Tracking clinical genetic services for newborns identified through newborn dried bloodspot screening in the United States—lessons learned

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Abstract To determine how US newborn dried bloodspot screening (NDBS) programs obtain patient-level data on clinical genetic counseling services offered to families of newborns identified through newborn NDBS and the extent to which newborns and their families receive these services. These data should serve to inform programs and lead to improved NDBS follow-up services. Collaborations were established with three state NDBS programs that reported

systematically tracking genetic counseling services to newborns and their families identified through NDBS. A study protocol and data abstraction form were developed and IRB approvals obtained. Data from three state NDBS programs on a total of 151 patients indicated that genetic services are documented systematically only by metabolic clinics, most often by genetic counselors. Data from 69 endocrinology patients indicated infrequent referrals for

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genetic services; as expected higher for congenital adrenal hyperplasia than congenital hypothyroidism. Endocrinology patients were often counseled by physicians. While systematic tracking of genetic counseling services may be desirable for quality assurance of NDBS follow-up services, current systems do not appear conducive to this practice. Clinical records are not typically shared with NDBS programs and tracking of follow-up clinical genetic services has not been generally defined as a NDBS program responsibility. Rather, tracking of clinical services, while recognized as useful data, has been viewed by NDBS programs as a research project. The associated IRB requirements for patient-related research may pose an additional challenge. National guidance for NDBS programs that define quality genetic service indicators and monitoring responsibilities are needed. US experiences in this regard may provide information that can assist developing programs in avoiding tracking issues.

Keywords Newborn dried bloodspot screening · Follow-up · Genetic services · Genetic counseling · IRB

Introduction

Newborn Dried Bloodspot Screening (NDBS) is an essential preventive public health program in most developed countries that identifies infants at high risk for particular congenital and hereditary conditions. NDBS program development is continuing to expand in developing countries, particularly in the Asia Pacific (Padilla and Therrell 2007; Padilla et al. 2010), Middle East and North Africa (Saadallah and Rashed 2007; Krotoski et al. 2009), and Latin America (Borrajó 2007), providing early identification of conditions for which timely interventions can help eliminate or reduce associated mortality, morbidity, and disabilities. NDBS systems are generally viewed as having six components that include education, screening, follow-up, diagnosis, treatment/management, and evaluation/continuous quality improvement (Therrell 2001; Pass et al. 2000; Therrell et al. 1992). Data that document related activities, including the extent to which clinical genetic services such as genetic counseling are available and utilized, are important for quality assessment and improvement.

The American Academy of Pediatrics' Newborn Screening Task Force's (2000) report on US newborn screening noted, as an underlying assumption, that the primary objective of each state's NDBS system should be to ensure that every newborn and his/her family receive appropriate and timely services that relate to improving outcomes from his/her condition. Further, they noted that state public health agencies should assume responsibility for the core public health elements of assessment, assurance, and policy

development in the context of NDBS. No particular component of the public health agency was assigned responsibility for implementing or monitoring the accomplishment of these three tasks. However, state public health agencies are required, under Title V of the Social Security Act, to report annually on 18 national performance measures. National Performance Measure #1 is "the percent of screen positive newborns who received timely follow-up to definitive diagnosis and clinical management for condition (s) mandated by their state-sponsored newborn screening programs" (Title V Today 2010). However, the components of clinical management and assessment methods are not defined in the Act.

It is generally recognized that clinical genetic services are among the timely and appropriate services required by newborns and their families who have been identified with most conditions through NDBS (Pass et al. 2000; Ciske et al. 2001; American Academy of Pediatrics, Section on Hematology/Oncology, Committee on Genetics. 2002). The core functions of clinical genetic services have been defined as including accurate clinical and laboratory diagnosis, risk estimation, genetic counseling, linkage of patients with genetic service providers, linkage of patients with medical home, communication with subspecialty providers, and maintenance of DNA storage service (Kaye et al. 2007). Thus, for example, a family with a newborn diagnosed by NDBS with congenital adrenal hyperplasia (CAH), in addition to appropriate endocrinology subspecialty services, should also receive genetic counseling on recurrence risk in future offspring.

