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Factors that Influence Parents' Experience with Results Disclosure after Newborn Screening Identifies Genetic Carrier Status for Cystic Fibrosis or Sickle Cell Hemoglobinopathy

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Abstract

Objectives—Newborn screening (NBS) identifies genetic carriers for sickle cell hemoglobinopathy and cystic fibrosis. We aimed to identify factors during initial NBS carrier results disclosure by primary care providers (PCPs) that influenced parents' experiences and reactions.

Methods—Open-ended responses from telephone interviews with 270 parents of carriers were analyzed using mixed-methods. Conventional content analysis identified influential factors; chi-square tests analyzed relationships between factors and parent-reported reactions.

Results—Parents reported positive (35%) or negative (31%) reactions to results disclosure. Parents' experiences were influenced by specific factors: content messages (72%), PCP traits (47%), and aspects of the setting (30%). Including at least one of five specific content messages was associated ($p<0.05$) with positive parental reactions; omitting at least one of four specific content messages was associated ($p<0.05$) with negative parental reactions. Parents reported positive reactions when PCPs avoided jargon or were perceived as calm. Parents reported negative reactions to jargon usage and results disclosure by voicemail.

Conclusions—Parents identified aspects of PCP communication which influenced their reactions and results disclosure experience.

Practice Implications—Our findings suggest ways PCPs may improve communication of carrier results. PCPs should provide specific content messages and consider how their actions, characteristics, and setting can influence parental reactions.

Keywords

Newborn screening; cystic fibrosis carrier; sickle cell trait; results disclosure; heterozygote; genetic carrier detection; psychosocial outcomes; genetic counseling; patient-provider communication; patient preferences; parental anxiety

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1. Introduction

Newborns routinely undergo newborn screening (NBS) for genetic, endocrine, and metabolic conditions. These include cystic fibrosis (CF) and sickle cell hemoglobinopathy (SCH), two commonly inherited recessive disorders [1]. While the main goal of NBS is to identify infants affected with these conditions, screening also inadvertently identifies infants who are heterozygous genetic "carriers." Genetic carriers are healthy infants who inherited a single mutation for an autosomal recessive condition but do not show symptoms or have the disease. As NBS continues to expand and incorporate molecular-based testing methods, incidental identification of carriers will likely increase. Responsibility for disclosing NBS carrier results to families generally falls to primary care providers (PCPs). Because PCPs may be the only source of information for carrier families [2], it is essential that PCPs effectively communicate the NBS results.

However, communicating NBS carrier results is challenging because their meanings are complex and many PCPs have limited knowledge about NBS and genetics [3–6]. Parents of carrier infants often experience psychosocial complications after learning their infant's NBS results including confusion, misconceptions, anxiety, depression, stress, or vulnerable baby syndrome [6–13]. While previous studies have documented these psychosocial complications throughout the 'NBS process' (including follow-up, confirmatory testing, and periods of waiting or uncertainty), the focus has not been on *initial* NBS results disclosure. In fact, the distinction between initial NBS results disclosure and the 'NBS process' is rarely made. Initial NBS results disclosure is the *first* conversation, usually between parents and PCPs, when parents are told their infant's NBS results. Thus, there is limited literature addressing parents' experiences with initial NBS carrier results disclosure by PCPs, or with the effects of PCP communication on parental reactions resulting from this first conversation.

The purpose of this mixed-methods analysis was to focus on the initial NBS results disclosure by PCPs. We aimed to identify specific factors during initial NBS carrier results disclosure by PCPs that influenced parents' experiences and reactions by qualitatively analyzing parents' opinions of their experience, and quantitatively analyzing relationships between influential factors and parent-reported reactions.

2. Methods

2.1. Study Design

This mixed-method analysis focused on interviews conducted during follow-up telephone calls with parents after NBS. Open-ended questions solicited parents' opinions of initial NBS carrier results disclosure in a richer fashion than was possible solely through scaled questions. Analysis focused on parents' responses to three questions, capturing both positive and negative aspects of initial results disclosure:

1. What do you remember from when the newborn screening took place?
2. Looking back on the conversation with [PCP], what did you find most helpful about the way you were told your infant's results?
3. What could have been done better when discussing those results?

2.2. The Project

The Wisconsin Project on Improvement of Communication Process and Outcomes after Newborn Screening (hereafter called "the Project") was developed to evaluate parents' experiences and psychosocial reactions after NBS identified carriers of SCH or CF. The

Project evaluated the quality of PCP communication when delivering NBS results, assessed parents' psychosocial reactions after results disclosure, and provided counseling if parents had questions about the results. This quality improvement effort was a partnership between the Wisconsin State Laboratory of Hygiene (WSLH) NBS Program, Department of Health Services, and the Medical College of Wisconsin as a contracted project agent [14]. The Institutional Review Boards at the Medical College of Wisconsin and University of Wisconsin-Madison approved the Project.

