# EUROCAT Guide 1.3 and reference documents

# Instructions for the Registration and Surveillance of Congenital Anomalies

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ICD10-BPA extension codes

# Chapter 1

# Aims and Objectives of EUROCAT
# (Issued September 2005)

# Aims and Objectives of EUROCAT

The aim of EUROCAT is to carry out epidemiologic surveillance of congenital anomalies in Europe.

EUROCAT's objectives are:

- To provide essential epidemiologic information on congenital anomalies in Europe.
- To facilitate the early warning of 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 centre regarding clusters or exposures or risk factors for 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.
- To act as a catalyst for the setting up of registries throughout Europe collecting comparable, standardised data.

**Why European Collaboration?**

- Pooling of data
- Comparison of data
- Sharing of expertise
- Joint approach to European public health questions

**Why Register Congenital Anomalies?**

There are two main reasons why congenital anomaly registers are established:

1. To facilitate the identification of teratogenic exposures. Ever since thalidomide and rubella (german measles) were discovered as powerful teratogens, registries have been set up to facilitate research and surveillance concerning environmental causes of congenital anomalies, and to give early warning of new teratogenic exposures. Registers are also used for genetic studies, and increasingly for research into the interaction of genetic and environmental factors in causing congenital anomalies. Congenital anomalies are registered not only for this intrinsic importance, but also as indicators of other adverse pregnancy outcomes linked to teratogenic exposures such as spontaneous abortions and neurobehavioural outcomes that are not as amenable to surveillance.

2. For the planning and evaluation of health services. This includes primary prevention strategies such as periconceptional folic acid supplementation to prevent neural tube defects and vaccination against rubella to prevent congenital rubella syndrome, so-called "secondary prevention" by prenatal screening and diagnosis, and tertiary prevention through paediatric rehabilitative and other services. Population-based registries are a particularly powerful tool for the evaluation of health services, because they represent the experience of a whole community, not the outcomes of specialist units which may serve only a selected group of women or children or which may have atypical human or financial resources. Many birth defect registries in Europe have been set up to provide a mechanism for the audit of prenatal screening practice. A registry can provide data on the proportion of cases of congenital anomaly diagnosed prenatally, the proportion of positive prenatal screening results which were confirmed as cases of congenital anomaly, and the proportion of prenatally diagnosed cases which led to termination of pregnancy, as well as related information about prenatal screening methods.

**How Can A Register Be Used?**

Whether concerned with the identification of teratogenic exposures, or with planning and evaluation of health services, or both, registers can be used in two main ways:

1. As a basis for surveillance using routinely collected data. Every register routinely collects a core dataset of standard information on each malformed child and more limited information on non-malformed children in the population.
2. As a basis for special or ad-hoc studies, such as case-control studies, which require further data collection. A register of congenital anomaly cases with diagnostic information can greatly facilitate the conduct of ad-hoc studies that seek to address specific hypotheses concerning teratogenic exposures or effectiveness of health services.

The decision as to which data should be included in the routine dataset of a registry, and which data should be collected only in ad-hoc studies is a difficult one. Each data item that is included in the routine dataset uses resources that must be balanced against other uses of those resources. Collection of incomplete and inaccurate data is generally a waste of resources. Depending on local circumstances, it may be justifiable for the registry to concentrate almost entirely on data about the baby and its diagnosis in routine data collection, leaving most risk factor data for collection in ad-hoc studies. Some ad-hoc data collection will always be necessary to address new or more elaborate hypotheses. However, registers that do not record the identity of children for confidentiality reasons can experience difficulties in supporting ad-hoc studies effectively and efficiently.

In chapter 2 we define the EUROCAT core dataset to be collected by all registries, and an extended dataset.

In general, risk factor data must be present for both cases and controls (non-malformed children) in order to be interpretable. While all registers collect basic information about the number of births in their population by type of birth and maternal age, some registers routinely collect the same set of risk factor information on control babies as on case babies. There are also useful approaches to analyzing risk factor data among malformed cases only, where children with different malformations act as controls for each other in "proportionate" analyses. For example, specific associations between particular drugs and particular malformation types can be sought.

# Chapter 2

# Data Transmission

2.1 General Instructions for Data Transmission

2.2 Variables and Coding Instructions for Transmission of Data to EUROCAT Central Registry

- 2.2.1a Summary of Variables
- 2.2.1b Coding Instructions
- 2.2.2 EDMP Derived Variables
- 2.2.3 Recommended Local Variables
- 2.2.4 Template for Associate Member Registry Data Transmission

2.3 Template for Denominator Data

2.4 EUROCAT Data Management Program (EDMP) Instructions

2.5 Data Validation Routines

# Chapter 2.1

# General Instructions for Data Transmission (Issued September 2005)

## 2.1 General Instructions for Data Transmission

1. Full members of EUROCAT transmit to Central Registry an electronic file of individual records of all cases of congenital anomaly occurring in the population surveyed by the register in a single year. The full dataset is given in Chapter 2.2.1. Complete information on all core variables (see point 9) *must* be transmitted, while information on non-core variables can be omitted with Central Registry's agreement.

2. Associate members of EUROCAT transmit to Central Registry a file of case counts per anomaly subgroup, year, and type of birth. Maternal age information is sent for cases of gastroschisis and Down Syndrome. See chapter 2.2.4 for Associate Registry data collection template. Data transmission instructions (see points 5 to 9) are not applicable to associate members.

3. Full and associate members should transmit denominator information according to the template given in chapter 2.3 of this Guide.

4. Guide 1.3 is a revision of Guide 1.2 for use for all births from 1st January 2005. Guide 1.2 should continue to be used for births up to 31st December 2004. Guides 1.2 and 1.3 are compatible with the EUROCAT Data Management Program (EDMP Version 27/07/05).

5. All data files should be validated locally first using the EUROCAT Data Management Program (EDMP). The EDMP validates data using the validation routines specified in chapter 2.5 of this Guide.

6. All data transmitted to Central Registry must be exported from EDMP. There are two possibilities for the transmission of data to EUROCAT Central Registry:

    - The EDMP is used for data entry. When your data entry is finished, run the validation and duplicate checks, make any corrections necessary, and then use the "Export" function to create a file for transmission to Central Registry.
    - If you enter your data in your own local program, you should import your data into the EDMP and run the validation routines and duplicate checks. Correct your data according to the results of these checks, import the corrected file into EDMP (after deleting the incorrect file), and then use the "Export" function to create a file for transmission to Central Registry. This will mean that Central Registry receives standardised data in terms of formatting and basic validation checks.

7. Instructions on how to use EDMP are included in chapter 2.4 of this Guide. The EDMP is downloadable from the Membership Only area of the EUROCAT website or can be provided by Central Registry on a compact disk and will run on Microsoft Access 2000 / 2002/ or 2003 software.

8. If you are sending updated records for previous years (years already transmitted to EUROCAT Central Registry), transmit the complete set of records for that year, not just the updated individual record(s). Central Registry will REPLACE the old file with the new file for that year.

9. Core variables are the minimum EUROCAT dataset. Core variables are shaded in grey in the coding instructions (chapter 2.2.1 of this Guide). As part of the validation routine, EDMP will indicate where core data is missing so that you can make every effort to complete it. There is an option in the EDMP for registries that choose to transmit only core variables to Central Registry.

# Chapter 2.2

# Variables for and Coding Instructions for Transmission of Data to EUROCAT Central Registry

2.2.1 Variables for Transmission to Central Registry

- 2.2.1a Summary of Variables (Issued February 2012)
- 2.2.1b Coding Instructions (Revised February 2012)

2.2.2 EDMP Derived Variables (Revised January 2012)

2.2.3 Recommended Local Variables (Issued September 2005)

2.2.4 Template for Associate Member Registry Data Transmission (Revised March 2007)

## 2.2.1a Summary of Variables (core variables are shaded blue) (Revised February 2012)

| Variable Number | Variable Name | Variable heading |
|---|---|---|
| **Baby and Mother – Variables 1 to 17** | | |
| 1 | CENTRE | Centre Number |
| 2 | NUMLOC | Local ID |
| 3 | BIRTH_DATE | Date of Birth |
| 4 | SEX | Sex |
| 5 | NBRBABY | Number of babies/fetuses delivered |
| 6 * | SP_TWIN | Specify twin type of birth, like or unlike, zygosity |
| 7 | NBRMALF | Number of malformed in multiple set |
| 8 | TYPE | Type of Birth |
| 9 * | CIVREG | Civil registration status |
| 10 | WEIGHT | Birth weight |
| 11 | GESTLENGTH | Length of gestation in completed weeks |
| 12 # | SURVIVAL | Survival beyond one week of age |
| 13 ** | DEATH_DATE | Date of death |
| 14 ** | DATEMO | Date of birth of mother |
| 15 | AGEMO | Age of mother at delivery |
| 16 | RESIDMO | Mother's residence code |
| 17 | TOTPREG | Total number of previous pregnancies |
| **Diagnosis – Variables 18 to 45** | | |
| 18 ** | WHENDISC | When discovered |
| 19 | CONDISC | Condition at discovery |
| 20 | AGEDISC | If prenatally diagnosed, gestational age at discovery |
| 21 * | FIRSTPRE | First positive prenatal test |
| 22 * | SP_FIRSTPRE | Specify first prenatal test in text if coded 7 ("other test positive") |
| 23 ** | KARYO | Karyotype of infant/fetus |
| 24 | SP_KARYO | Specify karyotype |
| 25 | PM | Post mortem examination |
| 26 * | SURGERY | First surgery for malformation performed or planned |
| 27 | SYNDROME | Syndrome |
| 28 | SP_SYNDROME | Specify Syndrome |
| 29 | MALFO1 | Malformation |
| 30 | SP_MALFO1 | Specify malformation |
| 31 | MALFO2 | As MALFO1 |
| 32 | SP_MALFO2 | Specify malformation |
| 33 | MALFO3 | As MALFO1 |
| 34 | SP_MALFO3 | Specify malformation |
| 35 | MALFO4 | As MALFO1 |
| 36 | SP_MALFO4 | Specify malformation |
| 37 | MALFO5 | As MALFO1 |
| 38 | SP_MALFO5 | Specify malformation |
| 39 | MALFO6 | As MALFO1 |
| 40 | SP_MALFO6 | Specify malformation |
| 41 | MALFO7 | As MALFO1 |
| 42 | SP_MALFO7 | Specify malformation |

| | | |
|---|---|---|
| 43 | MALFO8 | As MALFO1 |
| 44 | SP_MALFO8 | Specify malformation |
| 45 | MCKUSICK | McKusick Code / Type of Mendelian Inheritance |
| 46 * | AETIOLOGY | Aetiological classification of malformation |
| **Exposure – Variables 47 to 63** | | |
| 47 ## | ASSCONCEPT | Assisted conception |
| 48 | OCCUPMO | Mother's occupation at time of conception |
| 49 ** | ILLBEF1 | Illness before pregnancy 1 |
| 50 * | ILLBEF2 | Illness before pregnancy 2 |
| 51 ** | ILLDUR1 | Illness during pregnancy |
| 52 ** | ILLDUR2 | Illness during pregnancy 2 |
| 53 * | FOLIC | Folic acid supplementation |
| 54 *** | DRUGS1 | Drugs |
| 55 | SP_DRUGS1 | Specify drug exposures |
| 56 *** | DRUGS2 | As for DRUGS1 |
| 57 | SP_DRUGS2 | Specify drug exposures |
| 58 *** | DRUGS3 | As for DRUGS1 |
| 59 | SP_DRUGS3 | Specify drug exposures |
| 60 *** | DRUGS4 | As for DRUGS1 |
| 61 * | SP_DRUGS4 | Specify drug exposures |
| 62 *** | DRUGS5 | As for DRUGS1 |
| 63* | SP_DRUGS5 | Specify drug exposures |
| **Family History – Variables 64 to 75** | | |
| 64 ** | CONSANG | Consanguinity |
| 65 * | SP_CONSANG | Specify text information on consanguinity |
| 66 ** | SIBANOM | Siblings with anomalies |
| 67 | SP_SIBANOM | Specify type of anomaly and describe the malformation |
| 68 | PREVSIB | Previous malformed sibs notified to EUROCAT |
| 69 | SIB1 | Local ID number notified to the Central Registry |
| 70 | SIB2 | As SIB1 |
| 71 | SIB3 | As SIB1 |
| 72 ** | MOANOM | Mother's family with anomalies |
| 73 | SP_MOANOM | Specify type of anomaly and describe the malformation |
| 74 ** | FAANOM | Father's family with anomalies |
| 75 | SP_FAANOM | Specify type of anomaly and describe the malformation |
| **Sociodemographic – Variables 76 to 79** | | |
| 76* | MATEDU | Maternal education |
| 77* | SOCM | Socioeconomic status of mother |
| 78* | SOCF | Socioeconomic status of father |
| 79* | MIGRANT | Migrant status |
| **General Comments – Variable 80** | | |
| 80 | GENREM | General additional comments |
| **Extra Drugs – Variable 81** | | |
| 81* | EXTRA_DRUGS | Extra drugs |

* New variable In Guide 1.3
** Variable compatible with Guide 1.2, but coding has been extended/modified
*** Variable in Guide 1.2, but new ATC coding
# Variable compatible with Guide 1.2, code "3" added in June 2008
## Variable compatible with Guide 1.2, but coding has been extended/modified. Code "10" added in March 2010

## 2.2.1b Coding Instructions (Revised February 2012)

### Baby and Mother (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|---|---|---|---|
| 1 | CENTRE | CENTRE NUMBER | Code allocated by Central Registry |
| 2 | NUMLOC | LOCAL ID<br>Each case has a unique identification. This number is a maximum of 11 characters long, consisting of either numbers, letters or both.<br><br>ID numbers should not repeat themselves in different years. | Up to 11 digits |
| 3 | BIRTH_DATE | DATE OF BIRTH<br>Please enter dates as a numeric string, not in date format (eg. do not use 28/02/89 or 28-02-89, instead use 280289). | Day, month, year<br>99 = Not known for day and month<br><br>DO NOT TRANSMIT RECORDS IF YEAR OF BIRTH IS NOT KNOWN |
| 4 | SEX | SEX<br>Indicate chromosomal sex, if known, in case of ambiguous genitalia and code malformations in variables 29-44.<br><br>Indicate indeterminate sex in case of ambiguous genitalia with unknown or abnormal sex chromosome complement.<br><br>If sex could not be determined at autopsy due to maceration or other problems, indicate as “not known”. | 1 = Male<br>2 = Female<br>3 = Indeterminate<br>9 = Not known |
| 5 | NBRBABY | NUMBER OF BABIES/FETUSES DELIVERED<br>Fill out a separate form for each malformed baby/fetus in a multiple set.<br><br>Only one form to be completed for conjoined twins (Siamese). The code is “2” for a conjoined twin, unless another baby was delivered at the same time (code “3”).<br><br>Conjoined twins have a specific ICD/BPA code, to be coded under “syndrome” (variable 27). Give full description of type of conjoined twinning in syndrome text field (variables 28).<br><br>Any other malformations are coded in variables 29-44. | 1 = Singleton<br>2 = Twins<br>3 = Triplets<br>4 = Quadruplets<br>5 = Quintuplets<br>6 = Sextuplets or more<br>9 = Not known |
| 6 | SP_TWIN | SPECIFY TWIN TYPE OF BIRTH (malformed and non-malformed), like or unlike sex, zygosity | Free text |

## Baby and Mother (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|---|---|---|---|
| 7 | NBRMALF | NUMBER OF MALFORMED IN MULTIPLE SET<br>To be completed for multiple delivery only.<br><br>Complete one form for each malformed baby in a multiple set. Remember to give local ID of co-twin in SIB1 field (variable 69) if more than one malformed. | 1 = One<br>2 = Two<br>3 = Three<br>4 = Four<br>5 = Five<br>6 = Six or more<br>9 = Not known |
| 8 | TYPE | TYPE OF BIRTH<br>Birth with type of birth not known should be transmitted to EUROCAT, but will be excluded from routine EUROCAT analysis.<br><br>EUROCAT includes all livebirths, fetal deaths with gestational age (GA) ≥20 weeks and terminations of pregnancy (at any gestational age) after prenatal diagnosis of malformation. Fetal deaths with GA < 20 weeks (code = 3) may be reported to EUROCAT but will not be included in prevalence data.<br><br>The distinction between stillbirth and spontaneous abortion should follow the definitions in use in your country (to be specified in your Registry Description). There is usually a lower gestational age limit or birthweight limit for stillbirths. This varies from country to country. Below this limit fetal deaths are called spontaneous abortions.<br><br>Terminations of pregnancy refer to cases where prenatal diagnosis was made of malformation in a live fetus and the pregnancy was then terminated. If the fetus died spontaneously in utero either before or after prenatal diagnosis of malformation then it should be coded as spontaneous abortion or stillbirth, not as termination of pregnancy. If a termination was performed for other reasons than malformation, the case should not be transmitted to Central Registry. This means that early terminations where there was no suspicion of malformation before termination should be excluded from the case files.<br><br>Stillbirths or perinatal deaths resulting from termination of pregnancy following prenatal diagnosis must be coded as terminations (value = 4), irrespective of civil registration status.<br><br>For a non-natural fetal reduction in a multiple pregnancy where one fetus is malformed, code 4 (in that case gestlength = gestational age at reduction; date of birth = date of reduction; and code carefully all multiple birth variables). | 1 = Live birth<br>2 = Stillbirth<br>3 = Spontaneous abortion<br>4 = Termination of Pregnancy<br>5 = Not known |

## Baby and Mother (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|---|---|---|---|
| 9 | CIVREG | CIVIL REGISTRATION STATUS<br>Livebirths and stillbirths are civilly registered leading to either a birth or stillbirth certificate and appear in official birth statistics for your region.<br><br>Code here whether this case fulfilled the conditions for live or stillbirth registration in your country. | 1 = Livebirth<br>2 = Stillbirth<br>3 = No civil registration<br>9 = Not known |
| 10 | WEIGHT | BIRTH WEIGHT<br>Give weight in grams. | 9999 = Not known<br><br>(Do not use 99 or 999 for "Not Known" as this will be considered the birth weight). |
| 11 | GESTLENGTH | LENGTH OF GESTATION IN COMPLETED WEEKS<br>Give best estimate based on last menstrual period (LMP) and/or ultrasound determination. If the case is the result of fetal reduction give GA at fetocide. | 99 = Not known |
| 12 | SURVIVAL | SURVIVAL BEYOND ONE WEEK OF AGE<br>Yes = Child known to be alive after one week.<br><br>No = Child known to have died before or during first week (including stillbirths and abortions).<br><br>Alive at discharge <1 week refers to cases that are alive at discharge from maternity units before one week of age. Please specify in your Registry Description the day when discharge from maternity units usually takes place.<br><br>If survival at one week is unknown, but survival at discharge from maternity unit less than one week is known, use the latter.<br><br>The definition of first week of life varies between countries. Follow your country's perinatal mortality definition and specify this in your Registry Description.<br><br>Not known = Not known if child has died during first week. | 1 = Yes<br>2 = No<br>3 = Alive at discharge <1 week<br>9 = Not known |

## Baby and Mother (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|---|---|---|---|
| 13 | DEATH_DATE | DATE OF DEATH<br>**For livebirths only.**<br><br>Please enter dates as a numeric string, not in date format (eg. do not use 28/02/89 or 28-02-89, instead use 280289). | Day, month, year<br>99= Died, not known day or month<br>44 =Died, not known year<br><br>Do not use 99 for "not known" year of death, as this will be read as died in 1999, day and month not known.<br><br>222222= Known to be alive at 1 year<br>333333= Not known if alive or dead at 1 year |
| 14 | DATEMO | DATE OF BIRTH OF MOTHER<br>Give as much information as is known eg. Feb 1963 = 990263, 1963 = 999963.<br><br>Please enter dates as a numeric string, not in date format (eg. do not use 28/02/89 or 28-02-89, instead use 280289).<br><br>This variable can be used to calculate maternal age at Expected Date of Delivery for preterm deliveries and terminations. | Day, month, year<br>99 = Not known day or month<br>44 = Not known year |
| 15 | AGEMO | AGE OF THE MOTHER AT DELIVERY<br>In completed years at the time of delivery. If only the year of birth is available, assume that the mother was born on 30 June. | 99 = Not known |
| 16 | RESIDMO | MOTHER'S RESIDENCE CODE<br>Use local code for locality of residence at time of delivery. | Local code (up to 10 digits) |
| 17 | TOTPREG | TOTAL NUMBER OF PREVIOUS PREGNANCIES<br>NOTE – The current reported pregnancy is NOT included.<br><br>Include all previous abortions whether spontaneous or induced. Multiple pregnancies count as 1 in the total | 00 = None<br>01 = One<br>02 = Two<br>03 = Three etc<br>20 = Twenty or more<br>99 = Not known |

## Diagnosis (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|---|---|---|---|
| 18 | WHENDISC | WHEN DISCOVERED<br>When the baby was first suspected to be malformed.<br><br>For prenatal diagnosis: when a major malformation was first suspected (EXCLUDING soft markers). If prenatal diagnosis is made when fetus is dead code 1 (for stillbirths) or 7 (for spontaneous abortions).<br>For livebirths: when first suspicion malformation was at death OR at postmortem, when discovered is age at death (eg. At birth, < 1 week, 1-4 weeks etc).<br>For stillbirths: when first suspicion of a malformation was at birth OR at postmortem, when discovered is at birth (eg. Code = 1).<br><br>Please also complete variables 12 "SURVIVAL", 13 "DEATH-DATE", 19 "CONDISC" and 25 "PM". | 1 = At birth<br>2 = Less than 1 week<br>3 = 1-4 weeks<br>4 = 1-12 months<br>5 = Over 12 months<br>6 = Prenatal diagnosis in live fetus<br>7 = At abortion (spontaneous)<br>9 = Not known<br>10 = Postnatal diagnosis, age not known |
| 19 | CONDISC | CONDITION AT DISCOVERY<br>Condition of fetus or baby when malformation was first suspected. | 1 = Alive<br>2 = Dead<br>9 = Not known |
| 20 | AGEDISC | IF PRENATALLY DIAGNOSED, GESTATIONAL AGE AT DISCOVERY IN COMPLETED WEEKS<br>GA as defined in variable gestlength.<br><br>Gestational age at which the fetus was first suspected to be malformed (EXCLUDING soft markers). Indicate time of examination rather than time when result known.<br><br>If no prenatal diagnosis please leave blank. | 99 = Not known |
| 21 | FIRSTPRE | FIRST POSITIVE PRENATAL TEST<br>This refers to the first prenatal test whether screening procedure or diagnostic test which indicated a possible congenital anomaly or need for further tests.<br><br>For code 7 = other specified test, give information in text field (variable 22).<br><br>If test performed and result negative, then the "When discovered" variable cannot be coded 6 (prenatal diagnosis). | 1 = Ultrasound at GA < 14 weeks<br>2 = Ultrasound at GA 14-21 weeks<br>3 = Ultrasound at GA ≥ 22 weeks<br>4 = Ultrasound GA not known<br>5 = Serum/ combined screening<br>6 = CVS or amniocentesis<br>7 = Other test positive<br>8 = Test(s) performed, result negative<br>9 = Not known<br>10 = No test performed |

## Diagnosis (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|---|---|---|---|
| 22 | SP_FIRSTPRE | SPECIFY "OTHER" FIRST PRENATAL TEST<br>If FIRSTPRE = 7, specify which positive prenatal test | Free text |
| 23 | KARYO | KARYOTYPE OF INFANT/FETUS<br>Specify result in variable 24.<br><br>If performed and results known, please specify (according to Paris nomenclature).<br><br>"Probe test performed" refers to FISH, PCR, or other analyses restricted to specific chromosomal anomalies.<br><br>"Failed" refers to a technical failure where a repeat examination could not be done and the karyotype is therefore unknown. | 1 = Performed, result known<br>2 = Performed, results not known<br>3 = Not performed<br>4 = Probe test performed<br>8 = Failed<br>9 = Not known |
| 24 | SP_KARYO | SPECIFY KARYOTYPE | Free text |
| 25 | PM | POST MORTEM EXAMINATION<br>If performed record the malformation(s) discovered in the "malformation" section in the form. If other findings record in the "comments" space (variable 80).<br><br>"Results known" means that the autopsy record has been reviewed by the registry.<br><br>"Results not known" means that the autopsy record was not available to the registry.<br><br>"Macerated fetus" means that although a post mortem was performed, maceration of the fetus prevented a full protocol from being followed. | 1 = Performed, results known<br>2 = Performed, results not known<br>3 = Not performed<br>4 = Macerated fetus<br>9 = Not known |
| 26 | SURGERY | FIRST SURGICAL PROCEDURE FOR MALFORMATION (PERFORMED OR EXPECTED)<br>If your registry does not have the resources to complete this variable for all cases, concentrate on malformations where a) performed or expected surgery indicates level of severity, for example hydrocephalus and hypospadias or b) where active decisions to withhold surgery may impact survival for example hypoplastic left heart<br><br>The variable surgery does not include insertions of catheters. Performed (or expected) means that this case has already, or will at the appropriate age, have surgery for one or more of the listed malformations.<br>"No surgery required" means that this case does not have a severe enough malformation, or that the malformation is not correctable by surgery.<br>"Too severe for surgery" means that there has been an active decision to withhold surgery due to low chances of survival or very poor prognosis. | 1 = Performed (or expected) in the first year of life<br>2 = Performed (or expected) after the first year of life<br>3 = Prenatal surgery<br>4 = No surgery required<br>5= Too severe for surgery<br>9 = Not known |

## Diagnosis (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|---|---|---|---|
| 27 | SYNDROME | SYNDROME OR ASSOCIATION<br>Refer to EUROCAT Guide on syndromes. Give name of syndrome or association in text variable 28. All the anomalies observed by the local clinician should be coded in the remaining boxes for malformations.<br><br>If not a recognised syndrome or association, leave blank.<br><br>When 2 syndromes are present in the same subject, code the more important one in the syndrome variables 27 and 28, and include the other one in variables 29 and 30 MALF01.<br><br>Ensure karyotype information is given in variables 23 and 24, and that autopsy and medical genetics reports have been reviewed, where appropriate.<br><br>In case of conjoined twins, give full description in syndrome text variable 28.<br><br>Local registries are advised to keep photographs and x-ray images of all syndrome cases, as the diagnosis is predominantly established on the basis of specific facial dysmorphism. | ICD 10<br><br>First 4 digits are ICD10<br>5th digit = BPA supplement or leave blank |
| 28 | SP-SYNDROME | SPECIFY SYNDROME<br>Please specify availability of photographs and x-ray images of syndrome case. | |
| 29 | MALFO1 | MALFORMATION<br>A baby/fetus with ONLY minor anomalies (see exclusion list, chapter 7) should not be transmitted to Central Registry.<br><br>When a major anomaly is present, code both major and minor anomalies.<br><br>Up to 8 malformations can be coded – if more that 8 are present, specify additional anomalies in the text variable for the 8th anomaly (text variable 44 SP_MALFO8).<br><br>Include in the 8 specified codes the most important ones, or those tabulated in EUROCAT Reports.<br><br>Give written description of the malformations available in malformation text variables 30, 32, 34, 36, 38, 40, 42 and 44. | ICD 10<br><br>First 4 digits are ICD<br>5th digit = BPA classification OR leave blank |

## Diagnosis (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|---|---|---|---|
| 30 | SP_MALFO1 | SPECIFY MALFORMATION | Free text |
| 31 | MALFO2 | AS MALFO1 | As MALF01 |
| 32 | SP_MALFO2 | SPECIFY MALFORMATION | Free text |
| 33 | MALFO3 | AS MALFO1 | As MALF01 |
| 34 | SP_MALFO3 | SPECIFY MALFORMATION | Free text |
| 35 | MALFO4 | AS MALFO1 | As MALF01 |
| 36 | SP_MALFO4 | SPECIFY MALFORMATION | Free text |
| 37 | MALFO5 | AS MALFO1 | As MALF01 |
| 38 | SP_MALFO5 | SPECIFY MALFORMATION | Free text |
| 39 | MALFO6 | AS MALFO1 | As MALF01 |
| 40 | SP_MALFO6 | SPECIFY MALFORMATION | Free text |
| 41 | MALFO7 | AS MALFO1 | As MALF01 |
| 42 | SP_MALFO7 | SPECIFY MALFORMATION | Free text |
| 43 | MALFO8 | AS MALFO1 | As MALF01 |
| 44 | SP_MALFO8 | SPECIFY MALFORMATION | Free text |
| 45 | MCKUSICK | MCKUSICK / TYPE OF MENDELIAN INHERITANCE<br>To be coded by medical geneticist or after advice from medical geneticist.<br><br>This code is to be used for cases with single gene origin only – Refer to EUROCAT Syndrome Guide.<br><br>The first digit may be filled in without the rest of the code if the full McKusick/OMIM code is not known.<br><br>Full codes can be found on the OMIM website http://www.ncbi.nlm.nih.gov/omim/ | First digit designates mode of inheritance<br>1 = Autosomal dominant<br>2 = Autosomal recessive<br>3 = X-linked<br><br>Further digits denote syndromes |
| 46 | AETIOLOGY | AETIOLOGICAL CLASSIFICATION OF MALFORMATION<br>This is an optional variable to be completed only by a medical geneticist or after advice from a medical geneticist.<br><br>Reference document: Wellesley D, Boyd P, Dolk H, Pattenden S **(2005)**, "An Aetiological Classification of Birth defects for Epidemiological Research", *Journal of Medical Genetics*, Vol 42, No 1, pp 54-57. Available at http://www.eurocat.ulster.ac.uk/pubdata/ | C = chromosomal<br>OG = other genomic<br>T = teratogen<br>ND = New Dominant<br>F = familial<br>S = syndrome<br>I = isolated<br>M = multiple |

## Exposure (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|---|---|---|---|
| 47 | ASSCONCEPT | ASSISTED CONCEPTION<br>IVF = In vitro fertilization<br>GIFT = Gamete intra fallopian transfer<br>ICSI = Intracytoplasmic sperm injection | 0 = No<br>1 = Induced ovulation only<br>2 = Artificial insemination<br>3 = IVF<br>4 = GIFT<br>5 = ICSI<br>6 = Egg donation<br>8 = Other<br>9 = Not known<br>10 = Assisted conception, type unknown |
| 48 | OCCUPMO | MOTHER'S OCCUPATION AT TIME OF CONCEPTION<br>Code main occupation at time of conception. (Code according to the 1988 International Standard Classification of Occupations [ISCO]).<br>Links for ISCO classifications:<br>http://www.ilo.org/public/english/bureau/stat/isco/index.htm (English)<br>http://www.warwick.ac.uk/ier/isco/brit/grplist.html (English)<br>http://europa.eu.int/comm/eurostat/ramon/ (English with French and Spanish description).<br>The 4 digit ISCO-88 codes give more specificity, so use these when possible<br><br>Main Groups:<br>1 = Legislators, Senior Officials and Managers (ISCO-88 range 1110 to 1319)<br>2 = Professionals (ISCO-88 2111 to 2460)<br>3 = Technicians and Associate Professionals (ISCO-88 3111 to 3480)<br>4 = Clerks (ISCO-88 4111 to 4223)<br>5 = Service Workers and Shop and Market Sales Workers (ISCO-88 5111 to 5230)<br>6 = Skilled Agricultural and Fishery Workers (ISCO-88 6111 to 6210)<br>7 = Craft and Related Trades Workers (ISCO-88 7111 to 7442)<br>8 = Plant and Machine Operators and Assemblers (ISCO-88 8111 to 8340)<br>9 = Elementary Occupations (ISCO-88 9111 to 9333)<br>EUROCAT Supplement:<br>94 = Armed Forces<br>95 = Housewife<br>96 = Student<br>97 = Unemployed | 4 digit code<br><br>9999 = Not known (do NOT use 9, 99 or 999) |

## Exposure (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|---|---|---|---|
| 49 | ILLBEF1 | ILLNESS BEFORE PREGNANCY 1<br>Record any illness whether chronic or acute with onset before pregnancy and that may affect fetal development (eg. childhood cancer, metabolic disease). Code according to ICD10. The codes mentioned below are only examples.<br><br>Any additional details may be entered in the general comments section (variable 80).<br><br>Do not insert the decimal point in the code (e.g. Code E05.0 as E050)<br><br>Abridged list:<br>Hyperthyroidism E050 - E059<br>Hypothyroidism E000 - E039<br>Diabetes Type 1 E100 - E109<br>Diabetes Type 2 E110 - E119<br>Obesity E660 - E669<br>Epilepsy G400 - G409<br>Asthma J450 - J459<br>Chronic alcoholism F102<br>Drug addict F112 - F122 - F132 - F142 F152 - F192 | ICD 10<br>0 = No illness<br>1 = Yes, but no information available<br>9 = Not known |
| 50 | ILLBEF2 | ILLNESS BEFORE PREGNANCY 2<br>AS FOR ILLBEF1 | |
| 51 | ILLDUR1 | ILLNESS DURING PREGNANCY<br>Record illnesses with chronic or acute onset during first half of the pregnancy including asymptomatic maternal infections.<br><br>(Any additional details may be entered in the general comments section, variable 80). For maternal infections, use chapters A and B of the ICD 10 coding (4 digits). Fetal infections and associated malformations should be coded under syndrome and malformation 1-8 codes (variable 27-44). Do not insert the decimal point in the code (eg. Code B34.1 as B341)<br>Coxsackie's B341<br>Cytomegalic Inclusion Diseases B250 - B259<br>Herpes Simplex B000 - B009<br>HIV (AIDS) B200 - B249<br>Influenza J100 - J119<br>Listeria A320 - A329<br>Mumps B260 - B269<br>Rubella B060 - B069<br>Syphillis A530 - A539<br>Toxoplasmosis B580 - B589<br>Varicella (Chicken Pox) B010 - B019<br>Viral Hepatitis B190 - B199 | ICD 10<br>0 = No<br>1 = Yes, but no information available<br>9 = Not known |

## Exposure (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|---|---|---|---|
| 52 | ILLDUR2 | ILLNESS DURING PREGNANCY<br>AS FOR ILLDUR1 | |
| 53 | FOLIC | FOLIC ACID SUPPLEMENTATION<br>Recommend to your local maternity hospitals or midwives to collect these data.<br><br>Regularly means at least 4 times per week.<br><br>Folic acid supplementations include folic acid only tablets or a multivitamin preparation which contains folic acid.<br><br>Code 8 refers to cases where the mother was not asked or the information was not recorded, although she attended a service which normally records this information. If your registry directly interviews mothers for this information, record 8 if the information is missing for this particular mother.<br><br>Code 9 refers to mothers attending services where this information is not recorded or not available to the Registry. | 1 = At least 0.4 milligrams folic acid supplement taken regularly, starting preconceptionally<br>2 = Folic acid supplement taken irregularly or starting post-conceptionally or at an unknown dose or a dose less than 0.4 milligrams<br>3 = No folic acid supplement taken<br>8 = Folic acid use not recorded for this woman<br>9 = Folic acid use not recorded by this maternity unit/clinician |
| 54 | DRUGS1 | DRUGS - 7 DIGITS MAXIMUM<br>Record any drug taken by the mother during the first trimester of pregnancy (from the 1st day of last menstrual period up to the 12th week of gestation). Drugs with long elimination half time and taken before conception should also be recorded (eg. Acitretin, etretinate etc).<br><br>If it is not known in which trimester the drug was taken, and this information cannot be obtained, code it but write in the space for comments that it is not sure whether the drug was taken in the first trimester.<br><br>Use ATC-coding and use as many digits as possible (from 3 to 7). Website http://www.whocc.no/atcddd/.<br><br>Do not record usual vitamins and mineral supplementation, but record unusual intakes of vitamins or minerals (eg. Vitamin A mega doses). The ATC coding system does not have a code for alternative drugs or herbs. If these are used, give the main code Z.<br><br>ATC example:<br>N03A: antiepileptic drug<br>N03AF01: carbamazepine | 0 = No drugs used<br>9 = Not known |

| Variable Number | Variable Name | Explanation and Instructions | Code |
|---|---|---|---|
| 54 (cnt'd) | | DRUGS – 7 DIGITS MAXIMUM (CNT'D)<br>Details on the dosage and timing should be given in text variable 55. Do not forget to mention in the appropriate section (disease during or before pregnancy) the indication for drug use.<br><br>Only drugs taken at physiologic doses to be recorded.<br><br>If a drug overdose or self-poisoning, this MUST be explained in text in Variable 55. | |
| 55 | SP_DRUGS1 | SPECIFY DRUG EXPOSURES | Free text |
| 56 | DRUGS2 | AS FOR DRUGS1<br>Please give details in text variable 57 SP_DRUGS2. | As for DRUGS1 |
| 57 | SP_DRUGS2 | SPECIFY DRUG EXPOSURES | Free text |
| 58 | DRUGS3 | AS FOR DRUGS1<br>Please give details in text variable 59 SP_DRUGS3. | As for DRUGS1 |
| 59 | SP_DRUGS3 | SPECIFY DRUG EXPOSURES | Free text |
| 60 | DRUGS4 | AS FOR DRUGS1<br>Please give details in text variable 61 SP_DRUGS3. | |
| 61 | SP_DRUGS4 | SPECIFY DRUG EXPOSURES | |
| 62 | DRUGS5 | AS FOR DRUGS1<br>Please give details in text variable 63 SP_DRUGS3. | |
| 63 | SP_DRUGS5 | SPECIFY DRUG EXPOSURES | |

## Family History (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|---|---|---|---|
| 64 | CONSANG | CONSANGUINITY<br>Restrictive definition of consanguinity: where the parents of the malformed case have one or more ancestors in common no more remote than a great-grandparent (=second cousins) | 0 = Not related or relationship more distant than second cousin<br>1 = Relationship of second cousin or closer<br>9 = Not known |
| 65 | SP_CONSANG | SPECIFY TEXT INFORMATION ON CONSANGUINITY | Free text |
| 66 | SIBANOM | SIBS WITH ANOMALIES<br>If the sibling (including twin) was notified to EUROCAT fill in variables 68-71 below. Make sure that the local identification numbers given correspond to those in the central database; otherwise give more information in text here.<br><br>If previous siblings were not notified to EUROCAT specify in text SP_SIBANOM the year of birth and malformations of each sibling.<br><br>If one sibling has both the same anomaly and a different anomaly, code under "same". If one sibling has the same anomaly and another sibling has a different anomaly, code under "same and other"<br><br>Always give details in text variable 66 SP_SIBANOM | 1 = Same<br>2 = Other<br>3 = Same and other<br>4 = No<br>9 = Not known |
| 67 | SP_SIBANOM | SPECIFY TYPE OF ANOMALY OF SIBLINGS | Free text |
| 68 | PREVSIB | PREVIOUS MALFORMED SIBLINGS NOTIFIED TO EUROCAT<br>If yes, give the local ID number in variables SIB1, SIB2 or SIB3 (variables 69-71).<br><br>Include malformed co-twins or siblings from the same pregnancy, irrespective of birth order within multiple set.<br><br>Exclude, conjoined twin. | 1 = Yes<br>2 = No<br>9 = Not known |
| 69 | SIB1 | SIB LOCAL ID NUMBER NOTIFIED TO THE CENTRAL REGISTRY<br>Enter here also the code numbers of co-twins or siblings from the same pregnancy, irrespective of birth order within multiple sets.<br><br>Leave blank if no previous siblings notified to EUROCAT. | Local ID |
| 70 | SIB2 | As SIB1 | Local ID |
| 71 | SIB3 | As SIB1 | Local ID |

## Family History (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|---|---|---|---|
| 72 | MOANOM | MOTHER'S FAMILY WITH ANOMALIES<br>Include mother herself as well as mother's family. Specify type of anomaly in written text and relation to the infant.<br><br>If the aetiology is known, "same" means the same aetiology, even if the spectrum of malformations present is slightly different.<br><br>If the aetiology is unknown or multifactorial, "same" is a matter of judgment by a qualified coder, but full specification of the anomaly should be given, whether other or the same.<br><br>"Same and other" refers to two different relatives. If a relative has both the same and another anomaly, code "same".<br><br>Restrict the family to first, second and third degree relatives (mother, father, siblings, grandparents, aunts, uncles, half-siblings, first cousins).<br><br>Always give details in text variable 73 SP_MOANOM. | 1 = Same<br>2 = Other<br>3 = Same and other<br>4 = No<br>9 = Not known |
| 73 | SP_MOANOM | SPECIFY TYPE OF ANOMALY AND DESCRIBE THE MALFORMATION | Free text |
| 74 | FAANOM | FATHER'S FAMILY WITH ANOMALIES<br>As MOANOM<br><br>Please give details in text variable 75 SP_FAANOM | As MOANOM |
| 75 | SP_FAANOM | SPECIFY TYPE OF ANOMALY AND DESCRIBE THE MALFORMATION | Free text |

## Sociodemographic (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|---|---|---|---|
| 76 | MATEDU | MATERNAL EDUCATION<br>Refer to International Standard Classification of Education 1997 for more information and Kunst et al (2001).<br><br>Assign according to the highest level of education completed (or for full-time students, level in progress).<br><br>Elementary and lower secondary refers to the period of compulsory education, usually to age 15/16. Upper secondary refers to the last two school or college years (usually to age 18) preparing students for tertiary education or the workforce. Tertiary refers to Bachelor's degree (English), Diploma (German), License (French) or equivalent, and to higher degrees (eg. doctorates), or to other forms of higher education. | 1 = Elementary and lower secondary<br>2 = Upper secondary<br>3 = Tertiary<br>9 = Not known |
| 77 | SOCM | SOCIOECONOMIC STATUS OF MOTHER<br>Current or last occupation.<br><br>Upper non-manual – professionals, administrators and managers eg. doctor, architect, lawyer, banker, manager, teacher, nurse, performer.<br>Lower non-manual – routine non-manual eg. Book-keeper, salesman, receptionist, secretary, computer operator, clerk, waiter.<br>Skilled manual – cook, butcher, carpenter.<br>Unskilled manual – semi and unskilled manual eg. factory worker, driver, agricultural worker, porter.<br>Self employed/artisan – owner of shop, restaurant or hotel, independent artisan.<br>Farmer – eg. self-employed farmer or fisherman.<br><br>If code 8 ("other/student"), please specify in text in space for general comments (variable 80).<br><br>For further information see Kunst et al (2001) | 1 = Upper non-manual<br>2 = Lower non-manual<br>3 = Skilled manual<br>4 = Unskilled manual<br>5 – Self employed/artisan<br>6 = Farmer<br>8 = Other/Student<br>9 = Not known |
| 78 | SOCF | SOCIOECONOMIC STATUS OF FATHER<br>As SOCM. | 0 = Single mother, no father recorded<br>1 = Upper non-manual<br>2 = Lower non-manual<br>3 = Skilled manual<br>4 = Unskilled manual<br>5 – Self employed/artisan<br>6 = Farmer<br>8 = Other/Student<br>9 = Not known |

## Sociodemographic (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|---|---|---|---|
| 79 | MIGRANT | MIGRANT STATUS<br>This variable is included to allow assessment of the extent to which services such as prenatal screening are reaching migrants. It does not ask for ethnicity.<br><br>If code 4, give text details in the general comments section (variable 79). | 1 = Mother migrated from outside EU during pregnancy<br>2 = Mother migrated from outside EU during adult life (from age 18)<br>3 = Mother not a migrant as defined in 1 or 2<br>4 = Other (specify in text)<br>9 = Not known |

Footnote: Kunst AE, Bos V, Mackenbach JP and the EU Working Group on Socio-economic Inequality in Health, "Monitoring Socio-Economic Inequalities in Health in the European Union: Guidelines and Illustrations", A Report to the Health Monitoring Programme of the European Commission.

