Trends in Utilization of Prenatal Cytogenetic Diagnosis by New York State Residents in 1979 and 1980

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Abstract: It is estimated that 35.3 per cent of pregnant New York State women age 35 or over underwent cytogenetic diagnosis in 1980 as compared to 28.7 per cent in 1979. Rates varied sharply by county. In several small counties far from genetic centers, no 1980 cytogenetic diagnostic studies were reported in women 35 or over while in New York City the rate was 41 per cent. In one county with an active genetic center the rate appears to have plateaued at 30 per cent. (*Am J Public Health* 1983; 73:198–202.)

Introduction

There has been a marked increase in the use of prenatal cytogenetic diagnosis since the report of the National Institutes of Health committee concerning the safety and efficacy of this procedure.[1] Here we report the geographic variation in utilization by New York State residents in 1980 and compare those results with observations in 1979 reviewed previously.[2,3]

Materials and Methods

Data were collected by the New York State Chromosome Registry from 27 laboratories known to have provided cytogenetic services to New York State residents in 1980. Twenty-five laboratories were affiliated with the Registry of which 14 are in New York City, seven in the rest of the state, and four outside New York State (See Acknowledgments). Details of the methods of the Registry are reported extensively elsewhere.[4]

In addition, data were included from non-affiliated centers on an estimated total of 30 women studied by St. Luke's Hospital in New York City in 1980 and an estimated 870 New York State residents studied by Metpath Laboratories in nearby New Jersey. These women had not also been studied by Registry centers. Those at St. Luke's were all reported to be age 35 or over and Manhattan (New York County) residents. Those studied at Metpath were subdivided by maternal age (<35, ≥35) according to the known distribution of all cases (704) from this laboratory, January to October of 1980. The distribution of residences of these cases from Metpath was estimated from the known distribution of cases studied by New York City and Long Island centers.

To our knowledge, there were in 1980 no other laboratories not affiliated with the Chromosome Registry which provided cytogenetic services to New York State residents. All laboratories with New York State cytogenetic permits for prenatal diagnosis were accounted for. It is possible a few resident pregnant women living near the state boundary may have sought both obstetrical and cytogenetic services at nearby out-of-state centers. However, many cytogenetic laboratories in New England and New Jersey are affiliated with the Registry and would have reported such studies. We would only be likely to miss the few women who sought such services in Pennsylvania, Quebec, or Ontario.

There was some difficulty interpreting reports from participating centers in the New York City metropolitan area who reported local residence as simply "New York", with county unspecified. This could refer to New York County (Manhattan) or to New York City which comprises five counties: Bronx, Kings (Brooklyn), New York, Queens, and Richmond (Staten Island). All such cases were treated in two ways: all were considered as New York County residents, and all were distributed to the five New York City counties according to the distribution of cases of known residence reported by all centers.

The proportion of pregnant women age 35 and over electing prenatal diagnosis was calculated by dividing the estimated total number of resident women 35 and over who underwent amniocentesis by the total number of live births and fetal deaths (gestational age 20 weeks or over) for the same maternal age group in each county in 1980. The latter data were reported to the Office of Biostatistics of the New York State Department of Health.

Results

Table 1 displays the overall distribution of utilization for the state both for 1979 and 1980. Figure 1 illustrates the statewide variation in 1980 by county. The five New York City counties are coded at the midpoint of their estimate ranges.* Schuyler and Hamilton Counties are not coded to avoid any bias that might result from their low number of live

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*These ranges were: Bronx 36%–44%, Kings 31%–38%, New York 40%–66%, Richmond 31%–38%, Queens 37%–45%.

