

Analytical Resources for Assessment of Clinical Genetics Services in Public Health: Current Status and Future Prospects

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ABSTRACT

Context — Genetics services are not well integrated into the public health programs of most states, nor has there been effective use of clinical and program databases in the design, evaluation, and monitoring of public health genetics services at the state level.

Objective — To evaluate the availability and current use of population-based clinical genetics databases, including birth defects surveillance programs, in state-level public health genetics programs.

Design — Mail survey to state genetics coordinators in 50 states and 3 territories during 1996 with an update in 1997.

Results — Thirty states had birth defects surveillance programs; data from these resources were used in public health genetics program planning and management in only 15 states. Thirty states or territories had clinical genetics services databases. Most states had newborn screening program databases; few linked these records to vital statistics for programmatic purposes. Only 24 states had individual record databases for the Children with Special Health Care Needs program; 8 states had databases for maternal serum alpha-fetoprotein screening, and 7 had statewide cytogenetics registries.

Conclusion — Population-based databases concerning aspects of public health genetics are largely unavailable at the state level. Where these databases exist, they are poorly integrated into state public health genetics program activities. More attention should be paid to the development and use of clinical data programs for the assessment, monitoring, and assurance of genetics issues with relevance to population health.

The Council of Regional Networks for Genetic Services (CORN) recently published guidelines for genetics services in public health (CORN, '97). These guidelines describe the purpose, function, appropriate staffing, and management of statewide public health genetics programs. The critical role of information for assessment, planning, evaluation, and as-

surance was incorporated into the guidelines. A major impetus for the preparation of the guidelines was a broadly held perception that genetics services are not well integrated into the public health programs of most states, nor are clinical or program databases for genetics services effectively used in the design, evaluation, and monitoring of public health genetics services.

To assess the current status of population-based databases concerning genetics services and health outcomes, the CORN Birth Defects Surveillance Committee, with support from the CORN office and the clerical staff of the lead author's department, conducted a nationwide survey of state genetics coordinators during the summer of 1996 with updating in early 1997. While the survey focused on the structure, methodology, data contents, and use of birth defects registries, these elements were viewed from the perspective of the public health genetics program in each state. Results of this survey are presented here.

METHODS

A survey was mailed from the CORN office to state genetics coordinators in all states, the District of Columbia, Puerto Rico, and the Virgin Islands. This survey included a series of questions about the birth defects surveil-

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lance program (if any) and the uses of these data by the public health genetics program. Respondents were instructed to contact the staff of their birth defects program for assistance in completing this portion of the questionnaire. The survey solicited information about a number of questions concerning birth defects surveillance programs, such as what agency the program is located in, whether the program is statewide or, if not, population-based for how many annual live births, what case-ascertainment methodology is used, the ages of children whose cases are ascertained, what birth defects and/or genetic diseases are under surveillance, what coding methods are used, how many years for which ascertainment has been completed, and whether data are linked to vital statistics records. Questions concerning the use of birth defects surveillance data in the public health genetics program included the following: Are data from the surveillance program used for planning and evaluating genetics services?; Is an annual report prepared?; and Does the surveillance program prepare special analyses on request? Questions concerning population-based clinical genetics databases ascertained the existence of statewide genetics services databases and their uses, as well as specific databases for newborn screening, maternal serum alpha-fetoprotein screening, cytogenetics, and the state Children with Special Health Care Needs (CSHCN) program. Additional information was collected regarding routine linkages among these databases and between the databases and vital-statistics and birth-defects surveillance program records.

Information concerning birth defects surveillance programs in each state was cross-referenced with data published in the first report of the National Birth Defects Prevention Network (Edmonds, '97); respondents were contacted by telephone to resolve any discrepancies. Where information concerning other public health genetics databases was unobtainable after repeated follow-up calls and e-mail,

we assumed that these data resources and/or related record linkages are not present in the states concerned. Results presented here are current as of the spring of 1997; although minor changes in programs and state activities may have occurred since then, these are likely to have been incremental and do not affect our general conclusions.

Definitions

In comparing birth defects surveillance systems and other population-based genetics services databases, we used the following standards and definitions:

Case-finding methodology:

- 1) Active: trained staff members visit health care facilities and abstract records on-site.
- 2) Passive: reporting sources file reports on program-specified forms.
 - a) aggressive: routine follow-up is conducted, with mandatory reporting and penalties for noncompliance
 - b) compliant: forms are accepted basically as filed, with minimal or no follow-up to ensure completeness or accuracy of reporting.
- 3) Impassive: information is received via automated record linkage from data sources designed for purposes completely unrelated to the surveillance of the disease or health condition.

