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Modified Uncertainty Theory and Parents' Perspectives about Equivocal Diagnostic Results for Cystic Fibrosis

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Abstract

A grounded, dimensional analysis described the experiences of five couples whose infants had equivocal diagnostic test results following positive genetic newborn screens for cystic fibrosis. We analyzed interview data collected at two times during each infant's first year. Uncertainty emerged as the central thematic dimension. Results showed that parents passed through a series of stages similar to the process described by Mishel's Uncertainty in Illness Theory (UIT), thus extending the application of the theory to circumstances in which the very presence of an illness is uncertain. Findings informed a modified version of the UIT comprised of five domains: stimuli frame, degree of uncertainty, opportunity-danger continuum, affective responses, and coping. This model incorporated Morse's conception of suffering. Three contextual domains influenced parents' experiences at various junctures along the uncertainty trajectory: individual characteristics, structure providers, and time. We discussed implications of the model for future research and clinical practice relative to genetic testing.

Keywords

cystic fibrosis; dimensional analysis; genetics; psychosocial issues; suffering; uncertainty

Rapid advances in genomic research and biotechnology have given rise to widespread genetic screening for inheritable conditions. Early detection permits prompt treatment and improves clinical outcomes. However, the increasing capacity of technology to detect previously unknown genetic mutations with undocumented clinical courses also introduces uncertainty and unanticipated psychosocial consequences. In this report, we examine parents' perspectives about equivocal diagnostic results for cystic fibrosis (CF); use the findings to build on an existing conceptual framework; and discuss the implications for practice and research.

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Psychosocial Consequences of Uncertainty in Genetic Testing

The reduction of uncertainty has been identified as a major reason for pursuing predictive genetic testing (Hutson, 2003). However, even unequivocal results of such testing have been associated with some degree of uncertainty and adverse psychosocial consequences. In a recent review, Tibben (2007) summarized the research on the psychosocial consequence of predictive testing of adults for Huntington's disease (HD), suggesting both negative and positive genetic screening results were associated with emotional distress. HD carriers, at risk for developing this debilitating neurodegenerative condition, reportedly struggled with psychological maladjustment, relationship problems, and guilt for potentially passing the defective gene to their children. O'Neill and colleagues (2006) found that uncertainty played a key role in predicting emotional distress following uninformative results for breast cancer genetic mutations BRCA-1 and 2. Women with a low tolerance for uncertainty were more likely to report coping difficulties after testing than women with a higher tolerance. Other research described uncertainty as a valuable tool for maintaining a sense of optimism. Whitmarsh, Davis, Skinner, and Bailey (2007) reported that parents of children with positive genetic test results for Klinefelter, Turner or Fragile X syndromes, conditions associated with developmental disabilities, created a "space for uncertainty" in their representations of the diagnoses. Uncertainty opened up the possibility for better outcomes than those projected by providers or documented in the literature. Thus, uncertainty has been recognized as a fundamental construct in the study of patients' responses to and coping with genetic testing.

Genetic testing in newborn screening (NBS) differs from other predictive genetic testing in several ways. In the aforementioned genetic tests, patients or parents of the patients actively choose to undergo the testing and they do so only after receiving extensive counseling about the implications of possible results. By contrast, NBS is mandated by state law and parents of infants receiving the test typically know little about the testing process unless the infant's results are abnormal and parents are notified accordingly (Thuczek, Orland, Nick, & Brown, in press). Consequently, the receipt of abnormal NBS results is both unexpected and unsolicited, giving rise to a high degree of parental distress and uncertainty (Thuczek, Kosciak, Farrell, & Rock, 2005; Thuczek, et al. 2006). Finally, this testing method can detect genetic variants of unknown clinical significance, thereby compounding the uncertainty of results.

Newborn Screening for and Diagnosis of Cystic Fibrosis

Cystic fibrosis (CF) is a life-shortening autosomal recessive disorder that has recently been added to state-mandated NBS programs and implemented in every state as of December 2009 (National Newborn Screening and Genetics Resource Center [NNSGRC], 2009). In Wisconsin, CF is one of 48 conditions screened from a blood specimen obtained from a heel prick performed on all infants during the first two days of life. The two step analysis, performed at a centralized state laboratory, begins by measuring immunoreactive trypsinogen (IRT). If the IRT is elevated, the specimen is analyzed for the 23 most common CF mutations. If one or two mutations are detected, the parents are notified about the abnormal results and the need for another diagnostic procedure, a sweat test, to measure the infant's sweat chloride. Results of the sweat test are classified as (a) positive - a CF diagnosis is probable, (b) intermediate - CF diagnosis is possible, or (c) negative - CF is very unlikely (Farrell et al., 2008). The wait between first notification of abnormal NBS and diagnostic sweat testing ranges from a couple of days to several weeks.

CF is a prime example of how the ever-increasing capacity to screen for and detect genetic mutations has transformed our understanding of a condition. Test results, once delivered as a dichotomy (presence or absence of CF), are now interpreted on a spectrum with variable clinical presentations ranging from asymptomatic to serious. Patients with positive diagnostic results

typically develop chronic lung infections and gastrointestinal malabsorption. Interventions focus on optimizing nutrition and minimizing lung damage but there is no cure (Accurso, 2008; Davies, Alton, & Bush, 2007). The average age of survival is currently about 37 years (Cystic Fibrosis Foundation [CFF], 2008; Ratjen, 2008). When findings fall within the intermediate range the results are equivocal leaving parents and providers with a high degree of uncertainty about if or when the infant will develop symptoms of this condition and the implications for the child's longevity are unknown.

