Experience with Parent Follow-Up for Communication Outcomes after Newborn Screening Identifies Carrier Status

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Objective To conduct interviews with a multiyear sample of parents of infants found to have heterozygous status for sickle cell hemoglobinopathy or cystic fibrosis during newborn blood screening (NBS).

Study design Interviewers with clinical backgrounds telephoned parents, and followed a structured script that blended follow-up and research purposes. Recruiting followed several steps to minimize recruiting bias as much as possible for a NBS study.

Results Follow-up calls were conducted with parents of 426 infant carriers of sickle cell hemoglobinopathy, and 288 parents of cystic fibrosis carriers (34.8% and 49.6% of those eligible). Among these, 27.5% and 7.8% had no recollection of being informed of NBS results. Of those who recalled a provider explanation, 8.6% and 13.0% appraised the explanation negatively. Overall, 7.4% and 13.2% were dissatisfied with the experience of learning about the NSB result. Mean anxiety levels were low but higher in the sickle cell hemoglobinopathy group ($P < .001$). Misconceptions that the infant might get the disease were present in 27.5% and 7.8% of parents (despite zero actual risk for disease). Several of these data were significantly predicted by NBS result, health literacy, parental age, and race/ethnicity factors.

Conclusions Patient-centered public health follow-up can be effective after NBS identifies carrier status. Psychosocial complications were uncommon, but harms were substantial enough to justify mitigation. *(J Pediatr 2020;224:37-43)*.

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Newborn blood screening (NBS) saves lives and reduces morbidity, but its population-wide benefits are accompanied by risk of adverse psychosocial consequences for families of infants with false positive results and/or heterozygote "carrier" detection.[1-28] Receipt of abnormal NBS results has been associated with parental stress, depressive symptoms, and anxiety/worry about the infant's well-being.[3-12,24-26] Parents may have misconceptions about results and implications.[20] Some parents have lingering concerns about their child's vulnerability, overuse health services, and have difficulty with future reproductive plans.[1,2,5,12-17] Concerns about the infant's paternity may arise.[17,21,23] Parents also may struggle with decisions about when, how, and what to communicate about genetic information to their child or other family members.[25,29]

Psychosocial complications have been cited in arguments to modify, delay, or even cease some NBS or genetics programs.[27,28,30] Given NBS' benefits, however, we developed a mechanism for follow-up and Communication Quality Assurance: the Wisconsin Project on Improvement of Communication Process and Outcomes after Newborn Screening (referred to hereafter as "the Project").[31-40] The Project's efforts focused on carrier results for sickle cell hemoglobinopathy (SCH) and cystic fibrosis (CF) because false-positive and carrier results are common with the laboratory methods used for NBS.[28,41,42]

Methods

This Project report presents analyses of data from a statewide cohort of parents whose newborns had been identified as carriers for either SCH or CF. Parents were contacted by trained nurses or a genetic counselor following a standardized

CF	Cystic fibrosis
ELSI	Ethical, legal, and social implication
IRB	Institutional Review Board
PCP	Primary care provider
SCH	Sickle cell hemoglobinopathy
NBS	Newborn blood screening

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script. The Project team functioned as a contracted agent of the Wisconsin State Laboratory of Hygiene (a public health entity responsible for Wisconsin's NBS program), and Institutional Review Board (IRB) approval was also obtained.

Participants

Participants were parents of infants found to have genetic carrier status for SCH or CF. The NBS methods were standard at the time we enrolled parents during 2008-2012. Specifically, hemoglobin molecular assessment was done by cellulose acetate electrophoresis. SCH carrier infants had an NBS result showing fetal, adult, and sickle hemoglobin (the "FAS" result). CF NBS used the 2-tier evaluation of immunoreactive trypsinogen with a 96th percentile cutoff and, if above this level, a panel of 23 mutations was used to identify CF-causing variants in the *CFTR* gene. CF carrier infants had an NBS result showing elevated immunoreactive trypsinogen and a single mutation in the CF transmembrane conductance regulator (*CFTR*) gene, followed by a normal result on the infant's sweat chloride testing. The term "likely CF carrier" was used in the Project for infants with an elevated immunoreactive trypsinogen and a single mutation on NBS, but who had not yet had a sweat test.

