Genetic Counseling and Risk Communication Services of Newborn Screening Programs

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Objectives: Newborn screening test results labeled “positive” can have uncertain implications for parents, especially when false-positive results occur or when heterozygous infants are detected using molecular tests for sickle cell hemoglobinopathy or cystic fibrosis. This study surveyed communication services across state newborn screening programs.

Methods: We surveyed newborn screening programs to identify current communication practices and the methods used for quality assessment. Two successive survey instruments with fixed-answer and free-answer questions were distributed to screening program follow-up coordinators or similar designated officials associated with 52 states and territories.

Results: Replies from 46 respondents (89% response rate) revealed that regional newborn screening programs vary widely in their approaches to counseling. Of the 46 respondents, 35 (76%) answered that they “routinely” provide counseling services to families of affected infants. Depending on the disease, an average of approximately one-half that number provide counseling after false-positive results or for heterozygous infants. Most respondents advocate nondirective counseling more than direct advice. Most programs reported that counseling was usually done by subspecialist physicians or specially trained nurses and counselors. Respondents reported a perception that the “quality” of counseling by these professionals is better than counseling by primary care physicians. Few programs reported systems for assessing quality assurance of counseling.

Conclusions: Newborn screening programs in the United States vary widely with regard to counseling practices, and no best practices are currently evident. Few programs provide counseling quality assurance. Further study and advocacy is needed to optimize communication services, preferably before implementation of molecular tests arising as a result of the Human Genome Project.


NEWBORN screening has been described as a population-based public health program applying preventive medicine in defined regions to reduce newborn morbidity and mortality from certain biochemical and genetic disorders by using presymptomatic detection/diagnosis with dried blood specimens analyzed in central laboratories employing automated procedures and linked to clinical follow-up systems.¹

This and other descriptions emphasize the comprehensiveness of newborn screening programs, but they are less specific about the nature of “clinical follow-up systems” and the communication services required for interacting with families. Undoubtedly, newborn screening tests provide valuable medical information to the families of millions of infants each year. Ever since phenylketonuria (PKU) screening was implemented during the 1960s, newborn screening programs have become an important public health service and a key element in preventive pediatrics.¹ Despite their great success, there is clearly a need for improvement and better consistency across regions.² Previous observations¹-³ suggest that more critical appraisal and efforts toward continuous quality improvement would help enhance these already competent programs. Attention is needed with regard to both laboratory test procedures² and follow-up activities for infants with false-positive and true-positive (disease-affected) test results. Depending on the condition, the false-positive population can include infants with a heterozygous trait or infants identified by an error in laboratory testing.

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METHODS

SURVEY INSTRUMENT DEVELOPMENT

We designed an initial, focused instrument addressing concerns from the genetic counseling and risk communication literature. The instrument was pretested during informal discussions with some newborn screening officials and other leaders. The final instrument was composed of 2 packets with different questions, depending on whether or not the responding program routinely performed posttest counseling. Thus, the respondents selected 1 of the 2 packets based on their response to a statement on the cover sheet: “Please fill out this packet if your state routinely provides counseling services to families of newborns with any positive screen.” The packets were each composed of 39 detailed multiple-choice questions and 17 free-response questions, with ample space for comments after most items. We included inquiries about operational issues, including counselor background, methods, venue, and which diseases are felt to merit counseling. We also asked each respondent about the use of tools for quality assessment or counselor evaluation. Reasoning that the opinions of our respondents could suggest future policies, we asked about the perceived goals of counseling, and attitudes about counseling from different types of professionals. To simplify the instrument, most questions were general, and we encouraged the respondents to comment separately about variation for different diseases or populations. All materials were reviewed and approved by institutional review boards at the University of Wisconsin, Madison, and the University of Michigan, Ann Arbor.

PARTICIPANTS

Since we were interested in posttest counseling, we attempted to direct the survey to each program’s designated follow-up coordinator, who might be expected to know the most about counseling procedures. In most cases, we identified these officials from a list provided by Brad Therell, PhD, of the Council on Regional Networks’ Newborn Screening Committee. For programs without an official follow-up coordinator, we encouraged the program directors to either select a designee or fill it out themselves. There was some reported overlap between program responsibilities, so a total of 52 surveys were sent to newborn screening programs associated with each of the 50 United States, the District of Columbia, Puerto Rico, and the US Virgin Islands. The respondents were assured of confidentiality. Surveys were mailed in mid-1998, and returned by early 1999. To attain our goal of a response rate, all survey recipients received at least 1 recruiting phone call. These calls provided valuable information through lengthy and sometimes passionate discussions. Indeed, the telephone interviews helped identify some concerns that had not been addressed in full detail within the format of the instrument.

