An Evaluation of Congenital Malformations Surveillance in New York State: An Application of Centers for Disease Control and Prevention (CDC) Guidelines for Evaluating Surveillance Systems

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SYNOPSIS

Established in 1982, the New York State Congenital Malformations Registry (NYCMR) is one of the largest statewide, population-based birth defects registries in the nation. In this article, we evaluate the surveillance of congenital malformations in New York State using the Centers for Disease Control and Prevention (CDC) guidelines for evaluating public health surveillance systems. In addition to the evaluation of selected qualitative and quantitative system attributes, we assess the public health significance and usefulness of the surveillance system and how well it is meeting its stated objectives.

The NYCMR uses passive case ascertainment, relying on reports from hospitals and physicians. A congenital malformation is defined as any structural, functional, or biochemical abnormality determined genetically or induced during gestation and not due to birthing events. In addition to being the primary source of congenital malformations surveillance data in New York State, the NYCMR also provides cases for traditional epidemiological studies to determine risk factors for specific congenital malformations.

The NYCMR has been working to meet its stated objectives while striving to improve its qualitative and quantitative attributes. Registry personnel have implemented several measures designed to enhance the simplicity of the data collection and data entry processes, as well as to maintain the acceptability of the surveillance system to the reporting sources. Because it is a statewide, population-based surveillance system, by far the strongest quantitative attribute of the NYCMR is representativeness. The sensitivity of the NYCMR is difficult to evaluate. Available estimates suggest, however, that the NYCMR identifies a large proportion of children born with congenital malformations in New York State and diagnosed from birth through two years of life. Finally, the NYCMR has in recent years been able to publish and disseminate annual reports describing the distribution of specific malformations in New York State on a timely basis.

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Across the US, many statewide population-based congenital malformations registries were established in the aftermath of the thalidomide tragedy of the 1960s.[1–8] The New York State Congenital Malformations Registry (NYCMR) is one of the largest such registries in the nation. The NYCMR developed out of the Love Canal pollution crisis, which revealed the inadequacies of relying on birth certificates to monitor and evaluate birth defects.

In 1980, cognizant of the need to collect more reliable and valid surveillance data for birth defects, the New York State legislature enacted a bill authorizing the Department of Health to require hospitals and physicians to report diseases and conditions that are designated "environmentally related." In April, 1981, the Public Health Council (a body appointed by the governor and statutorily charged with rule making) enacted Part 22 of the State Sanitary Code, which mandated statewide reporting to four registries that were to form the database for an Environmental Disease Surveillance Program to be established on October 1, 1982. One of those registries is the NYCMR.

To date, numerous papers that describe the operations and case-ascertainment strategies of several congenital malformations registries/surveillance systems have been published.[1–3,6,7,9,10] Corresponding publications on the evaluation of congenital malformations surveillance systems, however, are lacking. This can be explained in part by the fact that widely accepted and complete guidelines for evaluating surveillance systems did not exist until the Centers for Disease Control and Prevention (CDC) published theirs in 1988.[11]

Previous authors have emphasized varying criteria for evaluating public health surveillance systems, depending on the goals and objectives of each system. Although the uniqueness of each system should be taken into account, many authors have agreed on some central questions that evaluations should seek to answer:[2,11–17] What is the public health importance of the surveillance system? Is the system meeting its stated goals and objectives? Is the system useful? Should the system be continued? The variation in criteria comes into play when evaluating qualitative and quantitative system attributes. It is generally acknowledged that improving one attribute (e.g., sensitivity) may lessen another attribute (e.g., timeliness). For this reason, it is likely that surveillance systems will not meet all criteria equally and that evaluators should therefore focus on the criteria that are most important to the surveillance system of interest.[11]

The purpose of this article is to evaluate the surveillance of congenital malformations in New York State according to the CDC guidelines for public health surveillance systems while considering the specific challenges of congenital malformations.[3,11] In addition to evaluating selected qualitative and quantitative system attributes, we assess the public health significance and usefulness of the surveillance system, as well as how well it is meeting its stated objectives.

PUBLIC HEALTH IMPORTANCE

Congenital anomalies as a group are relatively common, affecting some 3% to 5% of live-born infants in the United States. In New York State the overall occurrence of major malformations diagnosed up to age 2 is 4.1% (Table 1).

