Validity of death and stillbirth certificates and hospital discharge summaries for the identification of neural tube defects in Quebec City

Fassiatou Tairou, Philippe De Wals and Adrien Bastide

Abstract

The objectives of this study were 1) to assess the validity of different databases which identify neural tube defect (NTD) cases in the population, and 2) to examine the temporal trends in NTD rates and the impact of prenatal diagnoses among pregnancies referred to a tertiary care hospital in Quebec City, Canada, from 1993 to 2002. Infant death and stillbirth certificates were a highly reliable source for ascertaining NTD cases, but their overall sensitivity was poor (13%). Med-Echo had very good sensitivity (92%), but there were many coding errors in the database and some diagnostic categories were not specific for NTD. The average NTD prevalence proportion was 6.5/1,000 births during the entire study period, decreasing from 12.2/1,000 in 1993 to 3.9/1,000 in 2002. Overall, 78.6% of NTD cases were diagnosed prenatally and the pregnancy was terminated in 52.6% of these. These two proportions were stable over the study years. To conclude, the combination of hospital discharge summaries and infant death and stillbirth certificates is a highly sensitive method for the ascertainment of NTD cases, including terminations of pregnancies, but medical records must be reviewed to exclude coding errors and to clarify unspecific diagnostic categories.

Key words: *ascertainment, birth defects, database, neural tube defects, prenatal diagnosis, surveillance, validity*

Introduction

In Canada, recommendations on the use of folic acid supplements by women planning a pregnancy or capable of becoming pregnant were issued in 1993–1994 by Health Canada, the Society of Obtetricians and Gynaecologists of Canada and the Canadian Task Force on the Periodic Health Examination.[1–3] Fortification of a large variety of cereal food products with folic acid became mandatory in 1998.[4] In Quebec, the first evaluation of the impact of this program on the epidemiology of neural tube defects (NTDs) was performed using provincial administrative databases: stillbirth certificates (SBC) and infant death certificates (IDC), as well as computerized hospital discharge summaries (Med-Echo).[5] In Quebec City, an additional source of information was available, namely the computerized results of prenatal ultrasound examinations (Res-Echo) performed at the Saint-François d'Assise Hospital (SFAH). The objectives of the present study were to assess the validity of the different sources for the identification of NTD cases, and to examine the temporal trends in NTD rates and the impact of prenatal diagnosis among pregnancies referred to this tertiary care hospital from 1993 to 2002.

Methods

The study population included live births, stillbirths and terminations of pregnancies (because of fetal anomaly) at the SFAH from late 1992 to 2002. NTD cases were classified according to the nomenclature proposed by Nevin and Weatherall.[6] The three main categories were anencephaly (including craniorachischisis and inencephaly), spina bifida or meningomyelocele, and encephalocele (including exencephaly). Spina bifida occulta and sacral lipomeningocele were excluded. Spina bifida occulta is a common defect that is not diagnosed during the neonatal period.[6] Sacral lipo(myelo)meningocele is thought to be embryologically distinct from (myelo)meningocele; folic acid does not seem to be effective for its prevention.[7]

Med-Echo records including a main or a secondary diagnostic code compatible with NTD were identified for the period July 1992 to March 2002. The ninth revision of the *International Classification of Diseases* (ICD-9) was in use during the study period and codes of interest were infants with a neural tube defect (ICD-9: 740.0 to 742.0) and women with a fetus affected by a central nervous system malformation (ICD-9: 655.0). For infant death and stillbirth certificates, the tenth revision of the *International Classification of Diseases* (ICD-10) was adopted in 2000. The provincial databases were reviewed to identify records with a code for anencephaly (ICD-9: 740; ICD-10: Q00), for spina bifida (ICD-9: 741; ICD-10: Q05), or for encephalocele (ICD-9: 742.0; ICD-10: Q01). A file generated by Res-Echo, a computerized system for recording results of obstetrical ultrasound examinations at the SFAH, provided a list of pregnant women who had an examination indicating a fetus with an abnormality of the central nervous system. Hospital records were identified and medical notes were reviewed to ascertain all diagnoses.

