



EUROCAT Guide 1.4 and Reference Documents
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EUROCAT SYNDROME GUIDE: Definition and Coding of Syndromes (Revised 2017)
ICD10-BPA Extension Codes

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| Chapter 1 Aims and Objectives |
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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**
- 2.2 Variables and Coding Instructions for Transmission of Data to EUROCAT Central Registry**
 - 2.2.1a Summary of Variables**
 - 2.2.1b Coding Instructions**
 - 2.2.2 EDMP Derived Variables**
 - 2.2.3 Recommended Local Variables**
 - 2.2.4 Template for Associate Member Registry Data Transmission**
- 2.3 Template for Denominator Data**
- 2.4 EUROCAT Data Management Program (EDMP) Instructions**
- 2.5 Data Validation Routines**

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. Guide 1.4 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_TWINS | 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 | DATAMO | 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 58 | | |
| 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 |

| Variable Number | Variable Name | Variable heading |
|--|---------------|---|
| 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 |
| 58§ | ORPHA | ORPHA code |
| Exposure – Variables 59to 79 | | |
| 59** | ASSCONCEPT | Assisted conception |
| 60## | OCCUPMO | Mother's occupation at time of conception |
| 61 | ILLBEF1 | Illness before pregnancy 1 |
| 62 | ILLBEF2 | Illness before pregnancy 2 |
| 63* | MATDIAB | Maternal Pregestational Diabetes |
| 64* | HbA1c | Glycated haemoglobin value |
| 65 | ILLDUR1 | Illness during pregnancy |
| 66 | ILLDUR2 | Illness during pregnancy 2 |
| 67* | FOLIC_G14 | Folic acid supplementation |
| 68* | FIRSTTRI | First trimester medication |
| 69 | DRUGS1 | Drugs |
| 70 | SP_DRUGS1 | Specify drug exposures |
| 71 | DRUGS2 | As for DRUGS1 |
| 72 | SP_DRUGS2 | Specify drug exposures |
| 73 | DRUGS3 | As for DRUGS1 |
| 74 | SP_DRUGS3 | Specify drug exposures |
| 75 | DRUGS4 | As for DRUGS1 |
| 76 | SP_DRUGS4 | Specify drug exposures |
| 77 | DRUGS5 | As for DRUGS1 |
| 78 | SP_DRUGS5 | Specify drug exposures |
| 79 | EXTRA_DRUGS | Extra drugs |
| Family History – Variables 80 to 91 | | |
| 80 | CONSANG | Consanguinity |
| 81 | SP_CONSANG | Specify text information on consanguinity |
| 82 | SIBANOM | Siblings with anomalies |
| 83 | SP_SIBANOM | Specify type of anomaly and describe the malformation |
| 84 | PREVSIB | Previous malformed sibs notified to EUROCAT |
| 85 | SIB1 | Local ID number notified to the Central Registry |
| 86 | SIB2 | As SIB1 |
| 87 | SIB3 | As SIB1 |
| 88 | MOANOM | Mother's family with anomalies |
| 89 | SP_MOANOM | Specify type of anomaly and describe the malformation |

| Variable Number | Variable Name | Variable heading |
|--|---------------|---|
| 90 | FAANOM | Father's family with anomalies |
| 91 | SP_FAANOM | Specify type of anomaly and describe the malformation |
| Sociodemographic – Variables 92 to 95 | | |
| 92 | MATEDU | Maternal education |
| 93 | SOCM | Socioeconomic status of mother |
| 94 | SOCF | Socioeconomic status of father |
| 95 | MIGRANT | Migrant status |
| General Comments – Variable 96 | | |
| 96 | 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
- § Variable added in February 2018

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2.2.1b Coding Instructions

Baby and Mother (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|-----------------|---------------|---|--|
| 1 | CENTRE | <u>CENTRE NUMBER</u> | Code allocated by Central Registry |
| 2 | NUMLOC | <u>LOCAL ID</u> 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 | <u>DATE OF BIRTH</u> 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). Cases reported with unknown date of birth will not be included in cluster analysis. | Day, month, year 99 = Not known for day and month DO NOT TRANSMIT RECORDS IF YEAR OF BIRTH IS NOT KNOWN |
| 4 | SEX | <u>SEX</u> 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 | <u>NUMBER OF BABIES/FETUSES DELIVERED</u> 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 "MALFO1" (variable 32). Give full description of type of conjoined twinning in MALFO1 text field (variable 33). 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 | <u>SPECIFY TWIN TYPE OF BIRTH</u> (malformed and non-malformed), like or unlike sex, zygosity | Free text |
| 7 | NBRMALF | <u>NUMBER OF MALFORMED IN MULTIPLE SET</u> 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 | <u>TYPE OF BIRTH</u> 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 | <p><u>CIVIL REGISTRATION STATUS</u> Livebirths and stillbirths are civilly registered leading to either a birth or stillbirth certificate and appear in official birth statistics for your region.</p> <p>Code here whether this case fulfilled the conditions for live or stillbirth registration in your country.</p> | <p>1 = Livebirth 2 = Stillbirth 3 = No civil registration 9 = Not known</p> |
| 10 | WEIGHT | <p><u>BIRTH WEIGHT</u> Give weight in grams.</p> | <p>9999 = Not known</p> <p>(Do not use 99 or 999 for "Not Known" as this will be considered the birth weight).</p> |
| 11 | GESTLENGTH | <p><u>LENGTH OF GESTATION IN COMPLETED WEEKS</u> 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.</p> | <p>99 = Not known</p> |
| 12 | SURVIVAL | <p><u>SURVIVAL BEYOND ONE WEEK OF AGE</u> Yes = Child known to be alive after one week.</p> <p>No = Child known to have died before or during first week (including stillbirths and abortions).</p> <p>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.</p> <p>If survival at one week is unknown, but survival at discharge from maternity unit less than one week is known, use the latter.</p> <p>The definition of first week of life varies between countries. Follow your country's perinatal mortality definition and specify this in your Registry Description.</p> <p>Not known = Not known if child has died during first week.</p> | <p>1 = Yes 2 = No 3 = Alive at discharge <1 week 9 = Not known</p> |

Baby and Mother (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|-----------------|---------------|---|---|
| 13 | DEATH_DATE | <p><u>DATE OF DEATH</u> For livebirths only.</p> <p>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).</p> | <p>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.)</p> <p>222222= Known to be alive at 1 year 333333= Not known if alive or dead at 1 year</p> |
| 14 | DATEMO | <p><u>DATE OF BIRTH OF MOTHER</u> 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).</p> <p>This variable can be used to calculate maternal age at Expected Date of Delivery for preterm deliveries and terminations.</p> | <p>Day, month, year 99 = Not known day or month 44 = Not known year</p> |
| 15 | AGEMO | <p><u>AGE OF THE MOTHER AT DELIVERY</u> 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.</p> | 99 = Not known |
| 16 | BMI | <p><u>MATERNAL BODY MASS INDEX</u> 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)</p> <p>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</p> | <p>2 digits Expected range 15 – 50</p> <p>97 = exact BMI NK but <30 98 = exact BMI NK but >=30 99 = Not known</p> |
| 17 | RESIDMO | <p><u>MOTHER’S RESIDENCE CODE</u> Use local code for locality of residence at time of delivery.</p> | Local code (up to 10 digits) |
| 18 | TOTPREG | <p><u>TOTAL NUMBER OF PREVIOUS PREGNANCIES</u> NOTE – The current reported pregnancy is NOT included.</p> <p>Include all previous abortions whether spontaneous or induced. Multiple pregnancies count as 1 in the total</p> | <p>00 = None 01 = One 02 = Two 03 = Three etc 20 = Twenty or more 99 = Not known</p> |

Diagnosis (core variables shaded blue)

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

Diagnosis (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|-----------------|---------------|---|---|
| 22 | FIRSTPRE | <p><u>FIRST POSITIVE PRENATAL TEST</u></p> <p>This refers to the first prenatal test whether screening procedure or diagnostic test which indicated a possible congenital anomaly or need for further tests.</p> <p>For code 7 = other specified test, give information in text field (variable 23).</p> <p>If test performed and result negative, then the “When discovered” variable cannot be coded 6 (prenatal diagnosis).</p> <p>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</p> | <p>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</p> |
| 23 | SP_FIRSTPRE | <p><u>SPECIFY “OTHER” FIRST PRENATAL TEST</u></p> <p>If FIRSTPRE = 7, specify which positive prenatal test</p> | Free text |
| 24 | KARYO | <p><u>KARYOTYPE OF INFANT/FETUS</u></p> <p>Specify result in variable 25. Array results count as a karyotype test</p> <p>Report only clearly pathogenic variants and if uncertain, include only copy number variants (CNVs) (duplications or deletions) larger than 1 MB. Only report cases with de novo CNVs unless the parent in familial cases also has clinical manifestations of the condition (dysmorphic features or congenital anomalies). (Coding Committee 2015)</p> <p>If performed and results known, please specify (according to the latest ISCN edition).</p> <p>“Probe test performed” refers to FISH, PCR, or other analyses restricted to specific chromosomal regions .</p> <p>“Failed” refers to a technical failure where a repeat examination could not be done and the karyotype is therefore unknown.</p> | <p>1 = Performed, result known 2 = Performed, results not known 3 = Not performed 4 = Probe test performed 8 = Failed 9 = Not known</p> |

Diagnosis (core variables shaded blue)

| Variable Number | Variable Name | EXPLANATION AND INSTRUCTIONS | Code |
|-----------------|---------------|---|---|
| 25 | SP_KARYO | <p><u>SPECIFY KARYOTYPE</u> OR CHROMOSOMAL MICROARRAY</p> <p>EXAMPLES:</p> <p>47,XY,+21</p> <p>46,XX,del(2)(p13p23)mat Description: An interstitial deletion resulting from mal-segregation of a maternal insertion</p> <p>46,XY,der(5)ins(5;2)(q31;p23p13)mat Description : A derivative chromosome 5 resulting from mal-segregation of a maternal insertion from chromosome 2</p> <p>arr(1-22)x3,(X)x2,(Y)x1 Description: Microarray analysis shows triploidy 69,XXY</p> <p>arr(8)x3,(21)x3 Description: Microarray analysis shows a single copy gain of chromosomes 8 and 21.</p> <p>46,XY,rsa(13,18,21)x2,(X,Y)x1 Description: normal chromosomes, only chromosome 13,18,21, X and Y investigated</p> <p>rsa 22q11.2('kit name')x1 Description: microdeletion 22q.11.2 diagnosed by multiple ligation probe amplification method (MLPA)</p> | Free text |
| 26 | GENTEST | <p><u>GENETIC TEST</u></p> <p>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</p> <p>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</p> <p>If the test is performed but the result not yet known, please wait for the result before reporting</p> | 1 = specific genetic test positive 2 = specific genetic test negative 3 = Specific genetic test not Performed 9 = Not Known if genetic test is performed or result not known |
| 27 | SP_GENTEST | <p><u>SPECIFY TYPE OF GENETIC TEST</u></p> <p>Give method used and the result of the test (type of mutation and which gene)</p> <p>Examples: Single gene analysis, exome sequencing, whole genome sequencing</p> | Free text |

Diagnosis (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|-----------------|---------------|--|---|
| 28 | PM | <p><u>POST MORTEM EXAMINATION</u></p> <p>If performed record the malformation(s) discovered in the “malformation” section in the form. If other findings record in the “comments” space (variable 95).</p> <p>“Results known” means that the autopsy record has been reviewed by the registry.</p> <p>“Results not known” means that the autopsy record was not available to the registry.</p> <p>“Macerated fetus” means that although a post mortem was performed, maceration of the fetus prevented a full protocol from being followed.</p> | <p>1 = Performed, results known 2 = Performed, results not known 3 = Not performed 4 = Macerated fetus 9 = Not known</p> |
| 29 | SURGERY | <p><u>FIRST SURGICAL PROCEDURE FOR MALFORMATION (PERFORMED OR EXPECTED)</u></p> <p>Complete for all livebirths (and fetal deaths, only if there was prenatal surgery)</p> <p>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.</p> <p>“No surgery required” means that this case does not have a severe enough malformation, or that the malformation is not correctable by surgery.</p> <p>“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.</p> | <p>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</p> |
| 30 | SYNDROME | <p><u>SYNDROME OR ASSOCIATION</u></p> <p>Use this variable for genetic syndromes, skeletal dysplasias, teratogenic syndromes, associations, microdeletions and chromosomal anomalies.</p> <p>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.</p> <p>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.</p> <p>Ensure karyotype information is given in variables 24 and 25 and that information on genetic tests are given in variable 26 and 27., Mention in variable 28 if the autopsy report has been reviewed, where appropriate.</p> <p>Local registries are advised to keep photographs and x-ray images of all syndrome cases if possible, as the diagnosis is predominantly established on the basis of specific facial dysmorphism.</p> | <p>ICD 10</p> <p>First 4 digits are ICD10 5th digit = BPA supplement or leave blank</p> |

Diagnosis (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|-----------------|---------------|--|---|
| 31 | SP-SYNDROME | <u>SPECIFY SYNDROME</u> Written text description of the ICD10 code in variable 30 | |
| 32 | MALFO1 | <u>MALFORMATION</u> 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. In case of conjoined twins, give full description in text in variable 33 Up to 8 malformations can be coded – if more than 8 are present, specify additional anomalies in the text variable for the 8 th 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 5 th digit = BPA classification OR leave blank |
| 33 | SP_MALFO1 | <u>SPECIFY MALFORMATION</u> | Free text |
| 34 | MALFO2 | As MALFO1 | As MALFO1 |
| 35 | SP_MALFO2 | <u>SPECIFY MALFORMATION</u> | Free text |
| 36 | MALFO3 | As MALFO1 | As MALFO1 |
| 37 | SP_MALFO3 | <u>SPECIFY MALFORMATION</u> | Free text |
| 38 | MALFO4 | As MALFO1 | As MALFO1 |
| 39 | SP_MALFO4 | <u>SPECIFY MALFORMATION</u> | Free text |
| 40 | MALFO5 | As MALFO1 | As MALFO1 |
| 41 | SP_MALFO5 | <u>SPECIFY MALFORMATION</u> | Free text |
| 42 | MALFO6 | As MALFO1 | As MALFO1 |
| 43 | SP_MALFO6 | <u>SPECIFY MALFORMATION</u> | Free text |
| 44 | MALFO7 | As MALFO1 | As MALFO1 |
| 45 | SP_MALFO7 | <u>SPECIFY MALFORMATION</u> | Free text |
| 46 | MALFO8 | As MALFO1 | As MALFO1 |
| 47 | SP_MALFO8 | <u>SPECIFY MALFORMATION</u> | Free text |

| Diagnosis (core variables shaded blue) | | | | | |
|--|---------------|--|---|--|---|
| Variable Number | Variable Name | Explanation and Instructions | | | Code |
| 48 | PRESYN | <u>PRENATAL DIAGNOSIS FOR SYNDROME</u> 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 <i>exact</i> 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 | | | 1 = Yes, this anomaly was diagnosed prenatally 2 = No, this anomaly was diagnosed postnatally 3 = This anomaly partially prenatally diagnosed 9 =Not known |
| | | <u>Prenatal Finding</u> Double bubble High risk screening (no amnio) Ventriculomegaly Ventriculomegaly Ventriculomegaly Significant heart anomaly Heart abnormality Cleft lip IUGR Anhydramnios Micrognathia Severe skeletal dysplasia Echogenic bowel Absent stomach bubble | <u>Postnatal finding</u> Duodenal atresia/stenosis T21 Agenesis corpus callosum Neuronal migration anomalies Hydrocephalus Any significant heart anomaly 22q11 del Cleft lip and palate Skeletal displasia Renal agenesis Pierre Robin/cleft palate Specific skeletal dysplasia eg thanatophoric/achondrogenesis CF Oesophageal atresia | <u>Prenatal/Postnatal/Partial</u> Prenatal Partial Partial Partial Prenatal Prenatal Partial Partial Postnatal Partial Prenatal Prenatal Partial Partial | |