Standard practice in Wisconsin is for parent notification to be done by primary care providers (PCPs), who have access to guidance and support materials provided by the clinicians and education subcommittee of Wisconsin's NBS program.

Infants were excluded if >1 abnormality was found on NBS, the NBS specimen was a repeat test, the gestational age was <35 weeks, the calendar age at the time of specimen collection was >180 days, or if a PCP could not be identified from NBS records or by calling the birthing facility or home birth provider. In the case of CF, several infants without a known PCP became our clinical responsibility and our IRBs required us to retrospectively censor them from the dataset. During a call to the PCP, infants were excluded if (1) the infant had spent >5 days in the hospital, (2) the infant was rehospitalized after discharge from the nursery, (3) the infant was undergoing evaluation for another serious medical condition, or (4) the parent(s) were reported to need a language interpreter for the interview (because all the interviews were conducted in English owing to limited resources). Finally, (5) infants with CF were excluded if they were found on a periodic review of sweat test results logged by the NBS laboratory.

A multistage process for informed consent was developed to prevent psychosocial harm, mitigate recruiting bias, and respect autonomy. For the Project's first access to results within the NBS program, a waiver was granted by our IRBs. Second, we enabled PCPs to decline participation on behalf of parents for any perceived contraindication.[31,33] Third, parents were mailed an introductory letter that did not describe the result, and only described the Project as an attempt to learn about parents' experiences after NBS. This letter included a decline of contact card that, unlike an opt-out card, enabled parents to decline participation without becoming fully informed about the purpose of the Project. During the telephone call, informed consent was sought at 2 points: an initial consent for audio-recording and a detailed consent once the parent knew about the NBS result and the study's purpose. We clarified that parents could participate in the clinical portion of the call, but decline from the research portion (some did this).

Data Collection and Analysis

The NBS result documents provided infants' gestational ages, birth weights, and birthdays. Birthdays were used to calculate infants' ages on the interview days before identifying information was scrubbed from the database.

A standardized call script was developed with input and feedback from convenience discussions with parents, clinicians, and NBS experts. The script's first draft was aimed at clinical follow-up to verify receipt of the NBS result, screen for misunderstanding, and provide counseling for mitigation of psychosocial complications. The script's wording was then restructured in places to facilitate collection of research data, often as fixed, ordinal-scale questions followed by an opportunity for an open-ended comment. Finally, some research-only questions were embedded in the script in such a way that they would not interfere with the call's clinical purpose. The counseling/support portions were excerpted into subscripts that could be implemented whenever the interviewer felt that the parent was becoming alarmed or confused. The resulting script (**Table I**; available at www.jpeds.com) was 9 pages long for a 20- to 30-minute interview (actual duration averaged 25.3 ± 8.5 minutes).

A set of questions about vulnerable baby syndrome were included, but these data have been reported in another article because of the complexity of the results.[16] The results of the debriefing questions were also reported separately.[35]

The telephone call was scheduled when each infant was between 3 and 5 months old to allow for ≥1 well-baby visit. Contact information for the mother was sought via publicly accessible databases such as telephone directories and the search website Intellius (Bellevue, Washington). The located mothers were mailed the introductory letter, the decline of contact card, and an offer of a $20 gift certificate.

When the call began, the interviewer identified himself or herself and asked to speak to "the mother or whoever takes the infant to doctor visits." Telephone calls were digitally audio-recorded, and transcribed without names or other identifying information. Interviewers kept written notes during the call, and both notes and transcripts were abstracted for fixed answers and other fields in the Project database.