## General Comments (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|---|---|---|---|
| 80 | GENREM | GENERAL ADDITIONAL COMMENTS | Free text |

## Extra Drugs (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|---|---|---|---|
| 81 | EXTRA_DRUGS | EXTRA DRUGS<br>This field is only to be used if drug fields 1-5 have already been filled.<br><br>Record any drug taken by the mother during the first trimester of pregnancy (from the 1st day of last menstrual period up to the 12th week of gestation). Drugs with long elimination half time and taken before conception should also be recorded (eg. Acitretin, etretinate etc).<br>If it is not known in which trimester the drug was taken, and this information cannot be obtained, code it but write in the space for comments that it is not sure whether the drug was taken in the first trimester.<br><br>Use ATC-coding and use as many digits as possible (from 3 to 7). Website http://www.whocc.no/atcddd/.<br><br>Do not record usual vitamins and mineral supplementation, but record unusual intakes of vitamins or minerals (eg. Vitamin A mega doses). The ATC coding system does not have a code for alternative drugs or herbs. If these are used, give the main code Z.<br><br>ATC example:<br>N03A: antiepileptic drug<br>N03AF01: carbamazepine<br><br>Details on the dosage and timing should be given in the drug description. Do not forget to mention in the appropriate section (disease during or before pregnancy) the indication for drug use.<br><br>Only drugs taken at physiologic doses to be recorded.<br><br>If a drug overdose or self-poisoning, this MUST be explained in the drug description.<br><br>If importing data from a local program, enter the ATC code and text description in the following format:<br><br><ATC code\|text description><br><br>If more than one extra drug is to be imported for a | |

| | | | |
|---|---|---|---|
| | | single case, then enter the ATC codes in the extra drugs field as follows:<br><br><ATC code\|text description><ATC code\|text description><br><br>For example a case with valproate and lamotrigine exposure is entered in the extra_drugs field as: <N03AG01\|Valproate><N03AX09\|Lamotrigine><br><br>(See chapter 2.4 of EDMP User Guide for further guidance) | |

# Chapter 2.2.2

# EDMP Derived Variables
# (Revised January 2012)

## 2.2.2 EDMP Derived Variables

| Variable | Description |
|---|---|
| erec | Case number generated by Central Registry database |
| byear | Baby: Year of birth |
| bunknown | Baby: Birth date not known |
| ddyear | Baby: Date of death (year) |
| dsyear | Baby: Date of discovery (year) |
| dayear | Mother's date of birth (year) |
| dfyear | Father's date of birth (year) used for cases born before 2005 |
| bwconfirm | Birth weight confirmed where gestational age outside normal range (see chapter 2.5) |
| gaconfirm | Gestational age confirmed where birthweight outside normal range (see chapter 2.5) |
| dobmconfirm | Mother's DOB confirmed where outside normal range (see chapter 2.5) |
| death_age | Create age at death in days from the baby's date of birth and date of death |
| mult_malf | Provisional identification of multiply malformed cases according to EUROCAT's multiple malformation algorithm (see Chapter 3.4) |
| corenon | Core data / non core data exported to Central Registry (1 = core only, 2 = core and non-core) |
| tot_malf | Total number of valid malformations, non-zero, non-blank and at least one letter and two digits (ICD10) |
| tot_minor | Total number of minor malformations (see chapter 3.2) |
| other_in_chapter | Total number of malformations in ICD10 (Q Chapter) that are not included in EUROCAT's congenital anomaly subgroups, but are included in the total congenital anomaly case counts |
| out_of_chapter | Total number of malformations outside ICD10 (Q Chapter) that are not included in EUROCAT's congenital anomaly subgroups, but are included in the total congenital anomaly case counts |
| birth_type | Definitions of stillbirths and spontaneous abortions vary between regions. This variable recodes birth type according to EUROCAT's specifications: cases with gestational age <20 weeks are re-coded as spontaneous abortions, cases with gestational age >=20 weeks are re-coded as "stillbirths" (irrespective of the local definition of stillbirth/spontaneous abortion). |
| casestatus | 0 = Not a EUROCAT case. This includes cases with only minor malformations (see Chapter 3.2)<br><br>1 = Case with one or more EUROCAT subgroups. This includes all cases with an ICD10 code specified in EUROCAT's congenital anomaly subgroups (see chapter 3.3) and al1-al95 below.<br><br>2 = Case with ICD10 Q chapter code not allocated to EUROCAT subgroup. This includes any case not already classified under casestatus = 0 or 1 with a congenital anomaly ICD10 code (within Q chapter) that is not included in one of EUROCAT's congenital anomaly subgroups<br><br>3 = Case with ICD10 code outside Q chapter not allocated to EUROCAT subgroup. This includes any case not already classified under casestatus = 0, 1 or 2 with a congenital anomaly code that is outside the ICD10 Q chapter, that is not included in one of EUROCAT's congenital anomaly subgroups. |
| al1 to al108 | EUROCAT subgroups: (0 = No, 1 = Yes). Based on EUROCAT coding (Chapter 3.3 of this Guide) |
| al1 | EUROCAT subgroup: All Anomalies |
| al2 | EUROCAT subgroup: Nervous system |
| al3 | EUROCAT subgroup: Neural Tube Defects |
| al4 | EUROCAT subgroup: Anencephalus and similar |

| Variable | Description |
|---|---|
| al5 | EUROCAT subgroup: Encephalocele |
| al6 | EUROCAT subgroup: Spina Bifida |
| al7 | EUROCAT subgroup: Hydrocephaly |
| al8 | EUROCAT subgroup: Microcephaly |
| al9 | EUROCAT subgroup: Arhinencephaly/holoprosencephaly |
| al10 | EUROCAT subgroup: Eye |
| al11 | EUROCAT subgroup: Anophthalmos/microphthalmos |
| al12 | EUROCAT subgroup: Anophthalmos |
| al13 | EUROCAT subgroup: Congenital cataract |
| al14 | EUROCAT subgroup: Congenital glaucoma |
| al15 | EUROCAT subgroup: Ear, face and neck |
| al16 | EUROCAT subgroup: Anotia |
| al17 | EUROCAT subgroup: Congenital heart disease |
| al97 | EUROCAT subgroup: Severe CHD |
| al18 | EUROCAT subgroup: Common arterial truncus |
| al19 | EUROCAT subgroup: Transposition of great vessels (complete) |
| al20 | EUROCAT subgroup: Single ventricle |
| al21 | EUROCAT subgroup: Ventricular septal defect |
| al22 | EUROCAT subgroup: Atrial septal defect |
| al23 | EUROCAT subgroup: Atrioventricular septal defect |
| al24 | EUROCAT subgroup: Tetralogy of Fallot |
| al25 | EUROCAT subgroup: Tricuspid atresia and stenosis |
| al26 | EUROCAT subgroup: Ebstein's anomaly |
| al27 | EUROCAT subgroup: Pulmonary valve stenosis |
| al28 | EUROCAT subgroup: Pulmonary valve atresia |
| al29 | EUROCAT subgroup: Aortic valve atresia/stenosis |
| al30 | EUROCAT subgroup: Hypoplastic left heart |
| al31 | EUROCAT subgroup: Hypoplastic right heart |
| al32 | EUROCAT subgroup: Coarctation of aorta |
| al33 | EUROCAT subgroup: Total anomalous pulm venous return |
| al100 | EUROCAT subgroup: PDA as only CHD in term infants (>=37 weeks) |
| al34 | EUROCAT subgroup: Respiratory |
| al35 | EUROCAT subgroup: Choanal atresia |
| al36 | EUROCAT subgroup: Cystic adenomatous malf of lung |
| al101 | EUROCAT subgroup: Oro-facial clefts |
| al102 | EUROCAT subgroup: Cleft lip with or without palate |
| al103 | EUROCAT subgroup: Cleft palate |
| al40 | EUROCAT subgroup: Digestive system |
| al41 | EUROCAT subgroup: Oesophageal atresia with or without tracheo-oesophageal fistula |
| al42 | EUROCAT subgroup: Duodenal atresia or stenosis |
| al43 | EUROCAT subgroup: Atresia or stenosis of other parts of small intestine |
| al44 | EUROCAT subgroup: Ano-rectal atresia and stenosis |
| al45 | EUROCAT subgroup: Hirschsprung's disease |
| al46 | EUROCAT subgroup: Atresia of bile ducts |
| al47 | EUROCAT subgroup: Annular pancreas |
| al48 | EUROCAT subgroup: Diaphragmatic hernia |
| al49 | EUROCAT subgroup: Abdominal wall defects |
| al50 | EUROCAT subgroup: Gastroschisis |

| Variable | Description |
|---|---|
| al51 | EUROCAT subgroup: Omphalocele |
| al52 | EUROCAT subgroup: Urinary |
| al53 | EUROCAT subgroup: Bilateral renal agenesis including Potter syndrome |
| al54 | EUROCAT subgroup: Renal Dysplasia |
| al55 | EUROCAT subgroup: Congenital hydronephrosis |
| al56 | EUROCAT subgroup: Bladder exstrophy and/or epispadia |
| al57 | EUROCAT subgroup: Posterior urethral valve and/or prune belly |
| al58 | EUROCAT subgroup: Genital |
| al59 | EUROCAT subgroup: Hypospadias |
| al60 | EUROCAT subgroup: Indeterminate sex |
| al61 | EUROCAT subgroup: Limb |
| al62 | EUROCAT subgroup: Limb reduction |
| al63 | EUROCAT subgroup: Upper limb reduction |
| al64 | EUROCAT subgroup: Lower limb reduction |
| al65 | EUROCAT subgroup: Complete absence of a limb |
| al66 | EUROCAT subgroup: Club foot - talipes equinovarus |
| al67 | EUROCAT subgroup: Hip dislocation and/or dysplasia |
| al68 | EUROCAT subgroup: Polydactyly |
| al69 | EUROCAT subgroup: Syndactyly |
| al104 | EUROCAT subgroup: Skeletal dysplasias |
| al75 | EUROCAT subgroup: Craniosynostosis |
| al76 | EUROCAT subgroup: Congenital constriction bands/amniotic band |
| al79 | EUROCAT subgroup: Situs inversus |
| al80 | EUROCAT subgroup: Conjoined twins |
| al81 | EUROCAT subgroup: Congenital skin disorders |
| al82 | EUROCAT subgroup: Teratogenic syndromes with malformations |
| al83 | EUROCAT subgroup: Fetal alcohol syndrome |
| al84 | EUROCAT subgroup: Valproate syndrome |
| al86 | EUROCAT subgroup: Maternal infections resulting in malformations |
| al105 | EUROCAT subgroup: Genetic syndromes + microdeletions |
| al108 | EUROCAT subgroup: Sequences |
| al88 | EUROCAT subgroup: Chromosomal |
| al89 | EUROCAT subgroup: Down syndrome |
| al90 | EUROCAT subgroup: Patau syndrome/trisomy 13 |
| al91 | EUROCAT subgroup: Edward syndrome/trisomy 18 |
| al92 | EUROCAT subgroup: Turner syndrome |
| al93 | EUROCAT subgroup: Klinefelter syndrome |

# Chapter 2.2.3

# Recommended Local Variables
# (Issued September 2005)

## 2.2.3 Recommended Local Variables

In addition to the variables described in Chapter 2.2.1, EUROCAT recommends that registries collect locally the following variables (not for transmission to the Central Registry).

| | |
|---|---|
| **Place of birth** | A code for each maternity unit and for home delivery. Selective referral of case to registry population. This variable is particularly important for those registries which are classified as Population-based II or III (see Chapter 5.1 for definition). This variable should identify cases for exclusion which were referred to a hospital within the registry area in order to received specialist services after prenatal diagnosis of malformation outside the registry hospitals. |
| **Prenatal diagnostic techniques** | Prenatal diagnostic tests and their results. A possible coding scheme can be found in EUROCAT Guide 1.2. It is important to distinguish whether the result of the test was positive or negative for malformation. Some registries may wish to identify which of multiple malformations were identified by any particular test. |
| **Previous pregnancies** | Record number of previous spontaneous abortions, induced abortions, stillbirths and livebirths separately. Remember that this should refer to the baby/fetus, not the pregnancy (eg. in the variable "previous pregnancies" in the main dataset, a twin pregnancy is counted once only. Here you would count a twin delivery twice ie. two livebirths, one live and one stillbirth). |
| **Age of father at delivery** | |
| **Geographic co-ordinates of mother's residence** | This is important for small area environmental studies or the identification of clustering. Consult local expertise in geographic information systems to decide on the coding to use. For example, there may be a postcoding system which can be later automatically translated to geographic co-ordinates, or you may wish to directly code latitude and longitude co-ordinates translated from the address. |
| **Sources of ascertainment - spontaneous** | This is an important variable for the management and quality assessment of your registry. You should record which sources of information notified the case (eg. maternity unit, paediatric surgery, cytogenetic laboratory, ultrasound department), devising a code for the difference sources of information used by your registry (see chapter 5.2 for more examples in the Registry Description Questionnaire). For example, if the co-ordinator at a maternity unity gives you a list of recent malformed births including this case, or if you consult the entire maternity records to obtain a list of cases including this case, then code the maternity unit as a source of information. If the same case is also found on a list from the paediatric surgery department, then code paediatric surgery as another source of ascertainment. It should be possible to calculate the proportion of cases ascertained from more than one source of notification each year. Generally a high proportion of "multiply ascertained" cases are an indicator of high data quality. |
| **Source of ascertainment - confirmatory** | Use the same code as above, but code here the sources which confirmed the case or gave you extra information on request. For example, if the maternity unit notified a case and told you that he/she had died and you then request a post mortem report for this specific child, then the autopsy department should be coded as a confirmatory, not a spontaneous, source of information. |
| **Maternal smoking** | Code smoking during first trimester eg. Number of cigarettes per day. Make sure that the code you use can distinguish high levels of smoking from yes/no. |
| **Maternal alcohol use** | Code alcohol intake during first trimester, making sure that alcoholism and high alcohol intake can be distinguished. If the child has fetal alcohol's syndrome, code under malformation using ICD code (Q860). |
| **When first reported** | |

# Chapter 2.2.4

# Template for Associate Member Registry Data Transmission (Revised August 2011)

# Template for Associate Member Registry Data Transmission

## Form for Collecting Year XXXX Data from Associate Registries (Aggregate Data)

**Centre Number:**

**Denominator Data Year XXXX:**

Livebirths:
Stillbirths:
Total births:

**Births (live and still) by Maternal Age:**

<20
20-24
25-29
30-34
35-39
40-44
45+
Not known

| **Down Syndrome by Maternal Age:** | **LB** | **FD** | **TOPFA** | **Total cases** |
|---|---|---|---|---|
| <20 years | | | | |
| 20-24 | | | | |
| 25-29 | | | | |
| 30-34 | | | | |
| 35-39 | | | | |
| 40-44 | | | | |
| 45+ years | | | | |
| Total | | | | |

| | **Includes Chromosomal cases** | | | | **Excludes Chromosomal cases** | | | |
|---|---|---|---|---|---|---|---|---|
| **Gastroschisis by Maternal Age:** | **LB** | **FD** | **TOPFA** | **Total cases** | **LB** | **FD** | **TOPFA** | **Total non-chromosomal cases** |
| <20 years | | | | | | | | |
| 20-24 | | | | | | | | |
| 25-29 | | | | | | | | |
| 30-34 | | | | | | | | |
| 35-39 | | | | | | | | |
| 40-44 | | | | | | | | |
| 45+ years | | | | | | | | |
| Total | | | | | | | | |

| | Includes Chromosomal cases | | | | Excludes Chromosomal cases | | | |
|---|---|---|---|---|---|---|---|---|
| | No of LB | No of FD | No of TOPFA | Total cases | No of LB | No of FD | No of TOPFA | Total non-chromosomal cases |
| **All Anomalies** | | | | | | | | |
| **Nervous system** | | | | | | | | |
| Neural Tube Defects: | | | | | | | | |
| Anencephalus and similar | | | | | | | | |
| Encephalocele | | | | | | | | |
| Spina Bifida | | | | | | | | |
| Hydrocephaly | | | | | | | | |
| Microcephaly | | | | | | | | |
| Arhinencephaly / holoprosencephaly | | | | | | | | |
| **Eye** | | | | | | | | |
| Anophthalmos / microphthalmos | | | | | | | | |
| Anophthalmos | | | | | | | | |
| Congenital cataract | | | | | | | | |
| Congenital glaucoma | | | | | | | | |
| **Ear, face and neck** | | | | | | | | |
| Anotia | | | | | | | | |
| **Congenital Heart Defects (CHD)** | | | | | | | | |
| Severe CHD | | | | | | | | |
| Common arterial truncus | | | | | | | | |
| Transposition of great vessels | | | | | | | | |
| Single ventricle | | | | | | | | |
| VSD | | | | | | | | |
| ASD | | | | | | | | |
| AVSD | | | | | | | | |
| Tetralogy of Fallot | | | | | | | | |
| Triscuspid atresia and stenosis | | | | | | | | |
| Ebstein's anomaly | | | | | | | | |
| Pulmonary valve stenosis | | | | | | | | |
| Pulmonary valve atresia | | | | | | | | |
| Aortic valve atresia/stenosis | | | | | | | | |
| Hypoplastic left heart | | | | | | | | |
| Hypoplastic right heart | | | | | | | | |
| Coarctation of aorta | | | | | | | | |
| Total anomalous pulm venous return | | | | | | | | |
| PDA as **only** CHD in term infants (GA 37+ weeks) | | | | | | | | |
| **Respiratory** | | | | | | | | |
| Choanal atresia | | | | | | | | |
| Cystic adenomatous malf of lung | | | | | | | | |

| | Includes Chromosomal cases | | | | Excludes Chromosomal cases | | | |
|---|---|---|---|---|---|---|---|---|
| | No of LB | No of FD | No of TOPFA | Total cases | No of LB | No of FD | No of TOPFA | Total non-chromosomal cases |
| **Oro-facial clefts** | | | | | | | | |
| Cleft lip with or without cleft palate | | | | | | | | |
| Cleft palate | | | | | | | | |
| **Digestive system** | | | | | | | | |
| Oesophageal atresia with/without tracheo-oesophageal fistula | | | | | | | | |
| Duodenal atresia or stenosis | | | | | | | | |
| Atresia or stenosis of other parts of small intestine | | | | | | | | |
| Ano-rectal atresia and stenosis | | | | | | | | |
| Hirschsprung's disease | | | | | | | | |
| Atresia of bile ducts | | | | | | | | |
| Annular pancreas | | | | | | | | |
| Diaphragmatic hernia | | | | | | | | |
| **Abdominal wall defects** | | | | | | | | |
| Gastroschisis | | | | | | | | |
| Omphalocele | | | | | | | | |
| **Urinary** | | | | | | | | |
| *Bilateral* renal agenesis including Potter syndrome | | | | | | | | |
| Renal Dysplasia | | | | | | | | |
| Congenital hydronephrosis | | | | | | | | |
| Bladder exstrophy and/or epispadia | | | | | | | | |
| Posterior urethral valve and/or prune belly | | | | | | | | |
| **Genital** | | | | | | | | |
| Hypospadias | | | | | | | | |
| Indeterminate sex | | | | | | | | |
| **Limb** | | | | | | | | |
| Limb reduction | | | | | | | | |
| Upper limb reduction | | | | | | | | |
| Lower limb reduction | | | | | | | | |
| Complete absence of a limb | | | | | | | | |
| Club foot - talipes equinovarus | | | | | | | | |
| Hip dislocation and/or dysplasia | | | | | | | | |
| Polydactyly | | | | | | | | |
| Syndactyly | | | | | | | | |
| **Other anomalies/syndromes** | | | | | | | | |
| Skeletal dysplasias | | | | | | | | |
| Craniosynostosis | | | | | | | | |
| Congenital constriction bands/amniotic band | | | | | | | | |
| Situs inversus | | | | | | | | |
| Conjoined twins | | | | | | | | |
| Congenital skin disorders | | | | | | | | |
| Teratogenic syndromes with malformations | | | | | | | | |
| Fetal alcohol syndrome | | | | | | | | |
| Valproate syndrome | | | | | | | | |

| | Includes Chromosomal cases | | | | Excludes Chromosomal cases | | | |
|---|---|---|---|---|---|---|---|---|
| | **No of LB** | **No of FD** | **No of TOPFA** | **Total cases** | **No of LB** | **No of FD** | **No of TOFA** | **Total non-chromosomal cases** |
| Maternal infections resulting in malformations | | | | | | | | |
| Genetic syndromes + microdeletions | | | | | | | | |
| Sequences | | | | | | | | |
| **Chromosomal** | | | | | | | | |
| Down's syndrome | | | | | | | | |
| Patau syndrome/trisomy 13 | | | | | | | | |
| Edward syndrome/trisomy 18 | | | | | | | | |
| Turner's syndrome | | | | | | | | |
| Klinefelter's syndrome | | | | | | | | |

LB= Live births
FD= Still births and fetal deaths from 20 weeks gestation
TOPFA = Termination of pregnancy for fetal anomaly following prenatal diagnosis

* All Anomalies = ALL cases of congenital anomaly, excluding cases with only minor anomalies as defined in Chapter 3.2. Cases with more than one anomaly are only counted once in the "All Anomalies" subgroup.

# Chapter 2.3

# Template for Denominator Data
# (Issued September 2005)

## 2.3 Template for Denominator Data

Please send denominator data with every new year of case data.

| **Centre:** | | | | | |
|---|---|---|---|---|---|
| **Year** | | | | | |
| Livebirths | | | | | |
| Stillbirths | | | | | |
| **TOTAL** | | | | | |
| Please give definition of stillbirths: | | | | | |
| **Age distribution:** | | | | | |
| Mother <20 | | | | | |
| Mother 20-24 | | | | | |
| Mother 25-29 | | | | | |
| Mother 30-34 | | | | | |
| Mother 35-39 | | | | | |
| Mother 40-44* | | | | | |
| Mother 45+ | | | | | |
| Unknown | | | | | |
| **TOTAL**** | | | | | |
| * Does age group 40-44 include births in mothers aged 45 and over? (please delete as appropriate) Yes/No<br>** Are stillbirths included in the maternal age distribution? (please delete as appropriate) Yes/No | | | | | |
| **Monthly distribution:** | | | | | |
| January | | | | | |
| February | | | | | |
| March | | | | | |
| April | | | | | |
| May | | | | | |
| June | | | | | |
| July | | | | | |
| August | | | | | |
| September | | | | | |
| October | | | | | |
| November | | | | | |
| December | | | | | |
| **TOTAL*** | | | | | |
| * Are stillbirths included in the distribution by month? (please delete as appropriate) Yes/No | | | | | |

# Chapter 2.4

# EUROCAT Data Management Program (EDMP) Instructions (Revised February 2012)

# EUROCAT Data Management Program
# EDMP

## Version (5.10) 4/01/2012

## For Guide 1.3
## User Guide

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# 1 Introduction

The EUROCAT Data Management Program (EDMP) has been designed as a flexible tool to assist you in the collection, management, reporting and analysis of congenital anomaly data.

The major improvement in this new version of EDMP is the ability to enter (new data entry tab), or import, information on more than five drugs. Please see section 10.1 regarding import data format for the extra drugs. Other changes include the new anomaly subgroups. The output of data to Excel has been modified to ensure consistency, especially for date fields.

Printing to pdf

You can print any of the EDMP reports to pdf file if you install a pdf print to file driver. A typical example is Adobe Acrobat (not Adobe reader). Dopdf is a good free pdf print to file driver available from www.dopdf.com. Once installed simply select your pdf print to file driver instead of your default printer when you print your report (use Ctrl and P or choose File then Print from the menu to bring up the print dialog box) and enter a file name to save the report as.

Changes in EDMP to accommodate Guide 1.3

EUROCAT Guide 1.3 format is used for 2005 births onwards. EUROCAT Guide 1.2 format must still be used for cases born up to the end of 2004. EDMP will switch between Guide 1.2 and 1.3 formats depending upon the year of birth.

If you want to continue to use, at local level, some Guide 1.2 variables which are not included in Guide 1.3 they are available to you as local variables in the 2005+ data entry screen. You can also add up to 32 extra local variables of your choice. These must be the same for all years (before and after 2005). EDMP will automatically select which variables to export to the Central Registry.

You should not have any problems continuing to use all variables of your choice after 2005. If you are unsure, contact EUROCAT Central Registry.

*Please take note of the backing up details given below in section 3.*

## 2 System requirements and program details

The EDMP database has been written in Microsoft Access and will therefore only run on PCs that have either Access 2000, 2002, 2003 or Access 2007 installed. It is not possible to run the EDMP database using earlier versions of Access (e.g. Access versions 1, 2, 95 or 97). The program will automatically scale itself to fit any screen resolution from 800x600 up to1280x1024. Please note that support for Access 97 has been withdrawn.

You must also have a full version of Excel installed on your PC.

The program is comprised of two database files, one to store the data (Edmpdata.mdb) and the other to provide the user interface (Edmp.mde). You can use the EDMP program on single or networked PC.

## 3 Backup

You need to make regular backups of your data, ideally you should create a new backup at the end of each day that you have used the EDMP program. Remember to keep a recent backup at a different location to your PC in order to guard against fire or theft.

The data file you need to backup on a regular basis is Edmpdata.mdb. The option 'Data Location' available from the system menu will tell you where this file is on your PC or network. There are many methods and programs available to create your backups. Windows 95, 98, NT and XP provide backup utilities (Microsoft Backup) which can be used if you do not have access to any other third party backup utility. Microsoft Backup is not always installed on initial Windows set-up but can be installed using the add/remove programs option under the settings menu.

## 4 Getting started

### 4.1 Installation

There are separate installation instructions for users already using EDMP and for new users.

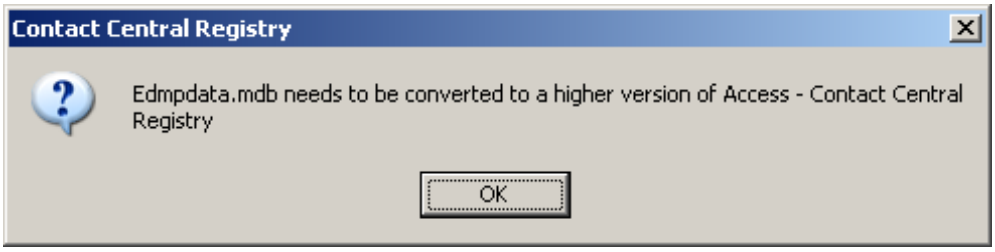
#### 4.1.1 Existing Users

**Before upgrading please make a backup of your data file Edmpdata.mdb.**

All you need to do is replace your existing copy of Edmp.mde. You can download Edmp.mde in various formats from the EUROCAT website www.eurocat-network.eu.

The first time you run the new version it will automatically apply any table updates to the data database file Edmpdata.mdb. If you are using a networked version of EDMP please ensure that no other EDMP programs are running at the same time.

If your existing Edmpdata.mdb data file is in Access 97 you will receive the following message:




If this happens you will need to send your data file Edmpdata.mdb to the Central Registry for conversion.

### 4.1.2 New Users

The EDMP program (EDMP.mde) can be downloaded from the EUROCAT website www.eurocat-network.eu. You will be sent a blank copy of the data file Edmpdata.mdb by Central Registry.

To install the EDMP program follow the instructions below.

a) Create a new folder on your hard drive using windows explorer or My computer. In this example we are using the folder called Edmp (C:\Edmp).
b) Copy the files Edmp.mde and Edmpdata.mdb to the newly created directory.
c) Create a shortcut by right clicking on Edmp.mde in windows explorer or My computer and then selecting 'create shortcut'. Drag and drop the shortcut onto your desktop.

Network installation

If you wish to install the EDMP program on a network then all you need to do is to follow the next two steps.

1) Move Edmpdata.mdb into a shared directory on the server and set any permissions as necessary.
2) Copy Edmp.mde onto each PC that will be running the program. Do not run Edmp.mde from the shared server directory, as there are a number of runtime processes that are individual to each session and are not suitable for sharing.

Although the EDMP program is network enabled it is not a true client/server version and will not provide satisfactory response times when used with dial-up remote access.

### 4.1.2 Access Security Settings

Access 2003 Security Settings - You may get a warning screen message regarding Macro Security when you run the program. To disable this message you need to close the program

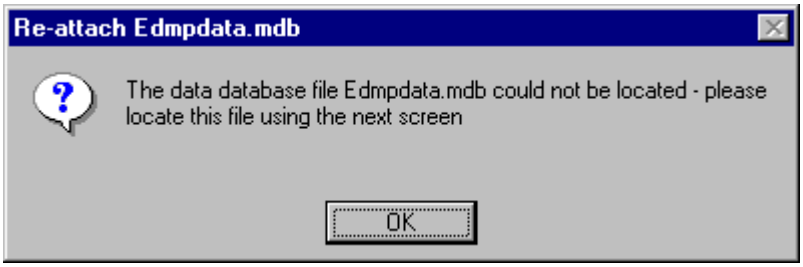
and then run Access without opening an existing database or creating a new one. Select the 'Tools' menu option then 'Macros' then 'Security' and choose the 'Low' setting.

Access 2007 Security Settings - You may get a warning screen message regarding Macro Security when you run program. To disable this message you need to close the program and then run Access without opening an existing database or creating a new one. Click on the Office button (round coloured button in the top left hand corner). At the bottom of the menu box click on the button labelled 'Access Options' then select 'Trust Center' then click on the 'Trust center settings' button. Now choose 'Macro settings' and select the 'Enable all macros' option.

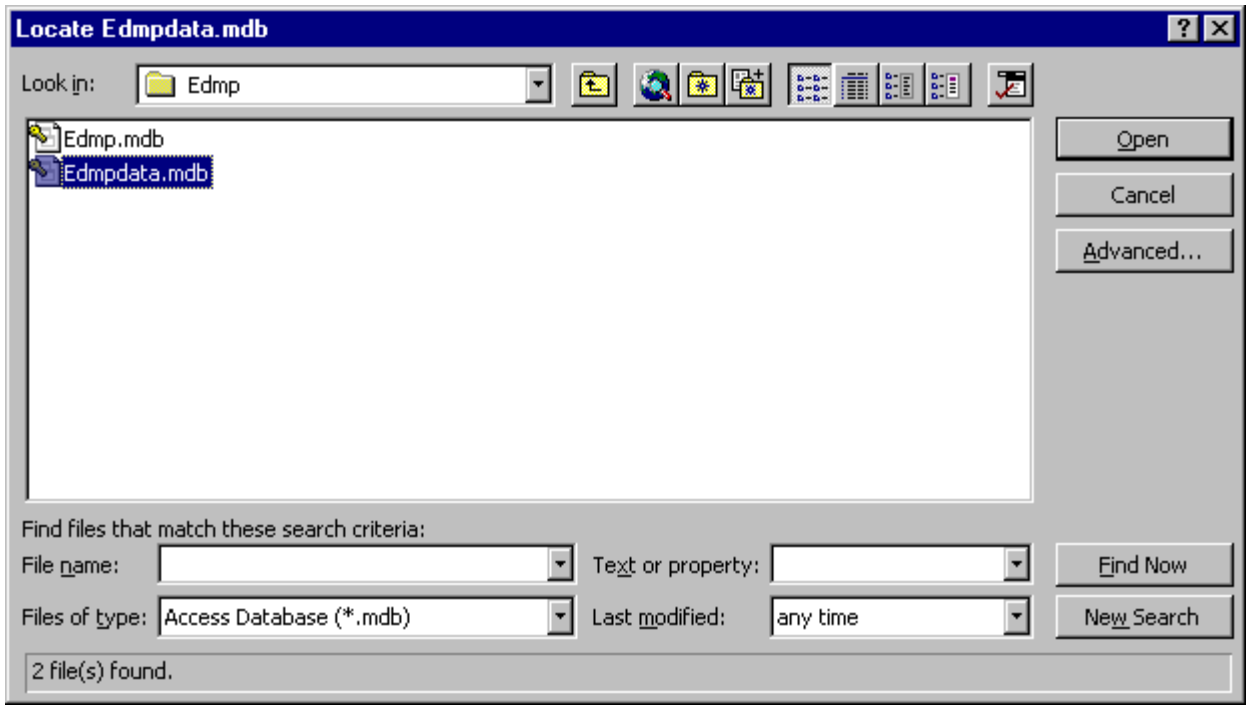
## 4.2 First time use

Logging into the EDMP program is normally a simple affair of typing in your username and password. However, the first time you run the program you may be prompted to locate Edmpdata.mdb (where the data is stored), specify your centre name and select the default printer.

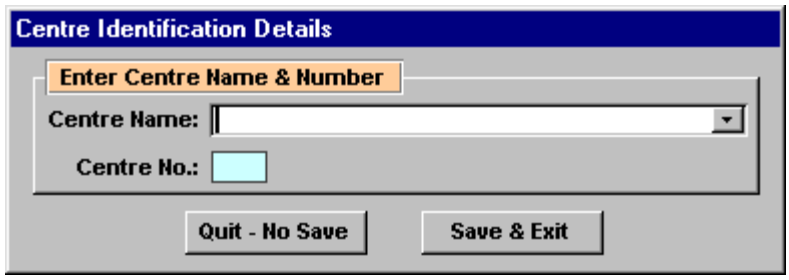
Each time you run the EDMP the first thing it does is to check to see that the program can locate the data file Edmpdata.mdb. If Edmpdata.mdb is not where it thinks it should be (i.e. after installation or if it has been moved to a different folder) then the following message will be displayed.




When you click on OK the following file selection screen will appear from which you select the folder and file (Edmpdata.mdb) in the usual Windows fashion and then click on the 'Open' button.