If, in the USA, it is the responsibility of state public health agencies to assure appropriate and timely services, it seems reasonable that some (if not all) state NDBS programs would document whether newborns and their families received the requisite clinical genetic services. In practice, monitoring and documenting provision of clinical genetic services would be expected to be a part of NDBS follow-up activities. Follow-up, both short- and long-term, is vital in facilitating early diagnosis, intervention, and management of affected newborns and their families. Short-term follow-up (STFU) and long-term follow-up (LTFU) have been variously defined, with definitions changing over time (Pass et al. 2000; Tuerck et al. 2006; American College of Medical Genetics' Expert Group. 2006; Kemper et al. 2008). The endpoint for STFU is understood generally to be the point at which a definitive diagnosis has been documented and appropriate treatment/management has been initiated. LTFU begins where STFU ends and extends throughout the lifetime (Tuerck et al. 2006).

The only published national/international guideline on newborn screening follow-up notes that, "All follow-up communications about an individual newborn should be documented in the newborn screening follow-up program's

record, according to applicable rules and regulations. The record should be kept current and at a minimum, contain the following: diagnosed cases, referral information (e.g. subspecialty providers, support services, *genetic counseling*), enrollment in early intervention, and long-term follow-up activities. For long-term follow-up of affected individuals, programs should collect data regarding use of appropriate follow-up services. These data should be collected in collaboration with primary care and subspecialty providers, other medical and support service providers, and/or parents/affected individuals” (Tuerck et al. 2006).

The U.S. Secretary of Health and Human Services’ Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children (SACHDNC) more recently defined LTFU as, “... comprising assurance and provision of quality chronic disease management, condition-specific treatment, and age-appropriate preventive care throughout the individual’s lifespan.” The SACHDNC further acknowledged LTFU as necessary to maximize the benefit of NDBS diagnosis, expanding the concept from data management to systematic, comprehensive care of affected individuals (Kemper et al. 2008).

By nature of their centrality in the larger state public health system, NDBS programs have been noted as strategically positioned to conduct assessment, assurance, and policy development of the entire NDBS system, all core public health functions (Institute of Medicine 1988). However, in a 2007 online survey of 35 state NDBS programs, 56% of responding programs reported that they collected no LTFU data. At issue is the extent to which state public health departments (and NDBS programs) should be involved in ensuring that NDBS is achieving its full potential (Hoff et al. 2007).

There are no published data regarding the extent to which genetic services, including genetic counseling, are provided to families of newborns whose disorders were identified through NDBS. In 2007, Kaye et al. surveyed state NDBS and/or state genetics programs to determine the extent to which they systematically documented clinical genetic services received by newborns and their families detected through NDBS (Kaye et al. 2007). This survey built on earlier research identifying multiple barriers to a state NDBS programs’ ability to do LTFU, such as financial constraints and lack of comprehensive quality assurance practices (Hoff et al. 2006). An ad hoc group of experts (details in report) developed a comprehensive definition for clinical genetics that identified the relevant service components (see Table 1). Survey data indicated that no state NDBS and/or state genetics program routinely tracked patient-level data on genetic services, including genetic counseling, for all of the NDBS conditions in their screening panel. However, 23% (eight of 35) reported systematically documenting whether newborns and their families were referred for and/

Table 1 NNSGRC Working Definition of Adequate Genetic Services

Genetic services for individuals and their families identified as affected by newborn screening include an integrated clinical and laboratory services system which offers the following core functions and associated attributes

Core Functions

- Accurate clinical diagnosis
- Accurate genetic laboratory diagnosis
- Risk estimation
- Genetic counseling
- Linkage of patient to genetic service providers
- Linkage of patient with medical home
- Communication with other specialty providers
- Maintenance of a DNA storage service

Attributes of the Genetic Service System (may be ongoing simultaneously with core functions)

- A family based approach where required
- Accessible information for families, other health professionals and patient support groups
- Support to individuals and families
- Prevention of a disorder or complications including family follow-up (e.g. Anticipatory care, prenatal care and testing)
- Participation in research and clinical quality assurance
- Maintenance of confidential family records

Kaye et al. 2007

or received genetic counseling for some of the screened conditions. Of these, 63% (five of eight) reported tracking genetic counseling with follow-up reporting forms, with 40% (two of five) of these using a searchable database. Of the three programs without searchable databases, various documentation methodologies were reported including: (1) as a contract requirement for the first visit for a condition identified by tandem mass spectrometry NDBS (other information was also available because of the NDBS program’s close working relationship with university genetic centers); (2) as part of the responsibilities of a contracted geneticist and NDBS care coordinators for metabolic, endocrine, and sickle cell/hemoglobinopathy disorders, and from cystic fibrosis follow-up centers; and (3) as part of a searchable database on genetic evaluation referrals and through patient records (genetic counseling) maintained by the state NDBS program.

