EUROCAT Guide 1.4 and Reference Documents

(2013)

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WHO Collaborating Centre for the Surveillance of Congenital Anomalies

Contents

Chapter 1 Aims and Objectives

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 three 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 for research into the interaction of genetic and environmental factors in causing congenital anomalies.

2. For the planning and evaluation of preventive 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, secondary prevention by prenatal diagnosis to prepare for birth and treatment, and tertiary prevention through paediatric surgery, 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 expertise or financial resources.

3. 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 and diagnostic methods. A population-based approach is important, for the reasons given above.

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.

In chapter 2 we define the EUROCAT core dataset to be collected by all registries, and an extended dataset with optional non-core data items. 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. 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 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.

With the advent of electronic healthcare databases, the opportunity arises to link registers with other databases e.g. with prescription databases for pharmacovigilance purposes. Registers can also be linked to spatial environmental databases through the place of residence of the case.

Risk factor data must be present for both cases and controls (non-malformed children) in order to be interpretable in terms of the risk of all anomalies combined. While all registers collect basic information about the number of births in their population by type of birth and maternal age, only a few registers routinely collect the same set of risk factor information on control babies as on case babies. There are useful approaches to analyzing risk factor data among malformed cases only, using a case-malformed control approach where children with different malformations act as controls for each other. 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

1. Full members of EUROCAT transmit to Central Registry an encrypted, password-protected 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 1.4 is a revision of Guide 1.3 for use for all births from 1st January 2013. Guide 1.3 should continue to be used for births between 1st January 2005 and 31st December 2012. Guide 1.2 should continue to be used for births up to 31st December 2004. Guide1.4 is compatible with the EUROCAT Data Management Program (EDMP v6.05 26/03/13 onwards).

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 program is downloadable from the Membership Only area of the EUROCAT website or can be provided by e-mail by Central Registry and will run on Microsoft Access 2000, 2002, 2003, 2007, or 2010 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 blue 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.

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

2.2.1a Summary of Variables (core variables are shaded blue)

Variable Number	Variable Name	Variable heading
Baby and Mother – Variables 1 to 18		
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*	BMI	Maternal Body Mass Index
17	RESIDMO	Mother's residence code
18	TOTPREG	Total number of previous pregnancies
Diagnosis – Variables 19 to 57		
19**	WHENDISC	When discovered
20	CONDISC	Condition at discovery
21	AGEDISC	If prenatally diagnosed, gestational age at discovery
22**	FIRSTPRE	First positive prenatal test
23	SP_FIRSTPRE	Specify first prenatal test in text if coded 7 ("other test positive")
24	KARYO	Karyotype of infant/fetus
25	SP_KARYO	Specify karyotype
26*	GENTEST	Genetic Test
27*	SP_GENTEST	Specify genetic test
28	PM	Post mortem examination
29**	SURGERY	First surgery for malformation performed or planned
30	SYNDROME	Syndrome
31	SP_SYNDROME	Specify Syndrome
32	MALFO1	Malformation
33	SP_MALFO1	Specify malformation
34	MALFO2	As MALFO1
35	SP_MALFO2	Specify malformation
36	MALFO3	As MALFO1
37	SP_MALFO3	Specify malformation
38	MALFO4	As MALFO1
39	SP_MALFO4	Specify malformation
40	MALFO5	As MALFO1

41	SP_MALFO5	Specify malformation
42	MALFO6	As MALFO1
43	SP_MALFO6	Specify malformation
44	MALFO7	As MALFO1
45	SP_MALFO7	Specify malformation
46	MALFO8	As MALFO1
47	SP_MALFO8	Specify malformation
48*	PRESYN	Prenatal diagnosis for syndrome
49*	PREMAL1	Prenatal diagnosis for malformation
50*	PREMAL2	As PREMAL1
51*	PREMAL3	As PREMAL1
52*	PREMAL4	As PREMAL1
53*	PREMAL5	As PREMAL1
54*	PREMAL6	As PREMAL1
55*	PREMAL7	As PREMAL1
56*	PREMAL8	As PREMAL1
57#	OMIM	OMIM code / Type of Mendelian Inheritance
Exposure – Variables 58 to 78		
58**	ASSCONCEPT	Assisted conception
59##	OCCUPMO	Mother's occupation at time of conception
60	ILLBEF1	Illness before pregnancy 1
61	ILLBEF2	Illness before pregnancy 2
62*	MATDIAB	Maternal Pregestational Diabetes
63*	HbA1c	Glycated haemoglobin value
64	ILLDUR1	Illness during pregnancy
65	ILLDUR2	Illness during pregnancy 2
66*	FOLIC_G14	Folic acid supplementation
67*	FIRSTTRI	First trimester medication
68	DRUGS1	Drugs
69	SP_DRUGS1	Specify drug exposures
70	DRUGS2	As for DRUGS1
71	SP_DRUGS2	Specify drug exposures
72	DRUGS3	As for DRUGS1
73	SP_DRUGS3	Specify drug exposures
74	DRUGS4	As for DRUGS1
75	SP_DRUGS4	Specify drug exposures
76	DRUGS5	As for DRUGS1
77	SP_DRUGS5	Specify drug exposures
78	EXTRA_DRUGS	Extra drugs
Family History – Variables 79 to 90		
79	CONSANG	Consanguinity
80	SP_CONSANG	Specify text information on consanguinity
81	SIBANOM	Siblings with anomalies
82	SP_SIBANOM	Specify type of anomaly and describe the malformation
83	PREVSIB	Previous malformed sibs notified to EUROCAT
84	SIB1	Local ID number notified to the Central Registry
85	SIB2	As SIB1
86	SIB3	As SIB1
87	MOANOM	Mother's family with anomalies
88	SP_MOANOM	Specify type of anomaly and describe the malformation
89	FAANOM	Father's family with anomalies
90	SP_FAANOM	Specify type of anomaly and describe the malformation

Sociodemographic – Variables 91 to 94		
91	MATEDU	Maternal education
92	SOCM	Socioeconomic status of mother
93	SOCF	Socioeconomic status of father
94	MIGRANT	Migrant status
General Comments – Variable 95		
95	GENREM	General additional comments

* New variable In Guide 1.4
** Variable compatible with Guide 1.3, but coding has been extended/modified
# Variable name change only
Guide 1.4 use ISCO-08 classifications

2.2.1b Coding Instructions

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 Each case has a unique identification. This number is a maximum of 11 characters long, consisting of numbers, letters or both. ID numbers should not repeat themselves in different years.	Up to 11 digits
3	BIRTH_DATE	DATE OF BIRTH 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 99 = Not known for day and month DO NOT TRANSMIT RECORDS IF YEAR OF BIRTH IS NOT KNOWN
4	SEX	SEX Indicate chromosomal sex, if known, in case of ambiguous genitalia code malformations in variables 32-47. Indicate indeterminate sex in case of ambiguous genitalia with unknown or abnormal sex chromosome complement. If sex could not be determined at autopsy due to maceration or other problems, indicate as “not known”.	1 = Male 2 = Female 3 = Indeterminate 9 = Not known
5	NBRBABY	NUMBER OF BABIES/FETUSES DELIVERED Fill out a separate form for each malformed baby/fetus in a multiple set. 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”). Conjoined twins have a specific ICD/BPA code, to be coded under “syndrome” (variable 30). Give full description of type of conjoined twinning in syndrome text field (variable 31). Any other anomalies are coded in variables 32-47. Notes. If code 8 is used, please specify in variable sp_twin the gestational age at which last known to be a multiple pregnancy and/or first known to be a singleton. The purpose of this coding system is to allow us to distinguish malformed cases which would have civil registration as singleton births from malformed cases which would have civil registration as multiple births. Please specify the sex and outcome (live, still) of the malformed/non-malformed co-twin and zygosity.	1 = Singleton 2 = Twins 3 = Triplets 4 = Quadruplets 5 = Quintuplets 6 = Sextuplets or more 7 = Multiple birth, number of babies not known 8 = Singleton at time of delivery/termination, but known to have been a multiple pregnancy at an earlier stage in pregnancy 9 = Not known

Baby and Mother (core variables shaded blue)

Variable Number	Variable Name	Explanation and Instructions	Code
6	SP_TWIN	SPECIFY TWIN TYPE OF BIRTH (malformed and non-malformed), like or unlike sex, zygosity	Free text
7	NBRMALF	NUMBER OF MALFORMED IN MULTIPLE SET To be completed for multiple delivery only. Remember to give local ID of co-twin in SIB1 field (variable 84) if more than one malformed.	1 = One 2 = Two 3 = Three 4 = Four 5 = Five 6 = Six or more 9 = Not known
8	TYPE	TYPE OF BIRTH Birth with type of birth not known should be transmitted to EUROCAT, but will be excluded from routine EUROCAT analysis. 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. 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. 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. Stillbirths or perinatal deaths resulting from termination of pregnancy following prenatal diagnosis must be coded as terminations (value = 4), irrespective of civil registration status. 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 2 = Stillbirth 3 = Spontaneous abortion 4 = TOPFA 9 = Not known

Baby and Mother (core variables shaded blue)

Variable Number	Variable Name	Explanation and Instructions	Code
9	CIVREG	CIVIL REGISTRATION STATUS Livebirths and stillbirths are civilly registered leading to either a birth or stillbirth certificate and appear in official birth statistics for your region. Code here whether this case fulfilled the conditions for live or stillbirth registration in your country.	1 = Livebirth 2 = Stillbirth 3 = No civil registration 9 = Not known
10	WEIGHT	BIRTH WEIGHT Give weight in grams.	9999 = Not known (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 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 Yes = Child known to be alive after one week. No = Child known to have died before or during first week (including stillbirths and abortions). 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. If survival at one week is unknown, but survival at discharge from maternity unit less than one week is known, use the latter. The definition of first week of life varies between countries. Follow your country's perinatal mortality definition and specify this in your Registry Description. Not known = Not known if child has died during first week.	1 = Yes 2 = No 3 = Alive at discharge <1 week 9 = Not known

Baby and Mother (core variables shaded blue)

Variable Number	Variable Name	Explanation and Instructions	Code
13	DEATH_DATE	DATE OF DEATH **For livebirths only.** 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 99= Died, not known day or month 44 =Died, not known year (Do not use 99 for "not known" year of death, as this will be read as died in 1999, day and month not known.) 222222= Known to be alive at 1 year 333333= Not known if alive or dead at 1 year
14	DATEMO	DATE OF BIRTH OF MOTHER Give as much information as is known eg. Feb 1963 = 990263, 1963 = 999963. 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). This variable can be used to calculate maternal age at Expected Date of Delivery for preterm deliveries and terminations.	Day, month, year 99 = Not known day or month 44 = Not known year
15	AGEMO	AGE OF THE MOTHER AT DELIVERY 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	BMI	MATERNAL BODY MASS INDEX Enter BMI (2 digits). The EDMP will also allow entry of maternal height (in centimetres) and weight (in kilograms) and calculate BMI automatically. Values measured at first antenatal visit are preferred, but pre-pregnancy self-reported values may be given. If mother known to be obese, enter code for obesity E660 in maternal illness before pregnancy (variable 60) Whilst BMI is a new variable in Guide 1.4 (for cases born from 2013 onwards) if any registry has this information for previous cases, EUROCAT is interested in collecting this information from 2005 onwards	2 digits Expected range 15 – 50 97 = exact BMI NK but <30 98 = exact BMI NK but >=30 99 = Not known
17	RESIDMO	MOTHER'S RESIDENCE CODE Use local code for locality of residence at time of delivery.	Local code (up to 10 digits)
18	TOTPREG	TOTAL NUMBER OF PREVIOUS PREGNANCIES NOTE – The current reported pregnancy is NOT included. Include all previous abortions whether spontaneous or induced. Multiple pregnancies count as 1 in the total	00 = None 01 = One 02 = Two 03 = Three etc 20 = Twenty or more 99 = Not known

Diagnosis (core variables shaded blue)

Variable Number	Variable Name	Explanation and Instructions	Code
19	WHENDISC	WHEN DISCOVERED When the baby was first suspected of having a congenital anomaly. For prenatal diagnosis: when a major congenital anomaly was first suspected (EXCLUDING soft markers except if nuchal translucency indicates a very high risk followed by confirmation of diagnosis at delivery/termination). If prenatal diagnosis is made when fetus is dead code 1 (for stillbirths) or 7 (for spontaneous abortions). For livebirths: when first suspicion of an anomaly was at death OR at postmortem, when discovered is age at death (eg. At birth, < 1 week, 1-4 weeks etc). For stillbirths: when first suspicion of an anomaly was at birth OR at postmortem, when discovered is at birth (eg. Code = 1). All cases MUST have been confirmed as having a congenital anomaly Please also complete variables 12 "SURVIVAL", 13 "DEATH-DATE", 20 "CONDISC" and 28 "PM".	1 = At birth 2 = Less than 1 week 3 = 1-4 weeks 4 = 1-12 months 5 = Over 12 months 6 = Prenatal diagnosis in live fetus 7 = At abortion (spontaneous) 9 = Not known 10 = Postnatal diagnosis, age not known
20	CONDISC	CONDITION AT DISCOVERY Condition of fetus or baby when malformation was first suspected.	1 = Alive 2 = Dead 9 = Not known
21	AGEDISC	IF PRENATALLY DIAGNOSED, GESTATIONAL AGE AT DISCOVERY IN COMPLETED WEEKS GA as defined in variable gestlength. 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. If no prenatal diagnosis please leave blank.	99 = Not known

Diagnosis (core variables shaded blue)

Variable Number	Variable Name	Explanation and Instructions	Code
22	FIRSTPRE	FIRST POSITIVE PRENATAL TEST This refers to the first prenatal test whether screening procedure or diagnostic test which indicated a possible congenital anomaly or need for further tests. For code 7 = other specified test, give information in text field (variable 23). If test performed and result negative, then the "When discovered" variable cannot be coded 6 (prenatal diagnosis). This field is to record what DID happen, not any possible plans or intentions. Ultrasound < 14 weeks means only ultrasound performed which may include a nuchal measurement. The serum/combined screening must involve a biochemical test	1 = Ultrasound at GA < 14 weeks 2 = Ultrasound at GA 14-21 weeks 3 = Ultrasound at GA ≥ 22 weeks 4 = Ultrasound GA not known 5 = Serum/ combined screening 6 = CVS or amniocentesis 7 = Other test positive 8 = Test(s) performed, result negative 9 = Not known 10 = No test performed 11 =Fetal karyotype on maternal blood
23	SP_FIRSTPRE	SPECIFY "OTHER" FIRST PRENATAL TEST If FIRSTPRE = 7, specify which positive prenatal test	Free text
24	KARYO	KARYOTYPE OF INFANT/FETUS Specify result in variable 25. Array results count as a karyotype test If performed and results known, please specify (according to Paris nomenclature). "Probe test performed" refers to FISH, PCR, or other analyses restricted to specific chromosomal anomalies. "Failed" refers to a technical failure where a repeat examination could not be done and the karyotype is therefore unknown.	1 = Performed, result known 2 = Performed, results not known 3 = Not performed 4 = Probe test performed 8 = Failed 9 = Not known
25	SP_KARYO	SPECIFY KARYOTYPE	Free text
26	GENTEST	GENETIC TEST For syndromes and single gene disorders, a genetic test may have confirmed the clinical diagnosis either prenatally or postnatally. Please complete for these cases. Karyotype should still be completed as per variables 24 & 25 Whilst GENETIC TEST is a new variable in Guide 1.4 (for cases born from 2013 onwards) if any registry has this information for previous cases, EUROCAT is interested in collecting this information from 2005 onwards	1 = Yes, diagnosis confirmed by genetic test 2 = No, diagnosis not confirmed by genetic test 3 = Not Performed 9 = Not Known
27	SP_GENTEST	SPECIFY TYPE OF GENETIC TEST	Free text

Diagnosis (core variables shaded blue)

Variable Number	Variable Name	Explanation and Instructions	Code
28	PM	POST MORTEM EXAMINATION If performed record the malformation(s) discovered in the "malformation" section in the form. If other findings record in the "comments" space (variable 95). "Results known" means that the autopsy record has been reviewed by the registry. "Results not known" means that the autopsy record was not available to the registry. "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 2 = Performed, results not known 3 = Not performed 4 = Macerated fetus 9 = Not known
29	SURGERY	FIRST SURGICAL PROCEDURE FOR MALFORMATION (PERFORMED OR EXPECTED) **Complete for all livebirths** **(and fetal deaths, only if there was prenatal surgery)** 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. "No surgery required" means that this case does not have a severe enough malformation, or that the malformation is not correctable by surgery. "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 2 = Performed (or expected) after the first year of life 3 = Prenatal surgery 4 = No surgery required 5= Too severe for surgery 6 = Died before surgery 9 = Not known
30	SYNDROME	SYNDROME OR ASSOCIATION Refer to EUROCAT Guide on syndromes. Give name of syndrome or association in text variable 31. All the anomalies observed by the local clinician should be coded in the remaining boxes for malformations.If not a recognised syndrome or association, leave blank. When 2 syndromes are present in the same subject, code the more important one in the syndrome variables 30 and 31, and include the other one in variables 32 and 33 MALF01. Ensure karyotype information is given in variables 24 and 25, and that autopsy and medical genetics reports have been reviewed, where appropriate. In case of conjoined twins, give full description in syndrome text variable 31. 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 First 4 digits are ICD10 5th digit = BPA supplement or leave blank