Parental Response to CF Diagnosis

Not surprisingly, research has shown that parents typically respond to their child's CF diagnosis with feelings of shock, depression, stress, and a profound sense of grief associated with the news of their child's CF diagnosis (Duff & Brownlee, 2008; Jedlicka-Köhler, Götz, & Eichler, 1996; Quittner, DiGirolamo, Michel, & Eigen, 1992). However, these reports predate the availability of NBS for CF and represent parents with children diagnosed by the presence of classic CF symptoms or sibling with CF, rather than by genetic testing. Although a diagnosis of CF is devastating to parents under any circumstances, for parents of ill children, it often validates their concerns about their child's health and offers them hopeful direction for treatment. In the context of NBS, Grob (2008) describes a shift in parents' perception about their seemingly healthy infants following the CF diagnosis. In the absence of symptoms, parents struggle to re-define their child from "healthy" to "sick." These findings are based on parents whose children had a definitive diagnosis of CF. To our knowledge, there are no reports describing the subjective experiences of parents of healthy infants who fall within the intermediate diagnostic category with equivocal results and uncertain health implications.

Purpose of Study

The purpose of this study was to examine the psychosocial consequences of the NBS process when it produces ambiguous or contradictory genetic information inconsistent with the clinical picture. Our intent was also to use the results to inform the development of a conceptual framework related to parents' experiences. Finally, we wanted to identify implications for clinical practice and future research.

Methods

Design

Grounded dimensional analysis procedures (Bowers & Schatzman, 2009) guided this secondary analysis conducted on interview data obtained from a subset of five couples who participated in a larger, mixed method, longitudinal project designed to examine the psychosocial consequences of applying genetic technology to NBS. The larger project spanned four medical centers in different parts of the state. Clinical staff at all four participating sites used convenience sampling methods to recruit parents during regularly scheduled clinic appointments following diagnostic sweat testing. Parents received verbal as well as written explanations about the study, had their questions answered, and then provided written consent to take part in the research. The Institutional Review Boards from all four participating medical centers approved this project.

Sample

The quantitative design component of the larger study precluded using the theoretical sampling of traditional grounded theory research. Therefore, we used a selective sampling method similar to that described by Draucker, Martsof, Ross and Rusk (2007) in their qualitative work. We identified interview data from three couples in the larger study whose infants' medical records documented abnormal NBS results plus two CF gene mutations and a normal or

intermediate sweat test. We began the analysis with the most data-rich interview. The first three interviews showed remarkably similar results and the analysis could have been considered saturated. To enhance our theoretical sensitivity, we chose to search for exceptions and found two couples whose infants had similar test results, but in which parents reported different perspectives based on divergent contextual factors. The final sample included 10 parents of five infants. All couples were married and of European American ethnicity. The mean age was 35.5 years with a range of 23 to 59. Education levels included high school (n=2), community college (n=2), baccalaureate degree (n=3), and graduate degree (n=3).

Data

Data for this study consisted of de-identified transcriptions of audio-taped interviews from the original study that were conducted with couples in their homes first when infants were 2 to 6 months old, and later at 12 months of age. Each interview included both members of the couple at the same time. We opted to analyze interview data from both data points for all five couples to see if parents' perspectives changed over time. Technical problems rendered one of the 12-month audiotapes inaudible. Therefore, the final data set for this study consisted of nine interviews (five when infants were two to six months old and four when infants were 12 months old). Interviews in the original study used a set of open-ended questions developed from selective coding of an earlier grounded theory study (Tluczek et al., 2005) and covered a wide range of psychosocial issues related to NBS. The authors of the current study were also involved in the original study.

The current study focused on portions of the interviews that pertained to parents' perspectives about their infants' ambiguous diagnostic findings. Data obtained from the following questions formed the basis for the current study: How did you learn about your infant's NBS results? What did you think the results meant? What did you know about CF at that time? What is your understanding of the sweat test results? What has it been like for you emotionally? How do you envision your child's future? What suggestions do you have for improving the NBS process?

Data Analysis

The analytic process followed the procedures advocated by Strauss and Corbin (1990, 1994) and Corbin and Strauss (2008). The research team performed open coding using line-by-line constant comparative analysis of transcribed data. Data were reduced to categories, related properties, dimensional profiles, and patterned relationships of categories; all of which were labeled and stored in a Word document until data were saturated and no new themes were found. Axial coding facilitated conceptualization of relationships among categories, contexts, intervening conditions, and strategies. For example, parents' prior knowledge about CF was an important context that affected their initial reactions to the news about the NBS results. Selective coding identified the central phenomenon of *uncertainty* that unified the conceptual framework and the model was refined. Throughout, memos recorded the analytic process and category refinement. We also incorporated the dimensional analytic techniques of "conjuring" up dimensions from the data, determining the relevance of dimension, and making inferences about the relationships among dimensions (Bowers & Schatzman, 2009). Theoretical notes documented the linkage among categories, time, and strategies to merge the constructs into a unified conceptual framework.

Trustworthiness and Validity of Data

During the original study, we maintained descriptive validity (Maxwell, 1992) by audiotaping the interviews, transcribing them verbatim, and having another team member check the transcripts for accuracy. Interviewers maintained interpretive validity (Maxwell, 1992) during the interview by seeking clarification, requesting examples, and summarizing the interviewee's

statements. During the second interview, when necessary, we asked participants to confirm or clarify information obtained from the previous interview. Additionally, our research team members challenged each other's interpretations to ensure the inductive reasoning was "grounded" in the data.

Results

Uncertainty emerged as the central dimension of parents' experience related to their infants' equivocal diagnostics for CF. Therefore, we revisited the literature and identified two evolving theories that resonated with our findings: Uncertainty in Illness Theory (UIT) and Suffering. UIT (Mishel, 1988, 1990) gives structure to the psychological processes involved in recognizing, appraising, and managing uncertain circumstances. According to this theory, uncertainty arises from the ambiguity, complexity, unpredictability, or lack of information about an illness. Uncertainty that results from an unexpected diagnosis, can create a shift in the person's usual patterns of functioning. If the required degree of shift falls within the person's range of capacities and resources, the person continues to function reasonably well. However, if the uncertainty exceeds the individual's threshold for adaptation, their functioning can be thrown off balance (Mishel, 1990). UIT (Mishel, 1988; Mishel & Braden, 1988) posits that there are three stages to the experience of uncertainty: (a) stimuli frame, (b) appraisal, and (c) coping strategies. The stimuli frame is the symptom profile that precipitates the uncertainty (Mishel & Braden, 1988). One's cognitive capacities influence whether the stimuli frame is coded as certain or uncertain. Structure providers, such as health professionals or social contacts, also influence one's perceptions of the stimuli frame by providing information that shapes interpretations of the event. When the stimuli are coded as uncertain, the state is appraised as either a source of danger or opportunity. Finally, the direction of the appraisal leads to coping strategies that reduce, maintain, or manage uncertainty.