Diagnosis (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|-----------------|---------------|---|------------------|
| 49 | PREMAL1 | <u>PRENATAL DIAGNOSIS FOR MALFORMATION</u> AS PRESYN | <u>AS PRESYN</u> |
| 50 | PREMAL2 | <u>PRENATAL DIAGNOSIS FOR MALFORMATION</u> AS PRESYN | <u>AS PRESYN</u> |
| 51 | PREMAL3 | <u>PRENATAL DIAGNOSIS FOR MALFORMATION</u> AS PRESYN | <u>AS PRESYN</u> |
| 52 | PREMAL4 | <u>PRENATAL DIAGNOSIS FOR MALFORMATION</u> AS PRESYN | <u>AS PRESYN</u> |
| 53 | PREMAL5 | <u>PRENATAL DIAGNOSIS FOR MALFORMATION</u> AS PRESYN | <u>AS PRESYN</u> |
| 54 | PREMAL6 | <u>PRENATAL DIAGNOSIS FOR MALFORMATION</u> AS PRESYN | <u>AS PRESYN</u> |
| 55 | PREMAL7 | <u>PRENATAL DIAGNOSIS FOR MALFORMATION</u> AS PRESYN | <u>AS PRESYN</u> |
| 56 | PREMAL8 | <u>PRENATAL DIAGNOSIS FOR MALFORMATION</u> AS PRESYN | <u>AS PRESYN</u> |
| 57 | OMIM | <p><u>OMIM / TYPE OF MENDELIAN INHERITANCE</u> To be coded by medical geneticist or after advice from medical geneticist.</p> <p>This code is to be used for cases with single gene origin only – Refer to EUROCAT Syndrome Guide.</p> <p>The first digit may be filled in without the rest of the code if the full OMIM code is not known.</p> <p>Full codes can be found on the OMIM website http://www.ncbi.nlm.nih.gov/omim/</p> | |
| 58 | ORPHA | <p>This code is to be used for rare diseases including congenital anomalies, chromosomal, teratogenic and genetic syndromes.</p> <p>Use the link and enter the name of the condition. if more than one code/disease appears, select the most specific orpha code. If you do not have specific information about genetic background or phenotype, select the most general orpha code.</p> <p>https://www.orpha.net/consor/cgi-bin/Disease.php?lng=EN</p> | |

Exposure (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|-----------------|---------------|--|--|
| 59 | ASSCONCEPT | <u>ASSISTED CONCEPTION</u> 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 |
| 60 | OCCUPMO | <u>MOTHER'S OCCUPATION AT TIME OF CONCEPTION</u> 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 | | | | | | | | | | | | | | | | | | | | | | | | | | |
|----------------------------|---------------------------|---|--|-------------|----------------|-------------|-----------------|-------------|-----------------|-------------|---------|-------------|---------------------|-------------|----------------------------|-----------|------------|-------------|----------|-------------|--------------|-------------|--------|-------------|--------------------|------|-------------|---------------------------|--|
| 61 | ILLBEF1 | <p><u>ILLNESS BEFORE PREGNANCY 1</u></p> <p>Record any illness whether chronic or acute with onset before pregnancy and that may affect fetal development (eg. childhood cancer, metabolic and endocrine disease, severe congenital anomaly). Code according to ICD10. The codes mentioned below are only examples.</p> <p>Historic maternal diseases may be coded using the Z-chapter in ICD10, example Z853 "Personal history of breast cancer".</p> <p>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)</p> <p>Abridged list:</p> <table><tr><td>Hyperthyroidism</td><td>E050 - E059</td></tr><tr><td>Hypothyroidism</td><td>E000 - E039</td></tr><tr><td>Diabetes Type 1</td><td>E100 - E109</td></tr><tr><td>Diabetes Type 2</td><td>E110 - E119</td></tr><tr><td>Obesity</td><td>E660 - E669</td></tr></table> <p>If maternal BMI ≥ 30 give code for obesity</p> <table><tr><td>Metabolic disorders</td><td>E700 – E889</td></tr><tr><td>Anorexia /eating disorders</td><td>F500-F509</td></tr><tr><td>Depression</td><td>F320 – F339</td></tr><tr><td>Epilepsy</td><td>G400 - G409</td></tr><tr><td>Hypertension</td><td>I100 - I159</td></tr><tr><td>Asthma</td><td>J450 - J459</td></tr><tr><td>Chronic alcoholism</td><td>F102</td></tr><tr><td>Drug addict</td><td>F112 - F122 - F132 - F142</td></tr></table> | Hyperthyroidism | E050 - E059 | Hypothyroidism | E000 - E039 | Diabetes Type 1 | E100 - E109 | Diabetes Type 2 | E110 - E119 | Obesity | E660 - E669 | Metabolic disorders | E700 – E889 | Anorexia /eating disorders | F500-F509 | Depression | F320 – F339 | Epilepsy | G400 - G409 | Hypertension | I100 - I159 | Asthma | J450 - J459 | Chronic alcoholism | F102 | Drug addict | F112 - F122 - F132 - F142 | ICD 10 0 = No illness 1 = Yes, but no information available 9 = Not known |
| Hyperthyroidism | E050 - E059 | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Hypothyroidism | E000 - E039 | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Diabetes Type 1 | E100 - E109 | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Diabetes Type 2 | E110 - E119 | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Obesity | E660 - E669 | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Metabolic disorders | E700 – E889 | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Anorexia /eating disorders | F500-F509 | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Depression | F320 – F339 | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Epilepsy | G400 - G409 | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Hypertension | I100 - I159 | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Asthma | J450 - J459 | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Chronic alcoholism | F102 | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Drug addict | F112 - F122 - F132 - F142 | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| 62 | ILLBEF2 | <u>ILLNESS BEFORE PREGNANCY 2 - AS FOR ILLBEF1</u> | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| 63 | MATDIAB | <p><u>MATERNAL PREGESTATIONAL DIABETES</u></p> <p>This variable is specifically for pregestational diabetes. Gestational diabetes is dealt with under the 'illness during pregnancy' variable (variable 64)</p> <p>Type 1 diabetes: characterized by hyperglycemia due to an absolute deficiency of the insulin hormone produced by the pancreas</p> <p>An HbA1c of 48mmol/mol is recommended as the cut-off point for diagnosing diabetes.</p> <p>Type 2 diabetes: characterized by hyperglycemia due to a defect in insulin secretion</p> <p>An HbA1c of 48mmol/mol is recommended as the cut-off point for diagnosing diabetes.</p> <p>*Maturity Onset Diabetes in the Young (MODY) displays an autosomal dominant pattern of inheritance</p> <p>An HbA1c of 48mmol/mol is recommended as the cut-off point for diagnosing diabetes.</p> <p>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.</p> <p>Diagnosed by fasting plasma glucose from 6.1 – 6.9 mmol/L (WHO criteria) http://www.who.int/diabetes/publications/en/</p> | 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)

| Variable Number | Variable Name | Explanation and Instructions | Code | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
|-----------------|---------------|--|------|------|------|------|------|------|------|------|-----|-----|-----|----------|----|----|----|----|----|----|----|----|----|----|---|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|----------|----|----|----|----|----|----|----|----|----|----|---|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|----------|----|----|----|----|----|----|----|----|----|----|---|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|----------|----|----|----|----|----|----|----|----|----|----|---|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|----------|----|----|----|----|----|----|----|----|----|----|---|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|----------|----|----|----|----|----|----|----|----|----|----|---|------|------|------|------|------|------|------|------|------|------|----------|----|----|----|----|----|----|----|----|----|----|---|------|------|------|------|------|------|------|------|------|------|----------|----|----|----|-----|-----|-----|-----|-----|-----|-----|---|------|------|------|------|------|------|------|------|------|------|----------|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|---|------|------|------|------|------|------|------|------|------|------|----------|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|--|
| 64 | HbA1c | <p><u>GLYCATED HAEMOGLOBIN (HbA1c) VALUE</u></p> <p>Give the first HbA1c value measured in the first trimester (in mmol/mol units)</p> <p>Normal values for non-diabetic individuals <48mmol/mol</p> <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> | % | 4.0 | 4.1 | 4.2 | 4.3 | 4.4 | 4.5 | 4.6 | 4.7 | 4.8 | 4.9 | mmol/mol | 20 | 21 | 22 | 23 | 25 | 26 | 27 | 28 | 29 | 30 | % | 5.0 | 5.1 | 5.2 | 5.3 | 5.4 | 5.5 | 5.6 | 5.7 | 5.8 | 5.9 | mmol/mol | 31 | 32 | 33 | 34 | 36 | 37 | 38 | 39 | 40 | 41 | % | 6.0 | 6.1 | 6.2 | 6.3 | 6.4 | 6.5 | 6.6 | 6.7 | 6.8 | 6.9 | mmol/mol | 42 | 43 | 44 | 45 | 46 | 48 | 49 | 50 | 51 | 52 | % | 7.0 | 7.1 | 7.2 | 7.3 | 7.4 | 7.5 | 7.6 | 7.7 | 7.8 | 7.9 | mmol/mol | 53 | 54 | 55 | 56 | 57 | 58 | 60 | 61 | 62 | 63 | % | 8.0 | 8.1 | 8.2 | 8.3 | 8.4 | 8.5 | 8.6 | 8.7 | 8.8 | 8.9 | mmol/mol | 64 | 65 | 66 | 67 | 68 | 69 | 70 | 72 | 73 | 74 | % | 9.0 | 9.1 | 9.2 | 9.3 | 9.4 | 9.5 | 9.6 | 9.7 | 9.8 | 9.9 | mmol/mol | 75 | 76 | 77 | 78 | 79 | 80 | 81 | 83 | 84 | 85 | % | 10.0 | 10.1 | 10.2 | 10.3 | 10.4 | 10.5 | 10.6 | 10.7 | 10.8 | 10.9 | mmol/mol | 86 | 87 | 88 | 89 | 90 | 91 | 92 | 93 | 95 | 96 | % | 11.0 | 11.1 | 11.2 | 11.3 | 11.4 | 11.5 | 11.6 | 11.7 | 11.8 | 11.9 | mmol/mol | 97 | 98 | 99 | 100 | 101 | 102 | 103 | 104 | 105 | 107 | % | 12.0 | 12.1 | 12.2 | 12.3 | 12.4 | 12.5 | 12.6 | 12.7 | 12.8 | 12.9 | mmol/mol | 108 | 109 | 110 | 111 | 112 | 113 | 114 | 115 | 116 | 117 | % | 13.0 | 13.1 | 13.2 | 13.3 | 13.4 | 13.5 | 13.6 | 13.7 | 13.8 | 13.9 | mmol/mol | 119 | 120 | 121 | 122 | 123 | 124 | 125 | 126 | 127 | 128 | <p>999 = Not known</p> <p>3 digits</p> |
| % | 4.0 | 4.1 | 4.2 | 4.3 | 4.4 | 4.5 | 4.6 | 4.7 | 4.8 | 4.9 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| mmol/mol | 20 | 21 | 22 | 23 | 25 | 26 | 27 | 28 | 29 | 30 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| % | 5.0 | 5.1 | 5.2 | 5.3 | 5.4 | 5.5 | 5.6 | 5.7 | 5.8 | 5.9 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| mmol/mol | 31 | 32 | 33 | 34 | 36 | 37 | 38 | 39 | 40 | 41 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| % | 6.0 | 6.1 | 6.2 | 6.3 | 6.4 | 6.5 | 6.6 | 6.7 | 6.8 | 6.9 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| mmol/mol | 42 | 43 | 44 | 45 | 46 | 48 | 49 | 50 | 51 | 52 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| % | 7.0 | 7.1 | 7.2 | 7.3 | 7.4 | 7.5 | 7.6 | 7.7 | 7.8 | 7.9 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| mmol/mol | 53 | 54 | 55 | 56 | 57 | 58 | 60 | 61 | 62 | 63 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| % | 8.0 | 8.1 | 8.2 | 8.3 | 8.4 | 8.5 | 8.6 | 8.7 | 8.8 | 8.9 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| mmol/mol | 64 | 65 | 66 | 67 | 68 | 69 | 70 | 72 | 73 | 74 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| % | 9.0 | 9.1 | 9.2 | 9.3 | 9.4 | 9.5 | 9.6 | 9.7 | 9.8 | 9.9 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| mmol/mol | 75 | 76 | 77 | 78 | 79 | 80 | 81 | 83 | 84 | 85 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| % | 10.0 | 10.1 | 10.2 | 10.3 | 10.4 | 10.5 | 10.6 | 10.7 | 10.8 | 10.9 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| mmol/mol | 86 | 87 | 88 | 89 | 90 | 91 | 92 | 93 | 95 | 96 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| % | 11.0 | 11.1 | 11.2 | 11.3 | 11.4 | 11.5 | 11.6 | 11.7 | 11.8 | 11.9 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| mmol/mol | 97 | 98 | 99 | 100 | 101 | 102 | 103 | 104 | 105 | 107 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| % | 12.0 | 12.1 | 12.2 | 12.3 | 12.4 | 12.5 | 12.6 | 12.7 | 12.8 | 12.9 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| mmol/mol | 108 | 109 | 110 | 111 | 112 | 113 | 114 | 115 | 116 | 117 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| % | 13.0 | 13.1 | 13.2 | 13.3 | 13.4 | 13.5 | 13.6 | 13.7 | 13.8 | 13.9 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| mmol/mol | 119 | 120 | 121 | 122 | 123 | 124 | 125 | 126 | 127 | 128 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |

