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# A Tailored Approach to Family-Centered Genetic Counseling for Cystic Fibrosis Newborn Screening: The Wisconsin Model

**Audrey Tluczek**[1], **Christina Zaleski**[2], **Dania Stachiw-Hietpas**[3], **Peggy Modaff**[4], **Craig R. Adamski**[1,4], **Megan R. Nelson**[1,4], **Catherine A. Reiser**[4], **Sumedha Ghate**[5], and **Kevin D. Josephson**[6]

[1] University of Wisconsin School of Nursing, Madison, WI

[2] Marshfield Clinic, Marshfield, WI

[3] Children's Hospital of Wisconsin, Milwaukee, WI

[4] University of Wisconsin School of Medicine and Public Health, Genetic Counselor Training Program, Madison, WI

[5] St. Vincent Hospital, Green Bay, WI

[6] Gundersen Lutheran, LaCrosse, WI

## Abstract

**Objective—**Develop a tailored family-centered approach to genetic counseling following abnormal newborn screening (NBS) for cystic fibrosis (CF).

**Method—**A genetic counseling consortium reviewed research literature, selected theoretical frameworks, and incorporated counseling psychology micro skills.

**Results—**This innovative intervention integrated theories and empirically validated techniques. Pilot testing and parent feedback confirmed satisfaction with and feasibility of the approach designed to (a) minimize parents' distress, (b) facilitate parents' understanding, (c) increase parents' capacities to use genetic information, and (d) enhance parents' experiences with genetic counseling. Counselors engage in a highly interactive process of evaluating parents' needs and tailoring assessments and interventions that include a therapeutic environment, the family's emotional needs, parents' informational needs, and a follow-up plan.

**Conclusion—**This promising new model is the first to establish a theory-driven, evidence-based standard for genetic counseling in the context of NBS for CF. Additional research will evaluate the model's efficacy in clinical practice.

### Keywords

cystic fibrosis; CF carrier; genetic counseling; newborn screening; micro skill; tailored intervention

Address for correspondence: Audrey Tluczek, PhD, RN, University of Wisconsin, School of Nursing, 600 Highland Ave. K6/346, Madison, WI 53792, Tele: 608-263-6111, Fax: 608-263-5458, atluczek@wisc.edu.

Institutes where the work was conducted:

- University of Wisconsin, Madison, WI
- Marshfield Clinic, Marshfield, WI
- Children's Hospital of Wisconsin, Milwaukee, WI

## Introduction

For the last half century, newborn screening (NBS) has been conducted nationally as a critical public health initiative to prevent significant infant mortality and morbidity. In 1991, DNA analysis was first introduced to NBS for cystic fibrosis (CF) in Wisconsin (Farrell et al., 1991; Gregg et al., 1993; Sharp & Rock, 2008). In the last decade implementation of NBS for CF quickly ensued in other states. As of December 2009, all NBS programs in the United States (US) included CF. This technology created a need for genetic counseling services to assist parents in understanding the meaning and implications of their infant's test results. However, there is no national standardized protocol documenting the most effective genetic counseling approach. This article provides an overview of NBS for CF that incorporates DNA analysis, and describes the development of an innovative theory-driven, evidence-based tailored approach to family-centered genetic counseling that builds upon and extends previous work in genetic counseling. Although the resulting model is designed for genetic counseling in the context of NBS, the model could be relevant to other disciplines and clinical populations.

### Cystic Fibrosis

Cystic fibrosis is one of the most common life-shortening inherited conditions in the US. Although CF is most common in populations with Northern European ancestry, as shown in Table 1, it has been identified in most ethnic groups. This condition affects about 30,000 individuals in the US and 70,000 worldwide. An estimated 10 million people in the US (about 1 in 31 people) carry one CF Transmembrane Conductance Regulator (*CFTR*) gene mutation (Cystic Fibrosis Foundation [CFF], 2009). In this article, "mutation" refers to a symptoms-producing variation of an allele. CF is an inherited autosomal recessive disorder. If both members of a couple are heterozygous for a *CFTR* mutation, each pregnancy has a 25% chance the child will be affected with CF; a 50% chance the child will be a heterozygous CF carrier, and a 25% chance the child will neither carrier a *CFTR* mutation nor have CF (Moskowitz, 2008).

*CFTR* mutations alter chloride channel functioning and produce multisystem pathology at the cellular level leading to abnormally viscous mucus production by exocrine glands. Symptom severity depends on a combination of genetic and environmental factors. Most individuals with CF have frequent pulmonary exacerbations of bacterial infections leading to chronic obstructive pulmonary disease, pancreatic insufficiency causing malnutrition, and congenital bilateral absence of the vas deferens in males resulting in infertility. The defective chloride channel also produces excessive chloride excretion into the sweat (Mishra et al., 2008). Although advances in research and treatment have significantly improved the longevity of individuals with CF, the diagnosis is still associated with a reduced life expectancy, with a median survival age of about 37 years (CFF, 2009). Early diagnosis and intervention resulting from NBS offer hope for decreased pulmonary complications (Waters et al., 1999; Montgomery & Howenstine, 2009), improved nutrition (Farrell et al., 1997; Jones & Helm, 2009), and increased longevity for affected individuals. Reproductive technologies have also provided options for addressing male infertility (Popli, Bourke, & Stewart, 2009).

### CF Newborn Screening and Diagnosis

The purpose of NBS for CF is to detect infants who have the disease as early as possible. However, NBS algorithms that include DNA analysis also identify infants who are carriers of only one *CFTR* mutation. For those infants, at least one of the parents is also a carrier. Furthermore, the DNA analysis performed by many state laboratories detects only about 85% of the mutations responsible for CF in children of Northern European descent. Thus,

infants with one or two CF mutations can be identified, but NBS does not detect all carriers or all types of CF.