Diagnosis (core variables shaded blue)

Variable Number	Variable Name	Explanation and Instructions	Code
31	SP-SYNDROME	SPECIFY SYNDROME Please specify availability of photographs and x-ray images of syndrome case.	
32	MALFO1	MALFORMATION A baby/fetus with ONLY minor anomalies (see exclusion list, chapter 7) should not be transmitted to Central Registry. When a major anomaly is present, code both major and minor anomalies. Up to 8 malformations can be coded – if more than 8 are present, specify additional anomalies in the text variable for the 8th anomaly (text variable 47 SP_MALFO8). Include in the 8 specified codes the most important ones, or those tabulated in EUROCAT Reports. Give written description of the malformations available in malformation text variables 33, 35, 37, 39, 41, 43, 45 and 47.	ICD 10 First 4 digits are ICD 5th digit = BPA classification OR leave blank
33	SP_MALFO1	SPECIFY MALFORMATION	Free text
34	MALFO2	AS MALFO1	As MALFO1
35	SP_MALFO2	SPECIFY MALFORMATION	Free text
36	MALFO3	AS MALFO1	As MALFO1
37	SP_MALFO3	SPECIFY MALFORMATION	Free text
38	MALFO4	AS MALFO1	As MALFO1
39	SP_MALFO4	SPECIFY MALFORMATION	Free text
40	MALFO5	AS MALFO1	As MALFO1
41	SP_MALFO5	SPECIFY MALFORMATION	Free text
42	MALFO6	AS MALFO1	As MALFO1
43	SP_MALFO6	SPECIFY MALFORMATION	Free text
44	MALFO7	AS MALFO1	As MALFO1
45	SP_MALFO7	SPECIFY MALFORMATION	Free text
46	MALFO8	AS MALFO1	As MALFO1
47	SP_MALFO8	SPECIFY MALFORMATION	Free text

Diagnosis (core variables shaded blue)

<table>
<tr><th>Variable Number</th><th>Variable Name</th><th>Explanation and Instructions</th><th>Code</th></tr>
<tr><td>48</td><td>PRESYN</td><td>PRENATAL DIAGNOSIS FOR SYNDROME
When each anomaly was first diagnosed.

The basis for this variable is to record whether the prenatal findings strongly suggest the postnatal diagnosis. This variable is not designed for fetal medicine specialists to assess the accuracy of their prenatal diagnosis. Thus the finding of a significant heart anomaly prenatally is considered to be prenatally detected, even if the exact anomaly was not correctly diagnosed. ‘Yes, prenatally diagnosed’, should be used when the prenatal finding is nearly 100% predictive of the congenital anomaly. ‘Partially’ means that the prenatal finding is consistent with the postnatal anomaly but has a lesser predictive value, being suggestive of more than one type of anomaly, an example here would be increased nuchal translucency. The examples below are to illustrate this principle and ensure consistency of coding. Queries about individual cases can be send to Central registry

<table>
<tr><th>Prenatal Finding</th><th>Postnatal finding</th><th>Prenatal/Postnatal/ Partial</th></tr>
<tr><td>Double bubble</td><td>Duodenal atresia/stenosis</td><td>Prenatal</td></tr>
<tr><td>High risk screening (no amnio)</td><td>T21</td><td>Partial</td></tr>
<tr><td>Ventriculomegaly</td><td>Agenesis corpus callosum</td><td>Partial</td></tr>
<tr><td>Ventriculomegaly</td><td>Neuronal migration anomalies</td><td>Partial</td></tr>
<tr><td>Ventriculomegaly</td><td>Hydrocephalus</td><td>Prenatal</td></tr>
<tr><td>Significant heart anomaly</td><td>Any significant heart anomaly</td><td>Prenatal</td></tr>
<tr><td>Heart abnormality</td><td>22q11 del</td><td>Partial</td></tr>
<tr><td>Cleft lip</td><td>Cleft lip and palate</td><td>Partial</td></tr>
<tr><td>IUGR</td><td>Skeletal displasia</td><td>Postnatal</td></tr>
<tr><td>Anhydramnios</td><td>Renal agenesis</td><td>Partial</td></tr>
<tr><td>Micrognathia</td><td>Pierre Robin/cleft palate</td><td>Prenatal</td></tr>
<tr><td>Severe skeletal dysplasia</td><td>Specific skeletal dysplasia eg thanatophoric/achondrogenesis</td><td>Prenatal</td></tr>
<tr><td>Echogenic bowel</td><td>CF</td><td>Partial</td></tr>
<tr><td>Absent stomach bubble</td><td>Oesophageal atresia</td><td>Partial</td></tr>
</table>
</td><td>1 = Yes, this anomaly was diagnosed prenatally
2 = No, this anomaly was diagnosed postnatally
3 = This anomaly partially prenatally diagnosed
9 =Not known</td></tr>
</table>

Diagnosis (core variables shaded blue)

49	PREMAL1	PRENATAL DIAGNOSIS FOR MALFORMATION AS PRESYN	AS PRESYN
50	PREMAL2	PRENATAL DIAGNOSIS FOR MALFORMATION AS PRESYN	AS PRESYN
51	PREMAL3	PRENATAL DIAGNOSIS FOR MALFORMATION AS PRESYN	AS PRESYN
52	PREMAL4	PRENATAL DIAGNOSIS FOR MALFORMATION AS PRESYN	AS PRESYN
53	PREMAL5	PRENATAL DIAGNOSIS FOR MALFORMATION AS PRESYN	AS PRESYN
54	PREMAL6	PRENATAL DIAGNOSIS FOR MALFORMATION AS PRESYN	AS PRESYN
55	PREMAL7	PRENATAL DIAGNOSIS FOR MALFORMATION AS PRESYN	AS PRESYN
56	PREMAL8	PRENATAL DIAGNOSIS FOR MALFORMATION AS PRESYN	AS PRESYN
57	OMIM	OMIM / TYPE OF MENDELIAN INHERITANCE To be coded by medical geneticist or after advice from medical geneticist. This code is to be used for cases with single gene origin only – Refer to EUROCAT Syndrome Guide. The first digit may be filled in without the rest of the code if the full OMIM code is not known. Full codes can be found on the OMIM website http://www.ncbi.nlm.nih.gov/omim/	

Exposure (core variables shaded blue)

Variable Number	Variable Name	Explanation and Instructions	Code
58	ASSCONCEPT	ASSISTED CONCEPTION IVF = In vitro fertilization GIFT = Gamete intra fallopian transfer ICSI = Intracytoplasmic sperm injection	0 = No 1 = Induced ovulation only 2 = Artificial insemination 3 = IVF 4 = GIFT 5 = ICSI 6 = Egg donation 8 = Other 9 = Not known 10 = Assisted conception, type unknown
59	OCCUPMO	MOTHER'S OCCUPATION AT TIME OF CONCEPTION Code main occupation at time of conception (or earliest known time in first trimester). Note that the main purpose of the variable relates to potential teratogenic occupational exposures in early pregnancy. Be as precise as possible. Code according to 2008 (ISCO-08) Classification for births with birth dates from 2013. Code according to the 1988 International Standard Classification of Occupations (ISCO-88) for births with birth dates up to 2012. Links for ISCO classifications: http://www.ilo.org/public/english/bureau/stat/isco/isco08/index.htm Available in many languages. The 4 digit codes give the necessary specificity. They are grouped into the following main groups: 0 = Armed Forces (NB – do not preface your codes with zero UNLESS it is an armed forces occupation. All database systems must accept a leading zero and not drop it). 1 = Managers 2 = Professionals 3 = Technicians and Associate Professionals 4 = Clerical Support Workers 5 = Service and Sales Workers 6 = Skilled agricultural, forestry and fishery workers 7 = Craft and related trades workers 8 = Plant and machine operators, and assemblers 9 = Elementary occupations EUROCAT Supplement: 9991 = Employed (including self-employed), but occupation unknown 9995 = Housewife 9996 = Student 9997 = Unemployed 9999 = Not known whether employed or not	**4 digit** code 9999 = Not known (do NOT use 9, 99 or 999 for not known)

Exposure (core variables shaded blue)

Variable Number	Variable Name	Explanation and Instructions	Code
60	ILLBEF1	ILLNESS BEFORE PREGNANCY 1 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. Any additional details may be entered in the general comments section (variable 95). Do not insert the decimal point in the code (e.g. Code E05.0 as E050) Abridged list: Hyperthyroidism E050 - E059 Hypothyroidism E000 - E039 Diabetes Type 1 E100 - E109 Diabetes Type 2 E110 - E119 Obesity E660 - E669 If maternal BMI ≥ 30 give code for obesity Anorexia /eating disorders F500-F509 Epilepsy G400 - G409 Asthma J450 - J459 Chronic alcoholism F102 Drug addict F112 - F122 - F132 - F142 F152 - F192	ICD 10 0 = No illness 1 = Yes, but no information available 9 = Not known
61	ILLBEF2	ILLNESS BEFORE PREGNANCY 2 AS FOR ILLBEF1	
62	MATDIAB	MATERNAL **PRE**GESTATIONAL DIABETES This variable is specifically for **pre**gestational diabetes. Gestational diabetes is dealt with under the 'illness during pregnancy' variable (variable 64) Type 1 diabetes: characterized by hyperglycemia due to an absolute deficiency of the insulin hormone produced by the pancreas An HbA1c of 48mmol/mol is recommended as the cut-off point for diagnosing diabetes. Type 2 diabetes: characterized by hyperglycemia due to a defect in insulin secretion An HbA1c of 48mmol/mol is recommended as the cut-off point for diagnosing diabetes. *Maturity Onset Diabetes in the Young (MODY) displays an autosomal dominant pattern of inheritance An HbA1c of 48mmol/mol is recommended as the cut-off point for diagnosing diabetes. Impaired Glucose Intolerance is a state of higher than normal blood (or plasma) glucose concentration, but less than the diagnostic cut-off for diabetes. Diagnosed before pregnancy. Diagnosed by fasting plasma glucose from 6.1 – 6.9 mmol/L (WHO criteria) http://www.who.int/diabetes/publications/en/	1= Yes, type 1 diabetes (IDDM) 2= Yes, type 2 diabetes (NIDDM) 3 = Yes, type MODY* (all types) 4 = Yes, type not known 5 = No, but impaired glucose intolerance 6 = No pregestational diabetes 9 = Not known

Exposure (core variables shaded blue)

<table>
<tr><th>Variable Number</th><th>Variable Name</th><th>Explanation and Instructions</th><th>Code</th></tr>
<tr><td>63</td><td>HbA1c</td><td>GLYCATED HAEMOGLOBIN (HbA1c) VALUE
Give the first HbA1c value measured in the first trimester (in mmol/mol units)

Normal values for non-diabetic individuals <48mmol/mol

<table>
<tr><td>%</td><td>4.0</td><td>4.1</td><td>4.2</td><td>4.3</td><td>4.4</td><td>4.5</td><td>4.6</td><td>4.7</td><td>4.8</td><td>4.9</td></tr>
<tr><td>mmol/mol</td><td>20</td><td>21</td><td>22</td><td>23</td><td>25</td><td>26</td><td>27</td><td>28</td><td>29</td><td>30</td></tr>
<tr><td>%</td><td>5.0</td><td>5.1</td><td>5.2</td><td>5.3</td><td>5.4</td><td>5.5</td><td>5.6</td><td>5.7</td><td>5.8</td><td>5.9</td></tr>
<tr><td>mmol/mol</td><td>31</td><td>32</td><td>33</td><td>34</td><td>36</td><td>37</td><td>38</td><td>39</td><td>40</td><td>41</td></tr>
<tr><td>%</td><td>6.0</td><td>6.1</td><td>6.2</td><td>6.3</td><td>6.4</td><td>6.5</td><td>6.6</td><td>6.7</td><td>6.8</td><td>6.9</td></tr>
<tr><td>mmol/mol</td><td>42</td><td>43</td><td>44</td><td>45</td><td>46</td><td>48</td><td>49</td><td>50</td><td>51</td><td>52</td></tr>
<tr><td>%</td><td>7.0</td><td>7.1</td><td>7.2</td><td>7.3</td><td>7.4</td><td>7.5</td><td>7.6</td><td>7.7</td><td>7.8</td><td>7.9</td></tr>
<tr><td>mmol/mol</td><td>53</td><td>54</td><td>55</td><td>56</td><td>57</td><td>58</td><td>60</td><td>61</td><td>62</td><td>63</td></tr>
<tr><td>%</td><td>8.0</td><td>8.1</td><td>8.2</td><td>8.3</td><td>8.4</td><td>8.5</td><td>8.6</td><td>8.7</td><td>8.8</td><td>8.9</td></tr>
<tr><td>mmol/mol</td><td>64</td><td>65</td><td>66</td><td>67</td><td>68</td><td>69</td><td>70</td><td>72</td><td>73</td><td>74</td></tr>
<tr><td>%</td><td>9.0</td><td>9.1</td><td>9.2</td><td>9.3</td><td>9.4</td><td>9.5</td><td>9.6</td><td>9.7</td><td>9.8</td><td>9.9</td></tr>
<tr><td>mmol/mol</td><td>75</td><td>76</td><td>77</td><td>78</td><td>79</td><td>80</td><td>81</td><td>83</td><td>84</td><td>85</td></tr>
<tr><td>%</td><td>10.0</td><td>10.1</td><td>10.2</td><td>10.3</td><td>10.4</td><td>10.5</td><td>10.6</td><td>10.7</td><td>10.8</td><td>10.9</td></tr>
<tr><td>mmol/mol</td><td>86</td><td>87</td><td>88</td><td>89</td><td>90</td><td>91</td><td>92</td><td>93</td><td>95</td><td>96</td></tr>
<tr><td>%</td><td>11.0</td><td>11.1</td><td>11.2</td><td>11.3</td><td>11.4</td><td>11.5</td><td>11.6</td><td>11.7</td><td>11.8</td><td>11.9</td></tr>
<tr><td>mmol/mol</td><td>97</td><td>98</td><td>99</td><td>100</td><td>101</td><td>102</td><td>103</td><td>104</td><td>105</td><td>107</td></tr>
<tr><td>%</td><td>12.0</td><td>12.1</td><td>12.2</td><td>12.3</td><td>12.4</td><td>12.5</td><td>12.6</td><td>12.7</td><td>12.8</td><td>12.9</td></tr>
<tr><td>mmol/mol</td><td>108</td><td>109</td><td>110</td><td>111</td><td>112</td><td>113</td><td>114</td><td>115</td><td>116</td><td>117</td></tr>
<tr><td>%</td><td>13.0</td><td>13.1</td><td>13.2</td><td>13.3</td><td>13.4</td><td>13.5</td><td>13.6</td><td>13.7</td><td>13.8</td><td>13.9</td></tr>
<tr><td>mmol/mol</td><td>119</td><td>120</td><td>121</td><td>122</td><td>123</td><td>124</td><td>125</td><td>126</td><td>127</td><td>128</td></tr>
</table>
</td><td>999 = Not known

3 digits</td></tr>
</table>

Exposure (core variables shaded blue)

Variable Number	Variable Name	Explanation and Instructions	Code
64	ILLDUR1	ILLNESS DURING PREGNANCY Record illnesses with chronic or acute onset during the first 20 weeks of pregnancy including asymptomatic maternal infections. For gestational diabetes include at any point in pregnancy (Any additional details may be entered in the general comments section, variable 95). 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 30-47). Do not insert the decimal point in the code (eg. Code B34.1 as B341) Coxsackie's B341 Cytomegalic Inclusion Diseases B250 - B259 Gestational Diabetes O244 – O249 Herpes Simplex B000 - B009 HIV (AIDS) B200 - B249 Influenza J100 - J119 Listeria A320 - A329 Mumps B260 - B269 Rubella B060 - B069 Syphillis A530 - A539 Toxoplasmosis B580 - B589 Varicella (Chicken Pox) B010 - B019 Viral Hepatitis B190 - B199 Drug poisoning T360-T509	ICD 10 0 = No 1 = Yes, but no information available 9 = Not known
65	ILLDUR2	ILLNESS DURING PREGNANCY AS FOR ILLDUR1	
66	FOLIC_G14	FOLIC ACID SUPPLEMENTATION Recommend to your local maternity hospitals or midwives to collect these data. Folic acid supplementations include folic acid only tablets, a multivitamin preparation which contains folic acid or contraceptive pills which contain folic acid. If the folic acid dose is high, please add the code B03BB01 in the drugs variable	1 = Folic acid taken pre and post-conceptionally 2 = Folic acid taken only post-conceptionally 3 = Folic acid not taken 4 = Folic acid taken, timing unknown 9 = Not known if folic acid taken