Exposure (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
|--------------------------------|---------------|--|---|-------------|-----------------------------|-------------|-------------|-------------|-------------|------|--------------------------------|-------------|----------------|-------------|------------|-------------|-----------|-------------|----------|-------------|-------|-------------|---------|-------------|----------|-------------|---------------|-------------|-------------------------|-------------|-----------------|-------------|------------|------|----------------|-----------|---|
| 65 | ILLDUR1 | <p><u>ILLNESS DURING PREGNANCY</u></p> <p>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</p> <p>For early pregnancy complications use the O chapter in ICD10. For maternal infections, use chapters A and B. (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)</p> <table><tr><td>Gestational Diabetes</td><td>O244 – O249</td></tr><tr><td>Haemorrhage early pregnancy</td><td>O200 – O209</td></tr><tr><td>Hyperemesis</td><td>O210 - O219</td></tr><tr><td>Coxsackie’s</td><td>B341</td></tr><tr><td>Cytomegalic Inclusion Diseases</td><td>B250 - B259</td></tr><tr><td>Herpes Simplex</td><td>B000 - B009</td></tr><tr><td>HIV (AIDS)</td><td>B200 - B249</td></tr><tr><td>Influenza</td><td>J100 - J119</td></tr><tr><td>Listeria</td><td>A320 - A329</td></tr><tr><td>Mumps</td><td>B260 - B269</td></tr><tr><td>Rubella</td><td>B060 - B069</td></tr><tr><td>Syphilis</td><td>A530 - A539</td></tr><tr><td>Toxoplasmosis</td><td>B580 - B589</td></tr><tr><td>Varicella (Chicken Pox)</td><td>B010 - B019</td></tr><tr><td>Viral Hepatitis</td><td>B190 - B199</td></tr><tr><td>Zika virus</td><td>A928</td></tr><tr><td>Drug poisoning</td><td>T360-T509</td></tr></table> | Gestational Diabetes | O244 – O249 | Haemorrhage early pregnancy | O200 – O209 | Hyperemesis | O210 - O219 | Coxsackie’s | B341 | Cytomegalic Inclusion Diseases | B250 - B259 | Herpes Simplex | B000 - B009 | HIV (AIDS) | B200 - B249 | Influenza | J100 - J119 | Listeria | A320 - A329 | Mumps | B260 - B269 | Rubella | B060 - B069 | Syphilis | A530 - A539 | Toxoplasmosis | B580 - B589 | Varicella (Chicken Pox) | B010 - B019 | Viral Hepatitis | B190 - B199 | Zika virus | A928 | Drug poisoning | T360-T509 | <p>ICD 10</p> <p>0 = No</p> <p>1 = Yes, but no information available</p> <p>9 = Not known</p> |
| Gestational Diabetes | O244 – O249 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Haemorrhage early pregnancy | O200 – O209 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Hyperemesis | O210 - O219 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Coxsackie’s | B341 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Cytomegalic Inclusion Diseases | B250 - B259 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Herpes Simplex | B000 - B009 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| HIV (AIDS) | B200 - B249 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Influenza | J100 - J119 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Listeria | A320 - A329 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Mumps | B260 - B269 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Rubella | B060 - B069 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Syphilis | A530 - A539 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Toxoplasmosis | B580 - B589 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Varicella (Chicken Pox) | B010 - B019 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Viral Hepatitis | B190 - B199 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Zika virus | A928 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Drug poisoning | T360-T509 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| 66 | ILLDUR2 | <p><u>ILLNESS DURING PREGNANCY</u></p> <p>AS FOR ILLDUR1</p> | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| 67 | FOLIC_G14 | <p><u>FOLIC ACID SUPPLEMENTATION</u></p> <p>Recommend to your local maternity hospitals or midwives to collect these data.</p> <p>Folic acid supplementations include folic acid only tablets, a multivitamin preparation which contains folic acid or contraceptive pills which contain folic acid.</p> <p>If the folic acid dose is high, please add the code B03BB01 in the drugs variable</p> | <p>1 = Folic acid taken pre and post-conceptionally</p> <p>2 = Folic acid taken only post-conceptionally</p> <p>3 = Folic acid not taken</p> <p>4 = Folic acid taken, timing unknown</p> <p>9 = Not known if folic acid taken</p> | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |

Exposure (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|-----------------|---------------|--|--|
| 68 | FIRSTTRI | <p><u>FIRST TRIMESTER MEDICATION</u></p> <p>“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.</p> <p>“Undetermined” means that the usual data sources were consulted, but</p> <ul style="list-style-type: none"> 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. <p>“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.</p> <p>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.</p> <p>“Not Known” means that the usual data sources were not found.</p> <p>Only fill in DRUGS1-5 and EXTRADRUGS if you have coded FIRSTTRI = 1 (Yes medication taken) or = 4 (Medication taken, but timing unknown).</p> <p>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</p> <ul style="list-style-type: none"> 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. <p>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</p> | <p>1 = Yes, medication taken in first trimester</p> <p>2 = No medication taken in first trimester</p> <p>3 = Undetermined</p> <p>4 = Medication taken, but timing unknown</p> <p>9 = Not Known</p> |

Exposure (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|-----------------|---------------|--|---------------|
| 69 | DRUGS1 | <p><u>DRUGS - 7 DIGITS MAXIMUM</u></p> <p>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).</p> <p>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.</p> <p>Use ATC-coding and use as many digits as possible (from 3 to 7). Website http://www.whooc.no/atcddd/.</p> <p>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.</p> <p>ATC example: N03A: antiepileptic drug N03AF01: carbamazepine</p> <p>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.</p> <p>If a drug overdose or self-poisoning, this MUST be explained in the drug description.</p> | |
| 70 | SP_DRUGS1 | <u>SPECIFY DRUG EXPOSURES</u> | Free text |
| 71 | DRUGS2 | <p><u>AS FOR DRUGS1</u></p> <p>Please give details in text variable 71 SP_DRUGS2.</p> | As for DRUGS1 |
| 72 | SP_DRUGS2 | <u>SPECIFY DRUG EXPOSURES</u> | Free text |
| 73 | DRUGS3 | <p><u>AS FOR DRUGS1</u></p> <p>Please give details in text variable 73 SP_DRUGS3.</p> | As for DRUGS1 |
| 74 | SP_DRUGS3 | <u>SPECIFY DRUG EXPOSURES</u> | Free text |
| 75 | DRUGS4 | <p><u>AS FOR DRUGS1</u></p> <p>Please give details in text variable 75 SP_DRUGS3.</p> | |
| 76 | SP_DRUGS4 | <u>SPECIFY DRUG EXPOSURES</u> | |
| 77 | DRUGS5 | <p><u>AS FOR DRUGS1</u></p> <p>Please give details in text variable 77 SP_DRUGS3.</p> | |
| 78 | SP_DRUGS5 | <u>SPECIFY DRUG EXPOSURES</u> | |

Exposure (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|-----------------|---------------|--|------|
| 79 | EXTRA_DRUGS | <p><u>EXTRA DRUGS</u></p> <p>This field is only to be used if drug fields 1-5 have already been filled.</p> <p>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).</p> <p>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.</p> <p>Use ATC-coding and use as many digits as possible (from 3 to 7). Website http://www.whocc.no/atcddd/.</p> <p>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.</p> <p>ATC example: N03A: antiepileptic drug N03AF01: carbamazepine</p> <p>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.</p> <p>Only drugs taken at physiologic doses to be recorded.</p> <p>If a drug overdose or self-poisoning, this MUST be explained in the drug description.</p> <p><u>If importing data from a local program</u>, enter the ATC code and text description in the following format:</p> <p style="text-align: center;"><ATC code text description></p> <p>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:</p> <p style="text-align: center;"><ATC code text description><ATC code text description></p> <p>For example a case with valproate and lamotrigine exposure is entered in the extra_drugs field as: <N03AG01 Valproate><N03AX09 Lamotrigine></p> <p>(See chapter 2.4 of EDMP User Guide for further guidance)</p> | |

Family History (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|-----------------|---------------|---|---|
| 80 | CONSANG | <u>CONSANGUINITY</u> 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 |
| 81 | SP_CONSANG | <u>SPECIFY TEXT INFORMATION ON CONSANGUINITY</u> | Free text |
| 82 | SIBANOM | <u>SIBS WITH ANOMALIES</u> 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 |
| 83 | SP_SIBANOM | <u>SPECIFY TYPE OF ANOMALY OF SIBLINGS</u> | Free text |
| 84 | PREVSIB | <u>PREVIOUS MALFORMED SIBLINGS NOTIFIED TO EUROCAT</u> 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 |
| 85 | SIB1 | <u>SIB LOCAL ID NUMBER NOTIFIED TO THE CENTRAL REGISTRY</u> 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 |
| 86 | SIB2 | <u>As SIB1</u> | Local ID |
| 87 | SIB3 | <u>As SIB1</u> | Local ID |

Family History (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|-----------------|---------------|--|---|
| 88 | MOANOM | <p><u>MOTHER'S FAMILY WITH ANOMALIES</u></p> <p>Include mother herself as well as mother's family. Specify type of anomaly in written text and relation to the infant.</p> <p>If the aetiology is known, "same" means the same aetiology, even if the spectrum of malformations present is slightly different.</p> <p>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.</p> <p>"Same and other" refers to two different relatives. If a relative has both the same and another anomaly, code "same".</p> <p>Restrict the family to first, second and third degree relatives (mother, father, siblings, grandparents, aunts, uncles, half-siblings, first cousins).</p> <p>Always give details in text variable 88 SP_MOANOM.</p> | <p>1 = Same 2 = Other 3 = Same and other 4 = No 9 = Not known</p> |
| 89 | SP_MOANOM | <u>SPECIFY TYPE OF ANOMALY AND DESCRIBE THE MALFORMATION</u> | Free text |
| 90 | FAANOM | <p><u>FATHER'S FAMILY WITH ANOMALIES</u></p> <p>As MOANOM</p> <p>Please give details in text variable 90 SP_FAANOM</p> | As MOANOM |
| 91 | SP_FAANOM | <u>SPECIFY TYPE OF ANOMALY AND DESCRIBE THE MALFORMATION</u> | Free text |

Sociodemographic (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|-----------------|---------------|---|---|
| 92 | MATEDU | <p><u>MATERNAL EDUCATION</u> Refer to International Standard Classification of Education 1997 for more information and Kunst et al (2001).</p> <p>Assign according to the highest level of education completed (or for full-time students, level in progress).</p> <p>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.</p> | <p>1 = Elementary and lower secondary 2 = Upper secondary 3 = Tertiary 9 = Not known</p> |
| 93 | SOCM | <p><u>SOCIOECONOMIC STATUS OF MOTHER</u> Current or last occupation.</p> <p>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.</p> <p>If code 8 ("other/student"), please specify in text in space for general comments (variable 95).</p> <p>For further information see Kunst et al (2001)*</p> | <p>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</p> |
| 94 | SOCF | <p><u>SOCIOECONOMIC STATUS OF FATHER</u> As SOCM.</p> | <p>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</p> |

Sociodemographic (core variables shaded blue)

| Variable Number | Variable Name | Explanation and Instructions | Code |
|-----------------|---------------|---|---|
| 95 | MIGRANT | <p><u>MIGRANT STATUS</u></p> <p>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.</p> <p>If code 4, give text details in the general comments section (variable 95).</p> | <p>1 = Mother migrated from outside EU during pregnancy</p> <p>2 = Mother migrated from outside EU during adult life (from age 18)</p> <p>3 = Mother not a migrant as defined in 1 or 2</p> <p>4 = Other (specify in text)</p> <p>9 = Not known</p> |

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 |
|-----------------|---------------|------------------------------------|-----------|
| 96 | GENREM | <u>GENERAL ADDITIONAL COMMENTS</u> | Free text |

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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 | <p>0 = Not a EUROCAT case. This includes cases with only minor malformations (see Chapter 3.2)</p> <p>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 a11-a108 below.</p> <p>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</p> <p>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.</p> |
| a11 to a108 | EUROCAT subgroups: (0 = No, 1 = Yes). Based on EUROCAT coding (Chapter 3.3 of this Guide) |

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 unit 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 Severe microcephaly Arhinencephaly / holoprosencephaly Eye Anophthalmos / microphthalmos Anophthalmos Congenital cataract Congenital glaucoma Ear, face and neck Anotia Congenital Heart Defects Severe CHD Common arterial truncus Double outlet right ventricle Transposition of great vessels Single ventricle VSD ASD AVSD Tetralogy of Fallot Tricuspid atresia and stenosis Ebstein's anomaly Pulmonary valve stenosis Pulmonary valve atresia Aortic valve atresia/stenosis Mitral valve anomalies Aortic atresia/interrupted aortic arch 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 malformation 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 <i>Bilateral</i> renal agenesis including Potter syndrome Multicystic renal dysplasia Congenital hydronephrosis Bladder exstrophy Posterior urethral valve and/or prune belly Genital Hypospadias Indeterminate sex Limb Limb reduction defects Club foot - talipes equinovarus Hip dislocation and/or dysplasia Polydactyly Syndactyly | | | | | | | | |

| | 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 |
| Other anomalies/syndromes Skeletal dysplasias Craniosynostosis Congenital constriction bands/amniotic band Situs inversus Conjoined twins Congenital skin disorders VATER/VACTERL Vascular disruption anomalies Laterality anomalies Teratogenic syndromes with malformations Fetal alcohol syndrome Valproate syndrome Maternal infections resulting in malformations Genetic syndromes + microdeletions 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.

Last update 27.10.2016

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** | | | | | |
| <p>*Please complete all / as many as possible of the age categories above</p> <p>** Are stillbirths included in the maternal age distribution? (please delete as appropriate) Yes/No</p> | | | | | |
| Monthly distribution: | | | | | |
| January | | | | | |
| February | | | | | |
| March | | | | | |
| April | | | | | |
| May | | | | | |
| June | | | | | |
| July | | | | | |
| August | | | | | |
| September | | | | | |
| October | | | | | |
| November | | | | | |
| December | | | | | |
| TOTAL* | | | | | |
| <p>*Are stillbirths included in the distribution by month? (please delete as appropriate) Yes/No</p> | | | | | |

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.

Characters to be avoided in the EDMP database

Do not use the ENTER key to separate information in any text field. Instead, use a full stop to separate information. If you use the ENTER key the information takes up more than one row which causes problems when it comes to importing/exporting files to Central Registry. Do not use inverted commas (" ") or semi-colons (;) in your data. These symbols cause fatal errors.

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 to 1280x1024. 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.

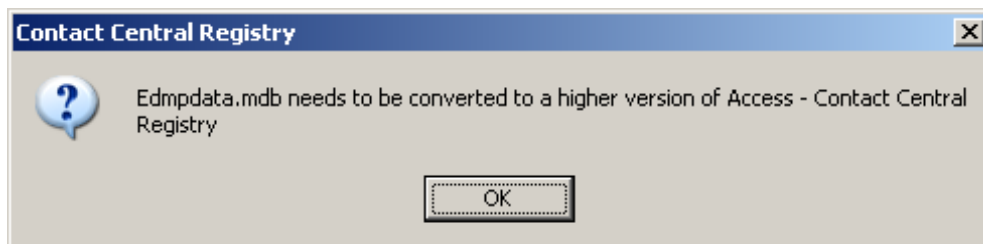
4.1.1 Existing Users

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.

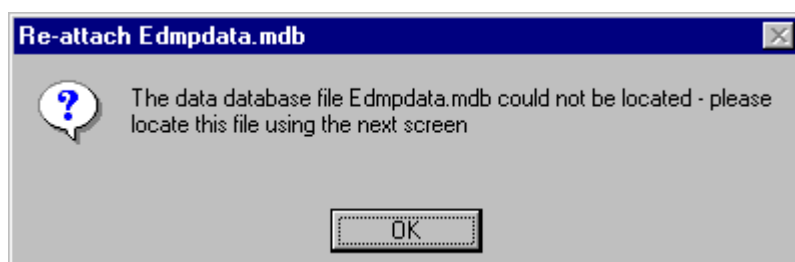
Access 2007 Security Settings

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.