Most CF NBS programs use a protocol similar to the Wisconsin two-tiered algorithm. Blood specimens, obtained during the infant's first 24–48 hours of life, are examined for immunoreactive trypsinogen (IRT), a substance elevated in infants with CF. IRT levels in the top four percent for the day prompt DNA analysis to detect the 23 *CFTR* mutations most commonly associated with CF in populations of Northern European decent. This procedure detects a small fraction of the more than 1700 known variants of the *CFTR* gene (Cystic Fibrosis Mutation Database, 2010). If one or two *CFTR* mutations are found, a sweat test using quantitative pilocarpine iontophoresis is indicated to rule out or confirm a CF diagnosis (Moskowitz et al., 2008). Sweat chloride levels ≥ 60 mmol/L are considered positive and indicative of CF. In newborns and infants less than 6 months, chloride levels from 30–59 mmol/L are classified as intermediate, for which a CF diagnosis is possible. Levels ≤ 29 mmol/L are within the normal range, which is a negative result, and a CF diagnosis is unlikely (Farrell et al., 2008).

In the Wisconsin NBS program about 97% of infants that have an abnormal CF NBS with only one identified *CFTR* mutation subsequently have normal sweat tests. These abnormal CF NBS results followed by normal sweat testing are considered to be false-positive screens (Rock et al., 2005). Thus, the sweat test rules out a CF diagnosis for most families with a positive CF NBS. About 3–10% of infants with a positive CF NBS have sweat test results in the intermediate range (Farrell et al., 2008; Soultan, Foster, Newman, & Anbar, 2008). Additional genetic testing is often recommended to search for *CFTR* mutations not included in the NBS panel, as well as repeat sweat testing. Intermediate sweat test results combined with the presence of two *CFTR* mutations, including one or more associated with minimal or late-onset symptoms, are classified as *CFTR*-related metabolic syndrome (Farrell et al., 2008; Borowitz et al., 2009; Voter & Ren, 2008).

The interpretation of genetic results is complicated by the variability of *CFTR* alleles, range of phenotypic presentations, and lack of documented clinical relevance for many variants. *CFTR* mutations have been classified into five categories based on the mechanism of genetic dysfunction and associated range in symptom severity (Voter & Ren, 2008; Rowntree & Harris, 2003). Class I–III mutations tend to produce significant illness, whereas Class IV–V mutations are associated with few, mild, or a delayed onset of symptoms. Patients with two Class I–III mutations typically develop pulmonary and gastrointestinal complications in childhood, but those with one Class I–III mutation and one Class IV–V are more likely to have mild, few, or delayed onset of symptoms (McKone, Goss, & Aitkens, 2006). There are no known adverse health consequences for individuals with only one *CFTR* mutation, regardless of class. To date, about 15% of known *CFTR* sequence variants have been identified as non-disease producing. Of the remaining variants, less than 2% have been proven via empirical evidence to be symptom causing mutations (Farrell et al., 2008). The clinical implications of other variants remain unclear. Many experts now view CF as a spectrum disorder with phenotypes forming a continuum from fewer, later onset of symptoms to more severe, multi-system disease. Given the increased complexity of the CF diagnostic procedure and its interpretation, it is essential that sweat testing centers include genetic counseling services to ensure parents understand the nuances of test results and implications for their infant, themselves, and extended family members.

## Parental Knowledge Deficits, Emotional Distress, and Sleep Deprivation

The motivation for developing the new model arose from research findings that repeatedly documented knowledge deficits (Ciske et al., 2001; Lewis, Curnow, Ross, & Massie, 2006; Tluczek et al., 1992), emotional distress (Moran, Quirk, Duff, & Brownlee, 2007), and

lingering worry about infant health (Tluczek, Orland, & Cavanagh, in press) among parents of infants identified as CF carriers. Additionally, genetic counseling that follows abnormal NBS results for CF is unique because the genetic testing used in NBS is mandated by law. Most parents know little about NBS in general and much less about the DNA analysis used in the procedure (Tluczek, Orland, Nick, & Brown, 2009). Therefore, the receipt of abnormal results comes as a complete shock to the majority of parents. Unlike a typical genetic counseling session in which clients actively seek genetic information, many parents affected by abnormal NBS results do not even know that they will meet with a genetic counselor until they arrive for their infant's sweat test appointment. Parents also have characterized the wait for their infants' sweat test appointment as extremely stressful. Tluczek and colleagues (2005) found significantly higher rates of depressive symptoms reported among parents of infants with abnormal CF NBS results than parents of infants with normal NBS results. The same study documented the well-known sleep deficits associated with nighttime feeding of infants; this lack of sleep might contribute to parents' inability to retain NBS information presented during counseling. Jedlicka-Kohler, Gotz, & Eichler (1996) observed that parents who were highly distressed following their child's CF diagnosis described difficulties in retaining new information providers attempted to share with them. In another study, parents attributed some of their anxiety to the way NBS test results were communicated (Parsons, Clarke, & Bradley, 2003). Such findings are not unique to NBS for CF. Waisbren and colleagues (2003) noted that mothers of infants with false-positive NBS results for metabolic disorders reported higher levels of parenting stress than mothers of infants with normal NBS results. Children with false-positive results were also more likely to be hospitalized than children with normal NBS results. Gurian and colleagues (2006) found parenting stress is associated with confusion about follow-up testing for metabolic disorders. The mandatory nature of NBS combined with the vulnerable emotional states of parents resulting from the receipt of unsolicited, unexpected abnormal results present unique challenges that may merit a more psychosocially oriented approach to genetic counseling than typically required.