2.2.1. Recruitment for the Project—The Project included NBS results for every infant born in Wisconsin showing carrier status for SCH or CF. This all-inclusive, systematic sampling allowed for a non-biased and representative study sample. The Project considered SCH carriers those infants with fetal, adult, and sickle hemoglobin ("F-A-S") on NBS. These F-A-S results were detected on NBS by hemoglobin electrophoresis and were definitively considered carriers, therefore confirmatory testing was not necessary. The Project considered CF carriers those infants whose initial NBS showed elevated immunoreactive trypsinogen (IRT) with a single *CFTR* mutation, and had a subsequent negative sweat test. The initial NBS lab result sent to PCPs indicated the infant was at least a CF carrier. After initial NBS results disclosure, PCPs then referred families for sweat testing due to a 2–5% chance the infant had an unmeasured second mutation, resulting in CF disease [15,16]. Infants were also excluded if they had multiple abnormal NBS results, were considered "medically fragile," or had their initial NBS performed after 180 days from birth. Parents of eligible infants were excluded if they had previously declined participation or PCPs identified contraindications to follow-up.

Remaining parents were mailed a recruitment letter when infants were 2–3 months old. Letters explained the study and contained a "decline of contact" card, providing parents an opportunity to decline participation without becoming fully informed about the Project. If no decline of contact card was returned, a study team member telephoned eligible parents to interview them about their experiences and reactions following NBS results disclosure. Parents who chose to participate in the telephone interview were mailed $20 gift certificates.

2.2.2. The Project's Interview—Interviews were conducted when infants were 3–5 months old. Interviews lasted approximately 20–30 minutes, but ranged from 12–84 minutes since parents were encouraged to talk as long as necessary to provide their opinions. The interview questions and counseling script were developed by a team of methodological and content experts in pediatric medicine, NBS, genetic counseling, nursing, communication, psychology, and bioethics. The script balanced research questions with informed consent, parent education, and emotional support. It contained 35 fixed-answer and scaled questions about parental anxiety, health literacy, infant vulnerability, and overall results disclosure satisfaction. Fifteen other open-ended questions assessed parents' experiences with the NBS process, results disclosure, previous knowledge or family history of carrier status, and future reproductive plans. The three open-ended questions for this analysis were the only interview questions inviting parents to fully elaborate on their initial results disclosure encounter. Probing questions such as "How so?" and "Can you tell me more?" were used to clarify parents' responses if needed.

Interviews were conducted by one member of a multidisciplinary team with backgrounds in nursing, medicine or genetic counseling; all trained to assess signs of parental emotional distress or confusion. If serious distress or confusion was identified, the parent or interviewer had the option of forgoing all research questions so the interview became a clinical intervention. All interviews ended by clarifying misconceptions and providing resources if parents had additional questions. Interviews were digitally audio-recorded, transcribed, and de-identified.

2.3. Sample for this mixed-methods analysis

A total of 317 parent interviews were conducted and transcribed between March 2008 and August 2010. Of these, 47 transcripts were excluded because interviews were incomplete or became clinical interventions, in which case the three research questions used in this analysis were not asked. The remaining 270 transcripts formed the dataset for this analysis. Demographic information was self-reported (Table 1).

2.4. Data Analysis

Parents' responses were extracted from transcripts and coded using adaptations of conventional content and dimensional analysis procedures [17–19]. Line-by-line coding was conducted on a subset of extracted data to identify categories and develop a coding schema [17,20–22]. Coders compared their initial impressions of data to capture key concepts. Codes represented specific words verbalized by parents in response to interview questions. Codes and key concepts with similarities were clustered into themes and continually refined until no new themes emerged. Themes were organized into subthemes and specific qualifying codes to form a codebook for the entire dataset [20,23,24]. Disagreements about coding were resolved through discussion, consensus ratings, and codebook revisions. A second subset of data was coded to reach saturation in the coding system.

Finally, group consensus coding was performed on 80% of the total transcripts over 10 rounds. Individual coding was performed between each round of consensus coding on 20% of the total transcript pool to promote discussion and avoid coding drift or group bias. Parents occasionally responded to interview questions using hypothetical scenarios, indicating an assumed preference for results disclosure (example: *"I would prefer like one-on-one but I guess I wouldn't want to, you know, have [PCP] waste her time and me waste my time by going to the doctor's office…")*. Coders did not code hypothetical scenarios, but rather focused exclusively on parent accounts of what actually happened during results disclosure and parental reactions. Final codes were analyzed using NVivo 8 [25]; t-tests and chi-square analyses were performed using SPSS [26].