ANALYSIS

Identifying information was removed from all returned materials in order to ensure the anonymity of the responding state and official. Responses were tabulated and organized in a database (Microsoft Excel; Microsoft Corp, Redmond, Wash), and investigators were blinded to the responding state. In most cases, the sample of responses represents a substantial fraction of the overall population of newborn screening programs. Thus, most responses are simply reported as a fraction of the eligible subset denominator. For some questions, the $\chi^2$ test was used to compare the response with the 50:50 ratio that might be expected by chance. Free response items and informal data are reported qualitatively where appropriate.

FOLLOW-UP SURVEY

Despite efforts to clarify the instrument, we were often uncertain as to whether the respondents’ comments about counseling related only to children diagnosed with the disease, or if programs also counseled families of infants with false-positive results or heterozygous trait status. In addition, some respondents’ comments seemed to blur the distinction between infants with false-positive results and heterozygous infants, perhaps simply to classify them as “not diseased.” To clarify whether programs give any priority to these infants, we decided to contact the respondent again with another questionnaire. This second survey consisted of 6 multiple-choice questions and 2 free-response questions with room for comment after each.

This false-positive group is about 50-fold larger than those groups actually diagnosed with congenital disorders and has received very little attention. The psychosocial and financial implications of newborn screening results are unclear, and families with an affected infant may face discrimination, reduced insurability, or other disadvantages. These adverse consequences may be especially disconcerting for families of infants with both false-positive results, or in whom a heterozygous or “trait” status is found, such as cystic fibrosis or sickle cell hemoglobinopathy. There are also concerns that parents may have impaired bonding and weakened long-term relationships with their essentially healthy children, or that they may become preoccupied with a pessimistic future for their child. Unfortunately, parental decision-making after a positive newborn screening test is poorly understood. Posttest counseling can potentially reduce confusion among families of affected infants, carriers, or healthy newborns with false-positive results and perhaps prevent psychosocial complications. As the Human Genome Project and other scientific advances create more opportunities for testing, the need for effective counseling also increases. This is true for tests that screen for congenital disorders, but it is just as important for many future tests that screen for disease risk in older individuals. The implementation of these potentially valuable tests may be limited by societal anxiety about the potential harms from the test results.

In developing the counseling services for the Wisconsin Cystic Fibrosis Neonatal Screening Project, we were aware of anecdotal reports of counseling in some

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newborn screening programs, but little was known about the prevalence of these services nationwide or about their effectiveness. In addition, the uncertainty about counseling skills and the complexity of the subject matter being communicated raise questions about what might be called the “quality” of counseling. We designed this study, therefore, to determine the status of risk counseling services in newborn screening programs in the United States, and to determine which services were known to be the most effective or “best” practices. We first held discussions with members of the group then named the Council on Regional Networks Newborn Screening Committee, and realized that little information was available and that no best practices were apparent. Consequently, we decided to develop an instrument that extended beyond simple practices into the goals and perceptions of program officials. The objective of this wider approach was to provide a foundation for further improvement of widespread counseling about complex tests.

### RESULTS

#### RESPONSES

After at least 1 telephone recruiting call per potential respondent, we achieved an 89% response rate (N = 46). Several respondents revealed strong feelings about the survey, both positive and negative, and many attached extensive comments and materials from their counseling programs. The group designated as follow-up coordinators comprised a wide variety of professionals, mostly including nurses and public health professionals, as well as a few social workers and physicians. Passive information available on the 6 nonresponding programs did not demonstrate systematic differences.

We were surprised by the volume of written comments and by the candor and interest expressed during the telephone interviews. During these calls, most potential respondents were very enthusiastic about the need for the project, but others objected strongly. Many said that the answers to our questions were “obvious,” although their answers often disagreed with the next person’s obvious answers. Some were concerned that “negative results” from the survey might be used by political opponents to argue against government-funded newborn screening programs or against counseling. Others had concerns that the results might be used in future litigation, perhaps identifying that a program is substandard or inferior in a certain way. Nevertheless, the most common comments came from coordinators who asked us about what the findings of our study had been thus far. Although some officials had concerns about the judicious publication of our results, many were interested in broadening their communication services but wanted guidance because they were uncertain as to what steps to take next.

