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Informing Parents about Positive Newborn Screen Results: Parents' Recommendations

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Abstract

This descriptive study examined parents' reactions to newborn screening (NBS) results and their recommendations for improving communication. Dimensional and content analyses were conducted on interviews with 203 parents of 106 infants having positive NBS results. Diagnostic results confirmed infants as having congenital hypothyroidism (n=37), cystic fibrosis (n=26), or being CF-carriers (n=43). Parents' reactions ranged from "very scary" to "not too concerned." Most reported feeling shocked, panic, and worry; some reported guilt. Parents in the CF and CF-carrier groups preferred face-to-face disclosure as the communication channel; whereas congenital hypothyroidism group parents supported telephone contacts. Parents recommended providers be well informed, honest, and calm; personalize disclosure, avoid jargon, listen carefully, encourage questions, recognize parental distress, offer realistic reassurance, pace amount and rate of information, assess parents' understanding, and refer to specialists. We conclude that provider-patient communication approach and channel can exacerbate or alleviate parents' negative reactions to positive NBS results.

Keywords

communication; newborn screening; cystic fibrosis; congenital hypothyroidism; content analysis

State-mandated newborn screening (NBS) detects neonates with serious or life-threatening conditions with the goal of preventing health complications via early intervention and treatment. Cystic fibrosis (CF) and congenital hypothyroidism are now included in all NBS panels (NIH website, 'Genetics Home Reference', 2011) throughout the United States (US). CF is the most common life-shortening autosomal recessive disease in the US (Cystic fibrosis foundation [CFF], 2011). This chronic, progressive, multisystem condition affects pulmonary, digestive, and reproductive systems. CF affects approximately 30,000 individuals nationwide with about 1,000 new cases diagnosed annually. Although early treatment is associated with improved nutritional health and pulmonary status for individuals with CF (Farrell, et al., 1997, 2001), the diagnosis is still associated with a reduced life expectancy of about 37 years (CFF, 2011). Congenital Hypothyroidism (hereafter referred to as hypothyroidism) is caused by diminished or absent thyroid function. Early treatment prevents mental retardation and promotes normal growth (Rastogi and LaFranchi, 2011). Approximately 3,000 to 4,000 infants are diagnosed with hypothyroidism annually in the US (NIH website, 'Genetics Home Reference', 2011).

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CF is relatively new to most NBS panels. NBS protocols using DNA analysis detect CF-carriers as well as infants with CF. Infants with CF have two symptom-causing mutations of their cystic fibrosis transmembrane conductance regulator (CFTR) genes. CF-carriers have only one mutation and develop no CF symptoms. Although there are no health benefits for detecting CF-carriers in infancy, they and their parents are at increased risk of having a child with CF. Regardless of whether NBS detects one or two mutations, a diagnostic sweat test is needed to confirm or rule-out the diagnosis. Thus, CF NBS is more complex than other screening procedures. By contrast, hypothyroidism has been included in NBS for about 40 years and does not involve genetic testing. Hypothyroidism is a prototype for NBS as it meets World Health Organization screening criteria (Wilson and Jungner, 1968). With treatment, the prognosis for hypothyroidism is favorable, whereas the impact of CF NBS and early diagnosis on longevity has not been well established. Positive NBS results for CF and hypothyroidism require follow-up diagnostic tests that are coordinated by primary care providers (PCPs). PCPs are typically responsible for informing families of these results (Dillard, et al., 2004), and there is no consensus guidelines for the initial conversation between PCPs and families.

Families of infants with abnormal NBS results for CF or hypothyroidism report feeling shocked and anxious about the unexpected results (Asplin, 2008; Beucher, et al., 2010; Jedlicka-Kohler, 1996). This reaction is partly due to a general lack of knowledge about NBS coupled with most infants being asymptomatic at the time of notification (Groose, et al., 2010; Tluczek, et al., 2005b, 2009). Families of infants with false-positive NBS for CF may also experience adverse psychosocial consequences attributed to confusion about test results and complexity of CF genetics (Tluczek, et al., 2009). Insufficient information leads to uncertainty and emotional distress in parents, which may be either alleviated or exacerbated by the methods used to communicate results to parents (Ciske, et al., 2001; La Pean and Farrell, 2005; Tluczek, et al., 2005b, 2010a).