3. Results

3.1. Themes

This section identifies factors reported by parents that most influenced their NBS results disclosure experiences and reactions, described as three themes: (1) specific content messages, (2) PCP traits, and (3) aspects of the setting. Parents' responses refer specifically to initial NBS results disclosure with PCPs.

3.1.1. Specific content messages that influenced parents' experiences and reactions (Table 2)—Parents often described specific content messages provided by PCPs as the most helpful aspect of results disclosure (n=195/270, 72%; parents of CF carriers 79/99, 80%; parents of SCH carriers 116/171, 68%).

Many parents reported content messages they appreciated hearing during results disclosure, while others reported content messages they wished the PCP had included in the explanation. Twelve discrete content messages emerged from parents' responses; however each message was not reported by every parent.

Parents reported a "better understanding" of the results or feeling "reassured," "relieved," "comforted," or "calm" when any of five specific content messages were present: infant is a carrier; infant does not have a disease; infant is healthy, not sick; parents should not worry about the results; and information on statistics, inheritance or family history. Parents reported feeling “scared,” “worried,” “confused” or “frustrated” when any of four content messages were omitted from results disclosure: infant is a carrier; infant does not have disease; explanation or clarification of carrier status; and information on sweat test process (CF only). Parents frequently reported an additional five content messages as helpful to hear: explanation of NBS process; explanation of disease; information on next steps for parents and infant; difference between having disease and carrier status; and the likelihood carriers will have health complications. Some parents (17%) reported that no explanation or little information was provided during the results disclosure encounter. Notably, no parents reported that being told their infant’s carrier status was unwanted information.

Several parents reported they appreciated receiving additional written information. This written material reportedly was the most helpful aspect of results disclosure for some (n=78/270, 29%; parents of CF carriers 19/99, 19%; parents of SCH carriers 59/171, 35%). Supplemental written information included pamphlets, brochures, illustrations, copy of NBS results, or contact information for specialists.

> *“It would definitely have to be the Sickle Cell trait pamphlet [PCP] gave to me. Yes, because she told me, but I guess we needed it on a sheet of paper to really know what it is – it’s just so much more helpful. It was – the pamphlet was awesome. It was not until that I got home and actually read the pamphlet and read it over and it was like, okay, this is what she means then I understood.”* (mother, SCH carrier infant)

3.1.2. PCP traits that influenced parents’ experiences and reactions (Table 3)

—Parents described several PCP behaviors and characteristics that influenced their experience (n=126/270, 47%; parents of CF carriers 63/99, 64%; parents of SCH carriers 63/171, 37%).

When PCPs took time to assess parents’ level of understanding or answer questions, parents reported feeling “reassured,” “confident,” “comfortable” or “less worried.” These behaviors reportedly allowed parents to ask questions and ensured their understanding of results. Though some parents reportedly were familiar with the NBS results due to family history or medical background, they reported it helpful when PCPs did not assume parental knowledge about the meaning of the result and repeated the information. Some parents reported having a “better understanding” of the results and feeling “comfortable” when PCPs avoided medical jargon. Conversely, parents reported feeling “scared” or “confused” when PCPs used jargon. Parents reported feeling “reassured” or “relieved” when PCPs were perceived as thorough in the explanation or knowledgeable about the result. Parents described these characteristics as “informative,” “educational” or “helpful.” If PCPs were not perceived as knowledgeable or thorough, parents noted this absence as problematic. Finally, parents reported feeling “calm,” “reassured,” “comfortable” or “less worried” when they perceived PCPs as calm.

3.1.3. Aspects of the setting that influenced parents’ experiences and reactions (Table 4)

—Parents identified aspects of the setting such as the method of disclosure, source of information and timing that influenced their experience (n=81/270, 30%; parents of CF carriers 45/99, 45%; parents of SCH carriers 36/171, 21%).

Some parents reported a preference for hearing the results over the phone because they could speak directly with PCPs and ask questions “immediately.” Other parents explained that receiving results over the phone was concerning because it took them by surprise, provided limited information, or occurred late at night. Telephone calls were further complicated by the potential for voicemails; every parent who reported hearing NBS results as voice messages described feeling “anxious” or “worried” as a consequence.

Parents who received NBS results from PCPs “face-to-face,” as part of a regularly scheduled appointment, said this method of disclosure was most helpful. The established relationship and personal contact with the PCP was described as “reassuring,” “comforting” or “personable.” However, parents reportedly found receiving results from unfamiliar persons (e.g., lab technicians or nurses) unsatisfactory because these individuals were generally brief, unable to answer questions or lacked information about the result. Finally, parents who received results on Fridays or before long holiday weekends described “waiting” was the most difficult aspect of results disclosure because it postponed immediate access to further information, resources, or scheduling follow-up appointments.

