

Current topic

Spina bifida—a vanishing nightmare?

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SUMMARY Detailed analysis is presented on the dramatic decline in spina bifida births and other congenital central nervous system defects in the past 12 years, in both Sheffield and the rest of Great Britain. In Sheffield, there was an average of 20 spina bifida births per year up to 1972, but since then there has been a progressive fall to only one in 1984. This decrease may be partly attributable to antenatal diagnosis and terminations of affected pregnancies, but there is no known explanation for the rest of the fall, which has also been experienced throughout Great Britain.

The virtual elimination of spina bifida is now possible, as long as the existing methods of prevention and antenatal diagnostic facilities are used even more fully. Relaxation of our effort and a reverse in the 'natural' trend could bring the incidence of spina bifida back to where it was 15 years ago.

The object of this report is to draw attention to the declining number of births with neural tube defects, with special reference to spina bifida in Sheffield, and to outline the role of antenatal diagnosis followed by termination of pregnancies in this phenomenon. It is now possible that spina bifida is a disappearing disorder. In the past, increases in spina bifida births occurred during war, famine, or economic disasters, and a corresponding tendency to steady decline was seen when these circumstances improved. Reductions in incidence, however, have never been of the order experienced in the past few years.

Experience in Sheffield

Sheffield is an industrial city with a population of 550 000 and an annual average of 6500 births. An accurate congenital anomalies registry was established 20 years ago, and is a most valuable source of information. The information is abstracted from birth certificates, on which every evident congenital abnormality is recorded. All information is cross-checked with hospital records, and is corrected if necessary.

Like elsewhere, there were no known measures for preventing spina bifida births until about 1965¹ when a large scale study established that the risk of

neural tube defects in a family after they had had a baby with spina bifida was approximately 5%. Prevention of recurrences, however, was only possible by avoiding further pregnancies. This made no measurable impact on the number of spina bifida births.

In 1972, after the first report on the value of α -fetoprotein estimation in the amniotic fluid,² amniocentesis was offered to high risk couples. Although this was most valuable to these couples, it made no measurable difference to the number of spina bifida births, because over 95% of these are firstborn. Soon after came the possibility of discovering a much higher number of families at high risk with the routine estimation of the serum α -fetoprotein concentration in pregnant women,³ and since then more and more 'normal' pregnancies have been monitored by serum tests. Routine serum tests started in January 1977 on women who presented at the antenatal clinic in good time. In 1983, over 90% of pregnant women had been screened. When indicated, the serum test was usually followed by amniocentesis and termination of pregnancy if the result indicated a neural tube defect.

Concurrently, increasing use was made of diagnostic ultrasound monitoring of pregnancies and this is now part of routine management.

Table 1 *Spina bifida and encephalocele births among Sheffield residents*

Years	Average number per year	Rate per 10 000 total births
1968–70	18	21
1971–73	10	16
1974–76	8	13
1977–79	6	11
1980–82	6	11
1982	2	3.6
1983	3	
1984	1	

The economic status of the city was improving in the 1960s. There was little unemployment, and the population's diet had probably never been as good. Yet, the incidence of spina bifida was around 20 per 10 000 births. The late 1970s saw a progressive deterioration in the standard of living for those who became unemployed, and by the 1980s some 15% of the population were jobless. It is not known whether and how this affected the nutrition of pregnant women.

Spina bifida and encephalocele. During the 1970s and 1980s the annual number of spina bifida and encephalocele births fell progressively from an average of 18 in 1968–70 to an average of two in 1982–4 (Table 1). In 1984, there was only one spina bifida birth. The rate per 10 000 total births was 21 in 1968–70 and only 3.6 per annum in 1982–4. In 1984, the rate was only 1.8 per 10 000 but this is based on very small figures. Furthermore, in recent years, a larger proportion than usual have suffered a simple meningocele, so it has been among the more severe cases, (those with myelomeningocele) that numbers have declined most steeply.

While the major reason for the nine-fold reduction in the annual number and a six-fold reduction in

the annual rate between 1968 and 1970, and 1982 and 1984 was the natural decline in pregnancies affected by spina bifida, antenatal diagnosis followed by terminations of pregnancies became more and more responsible for the reduction of spina bifida births in the last six years.

To establish the role of antenatal diagnosis and terminations of pregnancies, a thorough study was carried out for the years 1979 to 1984 inclusive, of all the notifications of births and deaths; all the antenatal α -fetoprotein tests; and the relevant obstetric records in the three large units where almost all the births and terminations in Sheffield take place.