Of the states that indicated systematic tracking of referral and/or receipt of genetic counseling, detailed information on the clinical genetic services provided was clearly lacking. In this report, we document our attempt to better understand the issues in acquiring and assessing patient-level follow-up data for genetic counseling, a surrogate for clinical genetic services. We analyzed patient-level data to better assess the extent to which affected children are receiving this service in US NDBS programs.

Methods

In order to expand on the 2007 survey data regarding tracking of clinical genetic services after NDBS (Kaye et al. 2007), we created a brief, second-tier survey that was submitted by email and telephone to the eight state NDBS programs who previously indicated that some genetic service tracking data were available. Our intent was to assess interest in, and program requirements for, sharing patient-level data on genetic counseling services. From the survey responses, the list was reduced to seven programs with potential interest, and ultimately to three state NDBS programs (Minnesota, Missouri, Rhode Island) willing and able to collaborate in obtaining and sharing data (including IRB approvals).

The Minnesota Department of Health NDBS Program (MN-NDBS) agreed to serve as the pilot program for collecting additional clinical data. They initially determined that de-identified data would be provided for infants identified with metabolic conditions and CAH from January 1, 2003 – December 31, 2005. Data were limited to infants with confirmed diagnoses. A medical record data abstraction form was developed, adapted from published guidance to states for tracking clinical genetic services for newborns (Kaye et al. 2007) (Table 2). Remuneration and/or personnel for data abstraction were offered but declined because of possible conflicts of interest.

A succinct proposal for providing data was developed from this pilot for consideration by the six other state NDBS programs that had initially expressed interest in the project. Contacts in these states were approached by email and telephone to further explore the feasibility of their participation. Interested states received a formal collaboration proposal for their consideration.

The Missouri State Department of Health and Senior Services' (MDHSS) NDBS program staff (MO-NDBS) and the Rhode Island Department of Health (RIDOH) state genetics contact, who was also responsible for NDBS follow-up, both expressed interest in the project in relatively short order. The collaboration offer was repeated several times to the remaining four states between September 2007 and February 2008. Two states declined participation, and two states did not respond to repeated communication efforts.

Results

States' ability to collect data

The MN-NDBS follow-up coordinator assumed responsibility for the project, obtained expedited institutional review board (IRB) approval from the University of Minnesota (UM), and abstracted the required data from UM medical

records. Final de-identified, patient-level data on metabolic disorders from January 1, 2004 to December 31, 2006 and CAH from January 1, 2000 to December 31, 2005 were provided in June of 2007.

During August and September 2007, two conference calls were made to the MDHSS genetics coordinator and MO-NDBS follow-up staff to move forward with the project. As a caveat to participation, the MDHSS required IRB review and exemption in order for the MO-NDBS to provide a patient listing for the clinical genetic service providers. To facilitate this process, we prepared an abstract of the project protocol, a template of the protocol and provided assistance in preparing the IRB application. The collaboration proposal was approved at the November 2007 meeting of the state genetics advisory committee.

Invitations were extended to four university-based clinical genetic service centers and four pediatric hemoglobinopathy centers to consider project participation. All four university-based clinical genetic service centers and one hemoglobinopathy center expressed interest in participating. Ultimately, only the four university-based clinical genetic service centers agreed to provide data on infants with confirmed diagnoses of inborn errors of metabolism and endocrine disorders from January 1, 2005 through December 31, 2006. Remuneration and/or personnel assistance for data abstraction were offered. In consultation with the clinical genetic center providers, a formula for reimbursement and a contractual agreement template were developed. One of the four collaborating centers was excluded from the study due to a leadership change. Two of the three remaining centers obtained expedited IRB approval through their respective institutions and the three centers all provided the requested data between September and December 2008. Only one center pursued and executed a subcontract.