3.2. Positive and negative reactions reported by parents (Table 5)

Parents reported one or more positive reactions (n=94/270, 35%; parents of CF carriers 31/99, 31%; parents of SCH carriers 63/171, 37%) and/or negative reactions (n=84/270, 31%; parents of CF carriers 29/99, 29%; parents of SCH carriers 55/171, 32%) resulting from their NBS results disclosure experience. Many parents did not report a reaction (n=124/270, 46%; parents of CF carriers 49/99, 49%; parents of SCH carriers 75/171, 44%).

Positive reactions were categorized as either emotional or cognitive. Positive emotional reactions included feeling “reassured,” “relieved,” “comforted,” “calm” or “happy.” Responses were considered positive cognitive reactions when parents reported an “understanding” of the NBS results or experienced increased “trust” in the PCP resulting from results disclosure. Negative reactions were categorized as emotional, cognitive, physical or social. Negative emotional reactions included feeling “scared,” “anxious,” “shocked,” “surprised,” “miserable,” “upset,” “stressed” or “frustrated.” Responses were considered negative cognitive reactions when parents reported “confusion,” thought infant had disease, stated “my mind went blank,” or did not remember what PCP said after hearing the results. Negative physical reactions experienced by parents included “crying,” “racing heart,” “sleepless nights” or becoming “sick.” Finally, negative social consequences were those parents who reportedly had no support system during results disclosure or changed PCPs due to dissatisfaction with their disclosure experience.

3.3. Associations between Factors and Reactions

Associations between influential factors and reactions reported by parents are designated in Tables 2–4 with distinct symbols. If parents reported factors they liked, we tested for associations with positive reactions; if parents reported factors they disliked, we tested for associations with negative reactions.

Including at least one of five specific content messages during results disclosure (Theme 1) was significantly associated ($p<0.05$) with positive parental reactions (Table 2). However, the omission of at least one of four specific content messages during results disclosure (Theme 1) was significantly associated ($p<0.05$) with negative parental reactions (Table 2). Furthermore, two of these content messages: infant is (likely) a carrier and infant (likely) does not have disease emerged as most important to include in results disclosure based on frequency and associations with parental reactions. If either of these two content messages was included in the discussion, an association with positive parental reactions was found ($p<0.001$). If either of these two content messages was omitted, parents reported the absence and the omission was associated with negative parental reactions ($p<0.01$).

Parents who reported that PCPs used medical jargon (Theme 2) were significantly more likely ($p<0.001$) to report negative reactions, while parents who reported PCPs avoided jargon were significantly more likely ($p<0.05$) to report positive reactions (Table 3). Parents who perceived PCPs as calm (Theme 2) were more likely ($p<0.05$) to report positive reactions (Table 3). An association ($p<0.001$) was found between parents who reportedly received their infant's NBS results as a voice message (Theme 3) and negative reactions (Table 4).

4. Discussion and Conclusion

4.1. Discussion

This analysis identified several factors and reactions associated with parent perceptions of PCP communication regarding NBS carrier results disclosure. Parents identified specific content messages, PCP traits, and aspects of the setting that influenced their reported positive or negative reactions. These findings suggest ways for PCPs to improve communication of carrier results, which may optimize parental understanding and minimize emotional distress related to NBS carrier results disclosure for CF and SCH.

By focusing on initial NBS results disclosure by PCPs, this analysis differs from previous research that has investigated psychosocial complications throughout the NBS process [3,27,28]. Additionally, we focused on opinions and reactions from parents of SCH carrier infants, a population whose NBS experiences have been understudied compared to CF carrier families.

This analysis found similar reactions reported in both SCH and CF parent groups (Table 5). Approximately one-third of parents in both groups reported positive emotional and cognitive reactions resulting from their NBS results disclosure encounter, such as understanding results or feeling reassured. These findings suggest that positive reactions can be achieved across patient populations, if PCPs are mindful of the parent-reported preferences identified in this analysis. We also found that one-third of parents in both groups reported negative reactions, consistent with previous reports of emotional distress and uncertainty [3,5,10,13,28–30]. Our findings extend previous research by reporting that parents may experience undesirable physical reactions resulting from NBS disclosure such as having a racing heart and becoming physically ill. Though infrequent, our parents reported social consequences such as not having support available during disclosure or changing infants' PCP based on a negative disclosure experience. Disclosure of NBS carrier results may be regarded by PCPs as "routine," however, these parental reports suggest that it has a greater impact on parents than PCPs may realize. Parents in both CF and SCH groups in this analysis provided detailed descriptions of their distress associated with the initial disclosure, even months after the encounter.