For more than two decades, congenital malformations have been a leading cause of infant mortality in the US.[4,18,19] Twenty percent of infant deaths are attributed to congenital malformations, a percentage that has increased over time. Among infants with malformations who do not survive, more than 70% die in the first month of life. In addition, congenital malformations are the fifth leading cause of years of potential life lost and a major cause of morbidity and mortality throughout childhood.[4,18,20] Approximately 12% of pediatric hospital admissions are for congenital malformations of various types, and hospitalizations for congenital malformations are longer and more expensive.[21] Infant mortality from birth defects has declined more slowly than has mortality associated with preterm birth or low birthweight.[22]

In New York State, congenital anomalies were the second leading cause of infant mortality in 1996, surpassed only by conditions originating in the perinatal period.[23] For children one to 9 years of age, congenital anomalies were the fourth leading cause of death after accidents, acquired immune deficiency syndrome (AIDS), and neoplasms. An examination of the infant mortality experience of infants with birth defects born from 1983 to 1988 in New York State found that for infants with major malformations the mortality rate was 68 per 1,000 population, a risk of death 6.2 times higher than the general population risk of 11 per 1,000 population.[24] A recent study in California found a risk of death that is nine-fold for black infants and 17.8-fold for white infants.[25]

Approximately 40% to 60% of congenital malformations are of unknown origin. Twenty percent may be due to a combination of heredity and other factors; 7.5% may be due to single gene mutations; 6% to chromosome abnormalities; and 5% to maternal illnesses, such as diabetes or infections, or to anticonvulsant drugs.[26–28]

Table 1. Prevalence of congenital malformations, New York State, 1997 Congenital Malformations Registry

	Both sexes			Males			Females		
Race and residence	*Infants*	*Total births*	*Percent*	*Infants*	*Total births*	*Percent*	*Infants*	*Total births*	*Percent*
New York State									
All races	10,457	256,976	4.1	6,324	131,353	4.8	4,128	125,619	3.3
White	7,307	183,073	4.0	4,466	93,469	4.8	2,837	89,601	3.2
Black	2,561	54,948	4.7	1,497	28,042	5.3	1,064	26,906	4.0
Other	558	17,847	3.1	340	9,286	3.7	217	8,560	2.5
New York State, excluding NYC									
All races	5,749	138,074	4.2	3,556	70,594	5.0	2,188	67,476	3.2
White	4,807	118,658	4.1	2,981	60,694	4.9	1,822	57,961	3.1
Black	786	14,467	5.4	468	7,375	6.3	318	7,092	4.5
Other	142	4,404	3.2	95	2,247	4.2	46	2,156	2.1
New York City									
All races	4,708	118,902	4.0	2,768	60,759	4.6	1,940	58,143	3.3
White	2,500	64,415	3.9	1,485	32,775	4.5	1,015	31,640	3.2
Black	1,755	40,481	4.3	1,029	20,667	5.0	746	19,814	3.8
Other	416	13,443	3.1	245	7,039	3.5	171	6,404	2.7

NOTE: Total includes unknowns within each category, thus row and column figures may not sum to totals.

OBJECTIVES

The initial goal of the NYCMR was to identify environmental exposures that may cause birth defects and to provide information for their prevention. Over time, other purposes for congenital malformations surveillance have evolved. Registries are the only way to obtain population-based data on congenital malformations because birth certificates are inadequate. According to Lynberg and Edmonds, the ideal congenital malformations surveillance system should provide "population-based information . . . reports in a timely manner . . . case ascertainment should be comprehensive . . . (with) . . . accurate and precise diagnosis."[3] The last is of special concern and presents a challenge because congenital malformations are a large, heterogeneous group. Specific objectives of the NYCMR include:

1. To determine the annual incidence of congenital malformations among New York live births;
2. To monitor the incidence and type of major malformations with regard to geographic distribution and community characteristics;
3. To investigate suspected increases in the incidence of malformations that may be associated with exposure to toxic substances;
4. To conduct epidemiological studies of specific malformations;
5. To respond to medical inquiries regarding congenital malformations;
6. To work with other bureaus within the Department of Health to ensure that services and quality care for children with malformations are provided;
7. To provide data for planning, advocacy, education, and other requests.

