnosed and confirmed in infants/fetuses born during the period 1st January to 31st December 2002 in Malta and Gozo and which were registered at the Malta Congenital Anomalies Registry by December 2004. All major anomalies as defined by EUROCAT guidelines are registered. Minor anomalies are only included when they occur in combination with major defects; a list of minor anomalies is given in Annex 2. 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, counting of defects and calculation of prevalence rates is performed as detailed previously. For this reason the total number of anomalies registered within each anomaly group do not add up to the number of infants registered.

The Malta Congenital Anomalies Registry continually updates its database to include any new information, more accurate diagnoses etc which may be received even some years after infant's date of birth. For this reason figures may sometimes vary slightly due to this continuous effort to update the database, making it as accurate and complete as possible.

A description of the functioning of the Malta Congenital Anomalies Registry and the most recent updated summary statistics may be found on the Department of Health Information web page: http://www.health.gov.mt/ministry/dhi/mcar.htm.

INFANTS / FETUSES REGISTERED

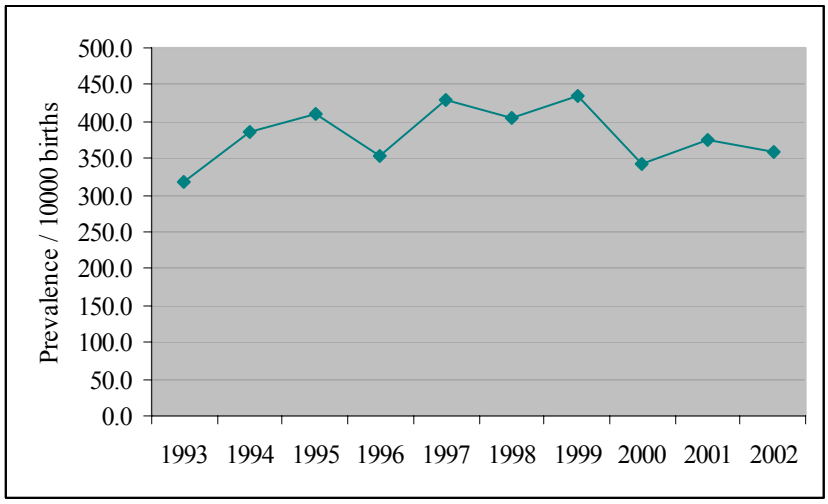
The total number of births (live and stillbirths) in Malta and Gozo during the year 2002 was 3826 (3805 livebirths and 21 fetal deaths). Of these, 137 were registered as having one or more major anomalies, giving an overall congenital anomaly birth prevalence rate of 358.1/10000 total births.

The table and figure below give the total birth prevalence rates of babies born with major congenital anomaly over the past 10 years.

Table 1 – Birth prevalence rates of infants/fetuses registered over the past 10 years

Year	Total Births (live and still births)	Births registered with anomaly	Prevalence / 10000 total births
1993	5172	164	317.1
1994	4863	188	386.6
1995	4633	190	410.1
1996	4978	176	353.6
1997	4864	209	429.7
1998	4511	183	405.7
1999	4339	189	435.6
2000	4272	146	341.8
2001	3883	146	376.0
2002	3826	137	358.1

Figure 1 – Birth prevalence rates of infants/fetuses registered 1993-2002



No significant trends are seen in the prevalence rates over these years.

TYPE OF BIRTH

Of the 137 registered infants/fetuses with congenital anomalies, 79 (58%) were male and 58 (42%) were female giving a male:female ratio of 1:0.7. A detailed breakdown is given in Table 2.

Table 2 - Infants / fetuses diagnosed with major congenital anomaly in 2002

	Sex	Total Births		With Congenital Anomalies	
		Live births*	Fetal Deaths**	Live births	Fetal Deaths
Malta	M	1840	14	72	1
	F	1677	5	54	3
Gozo	M	157	1	5	1
	F	131	1	1	0
Total		**3805**	**21**	**132**	**5**

* Source: Demographic Review of the Maltese Islands - 2002
** Source: National Mortality Register – Dept. of Health Information

Table 3 – Fetal and neonatal mortality rates

	All births	With Congenital Anomalies
Fetal deaths	21	5
Fetal Death Rate / 1000 livebirths	***5.5***	***36.5***
Neonatal deaths (livebirths dying before 28 days)	21	11
Neonatal death Rate / 1000 livebirths	***5.5***	***83.3***

Both the fetal and the neonatal death rates are significantly higher in babies born and diagnosed with congenital anomaly, making congenital anomalies a major cause of perinatal mortality.

FIRST 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.

For the majority of cases (66%), the first source of information came from the SLH obstetric and paediatric wards and cardiac lab., where registry staff members regularly go to collect information on neonates and echocardiography records respectively. The distribution of the major sources of information is given below.

Table 4 - First source of information for babies diagnosed with major congenital anomalies (2002)

First Source of information	Number	Percentage
Active collection from SLH Post natal wards and SCBU	65	47%
Cardiac lab records	25	18%
NOIS (National Obstetric Information Systems database)	12	9%
HAA (SLH Hospital Activity Analysis database)	6	4%
Genetic Clinic Records	0	0%
Death Register / Autopsy records	8	6%
Doctor / Midwife notification	8	6%
Others	13	9%

ISOLATED vs MULTIPLE ANOMALIES

The majority of babies registered in 2002 had isolated defects (69%), while 14% of infants / fetuses registered in 2002 had chromosomal anomalies and only 4% had recognised syndromes or sequences.

Table 5 - Distribution of infants / fetuses according to number of major anomalies (2002)

Anomalies	Number	Percentage
Isolated	94	69%
Multiple anomalies of same system	9	7%
Multiple anomalies of different systems	10	7%
Chromosomal Anomalies	19	14%
Recognised sequences / syndromes	5	4%

TIME OF FIRST DIAGNOSIS

55% of babies registered were diagnosed at birth. The great majority (82%) were diagnosed until one month of age, however a not insignificant 18% were diagnosed between 28 days and 1 year of age. This highlights the importance of continuing to register all anomalies diagnosed within the first year of life. The majority of defects diagnosed after 28 days of life were heart defects.

Table 6 - Distribution of infants by time of diagnosis

Time of Diagnosis	Number	Percentage
Prenatal	9	7%
At birth	75	55%
Birth to 7 days	21	15%
8 – 28 days	7	5%
29 days – 1 year	24	18%
Unknown	1	<1%

INFANTS BY GESTATIONAL AGE

In 2002, very few of the infants registered (8%) were 33 weeks gestation or less. The majority of cases (67%) were born between 38-41 weeks of gestation.

Table 7 - Distribution of infants registered by Gestational Age

Length of gestation (weeks)	Number of Infants	Percentage
<20	0	0%
20-21	0	0%
22-23	0	0%
24-25	0	0%
26-27	1	1%
28-29	2	1%
30-31	1	1%
32-33	7	5%
34-35	7	5%
36-37	21	15%
38-39	50	36%
40-41	42	31%
42+	6	4%

INFANTS BY BIRTH WEIGHT

The majority of babies registered in 2002 had birth weights between 2000g and 3999g. The average birth weight was 2965g. There was one baby over 5000g. A detail of the distribution of infants/ fetuses registered by birth weight is given below.

Table 8 - Distribution of babies delivered by birth weight

Birth weight (grams)	Infant Number	Percentage
<500	0	0%
500 – 749	2	1%
750 – 999	2	1%
1000 – 1249	0	0%
1250 – 1499	4	3%
1500 – 1749	3	2%
1750 – 1999	1	<1%
2000 – 2249	9	7%
2250 – 2499	8	6%
2500 – 2749	11	8%
2750 – 2999	22	16%
3000 – 3249	20	15%
3250 – 3499	24	18%
3500 – 3749	19	14%
3750 – 3999	5	4%
4000 – 4249	3	2%
4250 – 4499	2	1%
4500 +	1	<1%
Unknown	0	0%

MATERNAL AGE DISTRIBUTION

A very high rate of birth defects was recorded in the 40-44 year age group (137.5/1000births). The lowest maternal age group also had a high prevalence of babies with congenital anomaly – 42.4/1000 births. An overall prevalence of 53.3/1000 births with congenital anomaly was recorded in deliveries to mothers 35 years and over and a prevalence of 33.5/1000 births was recorded for mothers less than 35 years of age.

When considering only infants with non-chromosomal anomalies, the increased prevalence of birth defects in the older maternal age groups is less marked (Table 8, Figure 2).

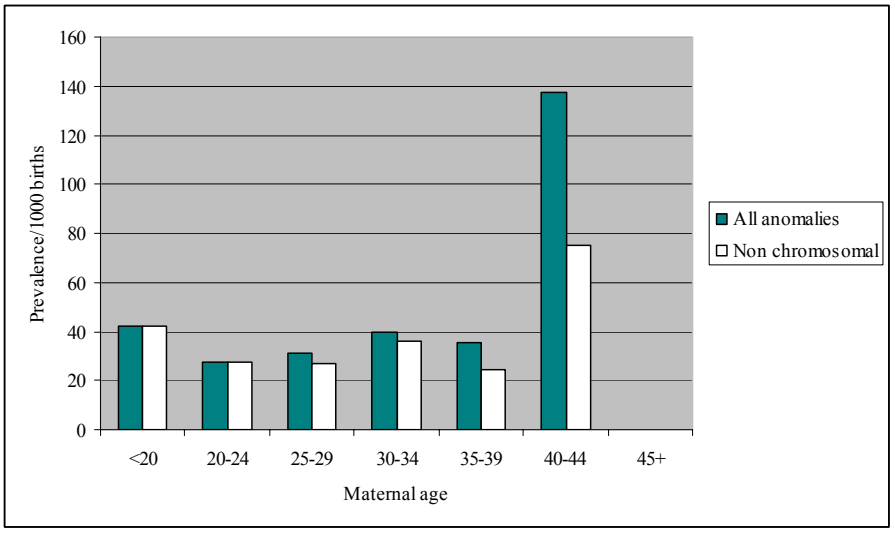
Table 9 - Prevalence of infants/fetuses with anomaly according to maternal age (2002)

Maternal age	Total Deliveries	Deliveries with anomaly	Prevalence /1000 births
<20	236	10	42.4
20-24	722	20	27.7
25-29	1534	48	31.3
30-34	884	35	39.6
35-39	369	13	35.2
40-44	80	11	137.5
45 and over	1	0	--

Table 10 - Prevalence of infants/fetuses with non-chromosomal anomalies according to maternal age (2002)

Maternal age	Total Deliveries	Deliveries with anomaly	Prevalence /1000 births
<20	236	10	42.4
20-24	722	20	27.7
25-29	1534	41	26.7
30-34	884	32	36.2
35-39	369	9	24.3
40-44	80	6	75.0
45 and over	1	0	--

Figure 2 - Maternal age distribution for all anomalies and for non-chromosomal anomalies (2002)



GEOGRAPHICAL DISTRIBUTION

The geographical break-down used in this analysis is that used by the National Statistics Office in the Demographic Review of the Maltese Islands - 2002. Details of this breakdown may be found in the latter publication.