Five outcome variables for communication were derived from the questions shown in **Table I**: (1) whether the parent recalled being told about the NBS result, (2) whether the parent recalled the provider giving an explanation, (3) parent's appraisal of the explanation, (4) parental satisfaction with the entire experience, and (5) misconception about risk for carrier status developing into the actual disease. The fifth outcome was operationalized as adverse if the parent chose any ordinal response other than "definitely not going to have the disease."

Table II. Participant characteristics

Characteristics	SCH carrier group	CF carrier group
Numeric data	Mean (SD)	Mean (SD)
Gestational age at birth, wk	38.9 (1.3)	39.1* (1.2)
Birth weight, g	3288 (484)	3413† (496)
Baby's age at interview, d	107.1 (23.9)	110.8 (25.8)
Infant is female	214 (50.2)	169 (58.7)
Parent's age, y	25.8 (5.9)	28.7 (5.6)
Categorical data	No. (%)	No. (%)
Interviewee is infant's mother	418 (98.1)	278 (96.5)
Parent knows another genetic carrier‡	325 (76.3)	137 (47.6)
Screen positive for health literacy problem	150 (37.7)	104 (36.5)
Parent race data§		
Race-included		
Black-included	280 (65.7)	20 (6.9)
White-included	87 (20.4)	250 (86.8)
Hispanic-included	29 (6.8)	8 (2.8)
Other-included	14 (3.3)	7 (2.4)
Race-only		
Black-only	265 (62.2)	16 (5.6)
White-only	72 (16.9)	246 (85.4)
Hispanic-only	19 (4.5)	5 (1.7)
Other-only	7 (16.4)	4 (1.4)
Multiracial unspecified	5 (1.2)	1 (0.4)
Not asked or answered	36 (8.4)	9 (3.1)
Infant race data§		
Race-included		
Black-included	330 (77.5)	24 (8.3)
White-included	82 (19.3)	255 (88.5)
Hispanic-included	38 (8.9)	14 (4.9)
Other-included	23 (5.4)	11 (3.8)
Race-only		
Black-only	249 (58.5)	14 (4.9)
White-only	13 (3.1)	235 (81.6)
Hispanic-only	15 (3.5)	4 (1.4)
Other-only	2 (0.5)	4 (1.4)
Multiracial unspecified	52 (12.0)	7 (2.4)
Not asked or answered	14 (3.2)	6 (2.1)

*$P < .04$.
†$P < .001$.
‡$P < .0001$.
§Columns for race data do not sum to 100% because categories are not mutually exclusive.

Table III. Communication outcomes after newborn screening identifies genetic carrier status

	Proportion of eligible cases					
	SCH carrier group			CF carrier group		
Adverse outcomes	No.	Eligibles	%	No.	Eligibles	%
1. Parent not informed about the NBS result.	60	426	14.1	n/a	–	–
2. An explanation was not given about the NBS result.	26	366	7.1	7	288	2.4*
3. Negative appraisal of provider's explanation about the NBS result.*	30	351	8.6	36	278	13.0†
4. Dissatisfaction with the experience of learning about the NBS result.	26	352	7.4	37	281	13.2†
5. Misconception about carrier status.	102	371	27.5	22	281	7.8†

n/a, not applicable.
*$P < .01$ vs the SCH group.
†$P < .0001$ vs the SCH group.

Anxiety was assessed using 2 approaches (Table I). First, anxiety at the time of the interview was measured using the Marteau version of the Spielberger State subscale.[18,43,44] The Marteau questions were asked immediately after the second informed consent section, so the parent had just been reminded about the NBS result. In the second approach to anxiety, we asked parents to think back to the time they first learned about the NBS result, and rate their original anxiety on an ordinal scale. Original anxiety responses were excluded from analysis if the parent had just learned about the NBS result from our call.

We also inquired about plans for subsequent pregnancy and genetic testing. However, it is worth clarifying that in our view reproductive and testing plans should be not considered outcomes of communication, because counseling is supposed to be nondirective.

Race/ethnicity data were obtained using open-ended questions (Table I) and abstracted responses into one or more binary fields for each of the standard National Institutes of Health categories.[45] For example, if a parent described his/her ethnicity as "mixed Latino and white" then the database fields for Hispanic and white were flagged.