This typology differs from the traditional approach in that the role of the surveillance program in collecting and assessing the data is more clearly delineated. Traditionally, case-finding strategies have been classified as "active" or "passive" (Teutsch and Churchill, '94; Halperin and Baker, '92). This traditional typology, however, does not identify whether data are collected for administrative or clinical purposes, nor does it reflect the fundamental relationships between surveillance programs and data-collection strategies (Iezzoni, '97). The active/passive/impassive typology used here highlights differences in data-collection strategies and allows researchers to grasp

quickly the potentials and limitations of surveillance systems in each state. The distinction between passive and impassive case-finding strategies is especially important. Surveillance programs using databases that collect information for other, primarily administrative purposes as inputs into case-finding algorithms must of necessity accept the data as reported. They have no opportunities to evaluate the diagnostic specificity of coding systems or their appropriateness for the surveillance of the disease(s) of interest. For example, if renal agenesis were a condition of interest, an impassive case-finding strategy that relies on databases using ICD-9-CM codes will be unable to differentiate between unilateral and bilateral cases or to distinguish between cases of renal agenesis and renal dysgenesis. A passive case-finding strategy that requires reporting sources to provide a text description of the birth defects will permit a more specific taxonomy of renal agenesis cases (Cunniff et al., '94).

Population-based database:

In order to be population-based, a data system must collect information concerning all occurrences of the primary condition or event of interest. Vital statistics registration systems are to all intents population-based, as these programs require the documentation of all live births, fetal deaths, and deaths occurring in the jurisdiction, with arrangements for the addition of records concerning members of the population for whom the vital events occur elsewhere. For a birth defects registry to be population-based, it must be able to obtain information concerning all birth defects cases diagnosed for the population of interest. A registry maintained by a hospital or health care organization may or may not be population-based, depending on the presence of other health facilities in its catchment area and the health-care-seeking propensities of the inhabitants of the region. Here, we considered birth defects registries to be population-based if they collect information from all obstetrical/pediatric hospitals in the

state or substate area, even if there are no formal arrangements for exchanging information with registries in adjoining states about residents of the surveillance area who receive care in the adjoining state. The same holds for databases concerning newborn screening, cytogenetics, and Children with Special Health Care Needs. In the case of clinical genetics services databases, our minimum expectation was that an individual-record database containing information on patients receiving services at all publicly funded or university-based centers in the state exists and could be accessed by the state genetics services coordinator.

Routine record linkage:

Record linkage refers to the processes whereby information concerning one individual or event that is stored in one database is associated with records on the same individual stored in another database. Some databases of interest to public health genetics contain data on all individuals, while others contain data only on affected individuals, those participating in a program or service, or those with a positive laboratory result. Here, we considered databases to be linked if the resultant data set contains records on the entire population (i.e., live-born infants) with genetics data appended to those records for whom it is available. To clarify, we did not consider a newborn screening program to be linked to vital statistics if only those infants with abnormal test results are linked, but we would if all newborn screening results are linked.

RESULTS

We received survey responses from 53 reporting units, including all states, the District of Columbia, Puerto Rico, and the Virgin Islands. For the purposes of this report, we use the word "state" to refer to all reporting units.

