Original Research

Prevalence and prenatal diagnosis of neural tube defects in Nova Scotia in 1980-84

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A survey of the records of all hospitals with obstetric services in Nova Scotia revealed that during 1980–84 there were 122 pregnancies involving a neural tube defect. The mean rate was 2/1000 births. Of the affected fetuses or infants 54% had spina bifida, 35% had anencephaly and 11% had encephalocele. The records showed that in the early part of the period studied at least one prenatal ultrasonographic examination had been performed in 60% of the pregnancies; in 1984 the rate was 74%. When examinations done before 16 weeks' gestation were excluded, the overall detection rates at the first ultrasonographic examination were 100% for anencephaly and 73% for spina bifida and encephalocele; the rates improved toward the end of the study period.

La revue des dossiers de tous les hôpitaux de Nouvelle-Écosse pourvus d'un service d'obstétrique montre, de 1980 à 1984, 122 grossesses où il est survenu chez le foetus des anomalies du tube neural. Le taux moyen est de 2 par 1000 naissances. Il s'agit de spina bifida (54%), d'anencéphalie (35%) et d'encéphalocèle (11%). Au début de cette période, on trouve mention d'au moins une ultrasonographie chez 60% des gestantes; en 1984 ce taux passe en 74%. Si on ne tient pas compte des examens qui ont eu lieu avant 16 semaines de gestation, on calcule un taux de détection de 100% pour l'anencéphalie et de 73% pour le spina bifida et l'encéphalocèle. Ce dernier taux s'améliore vers la fins de la période susdite.

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The occurrence of a neural tube defect, such as anencephaly, myelomeningocele, meningocele or encephalocele, has important implications for both the individual family and the health care system. Many affected babies do not survive infancy, and those who do survive may have severe disabilities. Although genetic factors have been implicated, over 90% of affected infants are born to parents with no family history of such defects. Thus, an effective program for prenatal detection of neural tube defects would involve some method of screening all pregnancies.

It has previously been reported that the prevalence of neural tube defects is higher in eastern Canada than in the rest of Canada and most of the United States.[1-3] The prevalence appears to be declining in some areas.[4,5]

The relation between elevated maternal serum α-fetoprotein levels and open neural tube defects has been well documented,[6] and population screening of pregnant women for elevated levels has been advocated for all those who wish to participate.[7] Such programs have not been implemented in Nova Scotia. However, there has recently been an increase in the availability and number of prenatal ultrasonographic examinations and thus an increase in the number of neural tube defects detected. In addition, measurement of the amniotic fluid α-fetoprotein level and supplementary testing are available to women with a family history of neural tube defects.

We undertook a study to estimate the prevalence at birth of neural tube defects in Nova Scotia, where there are about 12 000 births annually, and to assess the effects of the current practice of ultrasonography on the prenatal diagnosis of these defects.

Methods

All hospitals with obstetric services in Nova

Scotia were contacted and asked to supply names, date of birth, diagnosis and ultrasonographic results for all pregnancies in which a neural tube defect was detected in 1980–84. All fetuses and infants with spina bifida, anencephaly or encephalocele were included; those with hydrocephalus without spina bifida were not. If both spina bifida and anencephaly were recorded, the defect was classified as anencephaly. Records were identified by patient name and date of birth to avoid duplication when patients were transferred to another hospital. When possible, information about pregnancies that were terminated because of a neural tube defect was also obtained.

To ensure that the data were as complete as possible, birth records were cross-checked with hospital records at Izaak Walton Killam (IWK) Hospital for Children, Halifax (the only hospital in the province with pediatric neurosurgical services), and with records at the Atlantic Research Centre for Mental Retardation (which coordinates prenatal diagnosis for the province). It was possible to review the hospital records for most infants with neural tube defects to determine whether ultrasonography had been carried out during the pregnancy.

