Rare diseases, most of them genetic, pose a considerable burden to families, including diagnostic delays, lack of access to specialty care, and social and economic burdens. Despite the recommendations put forth to monitor rare diseases, there exists a lack of systematic collection of population-based epidemiologic data for many of these diseases. In particular, newborn screening for rare, single gene conditions has expanded rapidly in recent years, including integration of screening for early detection of hearing loss. The increase in the detection of newborn screening disorders has not been matched with an increase in systematic monitoring of outcomes of confirmed cases to better understand the epidemiology of such disorders. To accomplish this, requires integration of data collected by newborn screening programs with resources provided by established disease surveillance programs. This project, the Iowa Newborn Screening Surveillance Project (INSSP), proposes to expand the Iowa Registry for Congenital and Inherited Disorders (IRCID) to include confirmed cases from the Iowa Newborn Metabolic Screening Program (INMSP) and the Early Hearing Detection and Intervention (EHDI) Program identified from among all Iowa live births delivered from January 1, 2006 through December 31, 2008. Through its recent experience with expansion to include surveillance for muscular dystrophy and stillbirths, the IRCID is poised to begin surveillance activities for confirmed newborn screening cases. The overall goals of the INSSP are to: 1) conduct Iowa population-based (statewide) surveillance of confirmed newborn screening cases to advance the public health understanding of the impact and ongoing outcomes of children with these conditions; 2) provide accurate surveillance data to inform public health officials, health care providers, policy makers, and families; and 3) provide ongoing outcome information to allow for the monitoring and quality improvement of referral patterns to health care providers. To accomplish these goals, we propose to implement the following objectives during the initial project period: 1) produce timely data with well-defined, standardized data elements such as demographic information, genotypes when applicable, and timing and patterns of referral for specialty care; 2) enhance collaboration between the INMSP and the EHDI Program and the IRCID to integrate the collection and reporting of data for newborns with all confirmed screened disorders into the existing surveillance infrastructure, including modification of the existing IRCID Parental Notification Program; 3) contribute data at least semiannually to a centralized dataset providing de-identified, individual level data using standardized data elements; 4) involve appropriate partners, such as health care providers, the Heartland Regional Genetics and Newborn Screening Collaborative, neighboring states, and other organizations to assist with the collection of data; 5) enhance the referral component for affected children to ensure timely referral to services and include an evaluation component to assess service utilization; 6) disseminate quality data on enhanced population-based surveillance and ongoing outcomes of affected individuals; and on at least an annual basis, communicate information on project activities and results to partners and other state programs; 7) conduct a systematic project evaluation to include an assessment of the advantages and disadvantages of the pilot project including a description of the problems encountered, lessons learned, and potential improvements; and 8) if funds are provided beyond the initial project, continue the above activities for additional birth cohorts, and explore the feasibility of expansion to include South Dakota newborn screening data. Successful completion of the INSSP will promote the health and welfare of infants and children in Iowa.
Project Abstract

Enhancing California NBS Case Surveillance and Short Term Follow-up Data System
Funding number: CDC-RFA-DD08-810

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California’s Genetic Disease Screening Program (GDSP) runs one of the largest newborn screening (NBS) programs in the world, screening over 550,000 newborns each year for metabolic, endocrine and hemoglobin disorders, as well as cystic fibrosis. The program is supported by a comprehensive, web-based, computer application referred to as the Screening Information System, or “SIS” which supports an existing short-term follow-up data collection system that tracks all initial screen positive cases through the time the case is resolved as a confirmed disorder or until a disorder is definitively ruled out. Metabolic follow-up centers began using the short-term follow-up system in 2005; cystic fibrosis was added in 2007.

The short-term follow-up system allows follow-up centers to enter important case management milestones, including types of services provided, the type of professional delivering the service, the circumstances under which the services were provided; the mode of service delivery, the types of tests ordered, the types of treatments initiated; the newborn symptoms and health problems at each point of service; and an overall health assessment of the child. This data allows the GDSP to evaluate how services are being delivered and to better understand the natural history of the disorders detected through screening.

The project objectives focus on activities that support the enhancement of the existing data system through systematic identification of data problems and the development of tools to promote the standardization of the data. In addition, the GDSP will serve as a pilot case-finding site for confirmed disorders, and de-identified demographic and clinical data will be reported to the CDC on a bi-annual basis.

Due to the large scale of the California newborn screening system, it has been difficult for the GDSP to control the quality of the data being entered by the large number of users across the State. Problems such as incomplete and inconsistent data limit the validity of the data. In addition, some questions require a subjective assessment of the child and specific parameters need to be developed to help standardize responses.