## Long-term Consequences of NBS

Several studies identified long-term consequences of abnormal NBS results for CF. A dimensional analysis of interview data showed that parents continue to struggle with the uncertainty of their children's equivocal diagnostic results one year after the NBS procedure (Tluczek, McKechnie, & Lynam, 2010). In another study, some parents of infants with false-positive NBS results described the NBS experience as having a significant impact on them personally and on their interactions with other family members during the first year of the child's life (Tluczek et al., in press). In an 11–14 year follow-up evaluation of genetic counseling related to CF NBS, parents who received genetic counseling had a better understanding of the genetics of CF than those who had not received genetic counseling (Cavanagh, Compton, Tluczek, Brown, & Farrell, 2010). However, parents in both groups still had misconceptions and some wondered how best to inform their pre-adolescent children about being a CF carrier. Although the children from these studies are unlikely to need specialty care from the entire CF team, their parents might benefit from the ongoing availability of a team member, such as a genetic counselor or advanced practice nurse, to assist them in dealing with questions or concerns that arise related to their carrier status.

## Provider-Parent Communication

Effective communication is the foundation of any psycho-educational intervention. For CF NBS programs, the genetic counseling session conducted at the time of the sweat test represents a critical juncture for that process. Parents have an opportunity to meet with a CF expert who can answer their questions and share important information about NBS and their infants' test results. A qualitative analysis of parent interview data indicated that not all

parents shared the same preferences for content and timing of the counseling (Tluczek et al., 2006). Most parents reported wanting to know the probability of a CF diagnosis, how their infants were identified through NBS, about the sweat test procedure, facts about CF genetics, and implications for their children's future. Parents also expressed a desire for counseling personalized to their needs. Although most parents appreciated the opportunity to meet with a genetic counselor while awaiting the sweat test results, several parents explained that some information received during that particularly stressful time increased their worry. For example, parents inferred that detailed descriptions of the pathophysiology, symptomology, and treatment associated with CF meant that their child would be diagnosed with CF. Communication well matched to parents' needs engendered a sense of hopefulness and confidence about test results; whereas, mismatched counseling increased parents' already high levels of anxiety and produced confusion. Two other studies reported similar findings with recommendations for limiting the amount of information about CF symptoms before sweat test results are known (Moran et al., 2007; Parsons et al., 2003).

Dillard and colleagues (2008) delineate environmental factors adversely affecting the genetic counseling session. Infants' needs, e.g. for feeding or diaper changing, present the most common distractions. Active siblings often vie for parents' attention. Environmental noises, e.g. pagers, telephones, or the sweat test technician entering the room, can interfere with the flow of conversation during counseling sessions. Emotionally distraught parents and scripted counseling approaches create additional barriers to effective parent-counselor communication. The severity of the disruptions during genetic counseling sessions has been negatively correlated with parents' knowledge retention (Dillard, Shen, Tluczek, Modaff, & Farrell, 2007).

In a review of 18 studies of genetic counseling unrelated to NBS, Meiser and colleagues (2008) found counselors tend to dominate the conversation, focus on medical facts, and use a teaching approach more often than a psychosocial orientation. Favorable client outcomes were associated with a low ratio of counselor to client talk time, counselor expressions of empathy, and a follow-up letter summarizing the session. Pieterse and colleagues (2007) identified two factors associated with client satisfaction: receiving medical information and having opportunities to spend extended time with the counselor. These findings demarcate key considerations for genetic counseling interventions.

## Patient-centered Interventions

Ever-increasing empiric evidence supports the efficacy of patient-centered interventions across a range of health care disciplines including genetic counseling. In a review of 20 studies, Ryan and Lauver (2002) found that patients favored tailored psycho-educational interventions and retained more information from the tailored than the standard interventions. Behavioral health outcomes were significantly better in tailored interventions in half of the studies and results were equivocal in the remaining studies. Matloff and colleagues (2006; 2007) found that a tailored approach to genetic counseling regarding hormone replacement therapy for menopausal women with a family history of breast cancer improved women's knowledge and risk perceptions. Wallace and colleagues (2009) found that a patient-centered interdisciplinary intervention for patients with diabetes improved patient knowledge, self-efficacy, self-management, and emotional states. Patient-centered care in pediatric primary care was associated with increased trust among families (Aragon, McGuinn, Bavin, & Gesell, 2010).

## Summary of Literature Review

In short, the empirical literature documents significant parental distress and knowledge deficits associated with genetic NBS for CF. Although the genetic counseling literature

illustrates a long history of support for psychosocial interventions (Enpu, 1997; Kessler, 1997; LeRoy, McCarthy Veach, & Bartels, 2010; McCarthy Veach, LeRoy, & Bartels, 2003; Weil, 2000), there have been relatively few studies that identify the most effective techniques in the genetic counseling context or evidence-based models of genetic counseling. Therefore, the counseling model described in this article draws from the clinical investigational literature across multiple disciplines, as well as genetic counseling. The theoretical underpinnings of the tailored family-centered model are first discussed, followed by steps to the model's development, and a description of its components. Finally, implication for clinical practice and future research are presented.

## Theoretical Basis of the Tailored Family-Centered Model

### Emotion Regulation

We turned to neurobiological and psychological research for insights about how emotion and cognition operate as two independent but interactive processes (LeDoux, 1998). Ottowitz, Dougherty, and Savage (2002) documented the detrimental effects of depression on cognitive executive function, including the ability to organize thoughts, retrieve memory, attend to stimuli, and solve problems. In addition, decreased sleep can exacerbate the problem by further impairing cognitive functioning (Blagrove, Alexander, & Horne, 1995), decreasing attention (Doran, Van Dongen, & Dinges, 2001), altering mood, and reducing psychomotor performance (Dinges et al., 1997; Pilcher & Huffcutt, 1996).