Morse (2000, 2001) offers a conceptual model to explain suffering as comprised of two mutually exclusive affective states in response to significant loss: enduring and emotional suffering. Enduring involves focusing on present tasks, rather than the loss, and actively inhibiting emotional expression whereas emotional suffering allows one to dwell on the loss and openly release related feelings. Distinctive behaviors have been associated with each of these states and with the transition from enduring to emotional suffering (Morse, Beres, Spiers, Mayan, & Olson, 2003). Within this model both responses serve important functions in coping with loss. Enduring protects the person from the psychological pain that might interfere with performing essential activities of daily life. Emotional suffering is essential for the resolution of the loss because it helps the person "reformulate" her or his life and find hope for the future. The expression of emotional suffering also acts as a distress call to inform others of a need for consoling, and thus facilitates healing. Suffering requires more emotional energy than enduring; therefore, people alternate between enduring and emotional suffering as their circumstances require and their psychological reserves allow. If suffering is not allowed to occur, it can seep out as intense displays of emotion. On the other hand, people can also actively retreat from emotional suffering through diversion activities. According to this model, suffering is a natural dynamic shift between enduring and emotional suffering that moves an individual toward acceptance of the loss and a return to emotional well-being.

We found that our data matched most of the UIT categories in the existing conceptual models, e.g., stimuli frame, degree of uncertainty, uncertainty as danger or opportunity. When the affective categories in our data did not fit the model, we drew from Morse's (2000, 2001) work on suffering to form a new category we named affective response. This new component of our modified model complemented Mishel's cognitively-oriented UIT. Additionally, we added individual characteristics and time as contributing factors. We expanded the types of coping responses relative to uncertainty, e.g., reducing or managing uncertainty. We retained the

contextual contributions of structure providers but expanded it to include the Internet and popular media. As illustrated in Figure 1, the results produced a modified, blended model constructed of five sequential domains: stimuli frame, degree of uncertainty, opportunity-danger continuum, affective responses, and coping as a more comprehensive representation of the results than either model alone. The following narrative describes each component of the modified blended model with exemplars that use pseudonyms to protect participant confidentiality.

Stimuli Frame

Several factors, consistent with the UIT model, contributed to parents' sense of uncertainty. Ambiguity occurred at two junctures in the diagnostic process. First, the screening and diagnostic test results were contradictory. The presence of two CF mutations, usually resulting in an abnormal diagnostic test and CF symptoms, was very perplexing to parents because their infants had normal or borderline sweat test results and were healthy. Second, the lack of existing empirical knowledge about the natural history of the mutations left providers and parents with little certainty about the implications for the infants' future health. Furthermore, the difficulty in distinguishing normal childhood problems from CF symptoms caused parents to worry about whether seemingly minor illnesses were harbingers of serious CF complications. Finally, because the patients were newborns, parents had no history against which to compare their infants' health. Our findings of ambiguity, indistinguishability, and unfamiliarity as antecedents to perceptions of uncertainty matched the findings of Mishel and Braden (1988).

Individual Characteristics: Cognitive Resources, Emotional State, and Gender

Our analysis found three parent characteristics that contributed to their interpretations of and reactions to uncertainty relative to their infants' test results: educational level, emotional state, and gender. We renamed the cognitive capacities component of the original UIT (Mishel, 1988) to cognitive resources and moved education from the structure providers found in the original UIT to the cognitive resources subset because it offered a better conceptual fit in the modified model. Based on our analysis, we added emotional state and gender to the model. Cognition, emotional state, and gender are individual characteristics; therefore, we labeled them as such in our model.

Education level may have influenced parents' understanding of the genetic results. One couple with less than a college education misinterpreted their infant's test results and thought the infant carried only one CF mutation when in fact the child had two. They explained that several weeks after their infant's testing, they received "a bunch of pages with high-tech words that we didn't know." They made sense of the report as saying that the infant received a CF mutation from one parent who was a "CF carrier" and a "bad gene" that "apparently wasn't mutated enough" from the other parent. The couple concluded that the combination of these genes "caused the CF gene to stand out more" and therefore their infant was a CF carrier. Mishel (1988) suggested that the duration of uncertainty would be inversely related to educational level because less educated individuals might need more time to acquire and gain full appreciation of the meaning of information than persons with more education. In our study, the less educated couple reported no uncertainty about their infant's test results while the more highly educated parents struggled with much uncertainty. These results suggest that education level can influence one's perception of uncertainty in a variety of ways.

Although Mishel (1988) acknowledged the potential impact of emotions on cognitive functioning, her model does not explicitly include this construct. We added emotional state and its relationship to cognition to our model based on parents' testimonies about the adverse effects of intense feeling on their capacities to absorb the information even when reassurances were offered by primary care providers. Some parents reported hearing only worrisome

content, particularly the words “cystic fibrosis.” Hearing the NBS results in the context of disturbing connotations about CF dramatically undercut the exhilaration of having a healthy newborn and thrust parents on to a frightening emotional “rollercoaster.” Words like “scary,” “devastated,” “panic,” “constant crying,” and “stunned silence” depicted this initial psychological plummet during the first few postpartum days or weeks between learning of the abnormal NBS result and having the diagnostic sweat test. The detrimental effects of negative emotion on cognitive functioning reported by parents in this study have been well documented by emotion regulation research (Gross, 1999; Richards & Gross, 2000). Thus, our model explicitly identifies a link between emotional state and cognitive resources.