In Rhode Island, the proposal was accepted by the RI-NDBS advisory committee in January 2008, and the state genetics coordinator began the IRB approval process. An offer for outside assistance with the IRB was declined. During the next 6 months, it was determined that only hospital staff at the RI-NDBS referral clinical genetic service center would be allowed to abstract data for the study. In August 2008, the state genetics coordinator met with the hospital administrator who agreed to initiate an expedited hospital IRB application. In September 2008, the state genetics coordinator left the RI-NDBS for another position, and the project continued under the direction of the hospital administrator. In October 2008, the hospital administrator and the pediatrician responsible for clinical genetic services initiated a request for an expedited IRB review. In April 2009, the administrator reported that a full IRB application would be necessary, including a letter of support from the RIDOH. The IRB request was finally

Table 2 NNSGRC genetic services for newborns—data abstraction form

Record Number: _____ Calendar Year of Diagnosis: _____

Diagnosed Condition: _____

1. Was the diagnosis identified by NBS confirmed by a subspecialty provider? ☐ Yes ☐ No ☐ Unknown	6. Was a letter/literature on genetic issues provided to the parents or other primary care givers? ☐ Yes ☐ No ☐ Unknown
2. Was a genetic counseling referral made? ☐ Yes ☐ No ☐ Unknown	7. Was a follow up appointment to genetics clinic provided to the parents or other primary care providers? ☐ Yes ☐ No ☐ Unknown
3. Did genetic counseling take place? ☐ Yes ☐ No ☐ Unknown	8. Was a report sent to: ☐ Medical home ☐ Subspecialty provider ☐ NBS Follow up Program ☐ Unknown
4. If yes, was the provider: ☐ MD geneticist ☐ PhD geneticist ☐ Genetic Counselor BC/BE ☐ MD Sub specialist ☐ _____ ☐ Primary Care Provider ☐ Nurse ☐ Other, specify: ☐ _____	9. Were genetic tests provided? ☐ Yes Specify: a ☐ Chromosome analysis b ☐ DNA analysis c ☐ FAO probe d ☐ Other, specify ☐ _____ ☐ No ☐ Unknown
5. Were the following subjects covered in genetic counseling? ☐ Genetic basis of the disorder ☐ Risk estimation for future pregnancies ☐ Carrier testing ☐ Prenatal diagnosis (if appropriate) ☐ Prenatal treatment (if appropriate) ☐ Impact on family members ☐ Unknown	10. If genetic services were provided, how were they paid for? ☐ Private insurance ☐ Medicaid ☐ Self-pay ☐ Title V funds ☐ Public Health Department ☐ NBS fees ☐ Unknown

approved in August 2009, and a subcontract was executed in November 2009. In December 2009, de-identified data were provided on RI newborns diagnosed from January 1,

2005 through December 31, 2006, including inborn errors of metabolism, endocrine disorders, sickle cell and other hemoglobinopathies, and cystic fibrosis.

Assessment of reported data

De-identified data were obtained on a total of 151 patients referred to metabolic centers and 69 patients referred to endocrinology centers in three state NDBS programs. Select patient level data on these individuals are shown in Tables 3 and 4. In the case of infants and their families referred for a probable metabolic disorder, genetic counseling referrals were documented in all cases and genetic counseling could be documented in almost all cases (145/151=96%). At one metabolic center (Minnesota), most genetic counseling was provided exclusively by a genetic counselor. In centers in the other two states, genetic counseling was often provided by a medical geneticist or a metabolic geneticist collaborating with a genetic counselor.

Of the 69 patients with endocrine disorders who received care in the three states' endocrinology centers, only eight (11.5%) were referred for genetic counseling services. While only eight referrals could be documented, clinic records documented that 16 families received genetic

counseling services (23%; eight without a referral). For endocrinology patients, genetic counseling was provided by a genetic counselor with or without collaboration with a medical geneticist or a medical subspecialist in eight of 16 instances; the remainder received genetic counseling from a medical subspecialist or a medical geneticist.

Additional information in Minnesota indicated that metabolic patients who received genetic counseling services were more likely to receive a letter/literature on genetic issues from the genetic service provider, have a follow-up appointment with genetics clinic, and have a letter sent by the genetic service provider to their medical homes than CAH patients who received equivalent counseling services. In Missouri, although endocrine patients were less likely than metabolic patients to receive a letter/literature on genetic issues or have a follow-up appointment, in a majority of cases, a letter was sent to the affected child's medical home and/or state NDBS program. This is also true of Rhode Island genetics clinic which, in fact, reported sending a letter to the medical home of endocrine patients in 100% of cases.

