Birth defects surveillance

A pilot system in the Cape Peninsula

A. R. SAYED, D. E. BOURNE, J. M. NIXON, J. M. L. KLOPPER, J. OP'T HOF

Summary

A pilot birth defects surveillance system was established in 1982 as part of an epidemiological baseline study pertaining to potential changes in water quality in the Cape Peninsula. The methodology used for reporting birth defects for two information systems, one hospital-based and the other population-based, utilising statutory notifications of births, is described. Preliminary birth defect rates by cause are presented for a hospital-based system and are consistent with rates reported internationally. The system based on statutory notifications showed gross under-reporting.

S Afr Med J 1989; **76:** 5-7.

A birth defects surveillance system enables the frequency of clinically observable defects in newborn populations to be monitored. Any temporal or spatial clustering will provide information in formulating hypotheses about causes of these defects by means of epidemiological investigations. Although birth defect epidemiology is a relatively new field, there are a number of population- and hospital-based birth defect surveillance systems in operation throughout the world.[1-3]

The rationale for beginning the monitoring of birth defects in the Cape Peninsula was based on the hypothesis that changes in water quality, as a result of the possible future introduction of recycled water for potable use,[4] could possibly be related to carcinogenicity and mutagenicity. Since the effects of maternal exposure to teratogenic agents are not easily identified, and in view of the long and unknown latent periods usually associated with such effects, it was thought that the incidence of birth defects might provide an early indicator of such effects.[5-7] It has been reported from South Australia that significant risk increases occurred specifically for defects of the central nervous and musculoskeletal systems as a result of differences in the composition of water supplies.[8]

Methodology

One difficulty that arises in attempting to determine the total incidence of all birth defects is that some defects, although believed to be present at birth, cannot be diagnosed until later in life and require careful and complex investigations. Therefore, the group of defects studied had to be limited to those that could be diagnosed clinically at birth. Hence only defects that can be seen clinically within the first 7 days of life were recorded. This pragmatic limitation is utilised by many such birth defects surveillance systems.[3]

The data collection procedure for the surveillance of birth defects was initially based on the following two pilot systems.

Population-based system

The study population of births for the population-based system was confined geographically to areas which fell under the jurisdiction of the Combined Health Scheme of the former Divisional Council of the Cape (DCC). Since the inception of the surveillance system, the 'early notification of birth' form used by the DCC has required the reporting of both birth defects and birth weights. Those birth notification forms that contained an indication of a birth defect or defects were referred directly by the DCC Health Department to the Genetic Services Division of the Department of National Health and Population Development (DNHPD) for possible follow-up and verification of the cases reported. This information was then supplied to the Department of Community Health, University of Cape Town, where the information was coded and entered into a database.

These data are no longer used for surveillance purposes because it was established that there was gross under-reporting of birth defects. This source of data none the less continues to be of use to the local authority and genetic services for the provision of clinical health care to the patients concerned.

Hospital-based system

The pilot hospital-based system was restricted to delivery institutions, namely the Peninsula Maternity and Neonatal Service (PMNS), which is comprised of 5 maternity hospitals and 3 midwife obstetric units (MOUs). The area drained by these institutions is not the same as in the population-based system. On examination of each newborn, a detailed paediatric summary sheet is filled in by members of the Department of Paediatrics and Child Health of the University of Cape Town. The information is coded and entered on the PMNS computerised database. Since all cases of congenital malformations on the PMNS summary sheet are classified according to anatomical systems rather than specific conditions, detailed descriptive information of the specific defects had to be extracted retrospectively (for the 2-year period 1984-1985) from the mothers' and infants' hospital records for the purpose of taxonomy; they were then coded in the Department of Community Health.

The coding system used for classifying birth defects originated with the State of California Birth Defects Monitoring Program.[9] It is based on the Centers for Disease Control (Atlanta) modification of the British Paediatric Association coding system. This 6-digit code provides for the encoding of specific diagnostic details and also for comparison with other surveillance systems. Because of incomplete coverage of stillbirths, defects among this group were not included in our analysis.