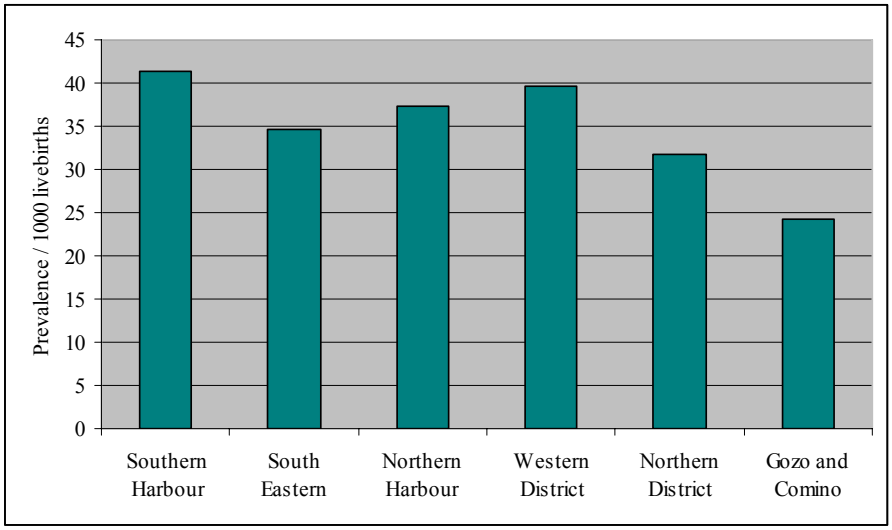
The highest prevalence of babies with birth defects in 2002 was registered for mothers residing in the Southern Harbour region (41.4 / 1000 births), while the lowest prevalence was recorded in Gozo (24.3 / 1000 births).

The differences in prevalence are however not statistically significant and as such may be due to chance.

Table 11 - Prevalence of infants/fetuses with anomaly according to geographical distribution (2002)

Locality	Total Livebirths	Babies with anomaly	Prevalence /1000
Southern Harbour	724	30	41.4
South Eastern	636	22	34.6
Northern Harbour	994	37	37.7
Western District	531	21	39.5
Northern District	632	20	31.6
Gozo and Comino	288	7	24.3
Total	3805	137	36.0

Figure 3 – Geographical distribution (2002)



DISTRIBUTION BY MONTH OF BIRTH

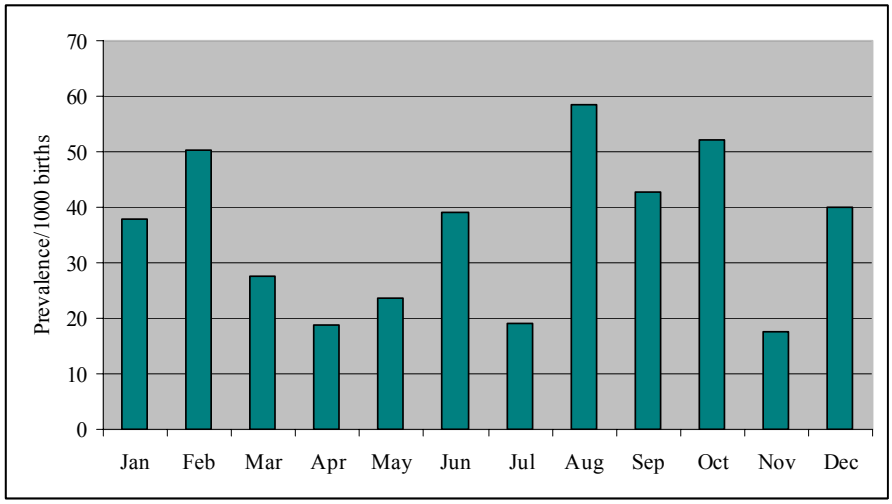
The highest prevalence of babies with birth defects in 2002 was registered in the months of August / September / October (58.5, 42.9, 52.0/1000 births respectively).

The lowest prevalence was recorded in November (17.4/1000 births). No significant variation in seasonal prevalence is observed.

Table 12 - Prevalence of infants/fetuses with anomaly according to month of birth (2002)

Month of birth	Total Births	Births with anomaly	Prevalence / 1000 births
January	318	12	37.7
February	298	15	50.3
March	289	8	27.7
April	319	6	18.8
May	295	7	23.7
June	306	12	39.2
July	312	6	19.2
August	325	19	58.5
September	350	15	42.9
October	346	18	52.0
November	344	6	17.4
December	324	13	40.1

Figure 4 - Distribution by month of birth (2002)



ANALYSIS OF INFANTS/FETUSES BY ANOMALY SYSTEM INVOLVED

When analysing and counting infants / fetuses by anomaly systems, babies with more than one anomaly within a specific subgroup/ICD classification group are counted only once within that class. Babies with anomalies affecting more than one specific subgroup/classification group are counted once within each class. For this reason the total number of babies in the anomaly groups is grater than the actual number of babies registered. This method of counting anomalies is that adopted by EUROCAT Central Registry for reporting purposes.

The most commonly encountered group of anomalies in the year 2002 was Congenital Heart Defects. These accounted for 33% of all anomalies registered. The next most frequently encountered group of anomalies were limb defects and defects of the external genital system (mainly hypospadias), which together accounted for 30% of all anomalies registered in 2002.

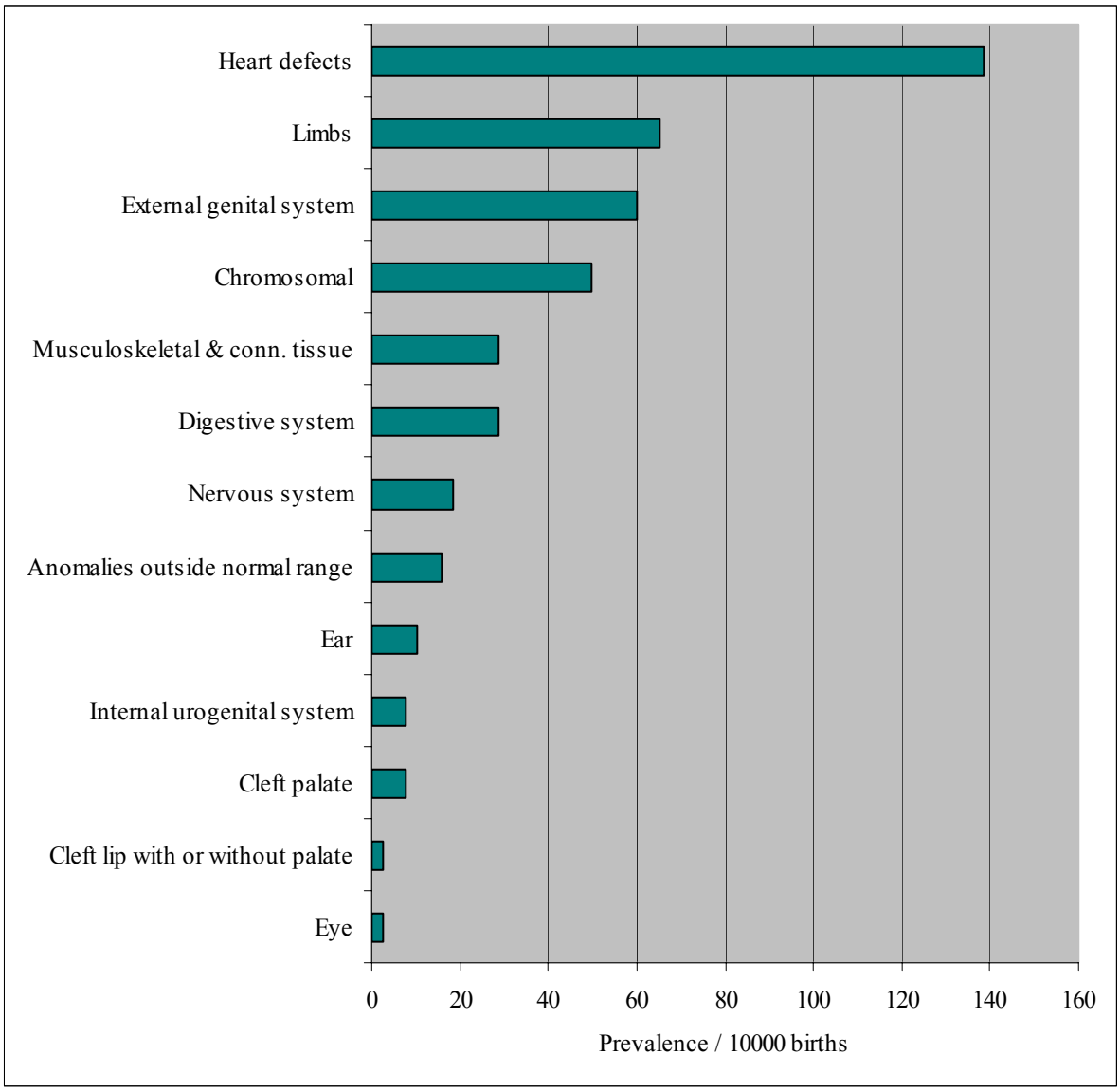
The distribution of anomalies by system affected is shown in Table 13 and Figure 5. More detailed breakdowns are found in TABLE A. The list of definitions for the various anomaly groups is given in Annex 4.