Table 3 Patient-level data on genetic counseling for metabolic and endocrine patients by state

State	Number of patients seen by calendar years (CY)	Metabolic disorder diagnosed	Genetic counseling referral made	Genetic counseling confirmed occurred	Percent receiving genetic counseling	Genetic counseling of patients by provider type
Minnesota	107 patients (CY 2004–2006)	107	107	105	98%	100 by genetic counselors; 3 by medical subspecialist; 1 by medical geneticist and genetic counselor; 1 by pediatric nurse practitioner
Missouri	36 patients (26 from CY 2005–2006; 10 from CY 2005–2007)	33	36	32	88%	14 by genetic counselors; 9 by medical geneticist, 6 of these patients also seen by metabolic specialist; 9 by medical geneticist, 6 of these patients also seen by genetic counselor
Rhode Island	8 patients (CY 2005–2006)	7	8	8	100%	8 by medical geneticist
All 3 States	151	147	151	145	96%	
State	Number of patients seen by calendar years (CY)	Endocrine disorder diagnosed	Genetic counseling referral made	Genetic counseling confirmed occurred	Percent receiving genetic counseling	Genetic counseling of patients by provider type
Minnesota	17 patients (CY 2000–2005)	16, all CAH	5	6	35%	3 by genetic counselor; 1 by MD subspecialist; 2 by medical geneticist and genetic counselor
Missouri	37 patients (CY 2005–2006)	31 CH 6 CAH	1 1	4 5	11% 14%	2 by MD subspecialist; 1 by genetic counselor; 1 by MD subspecialist, genetic counselor and MD geneticist
Rhode Island	15 patients (CY 2005–2006)	12 CH 3 CAH	1 0	1 0	6% 0%	1 by medical geneticist
All 3 States	69	68	8	16	23%	

Table 4 Follow-up communications on metabolic and endocrine patients by state

State	Number of patients seen by Calendar Years (CY)	Metabolic disorder diagnosed	Letter/literature on genetic issues provided to parents or other primary care givers	Follow-up appointment to genetics clinic provided to parents or other primary care givers	Report sent to Medical Home Subspecialty Provider NBS Follow-up Program Unknown
Minnesota	107 patients (CY 2004–2006)	107	71—Yes 30—No 6—Unknown	87—Yes 8—No 12—Unknown	42—Medical Home 62—Subspecialty 0—NBS Program 39—Unknown
Missouri	36 patients (26 from CY 2005–2006; 10 from CY 2005–2007)	33	29—Yes 3—No 3—Unknown 1—No response	30—Yes 4—No 1—Unknown 1—No response	33—Medical Home 1—Subspecialty 19—NBS Program 0—Unknown 2—No response
Rhode Island	8 patients (CY 2005–2006)	7	7—Yes 0—No 1—Unknown	8—Yes 0—No 0—Unknown	8—Medical Home 1—Subspecialty 0—NBS Program 0—Unknown
State	Number of patients seen by Calendar Years (CY)	Endocrine disorder diagnosed	Letter/literature on genetic issues provided to parents or other primary care givers	Follow-up appointment to genetics clinic provided to parents or other primary care givers	Report sent to Medical Home Subspecialty Provider NBS Follow-up Program Unknown
Minnesota	17 patients (CY 2000–2005)	16, all CAH	5—Yes 1—No 0—Unknown 11—No response	4—Yes 1—No 1—Unknown 11—No response	4—Medical Home 5—Subspecialty 0—NBS Program 0—Unknown 12—No response
Missouri	37 patients (CY 2005–2006)	31 CH 6 CAH	1—Yes 2—No 28—Unknown 1—Yes 0—No 5—Unknown	1—Yes 18—No 4—Unknown 8—No response 4—Yes 0—No 0—Unknown 2—No response	23—Medical Home 1—Subspecialty 14—NBS Program 8—Unknown 6—Medical Home 2—Subspecialty 3—NBS Program 0—Unknown
Rhode Island	15 patients (CY 2005–2006)	12 CH 3 CAH	1—Yes 4—No 7—Unknown 0—Yes 1—No 2—Unknown	1—Yes 9—No 2—Unknown 0—Yes 3—No 0—Unknown	12—Medical Home 0—Subspecialty 0—NBS Program 0—Unknown 3—Medical Home 0—Subspecialty 0—NBS Program 0—Unknown

Discussion

State NDBS programs have traditionally focused their follow-up activities and resources in the short term, taking extraordinary measures to prevent any newborns from being lost and documenting all communications concerning

screening test results, referral requests, confirmatory tests, and diagnoses. National guidelines suggest that facilitating referral for genetic counseling should be a follow-up function of state NDBS programs (Tuerck et al. 2006). However, tracking patient-level details on provision of services such as genetic counseling and other clinical

genetic services is generally considered a LTFU activity and in practice remains outside of the role self-defined by most state NDBS programs, as currently configured. Even when there is tracking, the methods are variable.