Rates of detection of neural tube defects were determined from a review of the ultrasonography reports. In a few cases the defect was scored as detected if the suspicion of an anomaly was mentioned in the report and a repeat examination scheduled. If both spina bifida and hydrocephalus were present and only hydrocephalus was mentioned in the first report, this was scored as if the neural tube defect were detected.

Results

Birth records were obtained from 37 of the 38 hospitals providing obstetric services. One hospital refused to provide information, but we estimated that there would have been only about 1200 births at the hospital in a 5-year period.

The number of cases of neural tube defects recorded in 1980–84 is shown in Table I. Eleven of the cases would have been missed if only hospital birth records had been used; six were identified from records at IWK Hospital for Children and five from records at the Atlantic Research Centre for Mental Retardation. At least 1 of these 11 babies was born at the hospital that refused to provide information. Four others were transferred to IWK Hospital for Children the day they were born, and this may partly explain why the defect was not coded at the birth hospital. The other discrepancies were probably due to differences in interpretation of the ICD-9[8] code or clerical errors. Another indication of the completeness of our data is that we identified 43 cases of anencephaly, compared with 31 reported by Statistics Canada for the same 5-year period.[9] Thus, for practical purposes the data on livebirths can be considered virtually complete. Some stillborn infants or fetuses with defects may have been missed.

Our survey did not include detailed long-term follow-up. However, of 71 liveborn infants with spina bifida or encephalocele, 65 (92%) had at least one admission to IWK Hospital for Children. The remaining six died at less than 1 year of age. As expected, none of the infants with anencephaly was transferred; one survived in hospital for 15 days and another for 9 days.

By the end of 1984, obstetric ultrasonography was available in all hospitals in Nova Scotia with more than 250 births per year. In 1984, 17 873 obstetric ultrasonographic examinations were recorded by the Nova Scotia Department of Health. It was not possible to determine from the data the number of examinations involving pregnancies that were not continued to term or the stage of the pregnancy at the time of examination. However,

Table I — Number of cases of neural tube defects in Nova Scotia in 1980–84

Defect	No. of cases					
	1980	1981	1982	1983	1984	Total
Anencephaly						
Births	5	5	5	6	5	26
Therapeutic abortions	2	1	2	6	6	17
Spina bifida						
Births	14	13	9†	16	10	62
Therapeutic abortions	0	0	0	2	2	4
Encephalocele						
Births	2	5	1	5	0	13
Therapeutic abortions	0	0	1	0	0	1
Total	23	24	18	35	23	123
Rate per 1000 births*	1.8	2.0	1.5	2.8	1.8	2.0

*Based on all identified neural tube defects including those in liveborn and stillborn infants and therapeutically aborted fetuses. The denominator was derived from vital statistics and includes stillbirths after 28 weeks' gestation. No data were available on spontaneous abortions.
†Includes one set of twins; thus, there were 122 affected pregnancies.

given the annual birth rate of about 12 000, it seems likely that in 1984 many pregnant women had at least one ultrasonographic examination at some time during their pregnancy.

During the study period 60% of the women with affected children had at least one ultrasonographic examination (Table II). In the last year of the study 74% of the women had at least one such examination. This is a minimum estimate, because if a woman had an ultrasonographic examination at a hospital other than the hospital of delivery the record of examination may not have been included in this survey. During the study period about half of the examinations were performed at Grace Maternity Hospital, Halifax, with real-time equipment with 3.5-mHz and 5.0-mHz transducers. The remaining examinations were performed at various hospitals throughout the province.

The results of the first recorded ultrasonographic examination are shown in Table III. In the one case of anencephaly that was not detected the examination was performed at an estimated gestation of 10 weeks, and no further examination was done during the pregnancy. The films were reviewed after the outcome of the pregnancy was known, and the anomaly could not be identified.