There’s just this moment when time has frozen. And you just think you can’t be possibly living this moment. Then, the shock and stunned silence when he says that the report has come back for possible cystic fibrosis. And then even though he goes on talking for five minutes without pausing so that you don’t panic, you really don’t hear any of it because you are in such a panic... I heard everything, but all I really heard was cystic fibrosis. (Belinda)

We also found gender differences in parents’ reported expressions of sadness. In contrast to mothers’ open display of distress, fathers suffered in more private ways. This quote illustrates how deeply fathers can be affected by uncertain diagnostic results in their infants. It also shows how socially prescribed gender differences can lead others to underestimate the extent of fathers’ emotional pain.

My wife had commented on a couple of occasions that it didn’t seem to be bothering me as much as it did her because I wasn’t crying. Well, when I was out mowing the lawn, and doing all of those things, I was crying. (Blake)

Structure Providers: Social Contacts, Internet, Popular Media, and Professionals

Most parents had some knowledge acquired through previous personal encounters with individuals having CF, conversations with family or friends, and information obtained from the Internet or popular media. These preconceptions served as filters for their reactions to the news of abnormal NBS for CF. They viewed CF as a serious condition associated with severe symptoms, painful treatments, and frequent hospitalizations. One parent recalled vivid memories from her youth when she shared a hospital room with a child who had CF. Another parent knew of a sports celebrity whose child had CF. Regardless of whether the informational source was from personal experience or the popular media, most parents viewed “cystic fibrosis” as a lethal illness and inconsistent with their infants’ apparent good health, amplifying their uncertainty. Given these findings, we revised the structure providers of the model to include the Internet and popular media as well as professional and social sources of information described by Mishel (1988).

I remember when I was in high school ... and at (hospital) I would hear this pounding sound and these kids coughing and we were like what is going on? And they said, “Oh that’s a child with cystic fibrosis ... ” The thing that stuck in my head was that sound and that horrible coughing, which was scary. (Abby)

There was a football player and his son had it. Their life expectancy is like 30 years old or something like that. (Adam)

I thought, I got to learn more about this in the next ten seconds. So ran to the Internet to educate myself. (Christine)

Similar to the findings of Mishel and Braden (1988), the credibility of the structure provider influenced the degree and perceived seriousness of the uncertainty reported by parents in this study. Parents explained that Internet sites were replete with details about the pathophysiology, symptomatology, and limited longevity associated with CF. Their efforts to find data about the

rare, non-disease producing CF genetic variants that their infants had were fruitless and frustrating. They described the information as limited, old, or irrelevant to their children. Parents agreed that genetic counselors were their most credible sources of up-to-date factual information and scientific evidence. The counselors' explanations of medical terminology helped parents understand CF genetics and the meaning of their infants' test results. Although counselors were unable to resolve parents' uncertainty about the prognosis for infants with intermediate diagnostic findings, they helped parents reframe the situation as more hopeful than parents initially believed it to be.

The lack of information is what was so frustrating, I'd say for both of us but I was feeling that way because I also like to have a lot of information so I can figure it out, get on top of the problem and figure out if there is a solution or how to understand something so you can move forward. And it was frustrating that there was no real information, it's so new, the discovery of it [their infant's genotype]. (Adam)

She [genetic counselor] was clearly prepared for our confusion, she handled it ... I went from absolutely knowing nothing and upset because the test didn't work to leaving comfortable with what we knew and what might happen with the results. She was absolutely fantastic. (Christine)

I was really worried and scared because I didn't have any information and I like to have information in my hands and I think we were confused actually for quite a while ... So it was just, it was like a roller coaster ride of trying to figure out what it all meant. (Abby)

Degree of Uncertainty and the Opportunity-Danger Continuum

Most of the parents in this study reported a high degree of uncertainty and at least initially viewed this state as cause for serious concern. An exception was found in one couple who had an older child with the same genotype as their newborn. With a clear understanding of the recessive genetics of CF, this couple chose to conceive another child because their older child remained very healthy. For them, the infant's test results were viewed as an opportunity to take extra precautions in maintaining each child's health. We observed a similar shift in the other parents' viewpoints between the first and second interviews. Consequently, we adjusted the model to depict danger and opportunity as a continuum rather than a dichotomy because a continuum allows for changes associated with time or other contextual influences.

So the range of emotions went from this is really bad to oh this is nothing ... I mean the range of emotion was that way because I realized I didn't know anything and there wasn't much information on it. I'm like, oh you know, at first you're just worried and you're thinking that this is really bad but as I was realizing there's not much information. I don't really know anything about it. I'm like well this really could be nothing. I don't want to overreact and act like there's something terrible when I really don't have a clue. (Adam)

Affective Responses to Uncertainty Over Time

Affective responses to uncertainty appeared to change as a function of time. All parents initially associated uncertainty with danger. As time passed and children remained healthy, the perceived seriousness of their circumstances lessened. Parents described less intense expressions of distress. Therefore, we added time as a contributing factor. Whitmarsh and colleagues (2007) also found that time was a factor that allowed parents to know and understand their children in ways that offered them a more favorable view of uncertainty.

Emotional Suffering and Enduring—The constructs of emotional suffering and enduring suffering (Morse, 2000, 2001; Morse et al., 2003) matched our findings and were incorporated

into the model. We found that most parents, particularly mothers, reported an initial emotional state punctuated by feelings of sadness, anger, guilt, blame, and somatic symptoms, e.g., insomnia, “couldn’t breathe.” An uncertain prognosis can be conceptualized as the potential loss of one’s healthy infant leading parents to engage in enduring and emotional suffering. These behaviors reportedly occurred around the time of and shortly after receiving the test results. The mother of an older child with the same diagnostic results as her infant described a similar sense of loss associated with the infant’s results, but an attenuated emotional reaction. This more restrained and accepting response matched the construct of enduring as illustrated in the following quote.