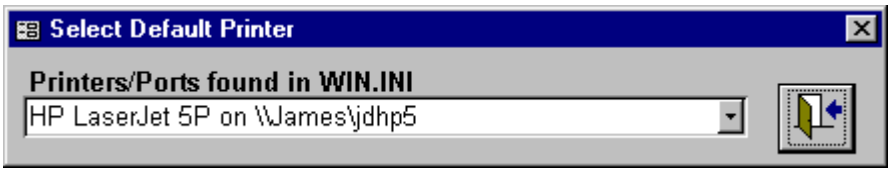

Once EDMP is happy that the data database has been located then you will need to select your centre from the list using the screen below.



Once you have selected your centre the login screen will be displayed. To login all you need to do is enter your username and password at the prompts provided. The program comes with the username '**m**' and password '**m**' already available. Please note that one of the first things that you should do is to add your own username and password and remove the installation defaults of '**m**' and '**m**' using the Users & Passwords facility under the System Menu (see below).

Normally the main menu is displayed after you have logged in successfully. However, sometimes (and usually after installation) the following message is displayed and this indicates that the default Windows printer has changed since the last time the program was run.

When this screen appears click on OK and then select the printer you require from the pull down list on the next screen. Once you have selected the required printer click on the exit button (button with door and arrow icon). All the printers, including network printers that are available to your PC will be displayed in the pull down list.




After you have logged in the main menu or navigation screen will be displayed.

## 5 Main menu

The main menu or navigation screen has been divided into logical sections. To access any of the functions simply click on the one you want. The cursor will change to a hand when it is positioned over an option that can be clicked on.

The new logical sections are:

- Manage Cases
- Analyse Data
- User Defined Categories
- Surveillance
- Import/Export Data
- Denominators
- System Menu

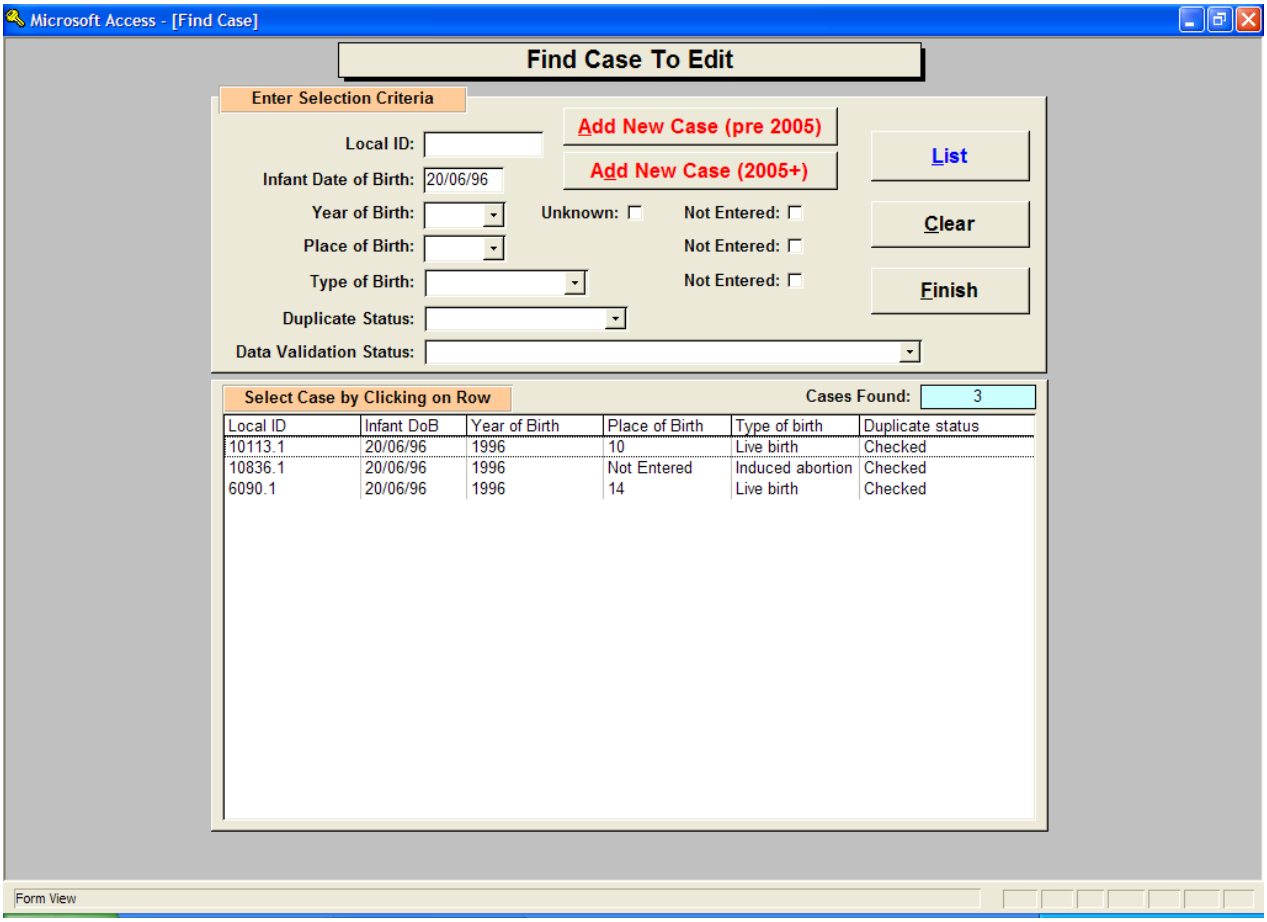
Access to each option will depend on individual permissions settings (see Manage Users under the System Menu.)

# 6 Manage Cases

## 6.1 Add / Edit Cases

In EDMP you use the 'Add / Edit Cases' menu option to both add new cases and also edit existing cases. You use the 'Find case to edit' screen, shown below, to either select an existing case to edit or to add a new case by clicking on the 'Add New Case' button. Please note the separate buttons for pre 2005 and 2005+ year of birth. The 2005+ option includes the new guide 1.3 variables. When you click on a row to edit a case EDMP will check the year of birth for that case and will use the correct data entry format which makes allowance for the new guide 1.3 variables.

The find case form is very easy to use. All you have to do is to enter any required selection criteria and then click on the 'List' button. Matching cases will be displayed in the list box and to edit a case simply click on the required row. In the example below three cases match the selection criteria of Infant DoB = 20/06/96.



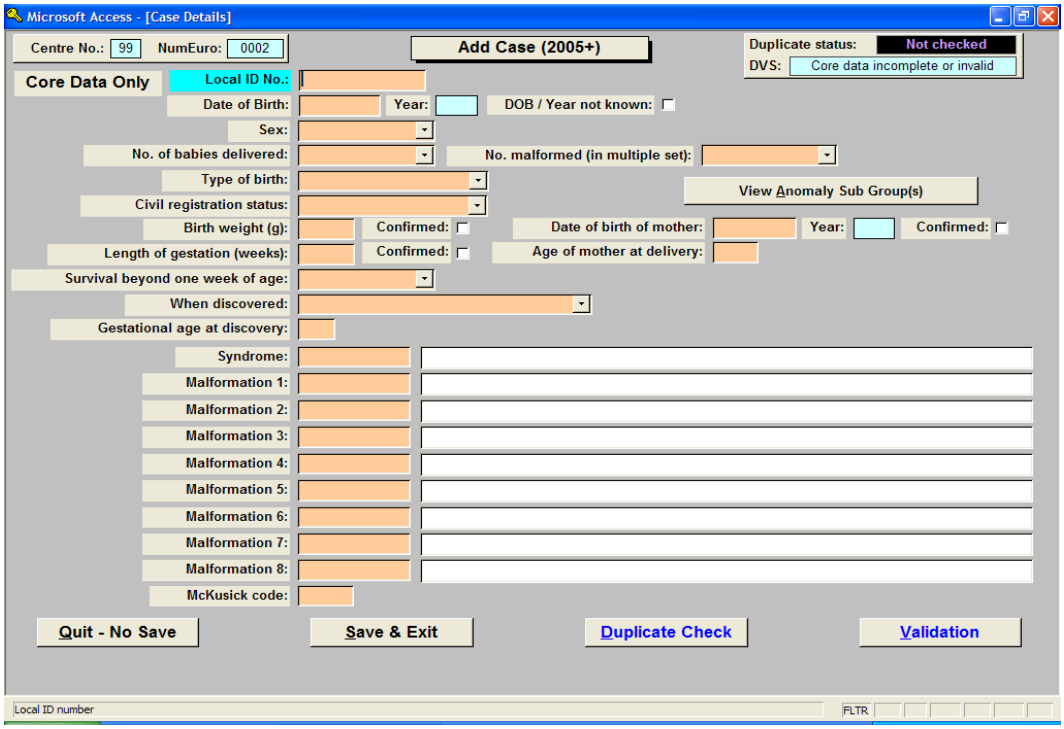

The EDMP display option chosen on the main menu (under System Menu) dictates which data entry form will be displayed. There are three different styles of data entry form, examples of each are shown below. The simplest is for the 'Core Data Only' option where there is a single page of data entry fields. The 'Core and Non-Core Data: Standard Format' screen is similar but has a number of pages which the user tabs through or selects by clicking on the required tab.

You navigate through the 'pages' of data by clicking on the relevant tab i.e. 'Infant', 'Mother' etc. This is also the same when editing or viewing a case. When viewing a case you cannot make changes to it, check for duplicates or do validation checking. Core data field backgrounds are highlighted in orange to make them easily identifiable. The page 'Local Variables' provides a number of fields that are for local use only and are not exported under the 'Export Data To EUROCAT' facility. It also includes five spare variables which you can rename to suit your own use.

Note in the example below the last page titled 'Extra Variables (1)'. There can be between none and two of these pages containing the extra variables (maximum of 32, 16 per page) as specified under the System Menu. As with the 'Local Variables' they are not included when exporting data to EUROCAT.

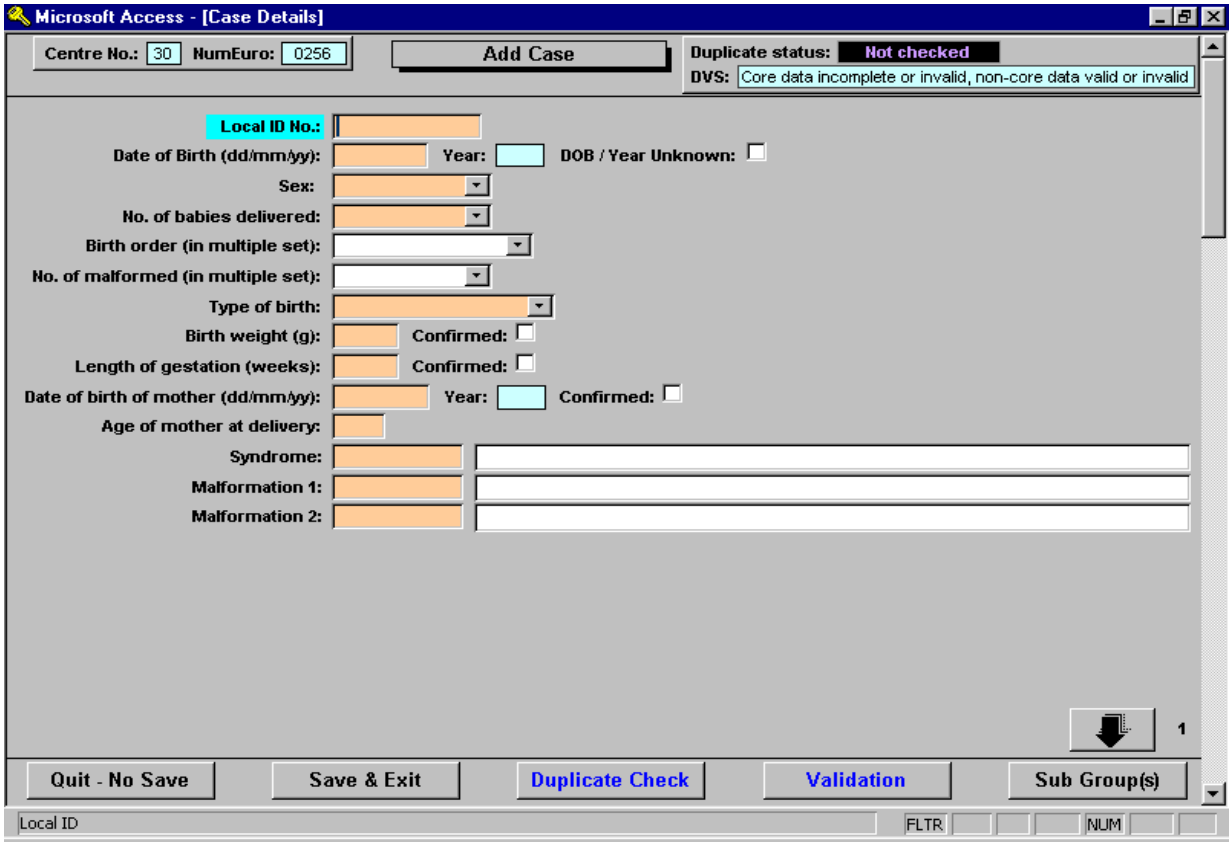
The final version of the data entry screen is for 'Core and Non-Core Data: User Defined Format' where the user has selected which variables (including any of the selected extra variables) to use and in which order to present them on screen

Core Data Only Screen (2005+)



# Core and Non-Core Data: Standard Format Screen (2005+)

Core and Non-Core Data: User Defined Format Screen (example)

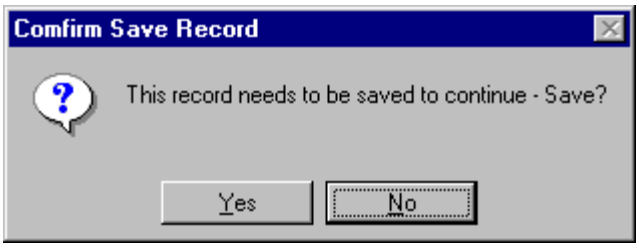



For the user defined data entry screen there are four ways to move between the pages. Firstly when you exit the last field of a page EDMP will automatically move you to the first field of the subsequent page (unless you use the mouse to click on a different part of the current page). Secondly there are 'up' and 'down' arrow buttons in the bottom right hand corner of each page which you can click on. Thirdly you can use the keyboard page up and page down keys and finally there is a vertical scroll bar on the right hand side of the screen.

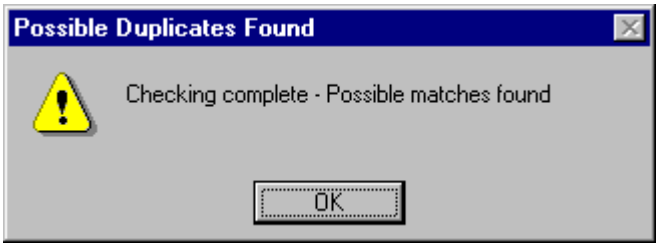
All the data entry screens offer the duplicate checking and validation buttons described below.

### 6.1.1 Duplicate Checking

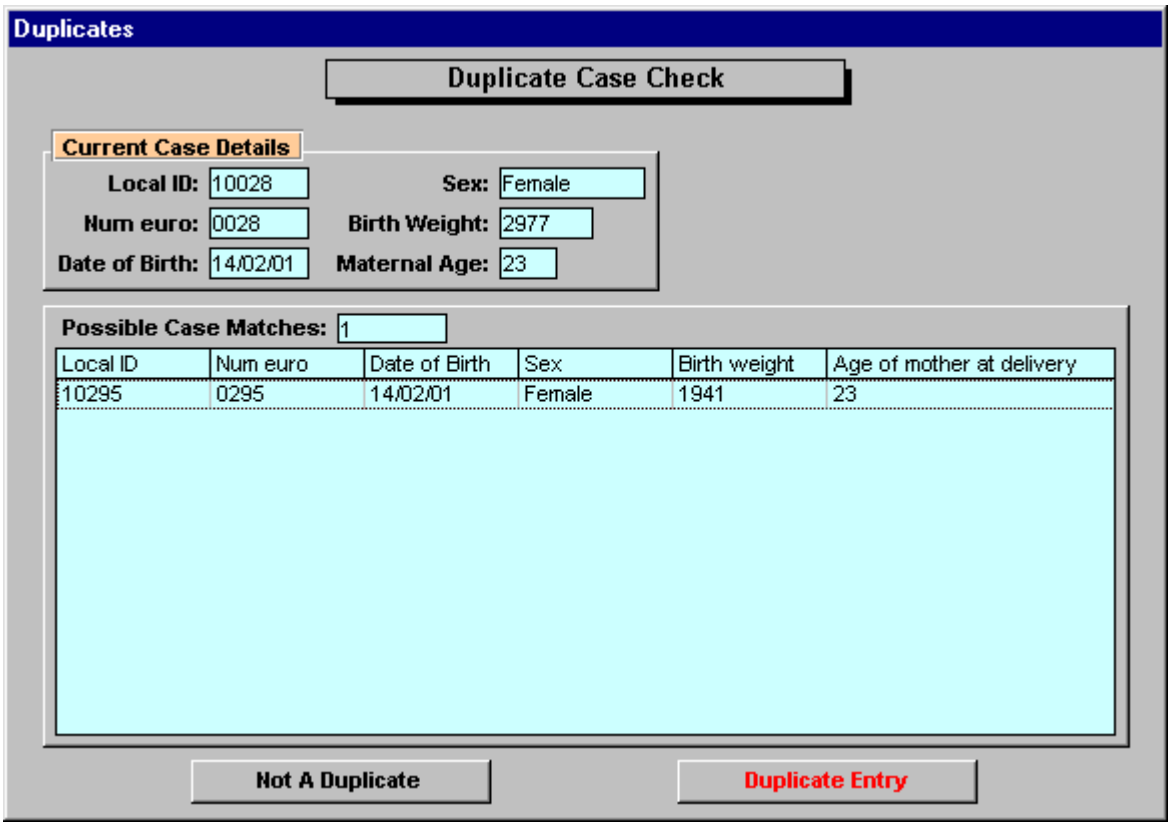
The 'Duplicate check' button when clicked checks for possible matches of other cases against the case you are currently adding or editing. You will be asked to save the record first as shown here:



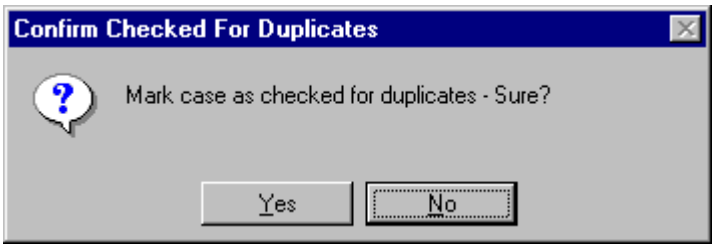

Clicking the 'Yes' Button then allows the checking to take place.
Once checking is completed if any matches are found a screen will pop up informing you that possible matches were found.




When you click the 'Ok' button a screen will be displayed showing the possible matches against the current case.

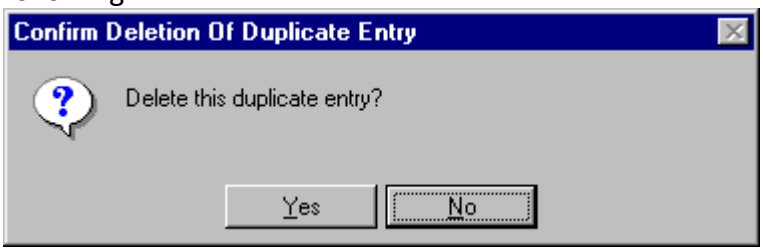


The screen above shows the current case at the top and then list's out the possible matches in the box below. As you can see from the above example there one possible match against the case, but it is not a match you would click the 'Not a duplicate' button. You would then be asked the following:




Hitting the 'Yes' button would mark the case as being checked for duplicates(no matches found).

If you had clicked on the 'Duplicate Entry' Button then you would have been asked the following:

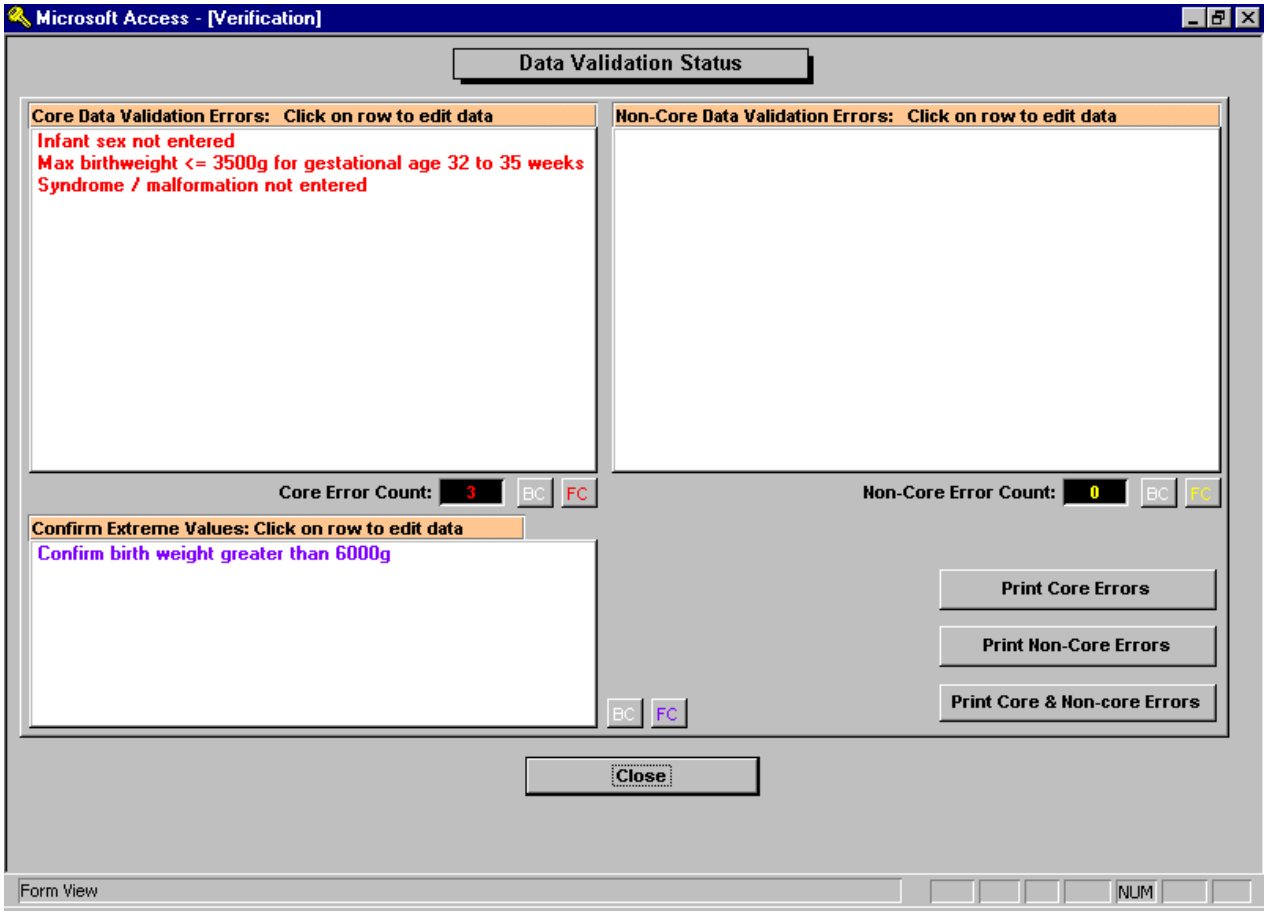



By hitting the 'Yes' button you would be confirming the deletion of the duplicate record.

### 6.1.2 Validation

Validation of the data runs checks on the data checking for possible errors.

The screen below shows the data validation status (DVS) of a case. As you can see from the example core data errors are shown in the top left box, non-core in the top right box and extreme value errors in the bottom left box. To go straight to the error just click on the relevant row. From this screen you also have the option to printout the errors for core and non-core data for that case. For extreme values there is a tick box by the relevant field to confirm that the value entered is correct. Once extreme values have been confirmed they will no longer be displayed as extreme at data validation.




## 6.2 View Existing Case

View existing case allows you to view case details, no editing, additions, duplicate checks or data validation can take place. Cases to view are selected in the same way as for editing a case.

## 6.3 Print Case

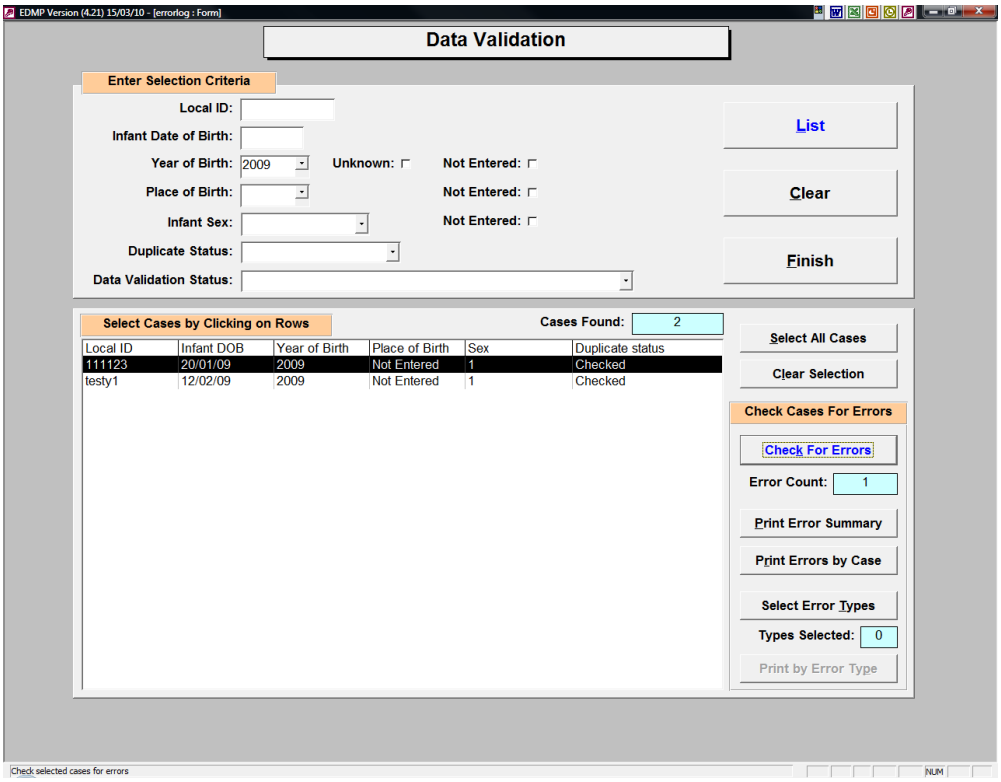
This provides a print out of an entire case where the amount of data printed is based on the current EDMP display option. All printouts and reports are displayed on the screen in preview mode. You can send the preview to the printer by clicking on the printer icon:

Alternatively you can send the report to Word, Excel or Notepad by clicking on the Office Links button:

## 6.4 Data Validation

In addition to the data validation available when you are adding or editing a case, EDMP allows you to multiple cases in one go. The data validation facility allows you to list all the Core, Non-Core and Extreme value errors for selected cases. Select the required cases to

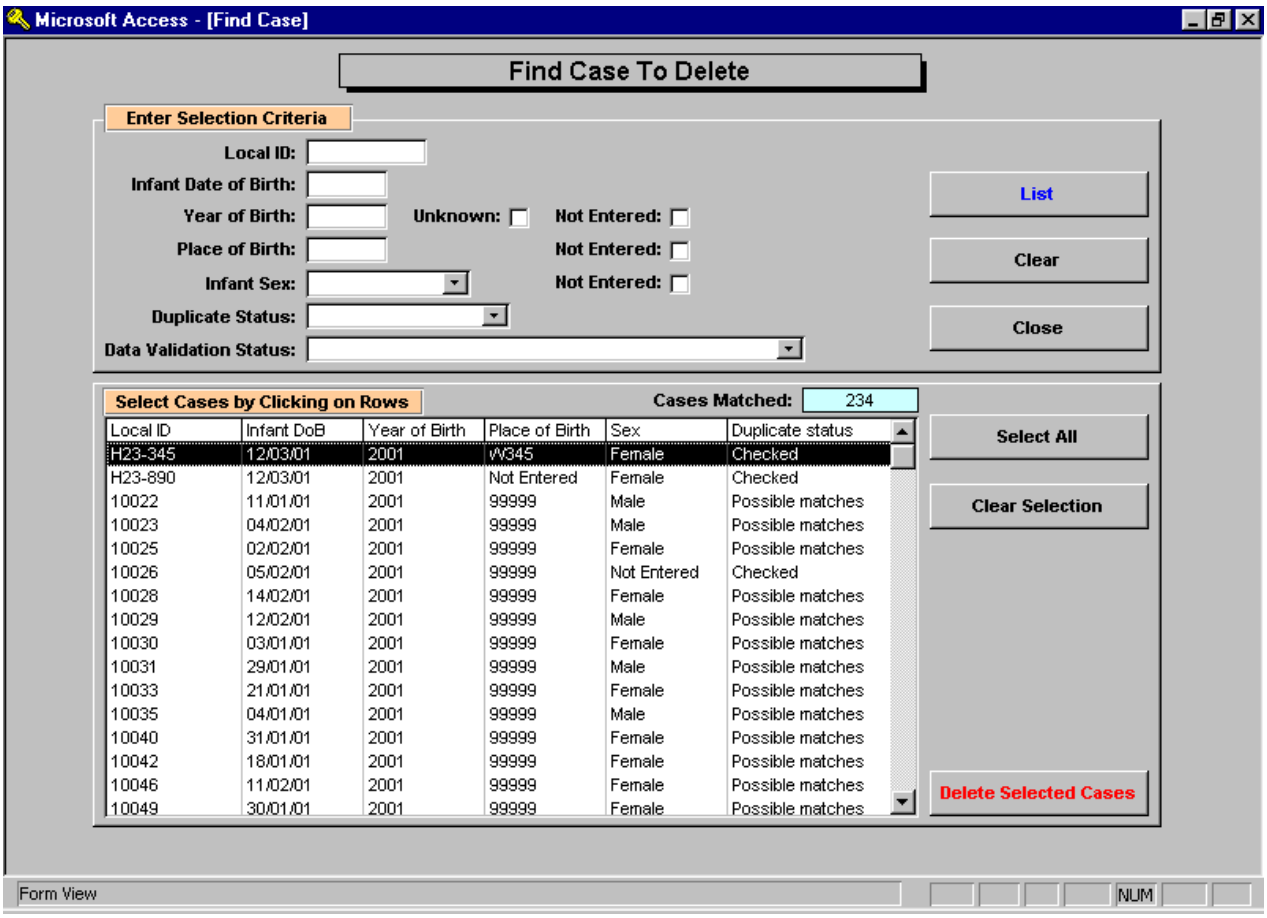
check using the screen shown below. Once you have selected the cases you can then print the error log either as a summary or as a list case by case using the 'Print Error Summary' and 'Print Errors By Case' buttons.




You can also select which of the found error type or types that you want to examine. Click on the 'Select Error Types' button and select one or more of the error categories listed and then print the error list again. EDMP will apply the correct checking rules depending upon whether each case relates to Guide 1.2 or 1.3.

## 6.5 Delete Case

The delete cases option allows you to delete records for selected case(s).
The list box, in the example below, shows case details. These include local ID number, year of birth, place of birth, sex of infant and duplicate status.
Select the case or cases you wish to delete and then click on the 'Delete Selected Cases' button. There is also a 'Select All' button which highlights / selects all cases. Please note that there is no 'undo' function available to recover deleted cases.




## 7 Analyse Data

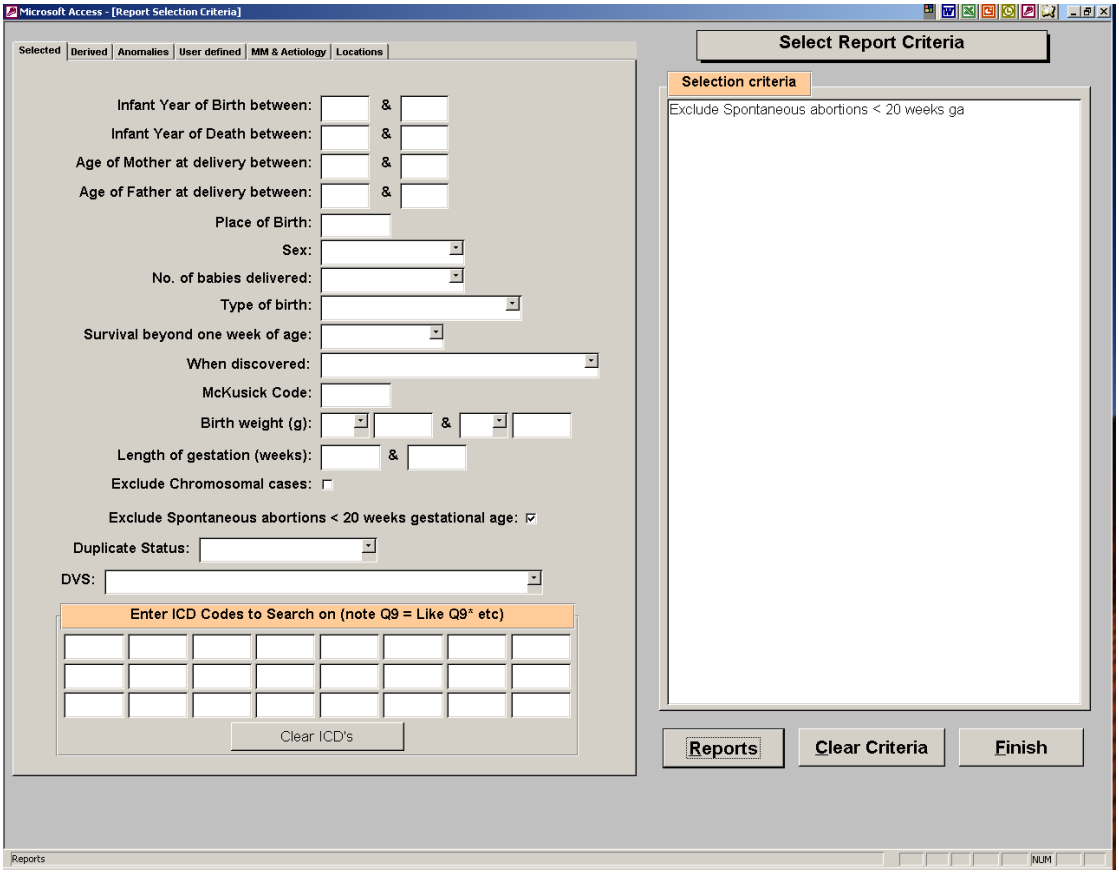
This section provides you with a simple and powerful way to analyse and describe your data. In EDMP there are now three categories of reports, List / Export Cases & Frequency Reports, Prevalence tables and missing values tabulation. Each category allows you to select which data to report on.

### 7.1 List / Export Cases & Frequency Reports

This option provides you with a selection criteria screen that allows you to report on defined subsets of your data. Once you have entered your selection criteria, if any, you can then run the standard reports on that subset of data. You analyse your data by comparing the reports for different selection criteria. For instance you can compare the numbers of males and female infants by listing the data after selecting males and again after selecting females. In EDMP there is now the option to write the reports out directly into an excel spreadsheet which you can save in the usual manner.

The selection screen is shown below and now contains five new pages of criteria relating to the derived variables, anomaly groups, user defined groups, multiple malformation and

aetiology categories and locations. EDMP automatically assigns each record into the correct anomaly sub-group or sub-groups and multiple malformation group at either data entry or edit and at import.




To make a selection enter the criteria as necessary. Your current selection will be displayed in the box on the right of the screen. When you are satisfied that the selection is correct click on the 'Reports' button to take you to the report sub menu.

The report menu offers you a number of standard reports you can run on the selected data. For clarity Reports are divided into three categories, namely "List Cases', 'Frequency Tabulations' and 'Export'. You can skip between the categories using the navigation buttons provided at the bottom of each screen. In addition, you can alter your selection by clicking on the 'New Selection Criteria' button and you will be returned to the selection screen. The selection criteria are printed on each report. You can also export selected core, core & non-core and all variables from the reports screen to a .csv file or to Excel. A new option allows you to save and recall named selections of variables to output.

## 7.2 Prevalence Tables

There are three categories of subgroups that you can produce prevalence tables for. They are 'EUROCAT Anomaly Subgroups', 'User Defined Anomaly Subgroups' and 'User Defined

Subgroups'. Once you have selected subgroup type you will be able to select between four different report formats (A1, A5, A6 and B3) which are the same as those available on the EUROCAT web site. These reports are all based on centre, birth year and anomaly or user defined subgroup. Simply select required report type and relevant selection criteria and the results will be output to an Excel spreadsheet.

### 7.3 Missing Values by Year

This option tabulates the number and percentage of non-missing, missing, unknown and invalid entries by year for selected variables. It is a useful tool in analysing the quality and completeness of your data.

## 8 User Defined Categories

EDMP provides you with the ability to define three types of definable categories that you can use in the analysis and reporting of your data. The categories are a valuable tool as they offer a great deal of flexibility in their definition and can save time as they are saved. Categories are automatically recalculated when cases are added, imported or edited.

### 8.1 Create User Defined Anomaly Subgroups

You can define up to ten subgroups with selection criteria, based on ICD codes, which you specify. This allows you to select cases from the database with different subgroup criteria than those already defined in the EUROCAT anomaly subgroups.

Setting up subgroups is simple and each group can contain up to fifty ICD selection criteria. Each ICD selection criteria can be in the form of 'like' (e.g. Q90*), 'exact' (e.g. Q901) or 'range' (e.g. Q90 to Q91). Further instructions are shown on the set up pages. When you save an user defined subgroup EDMP will check each case against the criteria and mark it as '1' if the criteria are met otherwise '0'. EDMP checks the syndrome and malfo1 to malfo8 fields against the criteria.

### 8.2 User Defined Subgroups

User defined subgroups are similar to user defined anomaly subgroups above except that you are not limited to ICD malformation codes and you can select any of the EDMP variables to enter criteria for. This is a powerful method of analysing your data as you can specify up to ten user defined subgroups based on a wide range of variables and values.