This analysis found that families of both SCH and CF carriers want to be initially informed about their infant's NBS results by someone who is knowledgeable about the results and provides a thorough explanation, a sentiment echoed in previous studies [5,6,13,31,32]. We suggest this may be partially achieved if PCPs include important content messages during disclosure. Previous studies suggest statements that carriers do not have disease; carriers are healthy, not sick; or information on genetics and inheritance may be important messages for parents [5,32,33]. Parents in our analysis frequently reported these same three content messages in addition to nine others (Table 2). A larger proportion of CF parents, compared to SCH parents, reported the most important three content messages were not to worry; infant is most likely a carrier; and infant likely does not have CF. Perhaps this is because NBS results are not definitive at the time of initial disclosure. Consequently, only CF parents wanted information about sweat testing procedures and CF disease symptoms. Alternatively, a greater proportion of SCH parents reported the most important two content messages were infant is healthy, not sick; and health complications associated with SCH carrier status. Parents of SCH carriers were most interested in information about carrier status, likely due to definitive NBS results. Similar to CF parents, though not proportionally greater, SCH parents reported it was important to receive the not to worry message. Though individual content messages may appear closely related, each message is unique and distinct. Although one message may infer another, parents in this study reported the desire to hear each message individually.

Both groups of parents varied their preferences for aspects of the results disclosure setting. Our results support previous findings that parents liked being told results in person at a regularly scheduled office visit [32], so it may be ideal to discuss results at a regularly scheduled two or four week well-child assessment. Other approaches may be used based on clinic practice or if NBS results are time-sensitive, however, PCPs should be aware that our analysis and others have found that parents who received NBS results via letter or telephone experienced stress and discomfort [3,5,6,32]. Though similar distress was reported by parents of SCH and CF carriers in this analysis, we should note that other parents in both groups reported appreciation for telephone disclosure. Regardless of parental preference for or against telephone disclosure, this analysis highlights the importance of not disclosing NBS results by voicemail or by telephone before a weekend or long holiday. Parents who received results in this manner universally reported disclosure as particularly anxiety-provoking. If parents were unavailable by telephone to discuss the results and a voicemail was necessary, parents often suggested that generic messages with requests for call-backs were favored over messages that referenced NBS results. Although these factors were mentioned by parents in the context of NBS results disclosure, they are likely relevant to PCP communication in results-disclosure encounters for other medical conditions or screening tests.

We acknowledge several limitations to this study. The retrospective parent interviews took place 3–5 months after initial results disclosure, which may have affected parental recall or influenced responses because of further PCP interactions or psychosocial issues unrelated to NBS. Also, parents may have responded to interview questions based on social desirability, potentially skewing the data toward favorable responses and opinions. Although both mothers and fathers were encouraged to participate, the majority of our study participants were mothers. This gender disproportion potentially limits the generalizability of the data. Though parents were informed that responses would not be shared with PCPs, it is possible that some parents felt uncomfortable reporting negative opinions. It may appear unremarkable that some of the parent-reported factors have low frequencies; however, some of these factors still met statistical significance for parental reactions. It should be noted that parents were not asked for an exhaustive list of *all* factors they found helpful or needed improvement. Rather, our interview questions targeted the *most* notable factors that were helpful or needed improvement during initial NBS results disclosure. Due to the qualitative nature of this analysis, not all parents commented on all factors.

4.2. Conclusion

Parents identified specific content messages, PCP traits, and aspects of the setting as factors that influenced their NBS results disclosure experience. Specific content messages provided by PCPs were most frequently identified by parents to influence their reactions. Parents appreciated when PCPs provided supplemental written information, avoided jargon, and personally disclosed NBS results. Parents reported negative reactions to receiving NBS results by voicemails or on Fridays. Future studies are needed to determine how PCPs should prioritize content messages during results disclosure and the impact of high quality communication techniques on parental outcomes.

4.3. Practice Implications

These data suggest that PCP communication during initial NBS carrier results disclosure can affect parental reactions, and parents identified several factors which may guide PCP communication of NBS carrier results to promote positive parental reactions. Though PCPs may refer patients to specialists for further counseling or follow-up, it is important for PCPs to be knowledgeable about the meaning of NBS carrier results and provide clear, concise content messages to parents during disclosure. We realize PCPs know their patients best and have preferences on how to explain these messages. The presentation and timing of content messages should be adapted to suit the informational needs of patients. PCPs may also consider reinforcing conversations with parents by providing written information. Additionally, PCPs should consider their own characteristics, as well as aspects of the disclosure setting as factors that may influence parental reactions and experiences. Initial NBS results disclosure can greatly impact parents; therefore, adequate PCP communication of carrier results with families is important.