SYSTEM DESCRIPTION

Case definition

A congenital malformation is defined as any structural, functional, or biochemical abnormality determined genetically or induced during gestation and not due to birthing events. Cases include infants and children diagnosed up to 2 years of age who were born or reside in New York State. Most congenital malformations systems include major structural malformations and some metabolic disorders. Most of these conditions are diagnosed in the first one or 2 years of life. Mental retardation, autism, and cerebral palsy are generally not included, because the issues in the surveillance of these conditions are very challenging and generally require that children be followed to school age, which requires additional sources for case ascertainment.[29]

Case ascertainment

State regulations require all physicians and other hospital staff to report major congenital malformations diagnosed at birth through the age of 24 months. In reality, reporting sources for the NYCMR consist primarily of hospitals that provide obstetric and pediatric services. Only a small number of reports are submitted by individual physicians. The mandatory reporting law provides some leverage when it becomes necessary to contact the reporting physician for clarification of a diagnosis. All new case reports are matched against existing registry data to eliminate duplication. As mentioned, a particular challenge in congenital malformations surveillance is accurate, complete, and specific diagnosis. To help accomplish this goal, the NYCMR provides written guidelines to hospitals and physicians describing what and how to report. Reporting physicians, hospitals, and genetics laboratories are asked to provide a narrative description of the congenital anomaly, and NYCMR staff review all such reports carefully. Incomplete reports and nonspecific diagnoses are followed-up with the reporting entity.

Virtually all reports are ascertained from inpatient hospital records, because malformations diagnosed on an outpatient basis are generally not well reported. Accurate clinical recognition of malformations depends on the clinical acumen and interest of hospital staff, particularly for conditions that are difficult to diagnose, such as fetal alcohol syndrome. As a consequence, the identification of malformations may vary by area and time. Areas with hospitals that provide a high level of care, and that may therefore make more thorough diagnoses, may be associated with higher rates. Similarly, areas with hospitals that report cases more completely will also appear to have higher rates. In regions with low numbers of births, small variations in the number of congenital malformation cases may produce large variations in the incidence proportion or prevalence rate.

Multiple malformation cases present a particular challenge. These can be categorized as known syndromes, recognized associations, sequences (where a primary defect causes other defects such as the paralysis from a spina bifida leading to talipes equinovarus), and unrecognized defect combinations.[30] Some investigators think that these associations are especially important in finding teratogens.[5,31] NYCMR staff recognize the importance of obtaining enough information on each of these cases, including chromosome reports, so that a classification can be made. Complex cases are reviewed with the medical director, who may follow up with the geneticist/dysmorphologist who originally saw the child. Because many syndromes are not diagnosed in the first two years of life, information about later diagnoses may not reach the NYCMR. Ideally, all multiple malformations would be reviewed by a geneticist/dysmorphologist. Although resources do not permit this routinely, a geneticist/dysmorphologist is usually consulted for etiologic studies.

Coding

A coding system with great specificity is needed to allow the analysis and categorization of congenital malformations, and it should be applied in a consistent fashion. To provide greater consistency in coding, NYCMR staff code the narrative diagnostic reports of congenital malformations using the ninth revision of the International Classification of Diseases Coding Manual (ICD-9-CM) and a modified version of the British Pediatric Association (BPA) system.[32] The BPA coding scheme is used by a number of other congenital malformations registries and allows for greater specificity than does the ICD-9 system. For example, the ICD-9 code does not distinguish between gastroschisis and omphalocele, two similar but distinct conditions; the BPA code does.

Confidentiality

It is important that congenital malformations registries collect identifiers.[4] Many children are treated at more than one institution, in which case multiple reports will be submitted, some of which may contain added or updated information or diagnoses. Identifiers are also needed to follow up with families for studies and to send information on to the families about services available to their affected children. These needs create an obligation to hold all information reported to the registry in strict confidence. NYCMR information is protected by Section 206(1)(j) of the state's Public Health Law; access to data by anyone other than registry personnel is restricted and carefully monitored to preserve confidentiality. Families of children reported to the registry are not contacted for studies without prior consent of the Department of Health Institutional Review Board (IRB) and notification of the child's physician.