Health literacy was evaluated with a 3-item screening tool adapted from Chew et al.[46] Analyses were done as applicable for the nature of each variable (χ^2 test, *t* test, correlation, the Wilcoxon rank-sum test, or logistic and linear modeling) using JMP software (SAS Institute, Cary, North Carolina).

Results

We logged NBS results until we identified 1669 infants with SCH carrier status and 800 infants with likely CF carrier status and a known PCP (Figure 1; available at www.jpeds.com). After exclusion criteria were applied and parents agreed to participate in the research portion of the Project, the final sample consisted of 426 in the SCH group and 288 in the CF group (participation rates 34.8% and 49.6% of those eligible).

Table II lists participant characteristics. For nonparticipants there were no significant differences in the available data (gestational age, birth weight, or birthing facility). Also shown in Table II are the race/ethnicity data abstracted from answers to our open-ended questions into "included" and "only" categories for each of the National Institutes of Health categories. Both CF and SCH groups demonstrated diversity in the race/ethnicity data. For example, responses in the SCH group led to the black-only variable in only 62.2% of parents and 58.5% of infants.

Screening for limited health literacy was positive in similar proportions of the SCH and CF groups. Limited health literacy was more common for older parents ($P < .'0001$ on regression). We also found 2 race/ethnicity differences: health literacy limitations were rarest for parents with white-only race/ethnicity (32.8% vs 50.1%; $P < .03$ on χ^2), and more common for parents with Hispanic-included race (56.8% vs 36.1%; $P = .01$ on χ^2).

Communication Outcomes

The 5 outcome variables are presented in Table III. For the misconception question, 7.8% of parents in the CF group indicated some lingering question that their infant might still develop the disease. The SCH group had more misconceptions (27.5%; $P < .0001$ on χ^2). A favorable combination of outcomes in the SCH group was present for 73% of parents who were informed, recalled an explanation, and appraised the explanation positively. In the CF group, 84% of those informed recalled an explanation and appraised it positively.

There were several characteristics associated (using stepwise regression) with the 5 adverse outcomes. In the SCH group, failure to recall being informed about the result was independently associated with parental white-included race/ethnicity (OR, 9.1; $P < .0001$) and infant Hispanic-included race/ethnicity (OR, 6.7; $P < .03$). In the CF group, biracial/multiracial status was independently associated with failure to recall an explanation (OR, 12.5; $P < .01$), and negative appraisal of explanation if it was recalled (OR, 2.5; $P < .03$). A negative appraisal of SCH carrier explanations was independently associated with a positive health literacy screen (OR, 1.5; $P < .01$).

Dissatisfaction with the entire experience in both groups was independently associated with failure to recall an explanation (SCH group OR, 4.0; CF group OR, 3.8; both $P < .0001$). Dissatisfaction was also correlated with negative appraisal of the explanation (SCH group r = 0.71; CF group r = 0.67; both $P < .0001$). In the SCH group, dissatisfaction was independently associated with infant white-only race/ethnicity (OR, 1.8; $P < .03$) and parental bi/multiracial status (OR, 0.54; $P < .05$).

The misconception outcome was independently associated in the SCH group with younger parental age (OR, 1.13 per year; $P < .0001$) and in the CF group with biracial/multiracial status (OR, 7.7; $P < .03$).

Anxiety

Figure 2 depicts anxiety immediately after being reminded of the NBS result, with the Marteau data prorated to the 20-80 range used by Spielberger.[18,43,44] Median anxiety scores were 26.6 and 23.3, respectively, for the SCH group and CF group ($P < .002$ on Wilcoxon). On average these results indicate low anxiety levels, but about 7% of parents had scores of >50. In the SCH, group higher anxiety was associated with lower birth weight and limited health literacy.