Tluczek, McKechnie and Lynam (2010) observed that some parents of infants with abnormal NBS for CF used emotional enduring to manage their feelings. Morse (2000; 2001) described this type of coping as an adaptive attempt to control or suppress intense negative feelings. However, according to emotion regulation theory, such strategies actually deplete mental reserves from other cognitive functions, such as attention and memory (Gross, 1999; Richards & Gross, 2000). Therefore, parents' attempts to regulate their feelings at the time of their infant's sweat test appointment can reduce their cognitive functioning, which is already potentially compromised by sleep deprivation and depressive symptoms. Decreased cognitive functioning can diminish understanding or retention of genetic information. Thus, this research emphasized the importance of devising interventions to address potential risk factors imposed by psychological distress and sleep deprivation.

### Person-Centered Theory

To address parents' emotional needs, we incorporated the tenets of Person-Centered Psychotherapy (Rogers, 1965), a framework embraced by many genetic counseling programs (McCarthy Veach et al., 2003; Weil, 2000). Adopting an attitude of unconditional positive regard, empathy, and respect for personal autonomy creates a psychologically supportive climate in which parents feel comfortable sharing concerns. Rather than ignoring, or being passive to, parents' emotional status, genetic counselors acknowledge and encourage parents to express their feelings, while employing micro skills to reduce parental distress if needed. However, it should have the added benefit of improving parents' cognitive capacities, and preparing them psychologically to learn new information. Consequently, assessment of parents' emotional state and degree of fatigue by listening and responding represent critical components of the approach.

### Tailored Care

Although the principles of family-centered care, outlined in Table 2 (Johnson, Jeppson, & Redburn, 1992) and family-centered genetic counseling (Eunpu, 2010) have long been regarded as the norm, the concept of patient-centered care is a more recent development

(Mead & Bower, 2000;Lutz & Bowers, 2000;Lauver et al., 2002). Patient-centered interventions are those in which "(a) the content of an intervention is selected to address salient characteristics of patients' experiences (e.g., beliefs) or (b) the intervention is responsive to patients' goals or preferences" (Lauver et al., 2002). The degree to which patient-centered interventions are customized varies. For example, "targeted" interventions usually address only one patient characteristic, e.g., diagnosis. "Tailored" interventions typically focus on several areas, e.g. parental distress, knowledge, and learning style. Highly "individualized" interventions can have an infinite number of customized elements.

Given the empirical support for patient-centered intervention models (Aragon et al., 2010; Matloff et al., 2006; Ryan et al., 2002; Wallace et al., 2009), in addition to the family-centered principles already espoused by genetic counselors, we incorporated the tenets of patient-centered care to tailor sessions based on parent preferences as well as genetic counselor appraisals of parents' needs in four a priori domains: (a) parent psychological well-being, (b) parent prior knowledge, (c) infant health and NBS results, as well as (d) parent comprehension of new information.

### Adult Learning Theory

Recognizing the importance of parents' understanding and retaining complicated genetic information, we incorporated andragogical principles of adult learning proposed by Knowles, Holton, and Swanson (2005). In this theory, parents are viewed as self-directed learners motivated to gain knowledge when they understand the underlying rationale. The counselor's role is to guide and facilitate the learning process by following the client's lead in counseling. Elements central to adult learning theory include (a) preparing the learner by setting realistic expectations, (b) creating a climate conducive to learning, (c) mutually planning the session, (d) identifying the learner's informational needs, (e) engaging the learner in activities that advance their understanding of critical concepts, and (f) evaluating the learner's acquisition of new knowledge or skills.

## Developing the Model

### Initial Phase

The new approach was developed as part of a two-phase exploratory project approved by the institutional review boards at each of the three pilot sites. In the first phase, the principal investigator (PI) videotaped counseling sessions and then reviewed the taped sessions with 4 parents in their homes two weeks later. The intent was to gain insights about parents' perspectives regarding specific counseling techniques. These video-playback sessions offered valuable information about how parents interpreted the counselor's attempts to provide information. For example, before the diagnostic test results were known, parents asked counselors, "What will happen if s/he has CF?" When counselors responded in kind with, "S/he will need… and you will need to do…" and provided detailed information about the pathophysiology and treatment for CF, parents thought that the counselor believed their infant would be diagnosed with CF. Parents offered suggestions about how counselors could tailor their approaches to each parent's needs and preferences.

### Second Phase

During the second phase, a genetic counseling consortium was formed to advise the PI in the design of the new model. This group was comprised of one genetic counselor from each of the five sweat testing centers in Wisconsin, the director of the University of Wisconsin Genetic Counselor Training Program, as well as the principal investigator (PI), whose background includes nursing and counseling psychology. During the consortium's initial meeting, members discussed the special challenges of counseling distressed parents,

engaged in reflective appraisals of current genetic counseling practices, reviewed the research literature, and explored ways to better address the unique needs of parents in the context of abnormal NBS results. The group adopted theories to inform the model, delineated objectives to guide the intervention, identified specific counseling psychology micro skills to accomplish the goals, and discussed strategies to overcome potential barriers to implementation. Micro skills are the subset of communication techniques that emphasize listening and responding to clients (Eunpu, 2010; Ivey & Ivey, 2003; Weil, 2000). Based on our review of research, we concluded that our intervention would combine tailored and family-centered principles responsive to parents' psychosocial issues as well as their needs for factual information. The resulting model incorporates relevant theory and research from communication arts, emotion regulation, genetic counseling, nursing, and psychology. Listening to parents is the essential thread woven throughout each component of this model. As Mozdzierz, Peluso, and Lisiecki (2009) explain, to effectively respond to parents' needs, counselors must use their "ears, eyes, and open minds" to really hear the feelings, content, inferences, and what is left unsaid in parents' communications.

### Pilot Testing the Model

Three certified sweat testing sites in Wisconsin with the highest patient volumes piloted the new model over a 6 month period with 34 families. These three medical centers also provided us an opportunity to test the model with diverse populations in urban and rural communities. Each counselor was board certified by the American Board of Genetic Counseling and had between five and fourteen years of experience counseling in the context of NBS for CF. At each site, a counselor met with parents during the sweat test appointment, before their infant's sweat test result was known. At two sites counselors also met with parents immediately after the sweat test to discuss results. At the third site, results were shared by telephone on the same day as testing.