### COUNSELING SERVICES

Respondents indicated their overall counseling practices by choosing one or another of the labeled survey packets. Of the 46 respondents, 35 (76%) chose packet 1, thus reporting that they routinely perform some sort of genetic or other risk counseling to families of infants with positive newborn screen results. The remaining 11 respondents (24%) indicated that they do not usually provide counseling to families of infants with positive newborn screen results. Of those program coordinators who said they provide such counseling, 22 (63%) responded that they “always” do so. Separated by different diseases, the variation in counseling services was greater than we anticipated. As presented in Table 1, most counseling programs communicate about the 4 disorders most commonly included in newborn screening panels, namely phenylketonuria, congenital hypothyroidism, galactosemia, and hemoglobinopathies. Communication services are less commonly provided for the other conditions. In addition, most programs counsel only for infants with a diagnosed disease, and do not provide education for families of infants with false-positive or “trait” results. Results of the follow-up survey (response rate, 22/52 or 42%) suggested that little of this counseling is offered. We used a ratio to interpret the differences in counseling practices for the various diseases. These ratio values, listed in Table 1, consisted of the percentages of programs that counsel families of infants false-positive results, divided by the percentage of counseling services offered for the disease itself (ie, for families of infants with true-positive results). The ratios range from 38% (for congenital adrenal hyperplasia) to 68% (for hemoglobinopathies).

### COUNSELING PROVIDERS

Since counseling skills, interpersonal interactions, and framing effects can influence understanding, we asked for percentage estimates of the different types of counseling professionals. Responses varied widely, and were occasionally left blank. Some respondents said they were uncertain as to provider demographics. Of the 35 programs that provide routine counseling, coordinators at 17 (49%) estimated that most of their counselors were female, 11 (31%) of 35 had no clear idea about the sex of their counselors, and 3 (9%) of 35 reported male predominance.

The race or ethnicity of the counselors was reported by 28 respondents, and 22 (63%) estimated that their counselors were predominantly white, while 5 (14%) of 28 of program coordinators said that at least half of their counselors were African American. Only 3 of 28 re-
Table 2. US Newborn Screening Programs Counseling for True-Positive and False-Positive Tests*

<table>
<thead>
<tr>
<th>Disease</th>
<th>Programs Counseling True Positives</th>
<th>Programs Counseling False Positives</th>
<th>False-True Counseling Ratio†</th>
</tr>
</thead>
<tbody>
<tr>
<td>Phenylketonuria</td>
<td>21/22 (95)</td>
<td>13/22 (59)</td>
<td>62</td>
</tr>
<tr>
<td>Congenital hypothyroidism</td>
<td>17/22 (77)</td>
<td>11/22 (50)</td>
<td>65</td>
</tr>
<tr>
<td>Galactosemia</td>
<td>18/22 (82)</td>
<td>11/22 (50)</td>
<td>61</td>
</tr>
<tr>
<td>Sickle and other</td>
<td>19/22 (86)</td>
<td>13/22 (59)</td>
<td>68</td>
</tr>
<tr>
<td>hemoglobinopathies‡</td>
<td>Maple syrup urine disease 9/22 (41)</td>
<td>5/22 (23)</td>
<td>56</td>
</tr>
<tr>
<td></td>
<td>Congenital adrenal 8/22 (36)</td>
<td>3/22 (14)</td>
<td>38</td>
</tr>
<tr>
<td></td>
<td>Biotinidase deficiency 10/22 (45)</td>
<td>4/22 (18)</td>
<td>40</td>
</tr>
<tr>
<td></td>
<td>Cystic fibrosis‡</td>
<td>2/22 (9)</td>
<td>50</td>
</tr>
<tr>
<td></td>
<td>Homocystinuria</td>
<td>5/22 (23)</td>
<td>40</td>
</tr>
</tbody>
</table>

*Data are reported as a ratio (percentage) unless otherwise noted.
†The false-true counseling ratio is the percentage of programs that counsel for false-positive tests divided by the percentage that counsel for true-positive tests.
‡For these conditions, false positive is taken to include infants receiving a diagnosis of heterozygosity or trait as well as infants receiving a diagnosis because of a laboratory error.

COUNSELING SITES

We found that 19 (54%) of 35 respondents reported that counseling occurred at one of a few closely affiliated sites, while 7 (20%) of 35 respondents reported counseling at “numerous closely affiliated centers (4 or more),” and 3 (9%) of 35 reported that counseling occurs “at many widely dispersed sites, not affiliated with the newborn screening program.” A few respondents added that the county health departments helped provide counseling, and others mentioned toll-free telephone numbers to reach the program with questions.