It was easier this time because we knew she’ll never have classic CF. We knew going into it [the pregnancy], that there was a risk that we could have another child with CF, but we knew that it would only be the same mild form from our experience with (our older child) ... I was sad, it was hard but it wasn’t as devastating... People talk about worst days of their lives. You know, the worst day was when I found out (older child) had it, and the second worse day was when I found out (infant) had it. (Diane)

In sharp contrast with the previous exemplar, another couple shared their accounts of how the news of an abnormal NBS exacerbated preexisting postpartum depression and elicited thoughts of infanticide as well as suicide in both parents. These thoughts emanated from a profound sense of remorse for passing defective genes to their infant and a desire to protect the infant from future suffering. They acknowledged that acting on such ideations was unlikely. Still, these disturbing thoughts tarnished the nearly three weeks of waiting for their infant’s sweat test appointment. The negative cognitive schema characteristic of clinical depression most likely amplified their representations of uncertainty as danger. The following quotes illustrate this psychological pain that we classified as suffering and the powerful impact that emotional state can have on one’s interpretation of a stimuli frame, such as an abnormal NBS result.

Wondering whether it was wise to bring her into the world, thinking it wasn’t; considering the possibility of ending it—both her life and our own, which is something we seriously considered and we’re both, you know, as stable as anyone else. But we had no intention of bringing in a diseased or defective, quote unquote, child and she would suffer. (mother with postpartum depression during the first interview)

Well, we thought if she would face a life of suffering and misery, that, why should she? (spouse of depressed mother)

Remembering—The one year birthday was particularly poignant for these parents because it also marked the anniversary of their newborn screening experiences. All of the parents recounted distressing memories of that period. The recollections of the mother who had postpartum depression seemed to be more frequent and traumatic than the other mothers. She recognized retrospectively that her depression had a serious effect on her reaction to the NBS results and caused her to filter out any messages of hope and focus solely on the worst possible scenario for her infant. The prolonged waiting time for the sweat test appointment combined with her depression seemed to leave her with disturbing memories of all events surrounding NBS. For all of these parents, the joyfulness of their infants’ births was marred by their experiences of NBS. Although memory is generally viewed as a cognitive process, we included remembering in the affective response domain because the content and tone of parents’ memories emanated from their feelings rather than cognitive recall.

I still think about it, but not in the way that it consumes me...there aren’t any days that pass that I don’t think about it. (mother with PPD) (Her spouse added) I don’t think about it nearly as much as her I believe, but it’s still one of those things you’ll often wonder about. Just think about in the back of your mind.

I was so excited for her to be healthy and then to find out that there was always going to be this cloud hanging over her. No parent wants that for their child. (Diane)

We'll always wonder because of the screening, if we didn't know this [about the CF mutations], we would just assume he's a healthy kid, but now we have the wonder, "Is it CF? Could it be?" It's taken away a little bit of the joy. (Carl) A little bit. (added by his wife)

Worrying—Although children were healthy at 12 months and most parents had gained a better understanding of the available facts, the uncertainty about their children's future health continued to cause worry. They wondered if the defective genes might remain dormant for a period of time and become destructive later in life. Some parents also voiced concerns about the potential stigmatizing effects of a CF label. Even in the absence of symptoms, parents' worried that the documentation of two CF genes would lead health and life insurance companies to deny their children coverage because any respiratory problems would be considered a preexisting condition. Additionally, one parent cautioned about the potential for adverse effects on parents' perceptions about the affected child. This parent speculated that some parents might ration their resources in favor of the child who does not have the CF label, because he is more likely to survive into adulthood than the child with a CF label.

I think it's fair to say we believe that he's healthy, but we'll always have it in the back of our mind, could it be [CF] ... it's just now knowing in our minds one gene's on one side the other's on the other side and that's supposed to be a big deal I guess. But yet his sweat test is down so, really, can it change? I don't know. Do they [CF experts] know? (Carl)

The potential for genetic discrimination in terms of health and life insurance, or future employment is certainly a great concern now that I think is something that needs to be taken into account ... A child who is asymptomatic and later becomes symptomatic because many pre-existing condition clauses and health insurance plans specifically identify what is a pre-existing condition. (Belinda)

If you're so seriously affected by this that you might eventually say, "Well look, why put resources into this child? We'll only save for the other one to go to college because this one might not survive." (Belinda)

Feeling Isolated—Two mothers expressed a desire to talk with other parents of children with similar diagnostic situations. They wanted to learn how other children were doing medically and how other parents coped with the ambiguity of the diagnostic classification. Although parents found support groups and Internet chat rooms, these resources were designed for parents of symptomatic children. This inability to connect with others in similar circumstances led to a sense of isolation, particularly for mothers.

You feel like an island because there really isn't a lot of information. There isn't a support group [for parents in these circumstances]. (Abby)

Coping with Uncertainty: Reducing or Managing

Based on our analysis, we framed the coping domain of the model to directly relate to uncertainty. The results produced two categories: one with strategies aimed at reducing uncertainty and the other focused on managing uncertainty. The reducing category contained only the strategy of searching for information, whereas we found seven strategies classified as managing uncertainty. These findings suggested that when the potential for reducing uncertainty is limited parents invested their energies in managing the uncertainty.

Searching for Information—Searching for more information was the only strategy directed toward reducing uncertainty. Parents sought three types of information: (a) facts about the natural course of CF in people with the same genotype as their infants, (b) the source of the CF gene within their families, and (c) personal experiences of other parents of children with the same or similar genotypes. The Internet was the most immediately accessible source, but genetic counselors were considered to be among the most valued sources of information. Other sources included written literature and social networks. Parents also described a process of questioning relatives to find some indication of the genes in extended family members or earlier generations. Even though parents continued to engage in this strategy at the time of the 12-month interview, for some the search shifted to wanting contact with other parents to share experiences on a personal and emotional level as compared with the factual information sought earlier. Other researchers have observed similar attempts to reduce uncertainty in other populations with serious health problems (Donovan-Kicken & Bute, 2008; Oliffe, Davison, Pickles, & Mróz, 2009).