Author References

Fassiatou Tairou, Philippe De Wals, Department of Social and Preventive Medicine, Laval University, Quebec City, Quebec, Canada
Adrien Bastide, Maternal and Child Health Center, Quebec City University Hospital Center, Quebec, Canada
Correspondence: Philippe De Wals, Département de médecine sociale et préventive, Université Laval, Pavillon de l'Est, Local 1110, 2180, Chemin Sainte-Foy, Quebec City, Quebec, Canada G1K 7P4; fax: (418) 656-7759; e-mail: Philippe.Dewals@msp.ulaval.ca

The denominator figures for live births and stillbirths were provided by the SFAH. A large proportion of NTD-affected pregnancies were terminated, so to prevent classification bias in calculating yearly prevalence proportions, a theoretical birth date was calculated for each NTD case, assuming a gestation of 40 weeks (date of birth/abortion less gestation length in weeks plus 40 weeks). The predictive positive value (PPV) of a source or of a diagnostic code was defined as the proportion of records which were true NTDs. The relative sensitivity of a source or of a diagnostic code was defined as the proportion of NTD cases identified from this particular source or diagnostic code, relative to the total NTD cases identified from all sources and codes combined.

TABLE 1
Distribution of neural tube defect (NTD) cases, by source of ascertainment at the St-François d'Assise Hospital (Quebec City)

Source			Number of cases	% of total
Death and stillbirth certificates	Med-Echo[1]	Res-Echo[2]		
+	+	+	22	12.7
+	+	–	0	0.0
+	–	+	0	0.0
–	+	+	43	24.9
+	–	–	0	0.0
–	+	–	94	54.3
–	–	+	14	8.1
All sources			173	100.0

+ The case was ascertained by the source.

[1] Quebec computerized hospital discharged summary database

[2] Computerized results of prenatal ultrasound examinations at the Saint-François d'Assise Hospital

The data were analyzed using SAS software.[8] Prevalence proportions with their 95% confidence intervals were calculated using an exact method. The Cochrane-Armitage test for trends in proportions was performed with statistical significance set at five percent. The study protocol was approved by the Quebec University Hospital Centre's Research Ethics Committee and access to provincial databases was authorized by the Quebec Access to Information Commission.

Results

Validation of sources

Provincial death records identified 14 infant deaths and eight stillbirths at the SFAH with an NTD diagnosis. A review of the medical records showed that the main cause of death was correct in all cases; the PPV was thus 100 percent.

The Med-Echo file contained 235 records mentioning a mother who had a fetus with a malformation of the central nervous system. Duplicate records were eliminated, so 178 mothers remained. A NTD-affected pregnancy was confirmed in 99 cases, another anomaly of the central nervous system was present in 62 cases, a malformation of another system or organ in ten cases, and another condition not considered a congenital malformation in the remaining seven cases. The PPV of the ICD-9 code 655.0 was 90% (161/178) for any malformation of the central nervous system and 56% (99/178) for NTDs.

The Med-Echo file contained 41 records of infants less than one year old with an NTD diagnosis. The code was incorrect in five cases: Another malformation of the central nervous system was present in four cases and a congenital anomaly of another organ in the remaining case. Therefore, the PPV of ICD-9 codes 740.0 to 742.0 was 88% (36/41). In the Med-Echo file, 24 NTD cases were identified under the mother and again under the child.

From the SFAH's Res-Echo file, we identified 133 pregnant women whose computerized record mentioned an anomaly of the central nervous system. In five cases, the medical records could not be found. Therefore, we reviewed 128 cases: 79 matched the definition of NTD and 49 did not (another anomaly of the central nervous system was present in 19 cases, a congenital malformation of another system or organ in 15 cases and another fetal condition not considered as a congenital malformation in the remaining 15 cases). The PPV for NTDs of this source was therefore 62% (79/128).

