2.2.2 Coding Instructions for the variables

Baby and Mother (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
1	CENTRE	CENTRE NUMBER	Code allocated by Central Registry
2	NUMLOC	LOCAL ID Each case has a unique identification. This number is a maximum of 11 characters long, consisting of numbers, letters or both. ID numbers should not repeat themselves in different years.	Up to 11 characters
3	BIRTH_DATE	DATE OF BIRTH Please enter dates in one of the following format: dd/mm/yyyy, yyyy/mm/dd, yyyy/mm/xx, yyyy/xx/xx. Old formats are also accepted: ddmmyy, 99mmyy and 9999yy. Please make sure to choose one format and stick to it.	Day, month, year xx = Not known for day and month DO NOT TRANSMIT RECORDS IF YEAR OF BIRTH IS NOT KNOWN
4	SEX	SEX Indicate chromosomal sex, if known, in case of ambiguous genitalia code malformations in variables 32-47. Indicate indeterminate sex in case of ambiguous genitalia with unknown or abnormal sex chromosome complement. If sex could not be determined at autopsy due to maceration, very small fetus, or other problems, indicate as "not known".	1 = Male 2 = Female 3 = Indeterminate 9 = Not known
5	NBRBABY	NUMBER OF BABIES/FETUSES DELIVERED Fill out a separate form for each malformed baby/fetus in a multiple set. Only one form is 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 the 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	SPECIFY TWIN TYPE OF BIRTH (malformed and non-malformed), like or unlike sex, zygosity	Free text
7	NBRMALF	NUMBER OF MALFORMED IN MULTIPLE SET To be completed for multiple delivery only. Remember to give local ID of co-twin in SIB1 field (variable 92) if more than one malformed.	1 = One 2 = Two 3 = Three 4 = Four 5 = Five 6 = Six or more 9 = Not known
8	TYPE	TYPE OF BIRTH Birth with the type of birth not known should be transmitted to EUROCAT, but will be excluded from routine EUROCAT analysis. EUROCAT includes all live births, fetal deaths with gestational age (GA) ≥20 weeks, and terminations of pregnancy (at any gestational age) after prenatal diagnosis of malformation (TOPFA). Fetal deaths with gestational age (GA) < 20 weeks (code = 3) may be reported to EUROCAT but will not be included in prevalence data. All cases MUST have been confirmed as having a MAJOR congenital anomaly (see exclusion list, chapter 3.2). 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 a 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 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 =	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
		date of reduction; and code carefully all multiple birth variables).	
9	CIVREG	CIVIL REGISTRATION STATUS Live births and stillbirths are civilly registered leading to either birth or stillbirth certificates and appear in official birth statistics for your region. Code here whether this case fulfilled the conditions for live or stillbirth registration in your country.	1 = Live birth 2 = Stillbirth 3 = No civil registration 9 = Not known
10	WEIGHT	BIRTH WEIGHT Give weight in grams.	9999 = Not known (Do not use 99 or 999 for "Not Known" as this will be considered the birth weight).
11	GESTLENGTH	LENGTH OF GESTATION IN COMPLETED WEEKS Give best estimate based on last menstrual period (LMP) and/or ultrasound determination. If the case is the result of fetal reduction give GA at feticide. Check GA below 10 weeks or above 43 weeks.	99 = Not known
12	SURVIVAL	SURVIVAL BEYOND ONE WEEK OF AGE Yes = Child known to be alive after one week. No = Child known to have died before or during the first week (including stillbirths and abortions). Alive at discharge <1 week refers to cases that are alive at discharge from maternity units before one week of age. Please specify in your Registry Description the day when discharge from maternity units usually takes place. If survival at one week is unknown, but survival at discharge from maternity unit less than one week is known, use the latter. The definition of the first week of life varies between countries. Follow your country's perinatal mortality definition and specify this in your Registry Description. Not known = Not known if a child has died during first week.	1 = Yes 2 = No 3 = Alive at discharge <1 week 9 = Not known
13	DEATH_DATE	DATE OF DEATH For live births only. Please enter dates in one of the following format: dd/mm/yyyy, yyyy/mm/dd, yyyy/mm/xx, yyyy/xx/xx and xxxx/xx/xx. Old formats are also accepted: ddmmyy, 99mmyy, 9999yy, 999944, 222222 and 333333. Please make sure to choose one format and stick to it.	Day, month, year xx = Died, not known day or month xxxx =Died, not known year 2222/22/22 = Known to be alive at 1 year 3333/33/33 = Not known if alive or dead at 1 year

