Research report from EpC

Registration of Congenital Malformations in the Swedish Health Registers

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Preface

One of the principal functions of the Centre for Epidemiology (EpC) at the Swedish National Board of Health and Welfare is to analyse and report on the national population's state of health. EpC is responsible for several nationwide registers of medical data including information on congenital malformations.

This report is intended primarily for those who use or plan to use information recorded in various registers regarding congenital malformations. Professor Bengt Källén of the Tornblad Institute at the University of Lund wrote the report.

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Registers

Information on congenital malformations observed among new-born infants can be obtained from the following registers, kept by the National Board of Health and Welfare, Stockholm:

The Medical Birth Register where information on malformations is obtained from ICD (International Classification of Diseases) codes given to the new-born at the paediatric examination, or from neonatal units. Data are taken from copies of the medical documents kept at the delivery unit.

The Register of Congenital Malformations is an alarm register based on compulsory reporting of infants with relatively severe malformations, observed during the first few weeks after birth. Malformations should be reported verbatim and reports can be supplemented with marks on a drawing of a new-born baby, and also with copies of, e.g., autopsy reports. Up to and including 1998, malformations were coded with a special and detailed code and also with ICD codes. Since 1999, only ICD codes have been used. Sweden changed from ICD9 to ICD10 during 1997.

Up to 1998, cardiac defects diagnosed before the age of one were reported to the Register of Congenital Malformations from the child cardiology clinics in Sweden. Cardiac defects were classified with the ISC (International Society of Cardiology) codes, which are very detailed. From 1999 onwards, reports of cardiac defects are included in the Register of Congenital Malformations and have been coded with ICD codes.

Up to 1998, reports from the cytogenetic laboratories were sent in to the National Board of Health and Welfare and a **Register of Cytogenetic Diagnoses** was formed. This Register, however, was restricted to autosomal conditions and, from 1999 onwards, such reports have been incorporated in the Register of Congenital Malformations.

The Hospital Discharge Register contains discharge diagnoses for all children. Children with congenital malformations can be identified from these diagnoses, (based on ICD codes). Malformations not observed during the neonatal period (e.g., pylorostenosis) can also be identified, as can conditions such as cerebral palsy and mental retardation, or behavioural problems. While the latter groups are not covered completely (hospitalisation is needed) experience shows that a reasonable proportion can be identified.

Special registers include a Register of Visually Impaired Children with a visus <0.3.

These registers can be linked ad hoc using personal identification numbers (PIN). Every person living in Sweden has a unique, ten-digit PIN showing birth year, month and day, a three-digit birth number and a check digit. The PINs are extensively used throughout the community and in all health care. There is no total linkage of all registers.

The contribution of each register depends on the malformation studied and also varies with time.

Malformation prevalence

Medical Birth Register

The total annual number of births registered in the Medical Birth Register between 1995 and 2001 was:

Year	All births
1995	102,137
1996	94,733
1997	89,124
1998	85,883
1999	86,163
2000	89,368
2001	90,243

The Medical Birth Register employs three different marks for malformations:

- "Total", for *any* congenital malformation (i.e. with an ICD code from Chapter 14 in ICD9 and Chapter 17 in ICD10),
- "Significant", for some defined conditions of general clinical significance (all cardiac defects for instance, except PDA and SUA), and
- "Weeded", where a number of common and rather insignificant conditions have been removed from the register (e.g., preauricular tags, undescended testicles, unstable hips, SUA, PDA). With the change between ICD classifications, the definition of “significant” malformations changed somewhat. The rates of the different marks for malformations are given below:

Year	Malformation rates		
	Total	Significant	Weeded
1995	3.8	2.2	2.5
1996	3.6	2.1	2.4
1997	3.5	1.7	2.3
1998	3.4	1.7	2.2
1999	3.5	1.7	2.3
2000	3.3	1.6	2.2
2001	3.6	1.7	2.3
Total	3.5	1.8	2.3

There is a tendency towards a reduction in the rate of registered infants with congenital malformations. This is partly because data from neonatal units are often not sent to the delivery units in time for registration. Furthermore, it has been difficult to translate the specific conditions between the ICD codes editions. For all analyses of malformation rates in the register, adjustment for year of birth has to be made.

Register of Congenital Malformations

A table similar to that for the Medical Birth Register can be made for the Register of Congenital Malformations.

