2.2.3 Recommended Local Variables

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

The list below includes variables that previously have been included in the standard set of variables for EUROCAT

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

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

Examples for C

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

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

Examples for T

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

Thalidomide	
Thyroid disease requiring drug treatment	
Vitamin A analogues	
Warfarin	

Examples for G

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

Examples for U

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

Examples for I

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

Examples for M

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