Few studies have documented parents' perspectives about the best way to receive difficult medical news about their children. Due to the rarity of infants with CF or hypothyroidism in primary care practices, most PCPs report feeling unprepared and stressed when required to give difficult news to families (Harrison and Walling, 2010). A survey of general pediatricians and family practice physicians showed that although respondents thought that PCPs should be responsible for informing families about NBS results, about 10% of pediatricians and 33% of family practitioners reported feeling unprepared to discuss positive NBS results for either hypothyroidism or CF (Kemper, et al., 2005). Similarly, another study found that 54% of pediatricians did not feel comfortable talking about NBS results with families (Gennaccaro, et al., 2005). On the other hand, 54.5% of parents of infants with positive CF NBS results reported wanting more information when receiving results; 13.8% recommended that PCPs be better informed of the details and implications of positive NBS results (Ciske, et al., 2001). These studies emphasize the need for improved communication when informing parents of results.

Research has repeatedly identified problems with provider-parent communication about NBS results (Ciske, et al., 2001; Groose, et al., 2010; La Pean and Farrell, 2005; Lewis, 2006; Parsons, et al., 2003). Such communications are often associated with adverse psychosocial outcomes (Al-Jader, et al., 1990; Beucher, et al., 2010; Moran, et al., 2007; Tluczek, et al., 2005b). In a quality improvement project for CF NBS, parents' misunderstanding of genetic counseling information topped the list of 20 concerns; 5th was that PCPs lack knowledge about NBS results (Groose, et al., 2010). The purpose of this study was to address gaps in the empirical literature by obtaining parents' perspectives about how best to communicate positive NBS results. We wondered how parents' experience of the provider communication might vary based on the nature of the identified condition.

Therefore, we chose CF and hypothyroidism because these conditions differ in illness severity, prognosis, complexity of NBS procedures, and duration on NBS panels.

Methods

Design and Procedure

We used a subset of qualitative data obtained from interviews conducted from 2003 to 2008. Data were part of a larger, longitudinal mixed-method study examining the psychosocial consequences of genetic testing through NBS (Tluczek, et al., 2005a). Analyses of other data subsets have been published elsewhere (Tluczek, et al., 2009, 2010a, 2010b).

Recruitment and Consent

With approval from institutional review boards of all four participating medical centers, families of infants with abnormal NBS results for CF or were invited to participate in this study. Clinical staff used convenience sampling techniques that included giving or mailing invitational letters to parents of infants who met inclusion criteria. The consent procedure involved a discussion about the use of non-identifying quotes in publications and all participants provided written consent. The extensive statewide recruitment effort precluded documenting recruitment rates. Additional recruitment details can be found in previous publications (Tluczek, et al., 2010a, 2010c).

Sample

We analyzed 106 interview transcripts obtained from 203 parents of 106 infants less than 6 months old with positive NBS results. Infants were excluded if born before 32-weeks gestation or having perinatal complications or other serious health problems. Diagnostic test results confirmed infant status: hypothyroidism (n=37), CF-carrier (n=43), or CF (n=26). The CF group included five infants who had positive NBS identifying two CF mutations and normal or intermediate sweat test results. One infant with hypothyroidism had false-negative NBS results and was subsequently diagnosed due to family history, and another infant with hypothyroidism had borderline thyroid levels and was being monitored by a pediatric endocrinologist.

This sample included biological mothers (n=106) and fathers (n=97) who were predominantly of European ancestry (95%), married (80%), and college educated (45%) with mean ages of 29 and 32 years, respectively. Infants' gender was 57% female.

Data Collection

The principal investigator (PI) or specially trained assistants conducted 30 minute interviews that focused primarily on participants' experiences of NBS results disclosure. Data were collected in participants' homes when their infants were between 6 weeks and 6 months of age. Parents were interviewed together if both chose to participate. Analyses for this study specifically focused on parents' experience with the receipt of positive NBS results and their suggestions for improving the experience for other parents. Questions included: How were you informed about your infant's abnormal newborn screening result? Who informed you? Please describe your experience. What suggestions do you have for improving the NBS program? Additional, probing questions were used to encourage parents to expand their responses with greater detail and specific examples. One research team member transcribed audio-recorded interviews verbatim, omitting identifying information and another researcher crosschecked transcripts for accuracy.

