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Practices and Perceptions of Long-term Follow-up Among State Newborn Screening Programs

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ABSTRACT

OBJECTIVES. The purposes of the study were to describe and to analyze the types of policies and practices that exist currently among state newborn screening programs in relation to long-term follow-up oversight and activities for newborns with confirmed disorders and to examine the perceptions of newborn screening program leaders regarding long-term follow-up activities.

METHODS. A 23-question survey was administered to state newborn screening program coordinators in each of the 50 states, the District of Columbia, Puerto Rico, and the US Virgin Islands in January to February 2005.

RESULTS. Survey findings revealed significant variation in terms of how long-term follow-up is defined, staffed, and conducted within state newborn screening programs. This variation does not seem to be related to existing state program capacity, in terms of size of newborn screening programs or available resources. In addition, at present many state programs do not conduct long-term follow-up oversight or activities for newborns with diagnosed disorders, and many of those that do seem to lack necessary staffing and quality assurance mechanisms for effective long-term follow-up.

CONCLUSIONS. The results point generally to a need for greater attention in aligning state newborn screening program capacity with long-term follow-up, increased emphasis on standardization for long-term follow-up activities, and development of stronger quality assurance oversight from state newborn screening programs if effective long-term follow-up oversight is to occur nationally. Given the present expansion of newborn screening in many states, additional research and policy-making with respect to long-term follow-up seem warranted.

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Key Words

follow-up, newborn screening, public health, health policy, newborn screening programs

Abbreviations

LTFU—long-term follow-up
MS/MS—tandem mass spectrometry
NNSGRC—National Newborn Screening and Genetics Resource Center
FTE—full-time employee

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NEWBORN SCREENING IS an important activity in health care. It is also ideally a system in which testing, diagnosis, treatment, and ongoing evaluation occur in a timely, coordinated manner. In 2001, more than \$120 million was spent nationally on newborn screening.¹ Newborn screening facilitates the early detection and prevention of disease among infants and children, reduces morbidity associated with disease, and improves the quality of life and longevity for individuals afflicted with or susceptible to certain disorders.²⁻⁴ A recent report on newborn screening sponsored by the federal government recommended expansion of state testing to a minimum of 29 specific disorders.⁵ Currently, newborn screening programs exist in all 50 states, the District of Columbia, and some US territories. States screen for 8 to 50 genetic and metabolic disorders, although, with the new recommended minimal panel, many states are in the process of expanding screening to ≥ 30 disorders.

Traditionally, the value of newborn screening as a public health activity lay in having valid and reliable tests to identify the presence of treatable disorders.⁶ However, this traditional view has given way recently to a more-expansive view of newborn screening. The latter view has accompanied technological breakthroughs such as tandem mass spectrometry (MS/MS), which make screening for more disorders cheaper and easier. The use of MS/MS also aids in screening for a larger array of preventable, treatable disorders. The expansive view justifies testing not only to prevent morbidity and death with early intervention but also to identify carriers for specific disorders, to reduce parental stress, and ultimately to detect genetic mutations that could lead to adult-onset disease within an individual.^{7,8}

The expansion of state newborn screening programs is an emerging reality, given the technology available, increasing public and political pressure for more testing, and the increasing availability of for-profit, private testing laboratories. However, there is debate in the field regarding whether proposed expansions will offer added value from a public health perspective.⁹⁻¹¹ Some disorders that can be detected with new MS/MS technology are not treatable, prompting debate regarding the ultimate benefits of testing for the newborn. Other disorders are treatable but not curable or preventable. In addition, screening for diseases such as cystic fibrosis may create complications and anxiety for patients and their families in the absence of actual disease; for example, the screening process may lead only to carrier identification. These realities make testing potentially more expensive, less reliable, and of less benefit.^{7,8}