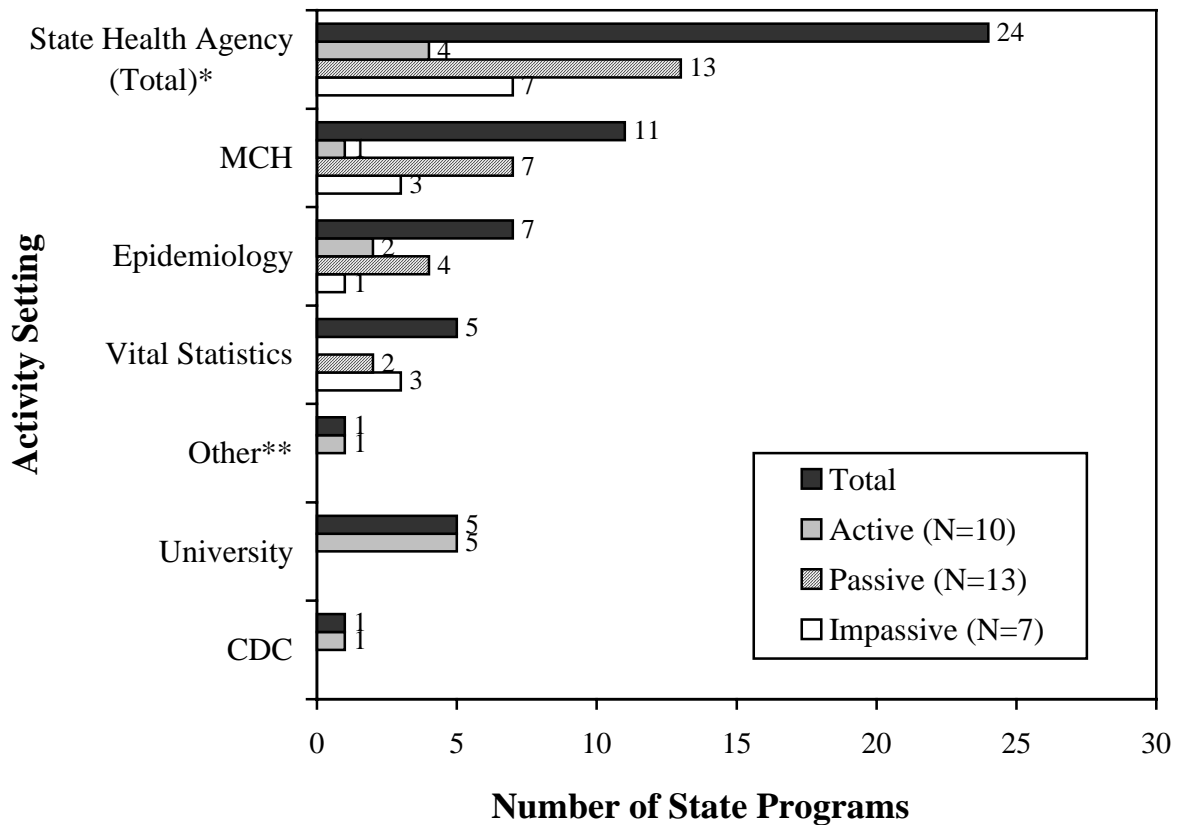
Birth Defects Surveillance

As of the first quarter of 1997, there were 30 birth defects surveillance programs. Of these, 10 used active, 13 used passive, and 7 used impassive methods of case-finding. Two states that previously had BDS programs no longer had them. Three additional states were in the planning stages. The remaining 18 states had no system or indicated no plans to implement one in the near future. Of the 30 BDS pro-

grams, 25 are statewide. Most programs are of recent origin; two began operations in 1996, and seven others have collected data only for birth cohorts beginning in 1993 or later. Data for births occurring before 1985 are available from only nine states.

Birth defects surveillance programs are housed in a variety of settings (Figure 1). It is interesting to note that none of the 5 programs housed in state health statistics units use active

Figure 1. Birth Defects Surveillance Programs: Agency Setting and Type of Surveillance Case-Finding Methodology.



* Includes MCH, Epidemiology, Vital Statistics, and Other.

** Administered by March of Dimes, under contract from state health agency.

case-finding methods and that only 4 of 24 programs housed in state health agencies use these methods. In contrast, all university programs use active case-finding methodologies, as does the Centers for Disease Control and Prevention's (CDC's) Metropolitan Atlanta Congenital Defects Program. Programs use a variety of definitions, sources, coding strategies, and age limits; although the standards recommended by CDC are more common, uniformity is by no means evident (Lynberg and Edmonds, '92; Lynberg and Edmonds, '95).

Of the 30 BDS programs, 21 indicate routine record linkage with vital statistics data (birth, fetal death, and infant death certificates); 4 additional programs plan to implement this linkage soon. However, state genetics services coordinators reported that BDS data are used in managing and planning for the state public health genetics program in only 15 of the 30 states with BDS programs. The specific uses for these BDS data varied considerably, with most applications involving descriptive statistics and relatively low-level applications.