For the 10 undetected cases in which the examination was performed at 16 weeks' gestation or later, five examinations were performed at Grace Maternity Hospital and the other five at other hospitals. All 10 pregnancies occurred in the early years of the study, and no defects were missed after 16 weeks' gestation in 1983 or 1984.

Of the 15 cases of spina bifida or encephalocele not detected at the first examination, 6 were detected at a later stage of pregnancy, and 6 were undetected despite subsequent examination (Table IV). In the remaining three cases there was no record of further examination.

Discussion

The prevalence rate of neural tube defects in Nova Scotia in 1980–84 was 2/1000 births (1.6 if therapeutic abortions were excluded). This compares with 1.7/1000 livebirths in eastern Ontario and western Quebec in 1969–81,[10] 3.2/1000 births in Newfoundland in 1976–83[11] and 2.5/1000 births in New Brunswick in 1977–81 (Greg Sherman, Canadian Congenital Anomalies Surveillance System: personal communication, 1985). Therapeutic abortions in these comparison groups were not specifically mentioned.

Detailed information on the prevalence of spina bifida in Nova Scotia before 1980 was not available for comparison. We found a rate of anencephaly of 0.7/1000 births in 1980–84, compared with 1.38/1000 births reported by Elwood[12] for 1943–70.

Table II — Distribution of ultrasonographic examinations during pregnancy, by year of birth of infant with neural tube defect and time of examination

Year	No. of women: No record of examination	No. of women: Examination at 20 weeks' gestation or earlier	No. of women: Examination after 20 weeks' gestation only	% of women who underwent at least one examination
1980	14	6	3	39
1981	10	5	9	58
1982	9	4	4	47
1983	10	13	12	71
1984	6	11	6	74
Total	49	39	34	60

Table III — Results of the first ultrasonographic examination in 73 pregnancies

Estimated gestation, wk	No. of pregnancies;* defect: Anencephaly: Detected	Anencephaly: Not detected	Spina bifida or encephalocele: Detected	Spina bifida or encephalocele: Not detected
≤ 15	2	1	1	5
16–20	8 (10)	0	4 (2)	6
21–27	2	0	6	1
≥ 28	7	0	15	3
Total	29	1	28	15

*Numbers in parenthesis represent pregnancies that were terminated; the defect was most likely detected at 16 to 20 weeks' gestation.

At an international workshop on prenatal diagnosis in 1979 Hobbins and colleagues[13] stated that "we expect that in competent hands anencephaly can be diagnosed as early as 14 weeks and should not be missed after 16 weeks". In our survey all 27 cases of anencephaly in fetuses examined after 16 weeks' gestation were detected.

For spina bifida and encephalocele, only 12 women had their first ultrasonographic examination at 16 to 20 weeks' gestation, and 50% of the defects were detected. When examination at a later stage and repeat examinations are included, 79% of the cases of spina bifida and encephalocele were detected before delivery.

These figures are based on a retrospective review of the hospital ultrasonography records. They represent the actual practice in Nova Scotia during 1980–84 and should be interpreted with caution, because the timing of the examination, the size and form of the lesion, the type of equipment used and the experience of the examiner were not fully assessed. Also, it can be assumed that in most instances there was no reason to suspect a neural tube defect before the first examination. In only three cases of spina bifida was it known that the woman had previously had an affected child. In all three cases the defect was detected early, one at 11 weeks' gestation and the other two at 16 weeks. The detection rate appears to be highly dependent on the timing of the examination. One of us (B.S.B.) previously reported a case of extensive spina bifida that was not identifiable by ultrasonography at 16 weeks' gestation but was clearly identified at 20 weeks.[14]

It is difficult to compare our rates of detection of spina bifida with those in the literature, because most reports are based on high-risk populations. Our patients were a heterogeneous group: some had an obstetric ultrasonographic examination for unspecified reasons, while a few were referred because of complications of pregnancy or a family history of abnormalities. In 1983 Roberts and associates[15] reported rates of detection of open spina bifida of 5/14 from April 1977 to March 1980 and 16/20 from April 1980 to March 1983. All examinations were done before 20 weeks' gestation. The authors attributed the change over time to improvement in equipment and experience gained by the operators. Milunsky and Alpert[16] reported that two cases of open neural tube defects were missed despite "sophisticated ultrasound scanning at two different first-rate institutions in Boston" and despite awareness of abnormal amniotic fluid α-fetoprotein and acetylcholinesterase levels.