### 8.3 User Defined Location Categories

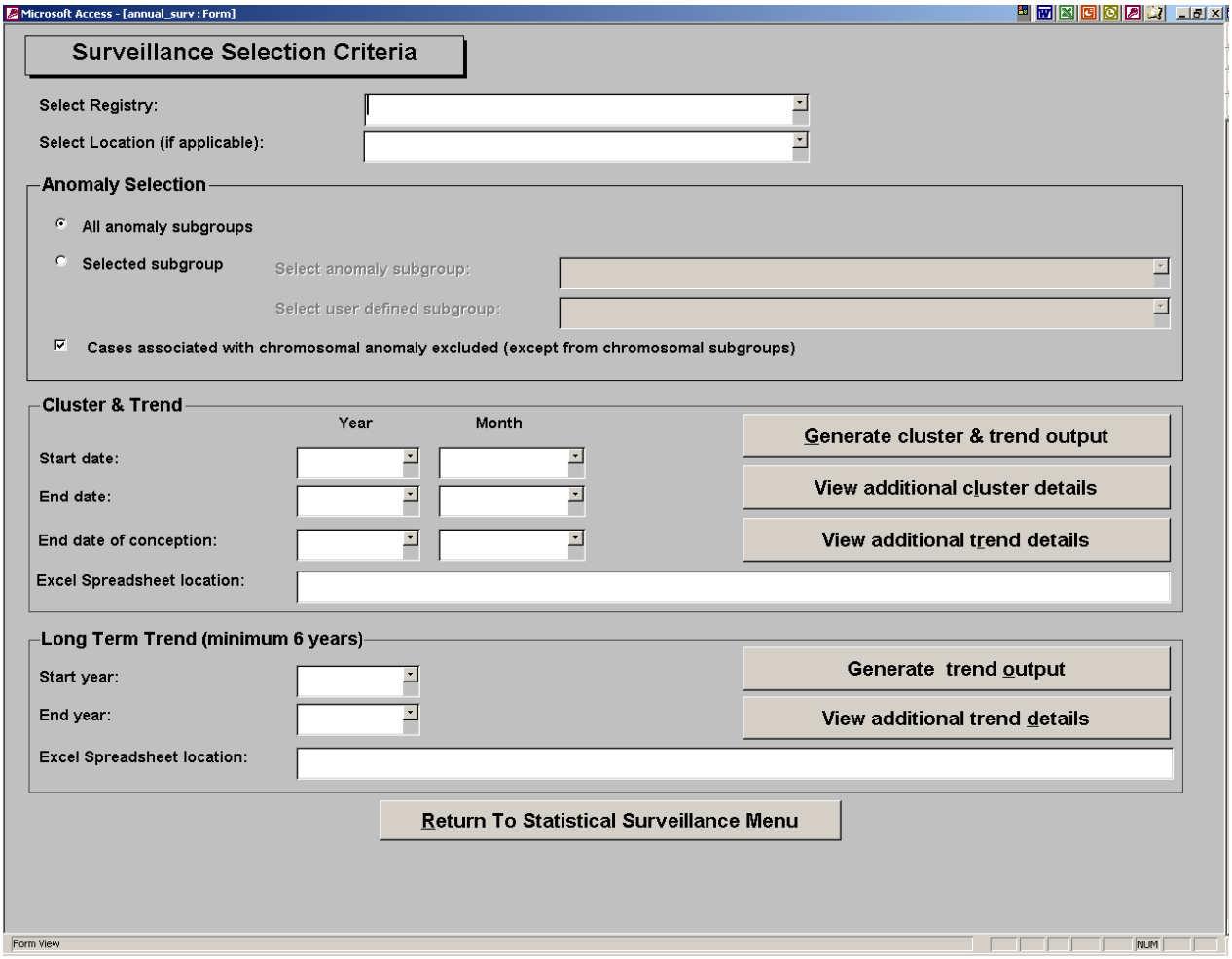
User defined location category allows you to specify up to ten location categories which can be used as selection criteria for reports and for statistical surveillance. The variables available to create a location subgroup are Place of birth, Mothers residence code and any of the extra variables that you have defined (excluding the 'Date' types). Extra variables are defined using the Extra Variables & Layout option under the System Menu.

# 9 Surveillance

The Surveillance option allows you to scan your data for trends and clusters by EUROCAT and user defined anomaly subgroups. In addition you can now select user defined location categories. Help on trends and clusters is provided on the statistical monitoring screen. For both trends and clusters you can choose to output the results for all anomaly sub groups to Excel or examine individual subgroups in further detail on screen.

There have been significant changes made to the way you perform routine checks for trends and clusters. To perform routine surveillance you need to select your centre (and optionally a location category). In the anomaly selection box you can choose to select all anomaly subgroups or a selected anomaly subgroup or user defined anomaly subgroup. If you select all anomaly subgroups then output will be to Excel with the option to print details for significant trends or clusters. For individual selected anomaly subgroups the results are displayed on screen with the option to print the results. Cases associated with chromosomal anomaly are always excluded (except from chromosomal subgroups) when the all anomaly subgroups option is selected but is optional for individually selected subgroups.

For the clusters & trend option you need to select the start and end years (by date of birth) and alter the months as required. EDMP will automatically calculate the new end date for use with date of conception as nine months less than the end date by date of birth. This is done to ensure that there is no bias due to excluding births with longer gestations that would be delivered after the study period.




When you click the 'Generate cluster & trend output' button you will be prompted for the name and location for the results spreadsheet if you have selected the all anomaly subgroups option. EDMP will then create this spreadsheet containing sheets for trend and cluster results. Cluster checking is not performed on major anomaly groups which are considered to be uninformative (i.e. All Anomalies, Nervous System, Eye etc.). Cases associated with chromosomal anomaly are excluded from trends and clusters except for subgroups which are chromosomal.

Trends are always based on date of birth and now include a test for heterogeneity of prevalence over time (change over time without increasing or decreasing). The trend option uses a Chi-square test to test for significant increases or decreases (or heterogeneity of slope) in the number of cases per year per 10,000 births by anomaly sub group. You must enter denominator data into EDMP in order to be able to use the trend analysis.

Clusters are based on date of conception where possible, if the number of estimated gestations exceeds 10% then the cluster is checked by date of birth. Where gestational age is missing EDMP will use estimated GA based on the average GA by birth year and type of birth. Date of conception is calculated as date of birth minus days gestation (GA weeks * 7).

Clusters are only shown which do not exceed 18 months duration and the last case in the cluster must be within two years of the end date.

You can print out the results of any significant trends or clusters by clicking on the relevant print button. The trend output includes a graphical representation including the trend line where the slope is not heterogeneous. The cluster output lists cases, cluster details for each significant cluster and a graphical 'time line' distribution of cases and the limits of each cluster.

The long term trend (minimum 6 years) option allows you to test for trends either for all anomaly subgroups (including the 'heterogeneous' subgroups excluded from cluster detection) or for selected subgroups. Results for the all anomaly subgroups option are output to Excel with the option to print significant results. Analysis for selected individual subgroups is displayed on screen with the option to print the results.

If the observed average number of cases per year is below the minimum of 5 then EDMP will group the data into two year intervals. If the grouped data has an observed average number of cases per two year interval of 5 or greater then a test for trend will be performed.

## 10 Import / Export Data

### 10.1 Import Data

You can use the import facility to enter a batch of cases from file, rather than entering them via the screen. Typically you would import cases if you were converting to EDMP from a different data entry program or if you are using EDMP to validate your own data prior to transmission to the Central Registry.

For a file to be imported successfully it must fulfil the following criteria:

- The file must be in comma separated format (.csv)
- The field names must be in the first row of the data
- One of the field names must be 'centre' which is your EUROCAT centre number.
- The centre number must be present in every row of the data.
- Date fields must be in the format specified in the Data Transmission Form i.e. 6 characters wide and must include any leading zeros. For example the 7th May 2001 would be 070501. However, EDMP will try and read date fields that have lost their leading '0' and are only 5 characters long and the dates will be accepted if they convert to a valid date.

- Coded variables must conform to values specified in 'EUROCAT Data Transmission Form' with the exception that '0' entries for coded fields will be converted to blanks where '0' is not a valid entry.

- Blank lines must be removed from the data including trailing carriage returns and line feeds.

- If you are creating your import file using Excel the date fields will need to be formatted to ensure that the leading zeros are not removed from the .csv file. Use 'Text' or 'Custom' formats for these date fields, if you choose custom then specify 000000 as the format (six zeros).

- Extra drugs can be imported in the field named 'extra_drugs' by first creating an extra column in your data with the heading 'extra_drugs'. In this column, you will need to enter the ATC code and text description in the following format:

  <ATC code|text description>

  The ATC code and the text description are enclosed by the '<' and '>' characters.

  Also, the ATC code and the text description are separated by the pipe symbol '|'. To get the pipe symbol separating the ATC code and text description, hold down the alt key while typing 124 on the numeric keypad:

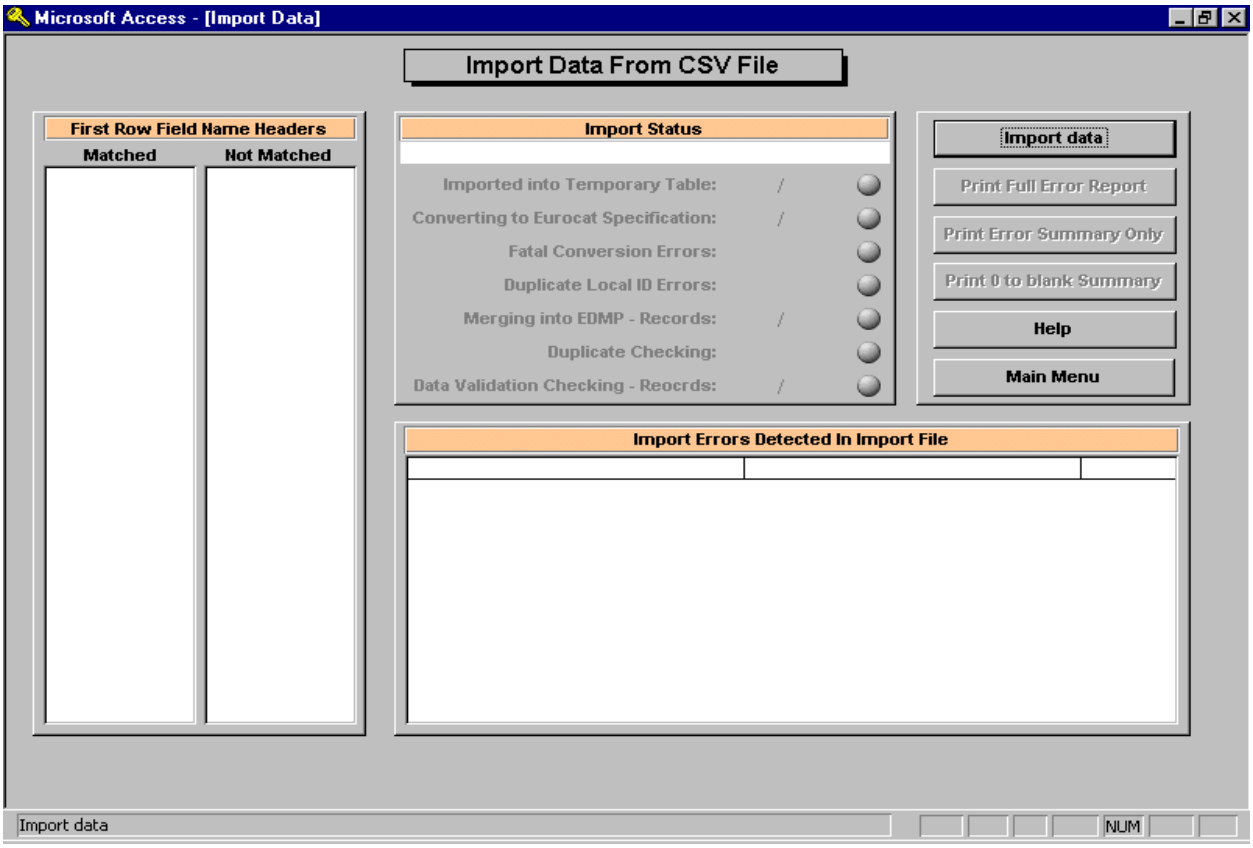
  Alt 124 = |

  If more than one extra drug is to be imported for a single case, then enter the ATC codes (in the same format as above) side by side in your extra drugs field:

  <ATC code|text description><ATC code|text description>

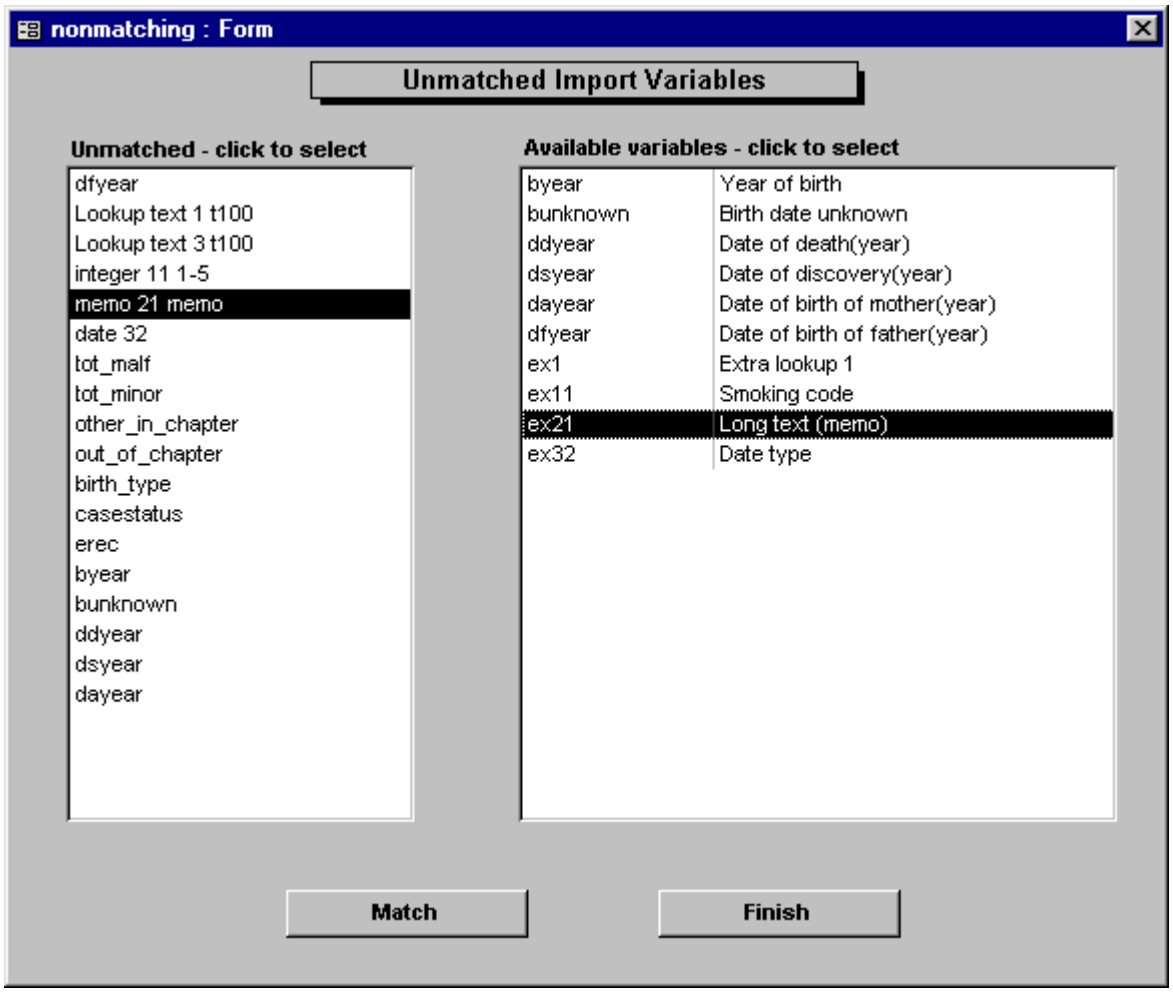
  So for example a case with valproate and lamotrigine exposure is entered in the extra_drugs field as: < N03AG01| Sodium Valproate>< N03AX09| Lamotrigine>

To import a .csv file click on the 'Import data' button as shown below:




You will then be prompted for the location of the file using the standard Windows file location screen. Once found the program will try to import the file.

EDMP has a new feature where the import variables that are unmatched (i.e. EDMP does not recognise their name) can be matched with variables not already being imported into. This feature is provided primarily to allow the importation of local data into the user definable extra variable fields but does provide a mechanism of importing files without having to edit all the field names. Please note that here must still be a field named 'Centre' present which contains your centre number.
In order to match variables simply select one each from the 'Unmatched' list (these are from the import file) and from the 'Available variables' list (available slots in EDMP) and then click on the 'Match' button (see below). You will be asked to verify the match and once matched the names will be removed from their respective lists. You can keep matching until done when you click on the 'Finish' button to continue with the import process.




The status of the import as it goes through various stages is also shown. The Stages of import are as follows:

Import Record

This is the import of the raw data from the import csv file into a temporary table within the EDMP. The variables whose name matches those specified in the Data Transmission Form or have been cross matched will be listed in 'Matched' box and those which do not match will be listed in the 'Not Matched' box. If any errors are encountered during import they will be listed in the 'Import Errors Detected In Import File' box. Access has been unable to import these rows and indicates serious data problems that need to be corrected in the raw data prior to import.

Converted Records

The data in the imported records are then converted into EDMP format.

Fatal Conversion Errors

If any fatal conversion errors are found the import process will be stopped and corrections need to be applied to the original data or the csv file.

Duplicate Local ID Errors

If any records within the import file contain the same local ID numbers that are already in the EDMP then again the process will be stopped.

Merged Records

Once checks are completed and passed the records will be merged into EDMP.

Duplicate Checking

Duplicate checking will then be performed on the newly imported records and each record will be marked with its matching status (Checked / Possible matches).

Data Validation Checking

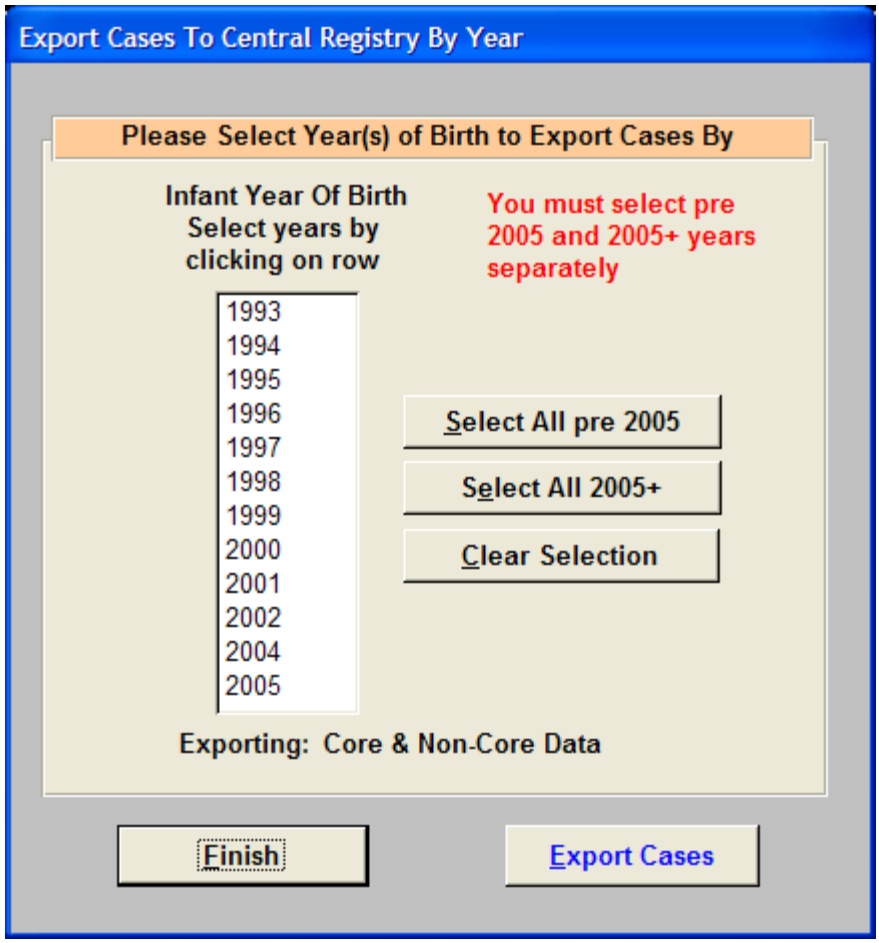
The records will then be validated and given a data validation status. Which are as follows:

1) Core data incomplete or invalid, non-core data valid or invalid
2) Core data complete & valid, non-core data invalid
3) Core data complete & valid, non-core data valid

Once the import has stopped you can print an error report for any errors found (Summary, Full or '0' to blank conversions). EDMP now displays the Local ID number for cases containing errors.

## 10.2 Export Data To EUROCAT

When sending data to the Central Registry you can now send the data for more than one year in a single file. To export data simply select the required year or years from the screen shown below and click on the 'Export Cases' button. You will need to specify the name and location of the export csv file in the usual manner. You can export either Core or Core & Non-core data depending upon the data input/output setting on the main menu. You can export different selections of data under the reports section. Please note that you must export pre 2005 and 2005 onwards cases separately as the new guide 1.3 variables are only available for 2005 and later births.

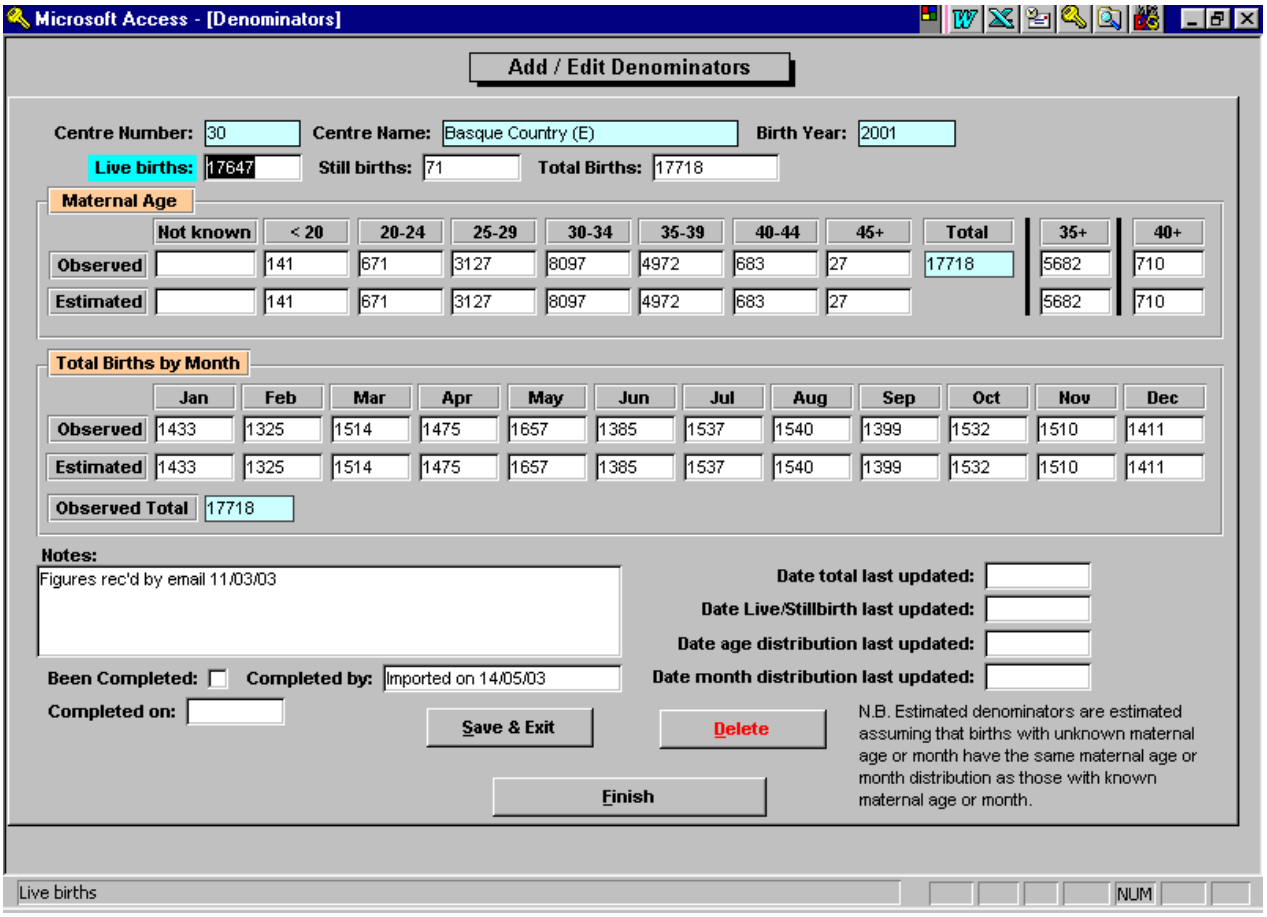



## 11 Denominators

This section allows you to add your denominator data to EDMP and then export it in a format suitable for transmission to the Central Registry. You also need denominator data for some of the reports and for the trend analysis under statistical surveillance.

### 11.1 Add / Edit Denominators

The Add/Edit option allows you to list and then select a year to edit or to add a new year. EDMP automatically detects what year and centre combinations are present in your data and creates a record ready for you to complete. The data entry/edit screen is shown below:




The data you enter is the same as that specified in the Template for Denominator Data in chapter 2.3 of the EUROCAT Guide 1.3. For both the maternal age and total births by month sections there are two rows of data namely 'observed' and 'estimated'. You enter your values in the observed row and EDMP will calculate the estimated values by multiplying each observed value by ratio of the total births entered (sum of live and still births) divided by the total of the values entered in the respective observed row.

## 11.2 Import Denominators

You can import denominator data much the same way as you import case data into EDMP. If you are importing data provided by the Central Registry then the file provided will be in the correct format. If you are importing data from a different source you will need to contact the Central Registry to obtain the required file format.

## 11.3 Export Denominators

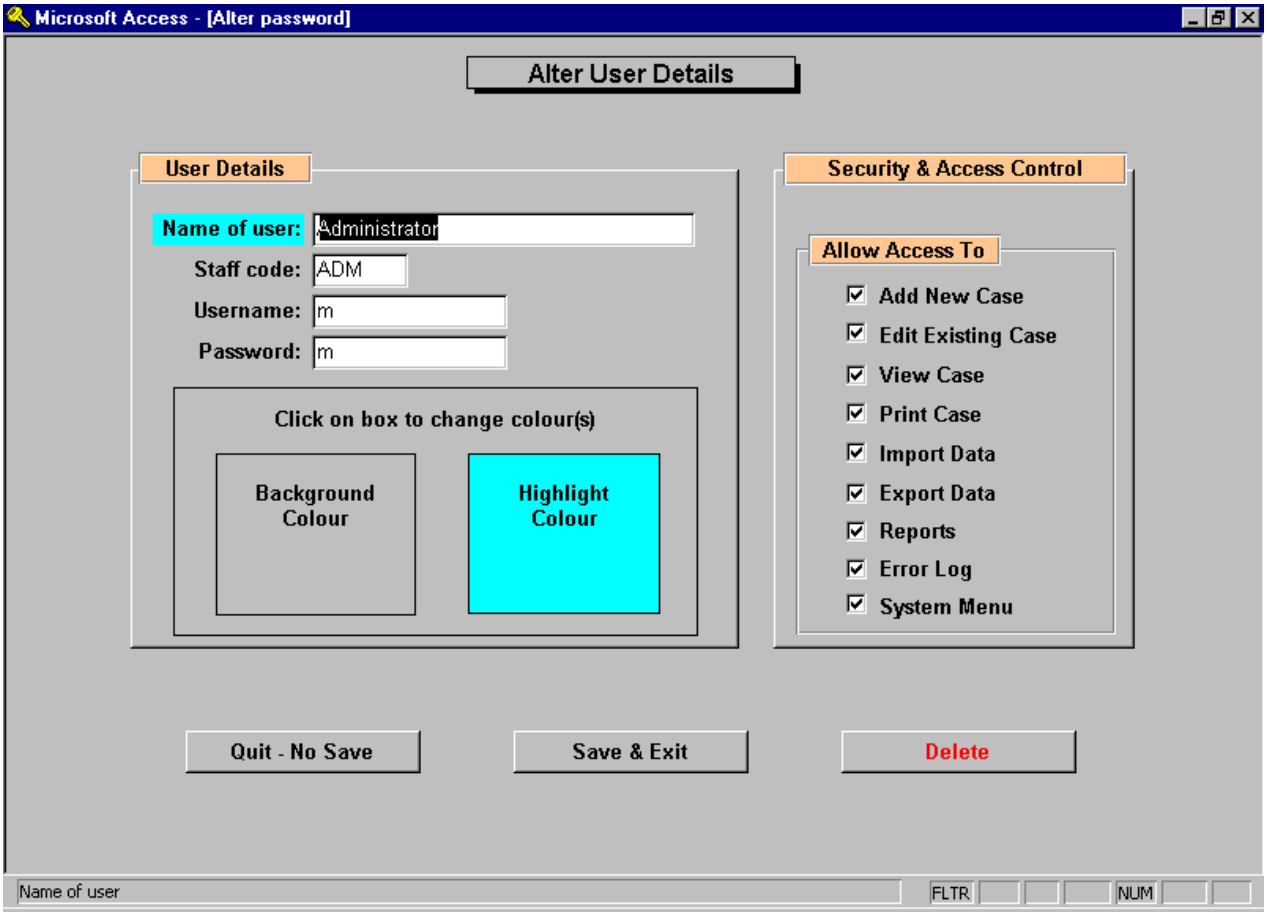
This option allows you to select denominator years and then create an export file in the format required by the Central Registry.

## 12 System menu

The system menu provides you with facilities to alter login details, change the default printer, determine the current location of the data, set your centre name and number (**note** that the centre name will appear on all reports) and delete selected cases. There are also two options relating to the anomaly subgroups and for the extra local variables and screen layout.

### 12.1 Manage Users

Once you have entered the Manage Users section you can either set up new user details (click the Add New button) or alter the details of existing users by clicking on the required row in the list box. Please remember to alter the user name and password for the 'Administrator' user, which is distributed with the program. The screen shown below will be displayed.




User details allows you to enter (or alter) user details including username and password as well as allow you to alter the personalised screen colour settings for each user. To change screen and highlight colours simply click on the required box on the screen and the standard Windows colour selection screen will appear. 'Security & Access Control' allows you to

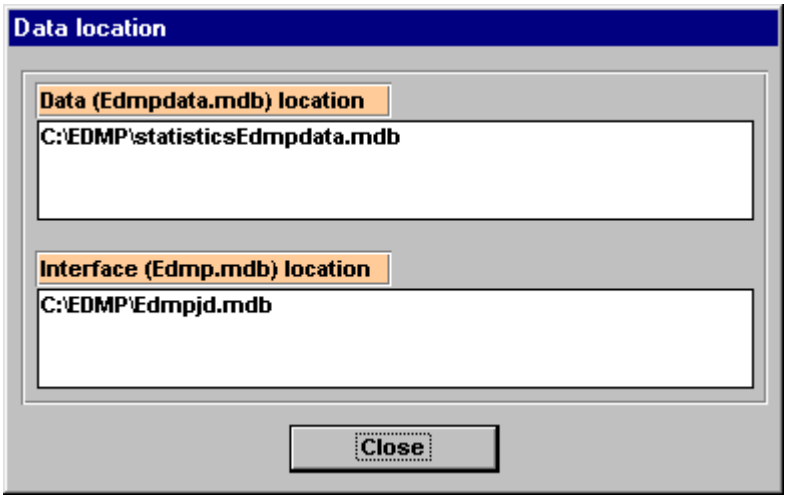
specify which parts of the program are accessible to each user. It is important that at least one user has access to the System Menu!

### 12.2 Set default printer

The default printer facility displays the currently selected default printer and also allows you to select a different default printer by clicking on the 'Change Printer' button. All the printers available to your PC will be displayed in the list for you to choose from.

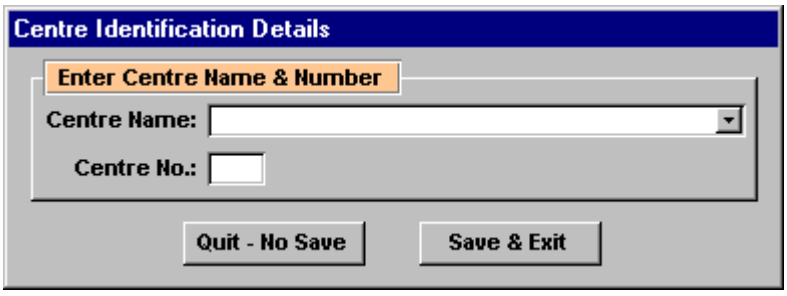
### 12.3 Data location

This facility displays the location of the data file Edmpdata.mdb that the program is currently using as well as the location of the 'front end' of EDMP (Edmp.mde).




### 12.4 Centre Name & Number

This is where you set your centre name and number. Note that the centre name will appear on all reports. You can select your centre name from the pull down list which will then automatically fill in the centre number, or enter the details yourself.




### 12.5 Recalculate Subgroups

Use this option to recalculate the user defined subgroups and categories and multiple malformation code for all cases in your database. This option is provided in case you

encounter any errors during data entry or import and you wish to recalculate to ensure subgroup integrity.

### 12.6 Extra Variables & Layout

This menu option allows you to define up to 32 extra variables for use in data entry, create custom data entry form layouts and also change the labels for the five EDMP spare variables.

#### 12.6.1 Extra Local Variables

You can select a maximum of 32 extra variables for use at data entry. For each variable you can specify its name, position and data type. Data types are 'lookup text' which are option lists to which you can add your own values at data entry, integer numeric with optional minimum and maximum values, unlimited text and finally date type.

#### 12.6.2 Data Entry Screen Layout

You can specify your own data entry layout using this option. Simply select which variable you want in each field position of the data entry form. EDMP will check that you have selected the minimum number of required (core) variables before allowing you to save the set-up.

Please note that you must define a screen layout for both pre 2005 and 2005 onwards cases as there are new variables (guide 1.3) for use in 2005 onwards.

You can only use the screen layout at resolutions of 800*600 and 1024*768 and you will also need to create separate layouts for each of the resolutions. Choose your preferred resolution prior to defining the layout.

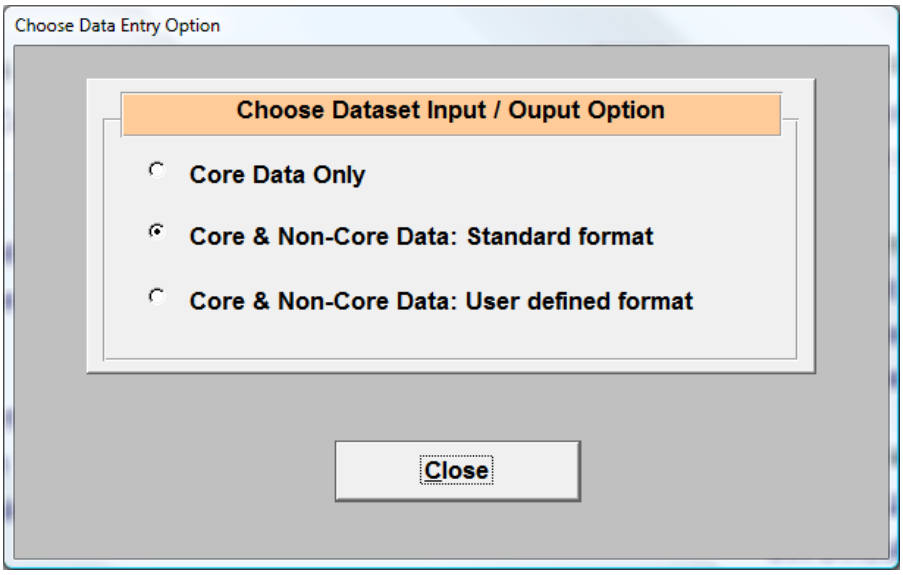
#### 12.6.3 Change Spare Variable Names

Use this option to change the label captions for the five EDMP supplied spare variables.

#### 12.6.4 Unhide 2005+ Local Variables

This option allows you to show or hide the sources of information(1-5) fields along with the social & racial status of mother and father.

## 12.7 EDMP Display Options




There are three options for EDMP Display Options: 'Core Data Only' and 'Core & Non-Core Data' either as Standard format or User defined format. When the 'Core Data Only' option is selected the data for adding, editing, viewing, printing and exporting is restricted to the core variables only. For the 'Core & Non-Core Data' options the standard format is the tabbed page by page data entry screen layout. The user defined format allows you to select which variables you wish to work with and also their order on the data entry screen. With this format the data entry page is continuous but with page divisions and offers a variety of page navigation methods.

# 13 Routine Maintenance

The file Edmp.mde may grow in size due to frequent use of the Import facility. To counter this you can repair and compact Edmp.mde. This is done by opening Microsoft Access without opening or creating a new database and then selecting 'Tools' from the menu bar. Select database utilities and then select the 'Repair Database' option. You will then be prompted for the location of Edmp.mde. Repeat the process selecting the 'Comp

# Chapter 2.5

# Data Validation Routines
# (Issued September 2005)

## 2.5 Data Validation Routines

Validation of data should be done using the EDMP before data is transmitted to Central Registry.

### 1 Essentials

- The local identity number within a registry cannot be duplicated
- Data with values outside the accepted range (as described in Chapter 2.2.1b) will not be imported to EDMP.

### 2 Duplication checks

- Cases with the same values for 4 key variables (date of birth, sex, birthweight +/- 100 g, maternal age) should be checked as possible duplicates. If value= unknown for any of the 4 matching variables, then case is not matched on that specific variable.
- If case is a twin or higher order multiple birth (NBRBABY=2, 3, 4, 5 or 6), then no duplicate check is carried out.
- It is not sufficient to rely on matching the name of the baby or mother for finding duplicates

### 3 "Core" information

All babies must have local identification number, date of birth, sex, number of babies delivered, number of malformed cases in multiple set, type of birth or abortion, civil registration status, birthweight, length of gestation, survival status, age of mother, when malformation was discovered, and at least one malformation or syndrome code. Before sending data to the Central Registry any cases with this "core" information lacking should be reviewed to find out if it is possible to complete the missing data.

### 4 Range Error Checks

Unusual values should be verified eg.

a) mother's age outside the range 15 to 50
b) total previous pregnancies greater than 12

### 5 Logical validation

The following checks of the logical relation between variables are suggested. Sometimes these checks only indicate unusual but possible relationships between

different items of information (for example a livebirth at 19 weeks gestation). The more unusual the information, the more likely that there is a coding error. Therefore, these cases should be checked to make sure that the information is correctly coded.