For newborns with abnormal screening results, additional concerns arise, related to the ability of each state's health care delivery system to provide confirmatory diagnoses for newborns in a timely manner, to provide similar access to treatment services across screened pop-

ulations, and to perform adequate follow-up monitoring and case management over time.¹¹⁻¹³ The issue of follow-up services for newborns with abnormal screening results is particularly important because newborn screening is screening and not diagnosis.⁷ Diagnosis should begin a longer-term process of monitoring, intervention, and treatment. From the state newborn screening program perspective, follow-up services may encompass a variety of oversight, care coordination, and direct service activities, including notification of providers of test results, notification of parents, confirmation of diagnosis and treatment for specific disorders, coordination of services for affected newborns, monitoring of service provision, and evaluation of clinical care for individuals.^{11,14}

Understanding more about the role of state newborn screening programs in follow-up activities requires an appreciation of the general context of newborn screening in the United States. Newborn screening is a state-controlled health care activity.^{7,15} States have complete discretion to determine how their newborn screening programs should be structured. This discretion produces wide variations in newborn screening programs across states.^{1,13,14,16,17} These variations are a function of a mixture of factors, such as state size, available financial resources, political and interest group pressures, type and authority of advisory groups for newborn screening, and availability of technology.^{10,11}

Less is known about the variations across state programs in long-term follow-up (LTFU) oversight and activities for newborns with diagnosed disorders. However, LTFU is the second major component of newborn screening that makes it a meaningful public health activity.^{7,11} Identifying infants who may have genetic or metabolic disorders is the first major component. However, if individuals with confirmed diagnoses cannot receive timely, accessible, appropriate care for their conditions, then identifying them is of less value. Gaining more insight into how state newborn screening programs, as one component of a state newborn screening system, perceive and practice LTFU care for newborns with confirmed disorders helps inform the ongoing dialogue regarding the public health benefits gained from the expansion of newborn screening in the United States. To our knowledge, this study is the first systematic effort to obtain more detailed information on a variety of LTFU practices and perceptions among state newborn screening programs. The following questions guide analysis. (1) What types of policies and practices exist currently among state newborn screening programs with respect to LTFU oversight and activities for newborns with abnormal screening results? (2) How do state newborn screening programs, whether or not they conduct LTFU activity, perceive it in terms of specific activities, required resources, and potential for standardization?

METHODS

Survey and Sample

A 23-question survey was prepared and sent to newborn screening programs in all 50 states, the District of Columbia, and the US Virgin Islands. The same survey was administered in both Internet-based and mail formats. The Internet-based version of the survey was sent to the e-mail addresses of both the laboratory and follow-up screening program coordinators listed in the most recent contact list maintained by the National Newborn Screening and Genetics Resource Center (NNSGRC).¹⁷ The printed version of the survey was sent to the mailing addresses of the same program contacts. Respondents were asked to submit only 1 version of the survey, whether they selected the Internet-based or printed survey. Only 2 respondents chose to reply by mail; the rest completed the Internet-based survey. The survey was administered during January and February 2005.

The survey focused on the practices and perceptions of LTFU services in state newborn screening programs. (The complete survey is available from the authors.) Coordinators were also asked to classify what they considered to be part of appropriate, state-level, LTFU oversight and activities. There is wide variation in the literature regarding what constitutes LTFU, which was another reason to explore how each program sought to define it. Questions in the survey asked respondents about items such as program-specific definitions of follow-up, whether or not states engaged in LTFU oversight and activities generally, whether or not states used specific quality assurance tools to conduct LTFU, the presence of formal evaluation activities in relation to LTFU, and current and desired staffing levels for follow-up. It also asked basic descriptive questions related to program budget size, number of disorders screened, positional title of respondent, and number of years working in the current position.

