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## **Congenital anomalies in Switzerland: Problems and solutions in data collection and their European integration experiences by Eurocat (Switzerland)**

Information about prevalence of congenital anomalies is one of the fundamental parameters of public health and vital statistics. The monitoring of its variation became even more important in the world of environmental disasters such as Seveso, Chernobyl or Schweizerhalle. Except individual hospital based studies no population based epidemiology study was existing in Switzerland prior to 1987. Thanks to a joint initiative of the Swiss Academy of Medical Sciences and the Swiss Society of Pediatrics a working group (U. Ackerman, J. Brinner, P. Extermann, T. Pexieder, A. Schnizel, T. Spühler and F. Stocker), headed by Prof. G. Duc, has recommended the pilot introduction of the Eurocat (European registry of congenital anomalies and twins) procedure for birth defects monitoring.

Using a standardized one page questionnaire, developed and used by 19 other European Eurocat registries, congenital anomalies are recorded during the first month of life by region (canton) based neonatologists who report them anonymously to the Swiss Eurocat coordination centre in Lausanne. In Lausanne the reports are checked for completeness and logical consistency and computer processed. Table 1 shows the dates of adhesion

of individual cantons to the Eurocat procedure, the number of births monitored and the name of the collaborating neonatologist or pediatrician. From 40% coverage of all deliveries in Switzerland in 1988 the system has grown up to a 90% coverage in 1991. Actually only 3 cantons (Thurgau, Solothurn and Schaffhausen), totalizing 6477 births out of annual 70000, do not yet have an Eurocat neonatologist. The computerized database contains at present data on 4719 children with birth defects born between 1987 and 1993. In 84.96% these are liveborn children. 2.97% are stillborn; 4.76% of cases came from spontaneous abortions and 7.30% from selective terminations of pregnancy.

For the purposes of monitoring, and to create a baseline against which eventual modification by e.g. environmental risk factors might be evaluated, it is necessary to achieve a very high degree of ascertainment. The level of ascertainment and the absence of its variations over time belongs to fundamental prerequisites to be achieved before any comparisons of regional (cantonal) prevalence of birth defects might be done. To estimate the degree of ascertainment we have calculated, as "gold standard", the expected prevalence

of all and individual malformations based on Eurocat (Bruxelles) data stratified by the period of data collection (first 30 days of life), birth status and center size.

Comparing the abnormal with the expected ("gold standard") prevalence of various defects has shown that there is a run-up time of at least two years before a reliable figures can be produced. This is illustrated in Figure 1 for chromosomal aberration in two regions, Zürich and Vaud. As in the everyday practice there is always a delay between the birth of a malformed baby, its discovery by collaborating neonatologist and the final reporting to the coordinating center and the computer processing, we have decided to consider for further discussion only the cases born during the period 1989–1991 i.e. from the second year of the regional reporting and one year before the reporting end point (1992).

Comparing the global prevalence of congenital anomalies with the European stratified average using the 95% confidence limits, shows (Figure 2) that the best ascertainment has been achieved in canton Vaud. Degree of ascertainment does not differ significantly from the European average in canton Valais, Neuchâtel, Graubünden and Jura. However

Canton	Data start registration	Annual number of deliveries	EUROCAT delegate
Aargau	01.03.91	5'865	Dr. Wopmann Dr. Hindermann
Baselland	01.05.87	2'532	Dr. Olafsson
Basel-Stadt	19.05.87	1'797	Prof. Nars
Bern	14.08.89	10'465	Prof. Bossi Dr. Biggler Dr. VonKänel Dr. Kindler Dr. Rotter
Fribourg	01.07.88	2'519	Dr. Ruckstuhl
Genève	01.09.91	4'280	Dr. Roch-Suzuki
Graubünden	01.03.90	2'166	Dr. Vischer Dr. Knoblauch Dr. Gambon Dr. Jörmann
Jura	12.05.89	821	Dr. Nicolet
Luzern (Nidwald, Obwald, Schwyz, Uri, Zug)	01.03.90	4'084	Dr. Schubiger
Neuchâtel	28.10.88	1'635	Dr. Pilloud
St. Gallen	01.01.91	5'336	Dr. Kind Dr. Sulmoni
Ticino	01.01.88	2'463	Dr. Tonella
Valais	03.03.89	2'996	Dr. Marcoz
Vaud	22.05.88	6'154	Dr. Addor
Zürich	01.02.88	13'398	Prof. Duc Prof. Schinzel Dr. Brändle Dr. Bühlmann Dr. Hunziker Dr. Sigg

**Table 1.** List of cantons taking part on EUROCAT(Switzerland). Are also indicated: Date of start of registration, annual number of deliveries and the name of the local delegate.

there is some degree of under-reporting in Tessin, Fribourg, Luzern, Basel-Land, Basel-Stadt and Zürich.