In our study, participating state NDBS program personnel were aware of the fact that they did not routinely collect patient-level data on genetic counseling services. However, both the NDBS program personnel and clinical genetic service providers perceived value in participating in the study in order to obtain these data for assessing NDBS activities.

As state NDBS programs begin to take a more active role in LTFU, their ability to access timely and reliable patient and provider-related data will be critical (Hoff et al. 2007). The experiences with all three states underscore some of the problems to be anticipated. Obtaining the small amount of patient-level data discussed here was a lengthy process, even in the best of circumstances, perhaps due to the fact that this was a special project that was not a priority for the programs. The cooperation and individual efforts of all personnel participating in completing this study cannot be overemphasized. They were dedicated in their efforts to provide data and have helped to contribute to a better understanding of the processes by which these data may be obtained. Due to lack of clarity in state statutes concerning responsibility for tracking patient-level data, all three of the participating state NDBS programs worked through IRB processes and within clinical genetic service center infrastructures to obtain data. All but one of the clinical genetic service providers, working with their own patients' medical records, also worked through IRB approval processes. IRB processes can be laborious, particularly for first-timers. However, given the public's privacy concerns and recent negative publicity regarding research use of stored residual bloodspot specimens (Grody and Howell 2010), it is probable that state NDBS programs, even when conducting certain types of program evaluation, will have to become more familiar and comfortable with these processes in the future. Alternatively, some state NDBS programs may consider the processes too time consuming and avoid the data collection that is necessary for program assurance, quality improvement, and generation of new medical knowledge.

The limited patient data obtained in this study suggest that metabolic centers are more likely to refer patients for genetic counseling and to utilize genetic counselors in providing these services than are endocrinology centers. Since congenital hypothyroidism is generally not a familial condition, some of this discrepancy is no doubt related to the perceived lack of need for genetic counseling for this condition. Of interest, more patients received genetic counseling for congenital hypothyroidism in Missouri than were actually referred, suggesting that some families may

seek information regarding recurrence risk for this condition. However, more data are needed to determine if these findings apply to other states and other subspecialty centers. If this is indeed the case, then genetic services providers should target their educational efforts to medical practitioners working with NDBS programs in the subspecialty areas outside of metabolic. Also worth noting is that in 52% of metabolic and 46% of endocrine patients, the Missouri genetic service providers also corresponded with the state NDBS program. Notations on the data abstraction forms provided by one of the genetic service centers indicated there is a form used for this reporting. Two of the three Missouri genetic service centers reported corresponding with the state NDBS program.

Over the past several years, LTFU has been the subject of increasing interest (Kemper et al. 2008; Hoff et al. 2007; Hoff et al. 2006; Newborn Screening Saves Lives Act. 2007; Hinman et al. 2009; Botkin et al. 2009; Alexander and van Dyck 2006). The following statements from a recent report documenting communications within newborn screening programs describe some of the issues: "Most states have individual protocols for the acquisition and analysis of dried bloodspot specimens, dissemination of screening results, and the mechanism of data input and information exchange. This does not allow for the easy flow and exchange of critical information between the public health and private sectors within the newborn screening system. This type of linkage is essential, particularly if LTFU is to be achieved. Therefore, current public health practice and research focuses on developing the seamless integration of clinical and public health information systems in newborn screening to increase the likelihood that every newborn receives the appropriate and requisite services" (Hinman et al. 2009).

Given our experience and results, we agree that better health information infrastructure and communications within NDBS systems are needed. Until information integration within the various NDBS system components occurs, whether newborns and their families with disorders identified through NDBS receive requisite clinical genetic services cannot be documented even though population-wide data likely exist. While the NDBS process mapping reported by Hinman et al. did not define exactly where genetic counseling falls on the follow-up spectrum, a public health care coordinator (PHCC) was proposed to provide oversight of LTFU. As described, the PHCC would assure care delivery, assess the completeness of care, and interact with a clinical care coordinator to gather information about the clinical services provided for evaluation. The PHCC's organizational location, support, legal, and other issues remain to be determined, but the concept is promising (Hinman et al. 2009).