Data Analysis

The two-part data analysis began with dimensional analysis (Bowers and Schatzman, 2009) followed by a directed and summative content analysis (Hsieh and Shannon, 2005; Krippendorff, 2004). Dimensional analysis involved line-by-line open coding of interview transcripts (Bowers and Schatzman, 2009). We abstracted transcript data related to parents' reported experiences of being informed about their infants' NBS results and their suggestions for improving related communication. We began with the most data rich interviews from the CF group, followed by CF-carrier and hypothyroidism groups, constantly comparing parent responses within and across study groups. Data were labeled with in vivo codes, e.g., using parents' words. When thematic saturation was reached, we developed a codebook by clustering codes with similar themes into categories and including quotes as exemplars. Throughout the coding process, categories were refined to maintain descriptive, interpretive, theoretical, and evaluative validity (Maxwell, 1992). Then, two coders attained 90% reliability in using the codebook as a guide to conduct a directed and summative content analysis of the entire data set (Krippendorff, 2004). Additional reliability checks were systematically performed on 20% of transcripts throughout the analysis. We also compared results across study groups to see if family reports differed by group membership. Frequencies were tabulated across each category and study group. Finally, we summed responses within categories and across groups.

Results

Parents' Reactions

Although parents reported a variety of reactions to the receipt of positive NBS results, their responses could be classified into the two categories of "very scary" with strong negative reactions or "not too concerned" which were characteristically less intense negative reactions. No parents reported a lack of concern. Table 1 shows the frequency of individual parent reactions by group. The majority of parents across all groups reported strong initial emotional reactions such as shock, panic, and anxiety about what results meant. Worry was common in parents in all groups, although reasons for worry varied, e.g., mental retardation associated with hypothyroidism vs. mortality associated with CF. Another response identified in all groups involved parents' report of guilt about their children having the conditions or being a CF-carrier. Responses unique to positive CF NBS results included fears of their infants dying, parental somatic symptoms, e.g., feeling "sick" and sensations of "suffocation," difficulty bonding with their infant, marital discord, and changed reproductive plans.

Across all study groups, there were a few parents who were not as surprised by the positive NBS results and consequently responded less emotionally. Reported reasons for their attenuated reactions included lack of family history of the conditions, having another child or family member with the condition who was doing well, or having an older child who also had positive NBS results for CF and a subsequent normal sweat test. Other parents attributed their relatively calm reactions to the calm and reassuring way their providers communicated results to them. One hypothyroidism group mother said, "My doctor said that... it wasn't too concerning as long as we got him on meds right away."

Parent-Provider Communication

Parent suggestions for ways to improve the NBS experience were classified into two categories. Those associated with parent-provider communication included provider characteristics and approach to informing parents (Table 2). Those associated with the NBS notification process included timing of notification, communication channel, care coordination, information, and family support (Table 3). Parents within couples tended to

share the same experience of and recommendations about parent-provider communication; therefore, data in Tables 2 and 3 represent the number of families rather than individuals by study group.

Provider Characteristics and Approach

Families wanted providers to be well informed about the implication of NBS results and able to provide additional information upon request. Although families appreciated knowing the PCP who notified them of results, some were willing to forego familiarity for knowledge. Of the twelve families who preferred known providers, all had previously established favorable relationships with their PCPs. A hypothyroidism group family described a dramatic illustration of the adverse psychological consequences of NBS results being conveyed by an individual without knowledge of the condition or meaning of test results. The mother explained, "The hospital... was trying to get a hold of us and they couldn't. So they actually had a (police) officer come over to our house and tell us. That was scary." Their distress was amplified by the informant's inability to offer them information about implications of a hypothyroidism diagnosis.