Table 13 - Distribution of infants / fetuses by anomaly system affected (2002)

Systems affected	Live Births (LB)	Fetal Deaths (FD)	LB + FD	Prevalence LB+FD/10000
Nervous system	5	2	7	18.30
Eye	1	0	1	2.61
Ear	4	0	4	10.45
Congenital heart disease	53	0	53	138.53
Cleft lip with or without palate	1	0	1	2.61
Cleft palate	3	0	3	7.84
Digestive system	11	0	11	28.75
Internal urogenital system-ovaries uterus and renal system	2	1	3	7.84
External genital system	23	0	23	60.12
Limb	25	0	25	65.34
Musculoskeletal and connective tissue	10	1	11	28.75
Chromosomal	19	0	19	49.66
Anomalies outside normal range	5	1	6	15.68

LB – Live Births, FD – Fetal deaths / Still Births from 20 weeks gestation

Figure 5 - Distribution of anomalies by system and in order of frequency (2002)



TABLES

TABLE A -
PREVALENCE OF ANOMALIES PER 10,000 BIRTHS FOR 2002

(Definitions for the anomaly groups are given in Annex 4)

Anomaly	LB* (n)	FD**(n)	LB+FD(n)	LB+FD/10000
Nervous system	**5**	**2**	**7**	**18.30**
Neural Tube Defects	3	2	5	13.07
Anencephalus and similar	0	2	2	5.23
Encephalocele	1	0	1	2.61
Spina Bifida	2	0	2	5.23
Hydrocephaly	0	0	0	0
Microcephaly	1	0	1	2.61
Arhinencephaly/holoprosencephaly	0	0	0	0
Eye	**1**	**0**	**1**	**2.61**
Anophthalmos/microthalmos	0	0	0	0
Anophthalmos	0	0	0	0
Microthalmos	0	0	0	0
Cataract	0	0	0	0
Ear	**4**	**0**	**4**	**10.45**
Anotia/microtia	0	0	0	0
Anotia	0	0	0	0
Microtia	0	0	0	0
Congenital heart disease	**53**	**0**	**53**	**138.53**
Anomalies of cardiac chambers and connections	1	0	1	2.61
Common arterial truncus	0	0	0	0
Transposition of great vessels (complete)	1	0	1	2.61
Single ventrical	0	0	0	0
Malformations of cardiac septa	44	0	44	115.00
Ventricular septal defect	17	0	17	44.43
Atrial septal defect	27	0	27	70.57
Atrioventricular septal defect	3	0	3	7.84
Tetralogy of Fallot	0	0	0	0
Malformations of valves	10	0	10	26.14
Tricuspid atresia and stenosis	0	0	0	0
Ebstein's anomaly	0	0	0	0
Aortic valve atresia/stenosis	1	0	1	2.61
Hypoplastic left heart	2	0	2	5.23
Malformations of the great arteries and veins	12	0	12	31.36
Coarctation of aorta	2	0	2	5.23
Cleft lip with or without palate	**1**	**0**	**1**	**2.61**
Cleft palate	**3**	**0**	**3**	**7.84**
Digestive system	**11**	**0**	**11**	**28.75**
Tracheo-oesophagal fistula-Oesophageal atresia and stenosis	1	0	1	2.61
Congenital absence, atresia and/or stenosis of small intestine	1	0	1	2.61
Congenital absence, atresia and/or stenosis of the duodenal	0	0	0	0
Congenital absence, atresia and/or stenosis of other specified parts of small intestine	1	0	1	2.61
Ano-rectal atresia and stenosis	4	0	4	10.45

Anomaly	LB* (n)	FD**(n)	LB+FD(n)	LB+FD/10000
Internal urogenital system-ovaries uterus and renal system	**2**	**1**	**3**	**7.84**
Bilateral renal agenesis	1	0	1	2.61
Cystic kidney disease	0	0	0	0
Congenital hydronephrosis	1	0	1	2.61
Bladder extrophy	0	0	0	0
External genital system	**23**	**0**	**23**	**60.12**
Hypospadias	8	0	8	20.91
Indeterminate sex	0	0	0	0
Limb	**25**	**0**	**25**	**65.34**
Limb reduction	2	0	2	5.23
Upper limb reduction	2	0	2	5.23
Complete absence of upper limb	0	0	0	0
Absence of upper arm and forearm with hand present	0	0	0	0
Absence of both forearm and hand	0	0	0	0
Absence of hand and fingers	0	0	0	0
Longitudinal reduction defect/shortening of arm	2	0	2	5.23
Lower limb reduction	0	0	0	0
Complete absence of lower limb	0	0	0	0
Absence of thigh and lower leg with foot present	0	0	0	0
Absence of both lower leg and foot	0	0	0	0
Absence of foot and toe	0	0	0	0
Longitudinal reduction defect/shortening of leg	0	0	0	0
Polydactyly	8	0	8	20.91
Syndactyly	5	0	5	13.07
Musculoskeletal and connective tissue	**10**	**1**	**11**	**28.75**
Choanal atresia	1	0	1	2.61
Craniosynostosis	1	0	1	2.61
Pierre Robin Syndrome	1	0	1	2.61
Mandibulofacial dystosis (Treacher-Collins and Franceschetti)	0	0	0	0
Oculomandibular dysostosis (Hallerman-Streiff	0	0	0	0
Goldenhar's Syndrome	0	0	0	0
Chondodystrophies and osteodystrophies	1	1	2	5.23
Diaphraghmatic hernia	4	0	4	10.45
Omphalocele	1	0	1	2.61
Gastroschisis	0	0	0	0
Prune Belly Syndrome	0	0	0	0
Chromosomal	**19**	**0**	**19**	**49.66**
Down Syndrome	15	0	15	39.21
Patau syndrome (trisomy 13)	0	0	0	0
Edward syndrome (trisomy 18)	2	0	2	5.23
Other trisomies and partial trisomies of autosomes	1	0	1	2.61
Monosomies and deletions from the autosomes	0	0	0	0
Turner's syndrome	0	0	0	0
Klinefelters syndrome	0	0	0	0
Anomalies outside normal range	**5**	**1**	**6**	**15.68**

*LB - Live Births;

**FD - Fetal Deaths/ Stillbirths from 20 weeks gestation

TABLE B - TIME TRENDS: CASES AND PREVALENCE PER 10,000 BIRTHS FOR SELECTED ANOMALY GROUPS 1993-2002

(Definitions for the various anomaly groups are given in Annex 4)

Cases and prevalence per 10,000 births for selected anomaly groups 1993-2002											
	1993	**1994**	**1995**	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**93-02**
Population:	5172	4863	4633	4978	4864	4511	4339	4272	3883	3826	45340
Totals											
Total cases:	387	456	454	478	509	489	480	427	377	362	4419
Live Births:	372	430	452	453	499	484	428	422	372	352	4264
Fetal Deaths:	15	26	2	25	10	5	52	5	5	10	155
Prevalence:	748.26	937.69	979.93	960.22	1046.46	1084.02	1106.25	999.53	970.9	946.16	974.64
Nervous system											
Total cases:	12	18	16	10	10	15	11	12	6	7	117
Live Births:	9	12	16	8	10	14	7	11	5	5	97
Fetal Deaths:	3	6	0	2	0	1	4	1	1	2	20
Prevalence:	23.2	37.01	34.53	20.09	20.56	33.25	25.35	28.09	15.45	18.3	25.81
Neural Tube Defects											
Total cases:	6	7	7	7	5	6	7	3	3	5	56
Live Births:	5	5	7	5	5	5	4	3	2	3	44
Fetal Deaths:	1	2	0	2	0	1	3	0	1	2	12
Prevalence:	11.6	14.39	15.11	14.06	10.28	13.3	16.13	7.02	7.73	13.07	12.35
Anencephalus and similar											
Total cases:	1	3	1	3	1	3	3	0	0	2	17
Live Births:	1	1	1	1	1	2	1	0	0	0	8
Fetal Deaths:	0	2	0	2	0	1	2	0	0	2	9
Prevalence:	1.93	6.17	2.16	6.03	2.06	6.65	6.91	0	0	5.23	3.75
Encephalocele											
Total cases:	1	1	0	1	2	0	1	2	1	1	10
Live Births:	1	1	0	1	2	0	1	2	0	1	9
Fetal Deaths:	0	0	0	0	0	0	0	0	1	0	1
Prevalence:	1.93	2.06	0	2.01	4.11	0	2.3	4.68	2.58	2.61	2.21
Spina Bifida											
Total cases:	4	3	6	3	2	3	3	1	2	2	29
Live Births:	3	3	6	3	2	3	2	1	2	2	27
Fetal Deaths:	1	0	0	0	0	0	1	0	0	0	2
Prevalence:	7.73	6.17	12.95	6.03	4.11	6.65	6.91	2.34	5.15	5.23	6.4
Hydrocephaly											
Total cases:	1	6	6	1	0	6	0	3	1	0	24
Live Births:	1	4	6	1	0	6	0	2	1	0	21
Fetal Deaths:	0	2	0	0	0	0	0	1	0	0	3
Prevalence:	1.93	12.34	12.95	2.01	0	13.3	0	7.02	2.58	0	5.29

Cases and prevalence per 10,000 births for selected anomaly groups 1993-2002											
	1993	**1994**	**1995**	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**93-02**
Population:	5172	4863	4633	4978	4864	4511	4339	4272	3883	3826	45340
Microcephaly											
Total cases:	2	3	2	2	1	0	2	2	1	1	16
Live Births:	1	2	2	2	1	0	1	2	1	1	13
Fetal Deaths:	1	1	0	0	0	0	1	0	0	0	3
Prevalence:	3.87	6.17	4.32	4.02	2.06	0	4.61	4.68	2.58	2.61	3.53
Arhinencephaly/holoprosencephaly											
Total cases:	0	1	0	0	1	1	1	0	0	0	4
Live Births:	0	0	0	0	1	1	1	0	0	0	3
Fetal Deaths:	0	1	0	0	0	0	0	0	0	0	1
Prevalence:	0	2.06	0	0	2.06	2.22	2.3	0	0	0	0.88
Eye											
Total cases:	3	2	3	3	3	2	2	4	2	1	25
Live Births:	3	1	3	3	3	2	2	4	2	1	24
Fetal Deaths:	0	1	0	0	0	0	0	0	0	0	1
Prevalence:	5.8	4.11	6.48	6.03	6.17	4.43	4.61	9.36	5.15	2.61	5.51
Anophthalmos/microthalmos											
Total cases:	0	1	0	1	2	0	1	1	0	0	6
Live Births:	0	0	0	1	2	0	1	1	0	0	5
Fetal Deaths:	0	1	0	0	0	0	0	0	0	0	1
Prevalence:	0	2.06	0	2.01	4.11	0	2.3	2.34	0	0	1.32
Anophthalmos											
Total cases:	0	1	0	0	0	0	0	0	0	0	1
Live Births:	0	0	0	0	0	0	0	0	0	0	0
Fetal Deaths:	0	1	0	0	0	0	0	0	0	0	1
Prevalence:	0	2.06	0	0	0	0	0	0	0	0	0.22
Microthalmos											
Total cases:	0	0	0	1	2	0	1	1	0	0	5
Live Births:	0	0	0	1	2	0	1	1	0	0	5
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	2.01	4.11	0	2.3	2.34	0	0	1.1
Cataract											
Total cases:	0	0	1	0	0	1	0	0	0	0	2
Live Births:	0	0	1	0	0	1	0	0	0	0	2
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	2.16	0	0	2.22	0	0	0	0	0.44
Ear											
Total cases:	2	5	9	6	7	4	3	6	4	4	50
Live Births:	2	5	9	6	5	4	2	6	4	4	47
Fetal Deaths:	0	0	0	0	2	0	1	0	0	0	3
Prevalence:	3.87	10.28	19.43	12.05	14.39	8.87	6.91	14.04	10.3	10.45	11.03