- Baby and Mother
    1. If SEX=3, then the code for indeterminate sex (Q56) MUST be entered in malformation field.
    2. If SEX=9 (unknown), then PM must be 3 (not performed) or 4 (macerated fetus) or 9 (Not known).
    3. If NBRBABY=2, 3, 4, 5 or 6, then NBRMALF must be filled in with a number other than 9 (Not known).
    4. If both twins of a twin pair are malformed (NBRMALF=2), then PREVSIB=1 (for both twins).
    5. If both twins of a twin pair are malformed (NBRMALF=2), then the local ID number of the co-twin should be entered in SIB1.
    6. If NBRBABY=2, 3, 4, 5 or 6, then specify twin or multiple type of birth in SP_TWIN field.

- Type of birth
    1. Type of birth, length of gestation and birthweight should be compatible according to the definitions used by the local registry (see instructions)
    2. If type of birth =2, 3 or 4 (SB/ SA/ or IA), then SURVIVAL must be 2 (No)
    3. If type of birth =4 (IA), then WHENDISC must be 6 (prenatal)

- Gestational age, and Birthweight
    1. Maximum birthweights for gestational age are usually:

        | | |
        |---|---|
        | 20-22 weeks | 750 g |
        | 23-25 weeks | 1000 g |
        | 26-27 weeks | 1500 g |
        | 28-31 weeks | 2000 g |
        | 32-35 weeks | 3500 g |
        | 36-37 weeks | 4000 g |
        | 38+ weeks | 6000 g |

        Birthweights outside these values should be checke
    2. Birthweights less than 500g should be verified if coded as a live or stillbirth

- Death
    1. If survival beyond a week of age =1 (yes), then TYPE =1 (livebirth).
    2. If survival beyond a week of age =2 (no), and TYPE =1 (livebirth), then date of death should be known and should be within one week of birth

- Parental age
  If date of birth of mother (father) is known, the age of mother (father) must also be completed. The age of mother (father) must be the number of completed years between the date of birth of the mother (father) and the date of birth of the baby.

- Previous reproductive history
  1. Total pregnancy validation – total pregnancies may not be equal to total births (twins=2 births, but 1 pregnancy)
  2. Implausible combinations of maternal age and number of previous pregnancies are age 15 or less with 2 or more previous pregnancies, or age 16-19 with 3 or more previous pregnancies.

- Diagnosis
  1. If “when discovered” =6 (prenatal) then AGEDISC should be completed.
  2. If “when discovered” =6 (prenatal) and “condition at discovery” =2 (dead), then type of birth should be a spontaneous abortion (code 3) or a stillbirth (code 2).
  3. If “when discovered”=7 (at abortion 7), then the type of birth should usually be a spontaneous abortion (code 3).
  4. If PM= 1, 2 or 4 (performed) and TYPE =1 (livebirth), then DATE OF DEATH must be entered
  5. If “when discovered” =6, then FIRST POSITIVE PRENATAL TEST must be coded as 1-7 or 9.

- Malformation codes
  1. If McKUSICK code is entered, there should be a valid ICD code entered in the Syndrome field
  2. All syndrome codes MUST be specified in the SP_SYNDROME field.
  3. If unspecified malformation code is entered, further information MUST be given in the SP_MALF fields.

- Specific malformation coding rules
  1. There is only one valid 2-digit ICD10 code, and that is microcephaly (Q02). All other malformations coded in ICD10 must have at least one letter (eg. Q) followed by 3 digits.
  2. If code is patent ductus Q250, gestational age must be at least 37 weeks
  3. If code is Q53, Q54, Q55 (male genital organs) sex must be 1
  4. If code is Q50, Q51, Q52 (female genital organs) sex must be 2
  5. If code Q96 and Q97 sex must be 2
  6. If code Q98 sex must be 1
  7. If code Q00 (anencephalus) "survival first week" must be 2
  8. If code Q05 (spina bifida) no separate code for hydrocephalus (Q03)

9. If code Q601 (bilateral renal agenesis) or Q606 (Potter syndrome) "survival first week" must be 2
10. Code Q897 or Q899 cannot be used as the only malformation code

- Family history
  1. If CONSANG=8 (other relation), text information MUST be specified in the SP_CONSANG field.
  2. If SIBANOM=1, 2 or 3 and PREVSIB=1, specify the local identification number in SIB1, SIB2 or SIB3.

## 6 Frequency checks

Before sending a batch of data to EUROCAT central registry, produce some frequency tables to ensure that the quality of the information corresponds to the aims of the local registry. NB: This check is not automatically performed by the EDMP.

- A high frequency of unknown values for any variable should prompt an investigation of how the recording of the variable can be improved, and the registry should communicate with Central Registry concerning how the variable can be used in analyses of data, or if there is selection bias in the distribution of known values.
- It may be useful to check that all malformation codes which have been used only once are valid codes.
- A high frequency of poorly specified malformation codes should prompt investigation.
- The number of cases where “total previous pregnancies” has been coded “0” should correspond approximately to the number of cases expected from the proportion of primiparous mothers in the population.
- Cross-tabulation of maternal age and number of previous pregnancies should show a distribution roughly corresponding to the distribution in the total birth population.

Data Validation Checking

The records will then be validated and given a data validation status. Which are as follows:

1) Core data incomplete or invalid, non-core data valid or invalid
2) Core data complete & valid, non-core data invalid
3) Core data complete & valid, non-core data valid

# Chapter 3

# Coding and Classification

3.1 Overview of EUROCAT Approach to Coding and Classification

3.2 Minor Anomalies for Exclusion

3.3 Coding of EUROCAT Subgroups of Congenital Anomalies

3.4 ~~Outside malformation chapter (Q) codes~~ – deleted, codes for all anomalies is given in Chapter 3.3 (July 2011)

3.5 Multiple Congenital Anomaly Algorithm

3.6 Detailed Congenital Anomaly Coding Guidelines

3.7 EUROCAT Description of the 2011 Congenital Anomaly Subgroups

# Chapter 3.1

# Overview of EUROCAT Approach to Coding and Classification
# (Revised July 2011)

## 3.1 Overview of EUROCAT Approach to Coding and Classification

Coding and Classification of congenital anomalies: a summary of the EUROCAT system

1. The purpose of coding congenital anomalies in EUROCAT registries is to summarise unstandardised written text in such a way that the data can be analysed for surveillance and research purposes. Registries can encourage but can rarely impose use of standard definitions and diagnostic tests and clinical follow-ups, and the coding system must allow for different levels of precision and accuracy of information provided by clinicians.

2. The purpose of a "classification" system in the sense that we use this term here is to group together anomalies which share aetiologic or clinical characteristics. There is a balance to be struck a) between "lumping" together heterogeneous sets of anomalies and "splitting" so finely that there are few cases in each group b) between creating groups based on great precision and accuracy of diagnosis and coding and creating groups which take into account what can be realistically found in medical records for most cases.

3. The standard dataset for each case (Chapter 2.2) allows for the text description and coding of up to eight malformations and one syndrome. Where there are more than eight malformations, additional malformations can be added in the text variable for the 8$^{th}$ malformation. When there is more than one syndrome, the second syndrome can be coded in the first malformation variable.

4. Malformations are coded to ICD10 with the British Paediatric Association (BPA) one digit extension. Syndromes are also coded to ICD10-BPA, but an OMIM (McKusick) code can be given in addition, but should be coded with caution and expertise. A specific EUROCAT guide to the coding of syndromes gives more detail (EUROCAT Guide 6, see Reference documents in Annex). Where ICD9-BPA codes are given in this Guide, this is solely for the purpose of analysing the EUROCAT database from 1980 to 2004. ICD9 codes should no longer be used.

5. "All anomalies": The core set of congenital anomalies to be registered by all member registries are structural malformations and chromosomal anomalies diagnosed in the fetus, baby or child. Particular attention is paid to complete ascertainment of those anomalies usually diagnosed in fetal life or the first year of life. In order to set clear boundaries to this group and achieve comparability of prevalence rates between registries, the count of cases with all anomalies includes all cases with one or more codes in the Q chapter of ICD10 and a very limited set of conditions coded outside the Q chapter, as specified in the definition of subgroups given in Chapter 3.3 (see subgroup "all anomalies").

6. In addition, registries may register other congenital conditions, including congenital neoplasms (ICD10 C and D codes), and congenital endocrine (E), metabolic (E), immunologic (D), and haematologic (D) conditions. These are analysed separately and not included in the prevalence of "all anomalies". Registries should make clear in their registry description which non-Q conditions they register to facilitate separate analysis of these data.

7. Impairments of function with a partially or wholly prenatal origin such as cerebral palsy or autism or mental retardation should not be transmitted to EUROCAT unless in association with specified structural malformations.

8. Cases with only minor anomalies as specified on the EUROCAT list of minor and unspecified anomalies for exclusion (Chapter 3.2) should not be transmitted to EUROCAT. Minor anomalies should be described in text, coded and transmitted to EUROCAT when they are in association with major anomalies. Where a case with one or more minor anomalies only is transmitted to EUROCAT in error, it will be excluded by computer if the minor anomalies have specific codes which allow recognition. Some minor anomalies as given in Chapter 3.2 do not however have specific codes and cases with such isolated anomalies must always be recognised and excluded at local level on the basis of the text description.

9. The EUROCAT subgroups as defined by their ICD10 codes (Chapter 3.3) are subgroups for which prevalence information is routinely produced. A selection of these subgroups are also the subject of routine statistical monitoring for trends and clusters in time. Subgroups have been defined according to one or more of the following criteria a) larger heterogeneous subgroups which show the relative health burden of anomalies in different organ systems b) subgroups which balance aetiologic homogeneity with the level of diagnostic specificity which can reasonably be expected by European registers c) subgroups which are relevant to health service provision, including prenatal diagnosis d) subgroups which are well defined and clinically diagnosed with a good level of consistency across Europe, and where specific codes are available e) subgroups that are consistent with the hierarchical classification of ICD10 f) subgroups of reasonable frequency such that a yearly European prevalence can be meaningful. Only major anomalies (ie not on the list for exclusion in Chapter 3.2) are allocated to subgroups, however where the same code specifies both a minor and major anomaly, minor anomalies may be included in subgroups.

10. Where appropriate for aetiologic analyses and statistical monitoring, prevalence will be calculated for "chromosomal" and "non-chromosomal" cases separately in order to achieve more aetiologic homogeneity. Non-chromosomal cases will be the number of cases in each subgroup excluding those which also have a chromosomal anomaly (eg. a case of Trisomy 18 with spina bifida will be allocated to the Trisomy 18 subgroup but not to the spina bifida subgroup).

11. All prevalence rates and counts for subgroups are based on cases, not malformations. Thus a baby with a VSD and valve stenosis will be counted ONCE in “all anomalies”, ONCE in “cardiac”, ONCE in “VSD”, ONCE in “valve stenosis”. A baby with encephalocele and renal dysplasia will be counted once in the count of “all anomalies”, once in the count of cases with central nervous system anomalies, once in the count of cases with neural tube defects, once in the count of cases with encephalocele, once in the count of cases with renal dysplasia and once in the count of cases with urinary anomalies. It follows that the number of cases in different subgroups CANNOT be added together to find the total number of cases, as one case can be counted in more than one subgroup. Higher prevalence of subgroups can be expected in areas where more detailed coding of multiply malformed babies is undertaken.

12. In addition to the EUROCAT subgroups, prevalence information will also be produced every five years, pooled across registries, for the rarer syndromes. These are not in the subgroup list but are in the database, and should be well coded and specified in written text. Priority for analysis of prevalence will be given to well defined syndromes diagnosed prenatally or in the first year of life with severe health consequences or of particular interest in relation to environmental risk factors or treatment possibilities.

13. Multiply malformed cases are the subject of a separate statistical monitoring exercise with the purpose of identifying teratogenic exposures that cause patterns of multiple malformations. Cases which are likely to be multiply malformed are identified using the hierarchical computer algorithm given in Chapter 3.5. Manual review of the identified potential multiply malformed cases will be done before statistical surveillance.

14. Further development of the algorithm in Chapter 3.5 will in future allow surveillance of prevalence rates of isolated cases (cases with one malformation only), and non-syndromic cases within each EUROCAT subgroup.

# Chapter 3.2

# Minor Anomalies for Exclusion
# (Revised October 2010)

## 3.2 Minor Anomalies for Exclusion

*For EUROCAT for use from 2005*

Reports of cases with the following anomalies are not to be transmitted to the EUROCAT Central Registry if the anomalies are isolated. It is, however, important to report all minor anomalies for cases with major malformations or syndromes.

"Minor" anomalies are excluded, when isolated, because they have lesser medical, functional or cosmetic consequences (although they may be indicators of other problems) and experience shows that their definition and diagnosis and reporting varies considerably. At the present time, it is not useful to collect data at a European level on these anomalies. We also exclude anomalies which are not always truly congenital in origin, sometimes associated with immaturity at birth. In addition, we exclude poorly specified conditions and recommend that for any such cases more specific information be sought from medical records.

Cases reported to EUROCAT should always be confirmed cases of congenital anomaly. Cases which had diagnosed ultrasound soft markers but who were found to be normal at birth or with unknown outcome should not be reported.

Note that exclusions should be made locally, where all information is available. Many minor anomalies do not have specific ICD10-BPA codes, but we give specific codes where they exist. For the codes given in the list, if any cases with only one or more of these codes has been inadvertently transmitted to Central Registry, they will be subsequently excluded from the central files on the basis of the code only. For allocation of cases to EUROCAT subgroups (see Chapter 8), only major malformations will be considered (codes for minor anomalies will be excluded).

| | **Specified ICD10-BPA – if present** |
|---|---|
| **Head** | |
| Aberrant scalp hair patterning | |
| Flat occiput | |
| Dolichocephaly | Q67.2 |
| Plagiocephaly – head asymmetry | Q67.3 |
| Bony occipital spur | |
| Third fontanel | |
| Macrocephalus | Q75.3 |
| Facial asymmetry | Q67.0 |
| Compression facies | Q67.1 |
| Other cong deformities of skull, face and jaw | Q67.4 |
| | |

| | |
|---|---|
| **Eyes** | |
| Epicanthic folds | |
| Epicanthus inversus | |
| Upward slanting palpebral fissures | |
| Downward slanting palpebral fissures | |
| Short palpebral fissures | |
| Congenital ectropion | Q10.1 |
| Congenital entropion | Q10.2 |
| Other congenital malformations of eyelid | Q10.3 |
| Dystopia canthorum | |
| Hypertelorism | Q75.2 |
| Hypotelorism | |
| Stenosis or stricture of lacrimal duct | Q10.5 |
| Synophrys | Q18.80 |
| Blue sclera | Q13.5 |
| Crocodile tears | Q07.82 |
| | |
| **Ears** | |
| Primitive shape | Q17.3 |
| Lack of helical fold | Q17.3 |
| Asymmetric size | Q17.3 |
| Posterior angulation | Q17.3 |
| Microtia | Q17.2 |
| Macrotia | Q17.1 |
| Protuberant ears | Q17.3 |
| Absent tragus | |
| Double lobule | Q17.0 |
| Accesorry auricle, preauricular appendage, tag or lobule | Q17.0 |
| Auricular pit | |
| Preauricular sinus or cyst | Q18.1 |
| Narrow external auditory meatus | |
| Low set ears | Q17.4 |
| Bat ear, prominent ear | Q17.5 |
| Unspecified and minor malformation of ear | Q17.9 |
| | |
| **Nose** | |
| Small nares | |
| Notched alas | |
| | |

| **Oral regions** | |
|---|---|
| Borderline small mandible/ minor micrognathia | |
| Aberrant frenula | |
| Enamel hypoplasia | |
| Malformed teeth | |
| High arched palate | Q38.50 |
| Tongue tie or cyst of tongue | Q38.1 |
| Macroglossia | Q38.2 |
| Macrostomia | Q18.4 |
| Microstomia | Q18.5 |
| Macrocheilia | Q18.6 |
| Microcheilia | Q18.7 |
| Ranula | |
| | |
| **Neck** | |
| Mild webbed neck | |
| Sinus, fistula or cyst of branchial cleft | Q18.0 |
| Preauricular sinus or cyst | Q18.1 |
| Other branchial cleft malformations | Q18.2 |
| Congenital malformation of face and neck, unspecified | Q18.9 |
| Torticollis | Q68.0 |
| | |
| **Hands** | |
| Duplication of thumbnail | |
| Enlarged or hypertrophic nails | Q84.5 |
| Single/abnormal palmar crease | Q82.80 |
| Unusual dermatoglyphics | |
| Clinodactyly (5$^{th}$ finger) | |
| Short fingers (4. 5. th finger) | |
| Accessorry carpal bones | Q74.00 |
| | |
| **Feet -Limb** | |
| Syndactyly (2nd-3rd toes) | |
| Gap between toes (1st-2nd) | |
| Short great toe | |
| Recessed toes (4th, 5th) | |
| Enlarged or hypertrophic nails | Q84.5 |
| Prominent calcaneus | |
| Clicking hip, subluxation or unstable hip | Q65.3-Q65.6 |
| Metatarsus varus or metatarsus adductus | Q66.2 |
| Hallux varus – other cong varus deformities of feet | Q66.3 |

| | |
|---|---|
| Talipes or pes calcaneovalgus | Q66.4 |
| Congenital pes planus | Q66.5 |
| Metatarsus varus – other cong valgus deformities of feet | Q66.6 |
| Pes cavus | Q66.7 |
| Clubfoot of postural origin - other cong deformities of feet | Q66.8 |
| Congenital deformity of feet, unspecified | Q66.9 |
| | |
| **Skin** | |
| Hemangioma (other than face or neck) | |
| Pigmented naevus – cong non-neoplastic naevus | Q82.5 |
| Neavus flammeus | Q82.50 |
| Strawberry naevus | Q82.51 |
| Lymphangioma | |
| Angioma | |
| Persistent lanugo | |
| Mongoloid spot (whites) | Q82.52 |
| Depigmented spot | |
| Unusual placement of nipples | |
| Accessory nipples | Q83.3 |
| Cafe-au-lait spot | |
| | |
| **Skeletal** | |
| Cubitus valgus | |
| Prominent sternum | Q67.7 |
| Depressed sternum | Q67.6 |
| Sternum bifidum | Q76.71 |
| Shieldlike chest, other cong deformities of chest | Q67.8 |
| Congenital deformity of spine | Q67.5 |
| Genua valgum | |
| Genua varum | |
| Genu recurvatum | Q68.21 |
| Congenital bowing of femur | Q68.3 |
| Congenital bowing of fibula and tibia | Q68.4 |
| Congenital bowing of long bones of leg, unspecified | Q68.5 |
| Spina bifida occulta | Q76.0 |
| Sacral dimple | |
| Cervical rib | Q76.5 |
| Absence of rib | Q76.60 |
| Accessory rib | Q76.62 |
| Congenital lordosis, postural | Q76.43 |
| | |

| | |
|---|---|
| **Brain** | |
| Arachnoid cyst | |
| Choroid plexus cyst | |
| Anomalies of septum pellucidum | |
| | |
| **Cardiovascular** | |
| Absence or hypoplasia of umbilical artery, single umbilical artery | Q27.0 |
| Functional or unspecified cardiac murmur | |
| Patent ductus arteriosus if GA < 37 weeks | Q25.0 if gestational age <37 weeks |
| Peripheral pulmonary artery stenosis | |
| Patent or persistent foramen ovale | Q21.11 |
| | |
| **Pulmonary** | |
| Accessory lobe of lung | Q33.1 |
| Congenital laryngeal stridor | Q31.4 |
| Laryngomalacia | Q31.4, Q31.5 |
| Tracheomalacia | Q32.0 |
| Azygos lobe of lung | Q33.10 |
| | |
| **Gastro-intestinal** | |
| Hiatus hernia | Q40.1 |
| Pyloric stenosis | Q40.0 |
| Diastasis recti | |
| Umbilical hernia | |
| Inguinal hernia | |
| Meckel's diverticulum | Q43.0 |
| Functional gastro-intestinal disorders | Q40.21, Q43.20, Q43.81, Q43.82 |
| Transient choledochal cyst | |
| Anterior anus | |
| | |
| **Renal** | |
| Vesico-ureteral-renal reflux | Q62.7 |
| Hydronephrosis with a pelvis dilatation less than 10 mm | |
| Hyperplastic and giant kidney | Q63.3 |
| Single renal cyst | Q61.0 |
| | |

| | |
|---|---|
| **External genitals** | |
| Deficient or hooded foreskin | |
| Undescended testicle | Q53 |
| Unspecified ectopic testis | |
| Retractile testis | Q55.20 |
| Hydrocele of testis | |
| Phymosis | |
| Bifid scrotum | Q55.21 |
| Curvature of penis lateral | |
| Hypoplasia of penis | |
| Hymen imperforatum | Q52.3 |
| Fusion of labia | Q52.5 |
| Prominent labia minora | |
| Enlarged clitoris | |
| Vaginal skin tag | |
| Cysts of vulva | |
| Transient ovarian cyst | |
| | |
| **Other** | |
| Congenital malformation, unspecified | Q89.9 |
| | |
| **Chromosomal** | |
| Balanced translocations or inversions in normal individuals | Q95.0, Q95.1 |

**"Non-congenital" anomalies**

Pyloric stenosis – there is controversy about the congenital nature of the majority of cases.

Patent ductus arteriosus in babies <37 weeks.

Hydrocephaly where a result of preterm birth rather than congenital: all cases among preterm births should be thoroughly checked before registration.

**Poorly specified anomalies**

Functional or unspecified cardiac murmur.

Laryngomalacia and tracheomalacia.

Functional gastro-intestinal disorders.

Undescended testicle. Registries may choose to record this locally if they can follow-up all babies to ascertain whether the testis descends normally.

Unspecified ectopic testis.

Vesico-ureteral reflux. Registries should record and transmit to EUROCAT the underlying anomaly, if present.

Clicking hip.

Clubfoot where there is no further specification of whether malformation or postural origin.

# Chapter 3.3

# EUROCAT Subgroups of Congenital Anomalies (Version 2012; implemented in EDMP 2012, used for website prevalence tables from April 2012) (Revised February 2012)

## 3.3 EUROCAT Subgroups of Congenital Anomalies (Version 2012; implemented in EDMP 2012, used for website prevalence tables from April 2012).

The subgroups are used to classify all cases of congenital anomaly in EUROCAT. The subgroups were revised to coincide with the implementation of Guide 1.3 in 2005 and uses ICD10-BPA codes only.
The ICD9 codes and the minor anomalies pre-2005 given in the table below are only used for retrospectively making subgroups for the earlier years of EUROCAT when these coding systems were used.

| EUROCAT Subgroups | ICD10-BPA | ICD9-BPA | Comments | Excluded minor anomalies post-2005 | Excluded minor anomalies pre-2005 | Subgroup binary variable number (al) |
|---|---|---|---|---|---|---|
| **All anomalies *** | Q-chapter, D215, D821, D1810, P350, P351, P371 | 74, 75, 27910, 2281, 76076, 76280, 7710, 7711, 77121 | | Exclude all minor anomalies as specified in Guide 1.3, Chapter 3.2 | Exclude all minor anomalies as specified in Guide 1.2 (ICD9 and ICD10) | al1 |
| **Nervous system** | Q00, Q01, Q02, Q03, Q04, Q05, Q06, Q07 | 740, 741, 742 | | Q0782 | | al2 |
| Neural Tube Defects | Q00, Q01, Q05 | 740, 741, 7420 | | | | al3 |
| Anencephalus and similar | Q00 | 740 | | | | al4 |
| Encephalocele | Q01 | 7420 | Exclude if associated with anencephalus subgroup | | | al5 |
| Spina Bifida | Q05 | 741 | Exclude if associated with anencephalus or encephalocele subgroups | | | al6 |
| Hydrocephalus | Q03 | 7423. Exclude 74232 | Exclude hydranencephaly. Exclude association with NTD subgroup | | | al7 |
| Microcephaly | Q02 | 7421 | Exclude association with NTD subgroup | | | al8 |
| Arhinencephaly / holoprosencephaly | Q041, Q042 | 74226 | | | | al9 |
| **Eye** | Q10-Q15 | 743 | | Q101-Q103, Q105, Q135 | 74365 | al10 |
| Anophthalmos / microphthalmos | Q110, Q111, Q112 | 7430, 7431 | | | | al11 |
| Anophthalmos | Q110, Q111 | 7430 | | | | al12 |
| Congenital cataract | Q120 | 74332 | | | | al13 |
| Congenital glaucoma | Q150 | 74320 | | | | al14 |
| **Ear, face and neck** | Q16, Q17, Q18 | 744 | | Q170-Q175, Q179, Q180-Q182, Q184-Q187, Q1880, Q189 | 74411, 74412, 7443, 74491 | al15 |
| Anotia | Q160 | 74401 | | | | al16 |

| EUROCAT Subgroups | ICD10-BPA | ICD9-BPA | Comments | Excluded minor anomalies post-2005 | Excluded minor anomalies pre-2005 | Subgroup binary variable number (al) |
|---|---|---|---|---|---|---|
| **Congenital Heart Defects** | Q20-Q26 | 745, 746, 7470-7474 | Exclude isolated PDA with GA<37 weeks | Q2111, Q250, 7470 if GA <37 weeks | Q250, 7470 if GA <37 weeks ** | al17 |
| Severe CHD | Q200, Q203, Q204, Q212, Q213, Q220, Q224, Q225, Q226, Q230, Q234, Q251, Q262 | 74500, 74510, 7452, 7453, 7456, 7461, 7462, 74600, 7463, 7467, 7471, 74742 | ICD9-BPA has no code for HRH | | | al97 |
| Common arterial truncus | Q200 | 74500 | | | | al18 |
| Transposition of great vessels | Q203 | 74510 | | | | al19 |
| Single ventricle | Q204 | 7453 | | | | al20 |
| VSD | Q210 | 7454 | | | | al21 |
| ASD | Q211 | 7455 | | Q2111 | | al22 |
| AVSD | Q212 | 7456 | | | | al23 |
| Tetralogy of Fallot | Q213 | 7452 | | | | al24 |
| Triscuspid atresia and stenosis | Q224 | 7461 | | | | al25 |
| Ebstein's anomaly | Q225 | 7462 | | | | al26 |
| Pulmonary valve stenosis | Q221 | 74601 | | | | al27 |
| Pulmonary valve atresia | Q220 | 74600 | | | | al28 |
| Aortic valve atresia/stenosis | Q230 | 7463 | ICD9-BPA has no code for atresia | | | al29 |
| Hypoplastic left heart | Q234 | 7467 | | | | al30 |
| Hypolastic right heart | Q226 | No code | | | | al31 |
| Coarctation of aorta | Q251 | 7471 | | | | al32 |
| Total anomalous pulm venous return | Q262 | 74742 | | | | al33 |
| PDA as **only** CHD in term infants (GA +37 weeks) | Q250 | 7470 | Livebirths only | | | al100 |
| **Respiratory** | Q30-Q34 | 748 | | Q314, Q315, Q320, Q331 | Q309, 74819 | al34 |
| Choanal atresia | Q300 | 7480 | | | | al35 |
| Cystic adenomatous malf of lung | Q3380 | No code | | | | al36 |
| **Oro-facial clefts** | Q35-Q37 | 7490, 7491, 7492 | Exclude association with holoprosencephaly or anencephaly subgroups | | | al101 |
| Cleft lip with or without cleft palate | Q36, Q37 | 7491, 7492 | Exclude association with holoprosencephaly or anencephaly subgroups | | | al102 |
| Cleft palate | Q35 | 7490 | Exclude association with cleft lip subgroup. Exclude association with holoprosencephaly or anencephaly subgroups | | | al103 |

| EUROCAT Subgroups | ICD10-BPA | ICD9-BPA | Comments | Excluded minor anomalies post-2005 | Excluded minor anomalies pre-2005 | Subgroup binary variable number (al) |
|---|---|---|---|---|---|---|
| **Digestive system** | Q38-Q45, Q790 | 750, 751, 7566 | | Exclude Q381, Q382, Q3850, Q400, Q401, Q4021, Q430, Q4320, Q4381, Q4382 | Q381, Q401, 7500, 7506 | al40 |
| Oesophageal atresia with or without trachea-oesophageal fistula | Q390-Q391 | 75030-75031 | | | | al41 |
| Duodenal atresia or stenosis | Q410 | 75110 | Exclude if also annular pancreas subgroup | | | al42 |
| Atresia or stenosis of other parts of small intestine | Q411-Q418 | 75111-75112 | | | | al43 |
| Ano-rectal atresia and stenosis | Q420-Q423 | 75121-75124 | | | | al44 |
| Hirschsprung's disease | Q431 | 75130-75133 | | | | al45 |
| Atresia of bile ducts | Q442 | 75165 | | | | al46 |
| Annular pancreas | Q451 | 75172 | | | | al47 |
| Diaphragmatic hernia | Q790 | 75661 | | | | al48 |
| **Abdominal wall defects** | Q792, Q793, Q795 | 75671, 75670, 75679 | | | | al49 |
| Gastroschisis | Q793 | 75671 | | | | al50 |
| Omphalocele | Q792 | 75670 | | | | al51 |
| **Urinary** | Q60-Q64, Q794 | 753, 75672, 75261 | | Q610, Q627, Q633 | | al52 |
| *Bilateral* renal agenesis including Potter syndrome | Q601, Q606 | 75300 | Exclude unilateral | | | al53 |
| Renal Dysplasia | Q614 | 75316 | | | | al54 |
| Congenital hydronephrosis | Q620 | 75320 | | | | al55 |
| Bladder exstrophy and / or epispadia | Q641, Q640 | 7535, 75261 | | | | al56 |
| Posterior urethral valve and / or prune belly | Q6420, Q794 | 75360, 75672 | | | | al57 |
| **Genital** | Q50-Q52, Q54-Q56 | 7520-7524, 75260, 75262, 7527-7529 | | Q523, Q525, Q5520, Q5521 | Q540, 75260# | al58 |
| Hypospadia | Q54 | 75260 | | | | al59 |
| Indeterminate sex | Q56 | 7527 | | | | al60 |
| **Limb** | Q65-Q74 | 7543-7548, 755 | | Q653-Q656, Q662-Q669, Q670-Q678, Q680, Q6821, Q683-Q685, Q7400 | 75432, 75452, 75460, 75473, 75481, 75560 | al61 |
| Limb reduction | Q71-Q73 | 7552-7554, 7556 | | | 75560 | al62 |
| Upper limb reduction | Q71 | 7552 | | | | al63 |
| Lower limb reduction | Q72 | 7553 | | | | al64 |
| Compete absence of a limb | Q710, Q720, Q730 | 75520, 75530, 75540 | | | | al65 |

| EUROCAT Subgroups | ICD10-BPA | ICD9-BPA | Comments | Excluded minor anomalies post-2005 | Excluded minor anomalies pre-2005 | Subgroup binary variable number (al) |
|---|---|---|---|---|---|---|
| Club foot – talipes equinovarus | Q660 | 75450 | | | | al66 |
| Hip dislocation and / or dyspasia | Q650-Q652, Q6580, Q6581 | 75430 | | | | al67 |
| Polydactyly | Q69 | 7550 | | | | al68 |
| Syndactyly | Q70 | 7551 | | | | al69 |
| **Other anomalies / syndromes** | | | | | | |
| Skeletal dysplasias | Q7402, Q77, Q7800, Q782-Q788, Q8716 | No code | | | | al104 |
| Craniosynostosis | Q750 | 75600 | | | | al75 |
| Congenital constriction bands / amniotic band | Q7980 | 76280 | | | | al76 |
| Situs inversus | Q893 | 7593 | | | | al79 |
| Conjoined twins | Q894 | 7594 | | | | al80 |
| Congenital skin disorders | Q80-Q82 | 7571, 7573 | | Q825, Q8280 | Q825, Q8280, Q8281, 75731, 75738 | al81 |
| Teratogenic syndromes with malformations | Q86, P350, P351, P371 | No code | | | | al82 |
| Fetal alcohol syndrome | Q860 | 76076 | | | | al83 |
| Valproate syndrome | Q8680 | No code | | | | al84 |
| Maternal infections resulting in malformations | P350, P351, P371 | 7710, 7711, 77121 | | | | al86 |
| Genetic syndromes + microdeletions | Q4471, Q6190, Q7484, Q751, Q754, Q7581, Q87, Q936, D821 Exclude Q8703, Q8704, Q8706, Q8708, Q8724, Q8726 | 7598, 27910 Exclude 759801,759895, 759844 | Exclude Associations and sequences | | | al105 |
| Sequences | Q606, Q6410, Q794, Q7980, Q8703, Q8708, Q8724, Q8980 | Only ICD9 codes for some sequences | | | | al108 |
| **Chromosomal** | Q90-Q92, Q93 , Q96-Q99 | 7580-7583, 7585-7589 | Exclude microdeletions Q936 | | | al88 |
| Down's syndrome | Q90 | 7580 | | | | al89 |
| Patau syndrome / trisomy 13 | Q914-Q917 | 7581 | | | | al90 |
| Edward syndrome / trisomy 18 | Q910-Q913 | 7582 | | | | al91 |
| Turner's syndrome | Q96 | 75860, 75861, 75862, 75869 | | | | al92 |
| Klinefleter's syndrome | Q980-Q984 | 7587 | | | | al93 |

* All Anomalies = ALL cases of congenital anomaly, excluding cases with only minor anomalies as defined in Chapter 3.2 for cases born post-2005. Cases with more than one anomaly are only counted once in the "All Anomalies" subgroup.
** The additional PDA exclusion (<2500 grams) listed in Guide 1.2 is not applied
**# The ICD9 code for hypospadias did not differentiate between the different types of hypospadias therefore minor cases of hypospadias (glandular l) were excluded at local registry level**

**Revised January 2013**

# Coding of EUROCAT Subgroups of Congenital Anomalies (Old Version)

**This version of the subgroups was used in EUROCAT publications from 1st July 2007 to 31st December 2011 (i.e. cases born between 2005 and 2009). Please note however, that there may be some overlap between older and newer versions of the subgroups at both the start and end of this period, as for example some papers may have been accepted in 2011, and yet not published until 2012.**

Guide 1.3 uses ICD10 codes only. The ICD9 codes given in the table below are only used for retrospectively making subgroups for the earlier years of EUROCAT when this coding system was used.

| EUROCAT Subgroups | ICD10-BPA | ICD9-BPA | Comments |
|---|---|---|---|
| **All anomalies *** | Q-chapter, D215, D821, D1810, P350, P351, P 371 | 74, 75, 27910, 2281, 7710, 7711, 77121, 76280 | **Exclude all minor anomalies listed in Chapter 3.2** |
| **Nervous system** | Q00, Q01, Q02, Q03, Q04, Q05, Q06, Q07 | 740, 741, 742 | |
| Neural Tube Defects | Q00, Q01, Q05 | 740, 741, 7420 | |
| Anencephalus and similar | Q00 | 740 | |
| Encephalocele | Q01 | 7420 | **Exclude if associated with anencephalus** |
| Spina Bifida | Q05 | 741 | **Exclude if associated with anencephalus or encephalocele** |
| Hydrocephalus | Q03 | 7423, Exclude 74232 | **Exclude hydranencephaly, Exclude association with NTDs** |
| Microcephaly | Q02 | 7421 | **Exclude association with NTDs** |
| Arhinencephaly / holoprosencephaly | Q041, Q042 | 74226 | |
| **Eye** | Q100, Q104, Q106-Q107, Q11-Q15 | 743 | **Exclude Q135** |
| Anophthalmos / microphthalmos | Q110, Q111, Q112 | 7430, 7431 | |
| Anophthalmos | Q110, Q111 | 7430 | |
| Congenital cataract | Q120 | 74332 | |
| Congenital glaucoma | Q150 | 74320 | |
| **Ear, face and neck** | Q16, Q178, Q183, Q188 | 744 | |
| Anotia | Q160 | 74401 | |
| **Congenital heart disease** | Q20-Q26 | 745, 746, 7470-7474 | **Exclude isolated PDA with GA <37 weeks (ICD9: 7470; ICD10: Q250)** |
| Severe CHD | Q200, Q203, Q204, Q212, Q213, Q220, Q224, Q225, Q226, Q230, Q234, Q251, Q262 | 74500, 74510, 7452, 7453, 7456, 7461, 7462, 74600, 7463, 7467, 7471, 74742 | **ICD9-BPA has no code for HRH** |
| Common arterial truncus | Q200 | 74500 | |
| Transposition of great vessels | Q203 | 74510 | |
| Single ventricle | Q204 | 7453 | |
| VSD | Q210 | 7454 | |
| ASD | Q211 | 7455 | |
| AVSD | Q212 | 7456 | |
| Tetralogy of Fallot | Q213 | 7452 | |
| Triscuspid atresia and stenosis | Q224 | 7461 | |
| Ebstein's anomaly | Q225 | 7462 | |

| EUROCAT Subgroups | ICD10-BPA | ICD9-BPA | Comments |
|---|---|---|---|
| Pulmonary valve stenosis | Q221 | 74601 | |
| Pulmonary valve atresia | Q220 | 74600 | |
| Aortic valve atresia/stenosis | Q230 | 7463 (no code for atresia) | |
| Hypoplastic left heart | Q234 | 7467 | |
| Hypoplastic right heart | Q226 | no code | |
| Coarctation of aorta | Q251 | 7471 | |
| Total anomalous pulm venous return | Q262 | 74742 | |
| **Respiratory** | Q30 - Q34 | 748 | **Exclude Q314, Q320** |
| Choanal atresia | Q300 | 7480 | |
| Cystic adenomatous malf of lung | Q3380 | no code | |
| **Oro-facial clefts** | Q35 - Q37 | 7490, 7491, 7492 | |
| Cleft lip with or without cleft palate | Q36, Q37 | 7491, 7492 | |
| Cleft palate | Q35 | 7490 | **Exclude association with cleft lip (7491-7492, Q36-Q37)** |
| **Digestive system** | Q38 – Q39, Q402-Q409, Q41-Q45, Q790 | 750, 751, 7566 | **Exclude Q381, Q382, Q3850, Q4021, Q430, Q4320, Q4381 Q4382** |
| Oesophageal atresia with or without tracheo-oesophageal fistula | Q390-Q391 | 75030-75031 | |
| Duodenal atresia or stenosis | Q410 | 75110 | **Exclude if also annular pancreas (Q451, 75172)** |
| Atresia or stenosis of other parts of small intestine | Q411-Q418 | 75111-75112 | |
| Ano-rectal atresia and stenosis | Q420 - Q423 | 75121-75124 | |
| Hirschsprung's disease | Q431 | 75130-75133 | |
| Atresia of bile ducts | Q442 | 75165 | |
| Annular pancreas | Q451 | 75172 | |
| Diaphragmatic hernia | Q790 | 75661 | |
| **Abdominal wall defects** | Q792, Q793, Q795 | 75671, 75670, 75679 | |
| Gastroschisis | Q793 | 75671 | |
| Omphalocele | Q792 | 75670 | |
| **Urinary** | Q60-Q64, Q794 | 753, 75672, 75261 | **Exclude Q627, Q633** |
| *Bilateral* renal agenesis including Potter syndrome | Q601, Q606 | 75300 | **Exclude unilateral** |
| Renal Dysplasia | Q614 | 75316 | |
| Congenital hydronephrosis | Q620 | 75320 | |
| Bladder exstrophy and/or epispadia | Q641, Q640 | 7535, 75261 | |
| Posterior urethral valve and/or prune belly | Q6420, Q794 | 75360, 75672 | |
| **Genital** | Q50-Q52 , Q54-56 | 7520-7524, 75260, 75262, 7527-7529 | **Exclude Q523, Q525** |
| Hypospadia | Q54 | 75260 | |
| Indeterminate sex | Q56 | 7527 | |
| **Limb** | Q650-Q652, Q658-Q659, Q660, Q681- Q682, Q688, Q69 - Q74 | 7543-7548, 755, | **Exclude Q6821** |
| Limb reduction | Q71-Q73 | 7552-7554, 75551 | |
| Upper limb reduction | Q71 | 7552 | |
| Lower limb reduction | Q72 | 7553 | |
| Complete absence of a limb | Q710, Q720, Q730 | 75520, 75530, 75540 | |
| Club foot - talipes equinovarus | Q660 | 75450 | |

| EUROCAT Subgroups | ICD10-BPA | ICD9-BPA | Comments |
|---|---|---|---|
| Hip dislocation and/or dysplasia | Q650-Q652, Q6580, Q6581 | 75430 | |
| Polydactyly | Q69 | 7550 | |
| Syndactyly | Q70 | 7551 | |
| Arthrogryposis multiplex congenital | Q743 | 75580 | |
| **Musculo-skeletal** | Q750-751, Q754-Q759, Q761-Q764, Q766-Q769, Q77, Q78, Q796-Q799 | 7540-7542, 7560-7565, 7568-7569, 76280 | |
| Thanatophoric dwarfism | Q771 | 756443, 756445 | |
| Jeunes syndrome | Q772 | 75640 | |
| Achondroplasia/ Hypochondroplasia | Q774 | 75643 | |
| Craniosynostosis | Q750 | 75600 | |
| Congenital constriction bands/amniotic band | Q7980 | 76280 | |
| **Other malformations** | Q27, Q28, Q80-Q85 Q89 | 7475-7479, 757, 7590-7597 | **Exclude Q270, Q825, Q8280, Q833, Q845, Q899** |
| Asplenia | Q8900 | 75900 | |
| Situs inversus | Q893 | 7593 | |
| Conjoined twins | Q894 | 7594 | |
| Disorders of skin | Q80-Q82 | 7571, 7573 | |
| **Teratogenic syndromes with malformations** | Q86, P350, P351, P371 | no code | |
| Fetal alcohol syndrome | Q860 | no code | |
| Valproate syndrome | Q8680 | no code | |
| Warfarin syndrome | Q862 | no code | |
| Maternal infections resulting in malformations | P350, P351, P371 | 7710, 7711, 77121 | |
| **Genetic syndromes + microdeletions** | Q87, Q936, D821 | 7598, 27910 | |
| **Chromosomal** | Q90-Q92, Q93 , Q96-Q99 | 7580-7583, 7585-7589 | **Exclude microdeletions Q936** |
| Down's syndrome | Q90 | 7580 | |
| Patau syndrome/trisomy 13 | Q914-Q917 | 7581 | |
| Edward syndrome/trisomy 18 | Q910-Q913 | 7582 | |
| Turner's syndrome | Q96 | 75860, 75861, 75862, 75869 | |
| Klinefelter's syndrome | Q980-Q984 | 7587 | |
| Cri-du-chat syndrome | Q934 | 75831 | |
| Wolff-Hirschorn syndrome | Q933 | 75832 | |

* All Anomalies = ALL cases of congenital anomaly, excluding cases with only minor anomalies as defined in Chapter 3.2. Cases with more than one anomaly are only counted once in the "All Anomalies" subgroup.