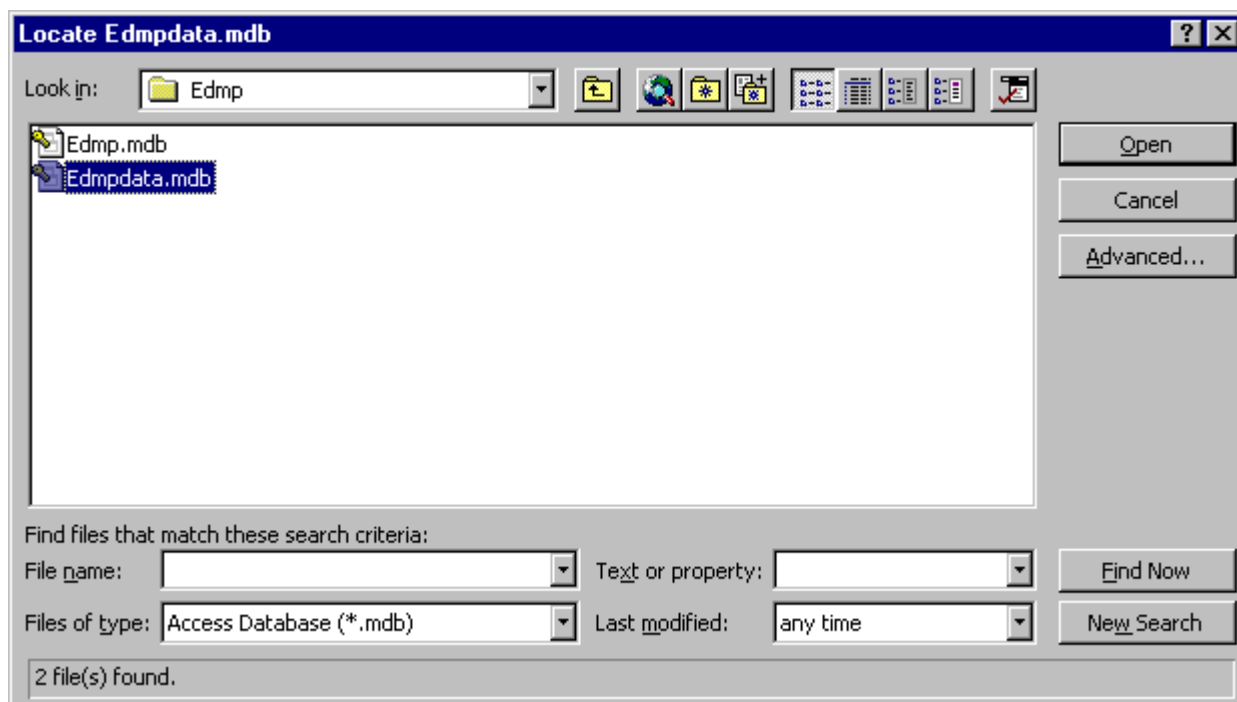
4.2 First time use

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.

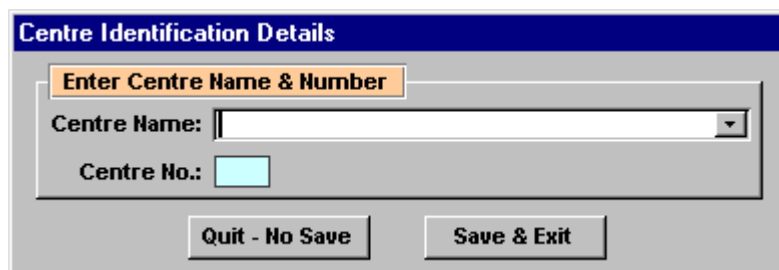
Each time you run the EDMP the first thing it does is to check to see that the program can locate the data file Edmpdata.mdb. If Edmpdata.mdb is not where it thinks it should be (i.e. after installation or if it has been moved to a different folder) then the following message will be displayed.



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 option that can be clicked on.

The new logical sections are:

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

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

6 Manage Cases

6.1 Add / Edit Cases

In EDMP you use the 'Add / Edit Cases' menu option to both add new cases and also edit existing cases. You use the 'Find case to edit' screen, shown below, to either select an existing case to edit or to add a new case by clicking on the 'Add New Case' button. Please note the separate buttons for pre 2005, 2005-2012 and 2013+ year of birth. The 2013+ option includes the new guide 1.4 variables.

When assigning ID numbers, it can be useful to prefix the unique local identifier number with the year of birth as this avoids duplication of ID numbers from one year to the next. For example cases born in 2009 could have ID numbers in the format 2009001, 2009002

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.

Find Case To Edit

Enter Selection Criteria

Local ID:

Infant Date of Birth:

Year of Birth:

Place of Birth:

Type of Birth:

Duplicate Status:

Data Validation Status:

Unknown: ☐ Not Entered: ☐

Not Entered: ☐

Not Entered: ☐

Add New Case (pre 2005)

Add New Case (2005-2012)

Add New Case (2013+)

List

Clear

Finish

Select Case by Clicking on Row

| Local ID | Infant DoB | Year of Birth | Place of Birth | Type of birth | Duplicate status |
|----------|------------|---------------|----------------|-------------------|------------------|
| 1234-67 | 20/06/96 | 1996 | 34 | Livebirth | Checked |
| 9090-87 | 20/06/96 | 1996 | Not Entered | Stillbirth | Checked |
| 1369-01 | 20/06/96 | 1996 | 1 | Termination of Pr | Checked |

Cases Found: 3

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+)

| | | | | |
|----------------------------------|-------------------------------------|--|--------------------------------------|--------------------------------------|
| Centre No.: 99 | NumEuro: 0004 | Add Case (2013+) | | Duplicate status: Not checked |
| Core Data Only | | Local ID No.: | DVS: Core data incomplete or invalid | |
| Date of Birth: | Year: | DOB / Year not known: <input type="checkbox"/> | | |
| Sex: | | | | |
| No. of babies delivered: | | | No. malformed (in multiple set): | |
| Type of birth: | | | | |
| Civil registration status: | View Anomaly Sub Group(s) | | | |
| Birth weight (g): | Confirmed: <input type="checkbox"/> | Date of birth of mother: | Year: | Confirmed: <input type="checkbox"/> |
| Length of gestation (weeks): | Confirmed: <input type="checkbox"/> | Age of mother at delivery: | | |
| Survival beyond one week of age: | | | | |
| When discovered: | | | | |
| Gestational age at discovery: | | | | |
| First surgical procedure: | | | | |
| Syndrome: | | | | |
| Malformation 1: | | | | |
| Malformation 2: | | | | |
| Malformation 3: | | | | |
| Malformation 4: | | | | |
| Malformation 5: | | | | |
| Malformation 6: | | | | |
| Malformation 7: | | | | |
| Malformation 8: | | | | |
| Quit - No Save | | Save & Exit | Duplicate Check | Validation |

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

Centre No.: 99 NumEuro: 0004 Add Case (2013+) Duplicate status: Not checked
DVS: Core data incomplete or invalid, non-core data valid or invalid

Baby & Mother Malformations Exposure Family History & Sociodemographic Local Vars(1) Local Vars(2) Extra Vars(1) Extra Drugs

Local ID No.:

Date of Birth (dd/mm/yy): Year: DOB / Year Unknown: ☐

Sex:

No. of babies delivered:

Specify twin type of birth, like or unlike sex, zygosity:

No. of malformed (in multiple set):

Type of birth:

Civil registration status:

Birth weight (g): Confirmed: ☐

Length of gestation (weeks): Confirmed: ☐

Survival beyond one week of age:

Date of death (dd/mm/yy): Year:

Date of birth of mother (dd/mm/yy): Year: Confirmed: ☐

Age of mother at delivery:

BMI (body mass index): Calculate BMI (& Codes)

Mother's residence code:

Total no. of previous pregnancies:

When discovered:

Condition at discovery:

Gestational age at discovery:

First positive prenatal test:

Specify 'other' prenatal test:

Karyotype of infant / fetus:

Specify karyotype:

Genetic test:

Specify genetic test:

Post mortem examination:

First surgical procedure:

Quit - No Save Save & Exit Duplicate Check Validation

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

Microsoft Access - [Case Details] Centre No.: 30 NumEuro: 0256 Add Case Duplicate status: Not checked
DVS: Core data incomplete or invalid, non-core data valid or invalid

Local ID No.:

Date of Birth (dd/mm/yy): Year: DOB / Year Unknown: ☐

Sex:

No. of babies delivered:

Birth order (in multiple set):

No. of malformed (in multiple set):

Type of birth:

Birth weight (g): Confirmed: ☐

Length of gestation (weeks): Confirmed: ☐

Date of birth of mother (dd/mm/yy): Year: Confirmed: ☐

Age of mother at delivery:

Syndrome:

Malformation 1:

Malformation 2:

Quit - No Save Save & Exit Duplicate Check Validation Sub Group(s)

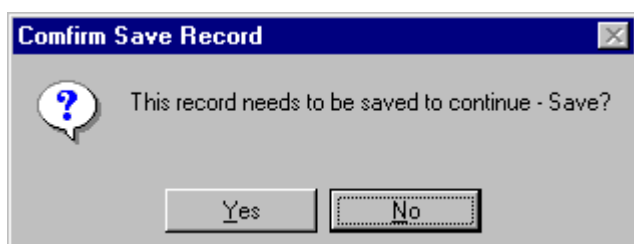
Local ID FLTR NUM

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.

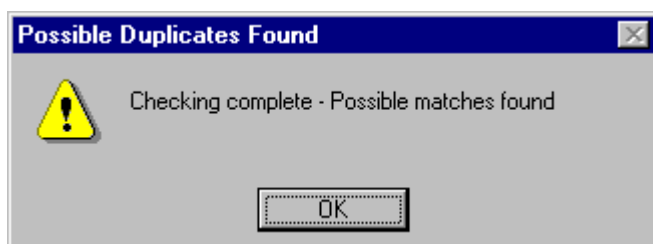
All the data entry screens offer the duplicate checking and validation buttons described below.

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.

Duplicates

Duplicate Case Check

Current Case Details

Local ID: 10028

Sex: Female

Num euro: 0028

Birth Weight: 2977

Date of Birth: 14/02/01

Maternal Age: 23

Possible Case Matches: 1

| Local ID | Num euro | Date of Birth | Sex | Birth weight | Age of mother at delivery |
|----------|----------|---------------|--------|--------------|---------------------------|
| 10295 | 0295 | 14/02/01 | Female | 1941 | 23 |

Not A Duplicate

Duplicate Entry

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:

Confirm Checked For Duplicates

?

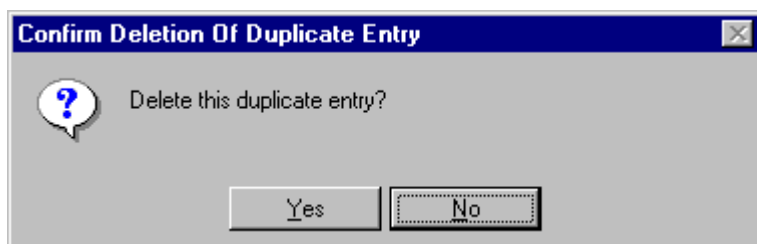
Mark case as checked for duplicates - Sure?

Yes

No

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.

Microsoft Access - [Verification]

Data Validation Status

| | |
|---|--|
| <p>Core Data Validation Errors: Click on row to edit data</p> <p>Infant sex not entered Max birthweight <= 3500g for gestational age 32 to 35 weeks Syndrome / malformation not entered</p> | <p>Non-Core Data Validation Errors: Click on row to edit data</p> |
|---|--|

Core Error Count: **3** BC FC

Non-Core Error Count: **0** BC FC

| | |
|--|---|
| <p>Confirm Extreme Values: Click on row to edit data</p> <p>Confirm birth weight greater than 6000g</p> | <p>Print Core Errors</p> <p>Print Non-Core Errors</p> <p>Print Core & Non-core Errors</p> |
|--|---|

BC FC

Close

Form View

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.

Find Case To Delete

Enter Selection Criteria

Local ID:

Infant Date of Birth:

Year of Birth: Unknown: ☐ Not Entered: ☐

Place of Birth: Not Entered: ☐

Infant Sex: Not Entered: ☐

Duplicate Status:

Data Validation Status:

List **Clear** **Close**

Select Cases by Clicking on Rows **Cases Matched: 234**

| Local ID | Infant DoB | Year of Birth | Place of Birth | Sex | Duplicate status |
|----------|------------|---------------|----------------|-------------|------------------|
| H23-345 | 12/03/01 | 2001 | w345 | Female | Checked |
| H23-890 | 12/03/01 | 2001 | Not Entered | Female | Checked |
| 10022 | 11/01/01 | 2001 | 99999 | Male | Possible matches |
| 10023 | 04/02/01 | 2001 | 99999 | Male | Possible matches |
| 10025 | 02/02/01 | 2001 | 99999 | Female | Possible matches |
| 10026 | 05/02/01 | 2001 | 99999 | Not Entered | Checked |
| 10028 | 14/02/01 | 2001 | 99999 | Female | Possible matches |
| 10029 | 12/02/01 | 2001 | 99999 | Male | Possible matches |
| 10030 | 03/01/01 | 2001 | 99999 | Female | Possible matches |
| 10031 | 29/01/01 | 2001 | 99999 | Male | Possible matches |
| 10033 | 21/01/01 | 2001 | 99999 | Female | Possible matches |
| 10035 | 04/01/01 | 2001 | 99999 | Male | Possible matches |
| 10040 | 31/01/01 | 2001 | 99999 | Female | Possible matches |
| 10042 | 18/01/01 | 2001 | 99999 | Female | Possible matches |
| 10046 | 11/02/01 | 2001 | 99999 | Female | Possible matches |
| 10049 | 30/01/01 | 2001 | 99999 | Female | Possible matches |

Select All **Clear Selection** **Delete Selected Cases**

Form View NUM

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.

Surveillance Selection Criteria

Select Registry:

Select Location (if applicable):

Anomaly Selection

☐ All anomaly subgroups

☐ Selected subgroup

Select anomaly subgroup:

Select user defined subgroup:

☒ Cases associated with chromosomal anomaly excluded (except from chromosomal subgroups)

Cluster & Trend

| | Year | Month |
|-------------------------|----------------------|----------------------|
| Start date: | <input type="text"/> | <input type="text"/> |
| End date: | <input type="text"/> | <input type="text"/> |
| End date of conception: | <input type="text"/> | <input type="text"/> |

Excel Spreadsheet location:

Long Term Trend (minimum 6 years)

Start year:

End year:

Excel Spreadsheet location:

Buttons:

- Generate cluster & trend output
- View additional cluster details
- View additional trend details
- Generate trend output
- View additional trend details
- Return To Statistical Surveillance Menu

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 screenshot shows a software window titled "nonmatching : Form". Inside, there is a section titled "Unmatched Import Variables". This section contains two columns of variables for selection.

| Unmatched - click to select | Available variables - click to select |
|-----------------------------|---------------------------------------|
| dfyear | byear |
| Lookup text 1 t100 | bunknown |
| Lookup text 3 t100 | ddyear |
| integer 11 1-5 | dsyear |
| memo 21 memo | dayear |
| date 32 | dfyear |
| tot_malf | ex1 |
| tot_minor | ex11 |
| other_in_chapter | ex21 |
| out_of_chapter | ex32 |
| birth_type | |
| casestatus | |
| erec | |
| byear | |
| bunknown | |
| ddyear | |
| dsyear | |
| dayear | |

At the bottom of the window, there are two buttons: "Match" and "Finish".

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.