Exposure (core variables shaded blue)

Variable Number	Variable Name	Explanation and Instructions	Code
67	FIRSTTRI	FIRST TRIMESTER MEDICATION "Yes" means that the data sources clearly state that medication was taken in the first trimester. "No" means that the data sources clearly state that no medication was taken in the first trimester. "Undetermined" means that the usual data sources were consulted, but • it was not clearly stated that medication was either taken or not taken • the information regarding medication use was illegible • Type of medication is unknown. "Medication taken but timing unknown" means that the usual data sources stated that medication was taken but the timing of use was not stated for **some** or **all** of the medications. Use this option also for cases in which the data sources clearly state that certain medication was taken in the first trimester, but for other medication the timing was unknown. Use SP_DRUGS fields to explain for each recorded medication whether it was taken in the first trimester, or if timing was unknown. "Not Known" means that the usual data sources were not found. Only fill in DRUGS1-5 and EXTRADRUGS if you have coded FIRSTTRI = 1 (Yes medication taken) or = 4 (Medication taken, but timing unknown). If you have coded FIRSTTRI = 2 (no medication taken), FIRSTTRI = 3 (undetermined) or FIRSTTRI = 9 (unknown), there shouldn't be any ATC codes in any of the DRUGS variables • Include any medication that was taken by the mother during the first trimester of pregnancy (from the 1st day of the last menstrual period up to the 12th week of gestation). Medication with long elimination half time and taken before conception should be included (eg. Acitretin, Etretinate, etc.). • Use of folic acid (either as folic acid only tablets or a multivitamin preparation which contains folic acid) should be registered in the folic acid variable • Do not include usual vitamins and mineral supplementation, but include unusual intakes of vitamins or minerals (eg. Vitamin A mega doses). • Only medication taken at physiologic doses should be included. Whilst FIRSTTRI is a new variable in Guide 1.4 (for cases born from 2013 onwards) if any registry has this information for previous cases, EUROCAT is interested in collecting this information from 2005 onwards	1 = Yes, medication taken in first trimester 2 = No medication taken in first trimester 3 = Undetermined 4 = Medication taken, but timing unknown 9 = Not Known

Exposure (core variables shaded blue)

Variable Number	Variable Name	Explanation and Instructions	Code
68	DRUGS1	DRUGS - 7 DIGITS MAXIMUM 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). 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. Use ATC-coding and use as many digits as possible (from 3 to 7). Website http://www.whocc.no/atcddd/. 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. ATC example: N03A: antiepileptic drug N03AF01: carbamazepine Details on the dosage and timing should be given in text variable 69. Do not forget to mention in the appropriate section (disease during or before pregnancy) the indication for drug use. Only drugs taken at physiologic doses to be recorded. If a drug overdose or self-poisoning, this MUST be explained in the drug description.	
69	SP_DRUGS1	SPECIFY DRUG EXPOSURES	Free text
70	DRUGS2	AS FOR DRUGS1 Please give details in text variable 71 SP_DRUGS2.	As for DRUGS1
71	SP_DRUGS2	SPECIFY DRUG EXPOSURES	Free text
72	DRUGS3	AS FOR DRUGS1 Please give details in text variable 73 SP_DRUGS3.	As for DRUGS1
73	SP_DRUGS3	SPECIFY DRUG EXPOSURES	Free text
74	DRUGS4	AS FOR DRUGS1 Please give details in text variable 75 SP_DRUGS3.	
75	SP_DRUGS4	SPECIFY DRUG EXPOSURES	
76	DRUGS5	AS FOR DRUGS1 Please give details in text variable 77 SP_DRUGS3.	
77	SP_DRUGS5	SPECIFY DRUG EXPOSURES	

Exposure (core variables shaded blue)

Variable Number	Variable Name	Explanation and Instructions	Code
78	EXTRA_DRUGS	EXTRA DRUGS This field is only to be used if drug fields 1-5 have already been filled. 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). 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. Use ATC-coding and use as many digits as possible (from 3 to 7). Website http://www.whocc.no/atcddd/. 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. ATC example: N03A: antiepileptic drug N03AF01: carbamazepine 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. Only drugs taken at physiologic doses to be recorded. If a drug overdose or self-poisoning, this MUST be explained in the drug description. If importing data from a local program, enter the ATC code and text description in the following format: <ATC code|text description> 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: <ATC code|text description><ATC code|text description> For example a case with valproate and lamotrigine exposure is entered in the extra_drugs field as: <N03AG01|Valproate><N03AX09|Lamotrigine> (See chapter 2.4 of EDMP User Guide for further guidance)	

Family History (core variables shaded blue)

Variable Number	Variable Name	Explanation and Instructions	Code
79	CONSANG	CONSANGUINITY 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 1 = Relationship of second cousin or closer 9 = Not known
80	SP_CONSANG	SPECIFY TEXT INFORMATION ON CONSANGUINITY	Free text
81	SIBANOM	SIBS WITH ANOMALIES If the sibling (including twin) was notified to EUROCAT fill in variables 83-86 below. Make sure that the local identification numbers given correspond to those in the central database; otherwise give more information in text here. If previous siblings were not notified to EUROCAT specify in text SP_SIBANOM the year of birth and malformations of each sibling. 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" Always give details in text variable 82 SP_SIBANOM	1 = Same 2 = Other 3 = Same and other 4 = No 9 = Not known
82	SP_SIBANOM	SPECIFY TYPE OF ANOMALY OF SIBLINGS	Free text
83	PREVSIB	PREVIOUS MALFORMED SIBLINGS NOTIFIED TO EUROCAT If yes, give the local ID number in variables SIB1, SIB2 or SIB3 (variables 84-86). Include malformed co-twins or siblings from the same pregnancy, irrespective of birth order within multiple set. Exclude, conjoined twin.	1 = Yes 2 = No 9 = Not known
84	SIB1	SIB LOCAL ID NUMBER NOTIFIED TO THE CENTRAL REGISTRY Enter here also the code numbers of co-twins or siblings from the same pregnancy, irrespective of birth order within multiple sets. Leave blank if no previous siblings notified to EUROCAT.	Local ID
85	SIB2	AS SIB1	Local ID
86	SIB3	AS SIB1	Local ID

Family History (core variables shaded blue)

Variable Number	Variable Name	Explanation and Instructions	Code
87	MOANOM	MOTHER'S FAMILY WITH ANOMALIES Include mother herself as well as mother's family. Specify type of anomaly in written text and relation to the infant. If the aetiology is known, "same" means the same aetiology, even if the spectrum of malformations present is slightly different. 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. "Same and other" refers to two different relatives. If a relative has both the same and another anomaly, code "same". Restrict the family to first, second and third degree relatives (mother, father, siblings, grandparents, aunts, uncles, half-siblings, first cousins). Always give details in text variable 88 SP_MOANOM.	1 = Same 2 = Other 3 = Same and other 4 = No 9 = Not known
88	SP_MOANOM	SPECIFY TYPE OF ANOMALY AND DESCRIBE THE MALFORMATION	Free text
89	FAANOM	FATHER'S FAMILY WITH ANOMALIES As MOANOM Please give details in text variable 90 SP_FAANOM	As MOANOM
90	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
91	MATEDU	MATERNAL EDUCATION Refer to International Standard Classification of Education 1997 for more information and Kunst et al (2001). Assign according to the highest level of education completed (or for full-time students, level in progress). 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 2 = Upper secondary 3 = Tertiary 9 = Not known
92	SOCM	SOCIOECONOMIC STATUS OF MOTHER Current or last occupation. Upper non-manual – professionals, administrators and managers eg. doctor, architect, lawyer, banker, manager, teacher, nurse, performer. Lower non-manual – routine non-manual eg. Book-keeper, salesman, receptionist, secretary, computer operator, clerk, waiter. Skilled manual – cook, butcher, carpenter. Unskilled manual – semi and unskilled manual eg. factory worker, driver, agricultural worker, porter. Self employed/artisan – owner of shop, restaurant or hotel, independent artisan. Farmer – eg. self-employed farmer or fisherman. If code 8 ("other/student"), please specify in text in space for general comments (variable 95). For further information see Kunst et al (2001)*	1 = Upper non-manual 2 = Lower non-manual 3 = Skilled manual 4 = Unskilled manual 5 – Self employed/artisan 6 = Farmer 8 = Other/Student 9 = Not known
93	SOCF	SOCIOECONOMIC STATUS OF FATHER As SOCM.	0 = Single mother, no father recorded 1 = Upper non-manual 2 = Lower non-manual 3 = Skilled manual 4 = Unskilled manual 5 – Self employed/artisan 6 = Farmer 8 = Other/Student 9 = Not known

Sociodemographic (core variables shaded blue)

Variable Number	Variable Name	Explanation and Instructions	Code
94	MIGRANT	MIGRANT STATUS 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. If code 4, give text details in the general comments section (variable 95).	1 = Mother migrated from outside EU during pregnancy 2 = Mother migrated from outside EU during adult life (from age 18) 3 = Mother not a migrant as defined in 1 or 2 4 = Other (specify in text) 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
95	GENREM	GENERAL ADDITIONAL COMMENTS	Free text

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) 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-al108 below. 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 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)

Variable	Description
al1	EUROCAT subgroup: All Anomalies
al2	EUROCAT subgroup: Nervous system
al3	EUROCAT subgroup: Neural Tube Defects
al4	EUROCAT subgroup: Anencephalus and similar
al5	EUROCAT subgroup: Encephalocele
al6	EUROCAT subgroup: Spina Bifida
al7	EUROCAT subgroup: Hydrocephalus
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
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 liveborn term infants (GA 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

Variable	Description
al49	EUROCAT subgroup: Abdominal wall defects
al50	EUROCAT subgroup: Gastroschisis
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: Edwards syndrome/trisomy 18
al92	EUROCAT subgroup: Turner syndrome
al93	EUROCAT subgroup: Klinefelter syndrome

2.2.3 Recommended Local Variables

In addition to the variables described in Chapter 2.2.1, EUROCAT recommends that registries collect other variables for use locally (not for transmission to the Central Registry).
The list below includes variables that previously have been included in the standard set of variables for EUROCAT

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).
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).
Age of father at delivery	
Sources of information (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.
When first reported	
Source of information (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.
Aetiology	Aetiological classification of anomaly to be completed by a medical geneticist or following the advice below. **C = All unbalanced Chromosome errors.** Included in C are all trisomies (+ mosaics), monosomies, triploidy, deletions, duplications, insertions, and unbalanced translocations including the syndromes in the table below.

T = Teratogens and prenatal infections. Includes maternal illness and teratogenic medications
G = All single gene disorders and genetic conditions not included in C
U = Non-genetic, non-chromosomal syndromes of unknown aetiology. Includes eg. Isomerism/Ivemark, Goldenhar, fetal akinesia of unknown aetiology
I = Isolated anomalies to include more than one anomaly from the same body system and sequences. Included here are:
(a) single anomalies such as gastroschisis, talipes, or cleft lip
(b) more than one anomaly from the same system such as a VSD and coarctation, or polydactyly of hands and feet
(c) more than one anomaly as part of a sequence such as spina bifida with talipes or hydrocephalus; lung hypoplasia and renal agenesis
M = Multiple anomalies, Associations and unclassifiable All that do not fit into above categories.
Z = Normal at birth. For cases with uncertain or abnormal findings prenatally that have not been confirmed postnatally.

Examples for C

Chromosome/Genetic Error	Syndrome Names
22q11 deletion	Di George, Velocardiofacial or Schprintzen syndrome
15q11 deletion, maternal or paternal disomy, imprinting mutations	Prader-Willi or Angelman syndrome
7q11 deletion, elastin mutation	Williams syndrome
11p15 duplication, paternal isodisomy, imprinting mutations	Beckwith-Wiedemann syndrome
20p12 deletion or JAG1 mutation	Alagille syndrome
16p13 deletion	Rubenstein-Taybi syndrome
5q35 deletion, NSD1 mutation	Sotos syndrome
11p13 deletion, PAX6 / WT1 mutn	Aniridia Wilms' tumour (WAGR)
17p13 deletion, LIS1 mutation	Miller-Dieker syndrome
1q21.1 deletion	TAR (thrombocytopaenia absent radius) syndrome

Whether diagnosed by a karyotype, an array or any other technique

Examples for T

Drugs and Maternal Illness	Infections
Abortifacients	Cytomegalovirus
ACE inhibitors	Herpes Simplex
Alcohol	Parvovirus
	Rubella
Cocaine and other illicit drugs	Toxoplasmosis
Cytotoxics	Varicella
Diabetes - uncontrolled	
Folic acid inhibitors	
Lithium	
Thalidomide	
Thyroid disease requiring drug treatment	

Vitamin A analogues	
Warfarin	

Examples for G

Common Single Gene Disorders	(Continued..)
Achondroplasia	Kabuki
Aperts	Meckel Gruber
ARPKD	
Campomelia	Noonan's syndrome
CHARGE syndrome	Osteogenesis imperfect
Cornelia de Lange syndrome	
Fragile X	Thanatophoric dysplasia
Jeune syndrome	Tuberous sclerosis

Examples for U

Non-genetic, non-chromosomal syndromes of unknown aetiology
Fetal akinesia/arthrogryposis not secondary to oligohydramnios (unless known to be genetic)
Goldenhar
Isomerism/Ivemark (unless known to be genetic)

Examples for I

Isolated anomalies to include more than one anomaly from the same body system and sequences Include here:	
(a) single anomalies such as gastroschisis, talipes, or cleft lip	
(b) more than one anomaly from the same system such as a VSD and coarctation, or polydactyly of hands and feet	
(c) more than one anomaly as part of a sequence such as spina bifida with talipes or hydrocephalus; lung hypoplasia and renal agenesis	
Acardiac twin	Limb body wall
Amniotic bands	Poland's
Hirschsprungs (unless known to be genetic)	Septo-optic dysplasia
Isolated hydrops, cystic hygroma or nuchal >3.5mm	Sirenomelia

Examples for M

Multiple, Associations and unclassifiable (use sparingly for this). All that do not fit into above categories should be multiple, unrelated anomalies from more than one system with no unifying diagnosis and recognised associations that are not regarded as a syndrome	
	Pentalogy of Cantrell
MURCS	VATER
OEIS	

2.2.4 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				

Gastroschisis by Maternal Age:	**Includes Chromosomal Cases**				**Excludes Chromosomal Cases**			
	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								
Hydrocephalus								
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 (LB and GA 37+ weeks)								

	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
Respiratory								
Choanal atresia								
Cystic adenomatous malf of lung								
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								

	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
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								
Maternal infections resulting in malformations								
Genetic syndromes + microdeletions								
Sequences								
Chromosomal								
Down syndrome								
Patau syndrome/trisomy 13								
Edwards syndrome/trisomy 18								
Turner syndrome								
Klinefelter syndrome								

LB= Live births
FD= Fetal deaths / Still births 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.

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+					
Mother 35+					
Mother 40+					
Unknown					
TOTAL**					
*Please complete all / as many as possible of the age categories above ** 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					

2.4 EUROCAT Data Management Program (EDMP) Instructions

EUROCAT Data Management Program

EDMP

Version (6.06) 07/10/2013

For Guide 1.4

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 improvements in this new version of EDMP are the ability to output both prenatal diagnosis and perinatal mortality data into Excel format. The prenatal diagnosis output enables you to check figures by outcome, by gestation, by maternal age and by indication. The perinatal mortality output allows you to check fetal deaths and early neonatal deaths by subgroup and also provides an overall perinatal mortality + TOPFA rate. There are new variables: maternal body mass index (BMI), genetic test, prenatal diagnosis per individual anomaly, maternal pregestational diabetes, HbA1c value, first trimester medication, and a new folic acid variable. Occupation of mother has been updated to ISCO-08 coding and McKusick has been re-named OMIM. These variables are compatible with Guide 1.4 which is for births from 01/01/2013 onwards.

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 EUROCAT instruction guides

EUROCAT Guide 1.2 format must still be used for cases born up to the end of 2004. EUROCAT Guide 1.3 format is used for births between 2005 and 2012 Guide 1.4 is for births from 2013 onwards EDMP will switch between Guide 1.2, 1.3 and 1.4 formats depending upon the year of birth.

If you want to continue to use, at local level, some variables which are no longer included in Guide 1.4 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, 2007 or 2010 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.