Table 2 shows that during these six years, eight babies were born with a simple meningocele; (none were terminated for this reason), 19 babies were born with myelomeningocele, and three with encephalocele. Without terminations nearly double this number would have been born with myelomeningocele and encephalocele because in the same six years 18 and 1 terminations respectively were carried out because the fetuses had these defects. During 1979–81 an average of 9.6, and in 1982–4 an average of 6.6 conceptions are known to have occurred with spina bifida (including simple meningocele) or encephalocele. During 1982–4 there were only two such births a year, so that in these three years antenatal diagnosis accounted for some two thirds of the reduction in spina bifida births.

Some of the 19 myelomeningocele births that occurred in the last six years could have been prevented. In two the diagnosis was made antenatally but the parents refused termination, in a further four a suggestive positive serum test result was not followed up by amniocentesis and in another five the serum gave false negative results and ultrasound was not used.

Finally, eight mothers of affected babies were not

Table 2 *Neural tube defects in children born in Sheffield between 1979 and 1984*

Year	Births					Total	Terminations			Total
	MMC*	MC†	EC‡	AC	MMC		AC	Total		
1979	3	2	1	1	7	4	7	11	18	
1980	6	2	1	1	10	1	8	9	19	
1981	5	3	1	—	9	—	5	5	14	
1982	1	1	—	—	2	6	4	10	12	
1983	3	—	—	—	3	4	7	11	14	
1984	1	—	—	1	2	3§	3	7	9	
Total	19	8	3	3	33	19	34	53	86	

MMC=myelomeningocele; MC=meningocele; EC=enkephalocele; AC=anencephalus.

*Of 19 cases of liveborn MMC, 5 are alive in 1985; †all 8 are alive; ‡all 3 died; §plus one with encephalocele.

There have been no terminations for meningocele.

screened, largely because they attended too late for the first antenatal visit. In this group there was a high proportion of immigrant women from the Asian subcontinent.

Fourteen of the 19 infants with myelomeningocele had severe, permanently handicapping lesions at birth within the criteria for non-treatment.⁴ They were under the care of several different consultants: none were treated and all 14 died. Two others, who were borderline cases, were not treated at birth but were operated on later; they survived with severe handicaps. The last three were far less severely affected, were treated, and survived with only mild to moderate handicap. During a similar period a decade earlier, some 60 infants survived with severe multisystem handicaps.

Anencephalus. Antenatal screening made an even more successful contribution to the elimination of anencephalic births. During the last six years there were 34 terminations of anencephalic fetuses and only three births (1.6 per 10 000) including stillbirths. There was only one anencephalic stillbirth in the last three years (0.5 per 10 000). (In this case no antenatal tests were carried out because the immigrant mother from the Third World did not attend an antenatal clinic.) Between 1968 and 1973 there were an average of 20 per 10 000 anencephalic births per year, so by 1982-4 there was a 40 fold reduction in births and a halving of known conceptions.

It is possible that there were more terminations for anencephalus than these figures indicate, if the diagnosis was made by ultrasound alone and if the reason for termination of pregnancy was not specified.

Experience in the major cities and NHS regions and in England and Wales

Major cities. Based on Office of Population Censuses and Surveys data there was a similar trend of a reduction in spina bifida births in most major cities.⁵ In London itself there were 47 spina bifida births in

1983; a rate of 5.0. As recently as 1979, the rate was 8.3.

The rate in 1983 for the seven provincial cities with births of over 5000 each was on average 4.4. Within these global figures there are, however, differences. For example, in Nottingham with over 7600 births, not a single baby was notified as having been born with spina bifida but in Bradford the rate had increased from 6.9 in 1979 to 8.7 in 1983. Such fluctuations are liable to occur when the numbers are small. Bradford has a large Asian minority but there is no indication whether the incidence was higher among Asian immigrants.

In the rest of England and Wales in 1983 there were 350 spina bifida births, a rate of 7.3. This is significantly higher ($P < 0.01$) than in the eight major cities. It is possible that the antenatal services in the major cities are better organised and detect more abnormal pregnancies. (Table 3)

Health regions. In most of the 14 English regions, and also in Wales, the incidence rates dropped considerably, but with individual variations.⁵ In 1979 the highest rate was in the Northern Regional Health Authority—22.9 per 10 000—and the lowest was in Oxford—9.0 per 10 000. These rates dropped to 7.6 and 4.8 respectively in 1983. Until recently East Anglia had experienced a low incidence for many years, but their rate has not decreased as in other regions. While in 1980 the rate was 9.0, in 1983 it was 10.3 per 10 000 which was the highest of all the 14 health regions and about the same as that of Wales (10.4). In East Anglia routine antenatal serum tests were abandoned in 1981 because this was considered to be uneconomic.⁶ In 1983 Trent, which includes Sheffield, had the lowest rate with only 4: a fourfold reduction since 1979 when it was 16.

