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

Member Registries should:

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

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

The attached questionnaire has been designed to allow you to fill it in directly on your computer and to subsequently return it by email. Please give as much detail as possible. If you have any problems with the completion of any of the questions, please do not hesitate to contact Central Registry.

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

- The questions are navigated by using the tab or arrow up/down keys on your keyboard.
- Tick boxes can be selected (☐) by clicking once on the left mouse button. Boxes can be unselected (☐) by repeating this process.
- Text can be typed into the rectangular grey shaded boxes. These boxes will expand to accommodate the text inserted. Answer the questions in detail - use as much space as needed.

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

EUROCAT REGISTRY DESCRIPTION QUESTIONNAIRE

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

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

A CONTACT INFORMATION

Date dd/mm/yy ☐

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

A2 **Name of Registry Leader** ☐

A3 **Registry address** ☐

A4 **Registry telephone number** ☐

A5 **Registry fax number** ☐

A6 **Registry email address** ☐

A7 **Registry web home page** ☐

B REGISTRY ORGANISATION

B1 **History of registry**

Year of establishment	☐
Year started collecting data	☐
First birth year collected	☐
Birth year from which you will send data to EUROCAT (Full and Associate Only)	☐

Membership of other international organisations
☐ none

☐ ICBDMS	Since year ☐
☐ ENTIS	Since year ☐
☐ OTIS	Since year ☐
☐ Other, name ☐	Since year ☐

B2 **Type of data to be supplied to EUROCAT** Please tick appropriate box
Full member: unidentifiable case data ☐
Associate member: aggregate data (ie. numbers of cases) ☐

B3 **Organisation of Registry (present status)**
(answer all A, B and C)

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

Further Information ☐

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

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

B4 **Main aims of the Registry at present**
(indicate importance by scoring:
1 = Very important
2 = Less important
3 = Not important)

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

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

C POPULATION COVERAGE

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

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

Choose only one box below. The definition refers to both malformed and denominator births
Delivery = LB (normal + malformed) + SB (normal +malformed) + TOPFA

☐ Population-based I ☐ % mothers delivering outside registry area *
☐ Population-based II ☐ % non-resident mothers delivering within registry area *
☐ Population-based III ☐ % non-resident mothers delivering within registry area *
☐ Hospital-based ☐ % deliveries in defined geographic area (specify below) occurring in the selected hospitals *

☐ Other, specify below

other ☐

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

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

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

If no, specify
Date ☐ Type of change, detail ☐
Date ☐ Type of change, detail ☐
Date ☐ Type of change, detail ☐
Date ☐ Type of change, detail ☐
Date ☐ Type of change, detail ☐

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

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

% covered by Registry ☐

C6 **Further information** ☐

D SOURCES AND ASCERTAINMENT

D1 Notification to the Registry is

☐ Voluntary
☐ Compulsory
☐ Other

Required by which law? ☐

Further information ☐

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

☐ notification of cases to the Registry by hospitals and other institutions
☐ active searching of patient notes by Registry staff
☐ active searching of hospital etc. discharge registers by Registry staff
☐ active searching of other local or national registers by Registry staff
☐ other, specify below
☐

D3 Sources of information of Registry

WHO?

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

0 = Not used as a source of information

1 = Occasional notification of malformed cases seen

2 = Virtually complete notification of all malformed cases seen

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

WHERE AND HOW?

Use the following score system:

1 = Registry routinely searches for new cases in their records

2 = Source notifies virtually all malformed cases seen to Registry

3 = Source occasionally notifies malformed cases seen to Registry

4 = Registry only consults this source for confirmatory or supplementary information about known cases

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

☐ Echocardiology labs
☐ Other registries

Specialised departments for:

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

General sources of health and civil registration records:

☐ Hospital discharge records
☐ Birth certificates
☐ Death certificates

☐ Other

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

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

How is this calculated? ☐

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

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

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

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

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

E STILLBIRTH AND EARLY FETAL DEATH

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

E2 Does Registry get notification of malformed cases among:

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

E3 If yes, does Registry get death certificates?