Department of Community Health, University of Cape Town
A. R. SAYED, B.SC. HONS, M.SC.
D. E. BOURNE, B.SC., B.PHIL.
J. M. NIXON, S.R.N., H.V.
J. M. L. KLOPPER, *O.B.E.*, M.B. CH.B., F.F.C.M., D.P.H.
Genetic Services Division, Department of National Health and Population Development, Pretoria
J. OP'T HOF, D.SC., M.S.A.

Accepted 28 Dec 1988.

Results

The reporting of birth defects in the population-based system revealed a comparatively low ascertainment rate and poor descriptive quality. For the period January - December 1984 an overall rate of 85,7/10 000 live births for all conditions was recorded out of a total of 17 484 live births. For the same period the hospital-based system (PMNS) provided a higher total ascertainment rate of 190,5/10 000 live births. For the study period (January 1984 - December 1985) the hospital-based birth defect rate for all conditions was 207,3/10 000 live births out of a total of 51 427 live births. Of the 1 118 infants identified as having congenital malformations in the PMNS computerised database, 52 (4,6%) hospital records were not accessible.

Table I shows birth defect rates with their 95% confidence intervals[10] from the PMNS hospital-based pilot study and rates from international monitoring systems for selected conditions. In comparing the PMNS rates with international hospital-based systems where stillbirths were also not included (Spain and Mexico), the ranges shown for the two countries overlap the 95% confidence intervals derived from the PMNS observed rates. The only exception was limb reduction defects, which were significantly lower than the rates reported for Spain and Mexico. At this stage there is no obvious explanation for this low figure since it is unlikely that this clinically obvious condition would be under-reported. However, on comparing the pilot study figure with the range shown for countries where stillbirths are included (Italy, Israel, Japan and the USA), the rates overlap the 95% confidence interval for each condition (Table I).

Discussion

A number of studies have indicated substantial under-reporting of birth defects based on statutory notifications of births compared with hospital records.[11-13] This tendency was also found to exist locally and a passive surveillance system based solely on routine birth notifications cannot be recommended.

The variation in the rates for the conditions specified in Table I can be attributed to the following factors: (*i*) exclusion of stillbirths in the PMNS and Mexican and Spanish hospital-based systems; (*ii*) varying autopsy rates of stillborns in international monitoring systems; (*iii*) variations in methods of data collection result in different levels of ascertainment; and (*iv*) the intensity of a search in specialised studies causes over-reporting of certain specific conditions.

The conditions listed in Table I (and Down syndrome) have been recommended for international comparison by the International Clearinghouse for Birth Defects Monitoring Systems.[3] Birth defect surveillance is not limited to these conditions only. They have been selected because they can be easily diagnosed at birth and, in general, there has been uniformity in their definition, thus minimising the effect of different diagnostic criteria. We have not included Down syndrome as we do not have sufficient data to express the rate of this defect by maternal age.

Since a considerable proportion of birth defects are found in stillborn infants, a detailed anatomical investigation of *all* stillbirths would greatly enhance the assessment of the situation in respect of causes in epidemiological investigations.

We conclude that the rates for the selected defects obtained locally in a hospital-based surveillance system are consistent with those reported internationally. More data are, however, required to reduce the statistical error in observed rates. The recording of birth defects based solely on statutory notifications showed gross under-reporting.

In view of the establishment of a national birth defects surveillance system by the DNHPD, it will be possible to compare data gathered from various centres and also to compare the ascertainment rate for selected conditions.

This study was supported financially by the Water Research Commission. We thank the former Director of Hospital Services of the Cape Provincial Administration, the Head of the Department of Paediatric and Child Health, University of Cape Town, and the Superintendents and Matrons of Groote Schuur, Somerset, St Monica's, Mowbray Maternity and Peninsula Maternity Hospitals for access to hospital records. Access to notification of birth forms was provided by the Medical Officer of Health for the Divisional Council of the Cape, Dr L. Tibbitt.

REFERENCES

1. Edmonds LD, Layde PM, James LM, Flynt JW, Erickson JD. Congenital malformations surveillance: two American systems. *Int J Epidemiol* 1981; **10:** 247-252.
2. Office of Population Censuses and Surveys. *Congenital Malformation Statisics 1971-1980.* (OPCS Series MB3 No. 1). London: HMSO, 1983.
3. International Clearinghouse for Birth Defects Monitoring Systems. *1985 Annual Report.* Sydney: ICBDMS, 1985.