During the early implementation stage, the PI met individually with each counselor at the pilot sites to review videotapes of their counseling sessions, identify counselor strengths, and explore additional opportunities for tailored interventions. After 6 months, the consortium reconvened to evaluate the process and refine the model. Members reviewed video and audio recordings with exemplars of micro skills and offered recommendations about modifications based on their experiences with implementing the model. Counselors at pilot sites reported that the model was feasible requiring only minor modifications to current practice.

## Components of the Tailored Family-Centered Model

The final model, referred to as a tailored family-centered approach to genetic counseling, is designed to (a) minimize parents' emotional distress, (b) facilitate parents' understanding of test results and genetics of CF, (c) increase parents' capacities to use genetic information resulting from the NBS, and (d) enhance parents' satisfaction with genetic counseling and NBS process. To achieve these outcomes, counselors engage in an iterative process of evaluating parents' needs and tailoring responses accordingly. Corresponding assessment and intervention domains include the therapeutic environment, parents' and infants' emotional needs, parents' informational needs, and the development of a follow-up plan.

### Creating a Therapeutic Environment

First, counselors create a psychologically and physically inviting environment. Attending to the family's physical comfort helps parents relax. Counselors establish rapport with parents by communicating genuine interest in parents' concerns and getting to know them as individuals. Inquiring about their well-being imparts a sense of compassion and caring

(Evans, 2006; Ivey & Ivey, 2003; Wright & Leahey, 2005). See Table 3 for descriptions and examples of counseling micro skills.

Minimally, the physical setting consists of a private space to protect parents' confidentiality while discussing personal health information. Ideally, this space contains comfortable furniture and provisions for meeting parent, infant and other family member needs, e.g. feeding and diaper changing. There should also be sufficient room to accommodate the infant's belongings, e.g., car seat or stroller. To minimize distractions from siblings, parents are encouraged when scheduling the sweat test appointment to either arrange in-home child care for their other children or bring another adult to supervise siblings during the appointment. A "distraction kit" containing washable toys serves as a diversion for the times when siblings are present during clinic appointments (Evans, 2006).

Counselors demonstrate respect for individual autonomy by following parents' lead in setting the agenda for the session (Weil, 2000). Within the tailored family-centered model, counselors collaborate with parents by explicitly addressing parents' expressed wishes. Although the agenda is established early in the session, counselors remain flexible about what and when particular information is discussed and readjust the agenda in response to parents' wishes. Parents view this personalized approach very favorably, as one parent stated, "I felt like I was in charge of what was discussed" rather than a passive recipient of a "predetermined speech that everyone receives."

Throughout the session counselors infuse cultural sensitivity in their interactions with all families, regardless of whether or not parents' demographics match those of the counselor (Lewis, 2010). This sensitivity involves remaining cognizant of one's own socio-cultural perspective and maintaining compassionate curiosity about parents' beliefs and values, particularly relative to genetics.

## Addressing Parents' Emotional Needs

Within the tailored family-centered model, counselors assess parents' emotional state early in the session by asking how long parents waited for the sweat test appointment and what the wait was like for them emotionally. In addition to being worried about the infant's abnormal NBS results, many women experience mild depressive symptoms during the early postpartum weeks commonly referred to as the "baby blues." About 10 to 15 % of women have more severe clinical levels of depression (Almond, 2009). Sleep deprivation and an abnormal NBS for CF can exacerbate preexisting depressive symptoms. Therefore, the assessment includes an inquiry about how much sleep parents have had and whether any lack of sleep was due to the infant's needs or their concerns about the infant. When parents report sleep deprivation, counselors appraise parents' understanding more frequently. If postpartum depression is suspected, counselors promptly refer the family for mental health services, symptoms of which are outlined in Table 4. It is equally important to avoid making assumptions that all parents are worried or distressed, thereby unwittingly sending a message suggesting that the infant will be diagnosed with CF or that the parents should be worried.

In addition to direct inquiry about parents' emotional states, throughout the session counselors observe parents' non-verbal signs of emotional reactions to information shared during the counseling process (Weil, 2000). Signs of distress can be recognized by subtle changes in a parent's facial expressions, breathing rate, tone of voice, gaze, posture, or their eyes welling with tears (Ivey & Ivey, 2003). Using parents' subjective reports and the counselor's objective observations, counselors form hypotheses about parents' emotional states and then test these hypotheses by explicitly asking parents about them, e.g., "It sounds like you are feeling fairly optimistic that your baby does not have CF; is that right?" or "I'm

wondering if you've been worried that your baby might have CF." This type of clarification offers the clinician direction about how to proceed with counseling and demonstrates that the counselor is listening to the parents.

Parents present with a range of emotional responses to information that can include stoic restraint, sad tearfulness, calm confidence, and nervous laughter. Throughout the session counselors attend to parents' emotional states by noticing subtle changes in parents' facial expressions (Weil, 2000) and checking with the parents about their reactions to whatever was just discussed. Tluczek and colleagues (2006) found that parents appreciated counselors' verbal and non-verbal communications that conveyed empathy for parents' circumstances and a sense of optimism about their children's future, regardless of test outcome. Empathy and compassion are conveyed explicitly through language and implicitly through facial expressions, tone of voice, eye contact, and body language (Evans, 2006; Ivey & Ivey, 2003). Counselors can also reflect the infant's feelings, e.g., for a crying infant "She seems so unhappy" or for a sleeping or a calm alert infant "He looks really content in your arms." If parents become tearful, counselors might intervene by pausing and asking parents if they would like to share what is bothering them. Emotion-focused interventions include reflecting parents' feelings, redirecting the conversation to address the source of their concerns, normalizing parents' reactions, pausing to allow time for them to regain their composure, and instilling realistic hope (Table 3).