Some of the survey questions were open-ended. The majority had forced choices, in terms of yes/no or Likert-scale responses (eg, strongly agree to strongly disagree). There were also a few questions in which respondents were asked to provide rankings. The survey items were developed through an iterative process that involved discussions with the director of the NNSGRC and review of the literature on newborn screening. The survey was pilot-tested with a group of 12 individuals at the authors' school of public health and several genetics professionals. To complete the data set, 1 additional variable was taken from a General Accounting Office 2003 survey of state newborn screening programs, 1 variable was taken from the NNSGRC June 2005 US national screening status report, and the current fees charged by programs were obtained from the NNSGRC. The variable taken from the General Accounting Office survey was the number of live births for each state.¹ The size of a state,

in terms of number of newborns who need to be screened, may affect follow-up practices, because it raises potential capacity issues related to the number of newborns with abnormal screening results. The number of mandated disorders for each state was obtained from the NNSGRC status report. Finally, program screening fees have been cited as a significant source of revenue used to run newborn screening programs.¹

Statistical Analyses

SPSS version 13.0 (SPSS, Chicago, IL) was used for analyses. Analyses were conducted by using a combination of univariate and bivariate statistical operations. These included descriptive values such as frequencies, means, and SDs; analysis of variance with independent and paired-sample *t* tests; and cross-tabulations with the χ^2 measure of association. Several key variables, such as whether or not the state conducted any LTFU oversight or activities, were used to stratify the data into subgroups a priori, for comparison analysis. Only 2 variables in the data set had missing data that amounted to >5% of total cases; these were the estimated budget for a state newborn screening program and the number of full-time employees (FTEs) necessary to conduct adequate LTFU service. Both variables were included in the analyses because they were deemed important for understanding overall practices and perceptions of LTFU. It was decided that the benefit of having data for these 2 variables for most of the states outweighed the disadvantage of not having data for a few states. Rather than the means for these variables being used in missing cases, the cases themselves were excluded from analysis.

RESULTS

Variation and Capacity Related to LTFU Services

Surveys were completed for 45 of 50 states, the District of Columbia, Puerto Rico, and the US Virgin Islands. This represents a 91% response rate. Nonresponding states included Massachusetts, Kansas, Tennessee, New Mexico, and South Dakota. Programs for these states were contacted by telephone, to determine whether the coordinators would complete and return the survey. Several could not be reached, and the others stated that they did not wish to complete the survey. We used only 1 completed survey for each state and territory and, in all cases except 5, the completed surveys came from the follow-up coordinators within each program. In the other 5 instances, we used laboratory coordinator survey responses, because it was communicated by the particular state programs that those individuals were in the best position to respond to questions about LTFU service.

Although the average number of disorders for which testing is required or has been mandated was ~28, 36% of all states and territories responding had required or mandated testing for >40 disorders (Table 1). There was

TABLE 1 Characteristics of State Newborn Screening Program Sample

Characteristic	Mean	Median	Range
Time employed (<i>n</i> = 48), y	10.07	9.25	0.50–28
No. of tested disorders required or mandated (2005) (<i>n</i> = 46)	28.66	30.00	8–52
Programs that perform LTFU service (<i>n</i> = 48), %	50	50	NA
Total no. of live births (2002) (<i>n</i> = 43)	81 727.02	49 578.00	5758–530 710
Fees charged (<i>n</i> = 45)	\$38.49	\$40.67	\$0–139
Program budget (<i>n</i> = 35)	\$3 910 522	\$2 118 562	\$39 000–\$37 000 000

Typical job titles for follow-up oversight were director, program manager, program administrator, genetics coordinator, research scientist, and nurse consultant. NA indicates not applicable.