TABLE I. SELECTED BIRTH DEFECTS — RATE PER 10 000 LIVE BIRTHS (PMNS AND INTERNATIONAL MONITORING SYSTEMS)

Defect	PMNS (95% CI)*	Spain*	Mexico*	Italy†	Israel†	Japan†	USA†
Anencephaly	**1,8 (0,8; 3,4)**	**3,0**	**3,8**	**2,3**	**4,5**	**4,2**	**5,2**
Spina bifida	**5,8 (4,0; 8,4)**	**3,9**	**11,4**	**3,8**	**4,5**	**2,7**	**7,2**
Hydrocephaly	**2,1 (1,1; 3,9)**	**1,7**	**4,4**	**3,6**	**3,4**	**2,4**	**4,7**
Cleft palate	**3,7 (2,3; 5,9)**	**5,0**	**2,9**	**5,2**	**3,9**	**6,1**	**5,3**
Total cleft lip	**5,1 (3,4; 7,5)**	**5,8**	**11,5**	**5,9**	**4,5**	**12,0**	**10,0**
Oesophageal atresia or stenosis	**1,4 (0,6; 2,9)**	**1,8**	**1,2**	**3,0**	**1,5**	**1,0**	**1,6**
Anorectal atresia or stenosis	**1,6 (0,7; 3,2)**	**2,1**	**2,9**	**3,4**	**3,2**	**5,4**	**3,6**
Hypospadias	**6,8 (4,8; 9,6)**	**18,3**	**4,0**	**19,2**	**25,8**	**1,7**	**21,7**
Limb reduction defects	**1,8 (0,8; 3,4)**	**6,4**	**5,4**	**6,3**	**2,9**	**6,1**	**3,1**
Omphalocele	**1,6 (0,7; 3,2)**	**1,4**	**1,6**	**2,1**	**2,2**	**3,4**	**—**

* Stillbirths not included: PMNS (1984-85); Spain (1978-83); Mexico (1980-84).
† Stillbirths included (20 weeks of gestation or over): Italy (1978-84); Israel (1978-84); Japan (April 1978 - March 1980); USA (1970-73).
CI = confidence interval.

4. Bourne DE, Sayed AR, Watermeyer GS, Klopper JML. *Epidemiological Studies Pertaining to the Possible Reclamation and Re-use of Purified Sewage Effluent in the Cape Peninsula* (Report No. WRC 74/1/86). Pretoria: Water Research Commission, 1987.
5. World Health Organization International Reference Centre for Community Water Supply. *Health Effects Relating to Direct and Indirent Re-use of Waste Water for Human Consumption* (Technical Paper Series No. 7). The Hague: WHO, 1975.
6. Water Pollution Control Federation. *Water Reuse: Manual of Practice SM-3 Systems Management*. Washington, DC: WPCF, 1983.
7. Hattingh WHJ, Bourne DE. Research on the health implications of the use of recycled water in South Africa. *S Afr Med J* 1989; **76:** 7-10 (this issue).
8. Dorsch M, Scragg RK, McMichael J, Baghurst A, Dyer F. Congenital malformations and maternal drinking water supply in rural South Australia: a case-control study. *Am J Epidemiol* 1984; **119:** 473-486.
9. California Birth Defects Monitoring Program. *Procedure Manual, Vol II: Classification of Birth Defects*. Berkeley, Calif.: California Department of Health Services, 1984.
10. Fleiss JL. *Statistical Methods for Rates and Proportions*. New York: John Wiley, 1981: 14.
11. Bock HB, Zimmerman JH. Study of selected congenital anomalies in Pennsylvania. *Public Health Rep* 1967; **82:** 446-450.
12. Hay S. Incidence of selected congenital malformations in Iowa. *Am J Epidemiol* 1971; **94:** 572-584.
13. Mackeprang M, Hay S. Reporting of congenital malformations on birth certificates. *Health Serv Res* 1972; **87:** 830-834.