Year	Reported cases	Percentage of total number of births
1995	1076	1.1
1996	910	1.0
1997	869	1.0
1998	799	0.9
1999	1008	1.2
2000	1059	1.2
2001	1180	1.3

Especially before 1999, however, only part of these was used for malformation monitoring because of restriction to relatively severe conditions. From 1999 onwards, a number of less significant malformations have also been registered and, in addition, data from the two auxiliary sources (child cardiology clinics and cytogenetic laboratories) mentioned above have been included directly in the Register.

The Hospital Discharge Register

Year	Reported cases	Percentage of all registered births
1995	2373	2.3
1996	2113	2.2
1997	1915	2.1
1998	1808	2.1
1999	1608	1.9
2000	1456	1.6
2001	1152	1.3

The declining rate is probably related to a shorter follow-up time (to the end of 2001).

Specific malformations

Total malformation rates or rates of "severe" or "major" malformations in infants give only a crude picture and, furthermore, data from different registers cannot be compared. More useful is the "complete" registration, based at least on both the Medical Birth Register and the Register of Congenital Malformations and perhaps also on the other sources. Some such estimates have been made. Most are published in the literature, most based on only the first two registers. Some of the data therein refer to earlier years, but the rates change little, except for conditions detected largely at prenatal diagnosis (e.g., neural tube defects, abdominal wall defects).

Malformation	Rate per 10,000 births	Reference
Spina bifida	2.1	Unpublished data, 1995-2001
An/microphthalmia	0.92	Int J Epidemiol 25:1009. 1996
Cyclopia	0.09	J Med Genet 29:30, 1992
Anotia/microtia	2.35	J Med Genet 33:1, 1996
Cleft lip/palate	12.7	J Craniofac Genet 16:234,1996
Median cleft palate	6.5	J Craniofac Genet 16:234, 1996
Cardiovascular defect	92.0	Reprod Toxicol 17:255, 2003
Choanal atresia	0.54	Pediatrics 99: 363, 1997
Esophageal atresia	2.84	Teratology 52:15, 1995
Gastric atresia	0.02	Teratology 52:15, 1995
Duodenal atresia	1.08	Teratology 52:15, 1995
Small gut atresia	0.59	Teratology 52:15, 1995
Colon atresia	0.08	Teratology 52:15, 1995
Anal atresia	3.48	Teratology 52:15, 1995
Diaphragmatic hernia	2.27	Eur. J Epidemiol 13:665, 1997
Abdominal wall defects	3.04	Unpublished data, 1995-2001
Bilateral kidney Malformation	2.61	Eur. J Epidemiol 16:985, 2000
Unilateral kidney Malformation	1.62	Eur. J Epidemiol 16:985, 2000
Bladder extrophy	0.04	Teratology 36:221, 1987
Epispadias	0.01	Teratology 36:221, 1987
Hypospadias	20.0	Int J Risk Safety Med 14:115, 2001
Conjoined twins	0.06	Acta Genet Med Gemel 40:325, 1991
Preaxial reduction	7.73	Reprod Toxicol 11:653, 1997
Thumb polydactyly	4.41	Reprod Toxicol 11:653, 1997
Sirenomelia	0.08	J Med Genet 29:30, 1992
Skeletal dysplasia	0.92	Int J Epidemiol 22:107, 1993

One further source of tabulation, based on the Medical Birth Register and the Register of Congenital Malformations, is the reports produced by the National Board of Health and Welfare. In 1999 the following rates per 10,000, valid for 1998-1999, were given for specific types of malformation:

Malformation	Rate per 10,000 births	
anencephaly	0.1	
spina bifida	2.1	
encephalocele	0.6	
hydrocephaly	1.3	
an/microphtalmia	0.7	
anotia/microtia	1.9	
cleft lip/palate	12.2	
isolated cleft palate	7.5	
choanal atresia	0.7	
cong. heart defect	22.5	(only severe forms, from child cardiology register)
diaphragmatic hernia	2.0	
abdominal wall defect	1.6	
esophageal atresia	3.2	
small gut atresia other	1.9	(stenosis does not include secondary to conditions such as malrotation or duodenal band)
anal atresia	2.7	
kidney dys/agenesis	1.8	
cystic kidney	2.2	
hypospadias	22.1	
craniostenosis	1.1	
generalised skeletal dysplasia	1.2	
limb reductions	5.1	
polydactyly	9.0	
syndactyly	7.8	
positional foot def.	17.6	
unstable hip	53.7	
Down's syndrome	13.0	(incl. Reg. of Cytogenetic Diagnoses)

The above figures give a general idea. For any scientific analysis, it is vital to avoid the use of any kind of "normal" rate of congenital malformation, and to calculate the expected numbers with account taken of the sources, year of birth and perhaps other confounders.