QUALITY AND ASSESSMENT OF COUNSELING OUTCOMES

Health service providers are subject to increasing pressure to document their credentials and the quality of their work. Many respondents mentioned that counseling providers were either officially certified, required to earn continuing education credits, or both. Some said that they use national guidelines to guide counseling most notably for sickle cell hemoglobinopathy (13/35 or 37%), but also for other conditions with less publicized guidelines (range, 2-11 or as high as 31%). Some respondents added that they have a separate quality control committee for counseling, some of which perform regular site visits of counseling services. When asked specifically, 7 (20%) of 35 respondents reported using some type of systematic measure of quality or outcome. Of the 35 programs that routinely provide counseling, 7 (20%) assess client satisfaction, and 3 (9%) have examined client’s mastery of the information provided in counseling sessions. Three respondents (9%) agreed that they “monitor and maintain a database of future pregnancies and babies born to at-risk couples.”

Since quality measurement of counseling is currently uncommon, we were interested in learning about the perceptions of coordinators regarding the quality of counseling as delivered by professionals with different backgrounds. To standardize the question, we suggested that quality might reflect the counselor’s experience, training background, or their ability to communicate to parents, and we asked the respondents to use a 5-point subjective scale (excellent, good, neutral, fair, poor). The distribution of responses is shown in the Figure, which reveals that the respondents were more likely to rate primary care physicians low, and the 3 specialty professions (including nurse specialists) high. These differences are significant both in...
comparison of simple means (P < .01) and in a χ² comparison of categories (P < .01).

### PURPOSES OF COUNSELING

Given the variation in services, we were curious about the reasons why programs provide counseling. To assess purposes intended, we supplied a list of possible goals identified from the literature, and asked each respondent to rate the importance of each. Table 4 summarizes these goals, categorized into education and advice goals. As a whole, the respondents were strongly in favor of promoting an understanding of inheritance and in sharing information with relatives. We found that 19 (54%) of 35 of respondents rated as important the advice statement to identify children who might be, “for genetic reasons, unsuitable choices for future reproduction.” This item, although worded to avoid a social desirability effect, was meant to at least partially suggest a concern for fitness of affected children to become parents themselves. There was less agreement on advising about future pregnancies in parents, including planning and suggest options for prenatal testing. There was a non-significant trend (χ² test, P = .09) for respondents to rate as unimportant the goal of understanding the difference between the affected homozygous children and any heterozygote carriers.

### COMMENT

Many would agree that families should receive some sort of counseling after positive newborn screening test results, but the best counseling practices and venues are not yet clear. This survey revealed substantial variation in communication services reported by US newborn screening programs. Some respondents reported a centralized process for counseling, but varied widely on specific practices and the professional backgrounds of counselors. In many states, counseling of families is left to the infant’s physician or perhaps not done at all. Few programs arrange, or even recommend, counseling after false-positive and trait results. The respondents said that primary care physicians were responsible for much of false-positive counseling, but also said that the quality of counseling delivered by primary care physicians was lower than that delivered by other professionals. When asked about directive and nondirective counseling techniques, responses were mixed. Some respondents said that they often use directive counseling to advise against subsequent pregnancies, while others are more nondirective and avoid definite recommendations about the next pregnancy.

We were struck by the large number of respondents agreeing with the statement about the “suitability for reproduction” of the patient. During development of the survey, we included this quaint wording with the expectation that most respondents would disagree with the phrasing reminiscent of eugenics. The fact that so many respondents agreed does not necessarily suggest discriminatory beliefs in the respondents, but it does reinforce the importance of further study of counseling content.

Variation among the programs is not entirely surprising. Each program was developed regionally in an autonomous fashion, often without best-practice guidelines for laboratory testing or counseling and with limited communication networks between programs. As a result, newborn screening programs tend to reflect local politics and populations. For example, programs working with large populations of African Americans have more experience counseling families of infants with the sickle cell hemoglobinopathy trait. In contrast, trypsinogen or DNA analysis for cystic fibrosis is a relatively new screening test, and few states have experience with it. Other sources of variation include external resource constraints, local preferences, and the individual creative projects of affiliated academic institutions.

Some believe that practice variation in health care should be eradicated, but that would only be appropriate when optimal practices exist and are not followed. As yet, the optimal practices for counseling after complex screening test results are not known. For example, there are no data to support a preferred professional background for counselors in newborn screening programs. The shortage of genetic counselors suggests that other health care professionals must also be involved. Counseling by primary care physicians seems desirable but could be limited by lack of time, poor understanding of genetics, and questionable counseling skills. Sub-specialist physicians may have a more specific knowledge base, but they are also limited by time and counseling skills. Nurses and other providers may relate well with patients and be cost-effective, but they may require special training and supervision. These and other sources of variation may actually be a strength, allowing comparison of the results or outcomes of counseling. Optimal counseling quality could be

