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The Work of Managing Phenylketonuria and Breastfeeding

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Abstract

Purpose: To describe the demands of breastfeeding in the context of Phenylketonuria (PKU) and mothers' efforts to manage the complicated feeding regimen.

Design and Methods: A qualitative design and methods were used to interview mothers (n=10) who had previously completed an online international survey about their feeding experiences with their infants with PKU. Mothers were recruited to participate in in-depth telephone interviews to further explore their breastfeeding experiences of their infants with PKU. Mothers were eligible for participating in the study if they were 21 years of age or older, had initiated breastfeeding an infant with PKU, and the infant was less than three years of age. There were no restrictions on the length of time mothers' breastfed.

Results: *The work of adapting breastfeeding to maintain phenylalanine levels* was the central theme of the breastfeeding experience. Mothers described two major components of the work and effort needed to breastfeed infants with PKU: maintaining phenylalanine levels and adapting breastfeeding.

Practice Implications: Mothers used various strategies to sustain breastfeeding while adhering to the PKU management plan in order to successfully manage their infants' PKU and breastfeeding.

Keywords: Breastfeeding; Inborn error of metabolism; Infant-newborn; Lactation; Phenylalanine; Phenylketonuria

Abbreviations

IRB: Institutional Review Board; kg: Kilogram; PAH: Phenylalanine Hydroxylase; Phe: Phenylalanine; PKU: Phenylketonuria; µmol/L: Micromole per Liter

Introduction

Phenylketonuria (PKU) is the most common inborn error in amino acid metabolism [1]. Although the occurrence of PKU varies across groups, it is present in every ethnic and racial group. Normally, the enzyme Phenylalanine Hydroxylase (PAH) metabolizes Phenylalanine (Phe) into tyrosine in the liver. In PKU, PAH is defective and results

in excessive concentrations of Phe in the blood and eventually toxic concentrations in the brain. The different mutations in the gene-coding PAH enzyme determines the clinical presentation and severity of the disorder. The combinations of the different mutations result in varying degrees of residual PAH activity [2]. The residual PAH enzyme reflects Phe tolerance.

Untreated PKU results in the slow insidious loss of neurocognitive skills resulting in permanent cognitive impairment as the child grows. Classical signs include eczema like skin rash, excessive restlessness, and a "musty" or "mousy" odor of the body, urine and perspiration due to phenylacetate accumulation. In addition, individuals affected with PKU have a lighter coloration of skin, hair, and eyes. Developmental problems, irritable behavior, gait disturbances, psychiatric symptoms, and impaired cognition become clinically present with increasing toxic accumulation within the body and brain.

Newborns with PKU appear normal at birth. In the past as they grew, these children were identified after the insidious insult had occurred and they had suffered permanent neurocognitive sequela [3]. Robert Guthrie's introduction of a simple diagnostic screening test for PKU allowed for mass screening and identification of PKU before the presence of clinical signs became detectable [4]. Early identification and prompt dietary intervention with a Phe-restricted diet means children with PKU can avoid the neurocognitive insult. The Phe-restricted diet limits daily dietary Phe intake and requires a supplementation of synthetic amino acid fortified medical foods and beverages to ensure low-Phe to Phe- free protein sources with adequate nutritional value for normal growth and development.

During the early years of PKU management, infants diagnosed with PKU were immediately weaned from breastfeeding. The dietary treatment consisted of a low-Phe medical formula in conjunction with standard commercial infant formula. This combination was believed to be the only effective way to manage the infant's Phe intake and allow for precise titration and measurement of Phe based on serial infant heel sticks [5]. This management strategy thus precluded breastfeeding infants with PKU [6-9].

However, breast milk has many advantages when offered as a primary source of nutrition for infants with PKU. These include the normal benefits of breastfeeding and breast milk for any infant

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Table 1: Descriptive Characteristics of Mothers and Infants (*n*=10).