The funds provided through the CDC will be used to hire one full time staff position; 50% of staff time will be devoted to data analysis and 50% will be spent on project development and data liaison activities. A formal data quality review process will be followed by the development of a Guidelines for Data Entry Manual, and staff training activities. A project Advisory Committee will provide input into the development of statewide and site-specific reports summarizing short-term follow-up milestones. Lastly, the short-term follow-up system will be expanded to include endocrine and hematology follow-up centers. With this expansion, the GDSP will have comprehensive follow-up information on all of the disorders that we target through our newborn screening program. Ultimately, the quality and completeness of the data we collect will determine its usefulness. This final step will help advance our understanding of the impact and short-term outcomes of children with newborn screening conditions.
Abstract

Studies have shown that linking data from population-based surveillance programs such as birth defects registries and newborn screening programs was helpful in understanding the epidemiology of some confirmed screening conditions and associated birth defects, and identifying possible causes of these congenital disorders. The main objective of this proposed project is to develop and implement a population-based surveillance and tracking system in New York State for children with confirmed newborn screening conditions through enhanced collaboration between the established newborn screening and birth defects surveillance programs. This integrated system is necessary to advance the public health understanding of the impact and short-term outcome of children with newborn screening conditions, ensure early access to services, treatment and cost savings for the health and education systems, prevent or reduce birth defects and developmental disabilities and ultimately help to meet Healthy People 2010 objectives and fulfill New York State Department of Health’s (NYSDOH) obligation to protect the health of all New Yorkers.

New York State is one of the largest states by population in the United States. Annually, about 250,000 children are born to all New York State residents, with about 125,000 these born to non-New York City residents. The New York State birth cohort (excluding New York City births) will be used as the study population for this project. The study cohort is diverse with regards to race and ethnicity. This diversity provides an opportunity to study population variation. The characteristics of the New York State Congenital Malformations Registry (CMR) and Newborn Screening Program (NSP) make them especially valuable for the proposed project: (1) Both are statewide and population-based; (2) The CMR receives a large number of cases (~10,000) of congenital malformations each year so even rare malformations occur with some frequency in this large population; (3) State regulations provide a mechanism to enforce reporting requirements to the CMR and the NSP and Department of Health enforcement procedures are well-established; (4) Children are reportable to the CMR up to 24 months of age, which allows time for malformations not readily apparent at birth to be detected; (5) A majority of the congenital disorders that NSP tests for are reportable to the CMR; and (6) The CMR’s database has been linked to other NYSDOH data sets such as birth and death certificates, hospital discharge files, as well as NSP data.

The collaborative team will include the NSP, the CMR and the DFH of the NYSDOH. To achieve the proposed objectives, the proposed activities include: (1) ensuring programmatic access to population-based surveillance data and vital records needed for the project, (2) developing data linkage programs using deterministic methods that use multiple criteria to establish a match between records, (3) matching the dataset of confirmed newborn screening conditions from the NSP to the CMR database to add CMR cases that were not ascertained by routine methods, to the death certificates to monitor the mortality and to the hospital discharge files to monitor the morbidity, (4) developing an integrated database that uniformly combines information from a multitude of sources such as the NSP, the CMR, death certificates and the hospital discharge data, (5) developing database management tools for data updating, retrieval and quality control, statistical analysis, and data submission and dissemination, (6) developing evaluation methods and performing program evaluation, (7) generating reports for statistical analysis and data dissemination and communication and (8) producing timely data with required standard data elements for submission.
Utah Newborn Metabolic Surveillance: Partnership of Newborn Screening and Birth Defects Surveillance

Newborn screening is the largest population-based genetic screening program in health care. It currently experiencing an unprecedented expansion—many metabolic conditions, which were the near-exclusive purview of specialized clinics until only a few years ago, are now being incorporated into mass screening. Such fast-paced expansion of newborn screening is outstripping the current public health understanding of many of these metabolic conditions.

There is an urgent need to understand the epidemiology and health service utilization of these conditions, using high-quality, population-based data. The proposed study is aimed at characterizing the distribution, genotype, and phenotype as identified in well-defined, population-based cohort of births undergoing expanded newborn screening (which can be very different from the information gathered by clinic-based series of cases identified because of clinical presentation). The study will also assess the health services utilization of this population, including the utilization of federally and state-funded services (e.g., Medicaid). Finally the study will evaluate outcomes, including morbidity and mortality at least through infancy. The overall programmatic objective is to pilot the enhancement of newborn screening surveillance based on the established infrastructure of birth defects surveillance.

The study will be conducted through a partnership with the newborn screening program and the birth defect surveillance program of the Utah Department of Health, as well as with clinical and laboratory partners at the University of Utah. The population will include Utah’s state-wide cohort of births from 2006 (when expanded newborn screening begun in Utah) through 2009, for a total of at least 200,000 births. The three-year cohort of births before expanded newborn screening (2003 to 2005) will provide a comparable reference group to assess the changing epidemiology and public health aspects of these conditions. The findings will be distributed to public health and clinical partners locally and nationally. The project has the potential to be expanded through collaborations with other states and potentially also with other federal organizations in the future.