Baby and Mother (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
14	DATEMO	DATE OF BIRTH OF MOTHER Give as much information as is known e. g. Feb 1963 = 1963/02/xx, 1963 = 1963/xx/xx. Please enter dates as dd/mm/yyyy, yyyy/mm/dd, yyyy/mm/xx, yyyy/xx/xx and xxxx/xx/xx. Old formats are also accepted: ddmmyy, 99mmyy, 9999yy and 999944. Please make sure to choose one format and stick to it. This variable can be used to calculate maternal age at Expected Date of Delivery for preterm deliveries and terminations.	Day, month, year xx = Not known day or month xxxx = Not known year
15	AGEMO	AGE OF THE MOTHER AT DELIVERY In completed years at the time of delivery. If only the year of birth is available, assume that the mother was born on 30 June.	99 = Not known
16	BMI	MATERNAL BODY MASS INDEX Enter BMI (rounded to one decimal place). The DMS 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 the mother is known to be obese, enter code for obesity E660 in maternal illness before pregnancy (variables 61-63). BMI was a new variable in Guide 1.4. If any registry has information on maternal BMI for previous years, EUROCAT is interested in collecting this information from 2005 onwards.	Rounded to one decimal place Expected range 15 – 50 96 = exact BMI not known but BMI < 18.5 97 = exact BMI not known but 18.5 to <30 98 = exact BMI not known but >=30 99 = Not known
17	RESIDMO	MOTHER'S RESIDENCE CODE Use local code for locality of residence at time of delivery.	Local code (up to 10 digits)
18	TOTPREG	TOTAL NUMBER OF PREVIOUS PREGNANCIES NOTE – The current reported pregnancy is NOT included. Include all previous abortions whether spontaneous or induced. Multiple pregnancies count as 1 in the total.	00 = None 01 = One 02 = Two 03 = Three, etc. 20 = Twenty or more 99 = Not known

Diagnosis (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
19	WHENDISC	WHEN DISCOVERED When the baby was first suspected of having a congenital anomaly. For prenatal diagnosis: when a major congenital anomaly was first suspected (EXCLUDING soft markers except if nuchal translucency indicates a very high risk followed by confirmation of diagnosis at delivery/termination). If a prenatal diagnosis is made when a fetus is dead code 1 (for stillbirths) or 7 (for spontaneous abortions). For live births: when the first suspicion of an anomaly was at death OR at *post mort*em, when discovered is the age at death (e.g. at birth, < 1 week, 1-4 weeks etc.). For stillbirths: when the first suspicion of an anomaly was at birth OR at *post mortem*, when discovered is at birth (e.g. Code = 1). All cases MUST have been confirmed as having a MAJOR congenital anomaly (see exclusion list, chapter 3.2). EUROCAT accepts a positive NIPT for trisomy 13, 18 and 21 without a full karyotype. Please also complete variables 12 "SURVIVAL", 13 "DEATH-DATE", 20 "CONDISC" and 28 "PM".	1 = At birth 2 = Less than 1 week 3 = 1-4 weeks 4 = 1-12 months 5 = Over 12 months 6 = Prenatal diagnosis in live fetus 7 = At abortion (spontaneous) 9 = Not known 10 = Postnatal diagnosis, age not known
20	CONDISC	CONDITION AT DISCOVERY Condition of fetus or baby when malformation was first suspected.	1 = Alive 2 = Dead 9 = Not known
21	AGEDISC	IF PRENATALLY DIAGNOSED, GESTATIONAL AGE AT DISCOVERY IN COMPLETED WEEKS GA as defined in variable GESTLENGTH. Gestational age at which the fetus was first suspected to be malformed (EXCLUDING soft markers). Indicate time of examination rather than time when the result was known. If no prenatal diagnosis please leave blank.	99 = Not known
22	FIRSTPRE	FIRST POSITIVE PRENATAL TEST This refers to the first prenatal test whether screening procedure or diagnostic test which indicated a possible congenital anomaly or need for further tests. For code 7 = other specified test, give information in the text field (variable 23). If test performed and result negative, then the "When discovered" variable cannot be coded 6 (prenatal diagnosis). This field is to record what DID happen, not any possible plans or intentions. Ultrasound < 14 weeks means only	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 = Chorion villus sampling or amniocentesis

Diagnosis (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
		ultrasound performed which may include a nuchal measurement. The serum/combined screening must involve a biochemical test	7 = Other test positive 8 = Test(s) performed, result negative 9 = Not known 10 = No test performed 11 = Fetal karyotype on maternal blood
23	SP_FIRSTPRE	SPECIFY "OTHER" FIRST PRENATAL TEST If FIRSTPRE = 7, specify which positive prenatal test	Free text
24	KARYO	TO BE FILLED IN FOR CASES BORN UNTIL 31/12/2025 KARYOTYPE OF INFANT/FETUS Specify the result in variable 25. Array results count as a karyotype test. 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) If performed and results known, please specify (according to the latest ISCN edition). "Probe test performed" refers to FISH, PCR, NIPT or other analyses restricted to specific chromosomal regions. "Failed" refers to a technical failure where a repeat examination could not be done and the karyotype is therefore unknown.	1 = Performed, result known 2 = Performed, results not known 3 = Not performed 4 = Probe test performed 8 = Failed 9 = Not known
25	SP_KARYO	TO BE FILLED IN FOR CASES BORN UNTIL 31/12/2025 SPECIFY KARYOTYPE OR CHROMOSOMAL MICROARRAY EXAMPLES: 47,XY,+21 46,XX,del(2)(p13p23)*mat* *Description: An interstitial deletion resulting from mal-segregation of a maternal insertion* 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* arr(1–22)×3,(X)×2,(Y)×1 *Description: Microarray analysis shows triploidy 69,XXY* arr(8)×3,(21)×3 *Description: Microarray analysis shows a single copy gain of chromosomes 8 and 21.*	Free text