Demographic Characteristics		Sample Characteristics
Ethnicity	Asian/Pacific Islander	1
	Caucasian	8
	Hispanic/Latino	1
Marital Status	Married	9
	Divorced	1
Gravida	Primipara	6
Previous breastfeeding experience	Yes	4
Education	Associates Degree	1
	Bachelors Degree	3
	Masters Degree	5
	Professional Degree	1
Employment, hr/wk	0	3
	≤ 32	1
	36 - 40	4
	> 40	2
Gross Annual Household Income, US Dollar	25,001 – 50,000	1
	50,001 – 75, 000	1
	75,001 – 100,000	3
	100,001 – 150,000	1
	> 150,000	4
Community Population Size	A town with a population < 10,000	2
	A city with a population between 10,000 - 50,000	6
	A metropolitan area with a population between 500,000 – 1 million	1
	A metropolitan area with a population > 1 million	1
Country of Residence	United States (7 states)	7
	Canada (3 provinces)	3
Maternal Age, mean (range) years		32.4 ± 2.8 (30-39)
Infant birth weight, mean (range), kg		3.7 ± 0.48 (3–4)
Phe level at diagnosis, mean (range), μmol/L		504.5 ± 143.6 (360-720)
Age in days at diagnosis, mean (range)		2± 1 (1–4)
Age in months at interview with mothers, mean (range)		20±8 (11–35)
Duration of breastfeeding, mean (range), months		11 ±7.3 (6–30)

Abbreviations: Phe: Phenylalanine.

[10,11]. In addition, Phe concentration is lower in mature breast milk (2,482 μmol/L) [12,13] than standard commercial infant formula, such as Mead Johnson Enfamil® (4,419 μmol/L) [8] making it possible to utilize a higher proportion of natural protein. For infants with PKU, Phe restriction means breast milk must be limited and adjusted based upon weekly blood Phe levels. Breast milk intake, whether from breastfeeding or breast milk feedings, must be adapted to maintain desired Phe levels (120-360 μmol/L) [14,15] that are dependent upon the infant's residual PAH activity. Therefore, the addition of Phe-free medical formula is essential in order to provide adequate caloric and protein intake while maintaining Phe restriction for normal cognitive and behavioral development.

Results from numerous studies [8, 9,16-20] consistently reported breast milk supplemented with Phe-free medical formula was an acceptable dietary treatment for infants with PKU because Phe levels could be maintained within the desired range despite the challenge that Phe intake was difficult to determine precisely in the context of breastfeeding. Yet, there are no universal approach to breastfeeding infants with PKU and supplementing with Phe-free medical formula. Consequently, mothers typically offer breast milk by one of three options based upon recommendations from their metabolic health care providers, usually a dietitian. Mothers provided 1) a fixed volume of Phe-free medical beverage followed by breastfeeding until satiety [9,21]; 2) a fixed volume of expressed breast milk or a fixed time limitation of breastfeeding followed by Phe-free medical beverage until satiety [22]; and 3) alternating feedings between breastfeeding and Phe-free medical beverage [19].

Table 2: Sample Questions from Interview Guide.

Family Management Topic	Question
Defining the Situation	
Self	Tell me how the initial few days of breastfeeding went with this infant.
Family	Tell me about your families and friends reaction to your infant's diagnosis of PKU.
Child	Tell me about your infant who has PKU.
Management Behaviors	
Self	Tell me what happens on a day-to-day basis with breastfeeding your infant with PKU after diagnosis.
Family	Tell me about what your family and friends did that helped or did not help you in managing your infant's PKU.
Child	Tell me about how you introduced the low-phenylalanine or phenylalanine-free medical formula to your infant.
Perceived Consequences	
Self	Tell me about your decision to continue to breastfeed your infant with PKU after diagnosis.
Family	Tell me about what happens on a day-to-day basis with your family due to breastfeeding an infant with PKU.
Child	Tell me about how you think PKU will affect your child as he/she grows.

PKU: Phenylketonuria

Table 3: Themes for the Work of Adapting Breastfeeding to Maintain Phenylalanine Levels.

Central Theme	Theme	Subtheme
The work of adapting breastfeeding to	Maintaining Phe levels	Orchestrating implementation of the PKU treatment plan
maintain phenylalanine		Documenting daily Phe intake
levels		Collecting blood samples to monitor Phe levels
		Adjusting to the every-changing treatment plan
		Developing a relationship with the metabolic team
	Adapting breastfeeding	Patience combining breast and bottle feedings
		Maintaining a milk supply
		Persevering to breastfeed

Phe: Phenylalanine; PKU: Phenylketonuria.