Export Cases To Central Registry By Year

Please Select Year(s) of Birth to Export Cases By

Infant Year Of Birth
 Select years by clicking on row

1996
 1999
 2000
 2005
 2006
 2007
 2008
 2009
 2010
 2011
 2012
 2013

You must select pre 2005, 2005-2012 and 2013+ years separately

Select All pre 2005
 Select All 2005-2012
 Select All 2013+
 Clear Selection

Exporting: Core & Non-Core Data

Finish Export Cases

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:

Microsoft Access - [Denominators]

Add / Edit Denominators

Centre Number: 30 Centre Name: Basque Country (E) Birth Year: 2001

Live births: 17647 Still births: 71 Total Births: 17718

Maternal Age

| | Not known | < 20 | 20-24 | 25-29 | 30-34 | 35-39 | 40-44 | 45+ | Total | 35+ | 40+ |
|-----------|-----------|------|-------|-------|-------|-------|-------|-----|-------|------|-----|
| Observed | | 141 | 671 | 3127 | 8097 | 4972 | 683 | 27 | 17718 | 5682 | 710 |
| Estimated | | 141 | 671 | 3127 | 8097 | 4972 | 683 | 27 | | 5682 | 710 |

Total Births by Month

| | Jan | Feb | Mar | Apr | May | Jun | Jul | Aug | Sep | Oct | Nov | Dec |
|-----------|------|------|------|------|------|------|------|------|------|------|------|------|
| Observed | 1433 | 1325 | 1514 | 1475 | 1657 | 1385 | 1537 | 1540 | 1399 | 1532 | 1510 | 1411 |
| Estimated | 1433 | 1325 | 1514 | 1475 | 1657 | 1385 | 1537 | 1540 | 1399 | 1532 | 1510 | 1411 |

Observed Total: 17718

Notes:
Figures rec'd by email 11/03/03

Date total last updated:
Date Live/Stillbirth last updated:
Date age distribution last updated:
Date month distribution last updated:

Been Completed: ☐ Completed by: Imported on 14/05/03
Completed on:

Save & Exit **Delete** **Finish**

N.B. Estimated denominators are estimated assuming that births with unknown maternal age or month have the same maternal age or month distribution as those with known maternal age or month.

Live births NUM

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.

The screenshot shows a Microsoft Access window titled "Microsoft Access - [Alter password]". The main form is titled "Alter User Details". It is divided into two panes. The left pane, "User Details", contains text boxes for "Name of user:" (containing "Administrator"), "Staff code:" (containing "ADM"), "Username:" (containing "m"), and "Password:" (containing "m"). Below these are two boxes labeled "Background Colour" and "Highlight Colour" with the instruction "Click on box to change colour(s)". The right pane, "Security & Access Control", contains a section "Allow Access To" with a list of permissions, each with a checked checkbox: "Add New Case", "Edit Existing Case", "View Case", "Print Case", "Import Data", "Export Data", "Reports", "Error Log", and "System Menu". At the bottom of the form are three buttons: "Quit - No Save", "Save & Exit", and "Delete". The status bar at the bottom shows "Name of user", "FLTR", and "NUM".

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

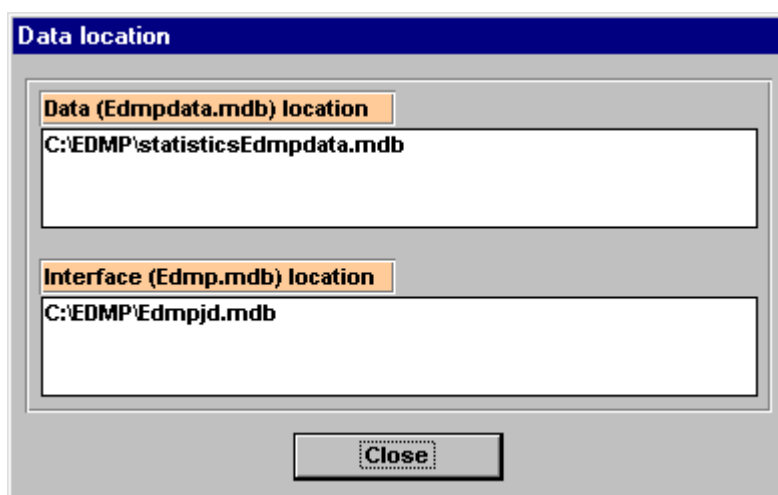
specify which parts of the program are accessible to each user. It is important that at least one user has access to the System Menu!

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.

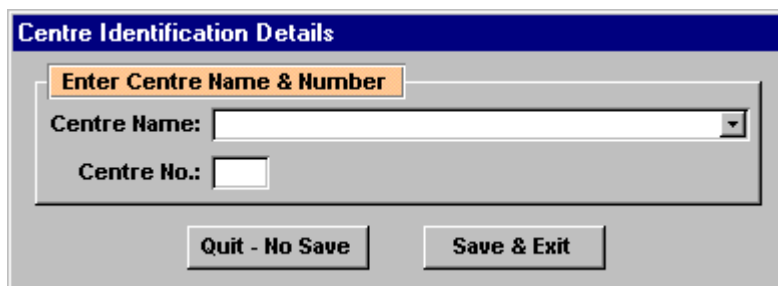
12.3 Data location

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.

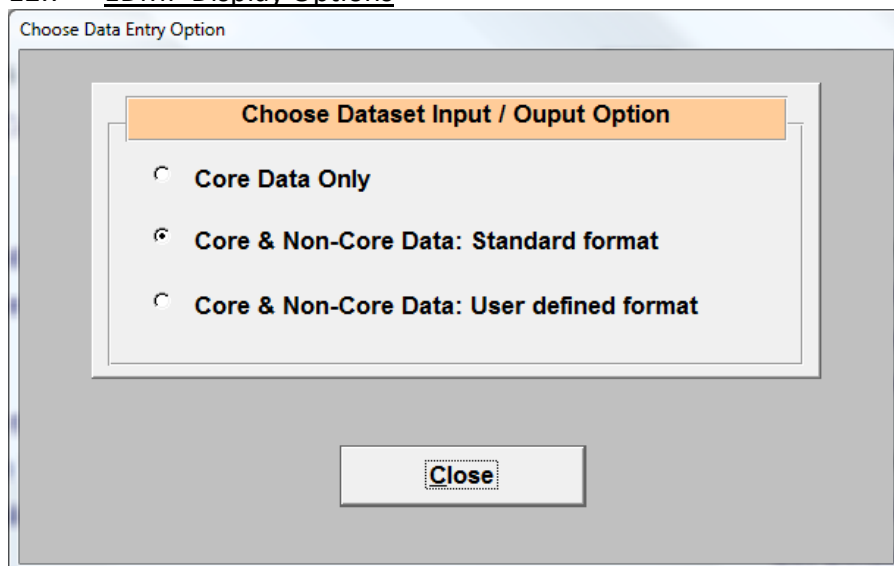
12.6.3 Change Spare Variable Names

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_TWAIN 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**
- 3.2 Minor Anomalies for Exclusion**
- 3.3 EUROCAT Subgroups of Congenital Anomalies**
- 3.4 Multiple Congenital Anomaly Algorithm**
- 3.5 Detailed Congenital Anomaly Coding Guidelines**
- 3.6 EUROCAT Description of the Congenital Anomaly Subgroups**

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 and other conditions for Exclusion

For EUROCAT use from 2005

Cases with only minor anomalies and unspecified anomalies for exclusion 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, it will be excluded by computer if the minor anomalies have specific codes which allow recognition. Some minor anomalies 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.

“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 preterm birth. In addition, we exclude poorly specified conditions and recommend that for any such cases more specific information be sought from medical records. Prenatal diagnosis of anomalies of uncertain severity should be confirmed after birth.

For allocation of cases to EUROCAT subgroups, only major malformations will be considered (codes for minor anomalies will be excluded). If a registry use major ICD10 codes to describe a minor anomaly or a syndrome feature, the prevalence will be higher than the true prevalence in the registry area and data will be less comparable to other regions. Use of major ICD10 codes for minor anomalies will also have a negative impact on the classification of cases by the multiple flowchart and the surveillance of multiple congenital anomalies.

Please note that the list is not exhaustive and not all dysmorphic features are mentioned. For some features commonly used by the registries, a code outside the Q-chapter is given.

Anomalies marked in red: added in 2018. The ICD10 codes are not yet implemented in the EUROCAT database (ECD and EDMP)

| | Specified ICD10-BPA – if present |
|---|---|
| Head | |
| Aberrant scalp hair patterning | |
| Bony occipital spur | |
| Brachycephaly | |
| Compression facies | Q671 |
| Depressions in skull, lacunar skull, temporal flattening | Q6740 |
| Dolichocephaly | Q672 |
| Dysmorphic face | Q189 |
| Broad, prominent forehead | |
| Coarse facies | |

| | |
|--|-------|
| Flattened face | |
| Frontal bossing / wide forehead | |
| Mid face hypoplasia | |
| Pointed facies | |
| Round head shape | |
| Sloping forehead | |
| Facial asymmetry | Q670 |
| Flat occiput | |
| Macrocephalus | Q753 |
| Metopic ridge, high metopic suture | |
| Other congenital deformities of skull, face and jaw (including all types of abnormally shaped skull without synostosis) | Q674 |
| Plagiocephaly – head/skull asymmetry | Q673 |
| Third fontanelle | |
| Skull, late closure | |
| Wormian bones | |
| Eyes | |
| Anisocoria | |
| Blue sclera | Q135 |
| Congenital ectropion | Q101 |
| Congenital entropion | Q102 |
| Crocodile tears | Q0782 |
| Dacryocystocele | H046 |
| Downward slanting palpebral fissures | Q103 |
| Dystopia canthorum | Q189 |
| Epicanthic folds | Q189 |
| Epicanthus inversus | Q189 |
| Exophthalmos | H052 |
| Hypertelorism | Q752 |
| Hypotelorism | Q189 |
| Other congenital malformations of eyelid | Q103 |
| Oval shaped pupils | |
| Prominent/protruding eyes | H052 |
| Short palpebral fissures | Q189 |
| Stenosis or stricture of lacrimal duct | Q105 |
| Synophrys | Q1880 |
| Upward slanting palpebral fissures | Q103 |
| Ears | |
| Absent tragus | |
| Accessory auricle, preauricular appendage, tag or lobule | Q170 |
| Asymmetric size | Q173 |
| Auricular pit | |
| Bat ear, prominent, protuberant ear | Q175 |
| Congenital absence of ear lobe | |
| Darwin's tubercle | |
| Double lobule | Q170 |
| Lack of helical fold | Q173 |

| | |
|--|-------------|
| Low set ears | Q174 |
| Macrotia | Q171 |
| Microtia/ small ears | Q172 |
| Narrow external auditory meatus | |
| Posterior angulation | Q173 |
| Primitive shape | Q173 |
| Pointed ear, Vulcan ear, simple ear | Q173 |
| Unspecified and minor malformation of ear | Q179 |
| Nose | |
| Anteverted nares | Q189 |
| Bifid tip of nose | Q189 |
| Broad nasal root, anomaly of nasal root | Q189 |
| Depressed nasal bridge | Q189 |
| Deviation of nasal septum | Q6741 |
| Dysmorphic nose | Q189 |
| Flat nose | Q189 |
| Flattened nasal bridge | Q189 |
| Notched alas | |
| Pinched nose | Q189 |
| Prominent nasal bridge | Q189 |
| Saddle nose | Q189 |
| Small/ hypoplastic nares | Q189 |
| Small pointed nose | Q189 |
| Underdeveloped nasal bones | Q189 |
| Upturned nose | Q189 |
| Wide nasal root | Q189 |
| Oral regions | |
| Aberrant frenula | |
| Absent /hypoplasia depressor anguli oris (asymmetric crying face) | |
| Alveolar crest | |
| Anomalies of philtrum, elongated philtrum | Q189 |
| Bifid uvula / cleft uvula | Q357 |
| Borderline small mandible/ minor micrognathia | |
| Disturbances in tooth eruption | |
| Enamel hypoplasia | |
| Glossoptosis | |
| High arched palate | Q3850 |
| Macrocheilia | Q186 |
| Macroglossia / hemi-hypertrophy of tongue | Q382 |
| Macrostomia | Q184 |
| Malformed teeth | |
| Microcheilia | Q187 |
| Microglossia | |
| Microstomia | Q185 |
| Mid-oral tongue position | |
| Neonatal teeth | |
| Prominent jaw | Q189 |

| | |
|--|-------------------|
| Ranula | |
| Retrognathia/ receding chin | Q674 |
| Short philtrum | Q189 |
| Thin lips | Q189 |
| Tongue tie or cyst of tongue | Q381 |
| Neck | |
| Broad neck | Q189 |
| Congenital malformation of face and neck, unspecified | Q189 |
| Congenital thymic hypoplasia | |
| Mild webbed neck | |
| Other branchial cleft malformations | Q182 |
| Preauricular sinus or cyst | Q181 |
| Short neck | Q189 |
| Sinus, fistula or cyst of branchial cleft | Q180 |
| Thymus involution | |
| Thyreoglossal cyst | |
| Torticollis | Q680 |
| Hands | |
| Accessory carpal bones | Q7400 |
| Arachnodactyly | |
| Clinodactyly (5 th finger) | Q6810 |
| Duplication of thumbnail | |
| Enlarged or hypertrophic nails | Q845 |
| Other congenital malformations of nails | Q846 |
| Overlapping fingers | |
| Short fingers (4. 5. th finger) | |
| Single/abnormal palmar crease | Q8280 |
| Small fingers | |
| Subluxation of phalangeal bones | |
| Unusual dermatoglyphics | |
| Feet -Limb | |
| Bulbous toes | |
| Clicking hip, subluxation or unstable hip | Q653-Q656 |
| Hip dysplasia and other specified/unspecified hip anomalies | Q658, Q659 |
| Clubfoot of postural origin - other cong deformities of feet | Q668 |
| Congenital deformity of feet, unspecified | Q669 |
| Congenital pes planus | Q665 |
| Enlarged or hypertrophic nails | Q845 |
| Gap between toes (1st-2nd) | |
| Hallux varus – other congenital varus deformities of feet | Q663 |
| Metatarsus varus – other congenital valgus deformities of feet | Q666 |
| Metatarsus varus or metatarsus adductus | Q662 |
| Overlapping toes | |
| Pes cavus | Q667 |
| Prominent calcaneus | |
| Recessed toes (4th, 5th) | |
| Rocker bottom feet | Q6680 |

| | |
|---|--------------|
| Short great toe | |
| Syndactyly (2nd-3rd toes) | |
| Talipes or pes calcaneovalgus | Q664 |
| Talipes calcaneovarus | Q661 |
| Skin | |
| Accessory nipples | Q833 |
| Accessory skin tags | Q8281 |
| Angioma | |
| Cafe-au-lait spot | |
| Depigmented spot | |
| Epibulbar dermoid | |
| Hemangioma if no treatment is required | |
| Heterochromia of hair | |
| Hypoplasia of toe nails | Q846 |
| Lymphangioma if no treatment is required | |
| Mongoloid spot (whites) | Q8252 |
| Neavus flammeus | Q8250 |
| Persistent lanugo | |
| Pigmented naevus – congenital non-neoplastic naevus | Q825 |
| Strawberry naevus | Q8251 |
| Unusual placement of nipples/ wide spaced nipples | |
| Skeletal | |
| Abortive 12 th rib | |
| Absence of rib/ hypoplastic rib | Q7660 |
| Accessory rib | Q7662 |
| Bipartite vertebrae | |
| Bifid ribs | |
| Cervical rib | Q765 |
| Congenital bowing of femur | Q683 |
| Congenital bowing of fibula and tibia | Q684 |
| Congenital bowing of long bones of leg, unspecified | Q685 |
| Congenital bowing of upper limb | |
| Congenital deformity of spine | Q675 |
| Congenital lordosis, postural | Q7643 |
| Coronal clefts of vertebrae, incomplete | |
| Cubitus valgus | |
| Depressed sternum | |
| Duplication of ribs | |
| Fused rib, single | |
| Genu recurvatum | Q6821 |
| Genua valgum | |
| Genua varum | |
| No ossification of os coccyx | |
| Ovoid configuration of vertebrae | |
| Prominent sternum | |
| Sacral dimple | L059 |
| Shieldlike chest, other congenital deformities of chest | Q678 |