Diagnosis (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
		46,XY.rsa(13,18,21)×2,(X,Y)×*1 Description: normal chromosomes, only chromosome 13,18,21, X and Y investigated* rsa 22q11.2('kit name')×1 *Description: microdeletion 22q.11.2 diagnosed by multiple ligation probe amplification method (MLPA)*	
26	GENETTEST	TO BE FILLED IN FOR CASES BORN FROM 01/01/2026 GENETIC TEST Indicate whether a genetic test was done pre- or postnatally. This includes any genetic test, except the NIPT (e.g. QF PCR, MLPA, FISH, karyotype, array, mitomycin C test, sequence analysis, gene panel, whole exome sequencing, genome sequencing, RNA analysis and methylation assays). The genetic test might have been performed for another indication than the congenital anomaly. One exception: if the NIPT diagnosed a trisomy and this was not confirmed with another genetic test, please register GENETTEST with label 1.	1 = performed, result known 2 = performed, result unknown 3 = not performed 8 = test failed 9 = not known if a genetic test was performed
27	GENETRESULT	TO BE FILLED IN FOR CASES BORN FROM 01/01/2026 GENETIC TEST RESULT Indicate whether a genetic cause has been found for the congenital anomaly. Only to be filled out if GENETTEST is 1. Genetic cause found for the congenital anomaly - Examples: pathogenic variant or likely pathogenic variant leading to the congenital anomaly or a syndrome, abnormal methylation, aneuploidy, microdeletion syndrome Unclear if the genetic cause for the congenital anomaly is found - Examples: variant of unknown significance (VUS), small deletion or duplication where the parents are not tested (please update to 1 or 3 if additional information becomes available), deletion or duplication with incomplete penetrance and variable expression that increases the risk of the congenital anomaly, gene variant that is thought to increase the risk of the congenital anomaly but the congenital anomaly will only occur in the presence of other gene variants or environmental factors NO genetic cause found for the congenital anomaly with the tests that were performed - Examples: normal test result, (likely) benign variant, polymorphism also found in healthy parent, small deletion or duplication not containing important genes, inversion or balanced translocation without interruption of an important gene, one heterozygous variant for an autosomal recessive disorder	1 = genetic cause found for the congenital anomaly 2 = unclear if the genetic cause for the congenital anomaly is found 3 = no genetic cause found for the congenital anomaly with the tests that were performed (they might be incomplete) 9 = not known from the information that the registry can access
28	SP_GENETRESULT	TO BE FILLED IN FOR CASES BORN FROM 01/01/2026 SPECIFY GENETIC TEST RESULT STATUS	Free text

Diagnosis (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
		specify the result of the genetic test. Only to be filled out if GENETTEST is 1. First specify the test which identified the genetic cause for the congenital anomaly. When a person has two genetic disorders, please specify both pathogenic results in SP_GENETRESULT. If a risk factor was found (and no genetic cause), specify this result. If there was one unclear result and all other tests were normal, specify the unclear result. If all tests had normal results, show the type of test and the result of the most extensive test. It is possible to state here if the test was done pre- or postnatally. Ranking of genetic tests from most to least extensive: whole genome sequencing (WGS), whole exome sequencing (WES), gene panel, sequence analysis, RNA analysis, methylation assay, array, karyotype, probe test (MLPA, QF PCR, NIPT, FISH). Examples: 47,XX,+21 Del22q11.21 of 2.57 Mb, de novo, leading to VCF Gene panel connective tissue diseases: pathogenic FBN1 mutation, inherited from parent with Marfan syndrome arr[GRCh37] 16p13.3(4204377_4686169)x1 dn, de novo del16p13.3 of 481.8 kb including several genes, not known to cause disease aCGH: no abnormalities explaining the prenatal ultrasound scan anomalies arr[GRCh37] 16p13.11(15124673_16310947)x3 dn, de novo dup16p13.11 of 1.2 Mb, risk factor for developmental delay and heart defects	
29	GENTEST	TO BE FILLED IN FOR CASES BORN UNTIL 31/12/2025 GENETIC TEST For syndromes and single gene disorders, a genetic test may have confirmed the clinical diagnosis either prenatally or postnatally. Please complete for these cases. Karyotype should still be completed as per variables 24 & 25 If any registry has this information for previous cases, EUROCAT is interested in collecting this information from 2005 onwards If the test is performed but the result not yet known, please wait for the result before reporting.	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
30	SP_GENTEST	TO BE FILLED IN FOR CASES BORN UNTIL 31/12/2025 SPECIFY TYPE OF GENETIC TEST	Free text