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Table 1

Participant Demographics*

Variable	Total		CF		SCH	
	n	**(%)**	**n**	**(%)**	**n**	**(%)**
Infants’ carrier NBS result[†]	270		99		171	
Parent gender						
Female	264	(98)	96	(97)	168	(98)
Male	6	(2)	3	(3)	3	(2)
Parent race/ethnicity						
White	122	(45)	87	(88)	35	(21)
Black	118	(44)	7	(7)	111	(65)
Latino	8	(3)	1	(1)	7	(4)
Other	17	(6)	4	(4)	13	(8)
Not reported	5	(2)	0	(0)	5	(3)
Parent age (years)						
<20	36	(13)	7	(7)	29	(17)
20–29	134	(50)	51	(52)	83	(49)
30–39	87	(32)	40	(40)	47	(28)
40–49	9	(3)	1	(1)	8	(5)
Not reported	4	(2)	0	(0)	4	(2)

[*] “SCH” refers to parents of SCH carrier infants; “CF” refers to parents of CF carrier infants; percents are rounded to the nearest whole percent

[†] CF carrier infants in this analysis had an elevated immunoreactive trypsinogen (IRT), a single CF-associated mutation, and a normal sweat chloride test. SCH carrier infants in this analysis had the presence of Fetal, Adult, and Sickle Hemoglobin (“F-A-S”) on NBS.

Table 2

Specific content messages that influenced parents' NBS results disclosure experiences and reactions (Theme 1)[*]

Content Message	Total % (n)	CF % (n)	SCH % (n)	Quotation
Infant is (likely[§]) a carrier [†] [‡]	20% (53/270)	39% (39/99)	8% (14/171)	"[PCP] did good. He explained it all to me and reassured me that [infant] most likely did not have [disease] but was most likely a carrier." (mother, CF carrier infant) "I think that it would've been nice…to be told that there is a possibility that [infant] could just be a carrier." (mother, CF carrier infant)
Infant (likely[§]) does not have disease [†] [‡]	19% (50/270)	26% (26/99)	14% (24/171)	"The most helpful thing is that [PCP] told me… [infant] has the trait, she doesn't have the disease….because at first I was like does that mean she have [the disease]? And when [PCP] told me, 'no' that she doesn't have it, she just has the trait, and I felt a little better." (mother, SCH carrier infant) "I was so scared and terrified because [PCP] didn't really give me any information, odds are [infant] is not going to have this disease. She didn't tell me that. I walked away believing that [infant] probably had it and, obviously, we were just freaking out. That was pretty scary there." (mother, CF carrier infant)
NBS result should not worry parent [†]	16% (44/270)	24% (24/99)	12% (20/171)	"[PCP] kinda broke it down like what, what [the NBS result] was and not to worry you know about it and stuff, but it was just helpful because it lets you know right away and don't get surprised by it." (father, CF carrier infant)
Information on statistics, inheritance or family history [†]	15% (40/270)	13% (13/99)	16% (27/171)	"[PCP] gave me a lot of specific information about, um, if my husband and I were carriers what that would mean for [infant]…I think she was trying to put our minds at ease by saying what would be the most likely reaction, and she was giving us actual statistics …they helped us visualize what was going on by [PCP] having these statistics to give to us… just focus on what she said regarding the statistics and the likelihood of [infant] actually having cystic fibrosis." (mother, CF carrier infant)
Explanation or clarification of carrier status[‡]	13% (34/270)	11% (11/99)	13% (23/171)	"[PCP] just said, '[infant] has trait' and she left out of the room and I kind of thought that was like a new disease because I had never heard of trait." (mother, SCH carrier infant)
Infant is healthy and not sick [†]	11% (31/270)	3% (3/99)	16% (28/171)	"Well first of all [PCP] said, um, she reassured me that it's not a health problem, and she said it's just something that [infant] has that's a little different from the rest, but it's not something that poses a great risk to his health. So that was pretty reassuring right away." (mother, SCH carrier infant)
Information on sweat test process[‡]	11% (30/270)	30% (30/99)	n/a	"[PCP] explained the chloride sweat test to us, but maybe there could be more detailed information on actually how they perform it and, you know, because I think the biggest thing that went through my mind was I was scared that they would, that it would hurt her or it would, you know, involve lots of needles…but I was more concerned about having her tested, you know, would it hurt her, you know, how long would it take, you know, that kind of stuff." (mother, CF carrier infant)
Explanation of NBS process	6% (17/270)	7% (7/99)	6% (10/171)	"We didn't really know what the newborn screening was up until the point that we found out that there was something that came up, we didn't still fully understand everything that they were looking for…I mean [PCP] explained that they did it on all babies and it was, you know, just testing but we didn't really know the detail of it." (mother, CF carrier infant)
Explanation of disease (symptoms)	5% (14/270)	13% (13/99)	<1% (1/171)	"I really think that I should have had more information because I felt like, first of all, I didn't even know what cystic fibrosis was and [PCP] just said "okay, a mutation on his cystic fibrosis gene." That all sounds really scary…and then, um, really, I think, for me personally, more information would've been better as far as what cystic fibrosis is and I think that it would've been nice to know." (mother, CF carrier infant)