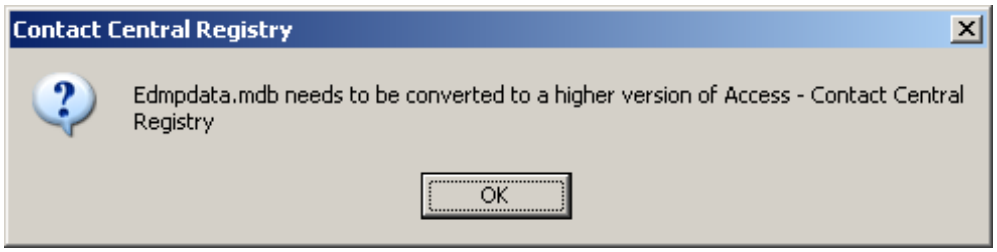
4.1.1 Existing Users

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.

4.1.3 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.

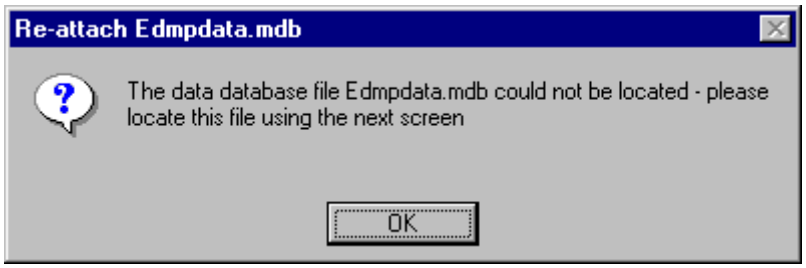
Access 2007 Security Settings

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.

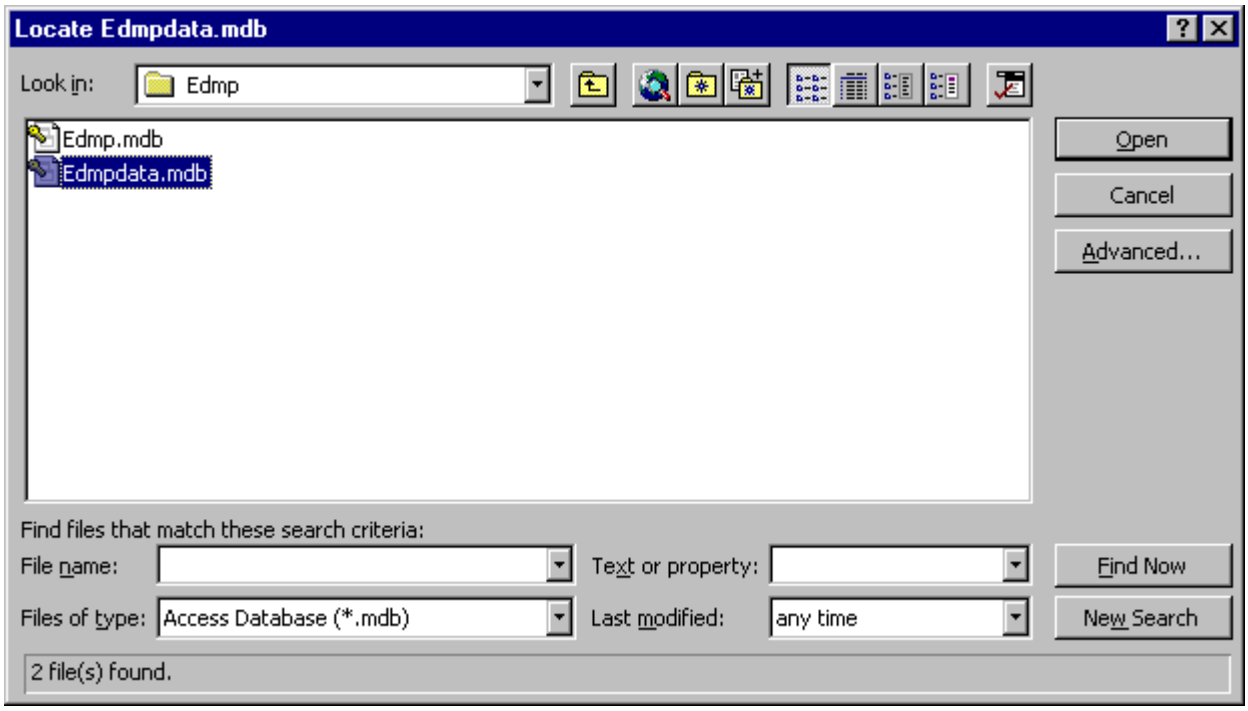
4.2 First time use

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.

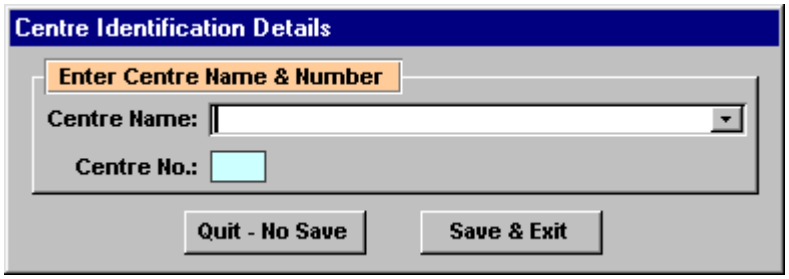
Each time you run the EDMP the first thing it does is to check to see that the program can locate the data file Edmpdata.mdb. If Edmpdata.mdb is not where it thinks it should be (i.e. after installation or if it has been moved to a different folder) then the following message will be displayed.



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.



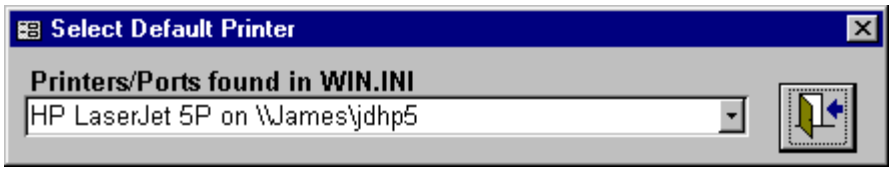
Once EDMP is happy that the data database has been located then you will need to select your centre from the list using the screen below.



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 op that can be clicked on.

The new logical sections are:

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

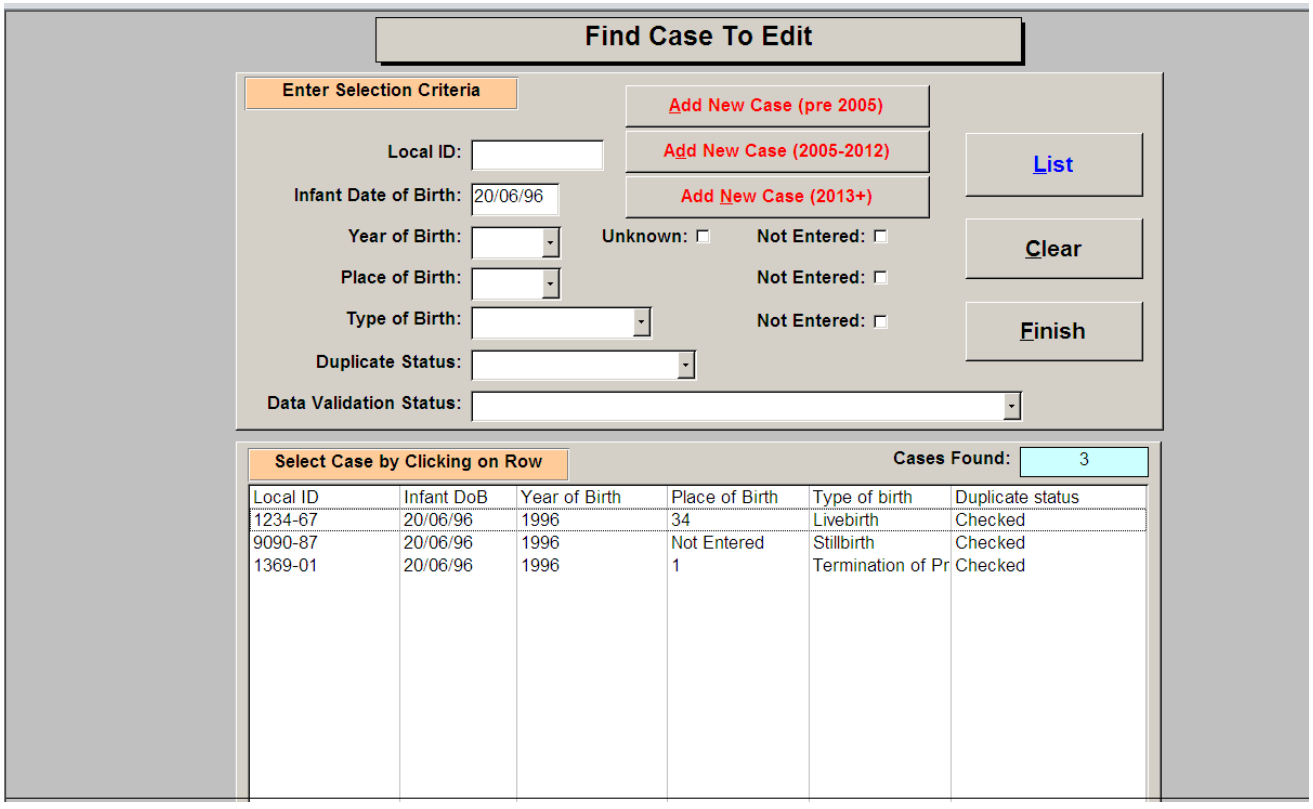
Access to each option will depend on individual permissions settings (see Manage Users under the System Menu.)

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, 2005-2012 and 2013+ year of birth. The 2013+ option includes the new guide 1.4 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 1.4 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.



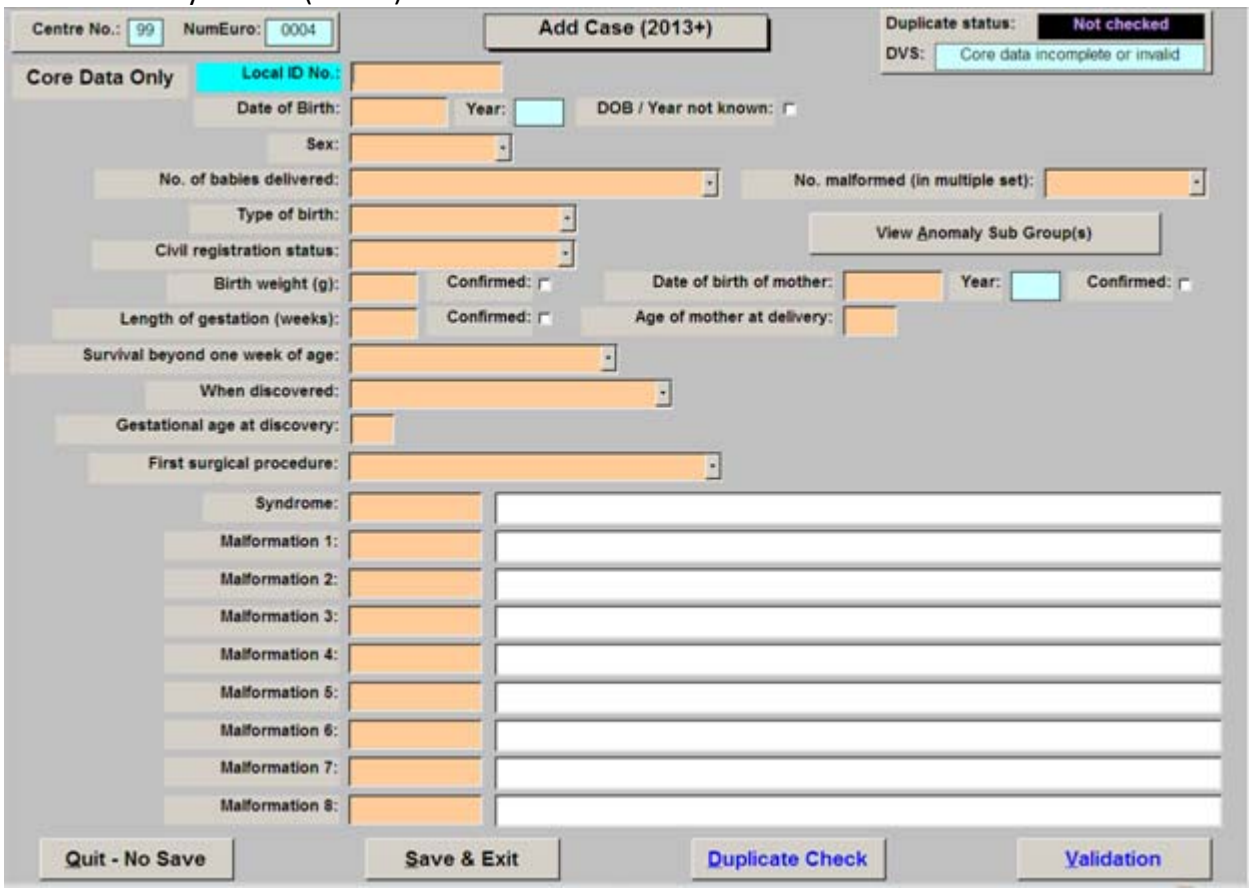
The EDMP display option chosen on the main menu (under System Menu) dictates which data entry form will be displayed. There are three different styles of data entry form, examples of each are shown below. The simplest is for the 'Core Data Only' option where there is a single page of data entry fields. 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. 'Baby & Mother', 'Malformations' 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.

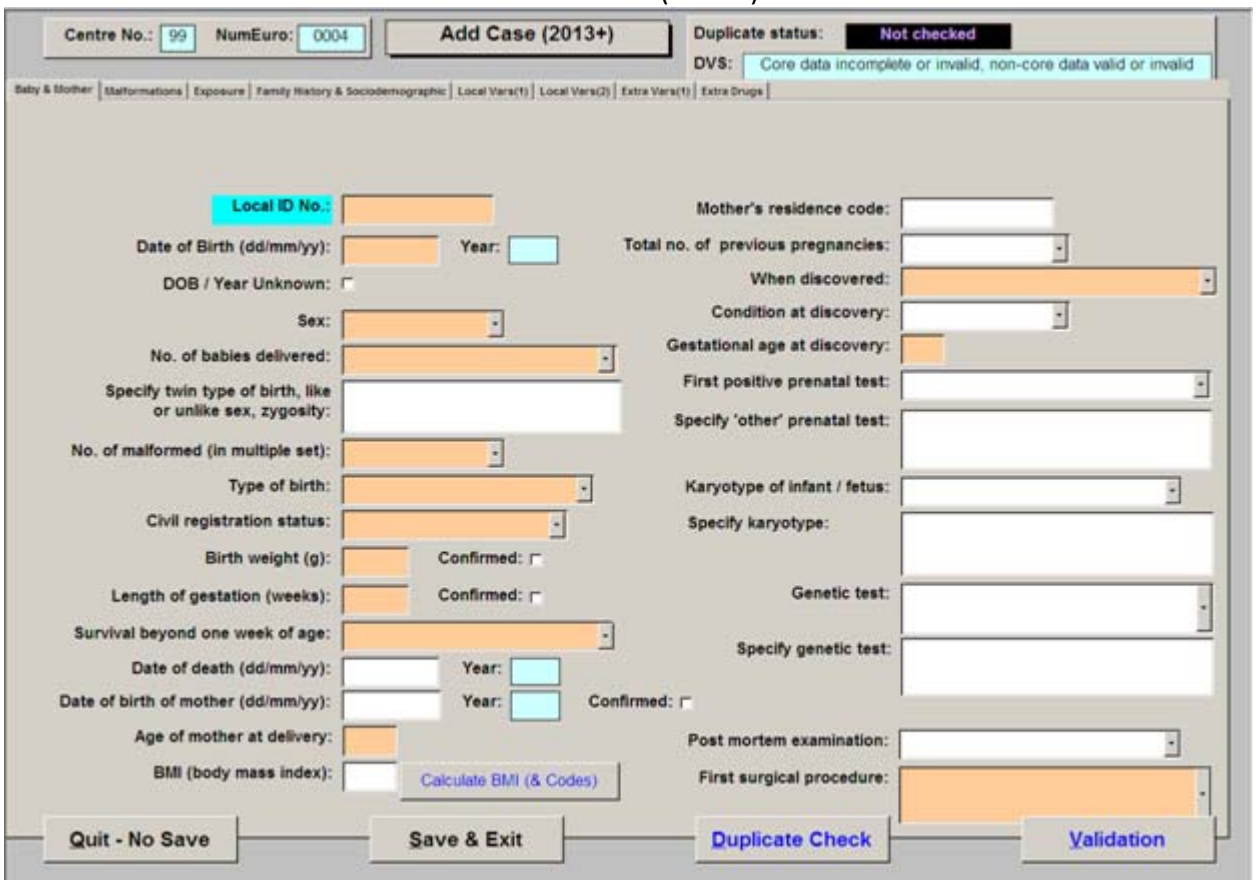
Note in the example below the last page titled 'Extra Variables (1)'. There can be between none and two of these pages containing the extra variables (maximum of 32, 16 per page) as specified under the System Menu. As with the 'Local Variables' they are not included when exporting data to EUROCAT.