Higher Marteau responses were correlated with more negative appraisal of the explanation for both groups (CF group, r = −0.15, $P < .02$; SCH group, r = −0.12, $P < .02$). In the SCH group, Marteau data correlated with dissatisfaction (r = −0.14, $P < .01$) and was associated with a misconception that the baby might develop sickle cell disease (OR, 1.05; $P < .0001$).

When parents were asked about anxiety from when they originally learned about the NBS result, their ordinal ratings were in the 3 most worried responses (of 5 options) in 35.6% of the SCH group and 77.2% of the CF group. Comparing all ordinal responses, the CF group recalled more anxiety than the SCH group (medians of 5 and 3, respectively; $P < .0001$ on Wilcoxon). In the SCH group,

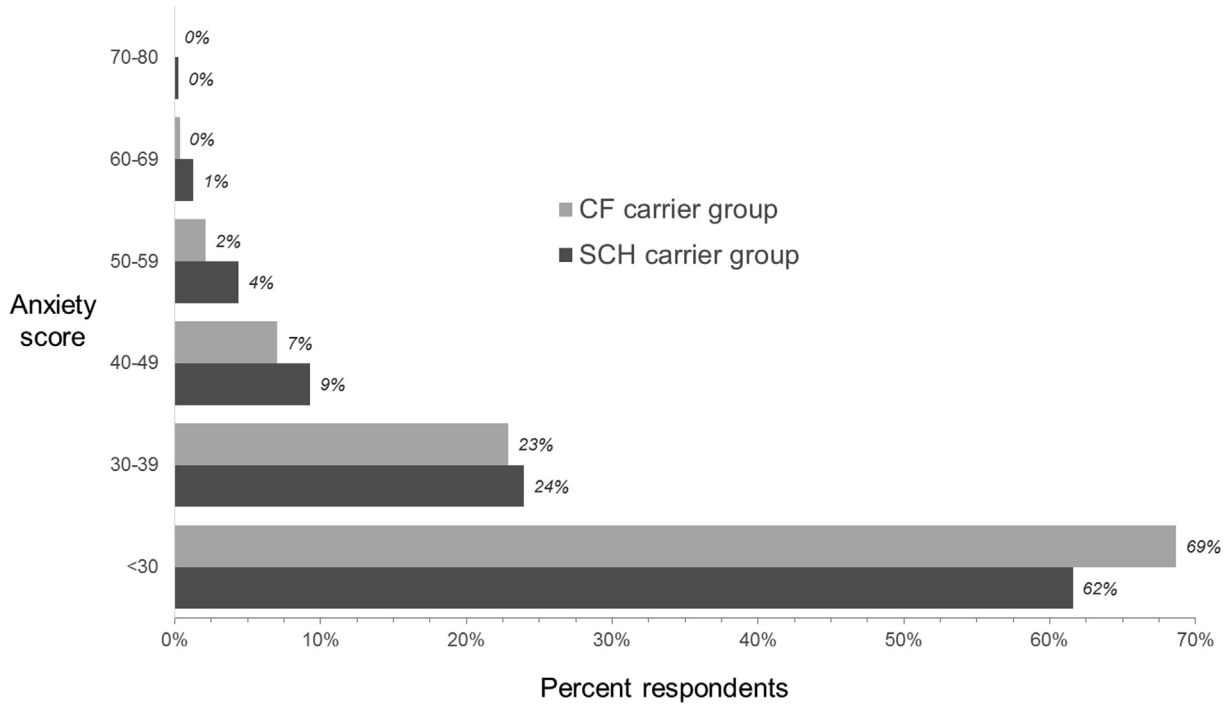

Figure 2. State anxiety assessed at the time of the interview.

rating of original anxiety was independently associated with older parental age (OR, 0.94 per year), infant race/ethnicity other-included (OR, 1.6), and for parents who knew another carrier (OR, 0.47).