USEFULNESS

The Bureau of Environmental and Occupational Epidemiology (BEOE) in the New York State Department of Health (DOH) is routinely called upon to assess the potential health impact associated with toxic substances throughout the state. The Bureau relies on the NYCMR to provide a means by which DOH can survey and monitor the New York State population for trends in congenital malformations.

In addition to being the primary source of congenital malformations surveillance data in New York State, the NYCMR is used for many other purposes. First, it can provide useful information to Maternal and Child Health programs. Tracking affected children to ensure quality care and enrollment in early intervention programs is an important part of disability prevention. Through data linkages between the NYCMR database and the Early Intervention database, the NYCMR has played an active role in ensuring that children with special needs are receiving available services. The NYCMR staff has collaborated with the Maternal and Child Health programs in the Department of Health to develop ways to inform families about programs and available services.

Second, the NYCMR surveillance data can help track the nation's progress toward the *Healthy People 2010* health promotion and disease prevention goals. Although a national surveillance system for congenital malformations does not yet exist, the CDC relies on data from statewide surveillance systems such as the NYCMR to monitor national trends in congenital malformations.[33] In an effort to facilitate the sharing of information from state birth defects surveillance programs, the CDC joined with representatives of numerous state health departments to form the National Birth Defects Prevention Network (NBDPN).[34] One activity of the NBDPN is developing rapid ascertainment of neural tube defects (NTD) to track the effectiveness of folic acid for NTD prevention in the population.

Third, birth defects registries can offer improvements over vital records when examining infant mortality in children with birth defects. Vital records are limited by a lack of completeness and accuracy in the reporting of birth defects.[9, 29,35] When "cause of death" was examined in the New York study, children with major malformations had higher mortality from causes other than congenital anomalies. The incidence of low birthweight, respiratory distress syndrome, other perinatal conditions, infectious diseases, and other systemic causes all were higher in children with major malformations.[24] For analysis of specific malformations, some studies use a case series from a hospital. However, such sources may be biased or incomplete in their ascertainment. Unless all of the children were born in the study hospital, mortality would be underestimated. A hospital case series would miss those cases born at outside institutions and considered unlikely to survive but never transferred for treatment.[36]

Fourth, because it is population-based, the NYCMR provides cases for descriptive and etiologic studies of birth defects; through the NYCMR, New York is one of seven states that participate with the CDC in the National Birth Defects Prevention Study.

SYSTEM ATTRIBUTES

Simplicity

For most efficient and timely reporting, the NYCMR report card should be filled out by hospital medical records personnel at the time the record is being reviewed and coded for billing purposes. The NYCMR staff has attempted to limit the number of items collected to the minimum needed. Demographic information is obtained by matching to the birth certificate file. If a case is selected for an etiologic study, additional information will be collected from hospital medical records to confirm the diagnosis.

Acceptability

Because the NYCMR relies on reports, staff members give periodic presentations and communicate frequently with hospitals about reporting issues. Hospitals have long complained about the burden of completing the CMR report card. In 1997, one of the hospital associations in New York State asked that the Commissioner of the Department of Health relieve hospitals of the requirement of submitting congenital anomalies by the report card. Completing the report cards is seen as burdensome, requiring too much staff time and resources. The new reporting systems described in the next section were developed to allow for the eventual phasing out of the report cards.

Flexibility

In response to declining resources for both the reporting hospitals and the NYCMR, new reporting systems have been developed. The first system abstracts congenital malformations information from hospital discharge data, which is already reported to the DOH, with hospitals entering the narrative diagnosis. The second system is a secure Web-based reporting system through which hospitals enter the data directly into a screen posted on the NYCMR website. While creation of these new systems was costly and time-consuming in the short run and caused interruption in routine CMR functions, the new systems will provide increased flexibility for database revision and data management, allow operations to continue with reduced staff, and relay more immediate feedback to the hospitals.

Representativeness

The NYCMR is a statewide, population-based registry that covers a large population of about 270,000 births in New York State each year. As stated earlier, state

regulations require reporting of all major malformations to the NYCMR. Around 25,000 such reports are received by the NYCMR annually on about 11,000 children with malformations. The population of the state is highly diverse; rare anomalies specific to certain population groups are represented among the large number of cases of congenital malformations received each year by the Registry. Therefore, the results of epidemiologic studies using NYCMR data can be generalized to the state population at large.