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

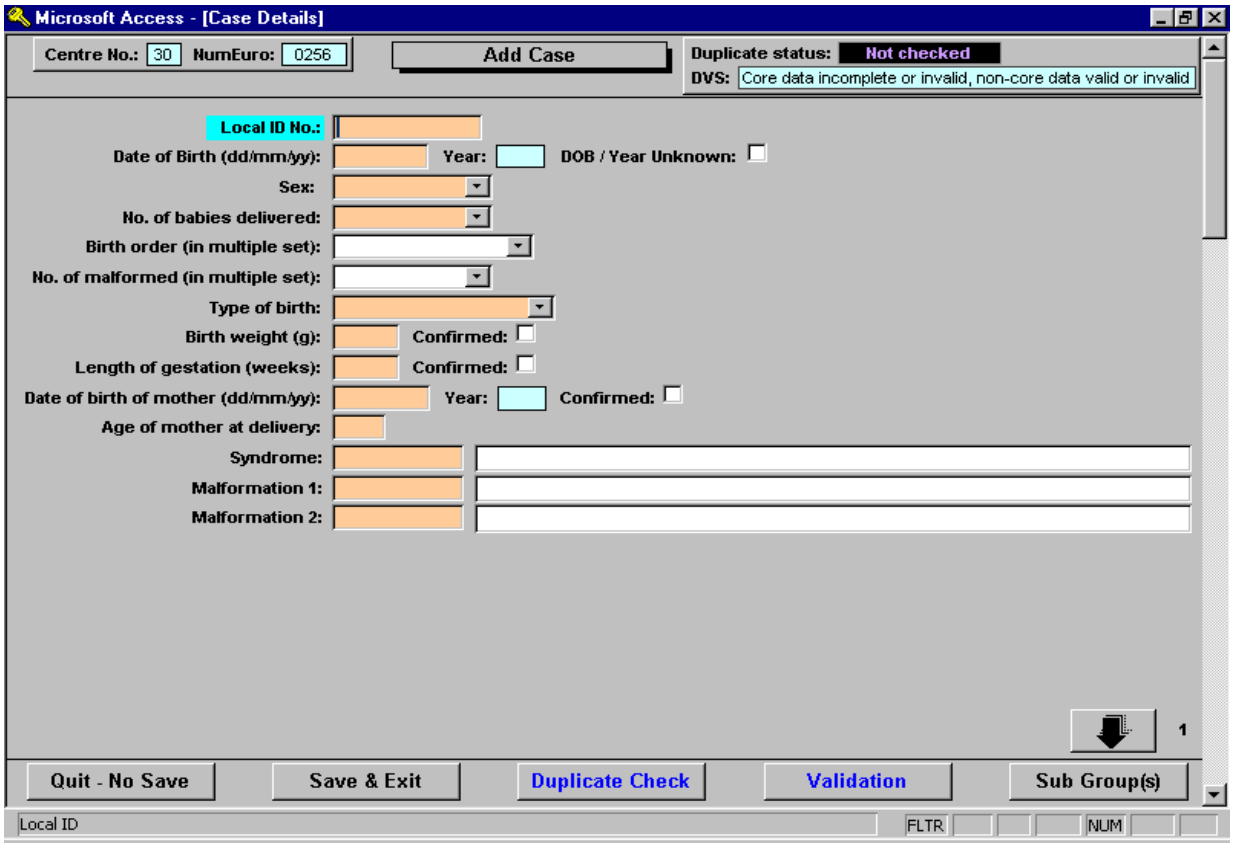
Core Data Only Screen (2013+)



Core and Non-Core Data: Standard Format Screen (2013+)



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

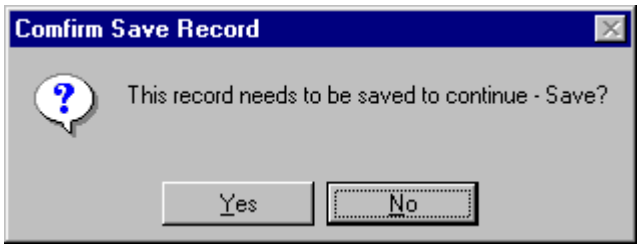


For the user defined data entry screen there are four ways to move between the pages. Firstly when you exit the last field of a page EDMP will automatically move you to the first field of the subsequent page (unless you use the mouse to click on a different part of the current page). Secondly there are '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.

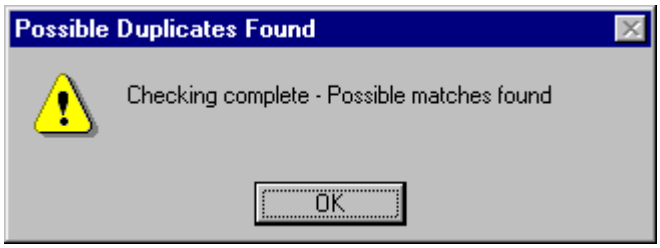
All the data entry screens offer the duplicate checking and validation buttons described below.

6.1.1 Duplicate Checking

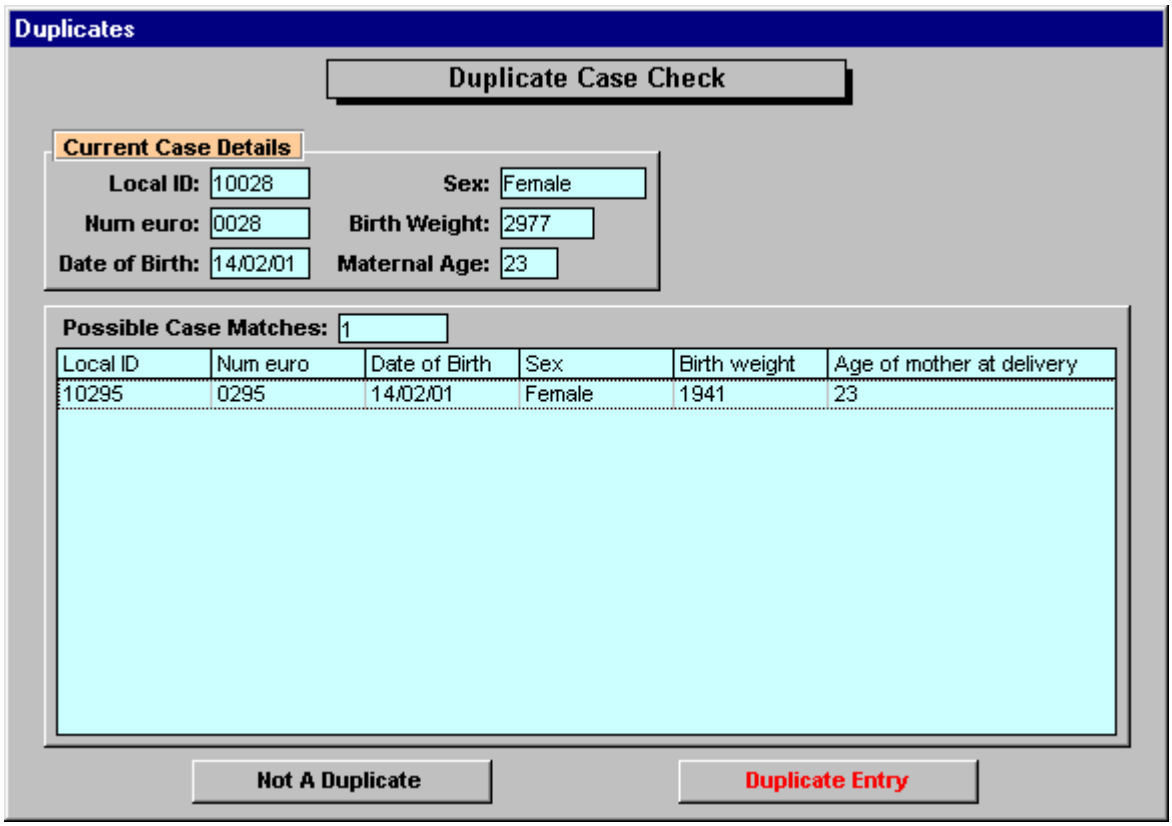
The 'Duplicate check' button when clicked checks for possible matches of other cases against the case you are currently adding or editing. You will be asked to save the record first as shown here:



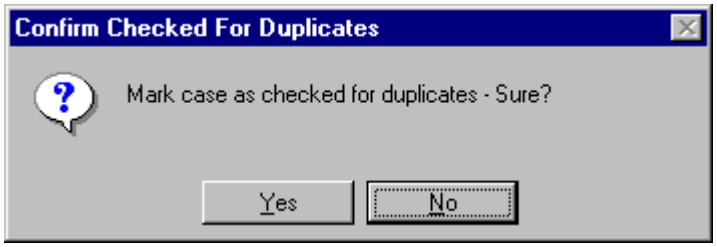
Clicking the 'Yes' Button then allows the checking to take place.
Once checking is completed if any matches are found a screen will pop up informing you that possible matches were found.



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

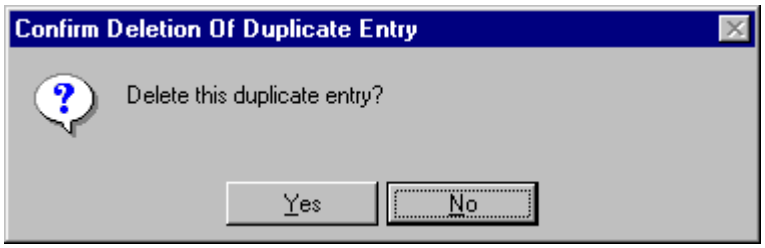


The screen above shows the current case at the top and then lists out the possible matches in the box below. As you can see from the above example there is one possible match against the case, but if it is not a match you would click the 'Not a duplicate' button. You would then be asked the following:



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:

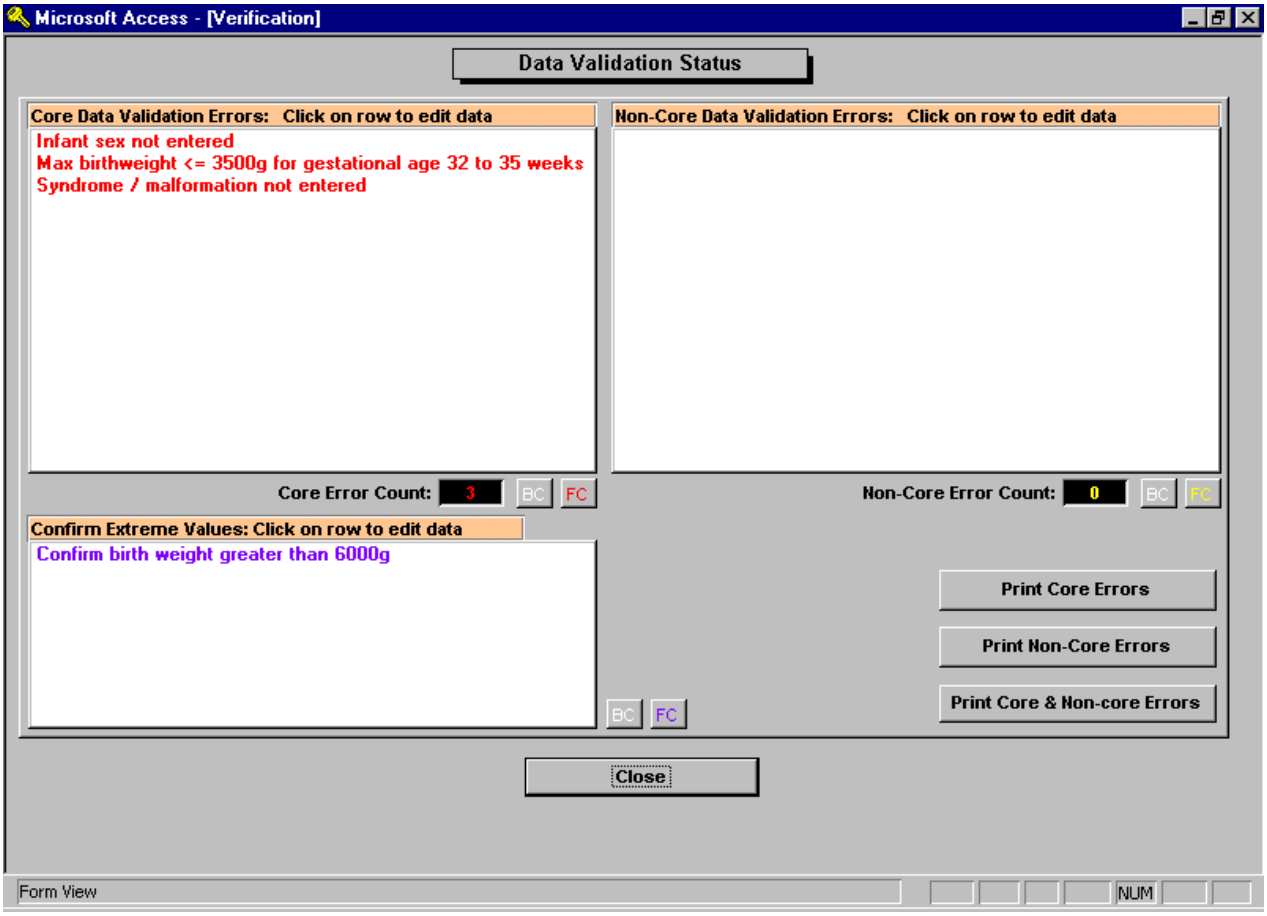


By hitting the 'Yes' button you would be confirming the deletion of the duplicate record.

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 print out 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.