Parents have identified hope as a critical component in genetic counseling (Tluczek et al., 2006; Tluczek et al., 2010). Within the tailored family-centered model, counselors make use of their assessment data, e.g. family history, infants' good health, and the infants' NBS results to offer parents hope. This hope can be conveyed by sharing factual information, e.g. 97% of infants with one *CFTR* mutation identified through NBS have normal sweat test results and do not have CF. Counselors can also note the benefits of early diagnosis and treatment in preventing complications and maintaining a child's good health. If a type IV–V *CFTR* mutation has been found, the counselor might explain that if diagnosed with CF, the child is likely to have fewer symptoms, or a later onset, and describe the spectrum of CF phenotypes. Some parents find it helpful to know that advancements in research and treatment continue to improve both the quality of life and longevity for people with CF. Finally, the counselor is ever cautious to avoid implying negative test results before actual results are available.

Anecdotal reports suggest that parents find these emotionally responsive techniques very reassuring. One mother described her experience of the approach in the following quote, "I was crying… She said that she was there for us. It was emotionally stressful and she was comforting." Another parent explained, "The counselor seemed hopeful and that made me feel hopeful."

### Addressing Parents' Informational Needs

Understanding parents' perceptions about the NBS results is central to tailoring the educational component of the tailored family-centered intervention. Based on previous research findings (Tluczek et al., 2006), this assessment involves a highly interactive discussion with parents about their knowledge about CF, understanding of the NBS procedures, and their perceptions of the likelihood that their infant has CF. Encouraging parents to share their personal stories provides the counselor a deeper appreciation for the multiple variables that shape a particular parent's interpretation of test results, e.g., personal experiences with individuals who have CF, information from the popular media including the internet, communications with their primary care providers, conversations with friends or family, their infant's health and perinatal history, as well as the parents' science background or their familiarity with genetics (Smith & Pollin, 2009). Collecting information

about the family pedigree can offer insights about parents' risk perceptions. There is evidence that parents' perceptions of genetic risk are more heavily influenced by the presence or absence of the disease in their family history than by the results of genetic testing (Tarini, Singer, Clark, & Davis, 2008). It is also important for counselors to appraise a parent's preferred learning style by directly asking them how they best learn new material. Jedlicka-Kohler et al. (1996) found most parents preferred a combination of reading, viewing, and listening. Thus, within the model, counselors use data from multiple sources to formulate an educational plan unique to each family.

The model embraces three quality indicators of optimal parent-provider communication recommended by Farrell, La Pean, and Ladouceur (2005). First, it includes key content, e.g. note infant's good health; high probability for a normal sweat test; high probability child is a CF carrier; non-association of carrier status with medical problems; siblings may also be carriers; parental testing is available; other relatives might benefit from learning about results. Second, facts supporting the prospect of favorable results should be shared early in the session, called "instilling hope" in our model. Third, maintain a low ratio (< 1:1) of the amount of standardized background information relative to the amount of individualized information, called "sharing talk time" in our model. La Pean and Farrell (2005) also caution against misleading statements, e.g. inferences that NBS results indicate the presence of illness. We also recommend avoiding the use of medical jargon (Farrell, Deuster, Donovan, & Christopher, 2008) and encourage the use of clearly understandable words like "normal" and "abnormal" rather than "negative" and "positive" to explain test results because the lay connotation of the latter terms frequently contradicts the clinical meaning.

Within the tailored family-centered model, the type and amount of information is titrated to individual parent requests. Counselors match the level of technical description in verbal explanations to the parents' baseline knowledge and explicit request for details (Tluczek et al., 2006; Maynard, 2003). Although counselors tailor the process to parents' expressed wishes, the core content listed in Table 5 is discussed with each family. Recognizing that parents' information processing capacities could be adversely affected by strong emotions and limited sleep, counselors assess parents' comprehension by using teach-back techniques, clarifying shared language, and pacing information delivery to match parents' cadence throughout the session. Teach-back techniques, which involve asking parents to use their own words to illustrate their understanding of the material discussed, are important measures to assure comprehension (Kripalani, Bengtzen, Henderson, & Jacobson, 2008; Lorenzen, Melby, & Earles, 2008).

Although the teach-back is a practical measure for assessing parents' comprehension of essential facts, it also provides valuable insights about which aspects of the counseling parents believe to be most salient. Clarification of shared language facilitates parental comprehension while building rapport. Pacing the verbal delivery to match a parent's discourse also promotes comprehension. When answering parents' questions about "what will happen if s/he has CF," counselors limit the amount of detail about CF or related treatment. They keep the discussion at a distance from the child by using the third person rather than the child's name, e.g., "Children with CF …" instead of "Your child will…" or "(Child's name) will…" Parents have expressed an appreciation for this tailored approach as illustrated in the following quote, "She (the counselor) explained the likelihood she (their child) would have it (CF). She talked a little about the future, but allowed us to control the conversation, what we wanted to know, specific to our child. She didn't scare us, didn't discuss CF-related things unless we asked."

Finally, recognizing that some disruptions are unavoidable, measures can be taken to minimize their impact. For example, counselors pause during the disruption and assist

parents in resolving the disruption, e.g. letting parents feed a hungry infant, or waiting until the sweat test technician completes a task before resuming counseling. Counselors are careful to avoid discussing important content while other environmental demands compete for parents' attention. After the distraction is resolved, counselors reiterate key points discussed prior to the disruption and assess parents' understanding before proceeding to new content.