Genetics Services

Thirty of the 53 reporting units indicated that a statewide clinical genetics services database is maintained in their state; all others reported that no statewide database for genetics services currently exists. However, many of the 30 statewide databases result from the participation of the single university-based clinical genetics center in the CORN minimum data set and are not used for planning or management purposes by the state public health agency. Among the 23 states with no database, 12 indicated that such a database is planned. Of the 30 states with BDS programs, 20 also had clinical genetics databases. A total of 13 states had neither a BDS program nor a clinical genetics database, while 10 had a BDS program without a clinical genetics database, and 10 had a clinical genetics database but no BDS program.

The survey also collected information on the

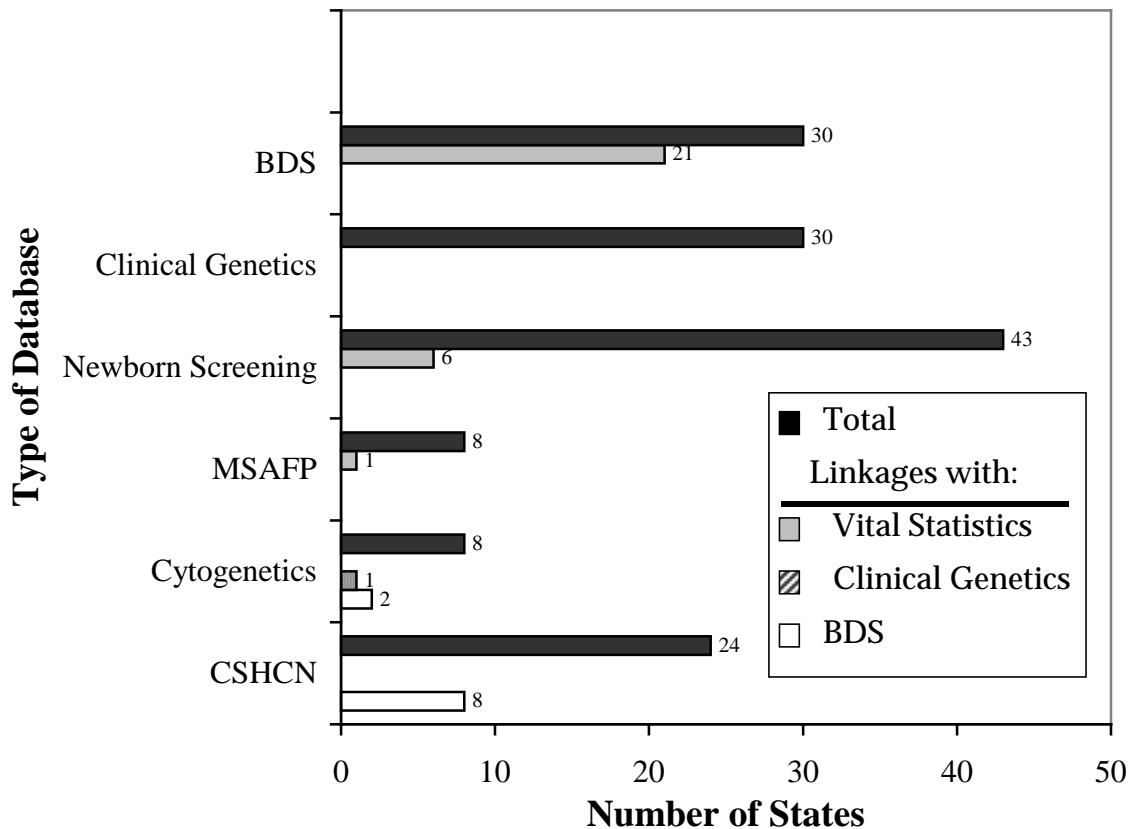
existence of several other population-based databases for genetics services in maternal and child health. These included newborn screening, maternal serum alpha-fetoprotein (MSAFP) screening, cytogenetics (chromosome), and Children with Special Health Care Needs (CSHCN). The results are shown in Figure 2. Most states have newborn screening (NBS) databases, although these are rarely linked to vital statistics records. Only eight states indicated that an MSAFP registry exists; only one state routinely links these data to vital statistics records. Seven states have statewide cytogenetics registries; only one state links these data to the state clinical genetics services database, and two use the registry as a source or resource for the BDS program.