Sensitivity

Like many other registries, especially those that rely on reports from hospitals, the NYCMR has always been concerned about completeness and accuracy of reporting. After a decline in reporting was noted in the mid-1990s, a new system for monitoring reports was developed. The Department of Health's Statewide Planning and Research Cooperative System (SPARCS), a hospital discharge database, can be matched with NYCMR data to ascertain the quality of reporting by hospitals. Use of the SPARCS/NYCMR comparison has enhanced the regularity of hospital audits performed each year from 1992 through 1997, yielding a greater than 30% increase in the number of reports.

Honein and Paulozzi recently reported that the sensitivity of the NYCMR was 86.4% on the basis of a capture-recapture estimate.[10] This sensitivity estimate is similar to that of the Metropolitan Atlanta Congenital Defects Program (MACDP) for infants up to one year of age. The MACDP has been used as the "gold standard" against which other surveillance systems are evaluated. Live birth prevalence rates of specific malformations in the NYCMR are regularly compared to those of other state registries.

Table 2 shows the NYCMR 1996 birth cohort prevalences for selected malformations compared to both the MACDP and the California Birth Defects Monitoring Program (CBDMP). These two programs employ active surveillance methods to identify cases. Field staff members visit hospitals to ascertain cases, and then follow children through the first year of life. Because more than 95% of NYCMR cases are reported in the first year, valid comparisons are possible. The major difference among the three registries is the prevalence of neural tube defects. The NYCMR contains only live-born cases, whereas the other two include stillborn and terminated cases. Because a high percentage of these pregnancies are terminated, the inclusion, or not, of terminated cases will affect the prevalence.[37]

Given the experience that has been gained at the NYCMR and many of the improvements that have been made in its operations since the mid-1980s, it is reasonable to conclude that the Registry now identifies a larger proportion of children born with congenital malformations in New York State. The issue of whether to include terminated cases will continue to be of concern as the use of prenatal diagnosis becomes more widespread.

Predictive value positive

Although the NYCMR has developed strategies to improve completeness, it has not yet developed a systematic review of the accuracy of submitted reports. Previously, special Registry studies have found that 80% to 85% of reports are correct when compared to the medical record (unpublished data). This comparison of data is an important activity and a priority for the future.

Timeliness

Timeliness is an attribute that is frequently difficult to combine with completeness and accuracy, and it is often the first attribute sacrificed. As mentioned, the audit process, while improving the completeness of case ascertainment, decreases timeliness. The Registry also does routine matching to the birth certificate file, which is important for two reasons: first, it validates registry data and helps eliminate duplicate cases that are reported under different names. Second, it allows for the abstraction of information from fields that are on the birth certificate but not on the NYCMR report. For example, birth certificate data are used to establish maternal residence at the time of the child's birth. In a compromise between completeness and efficiency, the standard goal has been to ensure that at least 95% of each NYCMR birth cohort has a match in the birth certificates database.

Before the early 1990s, the matching process was arduous and time-consuming, and was largely responsible for the delay in the completion and dissemination of annual reports. Recognizing that timely information is an important part of a surveillance system, a DOH staff member created an algorithm that uses a point system to match NYCMR cases with birth data in the State Vital Records database. As a result, the NYCMR has been able to produce annual reports on cohorts of infants and children diagnosed with congenital malformations on a more timely basis.

COST OF OPERATING THE SYSTEM

The basic operations of the NYCMR are funded through the Maternal and Child Health Block Grant, Preventive Health & Health Services Block Grant, and

Table 2. Comparison of selected malformation prevalence in the New York Congenital Malformations (NYCMR) Registry with two other birth defects registries—MACDP and CBDMP[a]