Studies reveal few mothers of infants with PKU persist in breastfeeding after diagnosis [7,9,16,18,19,23-27]. Overall, the incidence, prevalence, and duration of breastfeeding when infants have PKU are lower when compared to other full-term infants who do not have PKU. For example, in an Israeli study, breastfeeding rates were significantly lower in infants with PKU than in the general Israeli population [26]. In a more recent study involving mothers from the United States and Canada, the prevalence of breastfeeding infants with PKU before and after diagnosis revealed significantly fewer mothers were breastfeeding after diagnosis [25]. However, there are no published reports describing mothers' experiences breastfeeding infants with PKU to help understand how women sustain breastfeeding within this unique population. Therefore, the purpose of this study was to examine the demands of breastfeeding in the context of PKU and mothers' efforts to manage the complex feeding regimen.

Methods

Design

This study utilized a two-phase mixed methodology. Phase one was the quantitative phase with an Internet survey completed by mothers residing in the United States and Canada about their experiences feeding infants with PKU. Mothers were recruited from a variety of social media options including the PKU listserv, Facebook, Twitter, and national/regional PKU associations. The goal of the quantitative phase was to explore the rate of breastfeeding before and after diagnosis of PKU in the sample. In the phase one sample, 103 mothers had one child with PKU while 16 mothers had two or more children with PKU. Overall, mothers with one child with PKU were in their thirties (n=64, mode=34) but ranged from age 21 to 63 years, were married/partnered, and well-educated as all mothers were high school graduates and more than two-thirds (n=72) were college graduates. Initially 89 mothers with one child breastfed, but after the PKU diagnosis, only 72 mothers continued to breastfeed. This decrease in breastfeeding was significant (McNemar's $\chi 2 = 30.333$, $p < .001$; n = 72 versus n = 89) [25].

Phase two of the study was qualitative telephone interviews with a subset of mothers from phase one to provide a more detailed understanding of the maternal experience of breastfeeding infants with PKU and the meaning that the mothers attributed to their experiences. The study was approved by the institutional review board (IRB) of Oregon Health & Sciences University.

Sample and setting

Mothers were eligible for participating in the qualitative phase two if they were 21 years of age or older and had initiated breastfeeding an infant with PKU. There were no restrictions on the length of time mothers' breastfed. In addition, the breastfed infant with PKU needed to be less than 36 months old at the time of the telephone interview. Of the 103 mothers who participated in the Internet survey

and expressed a willingness to be interviewed, 23 mothers met the sample criteria for phase two of the study. This subset of mothers was predominantly Caucasian, well-educated, in their 30's, and had breastfed for at least four months. Ten mothers were purposely recruited to participate in the telephone interviews to reflect a broad range of demographic variables: 1) urban versus rural residence, 2) ethnicity, 3) gravida status, 4) education, 5) marital status, 6) employment, and 7) household income. Demographic characteristics of the mothers and infants are presented in the Table 1.

Data collection

Mothers who completed the online survey and met the inclusion criteria were informed about the phase two study with an email from the principal investigator. After obtaining participant consent, the principal investigator conducted digitally recorded in-depth telephone interviews at a convenient time of day with each of the mothers. Interviews lasted an average of 92 minutes (range 56 to 125 minutes). Mothers were encouraged to share their thoughts about and experience of breastfeeding their infant with PKU. A list of open ended interview questions guided each telephone interview. As a starting point, mothers were asked to "describe a typical day with your breastfed infant with PKU" to explore their breastfeeding experiences. Subsequent questions focused on gaining a detailed description of what mothers did to manage PKU and breastfeeding and the meanings that they ascribed to their management behaviors. A sample of the questions is provided in Table 2. Probes focused on issues such as learning to care for their child's PKU (e.g. obtaining weekly in-home Phe samples to be sent to the laboratory for Phe analysis, maintaining a milk supply, managing other responsibilities, concerns and stressors, and accessing needed resources). Field notes were compiled immediately after each interview. At the completion of the interview, mothers were sent a $20 gift certificate. Data collection occurred between February 2011 and June 2012.