Combining the various information sources, a total of 173 NTD cases were identified (Table 1). The relative sensitivity of the different sources for ascertaining NTD cases was as follows: death and stillbirth certificates – 13% (22/173), Med-Echo – 92% (159/173) and Res-Echo – 46% (79/173). All the NTD cases identified in death and stillbirth certificates were also identified in Med-Echo. The majority of the 14 NTD cases missed by both SBCs/IDCs and Med-Echo were medical terminations of pregnancy (three cases of anencephaly, two of encephalocele and six spina bifida cases). Two stillbirths were also missed (one case of anencephaly and one of encephalocele), and a single live birth with spina bifida was not identified.

NTD prevalence proportion

The ascertainment of NTD cases having an expected date of birth in 1992 was incomplete and rates were not provided for that year. When the data analysis is limited to cases where the expected date of birth was between January 1, 1993 and December 31, 2002, 168 NTD cases were identified: 68 were live births, 14 stillbirths and 86 pregnancy terminations (Table 2). During this period, there were 26,210 live births and 220 stillbirths at the SFAH. There was a general downtrend in the number of births, except for a sudden increase in 1997. If we consider the absolute frequency of NTDs, we note a decline that began in 1998, continued in 1999 and then stabilized. Because of the combined effect of the increase in the total number

TABLE 2
Prevalence of neural tube defects (NTDs) at the St-François d'Assise Hospital (Quebec City) by pregnancy outcome (1992–2002)

	Year											
	1992[1]	1993	1994	1995	1996	1997	1998	1999	2000	2001	2002	All years combined
Number of live births	–	2,182	2,161	2,173	2,003	3,435	3,300	3,066	2,908	2,437	2,545	26,210
Anencephaly	0	0	2	1	2	3	0	0	1	0	0	9
Encephalocele	0	0	2	1	2	0	3	0	0	0	0	8
Spina bifida	0	10	5	7	7	9	2	1	7	1	2	51
All NTDs	0	10	9	9	11	12	5	1	8	1	2	68
Proportion/1,000	–	4.6	4.2	4.1	5.5	3.5	1.5	0.3	2.7	0.4	0.8	2.4
Number of stillbirths	–	22	22	21	15	27	31	16	24	18	24	220
Anencephaly	0	1	1	0	1	0	2	1	1	1	0	8
Encephalocele	0	0	0	0	1	0	0	0	0	0	0	1
Spina bifida	0	2	0	0	0	2	0	1	0	0	0	5
All NTDs	0	3	1	0	2	2	2	2	1	1	0	14
Proportion/1,000	–	136.4	45.5	0	133.3	74.1	64.6	125.0	41.7	55.6	0.0	59.3
Number of pregnancy terminations												
Anencephaly	1	4	3	4	4	2	1	2	0	2	4	27
Encephalocele	0	3	0	1	0	0	2	0	0	0	2	8
Spina bifida	4	7	2	8	5	8	5	4	3	8	2	56
All NTDs	5	14	5	13	9	10	8	6	3	10	8	91
Total births	–	2,204	2,183	2,194	2,018	3,462	3,331	3,082	2,932	2,455	2,569	26,430
All NTDs	5	27	15	22	22	24	15	9	12	12	10	173
Proportion/1,000	–	12.2	6.9	10.0	10.9	6.9	4.5	2.9	4.1	4.9	3.9	6.5

[1] Ascertainment of NTD cases in 1992 was incomplete and proportions were not calculated.

of births in 1997 and the decline in the number of NTD cases in 1998, the total prevalence proportion started to decline substantially in 1997 and stabilized from 1998 on.

If we visually examine the long-term trend with two or three information sources (Figure 1), we reach the same conclusion, i.e., a decline in the prevalence proportion of NTDs, starting in 1997 and followed by stabilization.