### Table 4. Respondents’ Perceived Education vs Advice Goals for Counseling Parents

<table>
<thead>
<tr>
<th>Goal of Counseling</th>
<th>Rating by Respondents</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Education</strong></td>
<td>Important</td>
</tr>
<tr>
<td>Understanding of the inheritance pattern</td>
<td>25 (100)††</td>
</tr>
<tr>
<td>Provide information about risk that can be shared with relatives</td>
<td>25 (100)††</td>
</tr>
<tr>
<td>Understand the difference between the affected homozygote children and any heterozygote carriers</td>
<td>7 (30)</td>
</tr>
<tr>
<td><strong>Advice</strong></td>
<td>Important</td>
</tr>
<tr>
<td>Identify children who might be, for genetic reasons, unsuitable choices for future reproduction</td>
<td>19 (76)††</td>
</tr>
<tr>
<td>Whether parents would plan future pregnancies</td>
<td>13 (57)</td>
</tr>
<tr>
<td>Suggest options for prenatal diagnoses (amniocentesis or other prenatal testing) during future pregnancies</td>
<td>6 (24)</td>
</tr>
</tbody>
</table>

*Data are reported as number (percentage).
†Using χ² test, compared with even split, P < .05.

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defined by adherence to counseling practices known to be effective and that result in high patient satisfaction, and by the degree of autonomy in decision making desired by the patient.

The problem, of course, is that there is very little experience with measuring counseling effectiveness, especially on a large scale. Methods for more traditional measurement of quality of care are common in other areas of modern health care, and have even been implemented in newborn screening laboratories. In this study, however, we identified only a few programs that evaluate their own counseling with satisfaction surveys or assessment for mastery of counseled information. When asked about quality assurance methods for counseling, some respondents described a certification process and continuing education credits. Unfortunately, these may not necessarily guarantee good counseling practices, particularly in complex tasks. The lack of quality measurement may reflect a tradition where the highly educated are assumed to be good teachers; only recently have health educators been subject to the same scrutiny as other clinicians. This tradition may have to change. Children can be inadvertently harmed by the efforts of newborn screening programs, just as they can experience benefits. The potential for psychosocial harm has been apparent for decades but there have been only limited efforts to prevent problems such as the vulnerable child syndrome. The introduction of outcomes measurement for counseling assessment may not seem popular, but it will probably be necessary to ensure that newborn screening tests have more benefit than harm.

Although some of our findings are striking, they should be interpreted with caution. Our intention for this study was to provide a broad survey of all practices on a national scale, and we are unable to comment on what happens in an individual counseling session. The results are based on the knowledge and opinions of the persons designated as follow-up coordinators. It was obvious to us that some respondents had a more detailed grasp of counseling practices than did other respondents. Some seemed very confident in their answers, and attached materials to support their point. Others admitted in comment sections that they were unsure if their answers applied for their entire state. The analysis is only partially limited by the low number of respondents, since only 6 of the entire national complement of programs declined to participate in this survey.

Nevertheless, the lack of plans for measuring counseling quality and the previously noted lack of coordination between programs are disconcerting because more complex screening tests are likely to appear, either through the Human Genome Project or via other technological advances. If the counseling practices of newborn screening programs are optimized, these programs may become the natural choice for providing advanced tests, given their distinction as the nation’s premier preventive public health program for children. We believe that this study offers 3 implications to this emerging issue. First, newborn screening programs that wish to have more effective counseling in their state should consider closer ties with primary care physicians. This may improve the quality of counseling in the office and may allow access to families with concerns about test results. Second, we would consider diversity among the programs to be an asset, but only when they share their best practices with each other and with the academic community. In this way, improvements may be disseminated widely, and collaborative research can advance understanding of testing and counseling. Finally, the current lack of quality measurement for communication services raises concerns about whether any best practices will ever be identified.

We suggest future research that examines the efficient, effective provision of counseling services and the development of new models for studying the quality of risk communication with parents. For example, parental understanding of newborn screening results may benefit from new approaches to counseling, such as interactive computerized decision aids. A more widespread understanding of these and other counseling services could help improve the lives of affected families and ensure enhanced excellence in newborn screening programs. This agenda may prove critical to meeting the challenges posed by new technologies introduced in the 21st century. Lack of attention to enhancing the quality of newborn screening programs, especially risk communication practices, will almost certainly lead to inadvertent psychosocial harm. Although some efforts have already begun, we believe that addressing issues of communication and potential harm should have a higher priority and will require more effort by organized medicine, especially the American Academy of Pediatrics, Elk Grove Village, Ill, and the Centers for Disease Control and Prevention, Atlanta, Ga.

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