also a great deal of regional variation seen in the number of tested/mandated disorders. (“Tested” disorders were defined as disorders already included in a state’s newborn screening panel and “mandated” as disorders for which testing in a state has been required recently but has not yet been implemented.) Mountain and Southern states tended to have fewer tested/mandated disorders. However, only 5 states had required testing for <10 disorders. There was no association between the size of a state (measured in terms of live births) and the number of disorders tested. On average, programs charged a \$38.49 fee for each newborn screened. The median fee charged was \$40.67. However, 5 states and territories charged no fee, and 24 charged \$40 or more. For programs that performed LTFU activities and oversight, the average fee charged per newborn screened was \$36.70. For programs that did not conduct LTFU, the average fee was \$41.81. This difference was not statistically significant. Only 1 program that engaged in LTFU did not charge a fee, compared with 4 states that did not conduct LTFU. Approximately 43% of states engaged in LTFU charged \$40 or more in screening fees, compared with 58% of states that did not engage in LTFU. There was no association observed between the fees charged and the size of the state newborn screening program, as measured in terms of numbers of live births or size of total program budget. However, there was a significant association between the fees charged and the numbers of tested/mandated conditions within states ($r = 0.372$; $P < .01$).

Variation among state newborn programs was also seen in terms of leader tenure and position title in relation to the follow-up component. The average number of years follow-up coordinators had been employed in their current screening programs was 10. However, one third of all programs had follow-up coordinators who had been employed for <4 years, and 25% of coordinators had been employed for ≥ 15 years. Job titles for individuals in charge of follow-up activities (both short-term follow-up and LTFU activities) also varied meaningfully across state newborn programs (Table 1). At least 15 different titles were reported by respondents. Total budgets and numbers of FTEs for state newborn screening programs also varied greatly. The average annual state newborn screening program budget reported

by respondents was \$2.9 million, with a range between \$39 000 and \$37 000 000. However, the distribution of state program budgets was skewed toward lower budget amounts, with 35% of respondents reporting total program budgets of \$1 million or less and 50% reporting budgets of \$2.1 million or less. There was a strong positive correlation between state size, measured in number of live births, and program budget size ($r = 0.841$; $P < .001$). No relationship was found between the number of tested/mandated disorders in a state and budget size.

Of programs that conducted LTFU oversight and activities (*n* = 24), two thirds stated that they employed ≤ 5 FTEs for all follow-up activities, including both short-term follow-up and LTFU activities. However, when asked to identify the approximate percentage of total FTE follow-up time spent exclusively on LTFU activities, more than one half of these 24 programs stated that <20% of total FTE follow-up time was devoted to activities they considered LTFU. Given a mean of 5 FTEs reported for follow-up staffing in total and the fact that only a few respondents indicated employing ≥ 7 individuals for follow-up activities in total, we can infer that most state programs that perform LTFU allocate ~ 1 or 2 FTEs to this oversight and activity. When all 48 state and territory program respondents were asked how many FTEs they thought were needed in their programs to engage in adequate LTFU for newborns with abnormal screening results in their state, more than one half responded with >2 FTEs and 20% suggested ≥ 9 . Given existing staffing levels reported, a meaningful number of respondents thought that what was desirable in terms of staffing for effective LTFU amounted to 2 to 5 times more staff members than are employed presently in their programs. No differences in the desired number of FTEs needed to perform adequate LTFU were seen between state programs that did and did not conduct LTFU.

The definitions of what should constitute LTFU oversight and activity within state newborn screening programs were fairly uniform among survey respondents. Two thirds of responding programs thought that LTFU at the state level consisted of ensuring that patients have access to a medical home during their lifespans and ensuring that support services, such as transportation and information, are available to patients (the definition of “medical home” was provided to respondents in the

survey as “a regular and consistent source of care for a patient that is accessible, family-centered, comprehensive, and culturally competent”) (Table 2). The greatest agreement among respondents involved the components of ensuring that ongoing management/treatment is being provided to patients and ensuring that needed changes in management/treatment plans are identified and made in a timely manner. One half of all survey respondents identified these 4 components as appropriate parts of LTFU oversight and activity within state newborn screening programs.