Prenatal diagnosis and induced abortion

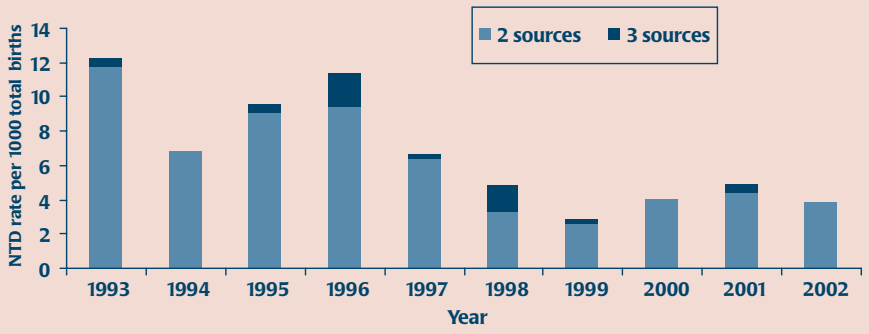
Of the 173 NTD cases ascertained in this study, 136 were diagnosed prenatally (78.6%). The primary diagnosis was made after an ultrasound examination in 92 cases and after an amniocentesis in the other three. No information was available for the remaining 41 cases. The percentage of cases with a prenatal diagnosis was 91% (40/44) for anencephaly, 76% (85/112) for spina bifida and 65% (11/17) for encephalocele. There was no significant trend in these percentages from 1993 to 2002. Overall, 52.6% (91/173) of NTD-affected pregnancies ended in induced abortion. The percentages were 61.4% (27/44) for anencephaly, 50% (56/112) for spina bifida and 47% (8/17) for encephalocele. There was no significant trend in these percentages between 1993 and 2002.

Discussion

Infant death and stillbirth certificates are a highly reliable source for ascertaining NTD cases, but their sensitivity is poor (13%). Indeed, NTD cases among terminations of pregnancy and living infants are not reported. On the other hand, the hospital administrative database Med-Echo has very good sensitivity (92%), but predictive value is low. It relies on the ICD-9 diagnostic category 655.0, which includes all anomalies of the central nervous system occurring in foetuses, but the database unfortunately contains many coding errors. Consequently, a valid analysis of the epidemiology of NTD based on hospital discharge summaries necessitates a complementary review of medical records. Currently, the Canadian Congenital Anomalies Surveillance System relies solely on this source and hospital records are not systematically reviewed.[9]

During the years covered by our study, the SFAH acted as a reference center for high-risk pregnancies in the Quebec City region. In 1996–1997, four maternity units were closed in Quebec City, resulting in a higher proportion of low-risk pregnancies referred to the SFAH. At the same time, food fortification was implemented in Canada. In the present study, it is impossible to disentan-

FIGURE 1
Total prevalence of NTDs, by number of sources* of information at the St-François d'Assise Hospital in Quebec City (1993–2002)



* Two sources = infant death and stillbirth certificates + Med-Echo;
Three sources = infant death and stillbirth certificates + Med-Echo + Res-Echo.

gle the possible effect of these two factors on the observed decreasing prevalence proportion of NTDs. Population-based studies, not hospital-based studies, are needed to assess the impact of folic acid food fortification on the epidemiology of NTDs. Such studies have been conducted in Newfoundland and Labrador[10] and in Nova Scotia,[11] and indicate a 78% to 54% decrease in the prevalence of NTD after fortification was implemented.

The prenatal ultrasound examination was introduced in Canada in the early 1970s and became routine in the mid 1980s. In 1994, the Society of Obstetricians and Gynaecologists of Canada recommended one examination between the 16th and 20th week of gestation to screen for malformations.[12] Under ideal conditions among high-risk pregnancies, prenatal sonography has been found highly sensitive (97%) and specific (100%) for the diagnosis of NTD.[13] Results may be different in a routine screening program. Among unselected pregnancies in 11 European regions, 96% of anencephaly cases were identified prenatally, but this occurred for only 68% of spina bifida cases.[14] In a 1992 study in the Estrie-Monteregie area of the Province of Quebec, all anencephaly and encephalocele cases were identified prenatally and the pregnancies terminated, but prenatal diagnoses were not made for 60% of spinal lesions.[15] At the SFAH, 79% of NTD cases were detected prenatally and the pregnancy was terminated in 53% of cases during the period 1993 to 2002. In the present study, there was no attempt to identify the reason why so many NTD cases were not detected prenatally nor why they were detected later on. This could be the objective of a subsequent quality control survey.