Data management and analysis

A descriptive qualitative approach was used to complete the analysis and identify themes reflecting mothers' breastfeeding experiences [28-30]. All digitally recorded interviews were transcribed verbatim and checked for accuracy against the recording. Digital recordings were kept confidential with no identifying names or locations used in the written transcripts. Based on multiple reviews of the interview transcripts by the research team, coding categories were identified and defined to insure that codes were applied consistently across interviews to identify commonalities and differences in mother's experiences [28,29]. Thematically similar categories and related data were grouped to create an overarching central theme with subthemes [31].

In this study, rigor was addressed through several techniques. Having one interviewer increased the dependability of the study by assuring a consistent style of interviewing. To address bias, the use of detailed descriptive field notes, including reflection, occurred immediately after each interview. To provide transparent description of the qualitative research steps taken in phase two from inclusion criteria to finalization of the central theme, an audit trail was maintained and included documentation decisions made regarding the coding and analysis of data. Two nurse researchers with expertise in families with children, who have chronic conditions, independently read the transcripts. Differences in coding were conferenced to seek resolution, and the overarching central theme and subthemes were agreed on [32].

Results

The work of adapting breastfeeding to maintain phenylalanine levels

The central theme of the breastfeeding experience that emerged from the interviews was "*the work of adapting breastfeeding to maintain Phe levels*". Mothers described their efforts to master a complex management routine that combined breastfeeding with a special Phe-free medical formula to maintain therapeutic Phe levels. This reflected mothers' efforts to continue breastfeeding in a uniquely challenging situation. Mothers vividly described how their everyday work of simultaneously managing PKU and breastfeeding was tied to their profound commitment to breastfeeding their infant despite PKU [33]. Mothers described two simultaneous aspects of the work of adapting breastfeeding to maintain Phe levels: *maintaining Phe levels and adapting breastfeeding*. The central theme and subthemes are outlined in Table 3.

Maintaining phe levels

The management of PKU was focused on maintaining Phe levels. The work of maintaining Phe levels included an array of responsibilities that concerned all mothers with young infants with PKU: orchestrating implementation of the PKU treatment plan, documenting daily Phe intake, collecting blood samples to monitor Phe levels, adjusting to the ever-changing treatment plan, and developing a positive working relationship with the metabolic team directing care.

Orchestrating implementation of the PKU treatment plan: As these mothers were the primary caregivers for their infants, orchestrating implementation of the PKU treatment plan included many responsibilities. After taking their infant home from the initial metabolic appointment, where they received directions on how to manage breastfeeding and PKU, mothers assumed responsibility for the infant's dietary adherence and managing the life-long chronic condition of PKU. Some mothers assumed responsibility for direct management of PKU while others delegated some activities to others, but they all saw themselves as ultimately responsible for assuring adherence to the prescribed guidelines. Fathers played an important role by assuming responsibility when mothers were busy, as one mother shared: "My husband was always on board and up to speed with what needed to be done . . . . He is fully versed in how to manage the diet that is crucial." Yet, even when others were involved in the care, mothers assumed responsibility for orchestrating care and making sure the special needs of their infants with PKU were met. Mothers developed different methods to educate day care providers and other family members' with explicit information about feeding the infant with PKU. However, constant assessment of the competence of others caring for the infant with PKU was required. Mothers described being satisfied with day care arrangements when they felt their infant's care included appropriate PKU management that followed their detailed instructions for infant feeding.

In contrast, mothers' sense of responsibility was further heightened by their recognition that the ability of some family

members and friends to provide some respite was either limited or lacking. The inability to find respite care was directly linked to the work of managing PKU. Only rarely were grandmothers, sisters, or sitters identified as available to provide child care. Initially, family and friends were described as offering babysitting services; yet, many did not follow through when asked. In addition, mothers described some friends and family who had misperceptions or struggled with implementation of the PKU feeding plan or who minimized the importance of adhering to the prescribed feeding plan. As one mother stated, we did leave our child with family when she was under a year . . . pretty much everyone that we left her with left screwed up the diet somehow . . . . Not that we stopped [leaving her with family] because of that. We did try to limit leaving her to one or two people who could really learn how to manage the diet. Of course, it's hard. It's a hands-on type of thing. It's easy for us because we do it every day; but for those who don't, it's tricky. Consequently, all mothers had concerns about the ability of day care providers, family, and friends to appropriately care for a young infant with PKU.