### Developing a Follow-up Plan

A follow-up plan is collaboratively developed with parents to provide them access to additional resources as needed. The plan includes mailing parents a letter summarizing the session, the counselor's contact information, and who to contact if additional questions or concerns arise. It might also include details about genetic testing for parents. When discussing the availability of carrier testing, counselors take a non-directive approach (Kessler, 2001), making it clear that carrier testing is optional. Parents may need time to contemplate the potential risks and value of such testing for themselves and their families (Archibald et al., 2009). A variety of variables contribute to whether, and to whom, parents share test results (Ormond, Mills, Lester, & Ross, 2003). Parents may appreciate knowing that they can re-contact counselors for assistance with communicating test results or sharing genetic information with other relatives. The final plan matches expressed needs and unique situations.

## Discussion

We developed an innovative tailored family-centered approach to genetic counseling that draws from multiple disciplines to deepen and expand upon the contemporary psychosocial and psycho-educational genetic counseling literature. The lively discussions and diverse perspectives shared among the multidisciplinary team members informed the model by addressing potential issues associated with implementing the model, respecting stylist differences in counseling, and assuring the model's practical application across settings. Counselors found that labeling strategies used in their communications with families provide a valuable common language for counselors to discuss within a self-reflective practice and for teaching students. The resulting model systematically employs specific counseling strategies to operationalize person/patient-centered principles by tailoring counseling to match parents' needs for information and emotional support.

Based on the feedback from the genetic counselors and anecdotal parent reports, the tailored family-centered approach appears to be well-received and feasible to implement. Data collection is currently underway to evaluate the initial efficacy of the new model in comparison with standard approaches. This evaluation will include parents' levels of emotional distress pre/post-counseling, knowledge of the CF genetics and test results pre/post-counseling, and satisfaction with counseling.

With the national mandate for NBS for CF, and the well-documented health benefits of early intervention for affected infants in this population, this new model could inform NBS programs throughout the US. Additionally, it will help fill the gap in the existing empirical literature regarding evidence-based genetic counseling practices and standards of care. The micro skills and components of the model offer a framework for other areas of genetic counseling practice as well as the professional development of genetic counselors and advanced practice nurses in genetics. Although the model is specific to the nuances of genetic counseling for NBS, many of the micro skills and the process taken in developing the model offer clinicians and researchers guidance for designing similar evidence-based models of genetic counseling and psychosocial education.

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**Table 1**

Cystic Fibrosis Carrier Frequency and Condition Prevalence

| Race/Ethnicity | Carrier Frequency[a] | Condition Prevalence Per Live Births[b] |
|---|---|---|
| European American | 1 in 29 | 1 in 2,500–3,500 |
| Hispanic American | 1 in 46 | 1 in 4,000–10,000 |
| African American | 1 in 65 | 1 in 15,000–20,000 |
| Asian American | 1 in 90 | 1 in 100,000 |

[a] CFF, 2010

[b] Voter and Ren, 2008

**Table 2**

American Association for the Care of Children's Health Principles for Family-Centered Care

| | |
|---|---|
| **1** | Value parents as constant in their child's life |
| **2** | Support parent-provider collaboration |
| **3** | Recognize the individuality of parents' coping styles |
| **4** | Keep parents well-informed |
| **5** | Encourage parent-to-parent support |
| **6** | Accommodate the child's developmental needs |
| **7** | Address parents' emotional needs |
| **8** | Develop services that are flexible and responsive to parents' needs. |

(Johnson, Jeppson & Redburn, 1992)

## Table 3

Counseling Micro Skills

| Micro Skill | Brief Description | Examples |
|---|---|---|
| **Core Skills Used Throughout Session** | | |
| Listens to parents | Listens for emotional, factual and inferred content of parents' communications. Uses verbal and non-verbal responses to indicate that they are listening to parents. | "Mm hmm." Or "Tell me more about that." Or quietly nods head. |
| Makes eye contact during discussion | Uses culturally appropriate eye contact with parents to facilitate conversation. | |
| Shows range of affect | Facial expressions suggest empathy, interest, and concern in response to parents' needs. | |
| Tone of voice communicates sincerity | Shows range of intonation to communicate sincerity, warmth, interest and concern in response to parents' needs. | |
| Uses attentive body language | Uses culturally appropriate facilitative posture and hand gestures to communicate interest and encourage parent participation. | |
| Shares talk time | Engages both parents to become active participants in the session so that parents and counselor contribute to conversation. | "So (mother's name) described what she would like to know more about; (father's name), I'm wondering about you. What questions do you have?" |
| Uses open-ended questions | Uses non-directed and semi-directed questions that prompt parents to share their thoughts, feelings, questions, or concerns. | Non-directed: "How can I best help you today?" Semi-directed: "What did your doctor tell you about (infant's name) NBS test results?" |
| Uses closed-ended questions | Uses questions that have one or two word responses when brevity is called for. | "How many children do you have?" "Are you interested in information about carrier testing? |
| Prompts questions | Uses verbal and non-verbal prompts to facilitate dialogue. Counselor remains alert for changes in parent's facial expression, pauses, and check to see if parents have questions. | "What questions do you have?" "Please feel free to ask questions." "I'm wondering if you have some questions about what we just discussed." |
| Uses minimal encouragers | Uses verbal and non-verbal prompts to reinforce and promote parents' contribution to the conversation | Nodding head "Uh ha," "mm" |
| Paraphrases comments | Uses parents' key words and/or summarizes. Comments confirm that counselor understands parent's perspective. | "It sounds like you learned from your primary physician what CF is and that it affects the lungs and digestive system. That's a great start for our discussion today." |
| Minimizes major disruptions | Stops counseling session during major disruptions. | "I'm just going to stop right now and let the technician collect the sweat pads from (baby's name)." |
| Minimizes minor and moderate disruptions | Uses pauses and other methods to address minor and moderate disruptions and does not amplify them. | "I apologize for that interruption; let's go back to where we were before the knock on the door. We were talking about…" |
| **Skills Associated with Emotional Support** | | |
| Supports parents' story-telling | Encourages parents to share stories about their experience with the NBS process and other relevant life experience. | "How was (infant's name) birth?" "What has the wait for the sweat test appointment been like for you?" "What do you know about CF?" |
| Assesses parents emotional needs | Asks parents if they have worries about their infant; explores specific sources of concerns including but not limited to the NBS results. Uses worry scale. | "On a scale of 0 to 10 with 0 meaning not worried at all and10 being the most worried ever, I'm wondering how you are feeling at this time." |
| Reflects parents' feelings | Labels feeling(s) that parents appear to be expressing verbally or non-verbally. | "You seem very worried." "It sounds like you're feeling pretty optimistic about how the sweat test will turn out today." |