Content Message	**Total** **% (n)**	**CF** **% (n)**	**SCH** **% (n)**	**Quotation**
Information on next steps for parent and infant	4% (11/270)	5% (5/99)	4% (6/171)	“If you’re going to call someone and say that their child could have a serious illness, you know, they want to know the next step as well as what that means to us and our family.” (mother, CF carrier infant)
Difference between disease and carrier	4% (10/270)	5% (5/99)	3% (5/171)	“[PCP] just went ahead and explained [trait] to me so, you know, it made it more clear because I was kind of…you don’t really know the difference between the trait and the actual sickle cell [disease].” (mother, SCH carrier infant)
Likelihood carriers will have health complications	3% (7/270)	0% (0/99)	4% (7/171)	“Could [infant] get sick? I mean, could he ever, his blood…I mean, anything else could happen to him? Or anything? Yeah, that’s what I wanted to know [from PCP].” (mother, SCH carrier infant)

[*] The abbreviation SCH refers to parents of SCH carriers; the abbreviation CF refers to parents of CF carriers; percents are rounded to the nearest whole percent

[†] If content message present, statistically significant correlation with parents’ report of **positive** reactions ($p<0.05$)

[‡] If content message not present, statistically significant correlation with parents’ report of **negative** reactions ($p<0.05$)

[§] The word “likely” should be included for CF results *only* because the infant’s carrier status is probable but not definitive at the time of results disclosure

Table 3

PCP traits that influenced parents' NBS results disclosure experiences and reactions (Theme 2) [*]

Behaviors and Characteristics	Total	CF	SCH	Quotation
	% (n)	% (n)	% (n)	
Assessed parents understanding and answered questions	14% (37/270)	16% (16/99)	12% (21/171)	"So like our conversation is I would say is, is very good. [PCP] tell me everything if I have a question she asks me do I got a question or if I ask her a question she answers it and she makes you all understand it. I mean um, it be good." (mother, SCH carrier infant)
Repeated information and did not assume parent knowledgeable	7% (19/270)	6% (6/99)	8% (13/171)	"And I, because I have two older sons, I had heard, you know, heard it before, but…if [infant] was my only child…then, you know, of course I probably would've been more concerned, more nervous, more questions, but [PCP] explained it to me and she didn't assume that I knew these things already." (mother, SCH carrier infant)
Avoided Jargon[†] [‡]	6% (16/270)	6% (6/99)	6% (10/171)	"But um, [PCP] is super great about explaining you know in great detail you know in ways that I can understand so you know so I'm not a doctor so I don't understand all that doctor stuff, but ah, he was really great about explaining it to me." (mother, SCH carrier infant)
Knowledgeable and Thorough	11% (30/270)	11% (11/99)	11% (16/171)	"Well, [PCP]'s just – she's very well informed herself about it and very reassuring. You know, she just – she has excellent information. She's obviously extremely smart and well educated and…make(s) sure that any question you might ever have has been answered." (mother, SCH carrier infant)
Calm[†]	5% (14/270)	7% (7/99)	4% (7/171)	"[PCP] just remained calm through the whole, you know, time, that's just who she is so, she's very soft spoken and very calm …um, I just always felt reassured with her, even though it was scary news." (mother, CF carrier infant)

[*] The abbreviation SCH refers to parents of SCH carrier infants; the abbreviation CF refers to parents of CF carrier infants; percents are rounded to the nearest whole percent

[†] Statistically significant for parents' report of **positive** reactions ($p<0.05$)

[‡] Using Jargon is statistically significant for parents' report of **negative** reactions ($p<0.001$)

Table 4

Aspects of the setting that influenced parents' NBS results disclosure experiences and reactions (Theme 3) [*]