Conclusion

The combination of hospital discharge summaries and infant death and stillbirth certificates is a highly sensitive method for the ascertainment of NTD cases in the population, but medical records must be reviewed to exclude coding errors and to clarify imprecise diagnostic categories. During the period 1993 to 2002, there was no apparent increase in the proportion of NTD cases with a prenatal diagnosis and pregnancy termination. A large population-based study is currently underway in Quebec, relying on hospital discharge summaries and infant death and stillbirth certificates—as well as the review of medical records—in order to assess the real impact of folic acid food fortification.

Acknowledgments

This study was funded by a grant from the Canadian Institutes of Health Research (MOP-57904). The authors thank Martine Six and Manale Ouakki, as well as Doctors Rachelle Laframbroise and Jean Gekas of the Quebec City University Hospital Centre, for their collaboration.

References

1. Health Canada. Folic acid: The vitamin that helps protect against neural tube (birth) defects. Issues April 9, 1993.

2. SOGC Genetics Committee. Recommendations on the use of folic acid for the prevention of neural tube defects. J Soc Obstet Gynaecol Can 1993; Supp 15:41–6.

3. Canadian Task Force on the Periodic Health Examination. Periodic health examination, 1994 update: 3. Primary and secondary prevention of neural tube defects. Can Med Assoc J 1994 151:159–66.

4. Regulatory impact analysis statement. SOR/98–550. Canada Gazette Part II 1998, 132:3029–33.

5. De Wals P, Rusen ID, Lee NS, Morin P, Niyonsenga T. Trend in prevalence of neural tube defects in Quebec. Birth Defects Res A Clin Mol Teratol 2003; 67:919–23.

6. Nevin NC, Weatherall JAC. Illustrated guide to malformations of the central nervous system at birth. Edinburgh: Churchill Livingstone; 1983.

7. McNeely PD, Howes WJ. Ineffectiveness of dietary folic acid supplementation on the incidence of lipomyelomeningocele: pathogenetic implications. J Neurosurg. 2004; 100(2 Suppl Pediatrics):98–100.

8. Statistical Analysis System (SAS). Version 8.1. Institute Inc; Cary, NC, USA, 1999–2000.

9. Health Canada. Congenital anomalies in Canada: A perinatal health report, 2002. Available at: www.phac-aspc.gc.ca/ublicat/cac-acc02/index.html

10. Liu S, West R, Randell E, et al. A comprehensive evaluation of food with folic acid for the primary prevention of neural tube defects. BMC Pregnancy Childbirth 2004;4:1–10.

11. Persad VL, Van den Hof MC, Dubé JM, Zimmer P. Incidence of open neural tube defects in Nova Scotia after folic acid fortification. Can Med Assoc J 2002; 167:241–45.

12. Société des Obstétriciens et Gynécologues du Canada (SOGC). Lignes directrices concernant l'exploration par ultrasons en obstétrique et en gynécologie. Directives Cliniques en Obstétrique 1994;8:1–7.

13. Lennon CA, Gray DL. Sensitivity and specificity of ultrasound for the detection of neural tube and ventral wall defects in a high-risk population. Obstet Gynecol 1999; 94:562–6.

14. Boyd PA, Wellesley DG, De Walle HE, et al. Evaluation of the prenatal diagnosis of neural tube defects by fetal ultrasonographic examination in different centres across Europe. J Med Screen 2000; 7:169–74.

15. De Wals P, Trochet C, Pinsonneault L. Prevalence of neural tube defects in the Province of Quebec, 1992. Can J Public Health 1999;90:237–39.