Diagnosis (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
		Give method used and the result of the test (type of mutation and which gene) Examples: Single gene analysis, exome sequencing, gene panel analysis, whole genome sequencing.	
31	PM	*POST MORTEM* EXAMINATION If performed, record the malformation(s) discovered in the "malformation" section in the form. If other findings, record in the "general remarks" space (variable 103). "Results known" means that the autopsy record has been reviewed by the registry. "Results not known" means that the autopsy record was not available to the registry. "Macerated fetus" means that although a *post mortem* was performed, maceration of the fetus prevented a full protocol from being followed.	1 = Performed, results known 2 = Performed, results not known 3 = Not performed 4 = Macerated fetus 9 = Not known
32	SURGERY	FIRST SURGICAL PROCEDURE FOR MALFORMATION (PERFORMED OR EXPECTED) Complete for all live births and fetal deaths (only if there was prenatal surgery). The variable surgery does not include insertions of catheters. Performed (or expected) means that this case has already, or will at the appropriate age, have surgery for one or more of the listed malformations. "No surgery required" means that this case does not have a severe enough malformation, or that the malformation is not correctable by surgery. "Too severe for surgery" means that there has been an active decision to withhold surgery due to low chances of survival or very poor prognosis.	1 = Performed (or expected) in the first year of life 2 = Performed (or expected) after the first year of life 3 = Prenatal surgery 4 = No surgery required 5= Too severe for surgery 6 = Died before surgery 9 = Not known
33	SYNDROME	SYNDROME OR ASSOCIATION All cases MUST have been confirmed as having a MAJOR congenital anomaly (see exclusion list, chapter 3.2). Use this variable for genetic syndromes, skeletal dysplasias, hereditary skin disorders teratogenic syndromes, associations, microdeletions and chromosomal anomalies. Refer to EUROCAT Guide on syndromes. Give the 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.	ICD 10 First 4 digits are ICD10 5th digit = BPA supplement or leave blank

Diagnosis (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
		When 2 syndromes are present in the same subject, code the more important one in the syndrome variables 30 and 31, and include the other one in variables 32 and 33 MALF01. Ensure karyotype information is given in variables 24 and 25 and that information on genetic tests are given in variable 26 and 27. Mention in variable 28 if the autopsy report has been reviewed, where appropriate. Local registries are advised to keep photographs and x-ray images of all syndrome cases if possible, as the diagnosis might be established on the basis of specific facial dysmorphism.	
34	SP_SYNDROME	SPECIFY SYNDROME Written text description of the ICD10 code in variable 30.	
35	MALFO1	MALFORMATION All cases MUST have been confirmed as having a MAJOR congenital anomaly (see exclusion list, chapter 3.2). A baby/fetus with ONLY minor anomalies (see exclusion list, chapter 3.2) should not be transmitted to Central Registry. This rule does not concern established syndromes (e.g. Down, Beckwith Wiedemann or Prader Willi syndrome without a major congenital anomaly are to be submitted). When a major anomaly is present, code both major and minor anomalies. In the case of conjoined twins, give a 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 8th anomaly (text variable 47 SP_MALFO8). Include in the 8 specified codes the most important ones, or those tabulated in EUROCAT Reports. Give written description of the malformations available in malformation text variables 33, 35, 37, 39, 41, 43, 45 and 47.	ICD 10 First 4 digits are ICD 5th digit = BPA classification OR leave blank
36	SP_MALFO1	SPECIFY MALFORMATION	Free text
37	MALFO2	AS MALFO1	AS MALFO1
38	SP_MALFO2	SPECIFY MALFORMATION	Free text
39	MALFO3	AS MALFO1	AS MALFO1
40	SP_MALFO3	SPECIFY MALFORMATION	Free text
41	MALFO4	AS MALFO1	AS MALFO1
42	SP_MALFO4	SPECIFY MALFORMATION	Free text
43	MALFO5	AS MALFO1	AS MALFO1
44	SP_MALFO5	SPECIFY MALFORMATION	Free text
45	MALFO6	AS MALFO1	AS MALFO1
46	SP_MALFO6	SPECIFY MALFORMATION	Free text
47	MALFO7	AS MALFO1	AS MALFO1