| | |
|--|-----------------------|
| Spina bifida occulta | Q760 |
| Sternum bifidum | Q7671 |
| Brain | |
| Anomalies of septum pellucidum | |
| Arachnoid cysts | |
| Asymmetric ventricles, normal size | |
| Banana shaped cerebellum | |
| Cerebellar hypoplasia, mild | |
| Cerebral atrophy | |
| Choroid plexus cysts | |
| Cyst of septum pellucidum | |
| Enlarged cisterna magna, isolated | |
| Jaw-winking syndrome, Marcus Gunn's syndrome | Q0780 |
| Periventricular leukomalacia | |
| Single congenital cerebral cyst | Q0461 |
| Thin or hypoplastic corpus callosum | |
| Ventriculomegaly < 15 mm | |
| Cardiovascular | |
| Absence or hypoplasia of umbilical artery, single umbilical artery | Q270 |
| Absence of vena cava superior | |
| Functional or unspecified cardiac murmur | R011 |
| Cardiomegaly | I517 |
| Cardiomyopathy | I429 |
| Deviation of the heart axis | |
| Patent ductus arteriosus if GA < 37 weeks | Q250 if GA < 37 weeks |
| Patent or persistent foramen ovale | Q2111 |
| Peripheral pulmonary artery stenosis | Q256 if GA < 37 weeks |
| Persistent left superior vena cava | Q261 |
| Persistent right aortic arch | Q2541 |
| Persistent right umbilical vein | |
| Pulmonary | |
| Accessory lobe of lung | Q331 |
| Azygos lobe of lung | Q3310 |
| Bronchomalacia | Q322 |
| Congenital laryngeal stridor | Q314 |
| Single cyst of the lung | Q3300 |
| Hyperplasia of thymus | |
| Laryngomalacia | Q3140 |
| Pleural effusion | |
| Pulmonary hypoplasia, secondary | |
| Relaxation of diaphragm | |
| Thymus involution | |
| Tracheomalacia | Q320 |
| Vocal cord palsy | |
| Gastro-intestinal | |
| Abdominal cyst not needing surgery | |
| Accessory spleen | |

| | |
|---|----------------------------|
| Anterior anus without surgery | |
| Choledochal cyst | Q444 |
| Congenital adrenal hypoplasia | Q8911 |
| Congenital cholestasis | |
| Congenital mesenteric cyst | Q4583 |
| Cyst of spleen | |
| Diastasis recti | |
| Dilatation of intestine | |
| Functional gastro-intestinal disorders | Q4021, Q4320, Q4381, Q4382 |
| Hepatomegaly | R160 |
| Hiatus hernia | Q401 |
| Inguinal hernia | K409 |
| Liver cyst | |
| Meckel's diverticulum | Q430 |
| Plica of anus | |
| Pyloric stenosis | Q400 |
| Splenomegaly | R161 |
| Transient choledochal cyst | |
| Umbilical hernia | |
| Renal | |
| Enlarged/thickened bladder | |
| Hydronephrosis with a pelvis dilatation less than 10 mm | |
| Hyperplastic and giant kidney | Q633 |
| Single renal cyst | Q610 |
| Vesico-ureteral-renal reflux | Q627 |
| External genitals | |
| Bifid scrotum | Q5521 |
| Buried penis | |
| Congenital chordee | Q544 |
| Congenital adrenogenital disorders | E250 |
| Congenital malformation of vulva | Q527 |
| Congenital torsion of ovary | Q502 |
| Curvature of penis | |
| Cysts of vulva | |
| Deficient or hooded foreskin/prepuce | N47 |
| Developmental ovarian cyst(s) | Q501, Q5010, Q5011 |
| Embryonic cyst of broad ligament | Q505 |
| Enlarged clitoris | |
| Foreskin tethered to the scrotum | N47 |
| Fusion of labia | Q525 |
| Hydrocele of testis | P835 |
| Hymen imperforate | Q523 |
| Hypertrophy of hymen | |
| Hypoplasia of penis/micropenis | |
| Phimosis | N47 |
| Prominent labia minora | |
| Retractile testis | Q5520 |

| | |
|---|------|
| Seminal vesicle cyst | |
| Testicular torsion | N44 |
| Transient ovarian cyst | |
| Undescended testicle | Q53 |
| Unspecified ectopic testis | Q530 |
| Vaginal skin tag | |
| Other | |
| Congenital malformation, unspecified | Q899 |
| Chromosomal | |
| Balanced chromosomal rearrangements | Q95 |
| Balanced translocations or inversions in normal individuals | |
| Balanced autosomal rearrangement in abnormal individual | Q952 |
| Individuals with marker heterochromatin | |
| Individuals with autosomal fragile site | |

“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

Version November 2018

3.3 EUROCAT Subgroups of Congenital Anomalies (Version 2014; implemented in EDMP December 2014, used for website prevalence tables from December 2014).

Guide 1.4 is used to code all cases of congenital anomaly and uses ICD10-BPA codes only. The ICD9-BPA codes and the minor anomalies pre-2005 are provided for retrospectively making subgroups pre-2005 when this coding system was 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 1.4, section 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 | | Q0461, 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 hydranencephaly 74232. Exclude association with NTD subgroup | | | al7 |
| Severe 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 PDA with GA <37 weeks Exclude peripheral pulmonary artery stenosis with GA < 37 weeks | Q2111, Q250 if GA <37 weeks, Q2541, Q256 if GA<37 weeks, Q261 | Q250, 7470 if GA <37 weeks ** | al17 |
| Severe CHD | Q200, Q201, Q203, Q204, Q212, Q213, Q220, Q224, Q225, Q226, Q230, Q232, Q233, Q234, Q251, Q252, Q262 | 74500, 74510, 7452, 7453, 7456, 7461, 7462, 74600, 7463, 7465, 7466, 7467, 7471, 74720, 74742 | ICD9-BPA has no code for HRH and double outlet right ventricle | | | al97 |
| Common arterial truncus | Q200 | 74500 | | | | al18 |
| Double outlet right ventricle | Q201 | No code | | | | al109 |
| 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 |
| Tricuspid 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 |
| Mitral valve anomalies | Q232, Q233 | 7465, 7466 | | | | al110 |
| Hypoplastic left heart | Q234 | 7467 | | | | al30 |
| Hypoplastic right heart | Q226 | No code | | | | al31 |
| Coarctation of aorta | Q251 | 7471 | | | | al32 |
| Aortic atresia / interrupted aortic arch | Q252 | 74720 | | | | al111 |
| Total anomalous pulm venous return | Q262 | 74742 | | | | al33 |
| PDA as only CHD in term infants (GA +37 weeks) | Q250 | 7470 | Livebirths only | | | al100 |
| Respiratory | Q300, Q32-Q34 | 7480, 7484, 74850, 74852, 74858, 7486, 7488 | Exclude Q336 | Q320, Q331 | Q309, 74819 | al34 |
| Choanal atresia | Q300 | 7480 | | | | al35 |
| Cystic adenomatous malf of lung | Q3380 | No code | | | | al36 |

| EUROCAT Subgroups | ICD10-BPA | ICD9-BPA | Comments | Excluded minor anomalies post-2005 | Excluded minor anomalies pre-2005 | Subgroup binary variable number (al) |
|---|------------------|---------------------|--|---|-----------------------------------|--------------------------------------|
| 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 |
| 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 | 75261, 753, 75672 | | Q610, Q627, Q633 | | al52 |
| Bilateral renal agenesis including Potter syndrome | Q601, Q606 | 75300 | Exclude unilateral | | | al53 |
| Multicystic renal dysplasia | Q6140, Q6141 | 75316 | | | | al54 |
| Congenital hydronephrosis | Q620 | 75320 | | | | al55 |
| Bladder exstrophy and / or epispadia | Q640, Q641 | 75261, 7535 | | | | al56 |
| Posterior urethral valve and / or prune belly | Q6420, Q794 | 75360, 75672 | | | | al57 |

| EUROCAT Subgroups | ICD10-BPA | ICD9-BPA | Comments | Excluded minor anomalies post-2005 | Excluded minor anomalies pre-2005 | Subgroup binary variable number (al) |
|--|--|------------------------------------|----------|---|--|--------------------------------------|
| Genital | Q50-Q52, Q54-Q56 | 7520-7524, 75260, 75262, 7527-7529 | | Q523, Q525, Q527, Q5520, Q5521 | Q540, 75260# | al58 |
| Hypospadias | Q54 | 75260 | | | Q540, 75260 | al59 |
| Indeterminate sex | Q56 | 7527 | | | | al60 |
| Limb | Q65-Q74 | 7543-7548, 755 | | Q653-Q656, Q662-Q669, Q670-Q678, Q680, Q6810, Q6821, Q683-Q685, Q7400 | 75432, 75452, 75460, 75473, 75481, 75560 | al61 |
| Limb reduction defects | Q71-Q73 | 7552-7554 | | | | al62 |
| Club foot – talipes equinovarus | Q660 | 75450 | | | | al66 |
| Hip dislocation and / or dysplasia | Q650-Q652, Q6580, Q6581 | 75430 | | | | al67 |
| Polydactyly | Q69 | 7550 | | | | al68 |
| Syndactyly | Q70 | 7551 | | | | al69 |
| Other anomalies / syndromes | | | | | | |
| Skeletal dysplasias | Q7402, Q77, Q7800, Q782-Q788, | 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 |
| VATER/VACTERL | Q8726 | 759895 | | | | al112 |
| Vascular disruption anomalies | Q0435, Q411, Q412, Q418, Q710, Q712, Q713, Q720, Q722, Q723, Q730, Q793, Q795, Q7980, Q7982, Q8706 | No code | | | | al113 |
| Laterality anomalies | Q206, Q240, Q3381, Q890, Q893 | No code | | | | al114 |
| 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 |

| EUROCAT Subgroups | ICD10-BPA | ICD9-BPA | Comments | Excluded minor anomalies post-2005 | Excluded minor anomalies pre-2005 | Subgroup binary variable number (al) |
|------------------------------------|---|----------------------------------|--|------------------------------------|-----------------------------------|--------------------------------------|
| Genetic syndromes + microdeletions | Q4471, Q6190, Q7484, Q751, Q754, Q7581, Q87, Q936, D821 | 75581, 75601, 75604, 7598, 27910 | Exclude Associations and sequences Exclude Q8703, Q8704, Q8706, Q8708, Q8724, Q8726 Exclude 759801, 759844, 759895 | | | al105 |
| 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 Section 3.2 in Guide 1.4 for cases born post-2005. Cases with more than one anomaly are only counted once in the "All Anomalies" subgroup.

^ ICD10 code D1810 (ICD 9 code 2281) is the code for cystic hygroma

** 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 I) are 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.

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 1.4 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

Accepted non Q codes: D215, D821, D1810, P350, P351, P371

ICD/BPA9: 27910, 2281, 76076, 76280, 7710, 7711, 77121

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

ICD/BPA9: all 759 cases except 7584

Q90-Q93 excluding Q936

Q96-Q99,

- Transfer to group C

Step 2

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

ICD/BPA9: 7598, 27910, 7571, 7573, 75581 (Larsen syndrome), 75601 (Crouzon), 75604

(Mandibulofacial dysostosis)

No ICD9 subgroup for skeletal dysplasia

Excluding 759801, 759895, 759844

Q87 Excluding Q8703, Q8704, Q8706, Q8708, Q8724, Q8726,

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

- Transfer to group B

Step 3

Exclude all cases with a code for teratogenic syndrome code

No ICD9 subgroup for teratogenic syndromes, only the codes for infection 7710, 7711, 77121 and fetal alcohol syndrome 76076

Q86, P350, P351, P371

- Transfer to group T

Step 4

Exclude all cases with a heterogenous syndrome code

ICD9: 756110, 75680, 759611, 3568 (not exactly the same as ICD10)

Q761, Q7982, Q8581, Q8706

- Transfer to group M

Step 5

Exclude all cases with only NTD codes

ICD/BPA9: 740-741, 7420

Q00-Q01, Q05

- Transfer to group N

Step 6

Exclude all cases with codes only in cardiac chapter

ICD/BPA9: 745-746, 7470-7474

Q20-Q26

- Transfer to group A

Step 7

Exclude all cases with codes only in renal chapter

ICD/BPA9: 753, 756.72, 752.61

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

(for our own purpose, transfer also cases with only cleft codes – chapter Q35-Q37)

ICD/BPA9: 740-759 (transfer to groups like ICD10 – see codes for group N, A, and R earlier)

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 or D1810 or D215/icd9 2281

- Transfer to group I

Step 9

Exclude all cases with codes only in eye chapter

ICD/BPA9: 743

Q10-Q15

- Transfer to group I

Step 10

Exclude all cases with codes only with limb reduction defects

ICD/BPA9: 7552-7554

Q71-Q73

- Transfer to group I

Step 11

Exclude all cases with codes only for hypospadias

ICD/BPA: 75260

Q54

- Transfer to group I

Step 12

Exclude all cases with codes only for polydactyly

ICD/BPA9: 7550

Q69

- Transfer to group I

Step 13

Exclude all cases with codes only for reduction defects of brain

ICD/BPA9: 7422

Q04

- Transfer to group I

Step 14

Exclude all cases with codes only for hip anomalies

ICD/BPA9: 75430

Q65

- Transfer to group I

Step 15

Exclude all cases with codes only for syndactyly

ICD/BPA9: 7551

Q70

- Transfer to group I

Step 16

Exclude all cases with codes only for syndactyly + polydactyly

ICD/BPA9: 7550 and 7551

Q69 and Q70

- Transfer to group I

Step 17

Exclude all cases with codes only for small intestinal atresia

ICD/BPA9: 75111, 75112, 75119

Q41

- Transfer to group I

Step 18

Exclude all cases with codes only for facial clefts

ICD/BPA9 7490, 7491, 7492

Q35, Q36, Q37

- Transfer to group I

Step 19

Exclude all cases with the code for balanced chromosomal rearrangements (Q95 or 7584) and only one other Q-code

ICD/BPA9: 7584 (transfer to groups like ICD10 – see codes for group N, A, and R earlier)

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)

ICD/BPA9: outside 740-759

2281 and 76280 accepted as outside malformation chapter codes

Not beginning with Q

D1810 and D215 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:

ICD/BPA9 741 coded with 7545 and/or 7423

Q05 coded with Q66 and/or Q03

- Transfer to group N

Bilateral renal aplasia/dysplasia – lung hypoplasia - talipes:

ICD/BPA9 75300 coded with 74851 and/or 7545

Q601/Q606 coded with Q336 and/or Q66

- Transfer to group R

Omphalocele/gastroschisis – malrotation of gut – small intestinal atresia

ICD/BPA9 75670/75671 coded with 7514 and/or 7511

Q792/Q793 coded with Q433 and/or Q41

- Transfer to group I

Anal atresia - rectovaginal fistula

ICD/BPA9 7512 coded with 75242

Q42 coded with Q522

- Transfer to group I

Diaphragmatic hernia – lung hypoplasia

ICD/BPA9 75661 coded with 74851

Q790 coded with Q336

- Transfer to group I

Anencephalus - adrenal hypoplasia

ICD/BPA9 740 coded with 75911

Q00 coded with Q891

- Transfer to group N

Unspecified hydrocephalus - reduction defect of brain

ICD/BPA9 74239 coded with 7422

Q039 coded with Q04

- Transfer to group I

Unspecified hydrocephalus – Arnold-Chiari

no ICD/BPA9 code for Arnold-Chiari

Q039 coded with Q070

- Transfer to group I

NTD – Arnold Chiari

no ICD/BPA9 code for Arnold-Chiari

Q01 or Q05 coded with Q070

- Transfer to group N

Amniotic band sequence

ICD/BPA9: 76280

All cases with the code Q7980

- Transfer to group I

Caudal dysplasia sequence

no ICD/BPA9 code for caudal dysplasia sequence

All cases with the code Q8980

- Transfer to group I

Sirenomelia sequence

ICD/BPA9: All cases coded with 759844

All cases coded with Q8724

- Transfer to group I

Cyclops sequence

ICD/BPA9: All cases coded with 759801

All cases coded with Q8703

- Transfer to group I

Pierre Robin sequence

ICD/BPA9: All cases coded with 756030 as only code or with 7490, 7491, 7492

All cases coded with Q8708 as only code or with Q35-Q37

- Transfer to group I

Holoprosencephaly – median cleft lip

ICD/BPA9: All cases coded with 74226 and 74912

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)

Ester Garne

19 November 2014

3.5 Detailed Congenital Anomaly Coding Guidelines

Remember always to give as specified code as possible

Other specified congenital anomaly codes

(Q188, Q178 Q308, Q288, Q742, Q764, Q758, Q445, Q638 etc)

Use these codes only to code major anomalies not specifically mentioned in Q chapter. Describe anomaly in the text. Do not use these codes for minor anomalies listed in the list of minor anomalies for exclusion.