TABLE 1—Estimated Utilization of Prenatal Cytogenetic Diagnosis by New York State Residents, 1979 and 1980

	Women Under Age 35		Women Age 35 and Over		All Ages	
	1979[a]	1980	1979[a]	1980	1979[a]	1980
New York City	626/94,931 = 0.66%	894/95,923 = 0.93%	2641/7852 = 33.6%	3413/8281 = 41.2%	3267/102,783 = 3.2%	4307/104,204 = 4.1%
Rest of State	579/126,878 = 0.46%	811/129,788 = 0.62%	1847/7800 = 23.7%	2246/7730 = 29.1%	2426/134,678 = 1.8%	3057/137,518 = 2.2%
Entire State	1205/221,809 = 0.54%	1705/225,711 = 0.76%	4488/15,652 = 28.7%	5659/16,011 = 35.3%	5693/237,461 = 2.4%	7364/241,722 = 3.0%

a = Data from reference 2.

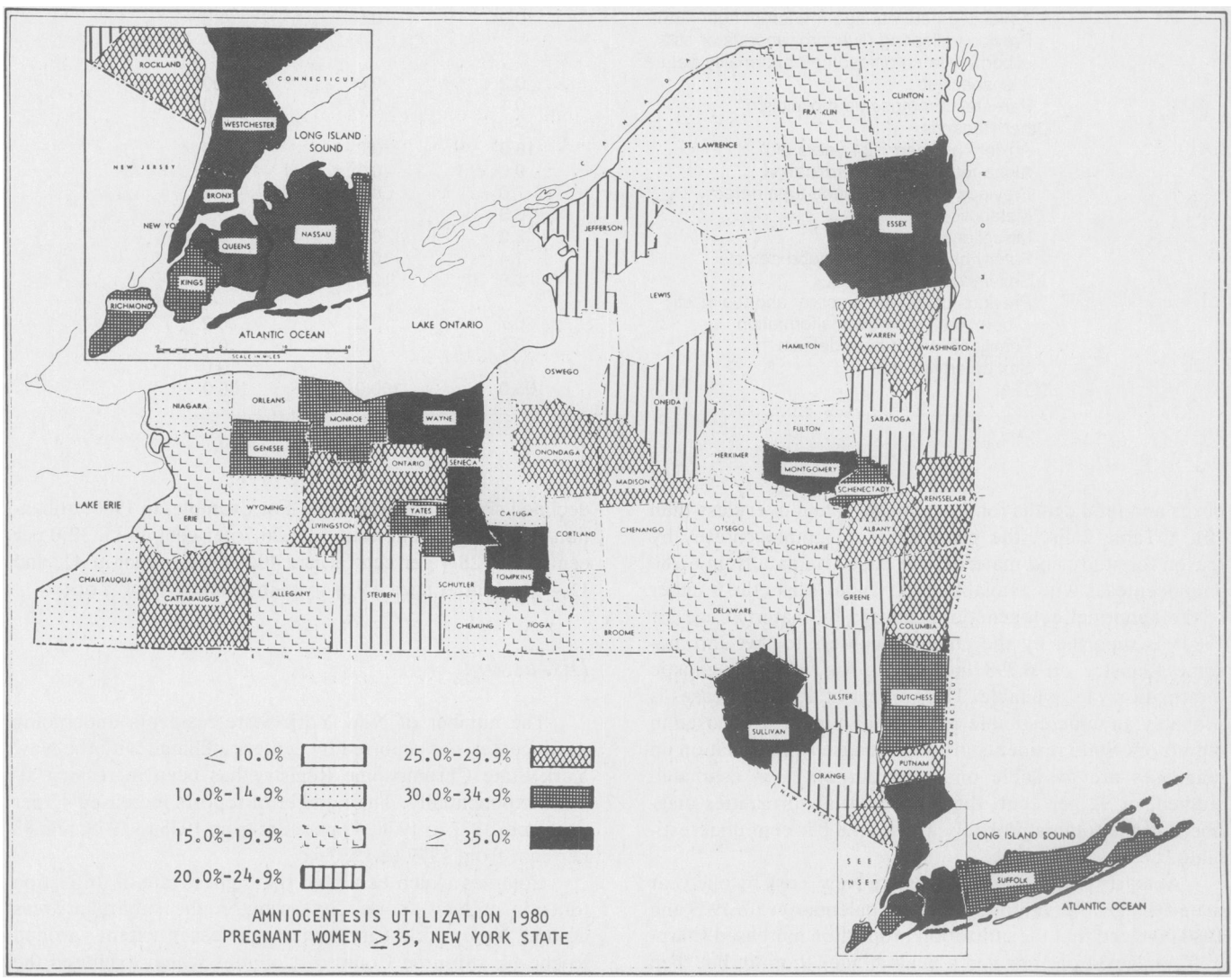

FIGURE 1—Estimated Proportions of Pregnant Women Age 35 or Over Electing Prenatal Cytogenetic Diagnosis, by Count of Residence in New York State, 1980

TABLE 2—Reason for Study by Maternal Age in Prenatal Studies in New York State Residents as Reported to the New York State Chromosome Registry in 1980

Reason for Study	Percentage of Amniocenteses by Maternal Age		
	<35	≥35	All Ages
Definite or Possible Chromosomal Risk Factors			
Elevated maternal age	30.6	97.8	82.6
Family history of Down's syndrome	7.8	0.3	2.0
Previous offspring (liveborn, abortus or stillborn) with Down's syndrome	6.3	0.3	1.6
Previous offspring (liveborn, abortus or stillborn) with chromosome abnormality other than Down's syndrome	4.1	0.4	1.2
Previous spontaneous abortion(s)	1.5	0.2	0.5
Previous offspring (liveborn, abortus or stillborn) with multiple malformations	1.7	0.0	0.4
Parent translocation carrier	1.2	0.1	0.3
Suspected chemical mutagen	1.2	0.0	0.3
Radiation	0.7	0.1	0.2
Family history of chromosome abnormality other than Down's syndrome	0.1	0.0	0.1
Family history of chromosome abnormality nature unspecified	0.4	0.0	0.1
Advanced paternal age (maternal age <35)	0.0	0.0	0.0