Cases and prevalence per 10,000 births for selected anomaly groups 1993-2002											
	1993	**1994**	**1995**	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**93-02**
Population:	5172	4863	4633	4978	4864	4511	4339	4272	3883	3826	45340
Anotia/microtia											
Total cases:	0	0	0	0	0	0	0	0	0	0	0
Live Births:	0	0	0	0	0	0	0	0	0	0	0
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	0	0	0	0	0	0	0	0
Anotia											
Total cases:	0	0	0	0	0	0	0	0	0	0	0
Live Births:	0	0	0	0	0	0	0	0	0	0	0
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	0	0	0	0	0	0	0	0
Microtia											
Total cases:	0	0	0	0	0	0	0	0	0	0	0
Live Births:	0	0	0	0	0	0	0	0	0	0	0
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	0	0	0	0	0	0	0	0
Congenital heart disease											
Total cases:	59	73	74	90	93	95	84	79	64	53	764
Live Births:	58	72	74	86	93	95	80	79	64	53	754
Fetal Deaths:	1	1	0	4	0	0	4	0	0	0	10
Prevalence:	114.08	150.11	159.72	180.8	191.2	210.6	193.59	184.93	164.82	138.53	168.5
Anomalies of cardiac chambers and connections											
Total cases:	2	3	1	4	3	4	2	4	3	1	27
Live Births:	2	3	1	3	3	4	2	4	3	1	26
Fetal Deaths:	0	0	0	1	0	0	0	0	0	0	1
Prevalence:	3.87	6.17	2.16	8.04	6.17	8.87	4.61	9.36	7.73	2.61	5.96
Common arterial truncus											
Total cases:	0	1	0	1	0	0	0	0	0	0	2
Live Births:	0	1	0	0	0	0	0	0	0	0	1
Fetal Deaths:	0	0	0	1	0	0	0	0	0	0	1
Prevalence:	0	2.06	0	2.01	0	0	0	0	0	0	0.44
Transposition of great vessels (complete)											
Total cases:	2	1	1	3	1	4	2	4	2	1	21
Live Births:	2	1	1	3	1	4	2	4	2	1	21
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	3.87	2.06	2.16	6.03	2.06	8.87	4.61	9.36	5.15	2.61	4.63
Single ventricle											
Total cases:	0	0	0	0	1	1	1	0	1	0	4
Live Births:	0	0	0	0	1	1	1	0	1	0	4
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	0	2.06	2.22	2.3	0	2.58	0	0.88

Cases and prevalence per 10,000 births for selected anomaly groups 1993-2002											
	1993	**1994**	**1995**	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**93-02**
Population:	5172	4863	4633	4978	4864	4511	4339	4272	3883	3826	45340
Malformations of cardiac septa											
Total cases:	42	58	62	72	78	75	63	64	49	44	607
Live Births:	42	57	62	70	78	75	59	64	49	44	600
Fetal Deaths:	0	1	0	2	0	0	4	0	0	0	7
Prevalence:	81.21	119.27	133.82	144.64	160.36	166.26	145.19	149.81	126.19	115	133.88
Ventricular septal defect											
Total cases:	23	26	35	35	37	32	31	37	26	17	299
Live Births:	23	26	35	34	37	32	27	37	26	17	294
Fetal Deaths:	0	0	0	1	0	0	4	0	0	0	5
Prevalence:	44.47	53.46	75.55	70.31	76.07	70.94	71.45	86.61	66.96	44.43	65.95
Atrial septal defect											
Total cases:	19	38	26	44	45	52	38	32	35	27	356
Live Births:	19	37	26	42	45	52	36	32	35	27	351
Fetal Deaths:	0	1	0	2	0	0	2	0	0	0	5
Prevalence:	36.74	78.14	56.12	88.39	92.52	115.27	87.58	74.91	90.14	70.57	78.52
Atrioventricular septal defect											
Total cases:	2	2	2	7	1	0	0	3	1	3	21
Live Births:	2	2	2	7	1	0	0	3	1	3	21
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	3.87	4.11	4.32	14.06	2.06	0	0	7.02	2.58	7.84	4.63
Tetralogy of Fallot											
Total cases:	2	0	2	1	4	2	2	3	1	0	17
Live Births:	2	0	2	1	4	2	2	3	1	0	17
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	3.87	0	4.32	2.01	8.22	4.43	4.61	7.02	2.58	0	3.75
Malformations of valves											
Total cases:	15	16	13	8	11	16	20	13	13	10	135
Live Births:	14	16	13	8	11	16	20	13	13	10	134
Fetal Deaths:	1	0	0	0	0	0	0	0	0	0	1
Prevalence:	29	32.9	28.06	16.07	22.62	35.47	46.09	30.43	33.48	26.14	29.78
Tricuspid atresia and stenosis											
Total cases:	1	0	0	0	0	2	0	1	0	0	4
Live Births:	1	0	0	0	0	2	0	1	0	0	4
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	1.93	0	0	0	0	4.43	0	2.34	0	0	0.88
Ebstein's anomaly											
Total cases:	0	0	1	1	1	1	0	0	0	0	4
Live Births:	0	0	1	1	1	1	0	0	0	0	4
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	2.16	2.01	2.06	2.22	0	0	0	0	0.88

Cases and prevalence per 10,000 births for selected anomaly groups 1993-2002											
	1993	**1994**	**1995**	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**93-02**
Population:	5172	4863	4633	4978	4864	4511	4339	4272	3883	3826	45340
Aortic valve atresia/stenosis											
Total cases:	2	0	2	1	1	2	1	3	1	1	14
Live Births:	2	0	2	1	1	2	1	3	1	1	14
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	3.87	0	4.32	2.01	2.06	4.43	2.3	7.02	2.58	2.61	3.09
Hypoplastic left heart											
Total cases:	1	0	2	0	0	0	2	0	2	2	9
Live Births:	0	0	2	0	0	0	2	0	2	2	8
Fetal Deaths:	1	0	0	0	0	0	0	0	0	0	1
Prevalence:	1.93	0	4.32	0	0	0	4.61	0	5.15	5.23	1.99
Malformations of the great arteries and veins											
Total cases:	7	12	3	16	12	16	13	10	16	12	117
Live Births:	7	12	3	16	12	16	13	10	16	12	117
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	13.53	24.68	6.48	32.14	24.67	35.47	29.96	23.41	41.21	31.36	25.81
Coarctation of aorta											
Total cases:	2	6	0	4	3	2	1	3	3	2	26
Live Births:	2	6	0	4	3	2	1	3	3	2	26
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	3.87	12.34	0	8.04	6.17	4.43	2.3	7.02	7.73	5.23	5.73
Cleft lip with or without palate											
Total cases:	5	5	5	2	4	3	6	7	4	1	42
Live Births:	5	5	5	1	4	3	5	7	4	1	40
Fetal Deaths:	0	0	0	1	0	0	1	0	0	0	2
Prevalence:	9.67	10.28	10.79	4.02	8.22	6.65	13.83	16.39	10.3	2.61	9.26
Cleft palate											
Total cases:	8	7	7	7	7	5	11	5	2	3	62
Live Births:	8	7	7	7	7	5	10	5	2	3	61
Fetal Deaths:	0	0	0	0	0	0	1	0	0	0	1
Prevalence:	15.47	14.39	15.11	14.06	14.39	11.08	25.35	11.7	5.15	7.84	13.67
Digestive system											
Total cases:	6	13	12	11	18	4	12	9	11	11	107
Live Births:	6	13	12	11	17	4	11	9	11	11	105
Fetal Deaths:	0	0	0	0	1	0	1	0	0	0	2
Prevalence:	11.6	26.73	25.9	22.1	37.01	8.87	27.66	21.07	28.33	28.75	23.6
Tracheo-oesophagal fistula-Oesophageal atresia and stenosis											
Total cases:	2	2	0	1	1	1	1	2	0	1	11
Live Births:	2	2	0	1	1	1	1	2	0	1	11
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	3.87	4.11	0	2.01	2.06	2.22	2.3	4.68	0	2.61	2.43

Cases and prevalence per 10,000 births for selected anomaly groups 1993-2002											
	1993	**1994**	**1995**	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**93-02**
Population:	5172	4863	4633	4978	4864	4511	4339	4272	3883	3826	45340
Congenital absence, atresia and/or stenosis of the small intestine											
Total cases:	0	0	1	1	1	0	2	0	2	1	8
Live Births:	0	0	1	1	1	0	1	0	2	1	7
Fetal Deaths:	0	0	0	0	0	0	1	0	0	0	1
Prevalence:	0	0	2.16	2.01	2.06	0	4.61	0	5.15	2.61	1.76
Congenital absence, atresia and/or stenosis of the duodenal											
Total cases:	0	0	0	1	1	0	1	0	0	0	3
Live Births:	0	0	0	1	1	0	1	0	0	0	3
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	2.01	2.06	0	2.3	0	0	0	0.66
Congenital absence, atresia and/or stenosis of other specified parts of small intestine											
Total cases:	0	0	1	0	0	0	1	0	1	1	4
Live Births:	0	0	1	0	0	0	0	0	1	1	3
Fetal Deaths:	0	0	0	0	0	0	1	0	0	0	1
Prevalence:	0	0	2.16	0	0	0	2.3	0	2.58	2.61	0.88
Ano-rectal atresia and stenosis											
Total cases:	2	0	3	4	1	2	2	0	3	4	21
Live Births:	2	0	3	4	0	2	2	0	3	4	20
Fetal Deaths:	0	0	0	0	1	0	0	0	0	0	1
Prevalence:	3.87	0	6.48	8.04	2.06	4.43	4.61	0	7.73	10.45	4.63
Internal urogenital system-ovaries uterus and renal system											
Total cases:	7	21	11	10	14	9	8	6	6	3	95
Live Births:	6	21	11	10	14	9	6	6	6	2	91
Fetal Deaths:	1	0	0	0	0	0	2	0	0	1	4
Prevalence:	13.53	43.18	23.74	20.09	28.78	19.95	18.44	14.04	15.45	7.84	20.95
Bilateral renal agenesis											
Total cases:	0	1	1	0	1	1	0	0	0	1	5
Live Births:	0	1	1	0	1	1	0	0	0	1	5
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	2.06	2.16	0	2.06	2.22	0	0	0	2.61	1.1
Cystic kidney disease											
Total cases:	1	3	3	1	2	3	1	2	0	0	16
Live Births:	1	3	3	1	2	3	0	2	0	0	15
Fetal Deaths:	0	0	0	0	0	0	1	0	0	0	1
Prevalence:	1.93	6.17	6.48	2.01	4.11	6.65	2.3	4.68	0	0	3.53
Congenital hydronephrosis											
Total cases:	3	8	5	3	5	2	3	3	2	1	35
Live Births:	3	8	5	3	5	2	3	3	2	1	35
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	5.8	16.45	10.79	6.03	10.28	4.43	6.91	7.02	5.15	2.61	7.72