I spent all of my time reading about cystic fibrosis and specifically that specific genotype. And I read any and all available literature that I could find on it. (Belinda-first visit)

We've actually asked as many people in our families as we can about our family history and there is no one that has cystic fibrosis. No one has ever had it. I mean as far back as we know of people being diagnosed with cystic fibrosis, there's been no family member ever diagnosed with cystic fibrosis. (Abby, first visit)

I would love for a mom to call me ... So I could ask, "Could you tell me how to feel? Can you tell me what's going to happen?" And we would compare notes ... I think it is so important to talk versus research and numbers and tests and doctor's appointments because only real people are going to tell you how to feel. (Christine, 12 month visit)

Mobilizing—After talking with CF experts, most parents had a better understanding of their infants' conditions which prompted a shift from emotion-focused responses to action-oriented coping. The emotional rollercoaster during mobilization changed from feelings of deep despair to a sense of hopeful agency. Information became the valuable commodity that might reduce uncertainty and alleviate their worry. They worked intensely to learn everything they could about their infants' genotypes so that they could do whatever was necessary to maintain their infants' good health. This shift in attitude was abrupt for some, gradual for others, but always deliberate and adaptive.

We almost went through the state, the grief process in like ten minutes and I was like okay guys suck it up. We can either sit here and cry about this right now, or we can learn what we're going to do ... And we're learning how we're going to fix it. (Christine)

Monitoring—Long after learning about the test results, parents remained vigilant for signs of CF-related illness in their children. This parental monitoring was similar to the intrapsychic coping method described by Mishel (1981). During the 12-month interviews parents tended to emphasize how healthy their children were with words like "preposterously healthy," "robust," and "really strong and energetic." Still, they shared doubts about their children's futures because their searches for prognostic information remained unsatisfying.

I will literally be living with that unknown [whether child will develop CF symptoms] for the rest of my life. (Belinda)

I'm still trying to understand how, how he can have these two genes and have nothing. So am I making myself a little anxious? Probably, I will always sleep with one eye open. I'm always going to be concerned that the reason he has a sinus infection is because the genes that we gave him." (Christine)

Relabeling—Another strategy for managing the uncertainty involved creating a new or different label for their children's condition that symbolized their interpretations of, and perhaps their hopes for, more favorable outcomes than those typically reported with CF. The way providers framed test results influenced some lexicon choices. One couple described their child as having a "mild case;" another referred to "non-classic CF." In the absence of technical language, parents created their own labels. One mother referred to her child as the "miracle baby" because the child's test results and good health defied medical literature that linked two mutations with CF symptoms. Still another parent used the term "double carrier" to indicate the presence of two mutations, but no disease. This form of distancing the child from the diagnosis has also been described by Whitmarsh and colleagues (2007).

Being Health Conscious—Uncertainty and related worry prompted parents to be vigilant, in some cases hypervigilant, for signs of CF. Parents wondered whether every respiratory symptom or gastrointestinal (GI) problem was caused by the defective CF genes and possibly the beginning of the child's decline into the dreaded complications of "classic CF." Given that most typical childhood ailments tend to be upper respiratory illnesses or GI problems, parents devoted a great deal of their energies to monitoring their children's health and communicating with primary care providers. They described taking precautionary health measures that, for some, included regular evaluations by a CF specialist and flu vaccinations for the whole family, as well as the affected child. In contrast, one couples' precautionary measures included avoiding CF clinics to prevent exposure to potential infection. A similar strategy, called "doing something extra," has been used by patients dealing with the uncertainties of prostate cancer (Olliffe et al., 2009).

I know that I need to do all of the right things to take care of him, like recommendations to get flu shots. We didn't normally get flu shots ... we both know that we just need to do what the doctors tell us to do and bring him in for the check-ups at the hospital as well as with the pediatrician. (Abby)

I think we are maybe a little bit quicker to go to the doctor with a cold that lingers. Typically they say, let it run about two weeks, and we might try to get them in a little bit sooner if we think that it's not going to go away on its own ... We try to get them to eat as healthy as they can. (Diane)

Initially they said, "You should follow up once a year [at a CF Center]." But, they back peddled on that and said it was it wasn't necessary. Even if they did say that, we wouldn't do it ... I don't need to have that kind of yearly reminder and there's also the issue of potential contamination. And there is no dirtier place on earth than any hospital setting. (Brenda)

Being Hopeful—Another coping strategy involved remaining hopeful about their child's future. Parents described their efforts to consciously set favorable expectations about their children's health. If extended family, particularly grandparents, voiced concerns about the infant's vulnerability to illness, parents attempted to convince their relatives that the children were healthy. Parents used evidence, such as the infant's growth parameters or professional opinions to support their arguments. Additionally, parents emphasized their children's good health, especially in the areas of nutrition and respiratory status, with the interviewers during the 12-month data collections. This idea of focusing on the child's non-symptoms echoed the

findings of Whitmarsh et al. (2007). Parry (2003) also noted that young adult survivors of childhood cancer also cling to hopefulness in coping with uncertainty.

I don't see him being hospitalized or being sick. I see him being very healthy and strong...I think that he's going to be just fine, like (his healthy older sibling). He's going to be totally normal, healthy. (Abby) 100% (added by her husband)

They [grandparents] act like he's sickly. That he's going to die or something. We go overboard to explain ... We get all of the percentages that he's 50% in his height and 75% in weight, that the doctors say, "He's doing great," so eventually after a few years, they'll get it. (Adam)

Gate Keeping Information—While some parents freely shared the diagnostic findings with the people in their lives, others were much more selective about divulging this information. This gate-keeping strategy was an attempt to protect the child from the potential stigmatizing effects of a "CF label" and perceptions associated with the illness. In one case, this recommendation reportedly came from a CF specialist.