**This version of the subgroups was used in EUROCAT publications from 1st July 2007 to 31st December 2011 (i.e. cases born between 2005 and 2009). Please note however, that there may be some overlap between older and newer versions of the subgroups at both the start and end of this period, as for example some papers may have been accepted in 2011, and yet not published until 2012.**

# Chapter 3.4

## ~~Outside Malformation Chapter (Q) Codes~~
## Deleted, codes for all anomalies included is given in Chapter 3.3 (July 2011)

# Chapter 3.5

# Multiple Congenital Anomaly Algorithm (Revised January 2011)

## 3.5 Multiple Congenital Anomaly Algorithm

**Multiple anomaly flow-chart for monitoring of multiple anomalies**
At the moment, this should be for the Central Database only.
Only valid for years with ICD 10 codes

**Definition of a multiple anomaly case (MCA):**
Two or more unrelated major structural malformations that cannot be explained by an underlying syndrome or sequence

This means that the process of the flowchart is to find cases with two or more codes within the Q chapter, unless the case is transferred to other groups according to the steps described below.

**Name for groups**:
C: chromosomal
B: genetic syndrome, skeletal dysplasia and monogenic disorder
N: NTD isolated
A: isolated cardiac
R: isolated renal
I: isolated other
O: non-syndrome outside malformation chapter
M: potential multiple anomalies
T: teratogenic syndrome

**Minor, unspecified and invalid codes.**
The following codes are ignored in the flowchart, but appear in individual case output:

Guide 1.3 list of minors post 2005 to be used for all years
Balanced chromosomal rearrangements (7584 or Q95) as the only code
Multiple Malformation code (7597 or Q897)
Unspecified malf code (7599 or Q899)
No valid ICD code

Group X contains cases with only the above-listed codes.

**Outside Q-chapter codes (except the few codes accepted in “all anomalies”)**
These codes are ignored by the flowchart process but appear in the individual case output

**The flow-chart**

For 3 and 4 digit codes mentioned here, the coding also includes the codes with more digits
Only Q-codes are valid for the process after step 2
This is a hierarchical procedure

Step 1
Exclude all cases with a chromosomal code

Q90-Q93 except Q936, Q96-Q99, • Transfer to group C

Step 2
Exclude all cases with genetic syndrome codes, skeletal dysplasia and congenital skin disorder codes

Q87, Q936, D821,
Q77, Q7800, Q782-788, Q7402
Q80-Q82

Q4471 Alagille syndrome, Q6190 Meckel-Gruber, Q7484 Larsen syndrome
Q751 Crouzon /craniofacial dysostosis, Q754 Mandibulofacial dysostosis (Treacher Collin)
Q7581Frontonasal dysplasia

Excluding Q8703, Q8704, Q8706, Q8708, Q8724, Q8726 • Transfer to group B

Step 3
Exclude all cases with a code for teratogenic syndrome code

Q86, P350, P351, P371 • Transfer to group T

Step 4
Exclude all cases with a heterogenous syndrome code

Q761, Q7982, Q8581, Q8706 • Transfer to group M

Step 5
Exclude all cases with only NTD codes

Q00-Q01, Q05 • Transfer to group N

Step 6
Exclude all cases with codes only in cardiac chapter

Q20-Q26 • Transfer to group A

Step 7
Exclude all cases with codes only in renal chapter

Q60 – Q64, Q794 • Transfer to group R

Step 8
Exclude all cases with **only one code** within Q chapter
Include known local coding variations/errors

| | |
|---|---|
| If Q00-Q01, Q05 | • Transfer to group N |
| If Q20-Q26 | • Transfer to group A |
| If Q60 – Q64, Q794 | • Transfer to group R |
| If only one other Q-code | • Transfer to group I |

Step 9
Exclude all cases with codes only in eye chapter

| | |
|---|---|
| Q10-Q15 | • Transfer to group I |

Step 10
Exclude all cases with codes only with limb reduction defects

| | |
|---|---|
| Q71-Q73 | • Transfer to group I |

Step 11
Exclude all cases with codes only for hypospadias

| | |
|---|---|
| Q54 | • Transfer to group I |

Step 12
Exclude all cases with codes only for polydactyly

| | |
|---|---|
| Q69 | • Transfer to group I |

Step 13
Exclude all cases with codes only for reduction defects of brain

| | |
|---|---|
| Q04 | • Transfer to group I |

Step 14
Exclude all cases with codes only for hip anomalies

| | |
|---|---|
| Q65 | • Transfer to group I |

Step 15
Exclude all cases with codes only for syndactyly

| | |
|---|---|
| Q70 | • Transfer to group I |

Step 16
Exclude all cases with codes only for syndactyly + polydactyly

| | |
|---|---|
| Q69 and Q70 | • Transfer to group I |

Step 17
Exclude all cases with codes only for small intestinal atresia

| | |
|---|---|
| Q41 | • Transfer to group I |

Step 18
Exclude all cases with codes only for facial clefts

| | |
|---|---|
| Q35, Q36, Q37 | • Transfer to group I |

Step 19
Exclude all cases with the code for balanced chromosomal rearrangements and only one Q-code

| | |
|---|---|
| Q95 | |
| If (Q00-Q01, Q05) and Q95 | • Transfer to group N |
| If Q20-Q26 and Q95 | • Transfer to group A |
| If (Q60 – Q64, Q794) and Q95 | • Transfer to group R |
| If only one other Q-code and Q95 | • Transfer to group I |

Step 20
Exclude all cases with only outside Q chapter codes (without Q-codes)

| | |
|---|---|
| Not beginning with Q | |
| D1810 accepted as outside Q-code | • Transfer to group O |

Step 21
Exclude all known sequences or combinations of anomalies without other anomaly codes
(NB: Anyone of these codes may be used more than once – disregard duplicate codes)

| | |
|---|---|
| Spina bifida – talipes – hydrocephalus: | |
| Q05 coded with Q66 and/or Q03 | • Transfer to group N |
| Renal aplasia/dysplasia – lung hypoplasia - talipes: | |
| Q601/Q606 coded with Q336 and/or Q66 | • Transfer to group R |
| Omphalocele/gastroschisis – malrotation of gut – small intestinal atresia | |
| Q792/Q793 coded with Q433 and/or Q41 | • Transfer to group I |
| Anal atresia - rectovaginal fistula | |
| Q42 coded with Q522 | • Transfer to group I |

Diaphragmatic hernia – lung hypoplasia
Q790 coded with Q336 • Transfer to group I

Anencephalus - adrenal hypoplasia
Q000 coded with Q891 • Transfer to group N

Unspecified hydrocephalus - reduction defect of brain
Q039 coded with Q04 • Transfer to group I

Unspecified hydrocephalus – Arnold-Chiari
Q039 coded with Q070 • Transfer to group I
NTD – Arnold Chiari
Q01 or Q05 coded with Q070 • Transfer to group N

Amniotic band sequence
All cases with the code Q7980 • Transfer to group I

Caudal dysplasia sequence
All cases with the code Q8980 • Transfer to group I

Sirenomelia sequence
All cases coded with Q8724 • Transfer to group I

Cyclops sequence
All cases coded with Q8703 • Transfer to group I

Pierre Robin sequence
All cases coded with Q8708 as only code or with Q35-Q37 • Transfer to group I

Holoprosencephaly – median cleft lip
All cases coded with Q042 and Q361 • Transfer to group I

Step 22

The remaining cases are group M: potential multiple anomalies. Manual evaluation of all remaining cases before final inclusion into multiple anomaly group - or inclusion in one of the other groups.

Notes:
Then need to output group M cases as individual case lists with text description of anomalies as well as codes plus variables: ID no, registry, year of birth, type of birth, twin, GA, BW, karyotype (including written text), postmortem examination, when discovered

For the website review of potential multiple cases, a subgroup for “poorly specified cases” has to be added (could go to group X)

# Chapter 3.6

# Detailed Congenital Anomaly Coding Guidelines (Revised August 2013)

# 3.6 Detailed Malformation Coding Guidelines

## Coding tips for Guide 1.3

Remember always to give as specified code as possible

### Q00 Anencephaly and similar malformations

### Q01 Encephalocele

### Q02 Microcephaly

### Q03 Congenital hydrocephalus

CONGENITAL HYDROCEPHALUS
Definition: Dilatation of ventricular system with impaired circulation and absorption of the cerebrospinal fluid. The dilatation should not be due to primary atrophy of the brain, with or without enlargement of the skull.
*Please always specify the size of the ventricles.*

Hydrocephalus cases can be coded using the following codes

Q030 Malformation of aqueduct of Sylvius

Q031 Atresia of foramina of Magendie and Luschka or Dandy-Walker anomaly
Approx 75% of cases with Dandy-Walker have hydrocephalus, but this code is the only way to report the Dandy-Walker anomaly

Q038 Congenital ventriculomegaly may not be due to fluid circulation abnormalities, but should be reported if the size of the ventricles is 15 mm or more. For less severe prenatally detected ventriculomegaly (10-14 mm) it is recommended to follow the case until further imaging and a final diagnosis has been found postnatally.

Q039 Unspecified congenital hydrocephalus

Coding Committee June 2011

Q0380 CLOVERLEAF SKULL: It is caused by the premature closure of several sutures and is apparent from birth. The ICD/BPA code is wrong. Use Q7503 in stead
Coding Committee June 2011

### Q04 Other congenital malformations of brain

Q040/Q0400 malformation of/ agenesis of corpus callosum: do not use a hydrocephalus code for the dilatation of the ventricles associated with this anomaly.
Coding Committee June 2011

Q0435 HYDRANENCEPHALY
Congenital absence of cerebral hemispheres with preservation of midbrain and cerebellum. May result from widespread vascular occlusion, infections, prolonged severe hydrocephalus. Coding Committee June 2011

## Q05 Spina bifida

CODING OF SPINA BIFIDA
In ICD/BPA 10 coding of spina bifida should be based on one code only. The codes in Q05 describe both the site of the defect and if hydrocephalus is present or not. Code the highest position of the defect (ex: thoracic if both thoracic and lumbar). Add the 4.th digit to describe if the defect is open or closed. The BPA extension can be found under (http://www.eurocat.ulster.ac.uk/pdf/Q-Chapter.pdf). Coding Committee meeting 2006 and EUROCAT Communication July 2006

CODING OF SPINA BIFIDA WITH ARNOLD CHIARI MALFORMATION.
In ICD/BPA9 there was a specified code for spina bifida with Arnold Chiari malformation. This code does not exist in ICD/BPA10. For coding spina bifida with Arnold Chiari malformation use the best possible code for spina bifida within Q05 (se coding tips) and add the code for Arnold Chiari: Q070 Coding Committee 2007

SPINA BIFIDA OCCULTA AND OTHER VARIATIONS
We include all spina bifida cases in EUROCAT - open or covered - in our prevalence.
We exclude spina bifida occulta if the only malformation is the vertebrae detected by x-ray and no neurological deficits.
If only tethered cord or lipomylmyelomeningocele is present we recommend you use the code Q068. This means that we record the case but outside the NTD subgroup.
We have followed the advice from Peter Harper: Practical genetic counselling.
Coding Committee August 2007

## Q06 Other congenital malformations of spinal cord

TETHERED CORD.
Use the code Q068 “Other specified malformation of spinal cord” and specify tethered cord and spinal location in written text.
Coding Committee August 2007

LIPOMYELOMENINGOCELE
Use the code Q068 “Other specified malformation of spinal cord” and specify the malformation including location in text
Coding Committee August 2007

## Q07 Other congenital malformations of nervous system

CODING OF SPINA BIFIDA WITH ARNOLD CHIARI MALFORMATION.

In ICD/BPA9 there was a specified code for spina bifida with Arnold Chiari malformation. This code does not exist in ICD/BPA10. For coding spina bifida with Arnold Chiari malformation use the best possible code for spina bifida within Q05 (se coding tips) and add the code for Arnold Chiari: Q070
Coding Committee 2007

## Q10 Congenital malformations of eyelid, lacrimal apparatus and orbit

## Q11 Anophthalmos, microphthalmos and macrophthalmos

## Q12 Congenital lens malformations

## Q13 Congenital malformations of anterior segment of eye

## Q14 Congenital malformations of posterior segment of eye

## Q15 Other congenital malformations of eye

## Q16 Congenital malformations of ear causing hearing impairment

## Q17 Other congenital malformations of ear

## Q18 Other congenital malformations of face and neck

Dysmorphic face.
If a case with one or more major malformations also has a dysmorphic face but no syndrome diagnosis or karyotype anomaly, use the code Q189: “malformation of face and neck, unspecified” and give the written text: dysmorphic face. This code is on the list of minors for exclusion and therefore will not affect our prevalence data and subgroups. The advantage is that we will be able to see which cases in the total database may later prove to have a syndrome.
Coding Committee August 2007

## Q20 Congenital malformations of cardiac chambers and connections

ATRIAL ISOMERISM AND IVEMARK SYNDROME WITH ASPLENIA/POLYSPLENIA
Q206 is the code for atrial isomerism or Ivemark syndrome with or without asplenia/polysplenia. Add a code for the spleen anomalies if present: Q8900 asplenia or Q8908 polysplenia. Additional codes for situs inversus may also be added if present
Coding Committee June 2013

## Q21 Congenital malformations of cardiac septa

Q211 ASD

For ASD use the 4-digit codes to distinguish between ASD secundum (Q2110) and persistent foramen ovale (Q2111). In registries where information is available for ASD secundum (Q2110) include only defects with flow across the defect still present 6 months after birth.
Coding Committee August 2007

TETRALOGY OF FALLOT
The ICD10-code for Tetralogy of Fallot is Q213. Do not use other additional cardiac codes for this malformation.
The cardiac malformation “VSD+pulmonary valve stenosis” is a different entity/disease than Tetralogy of Fallot as etiology, epidemiology and outcome are different.
EUROCAT Communication January 2005

**Q22 Congenital malformations of pulmonary and tricuspid valves**

**Q23 Congenital malformations of aortic and mitral valves**

**Q24 Other congenital malformations of heart**

**Q25 Congenital malformations of great arteries**

**Q26 Congenital malformations of great veins**

**Q27 Other congenital malformations of peripheral vascular system**

**Q28 Other congenital malformations of circulatory system**

**Q30 Congenital malformations of nose**

**Q31 Congenital malformations of larynx**

**Q32 Congenital malformations of trachea and bronchus**

**Q33 Congenital malformations of lung**

LUNG HYPOPLASIA
Lung hypoplasia associated with diaphragmatic hernia or bilateral renal agenesis is a consequence of the first malformation and it will be counted/considered as a single malformation. Lung hypoplasia after preterm rupture of the membranes is not a malformation and should therefore not be reported to EUROCAT as a case.
EUROCAT Communication November 2003

Q3380 CCAM - Congenital cystadenomatoid malformation of the lung
If a CCAM is detected antenatally, please code for this anomaly postnatally (and hence send the case to EUROCAT) whether or not the CCAM is confirmed by X-ray after birth. The clinical status of the baby, and whether the CCAM has been confirmed, should be added by text. This will allow us to accurately document the prevalence of this anomaly.

Coding Committee June 2013

## Q34 Other congenital malformations of respiratory system

## Q35 Cleft palate

CLEFT PALATE
Use only one code within chapter Q35-37. Find the code which describes the malformation in the best way. Cleft lip with cleft palate has a single code
EUROCAT Communication November 2003

CLEFT PALATE
The coding committee has decided to recommend the use of the WHO codes instead of the BPA codes for cleft palate. See table under Coding documents (link)
Coding Committee August 2007

## Q36 Cleft lip

CLEFT LIP
Use only one code within chapter Q35-37. Find the code which describes the malformation in the best way. Cleft lip with cleft palate has a single code
EUROCAT Communication November 2003

CLEFT LIP
The coding committee has decided to recommend the use of the WHO codes instead of the BPA codes for cleft lip. For Q369 we still recommend to use the BPA 4.th digit. See table under Coding documents (link)
Coding Committee August 2007

## Q37 Cleft palate with cleft lip

CLEFT LIP AND PALATE
Use only one code within chapter Q35-37. Find the code which describes the malformation in the best way. Cleft lip with cleft palate has a single code
EUROCAT Communication November 2003

CLEFT LIP AND PALATE
The coding committee has decided to recommend the use of the WHO codes instead of the BPA codes for cleft lip and palate. See table under Coding documents (link)
Coding Committee August 2007

## Q38 Other congenital malformations of tongue, mouth and pharynx

## Q39 Congenital malformations of oesophagus

## Q40 Other congenital malformations of upper alimentary tract

## Q41 Congenital absence, atresia and stenosis of small intestine

## Q42 Congenital absence, atresia and stenosis of large intestine

## Q43 Other congenital malformations of intestine

## Q44 Congenital malformations of gallbladder, bile ducts and liver

## Q45 Other congenital malformations of digestive system

## Q50 Congenital malformations of ovaries, fallopian tubes and broad ligaments

## Q51 Congenital malformations of uterus and cervix

## Q52 Other congenital malformations of female genitalia

## Q53 Undescended testicle

## Q54 Hypospadias

HYPOSPADIA
Definition: The urethral meatus is abnormally located and is displaced proximally on the ventral surface of the penis – in mild cases on the glans itself and in more severe cases at some points along the ventral surface of the penile shaft.
It is strongly recommended to use a specified code for hypospadia (Q540 to Q543) instead of the unspecified code Q549. Please also give written text description and fill in the surgery variable.
Note: Deficient or hooded foreskin by itself is not hypospadia.
Coding Committee August 2007

## Q55 Other congenital malformations of male genital organs

## Q56 Indeterminate sex and pseudohermaphroditism

Indeterminate sex to be coded under malformations, not as syndrome
Coding Committee 2002

INDETERMINATE SEX
Problem: Indeterminate sex (Q564) is often over used to describe genital anomalies (ambiguous genitalia) when the sex of the baby has already been assigned.
If known to be male with ambiguous genitalia use a code to describe the genital anomaly where possible or Q559 if further details are unknown or without a specified code
If known to be female with ambiguous genitalia use a code to describe the genital anomaly where possible or Q529 if further details are unknown or without a specified code Indeterminate sex (Q564)

is only to be used when the sex of the baby is not known or not determined by karyotype
Coding Committee June 2012

## Q60 Renal agenesis and other reduction defects of kidney

## Q61 Cystic kidney disease

Q61.40 Multicystic dysplastic kidney, unilateral
This is distinct from polycystic kidneys. MCDK is usually unilateral and involves cysts of varying sizes separated by dysplastic parenchyma. The shape of the kidney is irregular and the normal renal architecture is lost. Multicystic dysplastic kidneys often shrink and disappear but if they are seen first as MCDK they should be coded as this and not as renal agenesis.

61.41 Multicystic dysplastic kidney, bilateral
Approximately 20% of MCDK are bilateral. This is usually a lethal condition that is primarily detected prenatally. The features are as above.

Q61.8 Other cystic kidney disease
Included here should be cystic kidneys associated with a systemic condition such as Tuberous sclerosis, MODY 5 (Maternal diabetes and renal cysts), Bardet-Biedl etc

Q61.9 Cystic kidney disease, unspecified
Included here should be: Kidneys that have cysts but normal parenchyma in between and prenatally kidneys that appear particularly bright (and often larger) than normal that are not polycystic or classic multicystic dysplasia

Coding Committee June 2011

## Q62 Congenital obstructive defects of renal pelvis and congenital malformations of ureter

HYDRONEPRHOSIS
Only report hydronephrosis if renal pelvis is $\geq$ 10 mm after birth
Coding Committee 2003

Q620 HYDRONEPHROSIS
Defined as an obstruction of the urinary flow from kidney to bladder. Report only major cases defined as a renal pelvis at or above 10 mm after birth. Specify in written text if the hydronephrosis is unilateral or bilateral and give the maximum size of the renal pelvis measured postnatally. Hydronephrosis caused by vesico-ureteral reflux should not be reported to EUROCAT.
Coding Committee December 2007

## Q63 Other congenital malformations of kidney

## Q64 Other congenital malformations of urinary system

OEIS COMPLEX
Q6410 Cloacal exstrophy. This code will include cases with OEIS complex as the literature state that these conditions are within the same spectrum. For OEIS complex, give the code Q6410 in malformation 1 and add codes for all major malformations of the case.
Coding Committee May 2010

## Q65 Congenital deformities of hip

## Q66 Congenital deformities of feet

CODING OF CLUBFOOT
Congenital clubfoot (Q660) is a major malformation for inclusion in the EUROCAT database. Another name for congenital clubfoot is talipes equinovarus and this name is used in the ICD10 written text. Clubfoot of postural origin is on the EUROCAT list of minor anomalies for exclusion (Q668). Any isolated case with this code is currently EXCLUDED from the EUROCAT database, although the code includes unspecified clubfoot. If you have a case of congenital clubfoot, you must make sure that you use the correct codes above, or your case will be excluded from the subgroup.
EUROCAT Communication December 2002

## Q67 Congenital musculoskeletal deformities of head, face, spine and chest

## Q68 Other congenital musculoskeletal deformities

## Q69 Polydactyly

## Q70 Syndactyly

## Q71 Reduction defects of upper limb

## Q72 Reduction defects of lower limb

## Q73 Reduction defects of unspecified limb

## Q74 Other congenital malformations of limb(s)

## Q75 Other congenital malformations of skull and face bones

Q7503: CLOVERLEAF SKULL
ICD/BPA 10 recommends a code in the hydrocephalus chapter, which is wrong. Use Q7503 for this anomaly**.**
Coding Committee June 2011

Q75.4 Mandibulofacial dysostosis – Franceschetti and Treacher-Collins

WHO recommend the code Q754 and ICD/BPA10 recommend the code Q870A. Both codes will be given in the syndrome guide. EUROCAT recommend from now to use the code Q754, to give written text description and to use the OMIM code 154500 for definite Treacher- Collins syndrome. Use OMIM code only where family history and biological markers confirm the syndrome
Coding Committee August 2007

## Q76 Congenital malformations of spine and bony thorax

## Q77 Osteochondrodysplasia with defects of growth of tubular bones and spine

SKELETAL DYSPLASIA
If a final diagnosis of a lethal or severe skeletal dysplasia is not possible, as in TOP or neonatal deaths without post mortem examination, use the code Q788. For late diagnosed unspecified skeletal dysplasias use Q789
Coding Committee August 2007

## Q78 Other osteochondrodysplasias

SKELETAL DYSPLASIA
If a final diagnosis of a lethal or severe skeletal dysplasia is not possible, as in TOP or neonatal deaths without post mortem examination, use the code Q788. For late diagnosed unspecified skeletal dysplasias use Q789
Coding Committee August 2007

## Q79 Congenital malformations of the musculoskeletal system, not elsewhere classified

Limb-body-wall complex
Q795 "Other congenital malformations of the abdominal wall" is the recommended code to us e in malf 1 and always give written text. Code all major anomalies which include encephalocele and craniofacial defects, internal organ defects, limb defects (mainly LRD), clubfoot.
Coding Committee May 2010

## Q80 Congenital ichthyosis

## Q81 Epidermolysis bullosa

## Q82 Other congenital malformations of skin

## Q83 Congenital malformations of breast

## Q84 Other congenital malformations of integument

## Q85 Phakomatoses, not elsewhere classified

## Q86 Congenital malformation syndromes due to known exogenous causes, not elsewhere classified

SUBGROUP: Teratogenic syndromes with congenital anomalies
Definition: syndrome caused by an environmental teratogen
Include as a EUROCAT case if at least one major anomaly present and you are sure about the aetiology (drug exposure, maternal infection etc)
Put the appropriate code in the syndrome field and codes for the associated congenital anomalies in the congenital anomaly fields
Specified codes for teratogenic syndromes are listed in the EUROCAT syndrome Guide and in the ICD/BPA10 Q-chapter
Always give text description of the syndrome and the associated anomalies (including minor anomalies and dysmorphic features without using a code for a major anomaly)
Coding Committee June 2012

Q860 FETAL ALCOHOL SYNDROME (dysmorphic)
Cases reported to EUROCAT as fetal alcohol syndrome must as minimum have dysmorphic features and/or major anomalies. Alcohol consumption must be confirmed locally. Add codes for all major anomalies
Coding Committee May 2010

## Q87 Other specified congenital malformation syndromes affecting multiple systems

Q870A and Q75.4 Mandibulofacial dysostosis – Franceschetti and Treacher-Collins
WHO recommend the code Q754 and ICD/BPA10 recommend the code Q870A. Both codes will be given in the syndrome guide. EUROCAT recommend from now to use the code Q754, to give written text description and to use the OMIM code 154500 for definite Treacher- Collins syndrome.
Coding Committee August 2007

Q 8708 PIERRE ROBIN
Pierre Robin is a sequence derived from micrognathia (hypoplastic mandible) leading to displacement of the tongue and obstructing the closure of the palate. It may be part of a genetic syndrome, but otherwise considered an isolated malformation. Correct coding will include Q8708 and written text in malf 1, a code for micrognathia (K070) in malf 2 and a cleft palate code in malf 3
Coding Committee February 2013

Q878: OTHER SPECIFIED SYNDROME
This code must always be accompanied with a written text with the syndrome name
EUROCAT Communication November 2004

## Q89 Other congenital malformations, not elsewhere classified

## Q90 Down's syndrome

## Q91 Edward's syndrome and Patau's syndrome

## Q92 Other trisomies and partial trisomies of the autosomes, not elsewhere classified

Q923 to be used for partial chromosomal duplication or partial trisomy.
Coding Committee June 2011

### Q93 Monosomies and deletions from the autosomes, not elsewhere classified

Q935 to be used for partial chromosomal deletions or partial monosomies including those detected by array
Coding Committee June 2011

CODING OF MICRODELETIONS: We recommend coding of both the syndrome and the microdeletion. This means that the syndrome should be coded in the syndrome field using both the ICD10/BPA code and give the syndrome name in the text field. In malformation 1 give the code for microdeletion (Q936) and give the name of the microdeletion in written text. Please note that microdeletions are considered syndromes and not chromosomal anomalies. Coding example: Case with Prader-Willi syndrome and 15q11-13 del: Code Q8715 in syndrome field and write “Prader-Willi” in text field. In malformation 1 field use code Q936 and write “15q11-13 del” in text field.
Coding committee meeting 2005

### Q95 Balanced rearrangements and structural markers, not elsewhere classified

### Q96 Turner's syndrome

### Q97 Other sex chromosome abnormalities, female phenotype, not elsewhere classified

### Q98 Other sex chromosome abnormalities, male phenotype, not elsewhere classified

Q982 Klinefelter male with karyotype 46XX
This condition does not exist and the code should not be used
Coding Committee May 2010

Q984 Klinefelter, unspecified
Alternative codes will usually be possible and better
Coding Committee May 2010

### Q99 Other chromosome abnormalities, not elsewhere classified

### Outside Q-chapter:

Please remember that the correct code for cystic hygroma is D1810 and for sacral teratoma D215
Central registry January 2008

TRAP sequence:

**T**win **R**eversed **A**rterial **P**erfusion is a rare complication of monochorionic twin pregnancies, involving an acardiac parasitic twin and an otherwise normal "pump" twin. The acardiac twin fails to develop a head, arms and a heart.
Cases of TRAP sequence should have as **a minimum** the following essential codes and essential text:
P023 TRAP sequence
Q24.9 Acardia (this is better than Q89.8 as it at least specifies heart)
Q00.00 Anencephaly
Other common malformations in TRAP sequence (eg. absence of upper limbs, rudimentary alimentary tract) should also be coded, but the 3 codes above with text are suggested as a minimum.
Coding Committee February 2013

**CODING OF PRE-PREGNANCY DIABETES**
For surveillance and research on etiology it is important that we can find all cases in the EUROCAT database with pre-pregnancy diabetes. Further type-1 diabetes in increasing in prevalence among children and young people. Pre-pregnancy diabetes is coded very heterogeneous among registries. Not all registries code maternal disease before pregnancy or drug use.

At the coding committee meeting in Graz in 2006 we recommended to code illness before pregnancy with codes within E10-E14, drugs with ATC codes for insulin and to code P701 "infant of diabetic mother" in the malformation variable (not the syndrome variable), even if the case is a TOPFA

# Chapter 3.7

# EUROCAT Description of the 2011 Congenital Anomaly Subgroups (Revised January 2012)

## 3.7 EUROCAT Description of the 2011 Congenital Anomaly Subgroups

| EUROCAT Subgroup | Description | Often diagnosed after one week of age |
|---|---|---|
| Nervous System | | |
| Neural Tube Defects: | Neural tube defects inlcude anencephalus, encephalocele, spina bifida and iniencephalus | no |
| Anencephalus and similar | Total or partial absence of brain tissue and the cranial vault.The face and eyes are present. (incompatible with life) | no |
| Encephalocele | Cystic expansion of meninges and brain tissue outside the cranium. Covered by normal or atrophic skin. | no |
| Spina Bifida | Midline defect of the osseous spine usually affecting the posterior arches resulting in a herniation or exposure of the spinal cord and/or meninges | no |
| Hydrocephaly | Dilatation of ventricular system, not due to primary atrophy of the brain, with or without enlargement of the skull | no |
| Microcephaly | A reduction in the size of the brain with a skull circumference less than three standard deviations below the mean for sex, age and ethnic origin. Definitions known to vary between clinicians and regions. | yes |
| 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) | yes |
| **Eye** | | |
| Anophthalmos / microphthalmos | - | |
| Anophthalmos | Unilateral or bilateral absence of the eye tissue. Clinical diagnosis | no |
| Microphthalmos | Small eye/eyes with smaller than normal axial length. Clinical diagnosis | yes |
| Cataract | Alteration in the transparency of the crystalline lens | yes |
| Congenital glaucoma | Large ocular globe as a result of increased ocular pressure in fetal life | yes |
| **Ear** | | |
| Anotia | Absent pinna, with or without atresia of ear canal | no |
| **Congenital heart defects (CHD)** | | |
| Severe CHD | 13 subgroups of severe CHD as defined below | yes |
| 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. | yes |
| Transposition of great vessels, complete | Total separation of circulation with the aorta arising from the right ventricle | no |

| | | |
|---|---|---|
| | and the pulmonary artery from the left ventricle | |
| Single ventricle | Only one complete ventricle with an inlet valve and an outlet portion even though the outlet valve is atretic | no |
| VSD | Defect in the ventricular septum | yes |
| ASD | Defect in the atrial septum | yes |
| AVSD | Central defect of the cardiac septa and a common atrioventricular valve, includes primum ASD defects | yes |
| Tetralogy of Fallot | VSD close to the aortic valves, infundibular and pulmonary valve stenosis and over-riding aorta across the VSD | yes |
| Tricuspid atresia and stenosis | Obstruction of the tricuspid valve and hypoplasia of the right ventricle | no |
| Ebstein's anomaly | Tricuspid valve displaced with large right atrium and small right ventricle | no |
| Pulmonary valve stenosis | Obstruction or narrowing of the pulmonary valves which may impair blood flow through the valves | yes |
| Pulmonary valve atresia | Lack of patency or failure of formation altogether of the pulmonary valve, resulting in obstruction of the blood flow from the right ventricle to the pulmonary artery | no |
| Aortic valve atresia/stenosis | Occlusion of aortic valve or stenosis of varying degree, often associated with bicuspid valves | yes for stenosis |
| 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 | no |
| Hypoplastic right heart | Hypoplasia of the right ventricle, always associated with other cardiac malformations | no |
| Coarctation of aorta | Constriction in the region of aorta where the ductus joins aorta | yes |
| Total anomalous pulmonary venous return | All four pulmonary veins drain to right atrium or one of the venous tributaries | No |
| PDA as only CHD in term infants | Open duct in infancy or later and requiring invasive treatment | yes |
| **Respiratory** | | |
| Choanal atresia | Bony or membraneous choanae with no passage from nose to pharynx | Yes for unilateral |
| Cystic adenomatous malf of lung | Cystic structures of the lung, usually unilateral | No |
| **Orofacial clefts** | | |
| Cleft lip with and without cleft palate | Clefting of the upper lip with or without clefting of the maxillary alveolar process and hard and soft palate | |
| Cleft palate | Fissure defect of the soft and/or hard palate(s) or submucous cleft without cleft lip | No |
| **Digestive system** | | |
| Oesophageal atresia with or without tracheo-oesophageal fistula | Occlusion or narrowing of the oesophagus with or without tracheo-oesophagael fistula | no |

| | | |
|---|---|---|
| Duodenal atresia and stenosis | Occlusion or narrowing of duodenum | no |
| Atresia and stenosis of other parts of small intestine | Occlusion or narrowing of other parts of small intestine | no |
| 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 | no |
| Hirschsprung's disease | Absence of the parasympatic ganglion nerve cells (aganglionosis) of the wall of the colon or rectum. May result in cong megacolon | yes |
| Atresia of bile ducts | Congenital absence of the lumen of the extrahepatic bile ducts | yes |
| Annular pancreas | pancreas surrounds the duodenum causing stenosis | yes |
| Diaphragmatic hernia | Defect in the diaphragm with protrusion of abdominal content into the thoracic cavity. Various degree of lung hypoplasia on the affected side | no |
| **Abdominal wall defects** | | |
| Gastroschisis | Protrusion of abdominal contents through an abdominal wall defect lateral to an intact umbilical cord and not covered by a membrane | No |
| Omphalocele | Herniation of abdominal content through the umbilical ring, the contents being covered by a membrane sometimes ruptured at the time of delivery | No |
| **Urinary** | | |
| *Bilateral* renal agenesis including Potter syndrome | Bilateral absence, agenesis, dysplasia or hypoplasia of kidneys including Potter's syndrome. Incompatible with life | no |
| Renal dysplasia | Maldevelopment of kidney tissue | yes |
| Congenital hydronephrosis | Obstruction of the urinary flow from kidney to bladder. Only if renal pelvis is 10 mm or more after birth | yes |
| Bladder extrophy | Defect in the closure of the bladder and lower abdominal wall | no |
| Posterior urethral valve and/or prune belly | Urethral obstruction with dilatation of bladder and hydronephrosis. In severe cases also distended abdomen | no |
| **Genital** | | |
| Hypospadias | The urethral meatus is abnormally located and is displaced proximally on the ventral surface of the penis | Yes |
| Indeterminate sex | Includes true and pseudohermaphroditism male or female | No |
| **Limb** | | |
| Limb reduction | Total or partial absence or severe hypoplasia of skeletal structure of the limbs | no |
| Upper limb reduction | Total or partial absence or severe hypoplasia of skeletal structure of the upper limb(s) | no |
| Lower limb reduction | Total or partial absence or severe hypoplasia of skeletal structure of the lower limb(s) | no |
| Complete absence of a limb | Complete absence of a limb | no |

| | | |
|---|---|---|
| Club foot - talipes equinovarus | Foot anomaly with equinus of the heel, varus of the hindfoot and adductus of the forefoot | no |
| Hip dislocation and/or dysplasia | Location of the head of the femur outside its normal position | no |
| Polydactyly | Extra digit or extra toe | no |
| Syndactyly | Partial or total webbing between 2 or more digits includes minor forms | yes |
| **Other anomalies / syndromes** | | |
| Skeletal dysplasia | A large group of genetic diseases with developmental disorders of chondro-osseous tissue | Yes |
| Craniosynostosis | Premature closure of cranial sutures | Yes |
| Congenital constriction bands / Amniotic bands | Bands in the amniotic fluid that causes constriction of part of the brain, body or limbs, including limb-body-wall complex | No |
| Situs inversus | Inverse position of thoracic or abdominal organs or both | Yes |
| Conjoined twins | Siamese twins | No |
| Congenital skin disorders | A group of mainly genetic skin disorders in the newborn | No |
| Teratogenic syndromes with malformations | Congenital anomalies in pregnancies with known teratogenic exposure | Yes |
| Fetal alcohol syndrome | Fetal exposure to alcohol during pregnancy with following impact on fetal growth, facial appearance and development | Yes |
| Valproate syndrome | Fetal exposure to valproate during pregnancy with impact on fetal growth, facial appearance and development. Often associated with spina bifida | Yes |
| Maternal infections resulting in malformation | Specific maternal viral infections during pregnancy resulting in congenital anomalies in the fetus or infant | Yes |
| Genetic syndromes and microdeletions | Clinically or genetically diagnosed syndromes with dysmorphic features or congenital anomalies with or without a microdeletion | Yes |
| Sequences | Pattern of multiple anomalies derived from a single known or presumed prior anomaly or mechanical factor. | No |
| **Chromosomal** | | |
| Down's syndrome | karyotype 47,XX +21 or 47,XY +21 and translocations/mosaicism | no |
| Patau syndrome/trisomy 13 | karyotype 47,XX +13 or 47,XY +13 and translocations/mosaicism | No |
| Edward syndrome/trisomy 18 | karyotype 47,XX +18 or 47,XY +18 and translocations/mosaicism | No |
| Turner's syndrome | karyotype 45,X or structural anomalies of X chromosome | Yes |
| Klinefelter's syndrome | karyotype 47,XXY or additional X-chromosomes | yes |

# Chapter 4

# Prevalence Rates

4.1 Calculation of Prevalence Rates

4.2 Interpretation of Prevalence Rates

# Chapter 4.1

# Calculation of Prevalence Rates (Issued September 2005)

## 4.1 Calculation of Prevalence Rates

In EUROCAT Prevalence 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 a total number of babies/fetuses. A baby is counted once only in any given prevalence rate.

| Total Prevalence Rate = No. of Cases (LB + FD+IA) ÷ No. of Births (live and still) |
|---|

| | |
|---|---|
| Cases | Cases of congenital anomaly in population |
| LB | Liveborn |
| FD | Fetal Deaths from 20 weeks gestation |
| IA | Induced Abortion or termination of pregnancy after prenatal diagnosis, at any gestation age |
| Birth (live and still) | All live and still births in the population as declared on official birth registrations |

NOTE: Slight discrepancies are present between numerator and denominator, but are not great enough to have an important effect on prevalence rates.

| Prevalence Rate at Birth = No. of Cases (LB + FD) ÷ No. of Births (live and still) |
|---|

The two measures are equal if there are not terminations of pregnancy after prenatal diagnosis in the population.