Surprisingly, only 24 respondents indicated the existence of a statewide CSHCN database; 14 indicated there was none, 2 indicated that a database is in the planning stage, and the other 13 did not know. CSHCN records are linked with BDS data in only eight states. Sixteen states with clinical genetics databases also had CSHCN databases, while seven did not; the remaining 7 did not know whether a CSHCN database existed in their state.

PUBLIC HEALTH IMPLICATIONS

If maternal and child health programs define their populations broadly and focus on the range of health services across the continuum of care, more attention must be paid to the development, maintenance, and use of population-based data systems concerned with health aspects of genetics services. Friede et al. ('95) describe an emerging field, which they call *public health informatics*, in which public health databases and information services are seamlessly integrated into public health administration, decision-making, planning, evaluation, and assurance activities. In public health genetics, this will require a focused approach to data collection, database man-

Figure 2. Population-based Clinical Genetics Databases and Routine Record Linkages.



Note: Respondents were asked about specific record linkages; responses that were inconclusive are not included. States that are planning linkages or do partial linkages are not included.

agement, record linkage, and system administration that emphasizes the broad public health needs, issues, and concerns. As the survey results indicate, informatics for public health genetics is in its infancy in most state public health agencies. Comparatively few studies have examined the manner in which initiatives to collect, manage, and use population-based data for maternal and child health programs function at the state level (People and Miller, '83; Peoples-Sheps et al., '86). These studies do not address issues of concern to public health genetics programs in any detail.

Our survey reveals some weaknesses and un-

anticipated deficiencies in the availability and uses of population-based genetics databases across the United States. Not only do almost half the states have no statewide clinical genetics databases, but these databases are rarely used in planning, monitoring, or evaluating the public health genetics programs in each state. Most states do not have databases for cytogenetics or MSAFP screening. These are extremely difficult to establish now because of the complexities of the commercial laboratory environment, of health maintenance organizations, and of managed care and insurance contracts with diagnostic laboratories on a re-

gional or national basis, as well as legislative restrictions for data collection due to privacy/confidentiality concerns. Birth defects surveillance programs cannot, by themselves, take the place of MSAFP or cytogenetics registries. These databases have very different purposes, with an MSAFP database being particularly useful for assessing the degrees of access to and use of prenatal screening services, and a cytogenetics registry providing diagnostic confirmation of chromosomal abnormalities that might be lacking from in-patient hospital records or reports of birth defects surveillance programs.

A few states routinely link NBS records with vital statistics. This is an essential part of any population-based screening program. Without linking birth certificate records to newborn screening test results, it is impossible to determine the success of NBS programs in achieving their goals. The Maternal and Child Health Bureau is to be commended for creating a new state-level performance measure in this area; this standard should be upgraded to require a record linkage in order to meet the reporting requirement. It is insufficient merely to report the number of screening tests performed and divide that number by the number of live births in each jurisdiction. Without linking to birth certificates, programs cannot effectively ensure that re-screens are not counted as initial tests. This linkage would also identify infants born in other states but screened by the state program, as well as systematic patterns in screening efficiency by hospital, county, or region, or patient characteristics such as low birth weight, extreme immaturity, early discharge, neonatal transport, primiparity, plurality, or other risk factors. Spady et al. ('98) provide an excellent example of the opportunities for NBS program management through record linkage. In this survey, we did not ask about the existence or current function of registries for diseases screened by statewide NBS programs. These registries should also link to vital statistics records, both to obtain patient demographic in-

formation and characterize those with each disease, but also to ascertain the vital status of children with each disease or disorder. These registries should also follow patients on a routine basis to ascertain their health status and use of health, nutrition, and other services, and to provide health-education and disease-prevention information to families.

Finally, public health genetics programs do not make good use of birth defects surveillance data even when these data are available. Birth defects surveillance programs are rarely administered within the same organizational structure as public health genetics, nor is the focus of most registries oriented toward services, health education, or disease prevention/health promotion activities.

Public health genetics does not fit neatly into standard organizational charts for maternal and child health. The necessary data resources are typically complex and clinically or laboratory-based. More attention must be paid to the informatics environments within which these programs operate, and databases must be linked and otherwise shared with other public health programs involved in related activities. Confidentiality of information in individual records in these databases is a paramount concern, but this concern should not prevent the creation of clinical databases concerning genetics services. Without developing population-based data systems focusing on specific aspects of genetics services, health officials will be unable to meet the challenges that genetics technologies and changing health care delivery systems will pose for public health in the next century.

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