Diagnosis (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
48	SP_MALFO7	SPECIFY MALFORMATION	Free text
49	MALFO8	AS MALFO1	AS MALFO1
49	SP_MALFO8	SPECIFY MALFORMATION	Free text
50	PRESYN	PRENATAL DIAGNOSIS FOR SYNDROME When each anomaly was first diagnosed. The basis for this variable is to record whether the prenatal findings strongly suggest the postnatal diagnosis. This variable is not designed for fetal medicine specialists to assess the accuracy of their prenatal diagnosis. Thus, the finding of a significant heart anomaly prenatally is considered to be prenatally detected, even if the *exact* anomaly was not correctly diagnosed. *Note: A code "3=partially prenatally diagnosed" was in use until birth year 2021. It has been discontinued but there are cases with the code =3 in the database for some registries.*	1 = Yes, this anomaly was diagnosed prenatally 2 = No, this anomaly was diagnosed postnatally 9 = Not known
51	PREMAL1	PRENATAL DIAGNOSIS FOR MALFORMATION AS PRESYN	AS PRESYN
52	PREMAL2	PRENATAL DIAGNOSIS FOR MALFORMATION AS PRESYN	AS PRESYN
53	PREMAL3	PRENATAL DIAGNOSIS FOR MALFORMATION AS PRESYN	AS PRESYN
54	PREMAL4	PRENATAL DIAGNOSIS FOR MALFORMATION AS PRESYN	AS PRESYN
55	PREMAL5	PRENATAL DIAGNOSIS FOR MALFORMATION AS PRESYN	AS PRESYN
56	PREMAL6	PRENATAL DIAGNOSIS FOR MALFORMATION AS PRESYN	AS PRESYN
57	PREMAL7	PRENATAL DIAGNOSIS FOR MALFORMATION AS PRESYN	AS PRESYN
58	PREMAL8	PRENATAL DIAGNOSIS FOR MALFORMATION AS PRESYN	AS PRESYN
59	OMIM	OMIM / TYPE OF MENDELIAN INHERITANCE To be coded by medical geneticist or after advice from medical geneticist. For reporting OMIM refer to EUROCAT Syndrome Guide. The first digits may be filled in without the rest of the code if the full OMIM code is not known. Full codes can be found on the OMIM website http://www.ncbi.nlm.nih.gov/omim/	
60	ORPHA	This code is to be used for rare diseases including congenital anomalies, chromosomal, teratogenic and genetic syndromes. Use the link and enter the name of the condition. If more than one code/disease appears, select the most specific ORPHAcode. If you do not have specific information about genetic background or phenotype, select the most general ORPHAcode.	

Diagnosis (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
		https://www.orpha.net/consor/cgi-bin/Disease.php?lng=EN	

Exposure (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
61	ASSCONCEPT	ASSISTED CONCEPTION IVF = In vitro fertilization GIFT = Gamete intra fallopian transfer ICSI = Intracytoplasmic sperm injection	0 = No 1 = Induced ovulation only 2 = Artificial insemination 3 = IVF 4 = GIFT 5 = ICSI 6 = Egg donation 8 = Other 9 = Not known 10 = Assisted conception, type unknown
62	OCCUPMO	MOTHER'S OCCUPATION AT TIME OF CONCEPTION Code main occupation at time of conception (or earliest known time in first trimester). Note that the main purpose of the variable relates to potential teratogenic occupational exposures in early pregnancy. Be as precise as possible. Code according to the 2008 International Standard Classification of Occupations (ISCO-08) 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: International Standard Classification of Occupations (ISCO) 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)
63	ILLBEF1	ILLNESS BEFORE PREGNANCY 1	ICD 10 0 = No illness

Exposure (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
		Record any maternal illness whether chronic or acute with onset before pregnancy that may affect fetal development (e. g. childhood cancer, metabolic and endocrine disease, severe congenital anomaly). Code according to ICD10. The codes mentioned below are only examples (non-exhaustive list). ILLBEF1 and ILLBEF2 does NOT imply the disease in ILLBEF1 is more important/severe than the disease in ILLBEF2 or ILLBEF_EXTRA. If a mother has pregestational diabetes this should always be entered in variable 64 (MATDIAB). Historic maternal diseases may be coded using the Z-chapter in ICD10, example Z853 "Personal history of breast cancer". Any additional details may be entered in the general comments section (variable 103). Do not insert the decimal point in the code (e. g. Code E05.0 as E050) Examples, non-exhaustive list: Hyperthyroidism E050 - E059 Hypothyroidism E000 - E039 Diabetes Type 1 E100 - E109 Diabetes Type 2 E110 - E119 Obesity E660 - E669 If maternal BMI ≥ 30 give code for obesity 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 COVID-19 (only report if within 3 months before pregnancy): B342 (previously U071 and U072 were used).	1 = Yes, but no information available 9 = Not known
64	ILLBEF2	ILLNESS BEFORE PREGNANCY 2 - AS FOR ILLBEF1	
65	ILLBEF_EXTRA	OTHER ILLNESS(ES) BEFORE PREGNANCY This field is only to be used if "illnesses before pregnancy" fields 1 and 2 have already been filled, to record additional maternal illnesses before pregnancy. Follow the coding instructions of ILLBEF1. Please enter the ICD code in the following format: <ICD code> If more than one additional illness before pregnancy is to be reported for a single case, then enter the ICD codes in the ILLBEF_EXTRA field as follows: <ICD code><ICD code>	

Exposure (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
		For example, a case with hypothyroidism and depression exposure is entered in the ILLBEF_EXTRA field as: <E000><F320>	
66	MATDIAB	MATERNAL PREGESTATIONAL DIABETES This variable is specifically for pregestational diabetes. Gestational diabetes is dealt with under the 'illness during pregnancy' variable (variable 65) Type 1 diabetes: characterized by hyperglycaemia due to an absolute deficiency of the insulin hormone produced by the pancreas An HbA1c of 48mmol/mol is recommended as the cut-off point for diagnosing diabetes. Type 2 diabetes: characterized by hyperglycaemia due to a defect in insulin secretion An HbA1c of 48mmol/mol is recommended as the cut-off point for diagnosing diabetes. *Maturity Onset Diabetes in the Young (MODY) displays an autosomal dominant pattern of inheritance An HbA1c of 48mmol/mol is recommended as the cut-off point for diagnosing diabetes. Impaired Glucose Intolerance is a state of higher than normal blood (or plasma) glucose concentration, but less than the diagnostic cut-off for diabetes. Diagnosed before pregnancy. Diagnosed by fasting plasma glucose from 6.1 - 6.9 mmol/L (WHO criteria)	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
67	ILLDUR1	ILLNESS DURING PREGNANCY Record maternal 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. ILLDUR1 and ILLDUR2 does NOT imply the disease in ILLDUR1 is more important/severe than the disease in ILLDUR2 or ILLDUR_EXTRA. This variable aims to capture the maternal illnesses that may affect fetal development. 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). Examples (non-exhaustive list): Gestational diabetes O244 - O249 Haemorrhage early pregnancy O200 - O209 Hyperemesis O210 - O219 Fever R502 - R509 Hypertension I100 - I159	ICD 10 0 = No 1 = Yes, but no information available 9 = Not known