Setting	Total % (n)	CF % (n)	SCH % (n)	Quotation
Liked receiving results over the phone	4% (10/270)	4% (4/99)	4% (6/171)	"So I think maybe the phone call and the giving of information immediately was probably the best – at least for me – that was the best way to do it." (mother, CF carrier infant)
Disliked receiving results over the phone	3% (8/270)	6% (6/99)	1% (2/171)	"Just like I said, in the initial phone call when we found out that he had an abnormal screening, it was just high anxiety…we really didn't understand what was going on so it was just kind of hard that it all happened over the phone and I talked with [PCP] about it" (mother, CF carrier infant)
Liked receiving results from PCP	5% (14/270)	6% (6/99)	5% (8/171)	"Um, I liked the personal contact, that [PCP] herself called me, and it wasn't, it wasn't a nurse although I don't mind if a nurse calls me either but the fact that the doctor herself took the time to call, um, and made sure she spoke to me directly, um, and, you know, she was just, like I said, very matter of fact about it and reassuring and, um, she followed up with sending the printed material like she said she would." (mother, SCH carrier infant)
Liked receiving results in person	4% (12/270)	7% (7/99)	3% (5/171)	"I felt more comfortable by [PCP] talking to me about it face to face." (mother, SCH carrier infant)
Disliked receiving results on a Friday or before a holiday weekend	3% (7/270)	6% (6/99)	<1% (1/171)	"It was, um, well first of all it was just bad circumstances. It was the Friday before Labor Day weekend…and, well you know, it's Labor Day weekend and we couldn't even call to schedule an appointment until the following Tuesday, so we go through the entire holiday weekend completely miserable thinking that, you know, our poor infant is going to be, um, extremely ill for the rest of his life and not knowing, you know, the differences between, like I said, being a carrier and having the disease. And, um, you know, and then having to wait four days through a holiday weekend to even be able to call to schedule an appointment to find out if he had it – it just wasn't good…it was just unfortunate for us…it wasn't a good situation…It was a pretty miserable weekend." (mother, CF carrier infant)
Disliked receiving results by voice mail[‡]	2% (5/270)	4% (4/99)	<1% (1/171)	"[PCP] actually left me a voicemail saying that, "Some of the results came back…" I remember she used the word "irregular" I think and that I needed to call her back, and so that was the first time I heard anything about the newborn screening and the results, was on the voicemail…we played phone tag for probably a couple days, so without knowing what was irregular, you know, I'm at home with a brand new infant, extremely concerned….I'd say, uh, the first would be not leave a message…saying, you know, 'There's test results that came back' without elaborating on that at all. Of course as a new mom you think the worst right away." (mother, CF carrier infant)

[*] The abbreviation SCH refers to parents of SCH carrier infants; the abbreviation CF refers to parents of CF carrier infants; percents are rounded to the nearest whole percent

[‡] Statistically significant for parents' report of **negative** reactions (p<0.001)

Table 5

Positive and negative parental reactions resulting from NBS results disclosure experience *

Reactions	Total % (n)	CF % (n)	SCH % (n)	Quotation
Positive Emotional *Reassured, Relieved Comforted, Calm, Happy*	31% (84/270)	36% (36/99)	28% (48/171)	"And [PCP] just really comforted me and let me know and rest assured me, it's okay, and not to be concerned, and it was so meaningful, 'cuz that's how he is, and then when he gave me all the information… that was good too so, I just felt really reassured." (mother, SCH carrier infant)
Positive Cognitive *Understood result, Trusted provider*	8% (22/270)	7% (7/99)	9% (15/171)	"Um, well [PCP]…she broke it down into a picture and illustrated for me, which, um, I understood that better. You know, I got a better understanding." (mother, SCH carrier infant)
Negative Emotional *Scared, Anxious, Shocked, Surprised, Miserable, Upset Stressed, Frustrated*	26% (69/270)	48% (48/99)	12% (21/171)	"Um, it was kind of a – a scary, unsettling moment then. Um, because I knew that it was a genetic disorder, didn't know too much past that… it still left that uncertainty and – and worry in my mind right off the bat." (mother, CF carrier infant)
Negative Cognitive *Confused, Thought infant had disease, "My mind went blank"*	13% (35/270)	22% (22/99)	8% (13/171)	"You know when [PCP] said something about (the sweat test), she said, 'You should go do it' and then she started explaining about what it was and I…my head went blank, seriously. I was like, 'Oh, my God, something's going on. I got to go do this and oh, you know when should I do it?' and ah, honestly I didn't really even listen to her, you know. And then I'm like, 'Oh, my God, something could be wrong with my infant' …I didn't hear a thing." (mother, CF carrier infant)
Negative Physical *Crying, Racing heart, Sleepless nights, Sick*	4% (11/270)	10% (10/99)	<1% (1/171)	"I kind of lost it when [PCP] said cystic fibrosis and I guess I didn't understand what a carrier was at that time and I told my husband, I was pretty hysterical…[PCP] knows I'm a pretty sensitive person so I pretty much lost it on the phone. She was very understanding and was open to any questions that I had…" (mother, CF carrier infant)
Negative Social *Changed PCPs, No available social support*	2% (6/270)	5% (5/99)	<1% (1/171)	"Yes, so we've actually changed PCPs since then because of the situation that's happened from that [NBS] screening…I think, you know, the part that I feel really negative about the newborn screening, I think a lot of it was to do with my PCP herself and how it was handled." (mother, CF carrier infant)

* The abbreviation SCH refers to parents of SCH carrier infants; the abbreviation CF refers to parents of CF carrier infants; percents are rounded to the nearest whole percent