Coding Committee April 2018

Q00 Anencephaly and similar malformations

Q01 Encephalocele

Q02 Microcephaly

MICROCEPHALY

Report microcephaly if head circumference (occipito-frontal) is less than -3 SD for sex and GA. Add in written text the measurements and age at measurements. In case of maternal zika virus infection, use the code P358 for congenital viral infection in one of the malformation variables. Use local growth chart to confirm the diagnosis. Exclude secondary microcephaly (neonatal meningitis, birth asphyxia, extreme preterm birth)

Coding Committee June 2016

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 CORPUS CALLOSUM

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

Q043 OTHER REDUCTION DEFORMITIES OF BRAIN

Aicardi syndrome, Joubert syndrome, Miller-Dieker syndrome and Walker-Warburg syndrome:

Please code these genetic syndromes with the code Q878 and give the syndrome name in written text. Also give the code for the diagnosed cerebral anomaly in malf1 (Q043 for reduction deformity of brain, Q0433 for lissencephaly)

Coding Committee September 2018

Q0432 REDUCTION ANOMALIES OF CEREBELLUM

In livebirths this should be reported only if there is a significant reduction in the size of cerebellum.

Mild reduction of the size of cerebellum is considered a minor anomaly.

Coding Committee April 2018

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

Q048 OTHER SPECIFIED CONGENITAL MALFORMATIONS OF BRAIN

Congenital ventriculomegaly should be reported as hydrocephaly (Q03 subchapter) 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. If reported to EUROCAT, give written text description without a Q-code. Asymmetric ventricles and ventriculomegaly secondary to neonatal cerebral hemorrhage or meningitis should not be reported to EUROCAT

Coding Committee April 2018

Q048 OTHER SPECIFIED CONGENITAL MALFORMATIONS OF BRAIN

Colpocephaly is a congenital brain abnormality in which the occipital horns - the posterior or rear portion of the lateral ventricles (cavities) of the brain - are larger than normal because white matter in the posterior cerebrum has failed to develop or thicken. The term colpocephaly should not be used for asymmetric or mildly dilated ventricles, as these are minor anomalies.

Coding Committee April 2018

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 (see 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 lipomyelomeningocele 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

Q068 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

Q068 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

Q070 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 (see 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

Q183 WEBBING OF NECK/PTERYGIUM

Conditions leading to webbed neck and requiring surgery include a collapsed cystic hygroma and multiple pterygium syndrome for example. These must be coded as major anomaly Q183. Mild webbed neck should be regarded as a minor anomaly unless requiring surgery and coded Q189. Coding Committee April 2018

Q189 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

Q189 MILD WEBBED NECK

Mild webbed neck should be regarded as a minor anomaly unless requiring surgery and coded Q189. Conditions leading to webbing and requiring surgery include a collapsed cystic hygroma and multiple pterygium syndrome for example. These must be coded as major anomaly Q183.

Coding Committee April 2018

Q189 BIFID TIP OF NOSE

This is a minor anomaly often associated with chromosomal abnormalities. It can be coded as part of a description of dysmorphic features using Q189. The code Q302 should not be used.

Coding Committee April 2018

Q189 CONGENITAL MALFORMATION OF FACE AND NECK, UNSPECIFIED

Use this code for dysmorphic features/dysmorphic face and not Q759

Coding Committee April 2018

Q20 Congenital malformations of cardiac chambers and connections

Q201 DOUBLE OUTLET RIGHT VENTRICLE

For this anomaly, the aorta is overriding the ventricular septum with at least 50% of the size. The pulmonary artery arises from the right ventricle

Coding Committee December 2016

Q202 DOUBLE OUTLET LEFT VENTRICLE

For this anomaly the aorta and the pulmonary artery arises exclusively or predominantly from the left ventricle

Coding Committee December 2016

Q203 TRANSPOSITION OF GREAT ARTERIES (TGA)

This code is for classical transposition of great arteries (complete transposition, d-transposition) with aorta arising from the right ventricle and the pulmonary artery arising from the left ventricle and with normal or almost normal size of both ventricles. Some infants have a VSD, associated with later diagnosis. The VSD should be coded separately. The patent ductus should not be coded.

Malpositioned great arteries should be coded with Q208 unless more specified.

Coding Committee December 2016

Q204 SINGLE VENTRICLE, COMMON VENTRICLE, DOUBLE INLET LEFT VENTRICLE, COR TRILOCULARE BIATRIATUM

A single ventricle has 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

Q205 DISCORDANT ATRIOVENTRICULAR CONNECTION (Corrected transposition, levo-transposition, ventricular inversion)

This code is to be used when there is both atrioventricular discordance and ventriculo-arterial discordance. This means that the aorta and the pulmonary artery are transposed, with the aorta anterior and to the left of the pulmonary artery; the morphological left and right ventricles with their corresponding atrioventricular valves are also transposed

Coding Committee December 2016

ATRIAL ISOMERISM AND IVERMARK 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 (Q893*) 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

Q213 TETRALOGY OF FALLOT

The ICD10-code for Tetralogy of Fallot is Q213. Do not use other additional cardiac codes for this malformation except if there is pulmonary valve atresia (Q220).

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, edited by Coding Committee December 2016

Q22 Congenital malformations of pulmonary and tricuspid valves

Q226 HYPOPLASTIC RIGHT HEART

There is no clear definition of hypoplastic right heart, but in most cases it is a consequence of limited flow through the ventricle in fetal life due to tricuspid valve atresia or pulmonary (valve) atresia with

intact ventricular septum. The code Q226 may be used if the outcome is a univentricular heart. Add codes for the associated cardiac anomalies such as tricuspid atresia or pulmonary atresia.

Coding Committee December 2016

Q23 Congenital malformations of aortic and mitral valves

Q234 HYPOPLASTIC LEFT HEART

Hypoplastic left heart is a spectrum of cardiac defects characterized by severe underdevelopment of the left side of the heart. The definition includes atresia or marked hypoplasia of aortic orifice or valve with hypoplasia of ascending aorta and defective development of left ventricle (with or without mitral valve stenosis/atresia). The code Q234 includes all these anomalies on the left side of the heart. If there are anomalies on the right side of the heart, codes for these anomalies should be added. Do not code the patent ductus.

Coding Committee December 2016

Q24 Other congenital malformations of heart

Q248 OTHER SPECIFIED CHD

If a TOPFA is performed for a severe CHD without a final diagnosis, please use code Q248 for other specified CHD. Always give written text as specific as possible.

Coding Committee November 2017

Q249 UNSPECIFIED CARDIAC ANOMALY

Only for use if the cardiac diagnosis is certain and completely unknown. Do not use Q249 for a heart murmur. There must be a diagnosis of CHD before reporting to EUROCAT.

Do not use this code for cardiomyopathy – use I42 in the cardiac chapter.

Cardiac hypertrophy due to maternal diabetes is not a congenital heart defect and should not be reported with a CHD code, add in written text only.

Coding Committee November 2017

Q25 Congenital malformations of great arteries

Q250 PATENT DUCTUS (PDA)

Infants with patent ductus will be included as a major anomaly for term born babies only (GA \geq 37 weeks). To be reported only if the PDA is still present 6 months after birth or if surgery/catheter closure is required. Many critically ill neonates have an open PDA for days or weeks with spontaneous closure. These babies should not be reported to EUROCAT. Do not code the PDA if part of a ductus dependent CHD such as transposition of great arteries (Q203), hypoplastic left heart (Q234) and coarctation of aorta (Q2510).

Coding Committee December 2016

Q26 Congenital malformations of great veins

Q27 Other congenital malformations of peripheral vascular system

Q278 for MAPCA

The European Cardiology Society propose to use the code Q278 for MAPCA (multiple aorto-pulmonary collateral arteries). This is not a perfect code, but the best to recommend. This anomaly

is not of the aorta, but of the arteries coming off the aorta. This anomaly is usually associated with Tetralogy of Fallot, but occasionally occurs as an isolated anomaly.

Coding Committee April 2017

Q28 Other congenital malformations of circulatory system

Q30 Congenital malformations of nose

Q300 CHOANAL ATRESIA /CHARGE SYNDROME

For CHARGE syndrome please use the code Q878 for the syndrome name and add CHARGE syndrome in written text. If choanal atresia is present, code with Q300

Coding Committee September 2018

Q302 FISSURED, NOTCHED OR CLEFT NOSE

Bifid tip of nose is a minor anomaly and the code Q189 should be used together with written text.

Fissured, notched and cleft nose are major anomalies.

Coding Committee April 2018

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

Q336 HYPOPLASIA AND DYSPLASIA OF LUNGS

Bronchopulmonary dysplasia is an acquired condition due to preterm birth and the correct code is P270 or P271. These cases should not be reported to EUROCAT and Q-codes should not be used for this disease.

Coding Committee April 2018

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

Q35 CLEFT PALATE

There is no specific code for submucous cleft palate. We recommend to use the code Q353 cleft soft palate and give written text description about details (complete, incomplete)

Coding Committee November 2017

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

CLEFT LIP

If unilateral cleft lip give the side of defect in written text and state if the cleft lip is affecting both lip and gum/the alveolus.

Coding Committee November 2017

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

CLEFT LIP AND PALATE

Find the most appropriate code in Q37 for your case. If unilateral cleft lip give the side of defect in written text and state if the cleft lip is affecting both lip and gum/the alveolus. Also describe the position of the cleft palate in written text.

Coding Committee November 2017

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

Q433 CONGENITAL MALFORMATIONS OF INTESTINAL FIXATION

Q4330 MALROTATION OF COLON

Intestinal rotation physiologically ends around 11 gestational weeks, and can be coded as malrotation or major anomaly after 12 weeks of gestation. The period of intestinal fixation is a process that physiologically lasts until shortly after birth and can be coded as major anomaly if present after this time-period and needs surgery.

Coding Committee April 2018

Q435 ECTOPIC ANUS, MISPLACED ANUS

An anterior anus is one that is positioned closer than normal to the vagina or scrotum. It should only be reported to EUROCAT if surgery was required to re-position it.

Coding Committee April 2018

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 hypospadias (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 hypospadias.

Coding Committee August 2007

Q55 Other congenital malformations of male genital organs

Q556 OTHER CONGENITAL MALFORMATIONS OF PENIS

This code is for major anomalies of penis only. See list of minor anomalies and other conditions for exclusion.

Coding Committee April 2018

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

Q6140 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.

Coding Committee June 2011

6141 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.

Coding Committee June 2011

Q618 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

Coding Committee June 2011

Q619 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

Q620 HYDRONEPHROSIS

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

Q621-Q626 and Q628 CONGENITAL OBSTRUCTIVE DEFECTS OF RENAL PELVIS AND CONGENITAL MALFORMATIONS OF URETER

If these anomalies are diagnosed and associated with hydronephrosis with a diameter of 10 mm or more add the code for hydronephrosis Q620 and give measurement in written text – see coding tip for hydronephrosis.

Coding Committee December 2016

Q63 Other congenital malformations of kidney

Q64 Other congenital malformations of urinary system

Q644 MALFORMATION OF URACHUS

Report cases with urachus anomalies to EUROCAT if surgery is required and if the diagnosis is made before one year of age

Coding Committee April 2017

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

Q6420 POSTERIOR URETHRAL VALVES

Remember to use the 4-digit ICD/BPA10 code for this anomaly. If this anomaly is diagnosed and associated with hydronephrosis with a diameter of 10 mm or more add the code for hydronephrosis Q620 and give measurement in written text – see coding tip for hydronephrosis.

Coding Committee December 2016

Q6476 MEGACYSTIS

The diagnosis megacystis is usually a prenatal diagnosis. For livebirths please try to find the cause and add the code, eg for posterior urethral valves.

Coding Committee April 2017

Q65 Congenital deformities of hip

Q6580 and Q6581 HIP DYSPLASIA

Report to EUROCAT only if the dysplasia is still present at ultrasound examination 6 weeks after birth.

Coding Committee December 2016

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

Q7482 CONGENITAL UNDERGROWTH OF LIMBS .

Use this code for reporting short limbs without a specified diagnosis. Note that short limbs diagnosed prenatally must be followed up after birth for a final diagnosis. Do not use codes for limb reduction defects (Q718 and Q728) or codes for skeletal dysplasia unless specifically diagnosed.
Coding Committee November 2017

Q72 Reduction defects of lower limb

Q7482 CONGENITAL UNDERGROWTH OF LIMBS

Use this code for reporting short limbs without a specified diagnosis. Note that short limbs diagnosed prenatally must be followed up after birth for a final diagnosis. Do not use codes for limb reduction defects (Q718 and Q728) or codes for skeletal dysplasia unless specifically diagnosed.
Coding Committee November 2017

Q73 Reduction defects of unspecified limb

Q74 Other congenital malformations of limb(s)

Q7482 congenital undergrowth of limbs.

Use this code for reporting short limbs without a specified diagnosis. Note that short limbs diagnosed prenatally must be followed up after birth for a final diagnosis. Do not use codes for limb reduction defects (Q718 and Q728) or codes for skeletal dysplasia unless specifically diagnosed.
Coding Committee November 2017

Q75 Other congenital malformations of skull and face bones

Q7502 TRIGONENCEPHALY

This is often used to describe a somewhat triangular head shape but, for EUROCAT cases, should only be used where this is due to premature fusion of the metopic suture requiring treatment.
Coding Committee April 2018

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

Q754 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

Q759 CONGENITAL MALFORMATION OF SKULL AND FACE BONE, UNSPECIFIED

Do not use this code for dysmorphic features affecting face. Always use code Q189.