Exposure (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
		Depression F320 - F339 CMV B250 - B259 HIV (AIDS) B200 - B249 Influenza J100 - J119 Mumps B260 - B269 Rubella B060 - B069 Syphilis A530 - A539 Toxoplasmosis B580 - B589 Varicella (Chickenpox) B010 - B019 Viral hepatitis B190 - B199 Zika virus A925 Drug poisoning T360-T509 COVID-19 B342 (previously U071 and U072 were used).	
68	ILLDUR2	ILLNESS DURING PREGNANCY AS FOR ILLDUR1	
69	ILLDUR_EXTRA	EXTRA ILLNESS DURING PREGNANCY This field is only to be used if "illnesses during pregnancy" fields 1 and 2 have already been filled, to record additional maternal illnesses during pregnancy. Follow the coding instructions of ILLDUR1. Please enter the ICD code in the following format: <ICD code> If more than one additional illness during pregnancy is to be reported for a single case, then enter the ICD codes in the ILLDUR_EXTRA field as follows: <ICD code><ICD code> For example, a case with CMV and depression exposure is entered in the ILLDUR_EXTRA field as: <B250><F320>	
70	FOLIC_G14	FOLIC ACID SUPPLEMENTATION Recommend to your local maternity hospitals or midwives to collect these data. Folic acid supplementations include folic acid only tablets, a multivitamin preparation which contains folic acid or contraceptive pills which contain folic acid. If the folic acid dose is high (≥ 4 mg), please add the code B03BB01 in the drugs variable.	1 = Folic acid taken pre- and post-conceptionally 2 = Folic acid taken only post-conceptionally 3 = Folic acid not taken 4 = Folic acid taken, timing unknown 9 = Not known if folic acid taken
71	FIRSTTRI	FIRST TRIMESTER MEDICATION "Yes" means that the data sources clearly state that medication was taken in the first trimester. "No" means	1 = Yes, medication taken in first trimester

Exposure (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
		that the data sources clearly state that no medication was taken in the first trimester. Medications taken in the 2nd or 3rd trimester only should not be added to the DMS. "Medication taken but timing unknown" means that the usual data sources stated that medication was taken but the timing of use was not stated. "Not Known" means that the usual data sources were not found. Only fill in DRUGS1-5 and EXTRADRUGS if you have coded FIRSTTRI = 1 (Yes medication taken) or = 4 (Medication taken, but timing unknown). If you have coded FIRSTTRI = 2 (no medication taken), FIRSTTRI = 9 (unknown), there shouldn't be any ATC codes in any of the DRUGS variables. • Include any medication that was taken by the mother during the first trimester of pregnancy (from the 1st day of the last menstrual period up to the 12th week of gestation). Medication with long elimination half time and taken before conception should be included (e.g. 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, but if the folic acid dose is high (≥ 4 mg), please register FIRSTRI = 1 • Do not include usual vitamins and mineral supplementation, but include unusual intakes of vitamins or minerals (e.g. Vitamin A mega doses). • Only medication taken at physiologic doses should be included. Whilst FIRSTTRI is a new variable introduced 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. *Note: A code "3=undetermined" was in use until birth year 2022. It has been discontinued but there are cases with the code =3 in the database for some registries. It is advised to recode the variable as "9=Not know" when analysing the data.*	2 = No medication taken in first trimester 4 = Medication taken, but timing unknown 9 = Not Known
72	DRUGS1	DRUGS – 7 DIGITS MAXIMUM Record any drug 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). Drugs with long elimination half time and taken before conception should also be recorded (e.g. Acitretin, Etretinate, etc.). If it is not known in which trimester the drug was taken, and this information cannot be obtained, code it but write	