Previous offspring (liveborn, abortus or stillborn) with chromosome abnormality nature unspecified	0.2	0.0	0.0
Parental chromosome mosaicism	0.1	0.0	0.0
Other Reasons			
Anxiety (women ≤34 years old)	18.9	0.0	4.3
Alpha-fetoprotein determination	9.6	0.3	2.4
Previous child with neural tube defect	7.0	0.1	1.7
Metabolic disease	3.3	0.2	0.9
Miscellaneous	2.0	0.1	0.5
Family history of neural tube defects	1.4	0.1	0.4
Sex-linked disease	0.9	0.0	0.2
Previous offspring (liveborn, abortus or stillborn) with a single malformation	0.7	0.0	0.2
Family history of birth defects	0.3	0.0	0.1
Sex determination	0.0	0.0	0.0
TOTAL	100.0	100.0	100.0

births and fetal deaths for women age 35 and over (less than 10).** Table 2 lists the percentages of amniocenteses by reason for study and maternal age based on data from 6,780 amniocenteses with available data. Table 3 lists the number of the abnormal cytogenetic diagnoses by maternal age for 1980, as reported by the affiliated centers to the Chromosome Registry on 6,798 individuals for which diagnostic information was available. The number of abnormal cases is probably an underestimate of the total number diagnosed in New York State residents in 1980 because as information on diagnoses are available on 6,798 out of 7,364 estimated studied, or 92 per cent, the number of abnormalities diagnosed in the total is probably about an 8 per cent underestimate.

Analysis of utilization in Upstate New York by one year maternal age interval for each of the calendar years 1979 and 1980 revealed that the utilization proportion increased sharply from age 34 on, reached a peak at age 39 or 40, but then declined sharply at older ages. For example, in 1980, utilization was 8.9 per cent at age 34, 26.7 per cent at 35, 32.0 per cent at 39, 28.4 per cent at age 40, 24.4 per cent at 42, and 11.4 per cent at ages 45 or over in Upstate New York.