| Micro Skill | Brief Description | Examples |
|---|---|---|
| Shows empathy | Uses words, tone of voice, and facial expressions to communicate an appreciation for parents' emotions. | Mirrors parents' facial expressions of smiling or seriousness. "Given your past experiences, I can understand why you might feel…" |
| Normalizes parents' reported experience | Explains that many parents share similar feelings, concerns, responses or behaviors when in this, or similar, situations. | "Many parents come to the sweat test appointment with reactions much like yours." "The majority of parents that we see are not familiar with CF." |
| Uses silence effectively | Monitor's parents non-verbal language for cues that they need a moment to gather their thoughts. | Stops talking to allow parents time to formulate questions and express their feelings. |
| Clarifies meaning | Checks with parents to be sure that the counselor and parents share the same meaning and understanding of comments or questions. | "You said that you think your baby has been making odd noises during sleep. I'm wondering if you think it might be related to CF?" |
| Instills sense of hopefulness | Identifies contextual factors that offer reasons for optimism regarding infant's future. | "Although we still need the test results, everything that you have described about your baby sounds typical for a healthy newborn." |
| **Skills associated with Parent Education** | | |
| Foreshadows events | Outlines sequence of events that will occur during and/or after the session. | "While the technician is working on the sweat test analysis, I'd like to answer your questions and discuss the newborn screening process." Or "I will call you before 4:00 this afternoon with the sweat test results." |
| Gathers pertinent family information (brief pedigree) | Gathers family history related to CF, health of children, date and place of other children's births and Family history of CF or CF-related symptoms. | "Has anybody in your families ever been diagnosed with CF or needed to have a sweat test?" |
| Assesses parents' learning style | Asks parents how they learn best, e.g. visual, verbal, combination. | "I'm wondering how you learn new information best, such as reading, listening, or looking at visual aids." |
| Matches teaching method to parents' learning style | Uses visual aids, verbal explanations and/or hands-on activities to best fit the parents' needs. | "As you can see here in this diagram…" "I'm going to illustrate the genetics of CF in a drawing." |
| Matches language to parents' understanding | Defines technical terms, offers basic background information about genetics, and avoids medical jargon unless parents demonstrate clear understanding. | "A mutation is a change or 'misspelling' in a gene." "The sweat test was normal." |
| Provides verbal explanations | Provides clear, organized, and easily understandable verbal information. | "Today's sweat test will measure the amount of chloride or salt in (baby's name) sweat. Babies with CF have about 5 times more chloride in their sweat than babies that don't have CF. |
| Scaffolds information | Builds on parents' previous knowledge. | "So you read that the risk of having a child with CF is 1 in 4, if both parents are CF carriers. That's right. CF is a recessive disorder. That means that a baby must receive one changed CF gene from each parent in order to have the condition we call CF. |
| Normalizes carrier status | Explains how common genetic variations are and that being a CF carrier creates no health concerns. | "One in twenty-nine people whose ancestors came from Northern Europe is a CF carrier." or "At least one parent is a carrier and he or she is healthy." |
| Uses examples or metaphors | Uses illustrations or comparisons from everyday life to explain concepts to aid parents' understanding. | "Chromosomes are like our body's cookbooks. Genes are the individual recipes in those cookbooks." |
| Uses non-examples appropriately | States what something is not to aid the parents' understanding. | "CF carriers have one defective gene, but they do not have the disease" "Newborn screening is not designed to pick up all carriers of CF." |
| Conducts a teach back | Asks parents to explain their understanding of information using their own words. If misunderstanding is noted, a teach-to-goal is used, restating the information and the parent is again questioned to confirm comprehension. | "I'd like to ask you a few questions to be sure that I adequately explained all the information we just discussed." |

| Micro Skill | Brief Description | Examples |
|---|---|---|
| Summarizes information | Captures key points of the discussion in a clear and concise synopsis. | "Your baby's sweat test was normal. He does not have CF, but we know he is a carrier. This will not affect his health in any way." |
| Takes a non-directive approach | Remains neutral when presenting parents options for further carrier testing. | "Think about whether having carrier testing might be helpful for you and your future decisions" or "Some people choose to have carrier testing, and some choose not to. The decision is individual and I encourage you to think about what is best for you." |

**Table 4**

Symptoms of Postpartum Depression

| Five or more of the following symptoms suggest clinical levels of depression: |
|---|
| • Depressed mood most of time |
| • Loss of interest in pleasurable activities |
| • Feelings of guilt or worthlessness |
| • Indecisiveness or concentration problems |
| • Hopelessness |
| • Restlessness and agitation |
| • Disturbed sleep |
| • Low energy |
| • Change in appetite |
| • Weight loss or gain |
| • Persistent thoughts of death |
| • Thoughts of harm to self or others |

(American Psychiatric Association, 2000)

**Table 5**

Core Content

| Although genetic counseling is tailored to parents expressed wishes, the following content is typically discussed with each family |
|---|
| • Newborn screening procedures |
| • Interpretation of infant's NBS results |
| • Explanation about the sweat test |
| • Infant's sweat test result |
| • CF genetics |
| • Implications of NBS and sweat test results for infant |
| • Implications of NBS and sweat test results for parents and other family members |
| • Family history and/or pedigree |
| • Carrier testing options for parents and other family members |
| • Basic medical information about CF |