Families recommended that PCPs personalize the disclosure by taking time with parents to honestly and calmly discuss the meaning and implications of results using simple language. They suggested carefully listening to parent concerns, encouraging, and answering their questions. They advised PCPs to be sensitive to the distress parents are likely to experience; offer realistic reassurance and hope, e.g., the benefits of early detection. They cautioned PCPs not to overwhelm parents; to pace the amount and rate of information delivery. Finally, parents urged PCPs to make sure parents understand information discussed. The approaches to informing parents about their infants' NBS results had a profound effect on families' subsequent relationship with that provider. When dissatisfied, several families reportedly switched providers.

Timing of Notification

Most recommended that PCPs notify parents of positive NBS results as soon as possible. However, some advised against notifying parents on a Friday afternoon or a weekend because it left parents wondering and worrying about results without access to specialists who could answer their questions. Some said they would have preferred not to receive the NBS results until the diagnosis was confirmed. A few CF and CF-carrier families described waiting one to several weeks for the sweat test appointment, causing them to, as one parent said, "sit and wait and worry." Five families reported even longer delays because insufficient quantities of sweat obtained from the first sweat test necessitated scheduling a second sweat test, thus delaying their receipt of diagnostic findings. Less common suggestions included waiting until infants are out of neonatal intensive care units (hypothyroidism group), infants are older (CF group), or until parents can obtain life insurance for the infant (CF group).

Communication Channel

Twenty-four families described the communication channel they preferred or would have preferred regarding notification of positive NBS results. Many parents, especially those in the CF and CF-carrier groups, preferred receiving positive NBS results in person. More than half of those who preferred being notified by telephone were in the hypothyroidism group. Only one CF group family preferred telephone notification. Families in the CF-carrier group were fairly evenly divided in their preferences for or against receiving a letter describing NBS results. Additionally, families suggested that providers not leave messages about positive NBS results on their answering machines. One CF group family suggested that

when informing parents by phone, PCPs might offer parents opportunities for further discussion in the clinic soon thereafter.

Care Coordination

Families wanted to be connected with pulmonary or endocrine specialists as soon as possible. Within 24 hours of being informed about infants' NBS results, parents in the hypothyroidism group typically received a telephone call from an advance practice nurse in a pediatric endocrinology clinic. This rapid referral and subsequent telephone contact with families in the hypothyroidism group seemed to be motivated by the need to promptly initiate thyroid replacement therapy and to assure proper administration of medication by parents. However, this timely connection with a specialist had the added benefit of addressing families' concerns, thereby alleviating parental anxiety. Although CF and CF-carrier group families were also quickly referred for diagnostic sweat testing, they did not actually meet or speak with a specialist, usually a genetic counselor, until that appointment which could be days to weeks later. Families also described the importance of effective communication between PCPs and specialists, assistance in scheduling diagnostic appointments, referral information reaching specialty clinics, and referrals to mental health specialists when indicated.

Family Support

Families from all groups wanted to meet other parents who had children with the same NBS results. One CF group family explained that such encounters engendered feelings of hopefulness. Similarly, some families wished to meet individuals with the same condition as their children. CF group families also wanted access to support groups. Several parents in the CF and CF-carrier groups described their appreciation for the emotional support they received from CF specialists. Finally, families also identified a need for written materials regarding their infants' NBS results, access to additional information, and preparation for subsequent diagnostic procedures.

Discussion

These findings echo results of research in other clinical contexts by underscoring the importance of effective parent-provider communication (Avis Reardon, 2008; Midson and Carter 2010). This report provides clinicians and researchers additional documentation about parents' reactions to positive NBS results and practical suggestions for improving related communication. Many families described strong emotional reactions to their infants' positive NBS results. Some preferences, e.g., how to inform and informant characteristics, were universally described by families in all subgroups while other preferences, e.g., communication channel and timing of notification, were unique to the condition found to be abnormal through NBS. The CF and CF-carrier group parents appeared to be more likely to have strong negative reactions to the results than hypothyroidism group parents, suggesting that the prognosis and availability of effective treatment for the condition may influence parents' reaction to positive NBS results. Additionally, 88% of CF-carrier group parents recalled strong negative reactions to the receipt of positive CF NBS results similar to the reactions of parents of infants diagnosed with CF. This observation suggests that the CF-carrier group parents' retrospective recollection of the NBS experience was not altered by their subsequent knowledge that their infants did not have CF.

