

## Screening for congenital neural tube defects in a high-risk area: an epidemiological perspective

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**SUMMARY** Data from the Glasgow Register of Congenital Malformations were used to investigate the extent of the recent decline in the prevalence of anencephaly and spina bifida, and the contribution of antenatal screening to it. Over the period 1974–85 inclusive, 303 pregnancies with an anencephalic foetus were diagnosed, representing an “adjusted” prevalence of 1.9 per 1000 total births, of which 179 (59%) were terminated following antenatal screening. There were 364 pregnancies with a spina bifida foetus representing an “adjusted” prevalence of 2.3 per 1000 total births, of which 84 (23%) were terminated. Over the study period, the “adjusted” prevalence of anencephaly fell by 50% while the birth prevalence fell by 89%; the “adjusted” prevalence of spina bifida fell by 38% while the birth prevalence fell by 76%. It was concluded that although the birth prevalence of both defects (particularly anencephaly) would have declined substantially in the absence of screening, the West of Scotland programme should continue.

The population of Glasgow shares with Ireland, Wales and other “Celtic” areas of Britain a relatively high risk of neural tube defects. The development of antenatal screening for anencephaly and spina bifida (ASB) throughout the last decade or so has coincided with a decline in the birth prevalence of these defects in England and Wales.<sup>1</sup> An epidemiological analysis of Scottish (including Glasgow) data concluded that Scotland too had experienced a decline in prevalence (from 1971 to 1982), part of which was attributable to screening.<sup>2</sup>

The present study had two interrelated objectives: first, to describe the extent of the decline in the birth prevalence of ASB in Glasgow in recent years, and second, to assess the contribution of antenatal screening to that decline.

### Methods

Data on ASB were obtained from the Glasgow Register of Congenital Malformations for the years 1972–85 inclusive. All livebirths and stillbirths with a diagnosis (recorded by a doctor, midwife or health visitor) of anencephaly or spina bifida were registered, provided that the maternal address at birth lay within the boundaries of the area covered by the Greater

Glasgow Health Board (Glasgow City before 1974). Where both lesions occurred together, the defect was registered as anencephaly. Encephalocele, iniencephaly and spina bifida occulta were excluded. Terminations of pregnancy following antenatal screening (by ultrasound or serum alpha-fetoprotein assay) were recorded separately (providing the maternal residence criterion was fulfilled). Data on these were obtained from the Department of Medical Genetics, Yorkhill Hospitals. Since all fetuses aborted in Glasgow as a result of antenatal diagnosis are subjected to routine pathological examination, this information is likely to be both complete and reliable.

Two types of prevalence measure were calculated for anencephaly and spina bifida separately. *Birth prevalence rates* were obtained by restricting the numerator to affected births (live and still-); *adjusted prevalence rates* were obtained by adding terminations (following antenatal diagnosis) to affected births, thereby producing a numerator equivalent to the number of affected births which would presumably have occurred had there been no screening.<sup>2</sup> Denominator data consisted of the annual numbers of total births (live and still-) to residents of the Greater Glasgow Health Board area (Glasgow City Before 1974) and were obtained from the annual reports of the

Registrar General for Scotland. The prevalence rates were modelled using least squares regression techniques.

## Results

Between 1974 and 1985 there were 124 anencephaly births and 303 anencephaly pregnancies (births plus terminations), representing a birth prevalence for anencephaly of 0.8 per 1000 and an adjusted prevalence of 1.9 per 1000. Thus the majority (59%) of such pregnancies were terminated.

There were 280 spina bifida births and 364 spina bifida pregnancies (births plus terminations) during this time, representing a birth prevalence for spina bifida of 1.8 per 1000 and an adjusted prevalence of 2.3 per 1000. Thus more than three-quarters (77%) of spina bifida pregnancies reached term.

The declining annual birth and pregnancy prevalence rates for anencephaly and spina bifida are shown in tables 1 and 2 respectively. The greater impact of antenatal screening on anencephaly than on spina bifida prevalence is demonstrated by the derivation (from the 1972-85 data) of the least squares regression lines: exponential in the case of anencephaly births (fig 1) and linear in the case of spina bifida births (fig 2). All four prevalence rates declined significantly during the study period.

## Discussion

For technical reasons, the previously published study of anencephaly and spina bifida trends in Scotland was obliged to use provisional Glasgow Registry data generated by manual sorting.<sup>2</sup> Since then, automated data processing has enabled us both to identify and rectify inaccuracies in those figures and to update the analysis. Our findings, however, are broadly similar and indicate that a marked decline in the frequency of

Table 1 Birth and "adjusted" prevalence rates of anencephaly in Glasgow, 1974-85

Birth year	Total births (live and still-)	Birth prevalence (excluding terminations)		"Adjusted" prevalence (births plus terminations)	
		No	Rate/1000	No	Rate/1000
1974	14 880	29	1.9	33	2.2
1975	14 398	34	2.4	39	2.7
1976	12 889	18	1.4	34	2.6
1977	12 487	11	0.9	28	2.2
1978	12 491	10	0.8	30	2.4
1979	13 339	9	0.7	29	2.2
1980	13 438	5	0.4	24	1.8
1981	13 491	3	0.2	19	1.4
1982	12 884	0	0.0	19	1.5
1983	12 661	1	0.1	19	1.5
1984	12 783	1	0.1	14	1.1
1985	13 089	3	0.2	15	1.1