Our study had several limitations. Only 35 states participated in the original survey, and there may have been state NDBS programs who track patient-level data on genetic counseling services in the non-participating programs. Of the eight states that indicated systematic documentation of genetic counseling, only three states volunteered to participate. The resulting quantity of data is insufficient for identifying significant trends. Within the other five states, there may have been programs that track patient-level data on genetic services. Additionally, during the course of this project, LTFU has received increased attention at the national, regional and state levels. There may now be states actively involved in collecting patient-level data on genetic counseling services.

One study limitation concerned self-reported data, which was subject to bias and variable interpretation of the terms “tracking genetic counseling.” The three states participating in our study were reported by Hoff et al. (2006) as having no LTFU activities; however, all had data on genetic counseling referrals, even though they could not provide patient-level data on specific genetic counseling services without extraordinary effort. This illustrates a problem with self-reported survey data, which depends on the interpretation of the questions asked by the individual completing the survey. Even data abstracted from patient records are open to bias and errors in reporting.

This project corroborates the conclusion of other researchers that significant data-related challenges lie ahead in improving the ability of state NDBS programs to engage in LTFU (Hoff et al. 2007). It also highlights the need to develop seamless integration of clinical and public health information systems, including newborn screening, in order to improve healthcare delivery and outcome (Hinman et al. 2009). Further, if state NDBS programs are to be responsible for evaluating long-term outcomes, this responsibility needs to be defined, perhaps in guidance to the states from the SACHDNC, and made a priority with defined rules and designated resources. Specific indicators for performance and quality of services provided and received must be defined if successful evaluation and program improvement is to occur. If information sharing in this way presents special privacy issues that result in IRB interactions, then more efficient methods for proceeding through IRBs are needed. In fact, the Newborn Screening Translational Research Network, funded by National Institute of Child Health and Development, has begun to address issues related to consent, IRB, uniform data set, and standardization of language.

Should LTFU data collection and evaluation prove too burdensome for inclusion as a routine NDBS function, then perhaps registries with proper privacy protections embedded and follow-up through regional service delivery networks may be an alternative (Alexander and van Dyck

2006). One proposal for a national registry for conditions identifiable through NDBS acknowledged the need for a data collection system on long-term outcomes, using standardized data collection protocols. The elements of the system described included key clinical information (e.g., physical examination, etc.), laboratory results, a clinical history of illnesses or complications, treatment modalities and their side effects, assessments of compliance, and developmental evaluations. Additional information included psychosocial assessments and financial impacts. The authors were quick to note the possible prohibitive expense of such a system (Botkin et al. 2009).

Conclusion

The data obtained in this study, although limited, suggest that genetic counseling services are more likely to be documented for families of infants of metabolic disorders identified through NDBS than for families of infants with CAH. Apparent lack of a systematic method for documentation of such services at the level of state NDBS programs makes assurance of such services difficult. We strongly urge that documentation of patient-level data on clinical genetic services be included as part of relevant NDBS outcome data. The NNSGRC data abstraction form (Table 2) could serve as a template for creating data fields to capture these data. Sufficient data could be aggregated to document whether newborns and their families with disorders identified through NDBS are receiving appropriate clinical genetic services, and this in turn could be used to identify gaps and make service improvements. Better genetic services can help families receive the information they need to make informed decisions about their children’s health and planning for the future. At the national level, it is essential that there be some consensus on the data that should be maintained as a part of NDBS LTFU systems. Once accomplished, it follows that resources and data systems to meet this requirement will be necessary. Over time, these data could assist in evaluating system-wide service improvements.

The value of tracking this type of information for both patient care and public health functions cannot be over-emphasized. This effort, requiring the cooperation and collaboration of genetic service providers and public health practitioners, will assure that the identified child receives necessary services for optimal health. Further, it will enable quality assurance to improve program effectiveness and efficiency, and inform policy development for the benefit of children and their families.

We hope these preliminary findings contribute to national discussions on the need for better service tracking and follow-up data gathering across NDBS systems. Further, we hope that consensus can be reached at the

national level concerning the follow-up data to be monitored, including clinical genetic services, and the party(ies) responsible for their monitoring and maintenance within the public health system. By describing the US experiences, we hope that in countries developing NDBS programs, particularly those in which genetic counseling is considered to be a useful follow-up service, can benefit from lessons learned regarding the need for NDBS programs to document service delivery from the outset so that deficiencies can be better identified in order to deliver more effective follow-up services (Padilla et al. 2010).

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