No information about false-positive results of ultrasonography could be obtained in our study. Such results are obviously a concern if ultrasonography alone is used for diagnosis. In the 1982–83 pilot project on maternal serum α-fetoprotein in Manitoba, three patients were initially considered on the basis of results of ultrasonography to have fetuses with congenital anomalies (sacral meningocele, gastroschisis and hydrocephalus). The amniotic fluid α-fetoprotein and acetylcholinesterase levels were normal in all three, and the infants were unaffected at birth. No false-positive results have been reported since the pilot project; this is attributable to improvement in ultrasonography techniques (Manitoba Advisory Committee for Maternal Serum AFP Screening Project: personal communication, 1985).

During 1980–84, 30 out of 43 cases of anencephaly were studied; 29 of the 30 cases were detected by ultrasonography. Of the 20 pregnancies in which anencephaly was detected at 20 weeks' gestation or earlier, 17 were terminated. In the remaining three cases no information was available as to when the parents were informed of the diagnosis and whether therapeutic abortion was offered.

Six cases of spina bifida and one case of encephalocele were detected before 20 weeks' gestation, and three of the pregnancies involving spina bifida were terminated. Two further pregnancies were terminated after 20 weeks. In at least three cases prior knowledge of a fetal anomaly resulted in referral of the patient before delivery to Grace Maternity Hospital, where tertiary obstetric and neonatal care is available. Referral from community to regional hospitals also likely occurred, but this could not be determined from the records obtained for this study.

At an international consensus meeting in Berlin in November 1983 it was concluded that maternal serum α-fetoprotein screening will not become superfluous when routine ultrasonography is used more widely (or uniformly, as is now the

Table IV — Results of the second or third ultrasonographic examination in 12 cases of spina bifida or encephalocele not detected at the first examination

Estimated gestation, wk	No. of pregnancies; defect			
	Spina bifida		Encephalocele	
	Detected	Not detected	Detected	Not detected
25–29	1	1	0	0
30–35	2	2	1	1
≥ 36	2	2	0	0
Total	5	5	1	1

case in some countries) and that although obstetric ultrasonography can detect virtually all cases of anencephaly, it cannot reliably detect spina bifida at 16 to 20 weeks' gestation.[17] Hobbins and colleagues[18] considered that evaluation of the fetal spine during the second trimester is among the most difficult of diagnostic examinations to perform and that it is thus unrealistic to expect a perfect diagnostic record. They also considered that, given the time involved and experience required, it is currently not practical to suggest that all pregnant women undergo ultrasonography to screen for neural tube defects. This is debatable: at Grace Maternity Hospital detailed assessment of the fetal spine is an integral part of the examination as of 16 weeks' gestation.

In our experience the most appropriate time for ultrasonographic examination to detect neural tube defects is 16 to 20 weeks' gestation. Although anencephaly is virtually always recognizable at this stage, some cases of encephalocele and spina bifida may be less clearly identified at 16 weeks, and a repeat examination close to 20 weeks may be advisable. When there is doubt or when a high risk of neural tube defect is suspected, repeat examination is strongly recommended.

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New diseases

Some will allow no Diseases to be new, others think that many old ones are ceased; and that such which are esteemed new, will have but their time: However, the Mercy of God hath scattered the great heap of Diseases, and not loaded any one Country with all: some may be new in one Country which have been old in another. New Discoveries of the Earth discover new Diseases.

— Sir Thomas Browne (1605–1682)