Close up analysis of individual categories of birth defects shows however, that many of them (congenital heart disease, malformations of internal urinary system, malformations of the gastrointestinal tract, cleft lip, cleft palate and ear anomalies) have a prevalence in Switzerland that is not significantly different from their preva-

lence in other european countries. Major underreporting has been observed for limb anomalies, with the exception of the limb reduction deformities.

Regional variations in the observed prevalence of selected birth defects with a reasonable degree of ascertainment is displayed in Figure 3. When analyzing this graph one should not lose sight that at present stage of the study the observed variations represent a mixture of true preva-

lence differences, with bias due, in part, to variations in the ascertainment. Special statistical treatment of the data will be necessary to separate the true from artificial differences.

As an example of analysis of the various kinds of data collected we can mention the maternal age. Whereas in the majority of cantons there is no statistically significant difference between the age of mothers of normal and malformed babies, in the canton of Tessin and Luzern (plus central part of Switzerland) the mothers of newborns with malformations are older than mothers of normal babies.

In the study, as it is presently operated in Switzerland, more than 60% of all cases are diagnosed at delivery, 15% during the first week of life and 9% during the first month of life. 14% of all malformations recorded were discovered by prenatal diagnostic procedures. 2% of cases were observed during autopsy.

As far as the prenatal diagnosis of congenital anomalies is concerned there are also regional variations. For instance 21% of cases with congenital heart disease were discovered by prenatal diagnosis in Zürich, against 6% in Vaud. For internal urinary system malformations, Luzern and central Switzerland had a prenatal detection rate of 90% against 35% in Zürich.

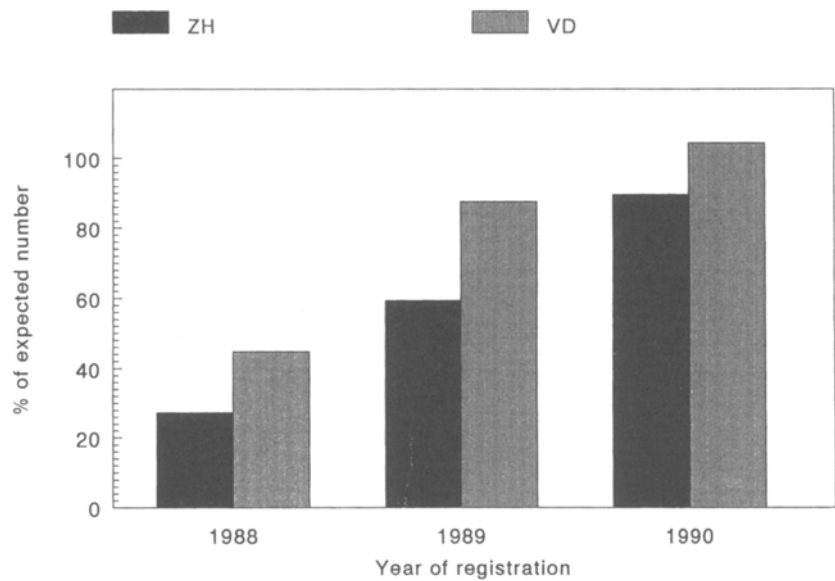
The analyses of the four years of operation and development of Eurocat (Switzerland) show that it is much more difficult to obtain a good ascertainment in large cantons with many maternity hospitals and physicians such as Zürich or Bern.

One of the reasons is the problem of obtaining satisfactory anamnestic data about the parents. This is why we have decided, to improve the degree of ascertainment in these two regions, to request only a minimal set of data concerning essentially the malformed baby, which can be provided more easily