All of our family knows, some of our friends know. None of her teachers know ... the doctor at the clinic said that because her case is mild, it was likely that if we told them that she had cystic fibrosis that she would be labeled as "that kid with cystic fibrosis" or she would be treated differently and we didn't want her to be known as "that sick kid." (Diane)

Thinking about the Future—In addition to uncertainty about future health problems, one mother talked about her son's potential infertility as "heartbreaking." She described the joys that having children brought the couple and was saddened to think her son might not have similar opportunities. The infertility issue also posed an interesting dilemma as to how and when to explain this possibility to their son. Even though he was still an infant, these parents were already planning the timing of this conversation with their son. They expressed reservations about informing him too early in his adolescence because they worried that without the deterrent of potentially impregnating a girlfriend, he might become sexually active and engage in unprotected sex. They planned to wait until they deemed him capable of responsible decision-making.

My big fear for a while was the lack of vas deferens and then I thought, "Oh no, he's not going to be able to have children ..." Because I just think how much I love our kids and so for him that was very heartbreaking for me to think that he wouldn't be able to have children of his own. (Abby)

The one thing we won't tell him is that he might not have a vas deferens because that's not an appropriate thing to tell a boy, "Oh by the way you can't get a girl pregnant by having intercourse." That wouldn't be a smart thing so that part might get left out until he's older and can handle that responsibility. (Adam)

As parents gained a better understanding of the genetics of CF and their infants' genotypes, some wondered if their older children might have the same CF genes. These concerns were amplified by the presence of any CF-type symptoms, particularly respiratory problems. On the other hand, the realization that the older child might have the same genotype offered them reassurances about the infant. If the older child's putative CF symptoms were perceived to be non-life-threatening and manageable, parents felt hopeful for the infant's future and confident in their capacities to meet the child's needs.

And then our oldest who has had sinus infections, pneumonia, well I'm wondering does she have (the same CFTR variants as infant) and if she does, if she is a mirror

image to what (infant) may be going through when he's two and three, I can handle that. (Christine)

Parents' Recommendations

When asked for feedback about improving the NBS process, parents offered the following specific recommendations: (a) assist parents in scheduling the sweat test right away, (b) create a new label for the CF genotypes associated with asymptomatic or mild forms of CF, (c) provide parents evidenced-based facts, and (d) connect parents with other parents of children with similar genotypes. These recommendations fit well with the structure provider component of the modified model and hold the potential to affect interpretations, appraisals and responses to uncertainty in a favorable direction.

Assist Parents in Scheduling Sweat Test Promptly—The lengths of time between notification about NBS results and the sweat test appointment ranged from one day to almost three weeks. All parents received the results of the sweat test on the same day of testing. Parents unanimously agreed that minimizing the waiting period for the diagnostic testing and communicating results was highly desirable and would lessen parental distress. As the following mother noted, foreshadowing the nature of the follow-up testing also reduced parental concerns.

She [genetic counselor] said, "You will come in for a sweat test, painless, fast, and you will get your results the same day ..." We get to learn tomorrow. I was ecstatic with relief. (Christine)

Create a New Label—Even though parents varied in the terms that they used to describe their infants' health status, they made it clear that their infants did not fit the category of CF patients described in the literature or on the Internet. The use of the CF label for their infants in the NBS notification process was a major source of concern. Parents found the label misleading and a source of substantial confusion and emotional distress. Several parents strongly advocated for a totally different label, such as naming the specific variant.

The objection is don't tell me that it's possible cystic fibrosis when in fact it is not possible cystic fibrosis. If you feel that I need to know that I'm a potential carrier, then notify me that I'm a potential carrier and that my child does have some genes that are associated with cystic fibrosis. But if you're going to do that then make it damn clear to me that my child does not have cystic fibrosis. (Blake)

My personal opinion is it should be called the allele [that the infant has] because the very words cystic fibrosis draws fear or concern that "wow this is a big deal." You see stuff on cystic fibrosis and people are already worried and concerned anyway and they can come to conclusions that are inaccurate. (Adam)

Provide Parents Facts and Figures—Parents suggested that primary care providers (PCP's) be better prepared with facts about NBS, be capable of answering questions, and offer parents literature explaining test results. Fathers tended to want empirical evidence documenting the statistical probability that their children would develop health problems. Most of these families chose to participate in this study because of the lack of research about children with non-disease producing CF variants.

I wish they would have said what it is, there's two genes, he has one, we have to test for the other. A little more information would have saved a lot of grief. (Carl).

... publications. A description of it, for instance having lots of patient data, lots of information on 1,000 people with the alleles. Say, "We've discovered this many; this percent have this happen." (Adam)

Connect Parents with Other Parents—Several mothers expressed the desire to talk with other parents of children in similar diagnostic situations. Interest in exchanging real life experiences with other families seemed to serve several purposes. It might alleviate fear of the unknown by offering parents a window into their own child's potential future. Second, it could serve as an antidote to parents' sense of isolation by validating their thoughts and feelings. Finally, it would provide parents a network of resources beyond professionals.