Exposure (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
		in the space for comments that it is not sure whether the drug was taken in the first trimester. Use ATC-coding and use as many digits as possible (from 3 to 7). Website http://www.whocc.no/atcddd/. Do not record usual vitamins and mineral supplementation, but record unusual intakes of vitamins or minerals (e.g. Vitamin A mega doses, folic acid dose ≥ 4 mg). The ATC coding system does not have a code for alternative drugs or herbs. If these are used, give the main code Z. ATC example: N03A: antiepileptic drug N03AF01: carbamazepine Details on the dosage and timing should be given in text variable 71. Do not forget to mention in the appropriate section (disease during or before pregnancy) the indication for drug use. Only drugs taken at physiologic doses to be recorded. If a drug overdose or self-poisoning, this MUST be explained in the drug description.	
73	SP_DRUGS1	SPECIFY DRUG EXPOSURES	Free text
74	DRUGS2	AS FOR DRUGS1 Please give details in text variable 73 SP_DRUGS2.	As for DRUGS1
75	SP_DRUGS2	SPECIFY DRUG EXPOSURES	Free text
76	DRUGS3	AS FOR DRUGS1 Please give details in text variable 75 SP_DRUGS3.	As for DRUGS1
77	SP_DRUGS3	SPECIFY DRUG EXPOSURES	Free text
78	DRUGS4	AS FOR DRUGS1 Please give details in text variable 77 SP_DRUGS4.	As for DRUGS1
79	SP_DRUGS4	SPECIFY DRUG EXPOSURES	Free text
80	DRUGS5	AS FOR DRUGS1 Please give details in text variable 79 SP_DRUGS5.	As for DRUGS1
81	SP_DRUGS5	SPECIFY DRUG EXPOSURES	Free text
82	EXTRA_DRUGS	EXTRA DRUGS This field is only to be used if drug fields 1-5 have already been filled. Record any drug taken by the mother during the first trimester of pregnancy (from the 1st day of the last menstrual period up to the 12th week of gestation). Drugs with long elimination half time and taken before conception should also be recorded (e.g. Acitretin, Etretinate, etc.).	

Exposure (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
		If it is not known in which trimester the drug was taken, and this information cannot be obtained, code it but write in the space for comments that it is not sure whether the drug was taken in the first trimester. Use ATC-coding and use as many digits as possible (from 3 to 7). Website http://www.whocc.no/atcddd/. Do not record usual vitamins and mineral supplementation, but record unusual intakes of vitamins or minerals (e.g. Vitamin A mega doses). The ATC coding system does not have a code for alternative drugs or herbs. If these are used, give the main code Z. ATC example: N03A: antiepileptic drug N03AF01: carbamazepine Details on the dosage and timing should be given in the drug description. Do not forget to mention in the appropriate section (disease during or before pregnancy) the indication for drug use. Only drugs taken at physiologic doses to be recorded. If a drug overdose or self-poisoning, this MUST be explained in the drug description. Please enter the ATC code and text description in the following format: <ATC code|text description> If more than one extra drug is to be imported for a single case, then enter the ATC codes in the extra drugs field as follows: <ATC code|text description><ATC code|text description> For example a case with valproate and lamotrigine exposure is entered in the extra_drugs field as: <N03AG01|Valproate><N03AX09|Lamotrigine>	
83	INF_COV_TEST	COVID-19 INFECTION STATUS OF THE MOTHER (PCR OR ANTIGENIC TEST) Use this variable to record the COVID-19 infection status of the mother obtained from a PCR or antigenic test only This field should be completed for all mothers registered since the pandemic outbreak of COVID-19 in 2020.	0 = No test 1 = Test positive 1st trimester 2 = Test positive 2nd trimester 3 = Test positive 3rd trimester 4 = Test negative

Exposure (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
		In the event of a mother being tested multiple times record the time of the first positive PCR or antigenic test in pregnancy.	9 = Not known if tested or test result not known
84	IMM_COV_TEST	COVID-19 IMMUNITY STATUS OF THE MOTHER (IGM ANTIBODY TEST) Use this variable to record the COVID-19 immunity status of the mother obtained from IgM antibody test This field should be completed for all mothers registered since the pandemic outbreak of COVID-19 in 2020. In the event of a mother being tested multiple times record the time of the first positive IgM antibody test	0 = No test 1 = Test positive 1st trimester 2 = Test positive 2nd trimester 3 = Test positive 3rd trimester 4 = Test negative 9 = Not known if tested or test result not known
85	OTH_COV_TEST	COVID-19 STATUS OF THE MOTHER (OTHER TESTS/EXAMS) Use this variable to record the COVID-19 status of the mother obtained from other tests/exams for diagnosis or confirmation of COVID-19 This field should be completed for all mothers registered since the pandemic outbreak of COVID-19 in 2020. In the event of a mother being tested multiple times record the time of the first positive test	0 = No test 1 = Test positive 1st trimester 2 = Test positive 2nd trimester 3 = Test positive 3rd trimester 4 = Test negative 9 = Not known if tested or test result not known
86	SP_OTH_COV_TEST	SPECIFY OTHER COVID-19 TESTS Specify the other tests (OTH_ COV_TEST) referred to above.	Free text
87	START_COV	START WEEK OF COVID-19 INFECTION Week of gestation at which the COVID-19 infection is considered to have started.	99 = Not known
88	COV_SEVERITY	SEVERITY OF COVID-19 INFECTION Use this variable to record the severity of COVID-19 infection in the mother.	0 = Asymptomatic 1 = Treated at home 2 = Hospitalised, no ICU 3 = Hospitalised in ICU 9 = Not known