Comparison of LTFU Activities, Models, and Quality Assurance

Table 2 also compares definitions of LTFU provided by programs that do and do not conduct such activities as a regular course of business. Several differences were found between the 2 groups with respect to how they defined the components constituting LTFU particularly. Respondents from programs that engaged in LTFU seemed to have a more expansive definition of the overall activity. For example, significantly greater proportions of LTFU programs thought that ensuring initial patient access to a medical home and ensuring that ongoing management/treatment is provided to patients were parts of LTFU ($P = .022$ and $.043$, respectively). In addition, one third of programs engaged in LTFU considered the component of ensuring that an initial management/treatment plan is designed and communicated to patients a legitimate part of LTFU (as opposed to being thought of exclusively as a short-term follow-up activity), compared with 13% of programs not engaged in LTFU. Comparisons were also made for a number of size and capacity characteristics among newborn screening programs that do and do not conduct LTFU for newborns with diagnosed disorders (Table 3). No significant differences were found in any of these characteristics.

An additional subgroup analysis was performed between state newborn screening programs that engaged

in LTFU oversight and activity for all tested/mandated disorders and those that engaged in LTFU activity only for select disorders (Table 4). These 2 groups were compared with respect to a number of size and resource characteristics. They were also compared in terms of the presence or absence of specific quality assurance practices. For example, respondents were asked whether they performed LTFU in-house or contracted out for it, whether they had a standardized LTFU policy or set of procedures in place to guide their oversight and activities, whether they used any sort of computerized tracking system to conduct follow-up monitoring over time, whether they had a formal evaluation plan to assess their program’s LTFU oversight and activities, and whether they tracked the LTFU costs for their program.

Few differences were found between the 2 LTFU groups (Table 4). The only significant difference was the greater proportion that tracked the costs of LTFU in their program in the group that engaged in LTFU activity only for select disorders, compared with the group that engaged in LTFU activity for all tested/mandated disorders (55% vs 8%; $P = .020$). A greater proportion of programs that performed LTFU for all disorders in their state had standardized LTFU procedures in place to guide activities (75% vs 36%; $P = .066$). Perhaps more interesting than the few differences found between these 2 LTFU groups were the lower percentages found in both groups with respect to having in place or conducting quality assurance and efficiency-focused activities. For example, fewer than one half of the 24 state programs that engaged in LTFU performed any of that service in-house, instead contracting it out to an external entity, thus creating a situation in which they might have less oversight ability.

In addition, only 5 of 24 programs reported having a formal evaluation plan in place to assess the effectiveness of their program’s LTFU activities. Two thirds of the 24 programs performing LTFU indicated that they did

TABLE 2 Components Identified by Program Coordinators as Appropriate Parts of State-Level LTFU Oversight and Activity

LTFU Component	Proportion of Programs, %			P for Difference
	Entire Sample (n = 48)	Conduct LTFU Service (n = 24)	Do Not Conduct LTFU Service (n = 24)	
Ensuring that patients have access to a medical home over their lifespans	69	67	71	.76
Ensuring that patients have initial access to a medical home	17	29	4	.02
Ensuring that newborns with abnormal screening results receive a diagnosis	15	17	13	.69
Ensuring that an initial management/treatment plan is designed and communicated to patients	23	33	13	.09
Ensuring that ongoing management/treatment or prevention is being provided to patients	92	100	83	.04
Ensuring that needed changes in management/treatment plans are identified and made in a timely manner	81	88	75	.28
Ensuring that support services, such as transportation and information, are available to patients	65	71	58	.38

TABLE 3 Comparison of Size and Capacity Characteristics Between Programs That Do and Do Not Engage in LTFU Oversight and Activities

Variable	Mean		P for Difference
	Conduct LTFU	Do Not Conduct LTFU	
Total no. of live births, 2002 ^a	97 102.00	65 620.00	.31
Fees charged ^b	\$36.70	\$41.81	.52
Approximate annual budget for program	\$4 864 113.36	\$2 639 066.67	.31
No. of tested disorders required or mandated (2005) ^c	18.65	20.96	.58

^a Data from 2003 General Accounting Office report.