Wales. In Wales, particularly the south, there had been a high incidence in the past, but between 1979 and 1983 the rate reduced from 15.7 to 10.4. The reduction in the three health districts of Glamorgan,

Table 3 *Spina bifida births in England and Wales in 1983*

	Total births	Spina bifida		Significance
		No	Rate per 10 000	
London	94 224	47	5.0	} ns
Seven other major cities*	57 370	25	4.4	
London and 7 major cities	151 594	72	4.7	
The rest of England and Wales	481 171	350	7.3	} $P < 0.01$
All England and Wales	632 765	422	6.7	

*Cases with 5000 or more births—Birmingham, Leeds, Nottingham, Liverpool, Manchester, Sheffield, and Bradford (in decreasing order of number of births). The spina bifida rate for London and the 7 major cities is significantly lower than the rest of England and Wales (99% level).

which includes Cardiff, was from 18.7 to 13.8. In Cardiff itself there has been no consistent decrease in spina bifida births during the period 1974 to 1983, with the lowest number of two in 1981 and the highest of seven in 1982.⁷

England and Wales. In England and Wales as a whole, the incidence of spina bifida was high, both absolutely and by international comparisons, up to 1972. There were only minor fluctuations in the rate, which was around 20 per 10 000 each year. In 1972, the last year before antenatal diagnosis became possible, 1380 babies were born with spina bifida unassociated with encephalocele or anencephalus (18.8 per 10 000) or approximately 1520 if we include an estimated 140 born with encephalocele not included with spina bifida births. This 1520 would give an annual incidence of 21 per 10 000. Since then there has been a rapidly progressive decline in spina bifida births, including encephaloceles, from 789 (12 per 10 000) in 1980 to 461 (7.3 per 10 000) in 1983 (Table 4).

The even greater reduction in anencephalic births from 14.7 per 10 000 total births in 1974 to only 2 in 1983 can be largely accounted for by antenatal diagnosis followed by termination of pregnancy.

During the same period there was also a consider-

able decrease in the number of hydrocephalic infants (unassociated with spina bifida) born from 4.8 to 3.1 per 10 000, although antenatal diagnosis and terminations played no part in this decline. Possibly, this is a true change in the incidence of the disorder and is similar to that which might have been observed in the case of neural tube defects had there been no medical interference. This suggestion is supported by the fact that in Northern Ireland, where therapeutic abortions are rarely carried out, the incidence of spina bifida decreased in a proportion similar⁸ to that observed for hydrocephalus in England and Wales. These figures for hydrocephalus must be viewed with some caution, however, firstly, because many cases are not diagnosed in the neonate, and secondly those diagnosed later may not be included in the statistics. On the other hand, some cases are not truly 'congenital', but are acquired in the neonatal period.

Role of antenatal diagnosis and terminations of pregnancies. The role of antenatal diagnosis in the reduction in babies born with neural tube defects is not precise as notifications of terminations of pregnancies because of fetal neural tube defects are almost certainly incomplete. The data available from the Office of Population Censuses and Surveys relates to 1980-3.⁹

In 1974 there were only 34 terminations but these increased progressively to 511 in 1983. Most were carried out because the fetus had anencephalus. The number of pregnancies terminated specifically because the fetus had spina bifida was never more than 130 (in 1982) unless terminations for spina bifida are included in those notifications with unspecified central nervous system malformations. Even so, it is unlikely that terminations for spina bifida exceeded 150 in any one year, and yet in 1982, 900 fewer babies were born with spina bifida than only 10 years earlier.

Table 4(a) *Spina bifida births (without anencephalus or encephalocele) in England and Wales between 1971 and 1978**

Year	No	Rate per 100 000 total births
1971	1404	17.7
1972	1380	18.8
1974	1078	16.7
1976	794	13.4
1978	765	12.7

†Number with encephalocele approximately 10% more.