Malformation	*NYCMR 1996*	*MACDP 1996*	*CBDMP 1995*
Anencephalus	0.6	2.2	2.0
Spina bifida	3.5	3.4	3.6
Hydrocephalus	7.8	8.1	5.8
Encephalocele	0.9	1.7	0.8
Microcephalus	5.8	8.8	—
An/Microphthalmos	1.4	3.4	2.8
Common truncus arteriosus	0.7	1.2	0.7
Transposition of the great vessels	4.0	6.1	2.8
Tetralogy of Fallot	4.2	5.1	3.8
Ventricular septal defect	38.8	32.3	16.9
Hypoplastic left heart syndrome	2.0	3.2	1.8
Coarctation of the aorta	4.3	4.4	4.7
Choanal atresia	2.2	2.2	1.0
Lung agenesis/hypoplasia	2.8	5.9	—
Cleft palate	6.1	6.1	7.3
Cleft lip ± cleft palate	8.2	9.8	10.0
Esophageal/tracheoesophageal atresia fistula	2.4	1.5	2.8
Rectal/large intestine atresia	3.3	2.2	3.4
Pyloric stenosis	17.8	14.9	17.8
Hirschsprung's disease	2.2	2.0	1.2
Biliary atresia	1.0	0.2	0.6
Renal agenesis/hypoplasia	3.0	6.1	—
Bladder exstroph	0.3	—	0.2
Hypo/epispadias	34.1	35.0	12.5
Reduction deformity of upper limb	2.7	3.9	4.1
Reduction deformity of lower limb	1.4	2.0	1.3
Diaphragmatic hernia	2.4	2.7	2.5
Omphalocele	1.4	1.7	1.6
Gastroschisis	1.6	1.7	2.6
Down syndrome	8.9	12.7	12.8
Trisomy 13	0.5	1.0	0.9
Trisomy 18	0.8	2.0	2.1
Fetal alcohol syndrome	1.6	3.2	0.8
Amniotic bands	0.4	1.7	1.2

[a]Metropolitan Atlanta Congenital Defects Program and California Birth Defects Monitoring Program

the State Superfund Program. The costs of operating the registry include salaries, travel for presentations about the NYCMR and hospital meetings, collection of surveillance data and the semiannual surveillance reports, data storage, compilation of annual reports, and responses to requests for data. Registry staff has declined over the years and now includes a full-time Program Director (1.0 full-time equivalent [FTE]), a Medical Director (1.0 FTE), Research Scientists (3.0 FTE), Clerks (2.0 FTE), and a Data Entry staff person (1.0 FTE). The approximate costs of basic registry operations during the 1998–1999 fiscal year were $474,250 for salaries, $140,378 for fringe benefits, and $157,775 for overhead, for a total of $772,403.

Program funding does not provide for major research activities. For the 1998–1999 fiscal year, research and special surveillance projects were funded through cooperative agreements with the CDC. In 1998, the NYCMR received $800,000 from the CDC to finance research activities related to the National Birth Defects Prevention Study, a study aimed at discovering the causes of congenital malformations. These funds

also allowed the NYCMR to collaborate with other DOH divisions, including the Wadsworth Center, which includes state-of-the-art research laboratories, and the Bureau of Child and Adolescent Health. In addition, the NYCMR received $100,000 for the Population-Based Surveillance of Fetal Alcohol Syndrome (FAS) and $50,000 for the Neural Tube Defect Surveillance Project. The Population-Based Surveillance of FAS is a collaborative effort of five states and the CDC to determine the prevalence of FAS within several geographically defined areas in the US. The Neural Tube Surveillance Project supports the development of NTD surveillance and prevention programs. Research staff for these three collaborative projects includes a Research Scientist (1.0 FTE), Field Staff for case ascertainment (3.5 FTE), a Clerk (0.5 FTE), and an Administrator (0.5 FTE).

SHOULD THE SYSTEM CONTINUE?

In an effort to meet the first two of its stated objectives—"the determination of the annual prevalence of congenital malformations" and "the monitoring of malformations according to type, geographic distribution, and community characteristics"—the NYCMR has published annual reports for the years 1983–1997. Each report presents the occurrence of congenital malformations among children in a specific birth cohort. The reports also present demographic characteristics of children reported to the registry, number of malformations, age at diagnosis, and distribution of specific malformations by geographic region. Recipients of NYCMR annual reports include, but are not limited to, hospitals, physicians' offices, several bureaus of the State Department of Health, County Health Departments, Maternal and Child Health programs, advocacy groups, and several non-profit organizations. To ensure timely dissemination of the surveillance data, the most recent reports are also accessible through the DOH website. The Registry's Birth Cohort reports are intended as a resource for programs providing primary, secondary, and tertiary preventive health care and for public officials concerned with reducing overall mortality and morbidity.