Group differences also emerged in families' preferences about communication channel, with more hypothyroidism families preferring telephone notification than the CF or CF-carrier groups. These differences may be due to the perceived characteristics of the disorder. Parents might prefer to have a face-to-face discussion about a life-shortening condition, like

CF, whereas the promptness of telephone contacts facilitates initiation of treatment critical to preventing cognitive impairment in infants with hypothyroidism. Group preferences could also reflect parents' favorable contacts with specialists. Hypothyroidism group families often receive telephone calls from advanced practice endocrine nurses, while the CF and CF-carrier group families meet face-to-face with genetic counselors. Parents tended to view these encounters as opportunities to talk with knowledgeable and credible sources of information.

Our results are consistent with and complementary to another project that was conducted in parallel with data collected from parents of infants born in subsequent years (Collins, et al., submitted manuscript). Although that study used similar methods to ours and was also conducted in Wisconsin, the Collins study included families whose infants had false-positive NBS results for CF or sickle cell anemia while our study included infants with true-positive results for CF and hypothyroidism, as well as false-positive results for CF. These studies independently drew similar conclusions about the pivotal role of PCP communication in ameliorating or exacerbating parental distress in response to positive NBS results across study populations. Both studies identified similar parental preferences and emotional reactions contingent upon the quality of provider communication. Findings unique to our study include parental reactions of guilt, difficulty bonding with infant, marital discord, and changed reproductive plans.

These findings suggest that PCPs are crucial to addressing parents' immediate needs for information and reassurance upon notification of positive NBS results. By becoming knowledgeable about their state's NBS program, using effective and empathic communication, and facilitating prompt referrals to specialists, PCPs may reduce the initial parental distress associated with positive NBS results. These strategies may be especially important to parents in the CF and CF-carrier groups because they tend to have relatively longer waiting periods between the time of notification about NBS results and their appointments with CF experts. The CF Foundation's new web page (<http://www.cff.org/AboutCF/Testing/NewbornScreening/>), which describes NBS for CF, may also serve as a valuable informational resource for PCPs as well as families.

Our findings suggest that personalizing the disclosure process to meet individual family needs may increase parents' confidence in providers and enhance parent-provider relationships. Given the high emotional distress reported by parents of infants with various positive NBS results (Beucher, et al., 2010; Hewlett and Waisbren, 2006; Tluczek, et al., 2005b), follow-up telephone practices, similar to those used with the hypothyroidism population, may benefit families with positive NBS results for other conditions. For example, CF specialists, e.g. genetic counselors or advanced practice nurses, might contact families prior to the diagnostic sweat test appointment to answer questions, explain sweat test procedures, and provide additional information about test results.

Limitations of this study include the sample's limited racial/ethnic and educational diversity and small study subgroups. Convenience sampling could have introduced selection bias. Although the subgroups in this study reflect a continuum of severity, they involved only two of the 48 conditions on the NBS panel in Wisconsin (Wisconsin State Laboratory of Hygiene, 2011). Future research is needed to document the degree to which these results can be generalized to other populations affected by positive NBS results.

Conclusions

Although positive NBS results are likely to engender parental concern, the quality of informants' communications with families can have a powerful affect in exacerbating or

alleviating parents' negative reactions arising from receipt of positive NBS results. The preferred communication channel may depend on the nature and severity of the condition. Sensitive and effective communication with parents about positive NBS results can have the added benefit of fostering mutually gratifying parent-provider relationships.