Documenting daily Phe intake: While orchestrating implementation of the PKU treatment plan, mothers also needed to record their infant's intake of Phe and Phe-free medical formula. The mothers described tracking daily Phe intake was an activity that required ongoing monitoring as well as flexibility and adaptability to insure appropriate PKU management. Mothers had to record the amount of Phe-free medical formula along with breast milk intake from either breastfeeding or bottle feeding expressed breast milk. All mothers developed their own systematic method of tracking their infant's daily Phe intake using some form of food logs and journals. Some mothers transitioned from food journals to Excel spread sheets to using new "apps," such as AccuGo© or the Dietwell© for PKU, available for iPhones, iPod touch, iPad, and Android phones. As one mother shared, if his dietitian wanted me to send his food journal, I would type it up and send it [the food journal] as an attachment in an email. If she wanted multiple dates, I would scan them in. I was always able to track it [Phe] on paper and then I had a binder, but now I track it on my iPhone with an app. Mothers adapted documenting daily Phe intake to their busy lives by increasingly embracing technology.

Collecting blood samples to monitor Phe levels: During the first six months following their child's birth, mothers described learning to perform in-home heel sticks once to twice weekly to collect blood samples that had to be mailed to a laboratory for Phe-level testing. These weekly routine in-home heel sticks provided the metabolic team with blood Phe results upon which treatment plans were constantly revised in order to maintain Phe levels within the desired range (120-360 μmol/L) [14,15]. In order to obtain and send routine blood Phe samples, mothers developed routines for monitoring Phe levels that accommodated other responsibilities and the family's usual schedule. One mother explained how she developed this routine to be less disruptive of family life: "I was drawing the blood on Monday nights, but now I am drawing on Sunday nights because sometimes Monday is hard as I am dressed up from work. So, I do it Sunday night."

Adjusting to the ever-changing treatment plan: Mothers also described regular discussions with dietitians to modify their child's feeding plan based on results of the most recent Phe level testing. One mother summarized treatment plan modifications: I breastfed her for 11 months . . . after the first month, the dietician felt the levels had stabilized enough and were within the desired range where we could draw the blood to send to the state [newborn screening laboratory] weekly from home [and not have to go to the clinic for a blood draw].

One mother reported her dietitian's description of modifying the treatment plan based upon serial Phe results: "This is an art, and not a science."

Mothers described ongoing challenges of trying to maintain optimal Phe levels during the inevitable changes in their child's rate of growth. Growth spurts and increased activity decreased Phe levels while a slowing of growth, less activity, and illness all increased Phe levels. With the ever- changing treatment plans related to the child's health and development, mothers felt the need to frequently clarify and discuss issues and concerns with the metabolic team.

Developing a positive working relationship with the metabolic team: Every mother interviewed stated that initially she felt a high level of trust in the metabolic team, especially the dietitian, as a source for the best information about PKU. However, as time progressed, some mothers reported becoming dissatisfied with their metabolic health care providers and began to seek a metabolic team that would better meet their needs and those of their infant. Mothers also reported being dissatisfied with the team's lack of availability for responding to questions and concerns; others questioned the credibility of some of the information they were receiving. Some mothers reported doubting the accuracy of the information received from their dietitian and expressed concern about the level of PKU expertise among members of the metabolic team. One mother stated: It was really stressful when I had an urgent question, about my daughter's levels and how we should make adjustments, deal with them, and not being able to speak with someone pretty quickly. I felt that she [dietitian] didn't really understand the urgency. I guess that was our main issue . . . she just wasn't available as she should be and she also gave us some contradictive information.