Cases and prevalence per 10,000 births for selected anomaly groups 1993-2002											
	1993	**1994**	**1995**	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**93-02**
Population:	5172	4863	4633	4978	4864	4511	4339	4272	3883	3826	45340
Bladder extrophy											
Total cases:	0	0	0	0	0	0	0	0	0	0	0
Live Births:	0	0	0	0	0	0	0	0	0	0	0
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	0	0	0	0	0	0	0	0
External genital system											
Total cases:	17	9	8	8	15	12	23	11	23	23	149
Live Births:	17	9	8	8	14	12	22	11	23	23	147
Fetal Deaths:	0	0	0	0	1	0	1	0	0	0	2
Prevalence:	32.87	18.51	17.27	16.07	30.84	26.6	53.01	25.75	59.23	60.12	32.86
Hypospadias											
Total cases:	11	1	4	4	9	8	15	7	11	8	78
Live Births:	11	1	4	4	9	8	14	7	11	8	77
Fetal Deaths:	0	0	0	0	0	0	1	0	0	0	1
Prevalence:	21.27	2.06	8.63	8.04	18.5	17.73	34.57	16.39	28.33	20.91	17.2
Indeterminate sex											
Total cases:	1	1	0	0	1	0	1	1	0	0	5
Live Births:	1	1	0	0	0	0	1	1	0	0	4
Fetal Deaths:	0	0	0	0	1	0	0	0	0	0	1
Prevalence:	1.93	2.06	0	0	2.06	0	2.3	2.34	0	0	1.1
Limb											
Total cases:	21	24	39	17	26	25	24	19	16	25	236
Live Births:	19	23	39	15	25	25	21	18	16	25	226
Fetal Deaths:	2	1	0	2	1	0	3	1	0	0	10
Prevalence:	40.6	49.35	84.18	34.15	53.45	55.42	55.31	44.48	41.21	65.34	52.05
Limb reduction											
Total cases:	3	4	4	1	2	2	2	5	4	2	29
Live Births:	3	4	4	1	2	2	1	5	4	2	28
Fetal Deaths:	0	0	0	0	0	0	1	0	0	0	1
Prevalence:	5.8	8.23	8.63	2.01	4.11	4.43	4.61	11.7	10.3	5.23	6.4
Upper limb reduction											
Total cases:	1	4	3	0	1	2	2	3	2	2	20
Live Births:	1	4	3	0	1	2	1	3	2	2	19
Fetal Deaths:	0	0	0	0	0	0	1	0	0	0	1
Prevalence:	1.93	8.23	6.48	0	2.06	4.43	4.61	7.02	5.15	5.23	4.41
Complete absence of upper limb											
Total cases:	0	0	0	0	0	0	0	0	0	0	0
Live Births:	0	0	0	0	0	0	0	0	0	0	0
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	0	0	0	0	0	0	0	0

Cases and prevalence per 10,000 births for selected anomaly groups 1993-2002											
	1993	**1994**	**1995**	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**93-02**
Population:	5172	4863	4633	4978	4864	4511	4339	4272	3883	3826	45340
Absence of upper arm and forearm with hand present											
Total cases:	0	0	0	0	0	0	0	0	0	0	0
Live Births:	0	0	0	0	0	0	0	0	0	0	0
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	0	0	0	0	0	0	0	0
Absence of both forearm and hand											
Total cases:	0	1	0	0	0	0	0	0	2	0	3
Live Births:	0	1	0	0	0	0	0	0	2	0	3
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	2.06	0	0	0	0	0	0	5.15	0	0.66
Absence of hand and fingers											
Total cases:	1	2	3	0	1	2	1	3	0	0	13
Live Births:	1	2	3	0	1	2	0	3	0	0	12
Fetal Deaths:	0	0	0	0	0	0	1	0	0	0	1
Prevalence:	1.93	4.11	6.48	0	2.06	4.43	2.3	7.02	0	0	2.87
Longitudinal reduction defect/shortening of arm											
Total cases:	0	2	0	0	0	0	1	0	0	2	5
Live Births:	0	2	0	0	0	0	1	0	0	2	5
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	4.11	0	0	0	0	2.3	0	0	5.23	1.1
Lower limb reduction											
Total cases:	2	0	2	1	1	1	0	2	2	0	11
Live Births:	2	0	2	1	1	1	0	2	2	0	11
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	3.87	0	4.32	2.01	2.06	2.22	0	4.68	5.15	0	2.43
Complete absence of lower limb											
Total cases:	0	0	0	0	0	0	0	0	0	0	0
Live Births:	0	0	0	0	0	0	0	0	0	0	0
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	0	0	0	0	0	0	0	0
Absence of thigh and lower leg with foot present											
Total cases:	0	0	0	0	0	0	0	0	0	0	0
Live Births:	0	0	0	0	0	0	0	0	0	0	0
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	0	0	0	0	0	0	0	0
Absence of both lower leg and foot											
Total cases:	0	0	0	0	0	0	0	0	0	0	0
Live Births:	0	0	0	0	0	0	0	0	0	0	0
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	0	0	0	0	0	0	0	0

Cases and prevalence per 10,000 births for selected anomaly groups 1993-2002											
	1993	**1994**	**1995**	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**93-02**
Population:	5172	4863	4633	4978	4864	4511	4339	4272	3883	3826	45340
Absence of foot and toe											
Total cases:	2	0	1	0	1	1	0	2	2	0	9
Live Births:	2	0	1	0	1	1	0	2	2	0	9
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	3.87	0	2.16	0	2.06	2.22	0	4.68	5.15	0	1.99
Longitudinal reduction defect/shortening of leg											
Total cases:	0	0	1	1	0	0	0	0	0	0	2
Live Births:	0	0	1	1	0	0	0	0	0	0	2
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	2.16	2.01	0	0	0	0	0	0	0.44
Polydactyly											
Total cases:	8	6	7	6	8	10	8	5	7	8	73
Live Births:	6	5	7	4	8	10	7	5	7	8	67
Fetal Deaths:	2	1	0	2	0	0	1	0	0	0	6
Prevalence:	15.47	12.34	15.11	12.05	16.45	22.17	18.44	11.7	18.03	20.91	16.1
Syndactyly											
Total cases:	5	4	5	2	5	7	9	3	2	5	47
Live Births:	5	4	5	2	5	7	6	3	2	5	44
Fetal Deaths:	0	0	0	0	0	0	3	0	0	0	3
Prevalence:	9.67	8.23	10.79	4.02	10.28	15.52	20.74	7.02	5.15	13.07	10.37
Musculoskeletal and connective tissue											
Total cases:	15	18	14	22	17	11	13	14	12	11	147
Live Births:	14	16	13	20	15	11	12	13	11	10	135
Fetal Deaths:	1	2	1	2	2	0	1	1	1	1	12
Prevalence:	29	37.01	30.22	44.19	34.95	24.38	29.96	32.77	30.9	28.75	32.42
Choanal atresia											
Total cases:	1	1	0	0	3	0	0	0	1	1	7
Live Births:	1	1	0	0	3	0	0	0	1	1	7
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	1.93	2.06	0	0	6.17	0	0	0	2.58	2.61	1.54
Craniosynostosis											
Total cases:	0	0	0	3	3	1	0	0	0	1	8
Live Births:	0	0	0	2	3	1	0	0	0	1	7
Fetal Deaths:	0	0	0	1	0	0	0	0	0	0	1
Prevalence:	0	0	0	6.03	6.17	2.22	0	0	0	2.61	1.76
Pierre Robin Syndrome											
Total cases:	3	1	1	0	0	1	3	0	1	1	11
Live Births:	3	1	1	0	0	1	3	0	1	1	11
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	5.8	2.06	2.16	0	0	2.22	6.91	0	2.58	2.61	2.43

Cases and prevalence per 10,000 births for selected anomaly groups 1993-2002											
	1993	**1994**	**1995**	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**93-02**
Population:	5172	4863	4633	4978	4864	4511	4339	4272	3883	3826	45340
Mandibulofacial dystosis (Treacher-Collins and Franceschetti)											
Total cases:	0	0	0	0	0	0	0	0	0	0	0
Live Births:	0	0	0	0	0	0	0	0	0	0	0
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	0	0	0	0	0	0	0	0
Oculomandibular dysostosis (Hallerman-Streiff											
Total cases:	0	0	0	0	0	0	0	0	0	0	0
Live Births:	0	0	0	0	0	0	0	0	0	0	0
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	0	0	0	0	0	0	0	0
Goldenhar's Syndrome											
Total cases:	0	0	0	0	0	0	0	0	0	0	0
Live Births:	0	0	0	0	0	0	0	0	0	0	0
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	0	0	0	0	0	0	0	0
Chondodystrophies and osteodystrophies											
Total cases:	0	2	1	1	1	1	0	2	2	2	12
Live Births:	0	1	1	1	1	1	0	2	1	1	9
Fetal Deaths:	0	1	0	0	0	0	0	0	1	1	3
Prevalence:	0	4.11	2.16	2.01	2.06	2.22	0	4.68	5.15	5.23	2.65
Diaphraghmatic hernia											
Total cases:	1	3	3	4	3	3	2	3	0	4	26
Live Births:	1	3	3	4	3	3	2	2	0	4	25
Fetal Deaths:	0	0	0	0	0	0	0	1	0	0	1
Prevalence:	1.93	6.17	6.48	8.04	6.17	6.65	4.61	7.02	0	10.45	5.73
Omphalocele											
Total cases:	1	2	1	2	1	1	0	0	1	1	10
Live Births:	1	1	0	2	0	1	0	0	1	1	7
Fetal Deaths:	0	1	1	0	1	0	0	0	0	0	3
Prevalence:	1.93	4.11	2.16	4.02	2.06	2.22	0	0	2.58	2.61	2.21
Gastroschisis											
Total cases:	2	0	0	0	1	0	0	1	1	0	5
Live Births:	2	0	0	0	1	0	0	1	1	0	5
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	3.87	0	0	0	2.06	0	0	2.34	2.58	0	1.1
Prune Belly Syndrome											
Total cases:	1	0	0	1	0	0	0	0	0	0	2
Live Births:	1	0	0	1	0	0	0	0	0	0	2
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	1.93	0	0	2.01	0	0	0	0	0	0	0.44