Discussion

The number of New York State residents undergoing amniocentesis as reported by centers affiliated with the New York State Chromosome Registry has been increasing almost exponentially. The number of reports increased 47 per cent from 1977 to 1978, 49 per cent from 1978 to 1979, and 43 per cent from 1979 to 1980.

Counties which exhibited the highest rate of utilization (outside of New York City) were in the suburban areas around New York City and, to a lesser extent, around Rochester (Monroe County). Counties which exhibited the lowest rate of utilization were in the southern tier on the Pennsylvania border and an almost concentric circle of

**Detailed figures by county available on request to author.

TABLE 3—Number and Proportion of Abnormal Cytogenetic Diagnoses Made Prenatally in 1980 as Reported to the New York State Chromosome Registry*

Diagnosis	Maternal Age									
	<35		≥35		Not Stated	All Ages	
Down's syndrome (All genotypes)	7	(0.46)	51	(0.97)	0	58	(0.85)
Edwards' syndrome (+18)	1	(0.07)	11	(0.21)	0	12	(0.18)
Patau's syndrome (+13, and other genotypes)	1	(0.07)	5	(0.10)	0	6	(0.09)
XXY	2	(0.13)	7	(0.13)	0	9	(0.13)
XXX	0	(0.00)	6	(0.11)	0	6	(0.09)
XYY	1	(0.07)	3	(0.06)	0	4	(0.06)
Turner's syndrome (All genotypes)	2	(0.13)	4	(0.08)	0	6	(0.09)
Other abnormal diagnoses	15	(0.98)	32	(0.61)	1	48	(0.71)
All abnormal diagnoses	29	(1.89)	119	(2.27)	1	149*	(2.19)
Normal diagnoses	1508	(98.11)	5124	(97.73)	17	6649	(97.81)
All diagnoses	1537	(100.00)	5243	(100.00)	18	6798	(100.00)

NOTE: Per cents given in parentheses.

*The number of abnormals are likely to be an 8% underestimate of the actual number detected in New York State residents in this period (see text).

counties around Syracuse (Onondaga County). Counties in both of these areas are some distance from active clinical genetics centers. New York State clinical genetics centers are located in New York City and in Albany, Erie, Monroe, Nassau, Onondaga, Rockland, Suffolk and Westchester Counties.

As awareness of and interest in prenatal cytogenetic diagnosis expands, utilization increases as demonstrated by the changes reported in New York State in this report. It is likely however, that there will always be a proportion of women who will decline the procedure because of concern about its risk and for other reasons that may relate to attitudes to selective termination of pregnancy. It is difficult to predict what the expected maximum rate of utilization is likely to be. In the New York City metropolitan area, current utilization is almost 40 per cent and may still be increasing. In Monroe County, however, where there is an active clinical genetics center, utilization was about 30 per cent in both 1980 and 1979.

Bernhardt and Bannerman have concluded from a review of data for New York State in 1979 that the most important variables influencing the overall rate of utilization are the knowledge, interest, and attitudes of obstetricians.[5] These may change with time, particularly because of increasing concern about medical-legal consequences of not informing parents about the availability and pertinence of prenatal diagnosis.[6-8] To our knowledge, however, while patients have won the legal right to sue over these issues,[6,7] no successful actions have been brought to date.

The decline in utilization over age 40, despite rapidly increasing rates of cytogenetic abnormality at these ages, has also been observed in Ohio.*** The trend is unexpected because it indicates that at these older ages some additional factor tends to inhibit utilization despite the advancing risk. Possibly, among pregnant women over age 35, there is a growing proportion with age who decline amniocentesis because they would not elect abortion in any circumstances, and over age 40 the numbers of such women outweigh the numbers of those who seek amniocentesis because of concern about the higher risks of abnormal offspring. Perhaps, also, pregnant women over age 40 who have had more offspring than those aged 35 to 39 are more likely to believe—because those born to date have been normal—that they have relatively little to benefit from prenatal diagnosis.

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*These centers are affiliated with the New York State Chromosome Registry, but did not report any diagnoses on New York State residents in 1980.