The majority of the mothers asserted that the metabolic team members did not recognize the overwhelming sense of responsibility, i.e., the "burden of" ongoing monitoring and the never-ending pressure felt by mothers. Despite these frustrations, all mothers eventually developed the ability to work with their original or new metabolic team. The mothers' goal was to obtain the information that they needed, when they needed it: "I think the dialogue with the clinic is important so that you can get your questions answered." As the mothers were managing PKU, they were also managing breastfeeding which was adapted to their unique situation.

Adapting breastfeeding

All mothers voiced a value for breastfeeding and breast milk for their infants and described how breastfeeding enhanced their feelings of emotional closeness with their infant. Shortly after delivery, the majority of mothers (n=9) started breastfeeding in the delivery room. After their infants' diagnosis of PKU, their breastfeeding plans changed from exclusively breastfeeding to combining breast and Phe-free medical formula feeding with ongoing adjustments based on serial Phe levels. Breastfeeding varied with the ever-changing treatment plan with some mothers breastfeeding only twice daily while others breastfed more frequently. Adapting breastfeeding

focused on patience and perseverance to continue breastfeeding despite the diagnosis.

Patience combining breast and bottle feeding: Mothers used a variety of approaches to combine breastfeeding and bottle feeding Phe-free medical formula for their infants. For the majority of mothers, the introduction of Phe-free medical formula was either before breastfeeding or they alternated breast milk and Phe-free medical formula feeding. Most mothers were able to introduce bottle feeding in conjunction with breastfeeding. However, introduction of the bottle was not without frustration and struggles for mothers whose infants preferred breastfeeding over bottle feeding. One mother described the struggle with the introduction of Phe-free medical formula to her newborn with PKU: After the first day of absolutely no bottle success, I was like all right; maybe if we put half an ounce of [Phe-free medical] formula in two ounces of breast milk into a bottle, we might be able to trick him into this whole bottle thing if we mix it with breast milk. So that was what I did . . . It took him two weeks to kind of get the hang of formula and we were still mixing it with breast milk and he would only take it out of this one bottle. These two glass bottles were for some reason the only bottles that he would drink formula out of. I basically carried these two glass bottles everywhere for the first three months.

Maintaining a milk supply: Although the majority of these mothers were breastfeeding their infants with PKU rather than providing expressed breast milk via bottle feeding, their infants were unable to exclusively breastfeed due to their limited Phe tolerance. Mothers identified having a breast pump as crucial to adequately emptying their breasts frequently enough to maintain their breast milk supply. Mothers reported appreciating a "powerful" electric breast pump to maintain an adequate breast milk supply. One mother stated: Having a good pump, a really good pump really did it because the issue that arise with breastfeeding and simultaneously bottle feeding are always related to supply. Because your infant has not nursed as frequently as an exclusively breastfed baby, your supply can wane. Even using the strategy of breast pumping, these mothers struggled to maintain their breast milk supply. One mother asserted, "[Breast milk] supply issues were the biggest challenge." As breast pumping alone was insufficient to maintain their breast milk supply, some mothers were prescribed domperidone to use in combination with breast pumping and breastfeeding. Other mothers were encouraged to use herbal supplements, such as fenugreek and blessed thistle, to boost their breast milk supply. Another issue for mothers was summarized by one mother: "finding the time to breast pump which wasn't easy" while continuing with their other responsibilities for household duties, child care, and for some, returning to work.

Persevering to breastfeed: After diagnosis, four of the mothers began to breast pump exclusively due to either a recommendation by the metabolic clinic, the child's inability to latch onto the breast, or work-related constraints. Of these mothers, two transitioned from exclusively breast pumping to limited breastfeeding. For one mother, the transition from exclusively breast pumping to limited breastfeeding illustrated her persistence and perseverance to breastfeed her infant. I mostly wanted to nurse her even though I was still pumping seven times a day. I called my local health clinic and I said I need a baby scale because I need to weigh her before and after. I am going to figure out how many milliliters it is. We did that for three weeks. We timed it. At first, it started out as one minute at a time, and then I would take her off and weigh her . . . we would nurse, and we would go back and forth. That worked actually quite well. By the time that she was a month old, we had timed it and she was able to nurse. . . Every morning and every evening, I was allowed to nurse her for eight minutes.