And I think it would be helpful and I know that I mean you have to have consent or whatever, but to have other people that have a child with non-classic cystic fibrosis available to talk to because it's a really scary time when you don't. When there isn't much information and there maybe a lack of information due to the lack of research because it's so new, it would certainly help alleviate fears if you were able to talk with someone else or even have them share their stories or experiences. (Abby)

Well and then you just sit there and you look at your computer and it's just calling you come read, read me, read all. And what I wanted to know, not what cystic fibrosis was necessarily, but what real people were writing on message boards about what their kids were feeling and doing physically. (Christine)

Discussion

This study was the first to examine parents' perspectives about their infants' equivocal diagnostic test results following genetic screening for CF. We found the construct of uncertainty to be the central thematic dimension in the results. Although uncertainty has been identified as a key concept in acute and chronic illness for adult and pediatric populations (Neville, 2003), its role in genetic screening has received far less attention. Therefore, we attempted to apply our findings to the UIT that was originally conceptualized for individuals with acute illness (Mishel, 1988) and chronic conditions (Mishel, 1990, 1999). The results produced an expanded version of the UIT (Mishel, 1988, 1990; Mishel & Braden, 1988) that (a) included emotional state and gender as well as cognitive capacities as individual characteristics contributing to perceptions of uncertainty, (b) added the Internet and popular media as structure providers, (c) conceptualized the appraisal of uncertainty along a continuum rather than distinctly separate entities, (d) introduced affective responses as a domain based on a model of suffering (Morse, 2001), (e) added time as a contributing factor, and (f) reframed coping strategies relative to affective responses and appraisal of uncertainty. We did not include the concepts of inference or illusion (Mishel, 1988; Mishel & Braden, 1988) in our model because they were not observed. Perhaps, the circumstances of the parents in this study did not allow for such appraisal methods. Moreover, the UIT has been conceptualized as a cognitive representation with little attention to affect. The results of this study and our previous work (Thuczek et al., 2006) support the inclusion of emotion as a critical contributing factor as well as an outcome to consider in predictive or explanatory models of uncertainty. Based on our findings, the expanded framework offers for a more comprehensive description of the dynamic psychological processes that contribute to representations of uncertainty and its consequences.

Research Implications

This study is limited by a small sample size and the homogeneity of its participants. However, the findings contribute to the literature by reconceptualizing uncertainty theory to include affective components and by extending the theory's application to genetic testing. This report also delineates previously undocumented psychosocial complications of genetic testing in the context of NBS. Additional research is needed to establish the utility and universality of the modified framework, particularly with larger samples and more culturally diverse participants. Additional investigation is warranted to (a) obtain empirical support for the proposed affective domains of the model, (b) examine how structure providers mediate appraisal of uncertainty,

(c) determine how professional structure provider interventions can mediate affective responses and enhance coping, and (d) evaluate the effect of time on each domain. Genomic health care has entered uncharted territory with varying degrees of uncertainty for affected patients and their families. The modified, blended model of uncertainty and suffering provides a conceptual framework to guide future investigation in this area.

Clinical Implications

All of the parents in this study reported some adverse psychosocial consequences related to the NBS and diagnostic process. Given the complexity of the CF genetic spectrum, even highly educated parents struggled to grasp the nuanced meaning of test results leading to uncertainty. Parents also described acute emotional distress similar to that described in the literature when a child has an abnormal sweat test and is diagnosed with CF (Jedlicka-Köhler et al., 1996; Quittner et al., 1992). Such findings suggest that parents of infants with abnormal NBS results plus equivocal diagnostic findings experience a similar degree of uncertainty and appraisal of uncertainty as a threat to their infants' well-being as parents with infants who have definitive diagnoses. Uncertainty theory offers guidance for two types of interventions: those that reduce the uncertainty or those that reframe it as an opportunity.

The following uncertainty-reduction strategies are informed by theory and grounded in the results of this study. First, without raising undue concern about the severity of the situation, the time between informing parents about abnormal NBS results and the appointment for diagnostic testing could be made as brief as possible. Primary care providers could expedite the scheduling process by reserving a tentative appointment at a CF Center before contacting parents with NBS test results. Second, to provide parents accurate and timely information, it is critical that informants have access to information about NBS and the meaning of the genetic findings. Such essentials can be made available and updated on Internet web sites sponsored by state and national professional organizations. Third, professionals need to recognize that emotional distress can alter parents' abilities to fully absorb details and augment verbal explanations with literature and Internet resources supporting parents' need for information. It might be helpful to ask parents if they would like a follow-up telephone call the next day to see how they are doing and to answer further questions. Such contacts also communicate a sense of compassion and caring.

Health care providers can use reframing strategies that apply a probabilistic perspective of uncertainty rather than deterministic position (Twomey, 2006). This approach can help parents regard uncertain results as an opportunity for hopeful outcomes. First, creating a uniform lexicon that differentiates the intermediate diagnostic classification from the more serious form of the condition might help parents frame their infant's situation in a more favorable light. Second, having a credible provider, e.g., expert in genetics and NBS, available to parents throughout the diagnostic process could also improve parents' appraisals of the uncertainty. Third, offering parents opportunities to contact other parents of children with similar test results could reduce the sense of isolation and enhance parent's capacities. Fourth, although gender differences might influence observable emotional reactions to perceived bad news, fathers are almost certain to be equally affected by abnormal results as mothers. Both parents should be offered information, follow-up communication, and contacts for questions. Finally, abnormal NBS results are being shared with parents during a particularly vulnerable time when they are likely to be overwhelmed, sleep-deprived, and emotionally drained. About 10–20% of women in the general population suffer from postpartum depression (Centers for Disease Control and Prevention [CDC], 2008; O'Hara & Swain, 1996; Vesga-López et al., 2008) and the rate of clinically significant depressive symptoms can be as high as 68% for women and 32% in men following abnormal NBS (Tluczek et al., 2005). Some women with postpartum depression have thoughts of suicide or infanticide. The findings from this study suggest that delivering

abnormal NBS results under the conditions of postpartum depression can trigger such ideation. It is critical for informants to be knowledgeable about and sensitive to the signs and symptoms of depression. When concerns arise, informants should empathically screen for depression and promptly refer parents to mental health resources as needed.

Conclusions

Genomic health care has entered uncharted territory with varying degrees of uncertainty for affected patients and their families. This secondary dimensional analysis of parent interviews about their infants' equivocal genetic test results for CF found uncertainty to be the central thematic dimension. Results showed that parents passed through a series of stages similar to the process described by Mishel's Uncertainty in Illness Theory (UIT). The findings informed modifications made to the original theory, extended the application of uncertainty theory to circumstances in which the very presence of an illness is uncertain, and offer guidance for future research and practice.

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Figure 1.
Modified Uncertainty Model