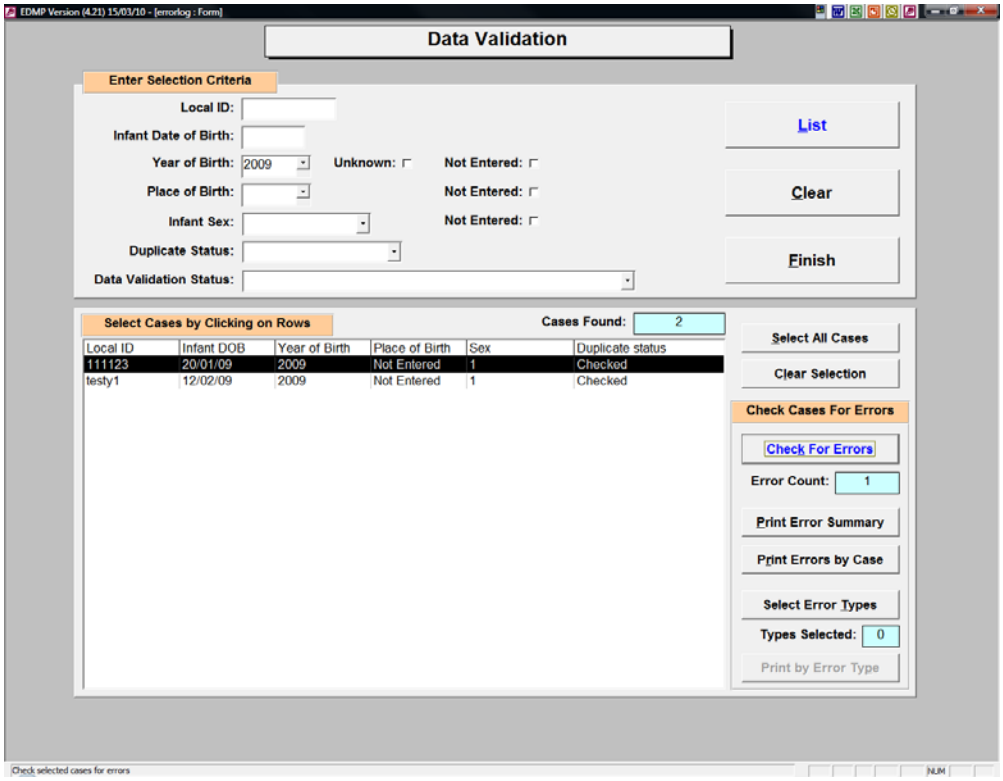
6.3 Print Case

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

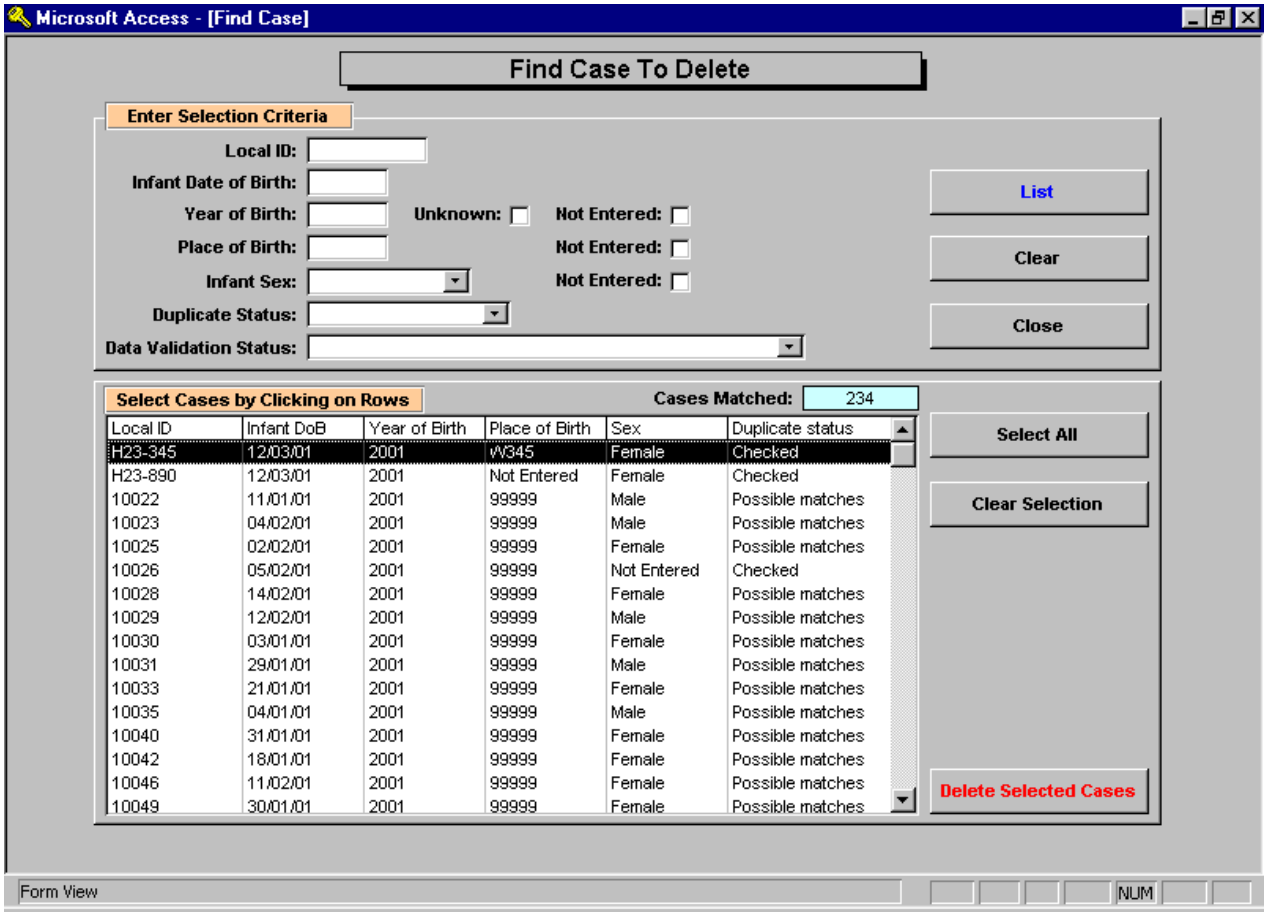
In addition to the data validation available when you are adding or editing a case, EDMP allows you to validate multiple cases in one go. The data validation facility allows you to list all the Core, Non-Core and Extreme value errors for selected cases. Select the required cases to check using the screen 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, 1.3 or 1.4.

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

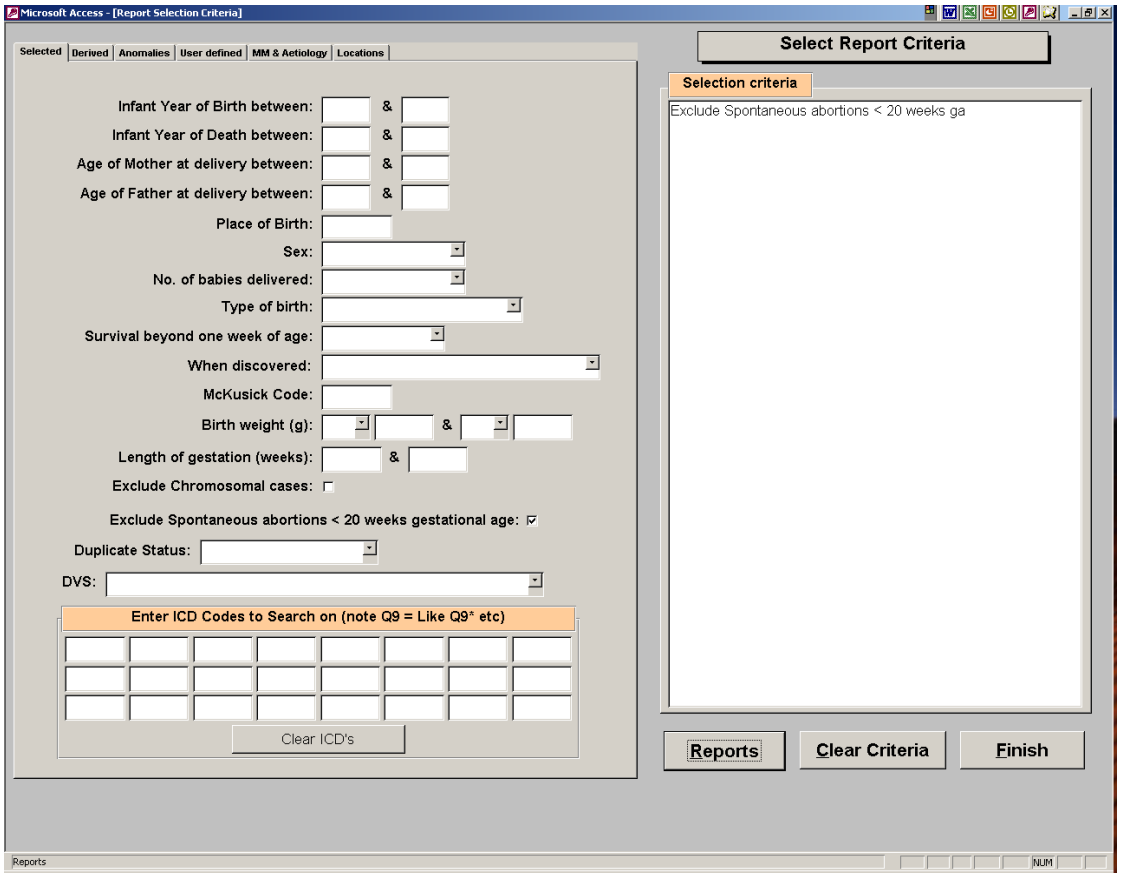
This section provides you with a simple and powerful way to analyse and describe your data. In EDMP there are now four categories of reports, List / Export Cases & Frequency Reports, Prevalence Tables, Prenatal Detection Rates and Missing Values tabulation. Each category allows you to select which data 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) similar to 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 Prenatal Detection Rates

This option allows you to output prenatal diagnosis data for a 4 or 5 year period. Ensure that no other Excel sheet is open on your computer. Enter your start and end dates, then click on 'Output Prenatal Diagnosis Data to Excel' and provide a file name. This then opens an Excel spreadsheet with 5 tabs named: Overall, By Outcome, By Gestation, By Maternal Age and By Indication

7.4 Perinatal Mortality Rates

This option allows you to output perinatal mortality data for a time period (5 years expected). Ensure that no other Excel sheet is open on your computer. Enter your start and end dates, then click on 'Output Perinatal Mortality Data to Excel' and provide a file name. This then automatically opens an Excel spreadsheet with 2 tabs – Table 1 and Table 2.

7.5 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.

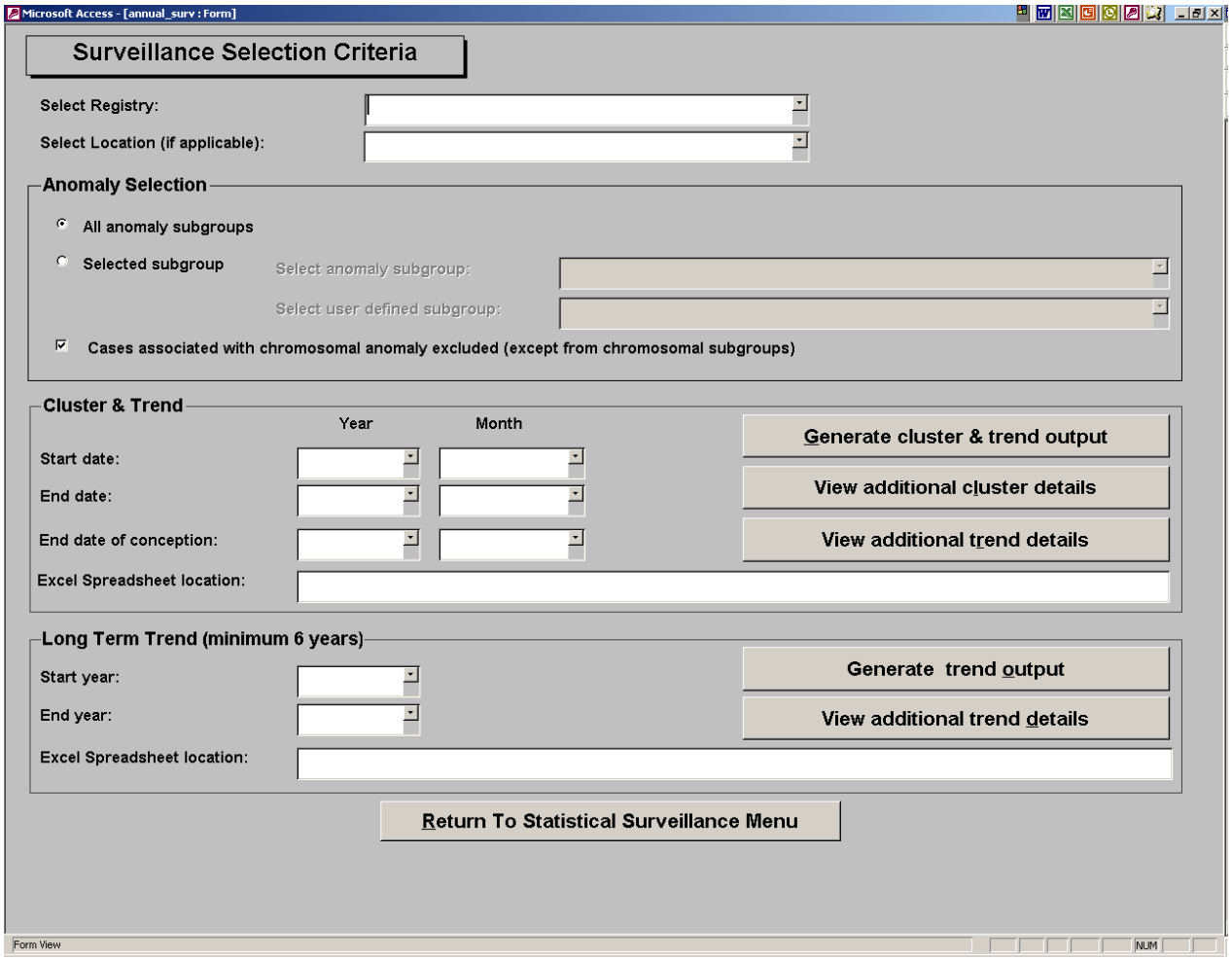
9 Surveillance

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 subgroups 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, genetic syndromes/microdeletions and skeletal dysplasia are excluded from trend and cluster analysis of the remaining subgroups.

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 8 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:

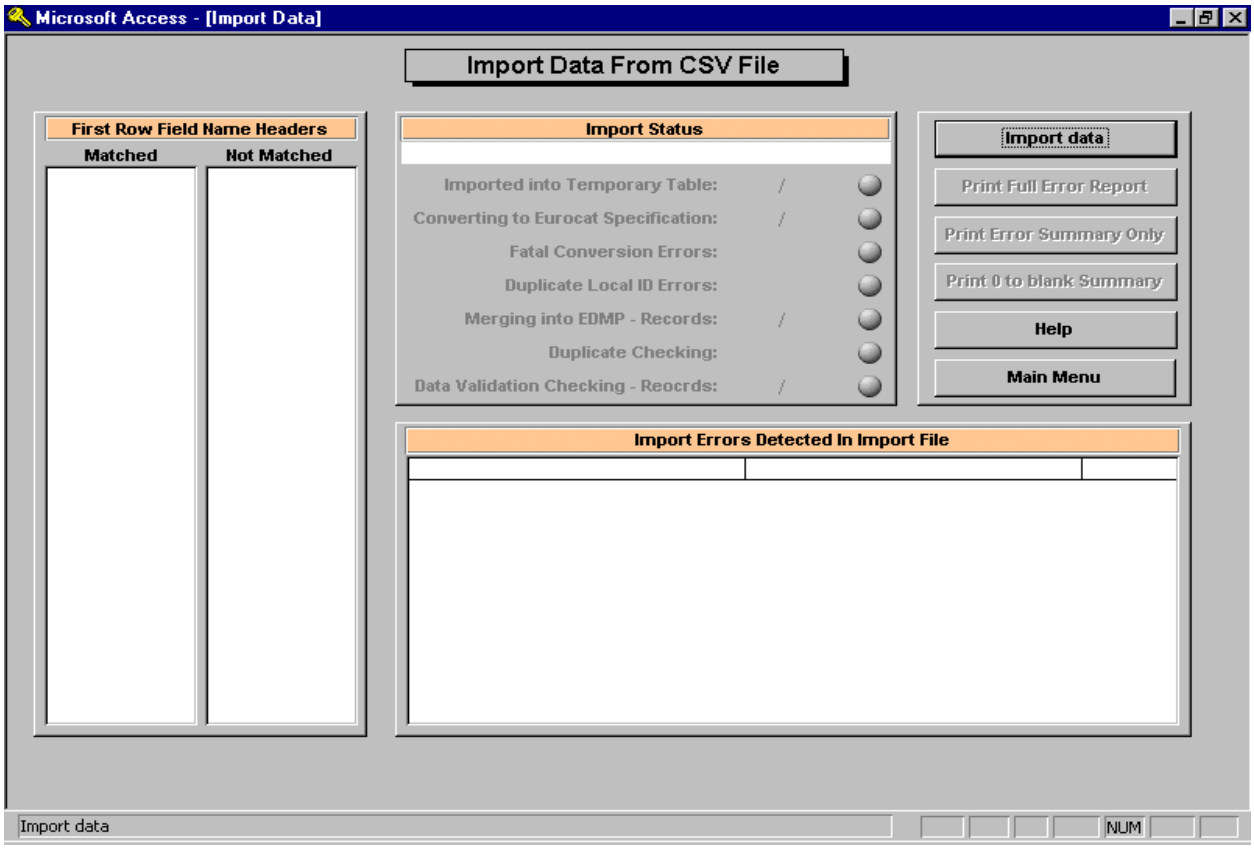
  Alt 124 = |

  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>

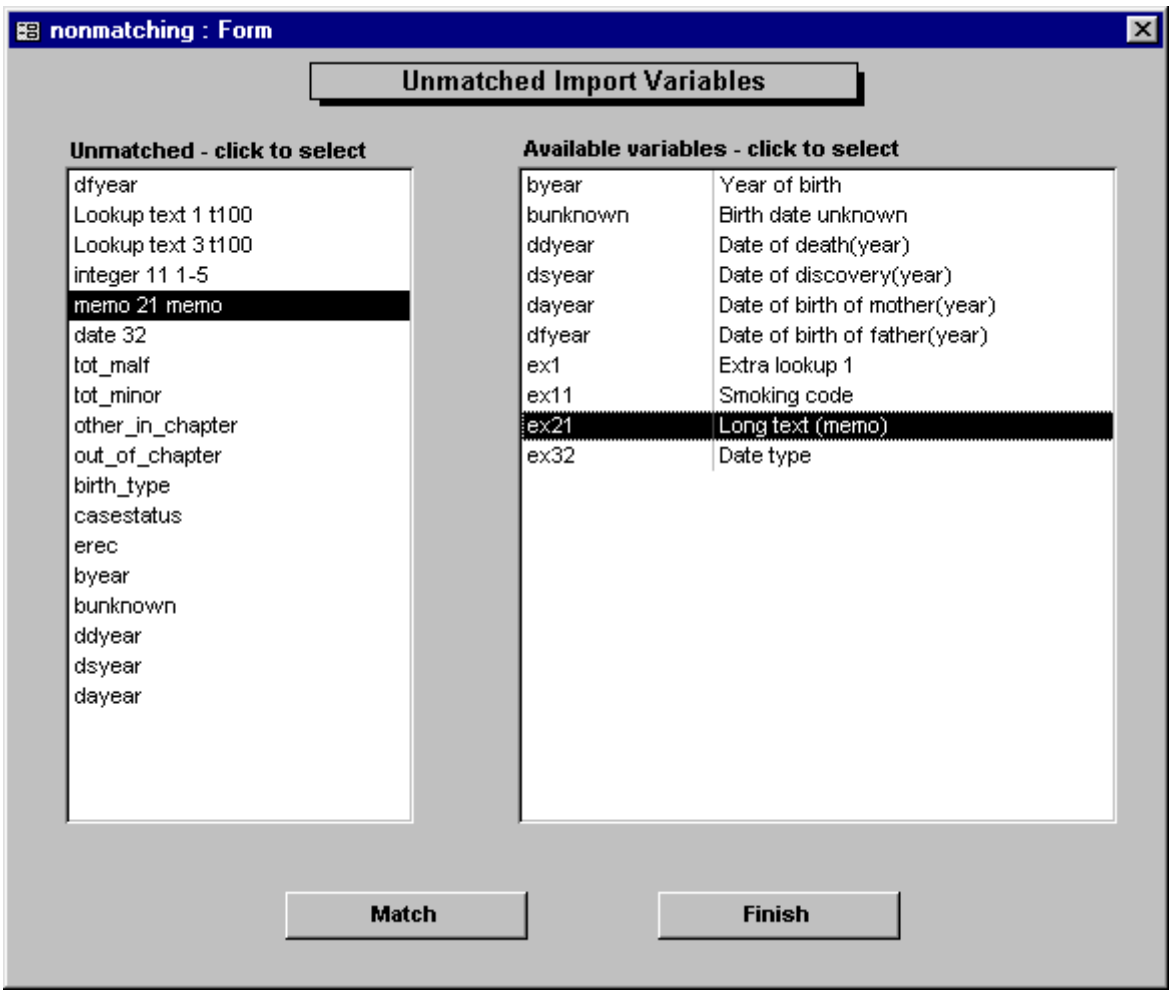
  So for example a case with valproate and lamotrigine exposure is entered in the extra_drugs field as: < N03AG01| Sodium Valproate>< N03AX09| Lamotrigine>