Differences in Total Prevalence Rates over time or between regions may reflect one or more of the following factors: genetic differences, environmental differences, differences in diagnostic services, differences in the methods of collecting epidemiological data, and even chance differences (see Chapter 4.2 Interpretation of Prevalence Rates).

Differences in Prevalence Rates at Birth over time or between regions may reflect the same factors as above, but also differences in prenatal screening policies and differences in frequency with which prenatal diagnosis if followed by termination of pregnancy.

| Livebirth Prevalence Rate = No. of Cases (LB) ÷ No. of Births (live) |
|---|

Livebirth prevalence rates are useful for the planning of services for affected children.

# Chapter 4.2

# Interpretation of Prevalence Rates (Issued September 2005)

## 4.2 Interpretation of Prevalence Rates

EUROCAT Registries follow a number of principles or organisation and registration to optimize the accuracy of estimation of prevalence rates and achieve standardization across regions. Many factors potentially affect the accuracy of estimation of prevalence rates at birth and total prevalence rates.

**Definition of the Population**
Registries can be "population-based" or "hospital-based". "Population-based" means that they cover residents of a defined geographical area. "Hospital-based" means that they cover births in selected hospitals. Where a registry is hospital-based, it is possible that there has been some selection of high-risk pregnancies towards or away from the selected hospitals, and thus estimated prevalence rates may be biased upwards or downwards. If a registry is population-based, it must ensure coverage of residents who deliver outside the geographic boundaries, who may also be at higher or lower risk than the rest of the population. In practice, there are also some variants of the above definitions based on knowledge of how information can be gathered and where mothers go to deliver. Assessing the potential for bias requires detailed knowledge of the local situation. All full member registries of EUROCAT are population-based. Three categories of population base are described in Chapter 5.1 (Template for Registry Description).

**Definition and Classification of Birth Defects and Diagnostic Practice**
Epidemiological data are derived from diagnoses made by clinicians working within given health service conditions. A registry is rarely in a position to impose a standard definition or diagnostic practice, though it may facilitate the adoption of standards. Minor anomalies are those which do not in themselves have serious medical, functional or cosmetic consequences for the child. EUROCAT applies a standard list of minor anomalies for Exclusion (see Chapter 3.2). These minor anomalies are not excluded if they appear in association with major anomalies. Some anomalies are present in gradations from minor to major forms, such as microphthalmia, microtia and microcephaly, the minor forms usually being much more common. Variable prevalence rates in these anomalies can be due to variable registration of their minor forms. Syndactyly can vary from slight webbing of the skin between two fingers to fusion of the bones between two fingers, and polydactyly can refer to the addition of a tiny digit to a fully formed digit. Unless a detailed description of syndactyly or polydactyly are given, it is difficult to standardize the exclusion of minor form.

Many variations in diagnostic practice may affect the reported prevalence of birth defects. For example, the accurate reporting of chromosomal anomalies (eg. Trisomy 13 or 18 and Down Syndrome) is dependent on karyotyping rates and indications for karyotyping. The autopsy rates for stillbirths and neonatal deaths will determine the likelihood that a birth defect is diagnosed, or the accuracy of the diagnosis, especially for conditions which are not externally visible such as serious congenital heart disease (eg. hypoplastic left heart syndrome).

Children with syndromes and multiple anomalies present particular classification problems. EUROCAT recommends recording of up to eight malformations, as well as a syndrome if present. Nevertheless, practice may vary as to whether all of the component malformations of a syndrome are recorded. Defects that are seen as consequences or other defects eg. "sequences" (ie. Hydrocephaly when associated with spina bifida) are counted only under the primary defect in EUROCAT prevalence rates (see Chapter 3.3)

**Ascertainment and Coding**

A diagnosis must not only be made, but also be recorded accurately, with the record reaching the registry, often through one or more intermediary records. There can be a loss of information between the place of diagnosis and the registry, either in terms of whether the child is recorded as having a birth defect at all, or in terms of the detail or accuracy of the diagnosis recorded (eg. is the baby recorded with congenital heart disease, or specifically with coarctation of aorta). Registries work hard to establish and maintain an information pathway which will lead to high case ascertainment (eg. the proportion of diagnosed cases who are registered), and accurate diagnostic information. Another step where there can be loss of information is between the text diagnostic information and the coding of that information. EUROCAT registries use versions 9 or 10 (more recently) of the International Classification of Disease, using the British Paediatric Association extension to allow more detail to be recorded. The McKusick (OMIM) Classification is used for conditions with Mendelian inheritance.

Overall case ascertainment probably never reaches 100%, and its level depends on a registry's methods of data collection. Registries need to use multiple sources of information. Under-ascertainment of some anomalies can occur if sources of information stop in the early neonatal period, as diagnoses may be made later than this. Specialist services treating children later than the postneonatal period are also vital for confirmation of diagnostic details. Some birth defects are now being discovered earlier in life due to prenatal and postnatal screening programmes. For example, cystic kidneys are more likely to be diagnosed early in life if there is ultrasound screening of the kidneys. This can lead to variation in prevalence rates between regions and over time as screening practice changes.

While EUROCAT recommends registration of fetal deaths from 20 weeks gestation, some registries have difficulties ascertaining fetal deaths outside the official stillbirth definition of their country (which may be 24 or 28 weeks or 500g). As malformed fetuses tend to be born prematurely or stillborn, ascertainment of fetal deaths of 20-27 weeks can influence prevalence rates substantially for certain congenital anomalies.

It is more difficult for registries covering very large populations to attain a high level of case ascertainment. Local contact with clinicians and other information sources is vital. The level of resources available to the registry or local office to employ suitable personnel, and the stability of those resources to retain experienced personnel, will also affect the quality of the

data collected. Data management itself can be complex, even the conceptually simple tasks such as not registering the same child twice.

**Termination of Pregnancy Following Prenatal Diagnosis**

We know that prenatal screening policies (and the resources for prenatal screening) vary enormously between different countries and between regions and even hospitals within countries. The "culture" in terms of how often prenatal diagnosis of a birth defect leads to termination of pregnancy also varies. For example, termination of pregnancy is very widespread for lethal conditions such as spina bifida. Thus, prenatal screening followed by termination of pregnancy introduces considerable geographic and temporal variation in prevalence rates at birth, and the proportion of terminations must be known or well estimated to assess whether there are real differences in "risk" between populations related to genetic or environmental risk factors.

Registries often require access to entirely different sources of information to ascertain terminations. Assessment of completeness of ascertainment of terminations requires detailed knowledge about local use of services (public and private) and information flows.

Ideally for epidemiologic purposes, terminations of pregnancy should be subject to the same rigour of diagnostic verification as live and stillbirths, but this is not always so. For example, autopsies may not be carried out to confirm the diagnosis, and a karyotype may not be performed where multiple malformations have been detected prenatally by ultrasound, to determine whether a chromosomal anomaly is present.

Reporting of terminations of pregnancy can lead to relative "over-ascertainment" of cases. The earlier in pregnancy the termination, the greater the probability that the pregnancy would, in other circumstances, have ended naturally in a spontaneous abortion. A spontaneous abortion would not necessarily have been examined for malformations or reported to the registry. These probabilities are generally small but, when the number of early terminations are high, it might result in a slight inflation of the total number of cases recorded compared to what would be expected if no terminations had been performed.

Prenatal screening and diagnosis, whether or not followed by termination, can also lead to relative "over-ascertainment" of cases when the average age of detection of a congenital anomaly is brought within the age coverage of the registry. This obviously depends on the age limit each registry applies to its information gathering, as well as diagnostic practice regarding the age when the anomaly would usually be detected postnatally. Similarly, the recorded proportion of all cases which are terminations of pregnancy may be inflated if prenatally diagnosed cases are ascertained by the registry more completely than postnatally diagnosed cases.

Laws and practices vary between countries as to the upper gestational age limit for termination (see Registry Descriptions of Member Registries for more detail – available on website).

The purpose of prenatal diagnosis is to increase the possibility of optimal management of the pregnancy and baby. While the issues of prenatal diagnosis and termination of pregnancy are intertwined in the evaluation of prevalence rates based on epidemiologic data, they are not intertwined in health service terms. Prenatal diagnosis can lead to beneficial outcomes such as effective early neonatal treatment or care. As outcomes improve, the practice of termination may well change.

# Chapter 5

# Registry Descriptions and Data Quality Indicators

5.1 Template for Registry Description

5.2 Registry description Questionnaire

5.3 Definition of Data Quality Indicators

# Chapter 5.1

# Template for Registry Description
# (Issued September 2005)

## 5.1 Template for Registry Description

This template provides the basis of the Registry Description which will be regularly updated on the EUROCAT website under "Member Registries". Please update on an annual basis at the same time as data transmission to Central Registry. Remember that the Registry Description should help to interpret your data for all years transmitted to EUROCAT, not just the most recent years.
The original Registry Description is based on the questionnaire in Chapter 5.2, but you have here the opportunity to give more detail and updates.

**Registry Country and Region (if applicable)**
**Registry Name**

**History and Funding**

1. Year started collecting data, first birth year collected, year joined EUROCAT, first birth year transmitted to EUROCAT
2. Who funds the registry – historical summary and current position (see Chapter 5.2, section B of Registry Description Questionnaire)
3. Describe the main aims of the registry (see chapter 5.2, QB4 of Registry Description Questionnaire)
4. Institution that hosts the registry and collaboration with regional and national institutions (Institutions to whom you regularly report your data)

**Population Coverage**

1. Population definition:
   - Population based I – All mothers resident in defined geographic area
   - Population based II – All mothers delivering within defined geographic area, irrespective of place of residence
   - Population based III – All mothers delivering in defined geographic area Excluding non-residents of that area
   - Hospital based – All mothers delivering in selected hospitals
2. Geographic area covered by registry (give year to which this relates)
3. Has the registry area been the same since its inception? (Please give an historical summary) Has there been an expansion/reduction of the registry area? Have there been any important demographic changes?
4. Annual number of births covered (give year to which this figure relates)
5. Percentage of births in country which registry covers (give year to which this figure relates)

**Sources of Ascertainment**

1. Whether voluntary/compulsory
2. Number of sources of cases ascertainment. Which sources of information eg. hospital, paediatric records, cytogenetic laboratory, pathology laboratory, child health services, specialised departments for diagnosis and treatment, midwives
3. What type of records and process of consultation/notification. Do you go through all records to find cases or rely on notification of cases by clinicians?
4. Do birth certificates include notification of congenital anomaly? Do you get this information to use as a source? How? When? (see QD3 of Registry Description Questionnaire)
5. Do death certificates allow for notification of congenital anomaly as cause of death? Do you get this information to use as a source? How? When? (See QD3 of Registry Description Questionnaire)
6. Percentage of cases spontaneously reported by more than one source (give year to which this refers)
7. What is the maximum age at diagnosis of livebirths for inclusion on the register (see Chapter 5.2 QD5 of Registry Description Questionnaire)

**Stillbirth Definition and Early Fetal Deaths**

1. Official stillbirth definition in your country (see Chapter 5.2 QE1 of Registry Description Questionnaire)
2. Do you obtain stillbirth certificates for cases of congenital anomaly? How?
3. Do you register spontaneous abortions registered? How do you obtain this information? What records are available? Is there a lower gestational age limit or weight for cases for inclusion in your registry, or for cases to be recorded in the sources you consult
4. Autopsy rates for stillbirths (see chapter 5.2, E5 of Registry Description Questionnaire). Give autopsy rates for stillbirths percentage (number), induced abortions following prenatal diagnosis percentage (number), early neonatal deaths (0-7 days) percentage (number), and deaths with congenital anomaly percentage (number). Do you obtain all autopsy reports for cases of congenital anomaly? How?

**Terminations**

1. Is termination of pregnancy following prenatal diagnosis of congenital anomaly legal – since what year?
2. If a congenital anomaly is diagnosed, what is the upper gestational age limit for termination? Does this differ for lethal anomalies?
3. What sources of information do you use to register terminations of pregnancy? How complete is ascertainment of terminations of pregnancy? What are the problems obtaining this information?
4. Prenatal diagnosis – describe the official policy in your region explicitly. What is offered (ultrasound/amniocentesis/chorionic villus sampling/ AFP/triple test, other. Why? When? How many? Is it free?

**Additional Exclusions and Availability of Exposure Data**

1. Please detail any feature of your registry which is unlike the typical EUROCAT registry requirements (eg. do you exclude any anomalies? Do you use a system of coding specific to your own registry or different to EUROCAT specifications?). If so, please explain giving the years when this applies
2. Conditions not recorded by your registry (see G4 of Registry Description Questionnaire): eg. metabolic disorders, Mendelian conditions, sacroccygeal/teratoma, Wilm's tumour, other tumours, infectious diseases
3. Please detail variables in Guide 1.3 which are not recorded (eg. which maternal exposure variables are not recorded)

**Denominators and Controls Information**

1. Where do you get your birth statistics from?
2. Do you record the number of births by maternal age group
3. Do you record the number of births/month
4. Do you collect information on controls? If so, how do you select controls?

**Ethics/Consent**

1. Please give details of procedure, percentage of cases where consent is withheld
2. Does the operation of your registry require the approval of an ethics committee or similar? Please give details

**Registry Staff and Computing**

1. How is your registry staffed (expertise and time)?
2. Give details of your computing system and software for data entry, data storage and data analysis

# Chapter 5.2

# Registry Description Questionnaire
# (Issued September 2005)

## 5.2 Registry Description Questionnaire

Dear Applicant Registry

The following information relating to Application for membership below is taken from the EUROCAT website (www.EUROCAT.ulster.ac.uk/memberreg/application.html):

1. A formal application (including Registry Description Questionnaire) for EUROCAT membership should be transmitted to the Project Leader.
2. An expertise and interest in the field of the epidemiology of congenital anomalies should be demonstrated.
3. The human and financial resources needed for running the registry should be made available at local level.
4. The registry should cover a geographically defined population.
5. The registration should include all types of congenital anomaly, should include registration of livebirths, stillbirths and induced abortions following prenatal diagnosis and should be based on multiple sources of ascertainment and emphasise high quality data. The registry should demonstrate the capacity to transmit to the Central Registry the EUROCAT standard data set on baby, diagnosis and risk factors (as specified in EUROCAT Guide 1.3)
6. There is an option for “Associate Members” of EUROCAT to transmit aggregate data only, in the form of number of cases by type of birth by year for a list of specified anomalies (See Chapter 3.3).

Applications must be approved by both the EUROCAT Steering Committee and the Registry Leaders.

**As a EUROCAT member applicant, we invite you to complete the following questionnaire. Please follow the instructions below and give as much detail as possible.**

INSTRUCTIONS TO ACCOMPANY QUESTIONNAIRE

The attached questionnaire has been designed for your convenience in Word 2000, which is compatible with lower versions of Word (eg. Word 97). This format allows you to fill it in directly on your computer and to subsequently return it by email. Please give as much detail as possible. If you have any problems with the completion of any of the questions, please do not hesitate to contact Central Registry at the email address below.

The following are useful instructions to simplify the process of questionnaire completion:

- The questions are navigated by using the tab or arrow up/down keys on your keyboard.
- Tick boxes can be selected (☐) by clicking once on the left mouse button. Boxes can be unselected (☐) by repeating this process.
- Text can be typed into the rectangular grey shaded boxes. These boxes will expand to accommodate the text inserted, so go on, answer the questions in detail - space is not a problem!!!!

Abbreviations used on the questionnaire are as follows:
LB = live births
SB = still births
TOP = terminations of pregnancy, following prenatal diagnosis of congenital anomaly

**Please return the questionnaire by email to:**
**EUROCAT Central Registry**
**EUROCAT@ulster.ac.uk**

# EUROCAT REGISTRY DESCRIPTION QUESTIONNAIRE

*Please read your information pack, Guide 1.3 and visit the EUROCAT website www.EUROCAT.ulster.ac.uk before completing this questionnaire*

I am applying for:
☐ Full Membership (complete entire questionnaire)
☐ Associate Membership (leave out questions: H1-5)

## *A CONTACT INFORMATION*

**Date** dd/mm/yy ☐

A1 **Name of Registry** (and acronym) ☐

A2 **Name of Registry Leader** ☐

A3 **Registry address** ☐

A4 **Registry telephone number** ☐

A5 **Registry fax number** ☐

A6 **Registry email address** ☐

A7 **Registry web home page** ☐

## *B REGISTRY ORGANISATION*

B1 **History of registry**

Year of establishment ☐
Year started collecting data ☐
First birth year collected ☐
Birth year from which you will send data to EUROCAT ☐

Membership of other international organisations
☐ none
☐ ICBDMS Since year ☐
☐ ENTIS Since year ☐
☐ OTIS Since year ☐
☐ Other, name ☐ Since year ☐

B2 **Type of data to be supplied to EUROCAT**

Please tick appropriate box

Full member: unidentifiable case data ☐

Associate member: aggregate data (ie. numbers of cases) ☐

B3 **Organisation of Registry** (present status)

(answer all A, B and C)

| A | B | C |
|---|---|---|
| ☐ Government/health authority | ☐ Ordered by national law | ☐ Steering committee |
| ☐ University or research institute | ☐ Regional/provincial law | ☐ No Steering committee |
| ☐ Hospital | ☐ Not ordered by law | |
| ☐ Private organisation | | |
| ☐ Other, specify below | | |

Further Information ☐

How is the registry funded? Please give name of the funder and explain what the funder's function is. ☐

How secure is the funding situation for the future of your registry ☐

B4 **Main aims of the Registry at present**

(indicate importance by scoring:
1 = Very important
2 = Less important
3 = Not important)

- ☐ Monitoring to detect new teratogenic exposures
- ☐ Producing statistics regarding prevalence
- ☐ Research, please give main areas ☐
- ☐ Audit of prenatal screening
- ☐ International cooperation
- ☐ Responding to public/lay requests/queries
- ☐ Assessment of reported clusters or environmental exposures
- ☐ Other, please detail

B5 **Why was the registry originally set up / funded** ☐

## *C POPULATION COVERAGE*

C1 **Type of Registry: Which of the following definitions are your prevalence rates based upon?**

Population-based: I = All mothers resident in defined geographic area
II = All mothers delivering within defined geographic area, irrespective of place of residence
III = All mothers delivering in defined geographic area excluding non-residents of that area

Hospital-based: All mothers delivering in selected hospitals, irrespective of place of residence

Choose only one box below. The definition refers to both malformed and denominator births

Delivery = LB (normal + malformed) + SB (normal +malformed) + TOP following prenatal diagnosis

☐ Population-based I ☐ % mothers delivering outside registry area*

☐ Population-based II ☐ % non-resident mothers delivering within registry area*

☐ Population-based III ☐ % non-resident mothers delivering within registry area*

☐ Hospital-based ☐ % deliveries in defined geographic area (specify below) occurring in the selected hospitals*

☐ Other, specify below

other ☐

* Year and source of information on which this estimate is based ☐

C2 **Geographical area covered by Registry *at present*** ☐
(give names of administrative boundaries, provinces, major cities etc. Attach a map if possible)

C3 **Has the registry population and geographic area been the same since the beginning of the Registry?** ☐ Yes ☐ No, specify below

If no, specify

Date ☐ Type of change, detail ☐

Date ☐ Type of change, detail ☐

Date ☐ Type of change, detail ☐

Date ☐ Type of change, detail ☐

Date ☐ Type of change, detail ☐

C4 **Annual number of births currently covered by Registry** (LB+SB) Number ☐ Year ☐ Name of source of information (e.g. Office for National Statistics for England and Wales) ☐

C5 **Births in the *country* in which Registry is situated** Number ☐ Year ☐ Name of source of information (e.g. Office for National Statistics for England and Wales) ☐

% covered by Registry ☐

C6 **Further information** ☐

# *D SOURCES AND ASCERTAINMENT*

D1 **Notification to the Registry is**
☐ Voluntary
☐ Compulsory Required by which law? ☐
☐ Other

**Further information** ☐

D2 **How are the malformed cases notified to the Registry? (tick all that apply)**
☐ notification of cases to the Registry by hospitals and other institutions
☐ active searching of patient notes by Registry staff
☐ active searching of hospital etc. discharge registers by Registry staff
☐ active searching of other local or national registers by Registry staff
☐ other, specify below
☐

D3 **Sources of information of Registry**

**WHO?**
Use the score system below, for all that apply, for each source
0 = Not used as a source of information
1 = Occasional notification of malformed cases seen
2 = Virtually complete notification of all malformed cases seen

| | |
|---|---|
| Community/GP doctors | ☐ |
| Hospital doctors | ☐ |
| Nurses | ☐ |
| Midwives | ☐ |
| Health visitors | ☐ |
| Other, specify | ☐ |

**WHERE AND HOW?**
Use the following score system :
1 = Registry routinely searches for new cases in their records
2 = Source notifies virtually all malformed cases seen to Registry
3 = Source occasionally notifies malformed cases seen to Registry
4 = Registry only consults this source for confirmatory or supplementary information about known cases

☐ Prenatal screening (ultrasound, serum testing, etc)
☐ Maternity units
☐ Paediatric departments
☐ Child health services
☐ Pathology labs
☐ Cytogenetic labs

☐ Echocardiology labs
☐ Other registries

Specialised departments for:

☐ Medical genetics
☐ Paediatric surgery
☐ Ophthalmology
☐ Orthopaedics
☐ Paediatric neurology
☐ Other, specify ☐

General sources of health and civil registration records:

☐ Hospital discharge records
☐ Birth certificates
☐ Death certificates

☐ Other

Please add any explanation that will help us understand how you ascertain cases to register ☐

D4 **% of cases reported by more than 1 source of information** ☐ % Year ☐

How is this calculated? ☐

D5 **What is the maximum age at postnatal diagnosis which would *routinely* result in a new notification to the Registry?**

☐ 1 week of life ☐ 1 month of life ☐ 1 year of life ☐ Childhood up to ☐ Years
☐ Other ☐ Any exceptions?

D6 **Are cases followed-up to find out more diagnostic details after a notification is received?** ☐ Yes, until age: ☐ Months ☐ Years ☐ No

Is follow-up applied to all or some anomalies? Please detail ☐

D7 **How are notifications and further details encouraged/ensured to reach the Registry? Please give details about where you think possible gaps in case/data ascertainment may be.** ☐

# *E STILLBIRTH AND EARLY FETAL DEATH*

E1 ***Detail the official stillbirth definition of your country which differentiates between stillbirths and spontaneous abortions in terms of*** **birthweight** ***and/or*** **gestational age** ☐

E2 **Does Registry get notification of malformed cases among:**

| | | | |
|---|---|---|---|
| Stillbirths | ☐ Yes | ☐ No | ☐ Sometimes |
| Early fetal deaths (spontaneous abortions) | ☐ Yes | ☐ No | ☐ Sometimes |
| Early neonatal deaths (0-7d) | ☐ Yes | ☐ No | ☐ Sometimes |
| Infant deaths (<1yr) | ☐ Yes | ☐ No | ☐ Sometimes |
| Other, specify ☐ | ☐ Yes | ☐ No | ☐ Sometimes |

E3 **If yes, does Registry get death certificates?**

| | Routinely | On request |
|---|---|---|
| Stillbirths | ☐ | ☐ |
| Early neonatal deaths | ☐ | ☐ |
| Infant deaths | ☐ | ☐ |

E4 **Is there a lower gestational age or weight limit to include early fetal deaths/spontaneous abortions in the Register?** ☐ Yes ☐ No

If yes, specify ☐ g ☐ and/ ☐ or ☐ wks of gestation

E5 **Autopsy rates (%) for most recent year of data available (state year)**

| | All deaths | Malformed cases on Register | Year |
|---|---|---|---|
| Stillbirths | ☐ | ☐ | ☐ |
| Early fetal deaths | ☐ | ☐ | ☐ |
| Early neonatal deaths | ☐ | ☐ | ☐ |
| Infant deaths | ☐ | ☐ | ☐ |
| Termination of pregnancy | ☐ | ☐ | ☐ |

Do the above autopsy rates refer exactly to your Registry population? ☐ Yes ☐ No
If not, explain ☐

Do specialist fetopathologists do most autopsies?

| | | |
|---|---|---|
| Stillbirths | ☐ Yes | ☐ No |
| Early fetal deaths | ☐ Yes | ☐ No |
| Early neonatal deaths | ☐ Yes | ☐ No |
| Infant deaths | ☐ Yes | ☐ No |
| Termination of pregnancy | ☐ Yes | ☐ No |

## *F PRENATAL SCREENING AND TERMINATION OF PREGNANCY*

F1 **Is termination of pregnancy for fetal malformation legal?** ☐ Yes ☐ No (Go to F6)

If yes, since (year) ☐

F2 **Up to what gestational age is termination legal for cases of fetal malformation?** ☐

F3 **Does the Registry register cases of termination of pregnancy for fetal malformation?** ☐ Yes since date dd/mm/yy ☐ ☐ No

F4 **Which sources provided notification of termination of pregnancy and how completely do you estimate terminations are notified to your registry? (please refer to D2 for list of sources)** ☐

F5 **Is there an official policy/policies for prenatal screening and diagnosis?** ☐ Yes ☐ No

If yes, this policy is
- ☐ National
- ☐ Regional (provinces, counties)
- ☐ Local (municipalities, hospitals
- ☐ Other, specify ☐

F6 **Is genetic screening performed routinely for mothers/parents during pregnancy?** ☐ Yes ☐ No

**What genetic diseases are screened for routinely ?**
- ☐ Sickle cell
- ☐ Thalassemias
- ☐ PKU
- ☐ Cystic fibrosis
- ☐ Other metabolic diseases Specify ☐
- ☐ Fragile X
- ☐ other Specify ☐

# *G REGISTRY INFORMATION: DIAGNOSES*

G1 **Does the Registry database contain diagnoses as text?** ☐ Yes ☐ No
**If yes, what diagnostic language is used in Registry?**
☐ English
☐ Other language, specify ☐

G2 **Coding system of congenital anomalies?**
☐ ICD10 original
☐ ICD10 national
☐ ICD10 BPA
☐ ICD9 original
☐ ICD9 national
☐ ICD9 BPA EUROCAT
☐ ICD9 Atlanta
☐ ICD9 BPA
☐ ICD8
☐ Other

If other, describe differences compared to ICD10 original ☐

G3 **Who does the diagnostic coding?**

☐ Registry leader
☐ Geneticist
☐ Other specialist, specify ☐
☐ Other Registry staff, specify ☐
☐ Notifier
☐ Other, specify ☐

Has any special training in coding been received? Please describe

G4 **Conditions <u>not</u> recorded in the Registry**
☐ specific structural malformations, *specify below*
☐ metabolic disorders
☐ Mendelian conditions (with no structural anomalies)
☐ sacrococcygeal teratoma / other teratomas
☐ tumours, malignancies
☐ other, *specify below*

Specify ☐

G5 **How many diagnoses can be registered per case?**
☐ 8 malformation codes and syndromes
☐ Less than 8
☐ Up to ☐ diagnoses
☐ No upper limit

G6 **Separate summary/syndrome diagnosis used in Registry?** ☐ Yes ☐ No

G7 ***Other special diagnostic codings used?***

☐ no

☐ McKusick - OMIM
☐ London Dysmorphology Data Base
☐ POSSUM
☐ Sakger
☐ Other, specify ☐

G8 **Do you exclude minor anomalies?** ☐ Yes ☐ No

If yes, which exclusion criteria do you use?

☐ EUROCAT (see EUROCAT Guide 1.3, Chapter 3.2)
☐ Other, specify ☐
☐ Registry's own criteria

If you do not use the EUROCAT exclusion criteria, please describe the differences or attach the list of exclusions you apply ☐

G9 **Are there any anomalies which you believe are not currently fully covered/could be improved by your Registry? Explain** ☐

G10 **Does your Registry collect information on surgical procedures for malformation? Explain** ☐

## *H AVAILABILITY OF EXPOSURE DATA AND INFORMATION ON PRENATAL SCREENING & DIAGNOSIS*

**Section H is to be completed by Applicants for full membership only**
**Guide 1.3 Chapter 2.2.1b details the variables and coding instructions for transmission to Central Registry. The core variables are shaded in grey. Please refer to the Guide when answering.**

H1 **Would you be able to transmit the core variables exactly as in Guide 1.3?** ☐ Yes ☐ No

If no, please detail the variable/s where there is difference and explain why you cannot transmit in standard format ☐

H2

| **Which of the following exposure variables do you record and which would you also transmit in standard Guide 1.3 format?** | **Record in the Register** | **Transmission in Guide 1.3 format possible** | **Quality of Registry data (completeness and accuracy)** | | **How is the data collected?** |
|---|---|---|---|---|---|
| | | | Good | Poor | |
| Occupation of mother | ☐ | ☐ | ☐ | ☐ | ☐ |
| Assisted conception | ☐ | ☐ | ☐ | ☐ | ☐ |
| Illness before pregnancy | ☐ | ☐ | ☐ | ☐ | ☐ |
| Illness during pregnancy | ☐ | ☐ | ☐ | ☐ | ☐ |
| Folic acid supplementation | ☐ | ☐ | ☐ | ☐ | ☐ |

| | | | | | |
|---|---|---|---|---|---|
| Drugs taken during first trimester | ☐ | ☐ | ☐ | ☐ | ☐ |
| Occupation of father | ☐ | ☐ | ☐ | ☐ | ☐ |
| Maternal education | | | | | |
| Social class of mother | | | | | |
| Social class of father | | | | | |
| Maternal migrant status | | | | | |

If you record but could not transmit, please explain why and would it be possible to send the data for special studies if requested? ☐

If you do not record the exposure data, please explain why and would it be possible to collect the data for special studies if requested? ☐

H3 **Can you transmit to EUROCAT the data from Guide 1.3 relating to whether the case was prenatally diagnosed, the gestational age at prenatal diagnosis and whether prenatal investigation was carried out? If yes, please specify below:** ☐ Yes ☐ No

Tick if you can send the data for the following EUROCAT Guide 1.3 variables:
- ☐ Whether prenatally diagnosed
- ☐ Gestational age at diagnosis
- ☐ First positive prenatal test

Further information ☐

H4 **Please read the information on EUROCAT's data management program (EDMP) before answering this question:**

**Are you willing to use this for <u>data transmission</u> to Central Registry?** ☐ Yes ☐ No

Further information ☐

H5 **Please tell us here about any coding which is specific to your registry which is different to EUROCAT specifications?** ☐

# I DENOMINATORS AND CONTROLS INFORMATION

I1 **Where do you obtain your birth statistics from?**

☐ National statistics, specify ☐

☐ Other, specify ☐

I2 **Do you record the number of births per maternal age group?** ☐ Yes ☐ No

If yes, please give figures below

Source ☐
Year ☐

| | Number of births (LB+SB) |
|---|---|
| <20 years | ☐ |
| 20-24 years | ☐ |
| 25-29 years | ☐ |
| 30-34 years | ☐ |
| 35-39 years | ☐ |
| 40-44 years | ☐ |
| >45 years | ☐ |

% of mothers >35 years old? ☐

I3 **Do you record the number of births per month?** ☐ Yes ☐ No

I4 **Do you collect information on controls?** ☐ Yes ☐ No

If yes, how do you select controls, how many per case and what information do you record about controls? ☐

# J ETHICS AND DATA SECURITY

J1 **Is there legislation in your country covering the protection of medical data?** ☐ Yes ☐ No

If yes, please specify and give year of implementation ☐

J2 **Do you require patient/parent consent for case registration?** ☐ Yes ☐ No

If yes, proportion of cases in which consent is given and comments: ☐

J3 **Do you have an advisory/steering committee?** ☐ Yes ☐ No

If yes, who is represented: ☐

## K EXPERTISE AVAILABLE TO THE REGISTRY

K1 **Who (name and institution) provides expertise to your Registry in the following areas:**

| | |
|---|---|
| Epidemiology | |
| Statistics | |
| Medical Genetics | |
| Obstetrics and Prenatal Diagnosis | |
| Paediatrics | |
| Public Health | |
| Other expertise, please state | |

K2 **Do you have easy access to computing expertise** ☐ Yes ☐ No

K3 **Which software packages do you use for data management?**

K4 **Which software packages do you use for data analysis?**

## L STAFF AT THE REGISTRY

L1 **List the staff at the registry, giving details requested**

Name, position, responsibility in the registry, academic/clinical background

## M SUMMARY OF RECENT CONGENITAL ANOMALY DATA COLLECTION

Please complete the tables below with summarised data for the last 5 years (or as many years as available) of completed data collection.