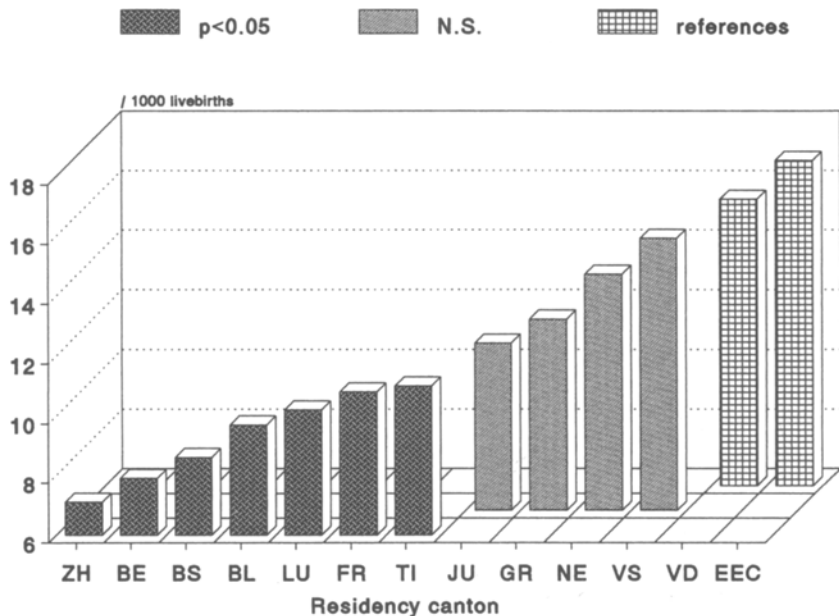
by the collaborating neonatologists or paediatricians. We have also speeded up the data flow and processing, by requesting immediate on the spot, instead of batch, reporting, so that eventual variations in prevalence will be detected more rapidly. The example of canton Vaud illustrates that most successful is the system where a physician whose principal, and not accessory, task is birth defects registration, searches actively various sources of information from delivery room records to echocardiography notebooks of pediatric cardiologists.

The future development of Eurocat (Switzerland) should consist in the extension of the surveyance period before and beyond the immediate perinatal period. In collaboration with the Swiss Society of Gynecology and Obstetrics we are seeking to introduce monitoring of the prevalence of spontaneous abortions occurring during the 8th to 11th week of gestation, a very sensitive and precious indicator of exposure to new teratogens. As some anomalies can be detected only later in life, extension of the registration up to the end of the first year of life is also planned. In the meantime statistical techniques for cleaning up of the collected data will have to be developed to allow to reach the final goal which is the answer to the question whether all regions of Switzerland have the same prevalence of birth defects and if not, to initiate special studies for detecting the causes of regional variations e.g. between the town and the country.

Finally we have to mention that Eurocat (Switzerland) collaborates closely with the Swiss Teratogen Information System (STIS) providing counselling on reproductive toxicology and risks and collecting prospectively the information on cases of teratogen exposure. STIS is member of ENTIS (European Network of Teratology Information Services).



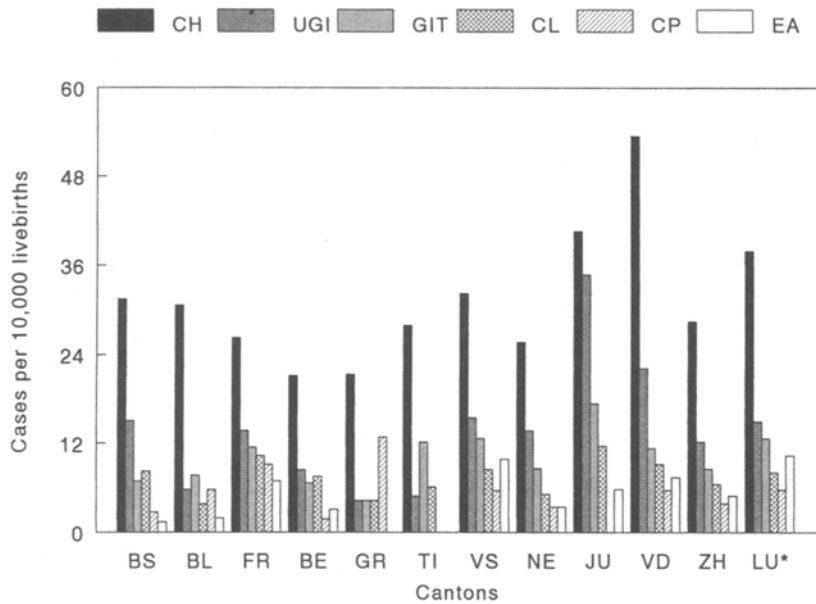
**Figure 1.** EUROCAT-Switzerland 1988–1990. Ascertainment of chromosomal aberrations.



**Figure 2.** EUROCAT (CH). Prevalence of congenital anomalies.

In conclusion we may state that there are, each year, about 1200 more or less severely handicapped children born in Switzerland, a situation similar to other European countries. The ultimate aim of a system such as Eurocat is to contri-

bute to the prevention of this source of emotional burden to the parents and public health costs to the society.



**Figure 3.** Abbreviations used.

CH = congenital heart disease, UGI = internal urogenital, GIT = gastro-intestinal tract, CL = cleft lip, CP = cleft palate, EA = ear anomalies.

**Figure 3.** Prevalence of major congenital anomalies. Eurocat-Switzerland 1989–1991.

### Acknowledgements

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### References

- 1 Lechat, M, Eurocat Working Group: EUROCAT Report 3- Surveillance of congenital anomalies. Years 1980–1986. 231 p. Dept. of Epidemiol. , UCL: Brussels, 1989.