Table 4(b) *Spina bifida and encephalocele births (without anencephalus) in England and Wales between 1979 and 1983**

Year	Spina bifida	Encephalocele	Total	Rate per 100 000 total births
1979	789	84	873	13.6
1980	712	77	789	11.9
1981	623	76	699	10.9
1982	491	67	558	8.9
1983	410	51	461	7.3

*These figures have been adjusted with the help of Office of Population Censuses and Surveys (OPCS).¹⁰ In the OPCS publications spina bifida with anencephalus is classified as spina bifida, and encephalocele are included among other disorders of the CNS.

Table 5 *Reported incidence of births with congenital malformations in England and Wales between 1974 and 1983*

Year	Rates per 10 000 total births			
	All malformations	CNS	%	Other than CNS
1974	196.7	37.9	(19.3)	158.8
1976	209.9	32.5	(15.5)	177.4
1978	212.2	29.2	(13.8)	183.0
1980	213.8	22.3	(10.4)	191.5
1982	210.9	16.1	(7.6)	194.7
1983	220.8	14.5	(6.6)	206.3

CNS=central nervous system.

Other congenital defects. Whatever are the cause or causes of the decline in central nervous system defects, these are not applicable to other types of congenital malformations. On the contrary, the incidence of all congenital malformations is on the increase. The rate for all congenital defects, excluding those of the central nervous system increased progressively from 159 in 1974 to 206 per 10 000 total births in 1983.⁵ (Table 5) In 1974, 19.3% of all notified congenital malformations were those of the central nervous system, and by 1983 this proportion had fallen to 6.6%.

Experience in Scotland

Considerable reductions in spina bifida births have been achieved in Scotland,^{10, 11} from 265 (30 per 10 000) in 1971 to 75 (11 per 10 000) in 1982. Some 40% of the fall is attributable to terminations during 1979–82. In 1982 the incidence, even without terminations, would have been down to 18 per 10 000.

In the West of Scotland, major effort was made to reduce the incidence of neural tube defects using routine serum α -fetoprotein tests.¹¹ By 1981, 73% of pregnant women had routine serum tests; only 1% needed amniocentesis and 56 terminations of pregnancy were carried out where the fetus had spina bifida.

In Greater Glasgow itself, spina bifida births declined from 40 per 10 000 in 1974 to 5 per 10 000 in 1983 (personal communication). Some 21 terminations were carried out in 1981 because of spina bifida, suggesting that 31 spina bifida conceptions took place, in contrast to the 56 some seven years earlier. There must be additional explanations for the remainder of the eightfold reduction in a mere nine years.¹⁰

Experience in Ireland

In Ireland the incidence of neural tube defects including spina bifida was always possibly the highest in the world. In Northern Ireland, as late as 1975, the rate of spina bifida births was 45 per 10 000. Subsequently, a major, progressive drop occurred to 20 in 1980,¹² but the rate was still as high as it had been in England 10 years earlier. Only 2% of this decrease is attributable to selective abortions because only women at high risk were offered antenatal tests.⁸

In Eire selective antenatal tests are rare and abortions are forbidden, yet in Dublin the rate fell from 32 in 1979 to 22 by 1982.¹³ In contrast, the 1982 rate in Sheffield was only three.

Discussion

The data illustrating the rapid decline in spina bifida and other neural tube defect births concentrate principally on Sheffield, and to a lesser extent on other parts of the British Isles. It is here that the greatest decline has occurred. An earlier, similar report also showed this trend in Liverpool up to 1980.¹⁴

The most rapid decline has occurred in the past few years and has been accelerated considerably by the termination of affected pregnancies. The trend was evident before, however, even in Ireland where terminations are not practised. The incidence of spina bifida in England is now about as low as anywhere in the world. In other countries,^{15–19} the decline is less dramatic, partly because they started from an already lower level, and partly because preventive methods have not been used.

There is a warning, too. In Hungary¹⁸ and in Australia (personal communication), the incidence of spina bifida has started to rise again. Such a rise could occur here, also, if we abandon our vigilance and fail to use preventive facilities fully.

Conclusion

If the present trends continue, then the virtual disappearance of spina bifida from the United Kingdom is now a real, none too distant possibility. Nevertheless, the current fall due to unknown 'natural' causes may stop or even be reversed. Failure to use the existing and perhaps future preventive measures could mean a return to the incidence that existed in the past. We must be on guard and not lull ourselves into false security just because, at present, spina bifida is on the decline for unknown epidemiological reasons, and continue to increase our efforts with active prevention.

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