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Abbreviations

CH	congenital hypothyroidism
CF	cystic fibrosis
CF-c	cystic fibrosis carrier
CFF	cystic fibrosis foundation
DNA	deoxyribonucleic acid
NBS	newborn screening
PCP	primary care provider
PI	principal investigator
US	United States

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Table 1

Parents' Reactions to Abnormal NBS Results % (n)

Initial Reactions & Parent Statements	Study Group (N=96)		
	CF	CF-c	CH
	N=24	N=38	N=34
“Very scary”			
Emotional distress He [primary care provider] said CF and I just lost it. I was, just crying and really, really upset. The sleep deprivation and the hormonal changes of just having a baby and ... the stress of having a newborn is overwhelming in itself and then to have something like that on top of it, I couldn't cope. I just...couldn't cope with it all. (CF-c group mother)	70.8 (17)	71.1 (27)	67.6 (23)
Stunned/Shocked I was in shock. I just wanted to know how serious it was. (CF group mother and father)	45.8 (11)	26.3 (10)	14.7 (5)
Fear of child dying Oh God, something's wrong. He's going to die. Oh my gosh, you know, I was crying. I called [husband] up right away on his cell phone and told him, get home now to go to the doctor's office with me and I was already in tears before we got there. I just could not stop crying, because I was just terrified of what I was going to hear. (CF-c group mother)	16.6 (4)	7.9 (3)	-
Guilt I was worried. I feel guilty, as if it was my fault. I was really upset. (CH group mother)	-	10.5 (4)	5.9 (2)
“Rollercoaster” It was like a rollercoaster ride trying to figure out what it meant. (CF group mother)	12.5 (3)	-	2.9 (1)
“Not too concerned”			
I wasn't real concerned... only because they did say that... if it comes back abnormal, it's abnormal. You go for the screening and knowing that we didn't have any family history at all was...something else... Neither one of us has any of the cystic fibrosis on either side. (CF-c group mother)	4.2 (1)	18.4 (7)	20.6 (7)

Note. NBS = newborn screening; CF = cystic fibrosis; CF-c = cystic fibrosis carrier; CH = congenital hypothyroidism.

N represents number of parents.

* Some parents identified more than one reaction.

Table 2

Family Suggestions for Improving Parent-Provider Communication % (n)

		Study Group (N=106)			
		CF	CF-c	CH	
Provider Characteristics (N=57)*		N=26	N=43	N=37	
Knowledgeable		87.7 (50)	42.3 (11)	41.9 (18)	56.8 (21)
Having [the PCP] be more informed, having more information about how to explain the genetics, how it works, the possibilities of your child actually having this disease, whereas just being a carrier. I think if our doctor had been prepared and been able to show us that, I think that would have probably, when we walked out of that doctor's office, we would have felt a lot more calm. (CF-c group mother)					
Known to family		21.1 (12)	11.5 (3)	4.7 (2)	18.9 (7)
I think having our pediatrician be the one that gave us the messages was a good idea. But, then again, we have a really good relationship with our pediatrician. (CF group father)					
Effective communicator		17.5 (10)	7.7 (2)	7.0 (3)	13.5 (5)
You've got to really listen to parents because they're sleep deprived and tired and probably dealing with kinds of healing issues. (CH group mother) I thought the experience with [PCP] was just really nice to be able sit down with us and could ask her questions and she really explained things at our level and let us ask questions so we would understand... I want someone to level with me and explain to me I mean not just give me a number but tell me what the number means. (CF-c group mother)					
Provider Approach (N=66)*					
Be calm and reassuring		54.6 (36)	23.1 (6)	27.9 (12)	48.6 (18)
When my doctor was telling this to me, my doctor didn't make me feel like he wasn't going to have it... like the tone and the way that he brought it to me, it was like here. You know, deal with it. It wasn't very... calming and comforting (CF-c group mother)					
Answer and encourage questions		33.3 (22)	23.1 (6)	20.9 (9)	18.9 (7)
They answered more than I could have even asked. (CH group mother)					
Make sure parents understand		24.2 (16)	11.5 (3)	13.0 (6)	18.9 (7)
So he [the doctor] really went out of his way to make sure... that we understood it. It meant a lot that he thought it was important enough to do it in person. (CF group mother)					
Don't downplay results, be honest		19.7 (13)	19.2 (5)	11.6 (5)	8.1 (3)
Telling us not to worry about it was like telling me not to breathe... It's impossible not to worry about it (CF-c group mother)					
Be sensitive, not harsh		16.7 (11)	11.5 (3)	4.7 (3)	13.5 (5)
The way she explained it to me was, "Her brain is screaming for help. And ... then I freaked out. (CH group mother)					
Use simple language, don't overwhelm		15.2 (10)	19.2 (5)	4.7 (2)	8.1 (3)
It was kind of like going back to school. I was trying hard not to fall asleep. I was like, ahh, just so much information tossed at you at one time, you know, you're brain is a sponge, but it can only hold so much. (CF group mother)					
Speak to both mother and father		12.1 (8)	15.4 (4)	2.3 (1)	8.1 (3)
[The pediatrician] called us up in the afternoon and talked it over with me. And then... he talked to (my husband). To make sure that we both got it from him as opposed to getting it passed on from me to (my husband). Just, because you know, things get lost in translation. Especially when emotions get involved. (CF group mother)					
Spend time, don't rush		12.1 (8)	15.4 (4)	9.3 (4)	-
[The doctor] spent a lifetime with us, and... it was wonderful. I felt that... we were humans not another number coming through and ok let's go, but that he actually cared about us, our family. (CF group father)					
Personalize information		7.6 (5)	3.8 (1)	2.3 (1)	8.1 (3)
Don't make it like a job, make it... more personal... if you're dealing with babies and parents that are concerned, worried, excited; all kinds of feelings are going on there (CF-c group father)					
Listen to parents		3.0 (2)	-	-	5.4 (2)
For the pediatricians I would say, you've got to really listen to parents because I think even though they're sleep deprived and tired and... probably dealing with ... healing issues, we					