Table 2 Birth and "adjusted" prevalence rates of spina bifida in Glasgow, 1974-85

Birth year	Total births (live and still-)	Birth prevalence (excluding terminations)		"Adjusted" prevalence (births plus terminations)	
		No	Rate/1000	No	Rate/1000
1974	14 880	51	3.4	51	3.4
1975	14 398	45	3.1	45	3.1
1976	12 889	22	1.7	25	1.9
1977	12 487	35	2.8	42	3.4
1978	12 491	25	2.0	28	2.2
1979	13 339	26	1.9	33	2.5
1980	13 438	25	1.9	36	2.7
1981	13 491	7	0.5	21	1.6
1982	12 884	18	1.4	23	1.8
1983	12 661	7	0.6	15	1.2
1984	12 783	8	0.6	17	1.3
1985	13 089	11	0.8	28	2.1

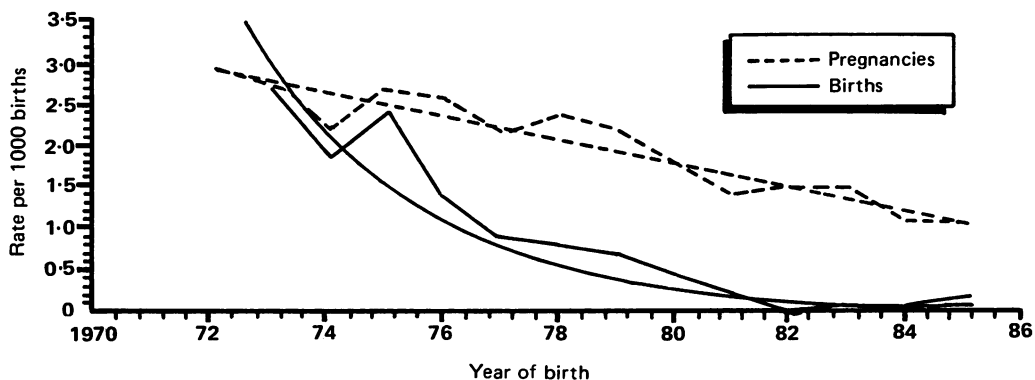


Fig 1 Prevalence of anencephaly in Glasgow 1972-85.

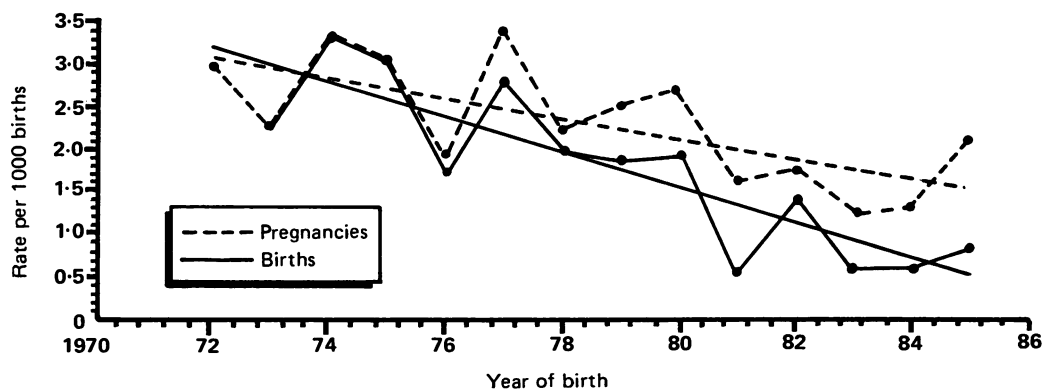


Fig 2 Prevalence of spina bifida in Glasgow 1972-85.

both anencephaly and spina bifida births would probably have occurred in Glasgow even in the absence of a programme of antenatal screening. The adjusted prevalence of anencephaly fell by half while that of spina bifida fell by more than a third between 1974 and 1985. This is the epidemiological context within which the impact of antenatal screening should be considered.

That screening accelerated this "natural" decline is evident from the sharper drop in the frequency of babies born with the defects. Anencephalic births declined by 89% while spina bifida births declined by 76%. The excess fall in birth prevalence as compared to adjusted prevalence is, of course, attributable entirely to antenatal screening.

Screening resulted in the termination of a larger proportion (59%) of anencephalic pregnancies than spina bifida pregnancies (of which 23% were terminated) over the period 1974-85 as a whole. This was due mainly to the greater sensitivity of serum-alpha-fetoprotein screening for anencephaly (97%) than for spina bifida (72%).<sup>3</sup> Any improvement in the efficacy of screening for spina bifida will therefore require either an increase in the sensitivity of the test or an increase in the proportion of the pregnant population screened (about 75% in 1985), or preferably both.

The practical implications of these findings are as follows. Firstly, because the prevalence of both defects would have declined (albeit to a lesser extent) in the absence of screening, we should continue to conduct aetiological and other research in an attempt to provide an explanation for the phenomenon. Secondly, continuous public health monitoring of the prevalence of ASB is essential since the population must be considered vulnerable to a future upturn in ASB frequency.<sup>4</sup> Finally, since our only effective (if partial) preventive response to the problem is antenatal screening, this should continue to be

implemented and improved until the "natural" decline in prevalence has reached the point where populations such as that of the West of Scotland need no longer be considered to be exposed to a "high risk". Precisely where that point lies is a matter for debate.<sup>5</sup>

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