Family History (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
89	CONSANG	CONSANGUINITY Restrictive definition of consanguinity: where the parents of the malformed case have one or more ancestors in common no more remote than a great-grandparent (=second cousins).	0 = Not related or relationship more distant than second cousin 1 = Relationship of second cousin or closer 9 = Not known
90	SP_CONSANG	SPECIFY TEXT INFORMATION ON CONSANGUINITY	Free text
91	SIBANOM	SIBS WITH ANOMALIES If the sibling (including twin) was notified to EUROCAT fill in variables 91-94 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 90 SP_SIBANOM.	1 = Same 2 = Other 3 = Same and other 4 = No 9 = Not known
92	SP_SIBANOM	SPECIFY TYPE OF ANOMALY OF SIBLINGS	Free text
93	PREVSIB	PREVIOUS MALFORMED SIBLINGS NOTIFIED TO EUROCAT If yes, give the local ID number in variables SIB1, SIB2 or SIB3 (variables 92-94). 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
94	SIB1	SIB LOCAL ID NUMBER NOTIFIED TO THE CENTRAL REGISTRY Enter here also the code numbers of co-twins or siblings from the same pregnancy, irrespective of birth order within multiple sets. Leave blank if no previous siblings notified to EUROCAT.	Local ID
95	SIB2	AS SIB1	Local ID
96	SIB3	AS SIB1	Local ID
97	MOANOM	MOTHER'S FAMILY WITH ANOMALIES Include mother herself as well as mother's family. Specify type of anomaly in written text and relation to the infant. If the aetiology is known, "same" means the same aetiology, even if the spectrum of malformations present is slightly different.	1 = Same 2 = Other 3 = Same and other 4 = No 9 = Not known

		If the aetiology is unknown or multifactorial, "same" is a matter of judgment by a qualified coder, but full specification of the anomaly should be given, whether other or the same. "Same and other" refers to two different relatives. If a relative has both the same and another anomaly, code "same". Restrict the family to first, second and third degree relatives (mother, father, siblings, grandparents, aunts, uncles, half-siblings, first cousins). Always give details in text variable 96 SP_MOANOM.	
98	SP_MOANOM	SPECIFY TYPE OF ANOMALY AND DESCRIBE THE MALFORMATION	Free text
99	FAANOM	FATHER'S FAMILY WITH ANOMALIES As MOANOM Please give details in text variable 98 SP_FAANOM	As MOANOM
100	SP_FAANOM	SPECIFY TYPE OF ANOMALY AND DESCRIBE THE MALFORMATION	Free text

Sociodemographic (core variables shaded blue)			
Variable Number	Variable Name	Explanation and Instructions	Code
101	MATEDU	MATERNAL EDUCATION Refer to International Standard Classification of Education 2011 (http://uis.unesco.org/sites/default/files/documents/international-standard-classification-of-education-isced-2011-en.pdf). Assign according to the highest level of education completed (or for full-time students, level in progress). Elementary and lower secondary refers to the period of compulsory education, usually to age 15/16. Upper secondary refers to the last two school or college years (usually to age 18) preparing students for tertiary education or the workforce. Tertiary refers to Bachelor's degree (English), Diploma (German), License (French) or equivalent, and to higher degrees (e.g. doctorates), or to other forms of higher education.	1 = Elementary and lower secondary 2 = Upper secondary 3 = Tertiary 9 = Not known
102	SOCM	SOCIOECONOMIC STATUS OF MOTHER Current or last occupation. Upper non-manual – professionals, administrators and managers e.g. doctor, architect, lawyer, banker, manager, teacher, nurse, performer. Lower non-manual – routine non-manual e.g. book-keeper, salesman, receptionist, secretary, computer operator, clerk, waiter. Skilled manual – cook, butcher, carpenter. Unskilled manual – semi and unskilled manual e.g. factory worker, driver, agricultural worker, porter. Self-employed/artisan – owner of shop, restaurant or hotel, independent artisan. Farmer – e.g. self-employed farmer or fisherman. If code 8 ("other/student"), please specify in text in space for general comments (variable 103). For further information see: http://uis.unesco.org/sites/default/files/documents/international-standard-classification-of-education-isced-2011-en.pdf	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
103	SOCF	SOCIOECONOMIC STATUS OF FATHER As SOCM.	0 = Single mother, no father recorded 1 = Upper non-manual 2 = Lower non-manual 3 = Skilled manual 4 = Unskilled manual

			5 – Self-employed/artisan 6 = Farmer 8 = Other/student 9 = Not known
104	MIGRANT	MIGRANT STATUS This variable is included to allow assessment of the extent to which services such as prenatal screening are reaching migrants. It does not ask for ethnicity. If code 4, give text details in the general comments section (variable 103).	1 = Mother migrated from outside EU during pregnancy 2 = Mother migrated from outside EU during adult life (from age 18) 3 = Mother not a migrant as defined in 1 or 2 4 = Other (specify in text) 9 = Not known

General Comments (core variables shaded blue)

Variable Number	Variable Name	Explanation and Instructions	Code
103	GENREM	GENERAL ADDITIONAL COMMENTS	Free text

Last update: 24.09.2026