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

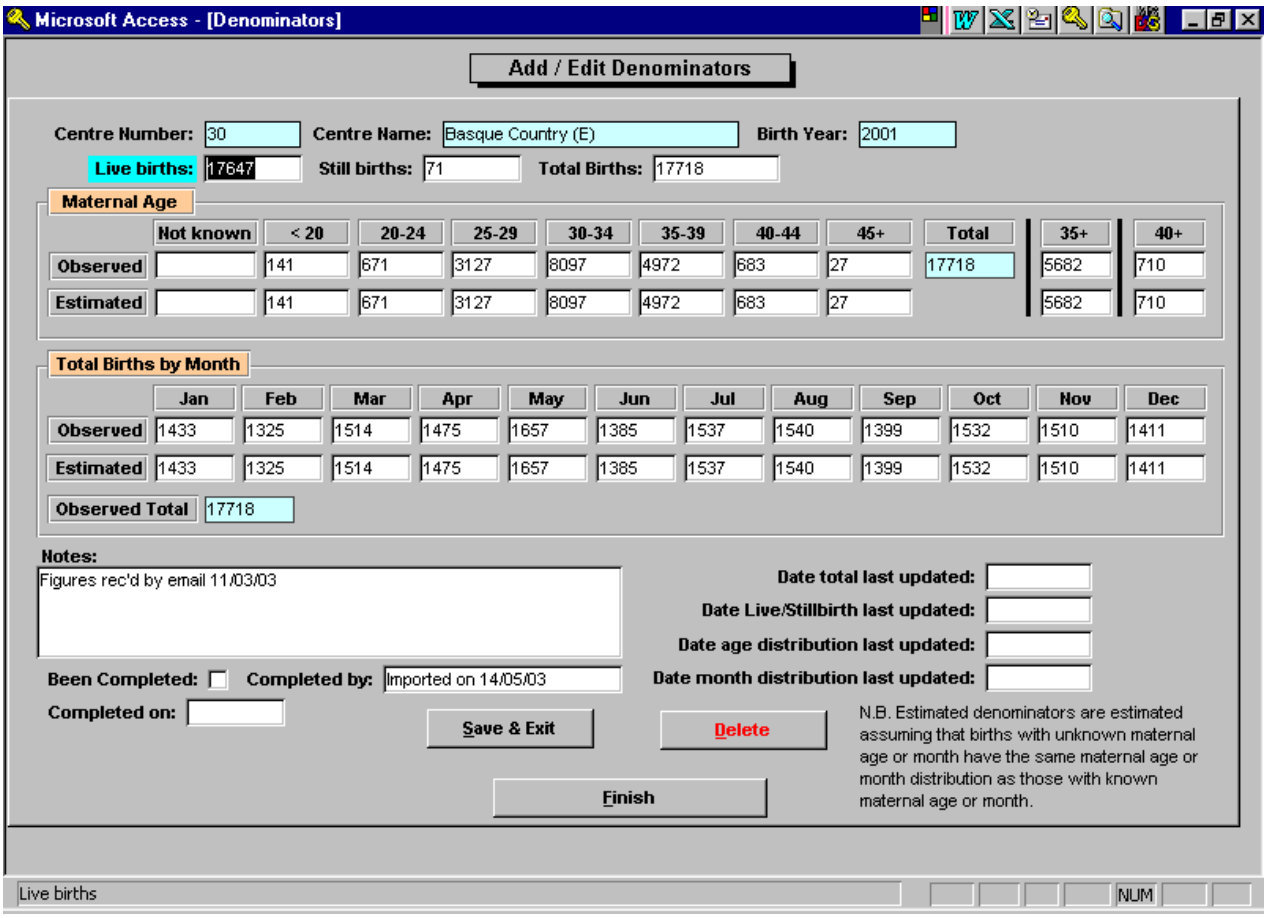
When sending data to the Central Registry you can now send the data for more than one year in a single file. To export data simply select the required year or years from the screen shown below and click on the 'Export Cases' button. You will need to specify the name and location of the export csv file 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, 2005-2012 and 2013+ cases separately.

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 1.4. 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

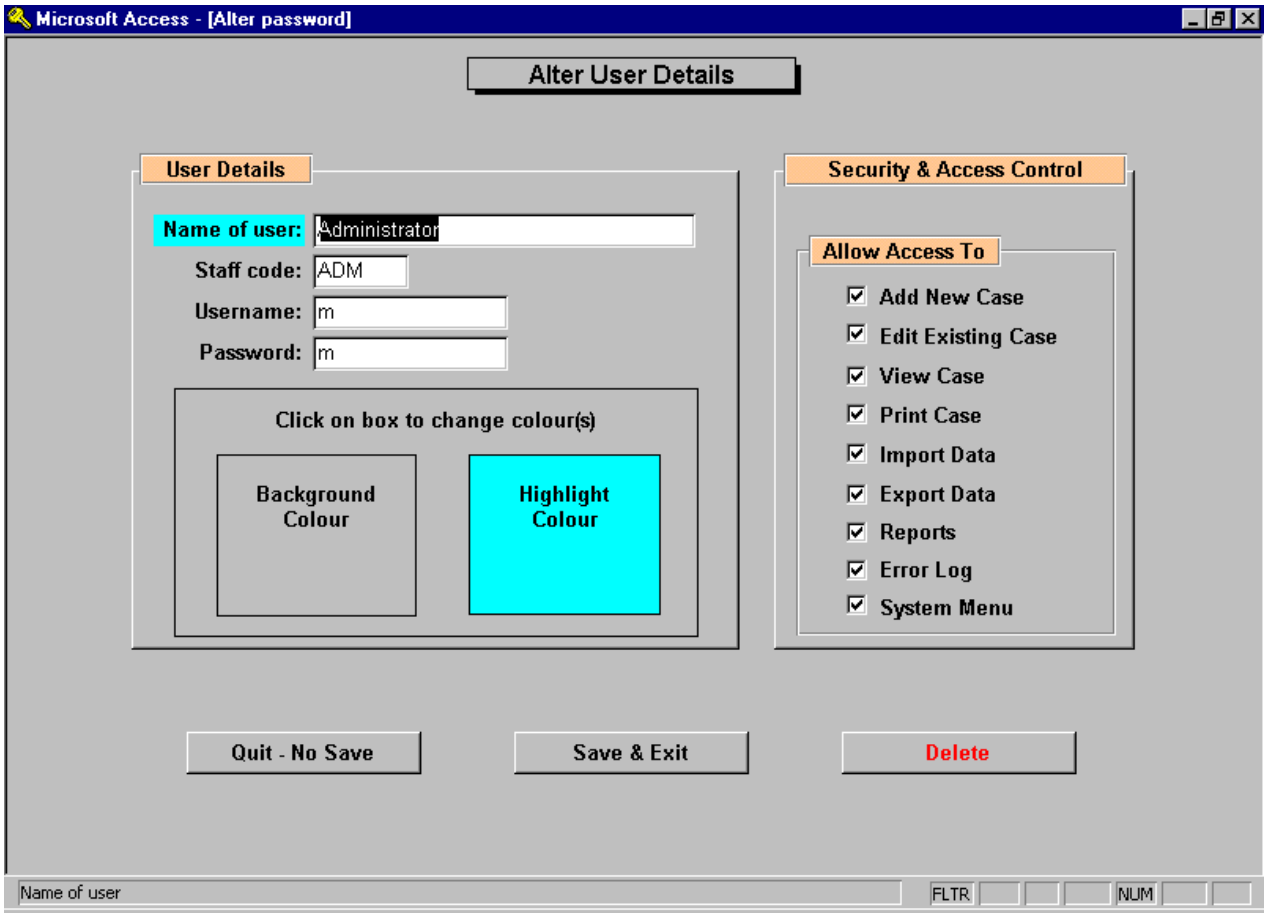
This option allows you to select denominator years and then create an export file in the format required by the Central Registry.

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.



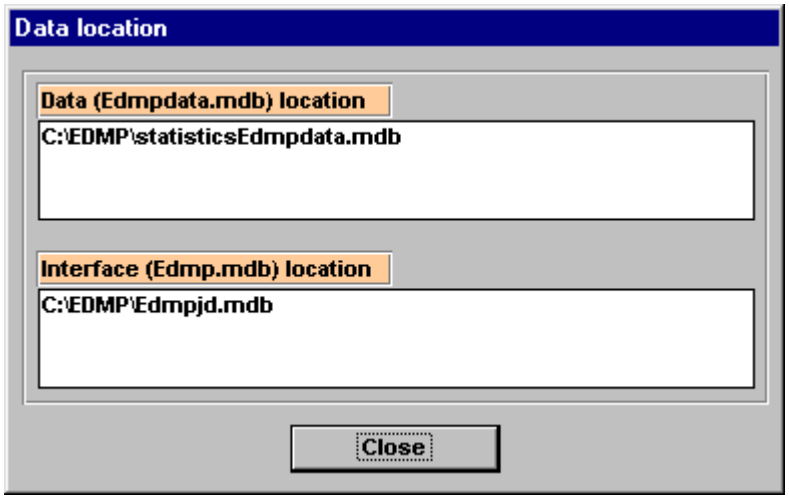
User details allows you to enter (or alter) user details including username and password as well as allow you to alter the personalised screen colour settings for each user. To change screen and highlight colours simply click on the required box on the screen and the standard Windows colour selection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

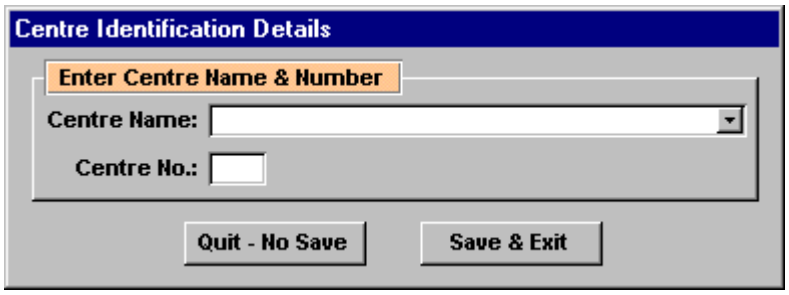
12.3 Data location

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.

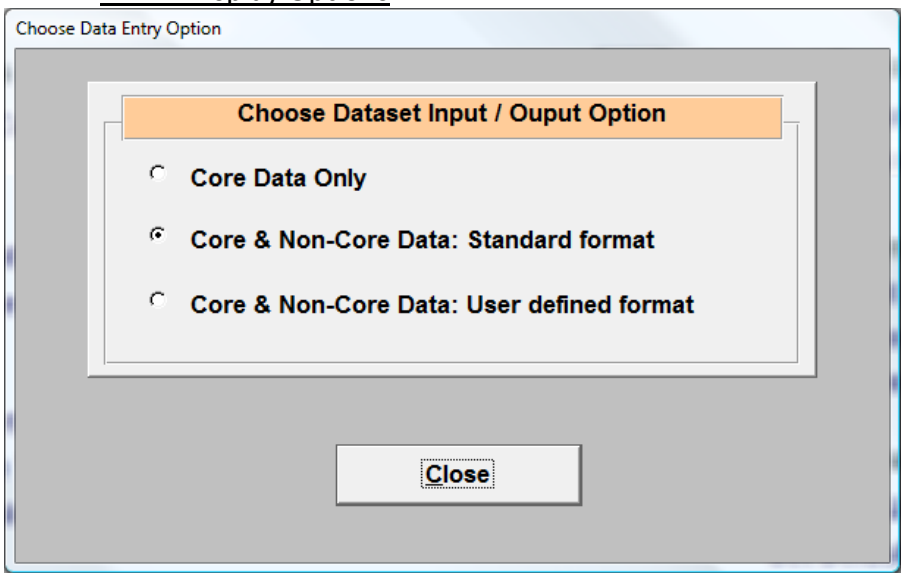
12.6.3 Change Spare Variable Names

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 & ethnic 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 'DatabaseTools' from the menu bar. Then select the 'Compact & Repair Databases' option. You will then be prompted for the location of Edmp.mde.

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 (variable numloc) within a registry cannot be duplicated
- Data with values outside the accepted range (as described in Chapter 2.2.1b) will not be imported into 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, 6 or 7), 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, civil registration status, birthweight, length of gestation, survival status, age of mother, when malformation was discovered, first surgery 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, 6 or 7, then NBRMALF must be entered.
  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, 6 or 7, 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 TOPFA), then SURVIVAL must be 2 (No)
  3. If type of birth =4 (TOPFA), 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 checked
  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, 9 or 11.

- Malformation codes
  1. If OMIM 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 in a livebirth to be counted in this anomaly subgroup
  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 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

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 and regional or national databases 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 8th 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 not included in the prevalence of "all anomalies". Registries should make clear in their registry description which non-Q conditions they register..

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, cases with chromosomal anomalies, skeletal dysplasia cases, genetic syndromes and microdeletions will be excluded from the analysis (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 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 3.4. Manual review of the identified potential multiply malformed cases (approx 10%) will be done before statistical surveillance.

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, 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 (5th 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 GA <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

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, 1.4 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	Q640, Q641	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 syndrome	Q90	7580				al89
Patau syndrome / trisomy 13	Q914-Q917	7581				al90
Edwards syndrome / trisomy 18	Q910-Q913	7582				al91
Turner syndrome	Q96	75860, 75861, 75862, 75869				al92
Klinefelter 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

3.4 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 congenital 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, 1.4 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)

3.5 Detailed Congenital Anomaly Coding Guidelines

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-network.eu/content/EUROCAT-Q-Chapter-2008.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

Q204 SINGLE VENTRICLE, COMMON VENTRICLE, DOUBLE INLET LEFT VENTRICLE, COR TRILOCULARE BIATRIATUM
A single ventricle has absence or near total absence of the ventricular septum. If there is a hypoplastic ventricle, the anomaly should be coded as hypoplastic left heart (Q234) or hypoplastic right heart (Q226)
Coding Committee November 2013

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 (see Q-Chapter under Malformation Coding Guides)
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 (see Q-Chapter under Malformation Coding Guides)
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 (see Q-Chapter under Malformation Coding Guides)
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 ≥ 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

Q660 CLUBFOOT/TALIPES EQUINOVARUS
Clubfoot cases requiring surgery or Ponsetti treatment should be reported to EUROCAT as a major congenital anomaly using the code Q660. If the foot anomaly is of postural origin and not receiving treatment as mentioned, use the code Q668 and the anomaly will be classified as a minor anomaly
Coding Committee November 2013

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

Q674 MICROGNATHIA /OTHER CONGENITAL DEFORMITIES OF SKULL, FACE AND JAW
This code SHOULD be used for MILD micrognathia – see coding tip for Pierre-Robin (Q8708). The code Q674 is classified as a minor anomaly
Coding Committee November 2013

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 syndrome

Q91 Edwards syndrome and Patau 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 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:

K070 Micrognathia
This code is the recommended code for SEVERE micrognathia. See coding tip for Pierre-Robin (Q8708)
Coding Committee November 2013

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

TRAP sequence:
Twin **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

3.6 EUROCAT Description of the 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 and the pulmonary artery from the left ventricle	no
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 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
Edwards syndrome/trisomy 18	karyotype 47,XX +18 or 47,XY +18 and translocations/mosaicism	No
Turner syndrome	karyotype 45,X or structural anomalies of X chromosome	Yes
Klinefelter syndrome	karyotype 47,XXY or additional X-chromosomes	yes

Chapter 4 – Prevalence Rates

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.