Satisfaction with the NBS experience was associated with rating of original anxiety (CF group OR, 0.70 [$P < .04$]; SCH group OR, 0.68 [$P < .04$]). In the SCH group only, rating of original anxiety was associated with a misconception that the infant might develop sickle cell disease (OR, 1.6; $P < .0001$). In the SCH group, there was a modest correlation between anxiety at the time of the interview and rating of original anxiety ($r = +0.18$; $P = .0007$). In the CF group, there was no correlation between the 2 anxiety assessments.

Parent's Reproductive Plans for the Future

Table IV shows the proportions of parents who planned a future pregnancy or testing for carrier status. The likelihood of planning another pregnancy was lower in both groups for older parents (OR, 0.9 per year; each $P < .0001$), and in the SCH group for parents with black-only race/ethnicity (OR, 0.58; $P < .05$). In the CF group, parents with a possible health literacy problem were more likely to plan another pregnancy (OR, 4.3; $P < .001$).

The likelihood of planned testing for self was higher for parents who reported knowing another carrier (for SCH group OR, 3.1; for CF group OR, 3.4; both $P < .01$), and for CF group parents with a possible health literacy problem (OR, 3.0; $P < .03$). For race/ethnicity, plans for testing were more likely with the black-included and black-only states (ORs of 5.7 and 7.4 [$P < .0001$] in the SCF group, and approaching certainty in the CF group [$P < .05$]). There was an independent effect for the white-included and white-only states (ORs of 0.13 and 0.09; $P < .0001$), and for biracial/multiracial status in the SCH group (OR, 0.24; $P < .0001$). Similar race/ethnicity factors were associated with likelihood of planning testing for a partner.

Another pregnancy was planned by more parents in the CF group than the SCH group (OR, 1.7; $P < .009$ on χ^2). All predictors of pregnancy planning were independent of each other. When a parent planned to have himself or herself tested, then she or he was very likely to plan having the infant's other parent tested (OR, 36.5 for CF group; OR, 24.9 for SCH group [$P < .0001$ for both]). However, significantly fewer of the CF group planned testing of partners than parents in the SCH group (OR, 0.53; $P < .008$). Parents were more likely to plan testing if they had higher ratings of original anxiety at the time of learning about the NBS result (OR, 2.3; $P < .03$).

Table IV. Parents' future plans for pregnancy and/or testing

	Proportion of eligible cases					
	SCH carrier group			CF carrier group		
Adverse outcomes	No.	Eligibles	%	No.	Eligibles	%
Parent plans another pregnancy	99	231	42.9	97	173	56.1*
Parent plans to have himself/herself tested for carrier status	251	289	86.9	189	225	84
Parent plans to have partner tested for carrier status	211	250	84.4	156	210	74.3*

*$P < .01$ vs the SCH group.

Discussion

Genetic screening programs for disease risk have been debated for decades, especially when test methods incidentally identify carriers. NBS is even more subject to debate because formal consent is not sought before testing. We sought to improve NBS safety, that is, mitigate harms that are incidental to NBS's benefits.[31-40] We describe our systematic experience with a NBS follow-up program for Communication Quality Assurance that was both public health and patient centered and provide updated epidemiologic insights about NBS after incidental findings.

Our experience was that a public health follow-up program for Communication Quality Assurance is clearly feasible, and we believe affordable (<2 full-time equivalents of personnel for our entire state's average 68 000 births per year). As part of a public health program, we theoretically could have been exempt from some IRB regulations, but we requested oversight because of the limited literature on follow-up programs. As a result, we lost a few hundred parents from the research portion of the Project (10.8% loss for SCH, 8.6% for CF), but only a small percentage of the lost parents could have known that we were studying communication and psychosocial complications related to carrier status. The final sample was large enough to detect modest effect sizes and adjust for covariance.

Adverse outcomes had a modest-sized incidence in our sample, but individually were still troubling. Our most concerning result may be the number of parents with the misconception that their infant might develop the disease. In the SCH group some misconceptions could be attributed to confusion about carrier adults' small risk of events.[47-51] Even so, both groups' misconceptions are surprising because all such parents in Wisconsin are offered genetic counseling.