	Study Group (N=106)		
	CF	CF-c	CH
Provider Characteristics (N=57)*	N=26	N=43	N=37
knew when he became a dramatically different child. And the first time they didn't listen and the second time we didn't give them a choice. (CH group mother)			
Match pace of discourse She [PCP] spoke too fast, and too in circles... she talked herself into confusion because by the time she was finished talking to us, we were under the impression that he wasn't coming home at all. (CF-c group mother)	1.5 (1)	-	2.3 (1)
Don't have others in the room when notifying It was hard too because the doctor had a medical student with him... I just, I felt a little, I don't know if I would say 'uncomfortable'? We were uncomfortable... uncomfortable getting, receiving news like that from a doctor and then a medical student being in there... That was kind of weird, you know. (CF group mother and father)	1.5% (1)	3.8 (1)	-
Assure quality health care for infant Instead of reassuring us that ... all of [baby's] symptoms were leading up to CF [and saying] I'm not really surprised that it's CF, maybe she could've been more, 'this is one of the best facilities...this is where your daughter is able to get the best health care possible for the sickness that [she] has (CF group mother)	1.5 (1)	3.8 (1)	-
Address the parents' feelings [The diagnosis] just hits like lightning... and that she immediately addressed my feelings, 'cause I knew she could tell on the phone that I was getting upset about it. (CH group mother)	1.5 (1)	-	2.7 (1)

Note. CF = cystic fibrosis; CF-c = cystic fibrosis carrier; CH = congenital hypothyroidism.

N represents number of families.

* Some families identified more than one suggestion

Table 3

Family Suggestions for Improving NBS Notification Process % (n)