Years of data submitted

Table 1. Total number of cases and births per year and by type of birth:
N.B. Please EXCLUDE any babies who have only minor anomalies as defined in Chapter 3.2, Minor anomalies for Exclusion in Guide 1.3. (Please add more rows if required.)

| Year | Total Cases: | | | Total Births: | | Total Prevalence per 10,000 | | |
|---|---|---|---|---|---|---|---|---|
| | Livebirths | Fetal Deaths * | Induced Abortions ** | Livebirths | Stillbirths | LB | LB+FD | LB+FD+IA |
| | | | | | | | | |
| TOTAL | | | | | | | | |

* 20 weeks gestation and above ** Following prenatal diagnosis

**Table 2. Prevalence rates of anomalies, including livebirths, fetal deaths and induced abortions following prenatal diagnosis.**

Notes:

1. Fetal deaths include stillbirths and late fetal deaths, EXCLUDING fetal deaths before 20 weeks gestation. Induced abortions include terminations of pregnancy after prenatal diagnosis of anomaly at any gestational age. The total prevalence rate per 10,000 births is calculated by dividing the total cases (LB+FD+IA) by the total births (live and still).
2. The definition/coding of each anomaly can be found in Chapter 3.3, EUROCAT subgroups, Guide 1.3.

| Anomalies | LB No. | FD No. | IA No. | Prevalence (LB+FD+IA/ total births) |
|---|---|---|---|---|
| **All anomalies** | | | | |
| **Nervous system** | | | | |
| Neural Tube Defects | | | | |
| Anencephalus and similar | | | | |
| Encephalocele | | | | |
| Spina Bifida | | | | |
| Hydrocephalus | | | | |
| Microcephaly | | | | |
| Arhinencephaly / holoprosencephaly | | | | |
| **Eye** | | | | |
| Anophthalmos / microphthalmos | | | | |
| Anophthalmos | | | | |
| Congenital cataract | | | | |
| Congenital glaucoma | | | | |
| **Ear, face and neck** | | | | |
| Anotia | | | | |
| **Congenital heart disease** | | | | |
| Common arterial truncus | | | | |
| Transposition of great vessels | | | | |
| Single ventricle | | | | |
| VSD | | | | |
| ASD | | | | |
| AVSD | | | | |
| Tetralogy of Fallot | | | | |
| Triscuspid atresia and stenosis | | | | |
| Ebstein's anomaly | | | | |
| Pulmonary valve stenosis | | | | |
| Pulmonary valve atresia | | | | |
| Aortic valve atresia/stenosis | | | | |
| Hypoplastic left heart | | | | |
| Hypoplastic right heart | | | | |
| Coarctation of aorta | | | | |
| Total anomalous pulm venous return | | | | |
| **Respiratory** | | | | |
| Choanal atresia | | | | |

| Anomalies | LB No. | FD No. | IA No. | Prevalence (LB+FD+IA/ total births) |
|---|---|---|---|---|
| Cystic adenomatous malf of lung | | | | |
| **Oro-facial clefts** | | | | |
| Cleft lip with or without cleft palate | | | | |
| Cleft palate | | | | |
| **Digestive system** | | | | |
| Oesophageal atresia with or without tracheo-oesophageal fistula | | | | |
| Duodenal atresia or stenosis | | | | |
| Atresia or stenosis of other parts of small intestine | | | | |
| Ano-rectal atresia and stenosis | | | | |
| Hirschsprung's disease | | | | |
| Atresia of bile ducts | | | | |
| Annular pancreas | | | | |
| Diaphragmatic hernia | | | | |
| **Abdominal wall defects** | | | | |
| Gastroschisis | | | | |
| Omphalocele | | | | |
| **Urinary** | | | | |
| *Bilateral* renal agenesis including Potter syndrome | | | | |
| Renal Dysplasia | | | | |
| Congenital hydronephrosis | | | | |
| Bladder exstrophy and/or epispadia | | | | |
| Posterior urethral valve and/or prune belly | | | | |
| **Genital** | | | | |
| Hypospadia | | | | |
| Indeterminate sex | | | | |
| **Limb** | | | | |
| Limb reduction | | | | |
| Upper limb reduction | | | | |
| Lower limb reduction | | | | |
| Complete absence of a limb | | | | |
| Club foot - talipes equinovarus | | | | |
| Hip dislocation and/or dysplasia | | | | |
| Polydactyly | | | | |
| Syndactyly | | | | |
| Arthrogryposis multiplex congenital | | | | |
| **Musculo-skeletal** | | | | |
| Thanatophoric dwarfism | | | | |
| Jeunes syndrome | | | | |
| Achondroplasia | | | | |
| Craniosynostosis | | | | |
| Congenital constriction bands/amniotic band | | | | |
| **Other malformations** | | | | |
| Asplenia | | | | |
| Situs inversus | | | | |

| Anomalies | LB No. | FD No. | IA No. | Prevalence (LB+FD+IA/ total births) |
|---|---|---|---|---|
| Conjoined twins | | | | |
| Disorders of skin | | | | |
| **Teratogenic syndromes with malformations** | | | | |
| Fetal alcohol syndrome | | | | |
| Valproate syndrome | | | | |
| Warfarin syndrome | | | | |
| Maternal infections resulting in malformations | | | | |
| **Genetic syndromes + microdeletions** | | | | |
| **Chromosomal** | | | | |
| Down's syndrome | | | | |
| Patau syndrome/trisomy 13 | | | | |
| Edward syndrome/trisomy 18 | | | | |
| Turner's syndrome | | | | |
| Klinefelter's syndrome | | | | |
| Cri-du-chat syndrome | | | | |
| Wolff-Hirschorn syndrome | | | | |

**Thank you for completing this questionnaire.**

**Please attach any helpful documentation eg. your local notification form, annual report, maps etc.**

**Please ensure that you have attached all specifically requested documents.**

# Chapter 5.3

# Definition of Data Quality Indicators (Revised May 2008)

## 5.3 Definition of Data Quality Indicators

- Introduction
- Definitions of Data Quality Indicators
- Interpretation of Data Quality Indicators (to be added later)

**Introduction**

Complex factors influence the quality of data collected by a congenital anomaly registry. How, where and when are diagnoses of congenital anomaly made to residents of the region? How does the registry obtain and verify this information and how does it code it? Registry data are never a "perfect" description of which babies have congenital anomalies in the population and precisely what those anomalies are. Moreover, diagnostic definitions and methods change over time.

Registries may need to make difficult decisions about which types of information to prioritize in order to allocate limited resources, particularly whether to devote resources to collection of exposure information as well as diagnostic information. Some types of information are easier to obtain in some regions than others, depending on specialist referral systems for affected children, confidentiality restrictions, what is recorded in medical notes, availability of computer databases, and willingness to collaborate of key professionals.
EUROCAT's policy is to strive for high quality, accompanied by transparency as to strengths and weaknesses in data quality. Making Registry Descriptions and Data Quality Indicators (DQI) freely available will allow registries to evaluate their performance in relation to other registries, and will allow appropriate interpretations to be made of the results of data analyses. The DQI only have relevance in relation to the objectives of EUROCAT. Different data strengths are needed to participate in timely statistical monitoring in relation to environmental teratogenic exposures, or to evaluate the population sensitivity of prenatal screening for detecting affected pregnancies, or to establish the prevalence of rare genetic syndromes.

The first part of the evaluation of data quality is to read the detailed registry description (available on the EUROCAT website). Key questions to address in evaluating these registry descriptions include the following:

1. Does the registry describe its methods in enough detail? If not, the data emanating from the registry must be regarded as uninterpretable.
2. Is the registry fully population-based? If it covers residents of an area, how does it ensure coverage of residents delivering or seeking paediatric services elsewhere? If it is not entirely population-based, how does it avoid inclusion of selective referrals of high risk pregnancies into registry hospitals?
3. How does the registry ensure coverage of liveborn cases diagnosed after the early neonatal period?
4. How does the registry ensure coverage of late fetal deaths and stillbirths?

5. By what process are terminations of pregnancy following prenatal diagnosis identified in the population?
6. What specialist services are accessed for information, and are these services used for case identification or only for follow-up of known cases?

The second part of the evaluation of data quality is to look at the data generated by the registers.

The following sections provide a set of Data Quality Indicators and a description of completeness of registry data transmitted to the EUROCAT Central Database. The DQI of a particular registry can be compared to the EUROCAT average. Strong deviations on either side of the average should be examined. The set of data quality indicators has been produced under the
headings:

Completeness of case ascertainment
Accuracy of Diagnosis
Completeness of Information on core and non-core EUROCAT variables
Timeliness of data transmission
Availability of Denominator Information

This first ever set of DQI was discussed in Poznan June 2005.

**Definiations of Data Quality Indicators (2001-2005)**

<u>Ascertainment</u>

- Total congenital malformations prevalence = All cases in malformation chapter ICD10 (Q chapter) or ICD9 (range 740-759), including outside malformation codes (D821, D1810, P350, P351, P371, D215, 27910, 2281, 7710, 7711, 77121). EUROCAT minor anomalies are excluded.

- Ratio of Spina bifida to Anencephalus.

- Prevalence NTD = Neural Tube Defects

- Prevalence selected cardiac malformations
  Hypoplastic left heart, Transposition of Great Vessels, Tetralogy of Fallot, Coarctation of aorta or Common arterial truncus.

- Prevalence selected postnatal diagnosed malformations
  Corpus callosum anomalies, Cataract, Coarctation of aorta, Hirschprung's disease, Unilateral renal agenesis, or Craniosynostosis.

- Prevalence non-chromosomal syndromes

*ICD/BPA9 codes:* 27910, 755500, 755501. 755551, 75581, 75601, 756041, 756050, 75606, 756110, 75640, 756421, 75643, 756540, 756441, 756442, 756443, 756512, 75652, 756540, 756551, 756581, 75680, 75683, 75685, 756883, 756885, 75712, 757303, 75735, 759340, 759611, 759802, 759804, 759807, 759820, 759821, 759822, 759823, 759826, 758927, 759828, 759840, 759841, 759842, 759845, 759846, 759860, 759870, 759872, 759874, 759875, 759890, 759892, 759895, 759896, 759897, 759898, 759899, 7710, 7711, 77121

*ICD10 codes:* D821, P350, P351, P371, Q4471, Q6190, Q7402, Q7484, Q751, Q754, Q7581, Q761, Q771, Q772, Q775, Q776, Q7780, Q7800, Q781, Q782, Q783, Q7840, Q796, Q7982, Q823, Q8580, Q8581, Q8582, Q8583, Q860, Q861, Q862, Q8680, Q8682, Q87, Q8934, Q936, Q992,
Excluding: Q8703, Q8706, Q8708, Q8724

- Prevalence malformed fetal deaths calculated using total births.

Accuracy of Diagnosis

- % of possible multiple malformations in database excluding chromosomal or syndrome cases using the EUROCAT flow-chart for multiple malformations

- % fetal deaths and terminations of pregnancy with post-mortem examination performed

- % of chromosomal cases with a karyotype performed

- % of non-chromosomal / non-syndrome multiple malformation cases in database with known karyotype
  Using the EUROCAT flow-chart for multiple malformations

- Prevalence of selected Q-BPA extension codes (restricted to registries that use ICD10 coding).
  Selected Q-BPA codes = Q00.00, Q00.20, Q04.00, Q04.35, Q21.10, Q21.21, Q25.10, Q25.11, Q26.20, Q33.80, Q37.10, Q39.11, Q44.20, Q61.41, Q64.20, Q71.31, Q89.80

- Prevalence of selected Q-chapter unspecified codes (restricted to registries that use ICD10 coding).
  Selected unspecified codes = Q04.9, Q05.9, Q24.9, Q33.9, Q43.9, Q54.9, Q63.9, Q74.9, Q79.9, Q89.9, Q99.9

Completeness of information

Completeness of information describes the amount of **complete valid data** transmitted to Central Registry (eg. “Not known” values, invalid values, or missing/blank fields are counted as incomplete information).

- There are 7 core variables (Sex, Nbrbaby, Type, Weight, Gestlength, Survival, Agemo).

- There are 39 non-core variables (Place, Birthord, Nbrmalf, Death_date, Datedisc, Whendisc, Agedisc, Condisc, Amnio, Chorvilsam, Othertech, Karyo, PM, Datemo, Residmo, SA, IA, LB, SB, Totpreg, Occupmo, Assconcept, Illbef, Illdur1, Habit1, Unusexp, Drugs1, Datefa, Agefa, Occupfa, Mckusick, Modetrans, Consang, Prevsib, Sib1, Sibanom, Moanom, Faanom)

  Please note that there are some changes to the list of core and non-core variables for cases born pre-2005 and post-2005.

- Written text for syndrome and malformations

# Chapter 6

# Data Protection and Access to Data

## 6.1 EUROCAT Central Database

## 6.2 Release of Data

## 6.3 Guidelines on Security and Confidentiality for Staff Working in EUROCAT Central Registry

## 6.4 Data Protection Principles

(Policy regarding Security, Confidentiality, Release and Publication of Data: Agreed December 2011)

These guidelines have been submitted to the Project Management Committee of EUROCAT and to the Data Protection Co-ordinator of the University of Ulster. They will be reviewed periodically by Central Registry at the University of Ulster, Jordanstown. EUROCAT is covered within the University of Ulster's registration with the Information Commissioner.

# Chapter 6.1

# EUROCAT Central Database
# (Revised December 2011)

## 6.1 EUROCAT Central Database

1. EUROCAT is a European network of population-based registries for the epidemiologic surveillance of congenital anomalies which was established in 1979 (see http://www.eurocat-network.eu/). EUROCAT Central Registry is currently located at the University of Ulster.

2. EUROCAT Association is the association of member registries who, as a group, elect a president and a Steering Committee.

3. The standardised central database is held at the University of Ulster at Jordanstown and currently holds information on more than 397,000 cases of congenital anomaly among livebirths, stillbirths and terminations of pregnancy for fetal anomaly. The database is updated annually. A description of the data (variables) held on each case can be found in Chapter 2.2.1.

4. Full member registries biannually transmit a file containing records of individual cases, with standard data as described in Chapter 2.2.1 of this guide. Associate members transmit the numbers of cases by year and type of birth for an agreed list of congenital anomaly subgroups (see Chapter 2.2.4).

5. Many of the member registries use named records at local level for one or more of the following reasons:

   - To link reports arriving from several sources, and so avoid duplicate registration
   - To allow the follow-up of cases to confirm the diagnosis and to study the outcome of malformed children
   - To trace the cases in order to conduct prospective or retrospective aetiological studies
   - To allow the delivery of information to the malformed children and their families

6. Central Registry does not hold the names or addresses of cases. Instead cases are identified for the purposes of communication with local registries by a unique identifier (a maximum of 11 characters long, consisting of either numbers, letters or both). Local codes are used for designating places of birth (e.g. hospital) and areas of residence (e.g. municipality). They do not include postcodes. These codes used for transmission to Central Registry are for sufficiently large groups so that individual cases cannot be identified at Central Registry.

7. Legislation imposes a need for IT security and steps must be taken to ensure compliance with relevant requirements. Currently legislation in UK includes:

   The Data Protection Act 1998 (see Chapter 6.4 for a list of the principles)

The Copyright, Designs and Patents Act 1988
The Computer Misuse Act 1990
The Freedom of Information Act 2000
The Human Rights Act 1998
The Crime and Disorder Act 1998
BS7799 – British Standard Code of Practice on Information Security Management
The EC Directive of Legal Protection of Databases 1993

8. This document takes cognisance of the following documents.
   a) The Declaration of Helsinki (amended October 2000; given in full in appendix C of the extended 'Guidelines on Security, Confidentiality, Release and Publication of Data' at: http://www.eurocat-network.eu/content/EUROCAT-Policy-Public-Version.pdf
   b) The GMC Guidance on Confidentiality 2000 (Revised 2001)
   c) The Caldicott Committee: Report on the Review of Patient Information. December 1997
   d) University of Ulster, Data Protection Policy (see http://www.ulster.ac.uk/privacy/#dataprotection)

9. Local registries operate according to their own national laws with regard to the need for consent.

# Chapter 6.2

# Release of Data
# (Revised December 2011)

## 6.2 Release of Data

1. Only the Project Leader has the authority to release data on approval of the Steering Committee. The Project Leader is responsible for correct procedures being followed in the event of data release.

2. Customised tables (according to user needs) indicating prevalence, public health indicators, perinatal mortality and prenatal diagnosis rates derived from aggregate data of congenital anomalies in the member registries, are available on the web at www.eurocat-network.eu, and will be updated twice yearly. A full publications list is available on the EUROCAT website and copies of publications published by Central Registry can be obtained from Central Registry.

3. Further information and data requests for research or policy purposes are welcome and Central Registry will endeavour to process requests in as timely a fashion as possible subject to resources. Enquiries should be addressed in the first instance to the EUROCAT Administrator (currently Barbara Norton). Information requests may concern (i) aggregate numbers of cases according to specified case characteristics where these are not available in EUROCAT publications or on the website or (ii) a request for a data file of individual records. See Requesting EUROCAT Data at http://www.eurocat-network.eu/aboutus/requestingeurocatdata.

4. Please note that access to data cannot be permitted if a EUROCAT member has already been given approval for the area of study under question.

5. A data contract must be signed to indicate agreement with EUROCAT terms and conditions. The details are available at http://www.eurocat-network.eu/aboutus/requestingeurocatdata.

6. All requests for access to Central Registry data should be completed on the appropriate application form (http://www.eurocat-network.eu/aboutus/requestingeurocatdata) and sent by email to the Administrator, Barbara Norton. The completed application form will then be considered by the Steering Committee. After approval from the Steering Committee, written permission will be requested from registries for use of their data.

7. Data will not be released until ethical approval, where necessary, has been obtained.

8. The Steering Committee may recommend that one or two EUROCAT staff or members collaborate in the proposed research, in order to advise on analysis and interpretation of EUROCAT data.

9. The formula for acknowledgement and/or authorship, is outlined in the terms and conditions of the contract (http://www.eurocat-network.eu/aboutus/requestingeurocatdata).

10. A draft of any intended publication of EUROCAT data should be submitted to the EUROCAT Steering Committee for comment. This will be advisory only, except where factual inaccuracies are seen. Approval for the paper should be sought from each contributing registry, and registries have a right to withdraw their data from an intended publication if they consider it to be factually inaccurate, in accordance with the terms and conditions of the contract (http://www.eurocat-network.eu/aboutus/requestingeurocatdata).

11. Advance notification of the acceptance of a manuscript for publication based on EUROCAT data should be notified to the Administrator at Central Registry as soon as possible. A copy of the subsequent publication/s should be sent to the Administrator at Central Registry.

# Chapter 6.3

# Guidelines on Security and Confidentiality for Staff Working in EUROCAT Central Registry (Revised December 2011)

## 6.3 Guidelines on Security and Confidentiality for Staff Working in EUROCAT Central Registry

1. All personal data are regarded as confidential.

2. The Data Protection Act 1998 legislated on the fair obtaining, processing, storing and disclosure of data held on computer, paper or in machine form.

3. Security of data within EUROCAT Central Registry is maintained by careful procedures to maintain:

   a) The physical environment

      i EUROCAT data are held on personal computers for which passwords are required. It is backed up to a network drive to which only Central registry staff have access.
      ii The EUROCAT office (administration and data management) is accessed through a door with a special security lock. Filing cabinets with case information are locked.
      iii Central registry staff will work on individual data within this office only, except where written permission is given by the Project Leader.

   b) Staff practices and procedures regarding security of EUROCAT premises:

      All windows and doors must be secured at night and during prolonged absence from the room.

      All visitors must be accompanied while on the premises.

      Only EUROCAT staff, the Head of Security, university security staff, out of hours security staff providers “Resource” and cleaning staff at the University of Ulster also provided by “Resource” hold the key to the EUROCAT office. Master keys are controlled by a tracker key management system, and are signed in and out.

      Room access for system support purposes will only be available between 8am and 6pm in the presence of other EUROCAT staff.

      Staff must always “log out” of their terminal/PCs when leaving the office to attend meetings if the office is not attended by another member of staff.

      A back-up of the Central Database will be taken weekly (or more often if required) and stored in a locked filing cabinet in the office.

Archives of data files used for publications or studies will be held for up to 5 years following publication, stored in a locked filing cabinet in the administrative office. Archive files will contain only the cases and the variables used for the study, together with the local ID number in the event that case lists need checking.

The passwords for the computer system will be frequently changed.

4 Central registry staff may analyse any data in the Central registry database for purposes in keeping with EUROCAT objectives approved by the Project Leader, and for internal communication. Any publication of data must first be approved by both the Steering Committee and contributing registries, and is subject to the agreed authorship guidelines (see EUROCAT Terms and Conditions at http://www.eurocat-network.eu/aboutus/requestingeurocatdata).

5 Transport of Data

(a) Transfer/transport of information will periodically be reviewed.

(b) Data files for transmission by local registries or Central Registry should be password protected where possible to ensure security and confidentiality. The password should be sent separately. Emailed data should be sent to Ruth Greenlees at r.greenlees@ulster.ac.uk. The next version of EUROCAT Data Management Program may include a data encryption facility.

(c) Day and month of birth should be removed from individual data files leaving Central Registry except where express permission has been obtained on the basis of declared need for this information, security of information, and destruction when no longer needed.

# Chapter 6.4

# Data Protection Principles
# (Revised December 2011)

## 6.4 Data Protection Principles

The Data Protection Act 1988 (UK) requires the registration of data relating to individuals and held on computer. For all such data it is essential to abide by eight principles which govern the care and use made of the data.

DATA PROTECTION PRINCIPLES

1. Personal data shall be processed fairly and lawfully and, in particular, shall not be processed unless at least one of the conditions in Schedule 2 is met (for information on Schedule 2 see http://www.legislation.gov.uk/ukpga/1998/29/schedule/2), and in the case of sensitive personal data, at least one of the conditions in Schedule 3 (for information on Schedule 3 see http://www.legislation.gov.uk/ukpga/1998/29/schedule/3) is also met.

2. Personal data shall be obtained only for one or more specified and lawful purposes, and shall not be further processed in a manner incompatible with that purpose or those purposes.

3. Personal data shall be adequate, relevant and not excessive in relation to the purpose or purposes for which they are processed.

4. Personal data shall be accurate and, whenever necessary, kept up-to-date.

5. Personal data processed for any purpose or purposes shall not be kept for longer than is necessary for that purpose or those purposes. Thereafter it will be disposed of in accordance with the EUROCAT Terms and Conditions available at http://www.eurocat-network.eu/aboutus/requestingeurocatdata.

6. Personal data shall be processed in accordance with the rights of data subjects under this Act.

7. Appropriate technical and organisational measures shall be taken against unauthorised or unlawful processing of personal data and against accidental loss or destruction of, or damage to, personal data.

8. Personal data shall not be transferred to a country outside the European Economic Area unless that country or territory ensures an adequate level of protection for the rights and freedoms of data subjects in relation to the processing of personal data.

# Chapter 7

# Statistical Monitoring
# (To be added later)

# 7. Statistical Monitoring

# Chapter 8

# Amendments to Guide 1.3 since its Publication in June 2005

8.1 Summary of amendments to Guide 1.3 (Revised May 2012)
8.2 EUROCAT Subgroups of Congenital Anomalies (old version) (Revised February 2011)

# 8.1 Summary of Amendments to Guide 1.3

## Chapter 2

**Section 2.2.1a Summary of Variables**

Section 2.2.1a Variable 47 (ASSCONCEPT) – Communication 2011/2

Change 47** to 47## with a new footnote "## Variable compatible with Guide 1.2, but coding has been extended/modified. Code 10 added in March 2010".

**Section 2.2.1b Coding Instructions**

Section 2.2.1b Variable 3 (BIRTH_DATE) – Communication 2007/7

Please note that if you report a case from a multiple pregnancy where fetal reduction is carried out after prenatal diagnosis of malformation, the case should be reported as a termination of pregnancy with date of birth as the date of the fetal reduction. GA at birth will be GA at the date of the fetal reduction. As always with multiple pregnancies, please code number of births as 2(twin) or more in NBRBABY, and give as much detail about the co-twin in the text SP_TWIN (ie. type of birth, GA at birth, sex).

Section 2.2.1b Variable 11 (GESTLENGTH) – Communication 2007/3

Add the sentence "If the case is the result of fetal reduction give GA at fetocide" after "Give best estimate based on LMP and/or ultrasound determination".

Section 2.2.1b Variable 13 (DEATH_DATE) – Communication 2007/3

Delete "If died after 1 year of age and date of death is known, enter date of death instead of 1 or 9 (survival status at 1 year)".

Section 2.2.1b Variable 18 (WHEN DISC) – Communication 2008/6

The following explanations need to be added to the WHEN DISCOVERED variable: "For livebirths: where first suspicion of a malformation was at death OR at postmortem, when discovered is age at death (eg. at birth, <1 week, 1-4 weeks etc). For stillbirths: where first suspicion of a malformation was at birth OR at postmortem, when discovered is at birth eg. code=1). Please also complete variables 12 "SURVIVAL", 13 "DEATH-DATE", 19 "CONDISC" and 25 "PM"."

Section 2.2.1b Variable 18 (WHENDISC) – Communication 2007/3

Delete "serum screening" from 3rd paragraph of Explanation and Instructions.

Section 2.2.1b Variable 20 (AGEDISC) – Communication 2007/3

Delete "serum screening" from 3rd paragraph of Explanations and Instructions.

Section 2.2.1b Variable 21 (FIRSTPRE) – Communication 2008/6
The following explanation needs to be added to the FIRSTPRE variable: "If test performed and result negative, then the "When discovered" variable cannot be coded 6 (prenatal diagnosis)"

Section 2.2.1b Variable 27 (SYNDROME) – Communication 2007/12
The title of this field under Explanation and Instructions has been changed to "Syndrome or Association". The variable name remains the same.

Section 2.2.1b Variable 47 (ASSCONCEPT) – Communication 2011/2
Add 10=Assisted conception, type unknown.

Section 2.2.1b Variable 48 (OCCUPMO) – Communication 2006/12
Please note that the first link to ISCO classifications (maternal occupation – Section 2.2.1B, page 23) has been changed as the original link is no longer active. It should read: http://www.ilo.org/public/english/bureau/stat/isco/index.htm.

Section 2.2.1b Variable 54 (DRUGS1) – Communication 2009/6
The codes N**O**3A: Antiepileptic Drug and N**O**3AF**O**1: Carbamazepine have been changed to N**0**3A: Antiepileptic Drug and N**0**3AF**0**1: Carbamazepine.

Section 2.2.1b Variable 77 (SOCM) – Communication 2009/6
Code 8 has been changed from "Other" to "Other/Student". This change has also been implemented in the explanation and instructions.

Section 2.2.1b Variable 79 (SOCF) – Communication 2009/6
Code 8 has been changed from "Other" to "Other/Student". This change has also been implemented in the explanation and instructions.

**Section 2.2.2 EDMP Derived Variables**
Section 2.2.2 Variable dfyear – Communciation 2008/6
Should read "Father's date of birth (year) used for cases born before 2005".

Section 2.2.2 Variable al54 – Communication 2008/6
Cystic Kidney Disease becomes known as Renal Dysplasia.

Section 2.2.2 Variable al74 – Communication 2007/3
Achondroplasia becomes known as Achondroplasia/Hypochondroplasia.

**Section 2.2.4 Template for Associate Member Registry Data Transmission**
Section 2.2.4 Births by Maternal Age – Communication 2009/6
Age categories have been corrected to read 35-39 and not 34-39.

Section 2.2.4 Urinary Subgroup – Communication 2008/6
Cystic Kidney Disease becomes known as Renal Dysplasia.

Section 2.2.4 Musculo-Skeletal Subgroup – Communication 2007/3 Achondroplasia becomes known as Achondroplasia/Hypochondroplasia.

**Section 2.4 EUROCAT Data Management Program (EDMP Instructions)**
Section 2.4 EUROCAT EDMP Instructions – Communication 2009/6 and Communication 2012/4
This section has been updated.

**Section 2.5 Data Validation Rountines**
Section 2.5 Diagnosis – Communication 2008/6
The following validation rule need to be added: If WHEN DISCOVERED=6, then FIRST POSITIVE PRENATAL TEST must be coded as 1-7 or 9."

## Chapter 3

**Section 3.2 Minor Anomalies for Exclusion**
Section 3.2 Skin Subgroup – Communication 2009/6
The word "white" has been deleted from "Mongoloid spot (whites)" to just read ""Mongoloid spot".

Section 3.2 Cardiovascular Subgroup – Communication 2008/6
Patent or persistent foramen ovale should read Q21.11 instead of 21.11.

Section 3.2 Pulmonary Subgroup – Communication 2007/3
Add code Q31.5 to Laryngomalacia.

Section 3.2 Neck Subgroup – Communication 2007/3
After "Other Branchial Cleft Malformations" add "Congenital Malformations of Face and Neck, Unspecified" with code Q18.9.

**Section 3.3 Coding of EUROCAT Subgroups of Congenital Anomalies**
Section 3.3 Coding of EUROCAT Subgroups of Congenital Anomalies – Communication 2012/4
Complete section updated.

Section 3.3 All Anomalies Subgroup – Communication 2008/6
Add the ICD9 code 76280.

Section 3.3 Musculo-Skeletal Subgroup – Communication 2008/6
Add the ICD9 code 76280.

Section 3.3 Urinary Subgroup – Communication 2008/6

Cystic Kidney is now called Renal Dysplasia and the code has been restricted to Q614 only (ICD9 75316).

Chapter 3.3 Digestive System Subgroup – Communication 2007/12
Add the ICD10 code Q790 for Diaphragmatic Hernia.

Section 3.3 Musculo-Skeletal Subgroup – Communication 2007/12
Add the ICD9 code 76280 for Congenital Constriction Bands/Amniotic Band.

Section 3.3 Ear, Face and Neck Subgroup – Communication 2007/3
The ICD10-BPA should read "Q16, Q178, Q183, Q187-188".

Section 3.3 Limb Subgroup – Communication 2007/3
The ICD9-BPA code for "Limb Reduction" should read "7552-7554, 75551".

Section 3.3 Musculo-Skeletal Subgroup – Communication2007/3
Achondroplasia becomes known as Achondroplasia/Hypochondroplasia.

**Section 3.4 Outside Malformation Chapter (Q) Codes**
Section 3.4 - Communication 2009/6
"Haemangioma (more than $4cm^2$)*" has been excluded.

Section 3.4 – Communication 2007/3
Congenital CMV should read "P351".

**Section 3.5 Multiple Malformations Algorithm**
Section 3.5 - Communication 2009/6
This section has been updated.

**Section 3.6 Detailed Malformation Coding Guidelines**
Section 3.6 – Communication 2012/8
New coding tips have been included for:

- Indeterminate sex
- Teratogenic syndromes

Section 3.6 – Communication 2011/9
New coding tips have been included for:

- Congenital hydrocephalus
- Hydracephalus
- Cystic kidney disease
- Cloverleaf skull
- Other trisomies, monsomies and deletions

Section 3.6 – Communication 2011/2
New Coding Tips have been included for:

- Cloacal Extrophy
- Limb Body Wall Complex
- Fetal Alcohol Syndrome
- Klinefelter.

Section 3.6 – Communication 2008/6
New coding tips have been included for:

- Spina Bifida Occulta and Other Variations
- Tethered cord
- Lipomyelomeningocele
- ASD
- Cleft Palate
- Cleft Lip and Palate
- Mandibulofacial dysostosis
- Skeletal Dysplasia.

**Section 3.7 EUROCAT Description of the 2011 Congenital Anomaly Subgroups**
Section 3.7 – Communication 2012/4
Complete section updated.

Section 3.7 – Communication 2007/12
This is a new section which has been added to Guide 1.3.

## Chapter 5

**Section 5.2 Registry Descriptions Questionnaire**
Section 5.2 Table 2 - Communication 2009/6
"Cystic Kidney Disease" has been changed to "Renal Dysplasia".

**Section 5.3 Definition of Data Quality Indicators**
Section 5.3 – Communication 2009/6
This section has been added.

## Chapter 6

**Section 6.1 EUROCAT Central Database**
Section 6.1 - Communication 2009/6
Point 3 has changed the figure "250,000" to"308,000".

**Section 6.3 Guidelines on Security & Confidentiality For Staff Working in EUROCAT Central Registry**

Section 6.3 – Communication 2009/6
Point 3ia has been reworded.

## Chapter 8

**Section 8.1 Summary of Amendments to Guide 1.3**
Section 8.1 – Communication 2012/4
Creation of new section within the Chapter.
Addition of Severe CHD as a new subgroup.

**Section 8.2 EUROCAT Subgroups of Congenital Anomalies (old Version)**
Section 8.2 – Communication 2012/4
Creation of a new section within the Chapter.

## 8.2 EUROCAT Subgroups of Congenital Anomalies (old version)

**This version of the subgroups was first issued in September 2005, with minor modifications over time as detailed in Chapter 8.1. These subgroups were used in EUROCAT papers and tables extracted from the EUROCAT database between 1 July 2007 to 31 March 2012.**

Guide 1.3 uses ICD10 codes only. The ICD9 codes given in the table below are only used for retrospectively making subgroups for the earlier years of EUROCAT when this coding system was used.

| EUROCAT Subgroups | ICD10-BPA | ICD9-BPA | Comments |
|---|---|---|---|
| **All anomalies *** | Q-chapter, D215, D821, D1810, P350, P351, P 371 | 74, 75, 27910, 2281, 7710, 7711, 77121, 76280 | **Exclude all minor anomalies listed in Chapter 3.2** |
| **Nervous system** | Q00, Q01, Q02, Q03, Q04, Q05, Q06, Q07 | 740, 741, 742 | |
| Neural Tube Defects | Q00, Q01, Q05 | 740, 741, 7420 | |
| Anencephalus and similar | Q00 | 740 | |
| Encephalocele | Q01 | 7420 | **Exclude if associated with anencephalus** |
| Spina Bifida | Q05 | 741 | **Exclude if associated with anencephalus or encephalocele** |
| Hydrocephalus | Q03 | 7423, Exclude 74232 | **Exclude hydranencephaly, Exclude association with NTDs** |
| Microcephaly | Q02 | 7421 | **Exclude association with NTDs** |
| Arhinencephaly / holoprosencephaly | Q041, Q042 | 74226 | |
| **Eye** | Q100, Q104, Q106-Q107, Q11-Q15 | 743 | **Exclude Q135** |
| Anophthalmos / microphthalmos | Q110, Q111, Q112 | 7430, 7431 | |
| Anophthalmos | Q110, Q111 | 7430 | |
| Congenital cataract | Q120 | 74332 | |
| Congenital glaucoma | Q150 | 74320 | |
| **Ear, face and neck** | Q16, Q178, Q183, Q188 | 744 | |
| Anotia | Q160 | 74401 | |
| **Congenital heart disease** | Q20-Q26 | 745, 746, 7470-7474 | **Exclude isolated PDA with GA <37 weeks (ICD9: 7470; ICD10: Q250)** |
| Severe CHD | Q200, Q203, Q204, Q212, Q213, Q220, Q224, Q225, Q226, Q230, Q234, Q251, Q262 | 74500, 74510, 7452, 7453, 7456, 7461, 7462, 74600, 7463, 7467, 7471, 74742 | **ICD9-BPA has no code for HRH** |
| Common arterial truncus | Q200 | 74500 | |
| Transposition of great vessels | Q203 | 74510 | |
| Single ventricle | Q204 | 7453 | |
| VSD | Q210 | 7454 | |
| ASD | Q211 | 7455 | |
| AVSD | Q212 | 7456 | |
| Tetralogy of Fallot | Q213 | 7452 | |
| Triscuspid atresia and stenosis | Q224 | 7461 | |

| EUROCAT Subgroups | ICD10-BPA | ICD9-BPA | Comments |
|---|---|---|---|
| Ebstein's anomaly | Q225 | 7462 | |
| Pulmonary valve stenosis | Q221 | 74601 | |
| Pulmonary valve atresia | Q220 | 74600 | |
| Aortic valve atresia/stenosis | Q230 | 7463 (no code for atresia) | |
| Hypoplastic left heart | Q234 | 7467 | |
| Hypoplastic right heart | Q226 | no code | |
| Coarctation of aorta | Q251 | 7471 | |
| Total anomalous pulm venous return | Q262 | 74742 | |
| **Respiratory** | Q30 - Q34 | 748 | **Exclude Q314, Q320** |
| Choanal atresia | Q300 | 7480 | |
| Cystic adenomatous malf of lung | Q3380 | no code | |
| **Oro-facial clefts** | Q35 - Q37 | 7490, 7491, 7492 | |
| Cleft lip with or without cleft palate | Q36, Q37 | 7491, 7492 | |
| Cleft palate | Q35 | 7490 | **Exclude association with cleft lip (7491-7492, Q36-Q37)** |
| **Digestive system** | Q38 – Q39, Q402-Q409, Q41-Q45, Q790 | 750, 751, 7566 | **Exclude Q381, Q382, Q3850, Q4021, Q430, Q4320, Q4381 Q4382** |
| Oesophageal atresia with or without tracheo-oesophageal fistula | Q390-Q391 | 75030-75031 | |
| Duodenal atresia or stenosis | Q410 | 75110 | **Exclude if also annular pancreas (Q451, 75172)** |
| Atresia or stenosis of other parts of small intestine | Q411-Q418 | 75111-75112 | |
| Ano-rectal atresia and stenosis | Q420 - Q423 | 75121-75124 | |
| Hirschsprung's disease | Q431 | 75130-75133 | |
| Atresia of bile ducts | Q442 | 75165 | |
| Annular pancreas | Q451 | 75172 | |
| Diaphragmatic hernia | Q790 | 75661 | |
| **Abdominal wall defects** | Q792, Q793, Q795 | 75671, 75670, 75679 | |
| Gastroschisis | Q793 | 75671 | |
| Omphalocele | Q792 | 75670 | |
| **Urinary** | Q60-Q64, Q794 | 753, 75672, 75261 | **Exclude Q627, Q633** |
| *Bilateral* renal agenesis including Potter syndrome | Q601, Q606 | 75300 | **Exclude unilateral** |
| Renal Dysplasia | Q614 | 75316 | |
| Congenital hydronephrosis | Q620 | 75320 | |
| Bladder exstrophy and/or epispadia | Q641, Q640 | 7535, 75261 | |
| Posterior urethral valve and/or prune belly | Q6420, Q794 | 75360, 75672 | |
| **Genital** | Q50-Q52 , Q54-56 | 7520-7524, 75260, 75262, 7527-7529 | **Exclude Q523, Q525** |
| Hypospadia | Q54 | 75260 | |
| Indeterminate sex | Q56 | 7527 | |
| **Limb** | Q650-Q652, Q658-Q659, Q660, Q681- Q682, Q688, Q69 - Q74 | 7543-7548, 755, | **Exclude Q6821** |
| Limb reduction | Q71-Q73 | 7552-7554, 75551 | |
| Upper limb reduction | Q71 | 7552 | |
| Lower limb reduction | Q72 | 7553 | |
| Complete absence of a limb | Q710, Q720, Q730 | 75520, 75530, 75540 | |

| EUROCAT Subgroups | ICD10-BPA | ICD9-BPA | Comments |
|---|---|---|---|
| Club foot - talipes equinovarus | Q660 | 75450 | |
| Hip dislocation and/or dysplasia | Q650-Q652, Q6580, Q6581 | 75430 | |
| Polydactyly | Q69 | 7550 | |
| Syndactyly | Q70 | 7551 | |
| Arthrogryposis multiplex congenital | Q743 | 75580 | |
| **Musculo-skeletal** | Q750-751, Q754-Q759, Q761-Q764, Q766-Q769, Q77, Q78, Q796-Q799 | 7540-7542, 7560-7565, 7568-7569, 76280 | |
| Thanatophoric dwarfism | Q771 | 756443, 756445 | |
| Jeunes syndrome | Q772 | 75640 | |
| Achondroplasia/ Hypochondroplasia | Q774 | 75643 | |
| Craniosynostosis | Q750 | 75600 | |
| Congenital constriction bands/amniotic band | Q7980 | 76280 | |
| **Other malformations** | Q27, Q28, Q80-Q85 Q89 | 7475-7479, 757, 7590-7597 | **Exclude Q270, Q825, Q8280, Q833, Q845, Q899** |
| Asplenia | Q8900 | 75900 | |
| Situs inversus | Q893 | 7593 | |
| Conjoined twins | Q894 | 7594 | |
| Disorders of skin | Q80-Q82 | 7571, 7573 | |
| **Teratogenic syndromes with malformations** | Q86, P350, P351, P371 | no code | |
| Fetal alcohol syndrome | Q860 | no code | |
| Valproate syndrome | Q8680 | no code | |
| Warfarin syndrome | Q862 | no code | |
| Maternal infections resulting in malformations | P350, P351, P371 | 7710, 7711, 77121 | |
| **Genetic syndromes + microdeletions** | Q87, Q936, D821 | 7598, 27910 | |
| **Chromosomal** | Q90-Q92, Q93 , Q96-Q99 | 7580-7583, 7585-7589 | **Exclude microdeletions Q936** |
| Down's syndrome | Q90 | 7580 | |
| Patau syndrome/trisomy 13 | Q914-Q917 | 7581 | |
| Edward syndrome/trisomy 18 | Q910-Q913 | 7582 | |
| Turner's syndrome | Q96 | 75860, 75861, 75862, 75869 | |
| Klinefelter's syndrome | Q980-Q984 | 7587 | |
| Cri-du-chat syndrome | Q934 | 75831 | |
| Wolff-Hirschorn syndrome | Q933 | 75832 | |

* All Anomalies = ALL cases of congenital anomaly, excluding cases with only minor anomalies as defined in Chapter 3.2. Cases with more than one anomaly are only counted once in the “All Anomalies” subgroup.