^b Data from the National Newborn Screening and Genetics Resource Center, October 2005.

^c Data from the National Newborn Screening and Genetics Resource Center, June 2005.

TABLE 4 Comparison Between Programs That Perform LTFU for Select Versus All Screened Disorders

Variable	Mean		P for Difference
	LTFU Conducted for Select Disorders (N = 12)	LTFU Conducted for All Screened Disorders (N = 12)	
Total no. of live births, 2002 ^a	94 414.42	100 327.00	.92
Fees charged ^b	\$35.18	\$38.36	.72
Approximate annual budget for program	\$5 730 104.32	\$3 998 122.40	.65
No. of tested disorders required or mandated (2005)	17.67	19.73	.74
LTFU performed in-house solely or partially, %	27	73	.04
Presence of standardized LTFU procedure, %	36	75	.06
Computerized tracking system used to conduct LTFU, %	55	67	.57
Presence of formal evaluation plan for LTFU, %	18	25	.71
Program tracks the costs of performing LTFU, %	55	8	.02
No. of FTEs involved in both short-term follow-up and LTFU activities	4.52	6.86	.30
Proportion of employee time spent on LTFU, %	27.09	24.71	.83

^a Data from 2003 General Accounting Office report.

^b Data from the National Newborn Screening and Genetics Resource Center, October 2005.

not track the costs of performing LTFU oversight in their state. Greater proportions indicated that they had both standardized procedures and computerized tracking systems to enhance the quality of LTFU oversight and activities in their programs. However, 35% to 40% of programs performing LTFU service had neither of those items (Table 4). Not unexpectedly, states that conducted LTFU activities for select disorders had fewer employee resources devoted to LTFU service generally within their state screening programs. For both groups, the amount of allotted FTE time, as indicated earlier, was small.

DISCUSSION

The results of this study raise issues in need of attention, with respect to state newborn screening program capacity to engage in LTFU service, standardization of that LTFU service, and improved quality management within state programs that engage in LTFU. First, there is the capacity issue. Most newborn screening programs currently are small, in terms of budgeted resources. There was no observed relationship between the fees charged and whether a state engaged in LTFU. In fact, the screening fee was higher, on average, for states that did not engage in LTFU activities. The lack of association observed here between capacity variables and whether a

state engages in any type of LTFU indicates that policy decisions creating increased workload for state newborn screening programs, such as adding new disorders for testing or performing extended follow-up for newborns with confirmed disorders, may not align with the existing capacity of a state newborn screening program to absorb that workload. This is an especially important point if there is an expectation, given newborn screening as a system, that state newborn screening programs have a role to play in overseeing and coordinating the management and treatment of newborns with diagnosed disorders.^{10,11,18}

The capacity issue for LTFU in state newborn screening programs is informed by other study results. One half of the states and territories that responded indicated that they currently do not engage in any LTFU oversight or activities. The findings also show that capacity is an issue even for state programs that perform some form of LTFU, with these programs allocating small amounts of resources to their LTFU oversight and activities, in the range of 1 to 2 FTEs. For state programs that screen for up to 50 disorders, these staff resources seem low, regardless of which specific tasks constitute the LTFU functions in those programs.

If resources (human and financial) to conduct ade-

quate LTFU activities and oversight in a state newborn screening program are not available, then the activities may fall in part to academic or other specialty centers. As seen in this study, LTFU services could be included as part of contracted services provided by these centers, in some cases. Subspecialty centers may have limited staffs with a variety of clinical priorities, yielding minimal time that can be devoted to LTFU. Subspecialty centers also may tend to care for patients with disorders in specific disease categories, such as metabolic or endocrine disorders, rather than caring for patients across multiple disease categories. There may be a variety of subspecialty centers in a given state, geographically distant from each other, that lack the resources for ongoing coordination and communication among themselves.