Cases and prevalence per 10,000 births for selected anomaly groups 1993-2002											
	1993	**1994**	**1995**	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**93-02**
Population:	5172	4863	4633	4978	4864	4511	4339	4272	3883	3826	45340
Chromosomal											
Total cases:	16	8	12	13	9	9	12	7	8	19	113
Live Births:	16	8	12	13	9	9	10	7	8	19	111
Fetal Deaths:	0	0	0	0	0	0	2	0	0	0	2
Prevalence:	30.94	16.45	25.9	26.11	18.5	19.95	27.66	16.39	20.6	49.66	24.92
Down Syndrome											
Total cases:	15	7	8	8	3	7	11	4	8	15	86
Live Births:	15	7	8	8	3	7	10	4	8	15	85
Fetal Deaths:	0	0	0	0	0	0	1	0	0	0	1
Prevalence:	29	14.39	17.27	16.07	6.17	15.52	25.35	9.36	20.6	39.21	18.97
Patau syndrome (trisomy 13)											
Total cases:	0	0	0	0	0	0	0	1	0	0	1
Live Births:	0	0	0	0	0	0	0	1	0	0	1
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	0	0	0	0	2.34	0	0	0.22
Edward syndrome (trisomy 18)											
Total cases:	0	1	2	1	4	1	1	1	0	2	13
Live Births:	0	1	2	1	4	1	0	1	0	2	12
Fetal Deaths:	0	0	0	0	0	0	1	0	0	0	1
Prevalence:	0	2.06	4.32	2.01	8.22	2.22	2.3	2.34	0	5.23	2.87
Other trisomies and partial trisomies of autosomes											
Total cases:	0	0	0	1	0	0	0	0	0	1	2
Live Births:	0	0	0	1	0	0	0	0	0	1	2
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	2.01	0	0	0	0	0	2.61	0.44
Monosomies and deletions from the autosomes											
Total cases:	1	0	2	1	0	0	0	1	0	0	5
Live Births:	1	0	2	1	0	0	0	1	0	0	5
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	1.93	0	4.32	2.01	0	0	0	2.34	0	0	1.1
Turner's syndrome											
Total cases:	0	0	0	0	1	0	0	0	0	0	1
Live Births:	0	0	0	0	1	0	0	0	0	0	1
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	0	2.06	0	0	0	0	0	0.22
Klinefelters syndrome											
Total cases:	0	0	0	0	0	0	0	0	0	0	0
Live Births:	0	0	0	0	0	0	0	0	0	0	0
Fetal Deaths:	0	0	0	0	0	0	0	0	0	0	0
Prevalence:	0	0	0	0	0	0	0	0	0	0	0

Cases and prevalence per 10,000 births for selected anomaly groups 1993-2002											
	1993	**1994**	**1995**	**1996**	**1997**	**1998**	**1999**	**2000**	**2001**	**2002**	**93-02**
Population:	5172	4863	4633	4978	4864	4511	4339	4272	3883	3826	45340
Anomalies outside normal range											
Total cases:	14	8	9	15	11	9	9	7	4	6	92
Live Births:	14	8	9	15	11	7	8	7	4	5	88
Fetal Deaths:	0	0	0	0	0	2	1	0	0	1	4
Prevalence:	27.07	16.45	19.43	30.13	22.62	19.95	20.74	16.39	10.3	15.68	20.29
Totals											
Total cases:	387	456	454	478	509	489	480	427	377	362	4419
Live Births:	372	430	452	453	499	484	428	422	372	352	4264
Fetal Deaths:	15	26	2	25	10	5	52	5	5	10	155
Prevalence:	748.26	937.69	979.93	960.22	1046.46	1084.02	1106.25	999.53	970.9	946.16	974.64

Fetal deaths include still births and spontaneous abortions from 20 weeks gestation

TABLE C - COMPARISON OF PREVALENCE RATES OF SELECTED ANOMALY GROUPS WITH OTHER EUROPEAN REGISTRIES WITHIN EUROCAT

Anomaly group	Malta	All EUROCAT Registries (Prevalence /10000)	
	LB+FD /10000	Average Rate	Lowest and Highest prevalence rates reported by EUROCAT Registries*
Nervous system			
Neural Tube Defects	12.35	8.48	2.90 (ECEMC**-Spain) – 24.63 (Mainz-Germany)
Anencephalus and similar	3.75	2.99	0.28 (South East Sicily) – 7.45 (Wales - UK)
Encephalocele	2.21	1.24	0.00 (Galway – Ireland) – 2.99 (Auvergne – France)
Spina Bifida	6.40	4.48	2.10 (ECEMC-Spain) – 17.18 (Mainz – Germany)
Hydrocephaly	5.29	4.96	0.56 (Galway-Ireland) – 18.62 (Mainz – Germany)
Microcephaly	3.31	1.80	0.11 (Sweden) – 6.95 (Cork and Kerry – Ireland)
Arhinencephaly/holoprosencephaly	0.88	1.30	0.00 (Galway-Ireland) – 4.58 (Mainz – Germany)
Eye			
Anophthalmos/microthalmos	1.32	1.18	0.00 (Oxford-UK and Auvergne-France) – 3.15 (Mainz – Germany)
Ear			
Anotia/microtia	0.00	1.12	0.00 (Malta, Galway and Auvergne) – 4.49 (Finland)
Congenital heart disease			
Anomalies of cardiac chambers & connections	5.95	5.81	1.72 (North East Italy) – 11.02 (South East Sicily)
Common arterial truncus	0.44	0.82	0.14 (Campania-Italy) – 7.46 (South East Sicily)
Transposition of great vessels (complete)	4.63	2.50	0.43 (Central East France) – 4.99 (Cork and Kerry – Ireland)
Atrioventricular septal defect	4.63	3.76	1.84 (Campania-Italy) – 8.88 (Mainz – Germany)
Tetralogy of Fallot	3.75	2.81	1.57 (Merseyside and Cheshire – UK) – 5.23 (Sofia – Bulgaria)
Hypoplastic left heart	1.76	2.23	0.65 (ECEMC - Spain) – 4.87 (Mainz – Germany)
Coarctation of aorta	5.73	3.58	0.82 (Zagreb - Croatia) – 9.40 (Finland)
Orofacial Clefts			
Cleft lip with or without palate	9.26	8.90	4.45 (Galway - Ireland) – 15.09 (Saxony-Anhalt – Germany)
Cleft palate	13.67	6.21	2.37 (Oxford - UK) – 14.16 (Finland)
Digestive system			
Tracheo-oesophagal fistula-Oesophageal atresia and stenosis	2.21	2.71	1.72 (South Portugal) – 7.16 (Mainz – Germany)
Congenital absence, atresia or stenosis small intestine	0.66	0.72	0.00 (Auvergne-France Zagreb-Croatia Merseyside and Oxford -UK) – 1.37 (Wessex – UK)
Congenital absence, atresia or stenosis of duodenum	0.66	1.32	0.33 (South Portugal) – 3.44 (Mainz – Germany)
Congenital absence, atresia or stenosis of other specified parts of small intestine	1.54	2.17	0.68 (Oxford – UK) – 5.16 (Mainz – Germany)
Ano-rectal atresia and stenosis	4.63	2.98	1.02 (Oxford – UK) – 8.31 (Mainz – Germany)
Internal urogenital system-ovaries uterus and renal system			
Bilateral renal agenesis	0.88	1.47	0.00 (Zagreb-Croatia) – 6.59 (Mainz – Germany)
Cystic kidney disease	3.75	4.59	0.24 (ECEMC-Spain) – 17.76 (Mainz – Germany)

Anomaly group	Malta	All EUROCAT Registries (Prevalence /10000)	
	LB+FD /10000	Average Rate	Lowest and Highest prevalence rates reported by EUROCAT Registries*
External genital system			
Hypospadias	17.42	8.08	1.46 (Wessex-UK) – 22.35 (Strasbourg – France)
Indeterminate sex	1.10	0.74	0.00 (Galway-Ireland) – 1.63 (Hainaut – Belgium)
Limb			
Limb reduction	6.18	5.67	1.67 (Galway-Ireland) – 10.88 (Mainz – Germany)
Upper limb reduction	4.41	4.06	0.56 (Galway-Ireland) – 7.20 (Glasgow – UK)
Lower limb reduction	2.21	1.93	0.00 (Oxford-UK) – 5.97 (Auvergne – France)
Polydactyly	16.10	8.75	1.80 (Wessex-UK) – 16.36 (Paris – France)
Syndactyly	10.37	5.80	1.02 (Oxford-UK) – 11.51 (Saxony Anhalt - Germany)
Musculoskeletal and connective tissue			
Chondodystrophies and osteodystrophies	2.65	2.15	0.60 (South Portugal) – 5.05 (Paris – France)
Diaphraghmatic hernia	5.73	2.61	0.00 (Wielkopolska – Poland) – 6.01 (Mainz – Germany)
Omphalocele	2.43	2.48	0.78 (ECEMC-Spain) – 5.31 (Paris – France)
Gastroschisis	1.10	1.69	0.36 (ECEMC-Spain) – 5.73 (Mainz – Germany)
Chromosomal			
Down Syndrome	18.97	17.54	8.36 (South Portugal) – 34.37 (Paris – France)
Patau syndrome (trisomy 13)	0.22	1.43	0.22 (Malta) – 4.58 (Mainz – Germany)
Edward syndrome (trisomy 18)	2.87	3.51	0.61 (ECEMC-Spain) – 9.19 (Paris – France)

LB - Live Births

FD - Fetal deaths / Still Births from 20 weeks gestation

*Prevalence rates taken from EUROCAT website: www.eurocat.ulst.ac.uk as at January 2005

** ECEMC – Spanish Collaborative Study

ANNEX 1 - LOCAL REPORT FORM

CONFIDENTIAL REPORT ON CONGENITAL ANOMALIES

(All data is collected and processed in accordance with the Data Protection Act 2001. The registry will not disclose personal information about the client to anyone outside the Health Division unless the law permits. The sensitive data being requested is required for the purposes of Public Health, Statistics and Research.)

