Chapter 1 Aims and objectives of EUROCAT

1.1 Congenital anomalies

EUROCAT adopts the WHO definition of congenital anomalies (WHO Birth Defect Surveillance. A manual for Programme Managers, 2014). According to this definition, major congenital anomalies are structural changes that have significant medical, surgical, social or cosmetic consequences for the affected individual, and typically require medical intervention. Examples include spina bifida, anencephaly, heart defects and orofacial clefts. Major anomalies account for most of the mortality, morbidity, and disability related to congenital anomalies.

Minor anomalies are structural changes that pose little or no significant health problem and tend to have limited social or cosmetic consequences for the affected individual. Therefore, cases with minor anomalies only are not included as EUROCAT cases. Minor anomalies are more common than major anomalies and can be a useful tool for clinicians to identify syndromes. Examples of minor anomalies include single palmar crease and clinodactyly (mild curvature of a finger).

1.2 Why register Congenital Anomalies?

There are four main reasons why congenital anomaly registries are established:

1. To provide essential epidemiological information on congenital anomalies in Europe.

   Having good baseline background data, which includes prevalence for all pregnancy outcomes for a wide range of major congenital anomalies is vital when evaluating possible environmental impacts, effects of policies, and identification of new teratogens.

2. 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. Registries are also used for genetic studies, and for research into the interaction of genetic and environmental factors in the etiology of congenital anomalies.

3. 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 pregnant women and children, or which may have atypical expertise or financial resources.

4. To provide a mechanism for the audit of prenatal screening practices.

   A registry can provide data on the proportion of cases with a congenital anomaly which were diagnosed prenatally, the proportion of positive prenatal screening results which were confirmed as cases of congenital anomaly, and the proportion of prenatally diagnosed cases that led to the 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.

1.3 How can a registry be used?

Whether concerned with the identification of teratogenic exposures, or with planning and evaluation of health services, or both, registries can be used in two main ways:

1. As a basis for surveillance using routinely collected data: Every registry routinely collects a core dataset of standard information on each child with congenital anomaly/-ies. Some registries collect also information about children without congenital anomalies.
2. As a basis for special or ad-hoc studies, such as case-malformed control studies, which may require further data collection: A registry 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.

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

There are useful approaches to analysing risk factor data among congenital anomaly 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.

1.4 Why European collaboration?

There is considerable added value of European collaboration for congenital anomalies, as it provides the opportunity to pool data (particularly relevant for rare anomalies), to compare data between regions and countries, to give a common response to European public health questions, and to share expertise and resources, including computing tools.

1.5 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.