EUROCAT prevalence is always cited as per 10,000 births.

Total prevalence = $$\frac{\text{No. Cases (LB + FD + TOPFA)}}{\text{No. Births (live and still)}} \times 10{,}000$$

Livebirth prevalence = $$\frac{\text{No. Cases (LB)}}{\text{No. Births (live)}} \times 10{,}000$$

Fetal death prevalence = $$\frac{\text{No. Cases (FD)}}{\text{No. Births (live and still)}} \times 10{,}000$$

TOPFA prevalence = $$\frac{\text{No. Cases (TOPFA)}}{\text{No. Births (live and still)}} \times 10{,}000$$

Cases =	Cases of congenital anomaly in population
LB =	Live birth
FD =	Fetal deaths from 20 weeks' gestation
TOPFA =	Termination of pregnancy for fetal anomaly after prenatal diagnosis, at any gestational 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 as terminations of pregnancy are included in the numerator but not the denominator, but are not great enough to have an important effect on prevalence.

Lower 95% confidence limit = $$\frac{\left(\frac{1.96}{2} - \sqrt{c + 0.02}\right)^2}{b} \times 10{,}000$$

Upper 95% confidence limit = $$\frac{\left(\frac{1.96}{2} + \sqrt{c + 0.96}\right)^2}{b} \times 10{,}000$$

c = No. Cases

b = No. Births

Note: The confidence intervals are calculated using the Poisson distribution.
Reference: Bégaud B, Martin K, Abouelfath A, Tubert-Bitter P, Moore N, Moride Y. Any easy to use method to approximate Poisson confidence limits. European Journal of Epidemiology (2005) 20: 213-216.

Differences in total prevalence 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 Interpretation of prevalence).

Differences in livebirth or fetal death prevalence 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 is followed by termination of pregnancy.

Calculation of Proportions and their 95% Confidence Intervals

Livebirth proportion = $$\frac{\text{No. Cases (LB)}}{\text{All Cases (LB + FD + TOPFA)}} \times 100$$

Fetal death proportion = $$\frac{\text{No. Cases (FD)}}{\text{All Cases (LB + FD + TOPFA)}} \times 100$$

TOPFA proportion = $$\frac{\text{No. Cases (TOPFA)}}{\text{All Cases (LB + FD + TOPFA)}} \times 100$$

Cases = Cases of congenital anomaly in population
LB = Live birth
FD = Fetal deaths from 20 weeks' gestation
TOPFA = Termination of pregnancy for fetal anomaly after prenatal diagnosis, at any gestational age

Lower 95% confidence limit = $$\frac{p + \frac{1.96^2}{2a} - 1.96\sqrt{\frac{p(1-p)}{a} + \frac{1.96^2}{4a^2}}}{1 + \frac{1.96^2}{a}} \times 100$$

Upper 95% confidence limit = $$\frac{p + \frac{1.96^2}{2a} + 1.96\sqrt{\frac{p(1-p)}{a} + \frac{1.96^2}{4a^2}}}{1 + \frac{1.96^2}{a}} \times 100$$

a = All cases (LB + FD + TOPFA)

p = Proportion/100

Note: The confidence intervals are calculated using the Binomial distribution.
Reference: Agresti A, Coull BA. Approximate is Better than "Exact" for Interval Estimation of Binomial Proportions. The American Statistician, Vol. 52, No. 2 (May, 1998), pp. 119-126.

4.2 Interpretation of Prevalence Rates

For the definition of "Prevalence" see Calculation of prevalence.

EUROCAT registries follow a number of principles of organisation and registration to optimise the accuracy of estimation of prevalence and achieve standardisation across regions. The following factors potentially affect the accuracy of estimation of prevalence.

Definition of the population

The definition of the population covered by each registry is given under Member Registries (http://www.eurocat-network.eu/aboutus/memberregistries). The majority of registries of EUROCAT are population-based, which means that they cover residents of a defined geographical area to obtain unbiased rates.

Definition and classification of birth defects and diagnostic practice

Epidemiological data are derived from diagnoses made by clinicians working within given health service conditions. Many variations in diagnostic practice may affect the reported prevalence of birth defects. For example, the accurate reporting of chromosomal anomalies (e.g. 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 (e.g. hypoplastic left heart syndrome). Renal dysplasia is more likely to be diagnosed early in life if there is ultrasound screening of the kidneys, which leads to variation in prevalence between regions and over time as screening practice changes.

Minor anomalies are those which do not in themselves have serious medical, functional or cosmetic consequences for the child. Cases with only minor anomalies are excluded from EUROCAT (see Guide 1.4, chapter 3.2 for list of minor anomalies). Minor anomalies are included if they appear in association with major anomalies. Some anomalies are present in gradations from minor to major forms and variable prevalence in these anomalies can be due to variable registration of their minor forms and lack of details in the medical notes allowing identification of the minor forms.

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 of other defects i.e. "sequences" (e.g. hydrocephaly when associated with spina bifida) are counted only under the primary defect in EUROCAT prevalence (see classification of subgroups, Guide 1.4 chapter 3.3).

Ascertainment and Coding

Registries work hard to establish and maintain an information pathway which will lead to high case ascertainment (i.e. the proportion of diagnosed cases who are registered), and accurate diagnostic information. EUROCAT registries use version 10 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.

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 post-neonatal period are also vital for confirmation of diagnostic details.

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 weeks to the stillbirth limit can influence prevalence substantially for certain congenital anomalies.

Termination of pregnancy for fetal anomaly following prenatal diagnosis

Prenatal screening policies (and the resources for prenatal screening) vary enormously between different countries and between regions and even hospitals within countries. Laws and practices vary between countries as to the upper gestational age limit for termination (see the Registry Descriptions of Member Registries for more detail, and the EUROCAT publications list). 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 anencephaly, but the practice is much more variable for conditions such as spina bifida. Thus, prenatal screening followed by termination of pregnancy introduces considerable geographic and temporal variation in prevalence, 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.

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 numbers of early terminations are high 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. For example children with sex chromosome trisomies are often not diagnosed until puberty. However some are now detected prenatally due to screening for Down's syndrome.

Chapter 5 – Registration Descriptions and Data Quality

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 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 case 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. 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
8. Specific congenital anomalies not recorded by your registry

Maximum Age at Diagnosis

1. What is the maximum age at diagnosis of livebirths for inclusion on the register (see Chapter 5.2 QD5 of Registry Description Questionnaire)

Termination of Pregnancy for Fetal Anomaly (TOPFA)

1. Is termination of pregnancy for fetal anomaly (TOPFA) following prenatal diagnosis 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 TOPFAs? How complete is ascertainment of TOPFAs? 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?)

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? 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), TOPFA 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?

Exposure Data Availability

Please detail variables in Guide 1.4 which are 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. Does the operation of your registry require the approval of an ethics committee or similar? Please give details
2. Does the operation of your registry require parental consent? If yes, please give details of procedure and percentage of cases where consent is withheld.

Address for Further Information

1. How is your registry staffed (expertise and time)?

5.2 Registry Description Questionnaire

Dear Applicant Registry

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.

EUROCAT will place a Member Registry Description of your registry on the EUROCAT website. To view examples of other Member Registry descriptions visit **http://www.eurocat-network.eu/aboutus/memberregistries**

Please transmit your completed Registry Description Questionnaire to EUROCAT Central Registry by emailing eurocat@ulster.ac.uk

Member Registries should:

- Have an expertise and interest in the field of the epidemiology of congenital anomalies
- Have the human and financial resources required at local level to run the registry
- Cover a geographically defined population
- Include all types of congenital anomaly, registration of live births, still births and terminations of pregnancy for fetal anomaly following prenatal diagnosis. Registration should be based on multiple sources of ascertainment with an emphasis on high quality data.
- Full Member registries only should demonstrate the capacity to transmit to the Central Registry the EUROCAT standard data set on baby, diagnosis and exposure (as specified in EUROCAT Guide 1.4 (http://www.eurocat-network.eu/content/EUROCAT-Guide-1.4-Section-2.4.pdf)
- 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 (Chapter 3.3 Guide 1.4).

Applications must be approved by both the Steering Committee and the Registry Advisory Service.

The attached questionnaire has been designed to allow 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.

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. Answer the questions in detail - use as much space as needed.

Abbreviations used on the questionnaire are as follows: LB = live births, SB = still births, TOPFA = terminations of pregnancy for fetal anomaly, following prenatal diagnosis.

EUROCAT REGISTRY DESCRIPTION QUESTIONNAIRE

Please read Guide 1.4 and visit the EUROCAT website http://www.eurocat-network.eu before completing this questionnaire

I am applying for:
☐ Full Membership (complete entire questionnaire)
☐ Associate, Affiliate, World Affiliate 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 (Full and Associate Only) ☐

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) + TOPFA

☐ 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 (eg. 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 (eg. 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
☐ Other

Required by which law? ☐

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/ ☐ ☐ wks of gestation
or

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	☐	☐	☐
TOPFA	☐	☐	☐

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
TOPFA	☐ Yes	☐ No

F PRENATAL SCREENING AND TERMINATION OF PREGNANCY

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

If yes, since (year) ☐

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

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

F4 Which sources provided notification of TOPFAs and how completely do you estimate TOPFAs 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 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 anomalies, *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 1.4, 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 anomalies? 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 1.4 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.
http://www.eurocat-network.eu/aboutus/datacollection/guidelinesforregistration/guide1_4

H1 Would you be able to transmit the core variables exactly as in Guide 1.4? ☐ 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 1.4 format?	**Record in the Register**	**Transmission in Guide 1.4 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 1.4 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 1.4 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:**
http://www.eurocat-network.eu/content/EUROCAT-Guide-1.4-Section-2.4.pdf
☐ Yes ☐ No

Are you willing to use this for data transmission to Central Registry?

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 1.4. (Please add more rows if required.)

Year	**Total Cases:**			**Total Births:**		*Total Prevalence per 10,000*		
	Livebirths	**Fetal Deaths ***	**TOPFAs ****	**Livebirths**	**Stillbirths**	**LB**	**LB+FD**	**LB+FD+TOPFA**
TOTAL								

* 20 weeks gestation and above ** Following prenatal diagnosis

Table 2. Prevalence rates of selected anomalies, including livebirths, fetal deaths and terminations of pregnancy for fetal anomaly following prenatal diagnosis.
Notes:

1. Fetal deaths include stillbirths and late fetal deaths, EXCLUDING fetal deaths before 20 weeks gestation. TOPFAs following prenatal diagnosis at any gestational age. The total prevalence rate per 10,000 births is calculated by dividing the total cases (LB+FD+TOPFA) by the total births (LB + SB).
2. The definition/coding of each anomaly can be found in Chapter 3.3, EUROCAT Subgroups, Guide 1.4

Anomalies	LB Number	FD Number	TOPFA Number	Prevalence (LB+FD+TOPFA/ 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				
Severe CHD				
Common arterial truncus				
Transposition great vessels				
Single ventricle				
Ventricular Septal Defect				
Atrial Septal Defect				
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 LB term infant (GA 37+ weeks)				
Respiratory				
Choanal atresia				

Anomalies	LB Number	FD Number	TOPFA Number	Prevalence (LB+FD+TOPFA/ 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 techeo-oesophageal fistula				
Duodenal atresia or stenosis				
Atresia or stenosis or 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				
Other anomalies / syndromes				
Skeletal dysplasias				
Craniosynostosis				
Congenital constriction bands/amniotic band				
Situs inversus				
Conjoined twins				

Anomalies	LB Number	FD Number	TOPFA Number	Prevalence (LB+FD+TOPFA/ total births)
Congenital skin disorders				
Teratogenic syndromes with malformations				
Fetal alcohol syndrome				
Valproate syndrome				
Maternal infections resulting in malformations				
Genetic syndromes + microdeletions				
Sequences				
Chromosomal				
Down syndrome				
Patau syndrome / trisomy 13				
Edwards syndrome / trisomy 18				
Turner syndrome				
Klinefelter 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.

5.3 Definition of Data Quality Indicators

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 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:

Ascertainment
Accuracy of Diagnosis
Completeness of Information
Timeliness
Denominator Information

List of Data Quality Indicators (DQI)

Ascertainment

- **Total number of cases**
- **Total congenital anomaly prevalence (>200 per 10,000 births expected), with 95% confidence intervals**
  All cases in malformation chapter ICD10 (Q chapter) or ICD9 (range 740-759), including outside malformation codes (D215, D821, D1810, P350, P351, P371, 74, 75, 27910, 2281, 76076, 76280, 7710, 7711, 77121). EUROCAT minor anomalies are excluded.
- **Prevalence of anencephalus**
- **Prevalence of severe cardiac defects**
- **Prevalence of selected postnatal diagnosis**
  Includes codes for corpus callosum anomalies (Q040), cataract (Q120), coarctation of aorta (Q251), Hirschprung's disease (Q431) and craniosynostosis (Q750).
- **Prevalence of genetic syndromes and microdeletions**
- **Prevalence of malformed fetal deaths**
- **Down syndrome: Observed/Expected ratio by maternal age**
  This calculates the ratio of Observed to Expected Down Syndrome cases. Observed (O) is the number of livebirth (LB) + fetal death (FD) ≥20 weeks gestational age + the number of TOPFA corrected for probability of fetal survival to 20 weeks.

  The calculation is:
  O = LB + FD + (TOPFA corrected to 20 weeks gestational age)

  Expected (E) is based on EUROCAT average 5 year maternal age-specific estimates (LB +FD + TOPFA corrected to 20 weeks) for the time period of analysis applied to the maternal age profile of each registry birth population.

Accuracy of diagnosis

- **% potential multiples according to the flowchart variable**
- **% fetal deaths with post-mortem examination carried out**
- **% TOPFA (GA ≥ 15 weeks) with post-mortem examination carried out**
- **% chromosomal cases (except trisomy 13, 18 and 21) with karyotype text**
- **% Non-chromosomal potential multiple cases with known karyotype**
- **Prevalence of selected exact 4-digit Q-BPA codes**
  Selected Q-BPA codes = Q0000, Q0020, Q0400, Q0435, Q2110, Q2121, Q2510, Q2511, Q2620, Q3380, Q3911, Q4420, Q6141, Q6420, Q7131, Q8980.
- **Prevalence of selected unspecified Q codes**
  Selected unspecified codes = Q049, Q059, Q249, Q339, Q439, Q549, Q639, Q749, Q799, Q899, Q999
- **% livebirths with ASD, VSD, hydronephrosis, hypospadia or club foot with known data on surgery**

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)

- **Number of core variables 90% complete (out of chosen 11)**
  Variables are: sex, number of babies/fetuses delivered (nbrbaby), number of malformed in multiple set (nbrmalf), type of birth (type), birth weight (weight), length of gestation in completed weeks (gestlength), survival beyond one week of age (survival), when discovered (whendisc), if prenatally diagnosed, gestational age at discovery in completed weeks (agedisc), age of mother at delivery (agemo), civil registration status (civreg).
- **Number of non-core variables 80% complete(out of chosen 26)**
  date of death (death-date), condition at discovery (condisc), karyotype of infant/fetus (karyo), post mortem examination (pm), date of birth of mother (datemo), mother's residence code (residmo), total number of previous pregnancies (totpreg), mother's occupation at time of conception (occupmo), assisted conception (assconcept), illness before pregnancy (illbef), illness during pregnancy (illdur1), drugs1, consanguinity (consang), previous malformed siblings notified to EUROCAT (prevsib), sibling ID number notified to Central Registry (sib1), siblings with anomalies (sibanom), mother's family with anomalies (moanom), father's family with anomalies (faanom), first postitive prenatal test (firstpre), first surgical procedure for malformation (surgery), folic acid supplementation (folic), maternal education (matedu), socioeconomic status of mother (socm), socioeconomic status of father (socf), migrant status (migrant), aetiological classification of malformation (aetiology).
- **% TOPFA with civil registration known**
- **% live births with one week survival known**
- **Medication exposure recorded using 7 digit ATC codes**
  Yes or No
- **% of ATC codes with 7 digits and in correct format**
- **% genetic syndromes + microdeletions with syndrome text complete**
- **% malformation 1 text complete**
- **Number of unresolved data edits (excluding free text fields)**

Timeliness

- **Timeliness for February deadline**
  Yes or No

Denominator Information

- **Years with 80% of maternal age denominators (out of 5)**
- **Years with monthly denominators (out of 5)**

Chapter 6 – Data Protection and Access to Data

Policy regarding Security, Confidentiality, Release and Publications 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.

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.

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.

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.

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 – Amendments to Guide 1.4 since its Publication in 2013

7.1 Summary of Amendments to Guide 1.4