CHILD'S NAME: _____ I.D. _____

ADDRESS: _____

_____

DATE OF BIRTH: _____

MOTHER'S NAME: _____ I.D. _____

DIAGNOSIS: _____

_____

_____

REFERRING DOCTOR: _____ DATE: _____

PLEASE SEND INFORMATION IN A SEALED ENVELOPE ADDRESSED TO:
MALTA CONGENITAL ANOMALIES REGISTRY, DEPT. OF HEALTH INFORMATION, 95 G'MANGIA HILL, G'MANGIA MSD 08

ANNEX 2 - LIST OF EXCLUSIONS

LIST OF MINOR ANOMALIES (AND ICD-9 CODES) WHICH ARE NOT INCLUDED IN REPORTS UNLESS IN COMBINATION WITH OTHER MAJOR ANOMALIES[1]

Anomalies of the eye
- Stenosis or stricture of lacrimal duct

Anomalies of the ear
- Minor or unspecified anomaly of the ear
- Preauricular appendage, tag or lobule
- Other appendage tag or lobule

Cardiovascular system
- Functional or unspecified cardiac murmur
- Absence or hypoplasia of umbilical artery, single umbilical artery
- Patent ductus arteriosus (in babies < 37 weeks or < 2500g)

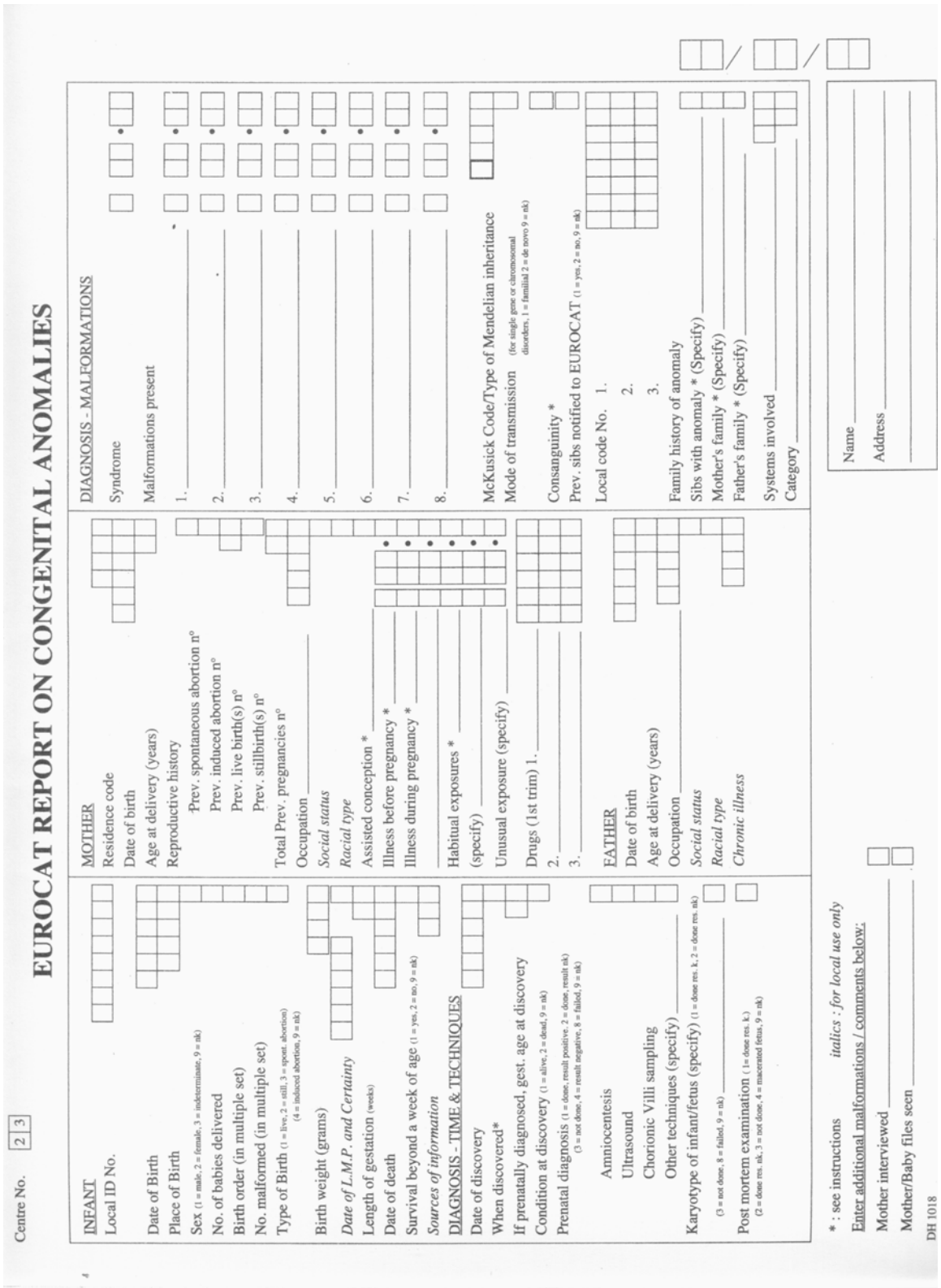
Digestive system
- Tongue tie

External genitals
- Undescended testicle and unspecified ectopic testis
- Congenital hydrocele or hydrocele of testis
- Phymosis
- Hypospadias when the meatus lies before the coronary sulcus, glandular or 1st degree hypospadias

Limbs
- Clicking hip
- Clubfoot of postural origin
- Postural or unspecified metatarsus varus or metatarsus adductus
- Postural or unspecified talipes calcaneovalgus or pes calcaneovalgus
- Minor or unspecified anomalies of toe such as hallux valgus, hallux varus or “orteil en marteau”

Other musculoskeletal anomalies and anomalies of the integument
- Spina bifida occulta uncomplicated
- Pectus excavatum
- Minor or unspecified anomaly of the nose
- Minor or unspecified deformity of the face
- Minor anomaly of nipple
- Accessory or ectopic nipple
- Congenital umbilical hernia, inguinal hernia, para umbilical, ventral or incisional, hiatus hernia
- Abnormal palmar crease
- Skin tag with surface area < 4 cm^2 : skin tag, naevus, angioma, haemangioma, glomus tumor, lymphangioma, birthmark
- Sacral dimple

ANNEX 3 - REGISTRY REPORT FORM



Centre No. 2 3

EUROCAT REPORT ON CONGENITAL ANOMALIES

INFANT
Local ID No.
Date of Birth
Place of Birth
Sex (1 = male, 2 = female, 3 = indeterminate, 9 = nk)
No. of babies delivered
Birth order (in multiple set)
No. malformed (in multiple set)
Type of Birth (1 = live, 2 = still, 3 = spont. abortion) (4 = induced abortion, 9 = nk)
Birth weight (grams)
Date of L.M.P. and Certainty
Length of gestation (weeks)
Date of death
Survival beyond a week of age (1 = yes, 2 = no, 9 = nk)
Sources of information

DIAGNOSIS - TIME & TECHNIQUES
Date of discovery
When discovered*
If prenatally diagnosed, gest. age at discovery
Condition at discovery (1 = alive, 2 = dead, 9 = nk)
Prenatal diagnosis (1 = done, result positive, 2 = done, result nk) (3 = not done, 4 = result negative, 8 = failed, 9 = nk)
Amniocentesis
Ultrasound
Chorionic Villi sampling
Other techniques (specify) _____
Karyotype of infant/fetus (specify) (1 = done res. k, 2 = done res. nk) (3 = not done, 8 = failed, 9 = nk) _____
Post mortem examination (1= done res. k.) (2 = done res. nk, 3 = not done, 4 = macerated fetus, 9 = nk)

MOTHER
Residence code
Date of birth
Age at delivery (years)
Reproductive history
Prev. spontaneous abortion n°
Prev. induced abortion n°
Prev. live birth(s) n°
Prev. stillbirth(s) n°
Total Prev. pregnancies n°
Occupation _____
Social status
Racial type
Assisted conception * _____
Illness before pregnancy * _____
Illness during pregnancy * _____
_____
Habitual exposures * _____
(specify) _____
Unusual exposure (specify) _____
Drugs (1st trim) 1. _____
2. _____
3. _____

FATHER
Date of birth
Age at delivery (years)
Occupation _____
Social status
Racial type
Chronic illness

DIAGNOSIS - MALFORMATIONS
Syndrome
Malformations present
1. _____
2. _____
3. _____
4. _____
5. _____
6. _____
7. _____
8. _____
McKusick Code/Type of Mendelian inheritance (for single gene or chromosomal disorders, 1 = familial 2 = de novo 9 = nk)
Mode of transmission
Consanguinity *
Prev. sibs notified to EUROCAT (1 = yes, 2 = no, 9 = nk)
Local code No. 1.
2.
3.
Family history of anomaly
Sibs with anomaly * (Specify) _____
Mother's family * (Specify) _____
Father's family * (Specify) _____
Systems involved _____
Category _____

Name _____
Address _____
_____

* : see instructions *italics : for local use only*
Enter additional malformations / comments below:
Mother interviewed _____
Mother/Baby files seen _____

DH 1018

ANNEX 4 - LIST OF ANOMALY SUBGROUPS

Anomaly	Description Of Anomaly	ICD9 codes	ICD10 codes
Nervous system	**Nervous system anomalies include neural tube defects, hydrocephaly, microcephaly and other anomalies of the brain, spinal cord and nervous system**	**740, 741, 7420-7425, 7428, 7429**	**Q00, Q01, Q02, Q03, Q04, Q05, Q06, Q07**
Neural Tube Defects	*Neural tube defects include anencephaly, encephalocele, spina bifida and iniencephacy*	*7400-7420*	*Q00, Q01, Q05*
Anencephalus and similar	Absence or deficience of a major portion of the cranial vault, the covering skin and the brain tissue. (incompatible with life)	7400-7402	Q00
Encephalocele	Herniation of the brain and/or meninges through a defect in the skull (exclude if associated with anencephalus)	7420	Q01
Spina Bifida	Midline defect of the osseous spine ususally affecting the posterior arches resulting in a herniation or exposure of the spinal cord and/or meninges (exclude if associated with anencephalus, or encephalocele)	7410-7419	Q05
Hydrocephaly	Dilation of ventricular system, not due to primary atrophy of the brain, with or without enlargement of the skull (exclude hydranencephaly, or association with NTDs)	7423, excl 74232	Q03
Microcephaly	Reduction in the size of the brain with a skull circumference less than three standard deviations below the mean for gestation or age and reduced growth during first year of life (exclude association with NTDs)	7421	Q02
Arhinencephaly/ holoprosencephaly	Absence of the first cranial (olfactory) nerve tract. There is a spectrum of anomalies from a normal brain, except for the first cranial nerve tract, to a single ventricle (holoprosencephaly)	74226	Q041, Q042
Eye		**7430-7436, 7438-7439 (excl. 74365)**	**Q10-Q15 (excl. Q105)**
Anophthalmos/micropthalmos		*7430, 7431*	*Q110 - Q112*
Anophthalmos	Unilateral or bilateral absence of the eye tissue.	7430	Q110, Q111
Micropthalmos	Small eye/eyes with smaller than normal axial length (exclude association with anophthalmos)	7431	Q112
Cataract	Alteration in the transparency of the crystalline lens	74332	Q120
Ear		**7440-7442 (excl. 74411, 74412)**	**Q16, Q17 (excl. Q179)**
Anotia/microtia		*74401, 74421*	*Q160, Q172*
Anotia	Absent pinna, with or without atresia of ear canal	74401	Q160
Microtia	Small or deformed pinna, with or without atresia of ear canal (exclude association with anotia)	74421	Q172
Congenital heart disease	**CHD includes malformations of heart, great vessels, and endocardial fibroelastosis (exclude PDA in preterm/ LBW babies (<2,500g or <37 weeks) - ICD9: 7470; ICD10: Q250)**	**7450-7459, 7460-7469, 7470-7474**	**Q20-Q26**
Anomalies of cardiac chambers and connections		*74500, 7451, 7453, 7457*	*Q20*
Common arterial truncus	Presence of a large single arterial vessel at the base of the heart (from which the aortic arch, pulmonary and coronary arteries originate), always accompanied by a large subvulvar septal defect.	74500	Q200
Transposition of great vessels (complete)	Total separation of circulation with the aorta arising from the right ventricle and the pulmonary artery from the left ventricle	74510	Q203