We identified risk factors that might help to improve follow-up by NBS programs or PCPs. Health literacy covaried with several factors, but health literacy's only independent association was for appraisal of explanations in the SCH group.

SCH results were independently less likely to be communicated by PCPs if the parent was white-included or the infant was Hispanic-included. CF results were less likely to be explained for biracial/multiracial families. We can only speculate that these effects were influenced by PCP confusion about genetic epidemiology of CF and SCH. Perhaps parents were also confused by stereotypes between genetics and race/ethnicity. Regardless, our Project clearly documented the

error of assuming that all SCH carriers are black, and all CF carriers are white. We believe that because NBS will continue to include CF and SCH (ie, infants with disease continue to need early identification), then NBS reports should be accompanied by better information for PCPs about carrier status and race/ethnicity.

Our findings about parents' reproductive and testing plans were informative. However, these parent decisions should not be considered outcomes of communication after NBS. We agree with the predominant view that NBS exists to identify infants with diseases (who therefore will benefit from treatment or surveillance), rather than for carrier identification or reproductive decision-making.

Some limitations are worth considering. Most participants were mothers because the NBS card identified the mother. We sought to minimize recruiting bias, but regret that limited resources did not allow us to include parents with language barriers. Health literacy was assessed with a widely used screening tool, but we recognize that some may be concerned about the tools' applicability to NBS studies. Our analyses are of association rather than causation, but such results are still important for projecting risk of adverse communication outcomes. Most of our regression modeling assumed linear relationships, but we could have missed more complex effects because linearity could not be guaranteed for the entire distribution. Many of the results depend on the parents' summative recall or heterogeneous experiences; a PCP could have explained the NBS result, but the parent may not have recalled it vividly enough to report in the interview. However, this limitation could be seen as a strength, because an explanation about an important issue is presumably ineffective if it is not memorable 2-3 months later. Notwithstanding these limitations, our epidemiologic findings and experience seem to emphasize the need for Communication Quality Assurance after NBS. We previously reported how our intervention seemed to have been effective and well-received by parents.[35]

Our experience and epidemiologic insights may also be informative for broader efforts with genetics in public health screening. Future study will be needed to test communication gaps, risk factors like race/ethnicity or health literacy, and the use of Communication Quality Assurance.

Finally, this Project has led us to reconsider some basic aspects of scholarship on the ethical, legal, and social implications (ELSIs) of genetics. Much of ELSI scholarship has considered whether genetics should (or should not) expand into new diseases or methods. We have seen NBS expansions prompted mostly by advocacy, politics, and the attractiveness of new laboratory techniques. We suspect that genome sequencing on blood spots will be routine within the coming generation, regardless of ELSI concerns. We suggest that geneticists and ELSI scholars adopt a safety perspective, and invest more effort to mitigate harm after molecular technologies.[52] We believe that the time has come to shift from the question, "Should we screen or not?" to ask, "How can we make DNA-based screening tests safe for all infants and families?" ■

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Data Statement

Data sharing statement available at www.jpeds.com.