Semiannual reports of the NYCMR, which reveal time-space clusters and track annual trends in malformations prevalences, achieve the third program objective—"the investigation of suspected increases in the incidence of malformations that may be associated with toxic substances."

With regard to the fourth objective—"the conduct of epidemiological studies of specific malformations"—NYCMR data have been used for numerous descriptive and etiologic studies. Studies have included the investigation of mortality rates in children with congenital malformations,[24,35] descriptive epidemiology of diverse birth defects such as hypertrophic pyloric stenosis,[38] prune belly syndrome,[36] holoprosencephaly,[39] limb reductions,[40] and heart malformations;[41] etiologic studies of birth defects and electric bed heating,[42] pesticides and limb reduction defects,[43] studies of the risk of congenital malformations and proximity to hazardous waste sites,[44,45] and effects of prenatal diagnosis on the prevalence of Down syndrome.[46]

To meet the fifth objective—"responding to medical inquiries regarding congenital malformations"—the NYCMR designates staff members who respond to inquiries from health care providers and members of the public. Inquiries have ranged from concerns about environmental hazards to suspected epidemics of specific malformations in some hospitals. When warranted, the NYCMR has conducted follow-up investigations to respond to inquiries made by the public or providers.

In line with the sixth objective—"working with other DOH bureaus to ensure services and quality care for children with malformations"—the NYCMR recently completed data linkage analyses in collaboration with the Bureau of Child and Adolescent Health (BCAH) to determine whether the Bureau's Early Intervention program reaches a significant proportion of children with congenital malformations in the state. When it appeared that a significant percentage of children was not receiving such services, NYCMR staff worked with BCAH staff to develop information packets to send to families. The strict confidentiality law prevents NYCMR staff from releasing names to service programs, so service programs have no basis for contacting the families of affected children. Therefore, NYCMR staff have assumed responsibility for sending information packets to families about services available to their children.

To meet the seventh objective—"provide data for planning, advocacy, education, and other requests"—NYCMR staff provide data to other bureaus in the health department, other government agencies, organizations, advocacy groups, researchers, and interested individuals.

In general, the NYCMR is fulfilling its objectives by collecting, organizing, and distributing information on an important public health problem. The major strengths of the NYCMR are the mandatory reporting law, relatively low cost for coverage of a large population, flexibility to respond to changing conditions and resources, the narrative diagnosis for specificity, and ongoing efforts to track and improve completeness of case ascertainment. Weaknesses include the lack of an ongoing system for evaluating the accuracy of reported

diagnoses, timeliness, and non-inclusion of terminated cases. Members of the NYCMR staff have been forthright in pointing out limitations that should be taken into account in the use of the registry data.

SUMMARY AND RECOMMENDATIONS

As resources in many state health departments have declined and as states seek to cut programs to reduce costs, congenital malformations registries have come under close scrutiny and have been called upon to demonstrate their usefulness. Health department management may have the idea that the only function of birth defects registries is time-space surveillance to look for clusters. Registry staff will have to point out that these functions have expanded beyond the original search for new teratogens in the environment. This comparison with existing guidelines for surveillance systems was a useful exercise for the NYCMR and may be helpful for other congenital malformations systems as well. It could help registry staff to communicate with public health personnel working in other areas. However, congenital malformations systems have very specific issues, and these should be included in evaluations—issues such as diagnostic specificity when literally hundreds of conditions are included, coding, and case classification.[3,4] In the future, the National Birth Defects Prevention Network will be developing a set of guidelines and standards for congenital malformations systems, and these should provide guidance and help to improve systems.

The evaluation of surveillance systems, as illustrated by this article, includes both process evaluation and outcome evaluation. The routine conduct of evaluations is of utmost importance in ensuring that the activities of a congenital malformation registry, be it a passive or an active surveillance system, maximize case ascertainment. It is only when a registry can consistently collect complete and accurate data on the incidence of malformations in a defined population that there will be an opportunity to produce meaningful results.

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