Anomaly	Description Of Anomaly	ICD9 codes	ICD10 codes
Single ventricle	Only one complete ventricle with an inlet valve and an outlet portion even though the outlet valve is atretic	7453	Q204
Malformations of cardiac septa		*74501, 7452, 7454-7456, 7458-7459*	*Q21*
Ventricular septal defect	Defect in the ventricular septum	7454	Q210
Atrial septal defect	Defect in the atrial septum	7455	Q211
Atrioventricular septal defect	Central defect of the cardiac septa and a common atrioventricular valve, includes primum ASD defects	7456	Q212
Tetralogy of Fallot	VSD close to the aortic valves, infundibular and pulmonary valve stenosis and over-riding aorta acroos the VSD	7452	Q213
Malformations of valves		*7460-7467*	*Q22-Q23*
Tricuspid atresia and stenosis	Obstruction of the tricuspid valve and hypoplasia of the right ventricle	7461	Q224
Ebstein's anomaly	Tricuspid valve displaced with large right atrium and small right ventricle	7462	Q225
Aortic valve atresia/stenosis	Occlusion of aortic valve or stenosis of varying degree, often associated with bicuspid valves	7463	Q230
Hypoplastic left heart	Hypoplasia of the left ventricle, outflow tract and ascending aorta resulting from an obstructive lesion of the left side of the heart	7467	Q234
Malformations of the great arteries and veins	*exclude PDA in preterm/LBW babies (<2,500g or <37 weeks) - ICD9: 7470; ICD10: Q250*	*7470, 7471, 7472, 7473, 74742, 74743*	*Q25-Q26*
Coarctation of aorta	Constriction in the region of aorta where the ductus joins aorta	7471	Q251
Orofacial Clefts		**7490-7492**	**Q35-Q36**
Cleft lip with or without palate	Clefting of the upper lip with or without clefting of the maxillary alveolar process and hard and soft palate	7491, 7492	Q36-Q37
Cleft palate	Fissure defect of the soft and/or hard palate(s) or submucous cleft without cleft lip (exclude association with cleft lip (7491-7492, Q36-Q37)	7490	Q35
Digestive system	**Includes tracheo-oesophagal fistula, oesophageal atresia and stenosis, atresia and stenosis of rectum and anal canal, atresia and stenosis of small intestine, Meckel's diverticulum, colon disorders, anomalies of intestinal fixation and anomalies of gallbladder, bile ducts and liver (exclude pyloric stenosis)**	**7503-7504, 7507-7519**	**Q39, Q402, Q403, Q408, Q409, Q41, Q42, Q43, Q44, Q45**
Tracheo-oesophagal fistula-Oesophageal atresia and stenosis	Occlusion or narrowing of the oesophagus with or without tracheo-oesophagael fistula	7503	Q390-Q394
Congenital absence, atresia and/or stenosis of the small intestine	Occlusion or narrowing of small intestine	7511	Q41
Congenital absence, atresia and/or stenosis of the duodenum	Occlusion or narrowing of duodenum	75110	Q410
Congenital absence, atresia and/or stenosis of other specified parts of small intestine	Occlusion or narrowing of jejumum or ileum	75111-75112	Q411-Q418
Ano-rectal atresia and stenosis	Imperforate anus or absence or narrowing of the communication canal between the rectum and anus with or without fistula to neighbouring organs	75121-75124	Q420, Q421, Q422, Q423

Anomaly	Description Of Anomaly	ICD9 codes	ICD10 codes
Internal urogenital system-ovaries uterus and renal system		**7520-7523, 7529, 7530, 7531, 7532, 7533, 7534-7539**	**Q50, Q510-Q514, Q517-Q519, Q60, Q61, Q62, Q63, Q641-Q649**
Bilateral renal agenesis	Bilateral absence, agensis, dysplasis or hypoplasia of kidneys including Potter's syndrome. Incompatible with life (exclude unilateral. exclude unspecified: Q602, Q605)	75300	Q601, Q604, Q606
Cystic kidney disease	Presence of single or multiple cyst(s) enlarging kidney tissue	7531	Q61
Congenital hydronephrosis	Obstruction of the urinary flow from kidney to bladder	75320	Q620
Bladder exstrophy	Defect in the closure of the bladder and lower abdominal wall	7535	Q641
External genital system	**Includes hypospadias, epispadias, indeterminate sex, and other anomalies such as absence of testis, aplasia or hypoplasia of scrotum, penis (exclude 7525, Q53)**	**7524, 7525, 7526, 7527, 7528**	**Q515, Q516, Q52, Q53, Q54, Q55, Q56, Q640**
Hypospadias	Agenesis of the distal urethra and opening of the urethra on the ventral side of the penis behind the coronary sulcus (exclude chordee: Q544; exclude glandular hypospadias Q540)	75260	Q541-Q543, Q548, Q549
Indeterminate sex	Includes true and pseudohermaphroditism male or female	7527	Q56
Limb	**Limb anomalies include limb reduction, polydactyly, syndactyly, congenital dislocation of the hip and other limb anomalies (exclude 75432, 75452, 75460, 75473, 75560, Q662, Q664, Q668)**	**7543-7544, 7545-7547, 7550-7551, 7552-7554, 7555-7556, 7558-7559**	**Q650-Q656, Q66, Q682-Q685, Q69, Q70, Q71, Q72, Q73, Q74**
Limb reduction	*Total or partial absence or severe hypoplasia of skeletal structure of the limbs*	*7552-7554*	*Q71-Q73*
Upper limb reduction		7552	Q71
Complete absence of upper limb		75520	Q710
Absence of upper arm and forearm with hand present		75521	Q711
Absence of both forearm and hand		75523	Q712
Absence of hand and fingers		75524	Q713
Longitudinal reduction defect/shortening of arm		75525-75527	Q714-Q718
Lower limb reduction		7553	Q72
Complete absence of lower limb		75530	Q720
Absence of thigh and lower leg with foot present		75531	Q721
Absence of both lower leg and foot		75533	Q722
Absence of foot and toe		75534	Q723
Longitudinal reduction defect/shortening of leg		75535, 75536	Q724-Q728
Polydactyly	Extra digit or extra toe	7550	Q69
Syndactyly	Partial or total webbing between 2 or more digits includes minor forms	7551	Q70

Anomaly	Description Of Anomaly	ICD9 codes	ICD10 codes
Musculoskeletal and connective tissue	**A heterogeneous group of anomalies including anomalies of diaphragm, abdominal wall, tongue, branchial cleft, auricular sinus, nose, face, skull, neck, thorax, bones, muscles and cartilages (exclude 74491, 74819, 75481,75610, 75636, Q189, Q309, Q676, Q760, Q767)**	**7444-7445, 7448-7449, 7480-7481, 7501-7502, 7540-7542, 7548, 7560-7568, 7569, 5240, 5249**	**Q18, Q30, Q380, Q382, Q389, Q67, Q680, Q688, Q75-Q79, Q8704, Q8705, Q8708,Q870A, K070, K079**
Choanal atresia	Bony or membraneous choanae with no passage from nose to pharynx	7480	Q300
Craniosynostosis	Premature closure of cranial sutures	75600	Q750
Pierre Robin Syndrome	Micrognatia, glossoptosis and often cleft palate	75603	Q8708
Mandibulofacial dystosis (Treacher-Collins and Franceschetti)	Malar and mandibular hypoplasia, malformation of ear, often cleft palate	75604	Q754, Q870A
Goldenhar's Syndrome	Facial, auriculo and vertebral malformations,usually unilateral	75606	Q8704
Chondrodystrophies and osteodystrophies	Heterogeneous group of dwarfism and other skeletal syndromes	7564, 7565	Q77, Q78
Diaphragmatic hernia	Defect in the diaphragm with protrusion of abdominal content into the thoracic cavity. Various degree of lung hypoplasia on the affected side	75661	Q790
Omphalocele	Herniation of abdominal content through the umbilical ring, the contents being covered by a membrane sometimes ruptured at the time of delivery	75670	Q792
Gastroschisis	Protrusion of abdominal contents through an abdominal wall defect lateral to an intact umbilical cord and not covered by a membrane	75671	Q793
Prune Belly Syndrome	Syndrome with deficient abdominal muscle and urinary obstruction/distension. May be secondary to urethral obstruction.	75672	Q794
Chromosomal	**(exclude balanced translocations: 7584, Q95)**	**7580-7583, 7585-7589**	**Q90-Q94, Q96-Q99**
Down Syndrome (trisomy 21)	karyotype 47xx +21 or 47xy +21	7580	Q90
Patau syndrome (trisomy 13)	karyotype 47xx +13 or 47xy +13	7581	Q914-Q917
Edward syndrome (trisomy 18)	karyotype 47xx +18 or 47xy +18	7582	Q910-Q913
Other trisomies and partial trisomies of autosomes		7585	Q92
Monosomies and deletions from the autosomes		7583	Q93
Turner's syndrome	karyotype 45x	75860, 75861, 75862, 75869	Q96
Klinefelters syndrome	karyotype 47xxy	7587	Q980-Q984
Anomalies outside normal range	**Relevant anomalies not coded within the range 740 to 759 of ICD 9 (International Classification of Disease, 9th edition, WHO Geneva 1992) or the Q chapter of ICD 10 (10th edition, WHO Geneva 1992)**		
All Cases	*Any case coded within the range 740 to 759 of ICD 9 (International Classification of Disease, 9th edition, WHO Geneva 1992) or the Q chapter of ICD 10 (10th edition, WHO Geneva 1992) or other relevant parts of chapters transmitted to EUROCAT, excluding minor anomalies according to the specifications in EUROCAT Guide 1.2*		