Coding Committee April 2018

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 use 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

Q8281 ACCESSORY SKIN TAGS

This is a minor anomaly.

Coding Committee April 2018

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 Q754 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

Q8708 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

Q878 OTHER SPECIFIED SYNDROMES

Aicardi syndrome, Joubert syndrome, Miller-Dieker syndrome and Walker-Warburg syndrome:

Please code these genetic syndromes with the code Q878 and give the syndrome name in written text. Also give the code for the diagnosed cerebral anomaly in malf1 (Q043 for reduction deformity of brain, Q0433 for lissencephaly)

Coding Committee September 2018

Q878 OTHER SPECIFIED SYNDROME

For CHARGE syndrome please use the code Q878 for the syndrome name and add CHARGE syndrome in written text. If choanal atresia is present, code with Q300

Coding Committee September 2018

Q89 Other congenital malformations, not elsewhere classified

Q897 MULTIPLE CONGENITAL MALFORMATIONS, NOT ELSEWHERE CLASSIFIED

This code should **always** be accompanied with codes (and text description) of all the separate major congenital anomalies that are present. The case will not be included in the surveillance of multiples unless specified codes are given.

Coding Committee April 2018

Q90-Q99 Chromosomal anomalies

Array results: Report only clearly pathogenic variants and if uncertain, include only copy number variants (CNVs) (duplications or deletions) larger than 1 MB. Only report cases with de novo CNVs unless the parent in familial cases also has clinical manifestations of the condition (dysmorphic features or congenital anomalies).

Coding Committee June 2015

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:

D180 is the correct code for haemangiomas

D181 is the correct code for lymphangiomas

Coding Committee September 2018

P351 CONGENITAL CMV INFECTION

Infants with congenital CMV infections should be reported to EUROCAT if there are associated major congenital anomalies (microcephaly according to EUROCAT definition, other structural anomalies).

Code P351 in the syndrome variable and major malformations in the malformation variables

Coding Committee December 2016

P358 CONGENITAL ZIKA VIRUS

This code to be used for all cases exposed to zika virus infection in pregnancy. Code P358 in the syndrome variable and major malformations (microcephaly, other cerebral anomalies and all other major and minor anomalies) in the malformation variables. Specify in text for all codes.

Since this is a new infection, report all cases irrespective of diagnosed congenital anomalies. Code the maternal illness during pregnancy as A928 – "other specified mosquito-borne viral fevers". These codes to be used only for zika virus.

Coding Committee December 2016

P371 CONGENITAL TOXOPLASMOSIS

Infants with congenital toxoplasmosis should be reported to EUROCAT if there are associated major congenital anomalies (hydrocephaly, other structural anomalies). Code P371 in the syndrome variable and major malformations in the malformation variables

Coding Committee December 2016

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 Reversed Arterial Perfusion 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

Q248 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 and November 2017

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 is 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

Coding Committee June 2006

PRETERM COMPLICATIONS

Most complications to preterm birth, including bronchopulmonary dysplasia and persistent fetal circulation, are reported with a code in the P-chapter and will not be relevant for EUROCAT.

Terminations, spontaneous abortions after GA 20 weeks and preterm birth with lung hypoplasia due to early rupture of membranes are not EUROCAT cases. Limb contractures and retrognathia due to early rupture of membranes are secondary diagnoses and not EUROCAT cases.

Coding Committee April 2017

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 include 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 |
| Severe 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 |

EUROCAT Guide 1.4 and Reference Documents

| EUROCAT Subgroup | Description | Often diagnosed after one week of age |
|--|---|---------------------------------------|
| 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 subvalvular septal defect. | yes |
| Double outlet right ventricle | Both aorta and the pulmonary artery connect to the right ventricle | 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 |
| Mitral valve anomalies | Atresia, stenosis or insufficiency of the mitral valve | Yes for stenosis and insufficiency |
| 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 |
| Aortic atresia/interrupted aortic arch | Atresia or interrupted connection of the aorta | |

EUROCAT Guide 1.4 and Reference Documents

| EUROCAT Subgroup | Description | Often diagnosed after one week of age |
|---|---|---------------------------------------|
| 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 membranous choanae with no passage from nose to pharynx | Yes for unilateral |
| Cystic adenomatous malformation 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 a long gap of the oesophagus with or without tracheo-oesophageal 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 parasympathetic ganglion nerve cells (aganglionosis) of the wall of the colon or rectum. May result in congenital 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 |

EUROCAT Guide 1.4 and Reference Documents

| EUROCAT Subgroup | Description | Often diagnosed after one week of age |
|--|--|---------------------------------------|
| Urinary | | |
| Bilateral renal agenesis including Potter syndrome | Bilateral absence, agenesis, dysplasia or hypoplasia of kidneys including Potter's syndrome. Incompatible with life | no |
| Multi cystic renal dysplasia | Multiple, non-communicating cysts of varying size in the kidney without functional 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 |
| 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 |

EUROCAT Guide 1.4 and Reference Documents

| EUROCAT Subgroup | Description | Often diagnosed after one week of age |
|---|---|---------------------------------------|
| VATER/VACTERL | Association with anomalies of Vertebra, anal atresia, cardiac, trachea-esophageal fistula, esophageal atresia, radial anomaly and limb defects | no |
| Vascular disruption anomalies (selected) | Anomalies likely to be due to vascular disruption | No |
| Laterality anomalies | Abnormal laterality mainly affecting heart and lungs | yes |
| 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 |
| 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**
- 4.2 Interpretation of 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.

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

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

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

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

| | |
|--------------------------|---|
| 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.

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

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

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

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

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

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

| | |
|---------|---|
| 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 |

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

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

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**
- 5.2 Registry Description Questionnaire**
- 5.3 Definition of Data Quality Indicators**

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 [JRC EUROCAT@ec.europa.eu](mailto:JRC_EUROCAT@ec.europa.eu)

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)**

A

B

C

(answer all A, B and C)

- ☐ Government/health authority
- ☐ University or research institute
- ☐ Hospital
- ☐ Private organisation
- ☐ Other, specify below

- ☐ Ordered by national law
- ☐ Regional/provincial law
- ☐ Not ordered by law

- ☐ Steering committee
- ☐ No steering committee

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

- | | |
|---|--|
| <input type="checkbox"/> Population-based I | <input type="text"/> % mothers delivering outside registry area * |
| <input type="checkbox"/> Population-based II | <input type="text"/> % non-resident mothers delivering within registry area * |
| <input type="checkbox"/> Population-based III | <input type="text"/> % non-resident mothers delivering within registry area * |
| <input type="checkbox"/> Hospital-based | <input type="text"/> % deliveries in defined geographic area (specify below) occurring in the selected hospitals * |
| <input type="checkbox"/> 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 <input type="text"/> | Type of change, detail <input type="text"/> |
| | Date <input type="text"/> | Type of change, detail <input type="text"/> |
| | Date <input type="text"/> | Type of change, detail <input type="text"/> |
| | Date <input type="text"/> | Type of change, detail <input type="text"/> |
| | Date <input type="text"/> | Type of change, detail <input type="text"/> |
| | Date <input type="text"/> | Type of change, detail <input type="text"/> |

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 Required by which law?
☐ Other

Further information

D2 How are the malformed cases notified to the Registry? (tick all that apply)

☐ notification of cases to the Registry by hospitals and other institutions

☐ active searching of patient notes by Registry staff

☐ active searching of hospital etc. discharge registers by Registry staff

☐ active searching of other local or national registers by Registry staff

☐ other, specify below

D3 Sources of information of Registry

WHO?

Use the score system below, for all that apply, for each source

0 = Not used as a source of information

1 = Occasional notification of malformed cases seen

2 = Virtually complete notification of all malformed cases seen

| | |
|----------------------|----------------------|
| Community/GP doctors | <input type="text"/> |
| Hospital doctors | <input type="text"/> |
| Nurses | <input type="text"/> |
| Midwives | <input type="text"/> |
| Health visitors | <input type="text"/> |
| Other, specify | <input type="text"/> |

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

| | |
|----------------------|---|
| <input type="text"/> | Prenatal screening (ultrasound, serum testing, etc) |
| <input type="text"/> | Maternity units |
| <input type="text"/> | Paediatric departments |
| <input type="text"/> | Child health services |
| <input type="text"/> | Pathology labs |
| <input type="text"/> | 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

Early fetal deaths (spontaneous abortions)

Early neonatal deaths (0-7d)

Infant deaths (<1yr)

Other, specify

| | | |
|------------------------------|-----------------------------|------------------------------------|
| <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Sometimes |
| <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Sometimes |
| <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Sometimes |
| <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Sometimes |
| <input type="checkbox"/> Yes | <input type="checkbox"/> No | <input type="checkbox"/> Sometimes |

E3 If yes, does Registry get death certificates?

Routinely

On request

Stillbirths

Early neonatal deaths

Infant deaths

| |
|--------------------------|
| <input type="checkbox"/> |
| <input type="checkbox"/> |
| <input type="checkbox"/> |

| |
|--------------------------|
| <input type="checkbox"/> |
| <input type="checkbox"/> |
| <input type="checkbox"/> |

E4 Is there a lower gestational age or weight limit to include early fetal deaths/spontaneous abortions in the Register?

☐ Yes

☐ No

If yes, specify

 g

☐ and/ ☐
or

 wks of gestation

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

All deaths

Malformed cases on Register

Year

Stillbirths

Early fetal deaths

Early neonatal deaths

Infant deaths

TOPFA

| |
|--|
| |
| |
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|--|
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| |

Do the above autopsy rates refer exactly to your Registry population?

☐ Yes

☐ No

If not, explain

Do specialist fetopathologists do most autopsies?

Stillbirths

Early fetal deaths

Early neonatal deaths

Infant deaths

TOPFA

☐ Yes

☐ No

☐ Yes

☐ No

☐ Yes

☐ No

☐ Yes

☐ No

☐ 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

G REGISTRY INFORMATION: DIAGNOSES

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

Please note that EUROCAT only accepts cases coded in ICD10 or ICD10 BPA

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 How many diagnoses can be registered per case?

- ☐ 8 malformation codes and syndromes
- ☐ Less than 8
- ☐ Up to diagnoses
- ☐ No upper limit

G5 Separate summary/syndrome diagnosis used in Registry?

☐ Yes ☐ No

G6 Other special diagnostic codings used?

☐ no

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

G7 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

G8 Are there any anomalies which you believe are not currently fully covered/could be improved by your Registry? Explain

G9 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 blue. 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 | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="text"/> |
| Assisted conception | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="text"/> |
| Illness before pregnancy | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="text"/> |
| Illness during pregnancy | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="text"/> |
| Folic Acid supplementation | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="text"/> |
| Drugs taken during first trimester | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="text"/> |
| Maternal education | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="text"/> |
| Social class of mother | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="text"/> |
| Social class of father | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="text"/> |
| Maternal migrant status | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | <input type="text"/> |

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 Which software packages do you use for data management?

K3 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+TOP FA |
| <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> |
| 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

| | LB Number | | FD Number | | TOPFA Number | | Prevalence (LB+FD+TOPFA/ total births) | |
|--|-----------|--|-----------|--|--------------|--|--|--|
| Anomalies | | | | | | | | |
| All anomalies | | | | | | | | |
| Nervous system | | | | | | | | |
| Neural tube defects | | | | | | | | |
| Anencephalus and similar | | | | | | | | |
| Encephalocele | | | | | | | | |
| Spina bifida | | | | | | | | |
| Hydrocephalus | | | | | | | | |
| Severe 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 | | | | | | | | |
| Double outlet right ventricle | | | | | | | | |
| Transposition great vessels | | | | | | | | |
| Single ventricle | | | | | | | | |
| Ventricular Septal Defect | | | | | | | | |
| Atrial Septal Defect | | | | | | | | |
| AVSD | | | | | | | | |
| Tetralogy of Fallot | | | | | | | | |
| Tricuspid atresia and stenosis | | | | | | | | |
| Ebstein's anomaly | | | | | | | | |
| Pulmonary valve stenosis | | | | | | | | |
| Pulmonary valve atresia | | | | | | | | |
| Aortic valve atresia/stenosis | | | | | | | | |
| Mitral valve anomalies | | | | | | | | |
| Hypoplastic left heart | | | | | | | | |
| Hypoplastic right heart | | | | | | | | |
| Coarctation of aorta | | | | | | | | |
| Aortic atresia/interrupted aortic arch | | | | | | | | |
| Total anomalous pulm venous return | | | | | | | | |
| PDA as only CHD in LB term infant (GA 37+ weeks) | | | | | | | | |
| Respiratory | | | | | | | | |
| Choanal atresia | | | | | | | | |

| | LB Number | FD Number | TOPFA Number | Prevalence (LB+FD+TOPFA/ total births) |
|--|-----------|-----------|--------------|--|
| Anomalies | | | | |
| Cystic adenomatous malformation of lung | | | | |
| Oro-facial clefts | | | | |
| Cleft lip with or without cleft palate | | | | |
| Cleft palate | | | | |
| Digestive system | | | | |
| Oesophageal atresia with or without tracheo-oesophageal fistula | | | | |
| Duodenal atresia or stenosis | | | | |
| Atresia or stenosis 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 | | | | |
| Multi cystic renal dysplasia | | | | |
| Congenital hydronephrosis | | | | |
| Bladder exstrophy and/or epispadia | | | | |
| Posterior urethral valve and/or prune belly | | | | |
| Genital | | | | |
| Hypospadias | | | | |
| Indeterminate sex | | | | |
| Limb | | | | |
| Limb reduction | | | | |
| 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 | | | | |

| | LB Number | FD Number | TOPFA Number | Prevalence (LB+FD+TOPFA/ total births) |
|--|----------------------|----------------------|----------------------|--|
| Anomalies | | | | |
| Congenital skin disorders | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> |
| VATER/VACTERL | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> |
| Vascular disruption anomalies | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> |
| Laterality anomalies | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> |
| Teratogenic syndromes with malformations | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> |
| Fetal alcohol syndrome | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> |
| Valproate syndrome | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> |
| Maternal infections resulting in malformations | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> |
| Genetic syndromes + microdeletions | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> |
| Chromosomal | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> |
| Down syndrome | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> |
| Patau syndrome / trisomy 13 | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> |
| Edwards syndrome / trisomy 18 | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> |
| Turner syndrome | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> |
| Klinefelter syndrome | <input type="text"/> | <input type="text"/> | <input type="text"/> | <input type="text"/> |

Thank you for completing this questionnaire.

Please attach any helpful documentation e.g. 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:

$$O = LB + FD + (TOPFA \text{ corrected to 20 weeks gestational age})$$

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, hypospadias 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 positive 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

- 6.1 EUROCAT Central Database**
- 6.2 Release of Data**
- 6.3 Guidelines on Security and Confidentiality for Staff Working in EUROCAT Central Registry**
- 6.4 Data Protection Principles**

Policy regarding Security, Confidentiality, Release and 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.

Note: Following the transfer of Central Registry to the European Commission's Joint Research Centre (01/01/2015), the new Data Protection and Access to Data procedures are under development.

Contact JRC-EUROCAT Central Registry for any query (JRC-EUROCAT@ec.europa.eu).

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 Joint Research Centre (JRC) of the European Commission, Health in Society Unit (JRC F1) , Directorate F - Health, Consumer and Reference Materials – Ispra Italy. It currently holds information on more than 430,000 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 JRC_EUROCAT@ec.europa.eu. 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.