A second issue raised by the expansion of newborn screening programs is related to the potential for standardization of LTFU program elements within state newborn screening programs. Standardization allows best practices to be identified, shared, and reproduced and helps to establish a national newborn screening system. The recent government-sponsored report on standardizing newborn screening panels across states is in the spirit of this idea.⁵ In the present study, several findings point to either a lack of standardization or potential difficulty in achieving greater standardization regarding the LTFU activities of state newborn screening programs. First, some variation was observed in how LTFU is perceived by state newborn screening programs that do and do not conduct LTFU activities (Table 2). Second, the findings that indicate few linkages between the state's decision to engage in LTFU and the state newborn screening program's capacity to perform LTFU activities mean that some states may be in a less-advantageous position than others in being able to incorporate elements of a standardized approach to LTFU, which might include resource-intensive systems in areas such as patient tracking and formal evaluation. Third, almost one half of all state programs that engaged in LTFU oversight and activities reported that they did not have any type of standardized policies or procedures in place at the present time. This may indicate screening program work cultures that are resistant to standardization or that require some degree of ongoing socialization to embrace standardization in their work.

Ensuring quality assurance for LTFU in the face of newborn screening expansion is a third issue worthy of greater exploration, on the basis of these findings. The quality issue is evident in the percentages of state newborn screening programs that do not have key elements in place as part of an overall quality management approach to LTFU (Table 4). Whether or not they conducted LTFU activities for all or only select tested/mandated disorders, many programs did not have a standardized LTFU protocol in place to guide their activities (as stated above), did not use any sort of computerized tracking system to monitor

care provided to newborns over time, did not have formal evaluation plans to gauge the success of their program's LTFU oversight and activities, and did not track what they spent on their LTFU activities and compare the costs and benefits of those activities. Interestingly, state size and available funding did not play meaningful roles in increasing the chances that a program would have more quality assurance elements in place. This raises the question of whether it is necessary to consider the federal government mandating (and perhaps helping to fund) minimal quality assurance standards, so that all state screening programs engaging in LTFU have similar abilities and incentives to create and to sustain LTFU quality components. Of course, other programs within a state that perform LTFU activities may possess these quality assurance elements to lesser or greater degrees. However, the question is to what extent state newborn screening programs need these elements when performing their own LTFU activities.

This study suffers from several key limitations. First, the findings are derived from self-reports from newborn screening program leaders. This raises a concern that answers to factual questions such as those on program size and budget might be reported erroneously. However, information from newborn screening programs generally is gathered through self-report at the present time, for example, information sent by programs to the NNSGRC for their annual reports. It was thought that those surveyed were in the best position to comment on both the scope and substance of state program LTFU activities and that the views of these individuals are important in determining how LTFU should be structured within their programs (because they are closest to the everyday program realities). This study did not focus on what state programs actually did in terms of specific LTFU oversight and activities, only whether programs performed LTFU oversight and activities as they defined them. An additional limitation of the study was that it asked only basic questions related to LTFU practices. As a result, additional investigation is needed to clarify the relationships found in the present study, perhaps through qualitative research with a subsample of different state newborn screening programs. Finally, this study focused only on state newborn screening programs. There may be other entities within a state that conduct LTFU activities for newborns with diagnosed disorders.

CONCLUSIONS

The present study represents the early phase of what needs to be extensive systematic exploration of the LTFU component within and across state newborn screening programs. This is imperative if we are to evaluate fully the success of expanded newborn screening throughout the nation. Currently, LTFU remains an overlooked component of newborn screening generally, and it re-

mains unclear what role state newborn screening programs should play in it. However, to comply fully with the spirit of recently released guidelines for LTFU,¹⁸ it will be important for state newborn screening programs to be cognizant of the issues related to LTFU and to take a leadership role in finding solutions that lead to more-comprehensive LTFU services. This means that ultimately every state NBS program should be involved in LTFU at some level.

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