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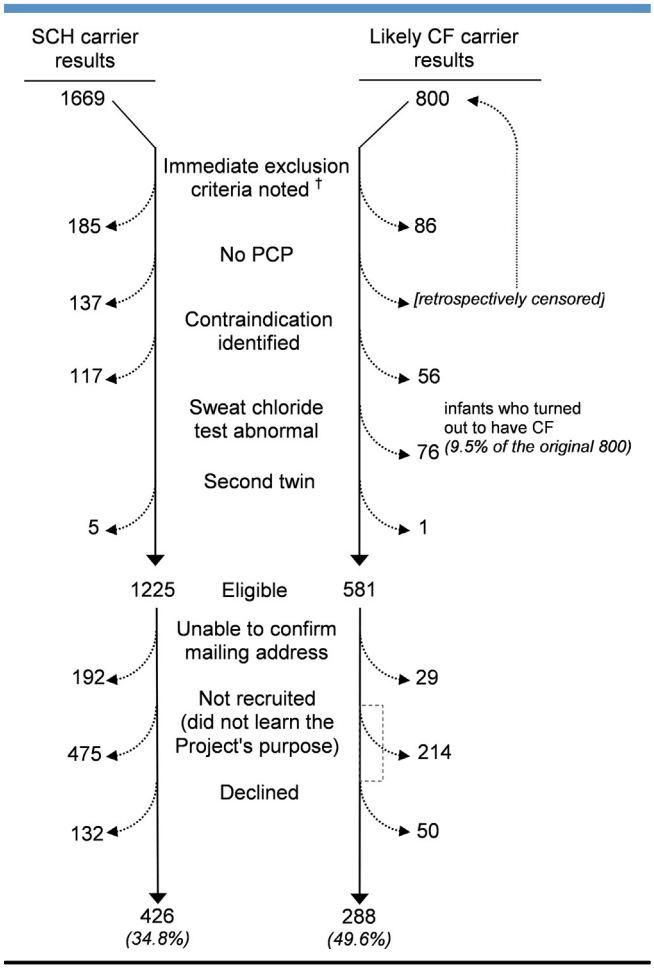

Figure 1. Exclusions and losses during recruitment.

Table I. Questions and timing of segments in the interview script*

Segments	Description
Information giving	Introduction, initial consent for recording a call about NBS
Data collection	Assessment of recall "What do you remember from when the NBS test took place?" If parent does not mention the result… "Do you follow up with [source] for [baby]?" "Do you remember having a conversation with [source] about the screening results?" If not result not known or recalled, inform result without details "Screening showed that [baby] is probably something called a carrier of the gene for a disease called [disease]. [If CF carrier, ask about sweat test.] Is that information you had heard before?"
Information giving	Detailed informed consent for interview about the specific NBS result
Data collection	Marteau instrument for anxiety at the time of the interview Health literacy questions adapted from Chew[39] "When you were in the hospital for [baby]'s delivery, how confident or comfortable did you feel with reading the brochures and handouts that the hospital gave you? 'Extremely,' 'Quite a bit,' 'Somewhat,' 'A little bit,' or 'Not at all?'" "When you were in the hospital for [baby]'s delivery, how much (if any) help did you need to fill out the medical forms related to the birth? 'Quite a bit,' 'Some,' or 'None?'" "Before [baby] was born, how often did you have a hard time learning about medical problems because of difficulty understanding things that are written down? 'Always,' 'Often,' 'Sometimes,' 'Occasionally,' or 'Never?'" Reaction to communication services (if parent had heard about the result) "You mentioned earlier that you heard about [baby's] result before … Can you rate on a scale of 1 to 5 how satisfied or happy you were with the way you heard about the result, with '5' being 'very satisfied' and '1' being 'very dissatisfied?'" "How well did the [source] explain the screening result to you, on a scale of 1 to 5, '5' being 'explained very well' and '1' being 'no explanation at all?'" "How worried did you feel when you first heard about the screening result, on a scale of 1 to 5, '5' being 'very anxious,' '3' being 'a little concerned,' and '1' being 'not worried at all?'" Age, race/ethnicity questions "Did you feel like your interactions, meetings, or conversations with the [source] were affected by differences between you and the [source], like maybe differences in age or differences in race or ethnicity? … How old are you? … How would you describe your race or ethnicity? … How would you describe your baby's race or ethnicity?" Misconception about risk for developing the disease "Based on what you know now, how likely is it that [baby] is going to have [disease], the disease, on a scale of 1 to 5, with '5' being 'definitely going to have it,' '3' being 'unsure' and '1' being 'definitely NOT going to have it?'"
Information giving	Detailed education, counseling, and support
Data collection	Plans for the future "So, do you mind if I asked about your plans now?" If necessary… "Are you planning to have another baby in the future?" "Do you think that you will get yourself and [other parent] tested to see if you are carriers, too?"
Information giving	Debriefing and closure

*Not shown: subscripts for ad hoc counseling and support.