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

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

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

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

	All deaths	Malformed cases on Register	Year
Stillbirths	☐	☐	☐
Early fetal deaths	☐	☐	☐
Early neonatal deaths	☐	☐	☐
Infant deaths	☐	☐	☐
TOPFA	☐	☐	☐

Do the above autopsy rates refer exactly to your Registry population? ☐ Yes ☐ No

If not, explain ☐

Do specialist fetopathologists do most autopsies?

Stillbirths	☐ Yes	☐ No
Early fetal deaths	☐ Yes	☐ No
Early neonatal deaths	☐ Yes	☐ No
Infant deaths	☐ Yes	☐ No
TOPFA	☐ Yes	☐ No

F PRENATAL SCREENING AND TERMINATION OF PREGNANCY

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

If yes, since (year) ☐

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

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

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

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

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

G REGISTRY INFORMATION: DIAGNOSES

G1 **Does the Registry database contain diagnoses as text?** ☐ Yes ☐ No

If yes, what diagnostic language is used in Registry?

☐ English
☐ Other language, specify ☐

G2 Coding system of congenital anomalies?

- ☐ ICD10 original
- ☐ ICD10 national
- ☐ ICD10 BPA
- ☐ ICD9 original
- ☐ ICD9 national
- ☐ ICD9 BPA EUROCAT
- ☐ ICD9 Atlanta
- ☐ ICD9 BPA
- ☐ ICD8
- ☐ Other

If other, describe differences compared to ICD10 original ☐

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	☐	☐	☐	☐	☐
Assisted conception	☐	☐	☐	☐	☐
Illness before pregnancy	☐	☐	☐	☐	☐
Illness during pregnancy	☐	☐	☐	☐	☐
Folic Acid supplementation	☐	☐	☐	☐	☐
Drugs taken during first trimester	☐	☐	☐	☐	☐
Maternal education	☐	☐	☐	☐	☐
Social class of mother	☐	☐	☐	☐	☐
Social class of father	☐	☐	☐	☐	☐
Maternal migrant status	☐	☐	☐	☐	☐

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

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

H3 **Can you transmit to EUROCAT the data from Guide 1.4 relating to whether the case was prenatally diagnosed, the gestational age at prenatal diagnosis and whether prenatal investigation was carried out? If yes, please specify below:** ☐ Yes ☐ No

Tick if you can send the data for the following EUROCAT Guide 1.4 variables:
☐ Whether prenatally diagnosed
☐ Gestational age at diagnosis
☐ First positive prenatal test

Further information ☐

H4 **Please read the information on EUROCAT's data management program (EDMP) before answering this question:**
http://www.eurocat-network.eu/content/Section%202.4-%2027_Oct2016.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+TOPFA**
TOTAL								

* 20 weeks gestation and above ** Following prenatal diagnosis

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

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

Anomalies	LB Number	FD Number	TOPFA Number	Prevalence (LB+FD+TOPFA/ total births)
All anomalies				
Nervous system				
Neural tube defects				
Anencephalus and similar				
Encephalocele				
Spina bifida				
Hydrocephalus				
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				
Triscuspid 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				

Anomalies	LB Number	FD Number	TOPFA Number	Prevalence (LB+FD+TOPFA/ total births)
Cystic adenomatous malf of lung				
Oro-facial clefts				
Cleft lip with or without cleft palate				
Cleft palate				
Digestive system				
Oesophageal atresia with or without techeo-oesophageal fistula				
Duodenal atresia or stenosis				
Atresia or stenosis or other parts of small intestine				
Ano-rectal atresia and stenosis				
Hirschsprung's disease				
Atresia of bile ducts				
Annular pancreas				
Diaphragmatic hernia				
Abdominal wall defects				
Gastroschisis				
Omphalocele				
Urinary				
Bilateral renal agenesis including Potter syndrome				
Multi cystic renal dysplasia				
Congenital hydronephrosis				
Bladder exstrophy and/or epispadia				
Posterior urethral valve and/or prune belly				
Genital				
Hypospadia				
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				

Anomalies	LB Number	FD Number	TOPFA Number	Prevalence (LB+FD+TOPFA/ total births)
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				

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.

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