		Study Group (N=106)		
		CF	CF-c	CH
Timing of Notification	34 (37)*	N=26	N=43	N=37
As soon as possible	67.6 (25)	15.4 (4)	20.9 (9)	32.4 (12)
I'm glad that we found out right away instead of later. (CH group mother)				
Not on a Friday afternoon	24.3 (9)	3.8 (1)	16.3 (7)	2.7 (1)
...instead of a phone call on a Friday afternoon saying that your kid has to go in for a cystic fibrosis test. That's not a very good way to start a weekend off and leave you unarmed in terms of information. (CF-c group father)				
When diagnosis can be confirmed	18.9 (7)	3.8 (1)	11.6 (5)	2.7 (1)
I wish I wouldn't have known until we were able to actually do the sweat test. Because it's like you sit for those weeks and that's all you think about. (CF-c group mother)				
Other	8.1 (3)	7.7 (2)	-	2.7 (1)
Communication Channel	23 (24)*			
In person	70.8 (17)	23.1 (6)	23.3 (10)	2.7 (1)
It's just personable, meeting one on one with somebody is much better then over the phone, especially when you're thinking, "Oh my gosh, my child might have cystic fibrosis." It's rather difficult. It's nice to meet with them one on one. (CF-c group mother)				
Phone call	29.2 (7)	3.8 (1)	4.7 (2)	10.8 (4)
[PCP] called and said that, "We've just receive a report from [infant's] newborn screening that one of her levels was high and so you just need to take her in for a blood, for a routine blood sample." Then she elaborated about the hypothyroid. So I think she handled it fine. (CH group mother)				
Not on answering machine	12.5 (3)	-	7.0 (3)	-
It would have been better if he had just tried to call back, you know, rather than leave us a message. (CF-c group mother)				
Send letter	8.3 (2)	-	4.7 (2)	-
A letter that let us know that this was just the next step... [would have been more reassuring]. (CF-c group father)				
Do not send letter	8.3 (2)	-	4.7 (2)	-
The doctor sent us a letter. (mother) It scared us. (father) We didn't really know. So immediately, we went on the Internet to look it up... And, it made us actually, it scared us even more... (mother) We would have wanted to be informed... and not just a letter. (father, CF-c group)				
Care Coordination	30 (32)*			
Refer to specialist right away	100.0 (32)	26.9 (7)	32.6 (14)	29.7 (11)
I wish we had much easier access to a specialist, within a week or two, instead of four weeks. (CH group father)				
PCP-specialist communication	15.6 (5)	7.7 (2)	-	8.1 (3)
[PCP] was really nice. Got the entire appointment and everything set up. All we had to do was go. (CF-group mother)				
Other	9.4 (3)	7.7 (2)	2.3 (1)	-
Information	30 (32)*			
Written materials	56.3 (18)	26.9 (7)	13.0 (6)	13.5 (5)
She [specialty nurse] just went through everything. She verbally told us everything and then she gave us a pamphlet and some articles she thought we would find useful... she said, 'you don't have to read everything, I have highlighted what you'll find useful.' She was very, very wonderful. (CH group mother)				
Follow up process	34.4 (11)	3.8 (1)	11.6 (5)	13.5 (5)

		Study Group (N=106)		
		CF	CF-c	CH
Timing of Notification	34 (37) *	N=26	N=43	N=37
She [specialty nurse] just told us everything that would go on at the appointment, what we could expect... She answered any questions that I had and she really put me at ease that he was going to be ok. (CH-group mother)				
Access to additional information	31.3 (10)	3.8 (1)	9.3 (4)	13.5 (5)
When you hear something like that initially that there's something wrong with your child, you're not in the right frame of mind. All these things run through your head and you may not be even paying attention to what people are saying to you. If there was a website that they gave you and said, "okay, if you want more information on this you can look up on the website here and you can get information on your own. (CH group father)				
Family Support	16 (17) *			
Other families	82.4 (14)	38.5 (10)	2.3 (1)	8.1 (3)
We had one of the fathers from a CH kid call us and say, 'Hey, I'm so-and-so. Just wanted to let you know that you're son is going to be just fine...' He had told us that his son was eight years old and he was playing soccer. (father) Which at that point was a breath of fresh air. (CH group mother)				
Individuals with the condition	35.3 (6)	15.4 (4)	-	5.4 (2)
Seeing or knowing somebody else that has it [CF] that's an adult gave me hope. (CF-group father)				
Providers	53.0 (9)	7.7 (5)	9.3 (4)	-
[Genetic counselor] was so personable. We could call her with any questions that we had or concerns...So that one on one, I guess, extra special treatment that we received. (CF-c group mother)				
Support groups	11.8 (2)	7.7 (2)	-	-
More people talking about it and saying what they've gone through in their life. As far as how their experiences have been. Cause, there's this and that statistic but what good are statistics? Just more of what people have experienced. Like how their life has it [CF] affected their life? (CF-group father)				

Note. NBS = newborn screening; CF = cystic fibrosis; CF-c = cystic fibrosis carrier; CH = congenital hypothyroidism; PCP = primary care provider.

N represents is the number of families.

* Some families identified more than one suggestion.