Despite the inability to exclusively breastfeed and the various challenges to breastfeeding their infants with PKU, but through patience and perseverance, mothers were successful in their efforts to breastfeed their infants. As one mother described her breastfeeding experience, "it [breastfeeding] was not ever easy . . . the [milk] production wasn't easy . . . I really had to persevere."

Discussion

Study results yielded a detailed description of how mothers simultaneously manage PKU and breastfeeding by adapting breastfeeding to maintain Phe levels. This simultaneous act of managing PKU and breastfeeding entailed considerable effort. Like mothers in other studies [11,34,35], participants in this study recognized the benefits of breastfeeding and breast milk and were willing to expend the extra effort required to insure their child received these benefits.

In this study, Phe levels were the focus of the feeding experience as care of the infant was "all about Phe levels" to maintain desired Phe levels. Phe levels dictated how often the infant with PKU could breastfeed, the amount of Phe-free medical formula needed for the infant, the frequency of in-home blood Phe samples, and the documentation of daily Phe intake. By focusing on Phe levels, mothers were able to orchestrate the successful management of PKU for their infants and breastfeed them. Mothers adapted breastfeeding in order to successfully manage PKU to have their infant maintain therapeutic Phe levels. Consequently, breastfeeding became a challenge; but through patience and perseveration, these mothers' breastfed and provided breast milk to their infants with PKU.

All the mothers, in this study, provided their infants breast milk for a minimum of six months. Mothers attributed their perseverance to their strong beliefs about the important benefits of breast milk and breastfeeding for their infants. They sought out information, technical assistance, and support from other mothers who had experienced breastfeeding infants with PKU. They described the input and advice they received from these mothers as contributing greatly to their own breastfeeding success.

Limitations

In this study, mothers interviewed were predominantly married, Caucasian, English speaking, and well-educated. These sample characteristics were consistent with mothers who are more likely to successfully breastfeed [36-38]. This necessarily limits the transferability of the findings, but still provides insight into the management of breastfeeding infants with PKU. In addition, these mothers more likely represent normal variation of breastfeeding infants with PKU. As such, further research is needed to include mothers of breastfed infants with PKU who breastfed for less than six month in order to provide an overall spectrum of breastfeeding

infants with PKU and to determine the challenges and issues of breastfeeding infants with PKU.

Conclusion

The findings from this qualitative study illuminate findings from a larger quantitative survey on mothers' perceptions of their experiences breastfeeding infants with PKU. These results begin an attempt to understand the work of managing PKU and breastfeeding by *adapting breastfeeding to maintain Phe levels*. Mothers described two major components of the special effort needed to continue to breastfeed their infant: *maintaining Phe levels* and *adapting breastfeeding*. Mothers individualized their infant's feeding routine in order to maintain metabolic control with appropriate Phe levels while adapting breastfeeding to their situation. As their newborns became active infants meeting developmental milestones, mothers became increasingly confident of their ability to successfully simultaneously manage PKU and breastfeed.

Implications for practice

The findings revealed the increased amount of maternal responsibility, commitment, and hard work that mothers assumed when choosing to continue to breastfeed an infant with PKU. Mothers desired to provide their infant the benefits of breastfeeding and breast milk while adhering to the ever-changing PKU management plan. It is imperative to acknowledge the challenges of care placed on mothers simultaneously managing PKU and breastfeeding. Mothers should be supported and encouraged to continue breastfeeding and receive anticipatory guidance about maintaining their breast milk supply while providing Phe-free medical formula to maintain desired Phe levels. In addition, mothers need to be encouraged to seek out assistance and support about challenges managing PKU, but also breastfeeding issues. This includes identifying qualified childcare options, lactation resources for obtaining a hospital grade electric breast pump immediately after diagnosis, to prevent a waning breast milk supply, and options for providing emotional and social support from other mothers who have breastfed infants with PKU. Designing interventions that support mothers adapting breastfeeding to maintain Phe levels, especially immediately after diagnosis, may also help provide them with strategies to sustain breastfeeding while adhering to the ever-changing PKU treatment plan.

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