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Challenges to Breastfeeding Infants With Phenylketonuria[1]

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Breastfeeding duration for infants with phenylketonuria (PKU) is less than other full-term infants. However, no study has examined the challenges encountered by mothers' breastfeeding infants with PKU. In 75 mothers of a child with PKU, three categories of breastfeeding challenges were identified: common breastfeeding issues, breastfeeding and PKU, and no challenges. The common breastfeeding issues can be identified in the literature but for these mothers, the issues are heightened due to frequent phenylalanine (Phe) monitoring. Even so, many mothers adapt breastfeeding to maintain desired Phe levels. A few mothers had no issues and were the exception, not the norm.

Background

PHENYLKETONURIA (PKU) IS an autosomal recessive disorder involving amino acid metabolism. The enzyme phenylalanine hydroxylase (PAH) which is required for metabolizing phenylalanine (Phe) to tyrosine in the liver is defective in PKU. This results in an excess of Phe in the blood and eventually the brain. Accumulation of Phe and its metabolites without proper intervention in the neonatal period results in progressive cognitive–neurologic damage in the infant. The signs of untreated PKU develop gradually and are not noticed until irreversible cognitive insult has occurred. In contrast, when identified via newborn screening and treatment begun prior to 1 month of age, cognitive–neurologic development is essentially normal. The recommended treatment for PKU is a Phe restricted diet for life.

In the past, mothers with newly diagnosed infants with PKU were not allowed to breastfeed since it is difficult to precisely estimate Phe intake with breastfeeding. However, breastfeeding may be advantageous since Phe concentration is lower in breast milk (2,482 μmol/L) than standard commercial infant formula (4,419 μmol/L) making it possible to utilize a higher proportion of natural protein (Lonnerdal et al., 1976; McCabe et al., 1989). For infants with PKU, Phe restriction means breast milk (which contains protein and therefore Phe) must be limited and recommended intake based upon weekly blood Phe levels. With each Phe level, breastfeeding is adapted to maintain desired Phe levels (120–360 μmol/L) that are dependent upon the infant's Phe tolerance (Blau et al., 2008, 2010). Consequently, the addition of Phe-free medical formula is vital in order to provide adequate protein and caloric intake while maintaining Phe restriction for normal cognitive and behavioral development. Phenylalanine is an essential amino acid; therefore in addition to Phe-free medical formula, infants with PKU need natural protein, such as breast milk or standard commercial infant formula, and later in infancy, cow's milk.

Breastfeeding infants with PKU are challenging. First, there is no universal approach to breastfeeding infants with PKU and

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supplementing with Phe-free medical formula. As a result, mothers offer breast milk and Phe-free medical formula by one of three methods based upon recommendations from their metabolic clinic. Mothers may 1) provide a fixed amount of Phe-free medical formula followed by breastfeeding until satiety, 2) provide a fixed amount of expressed breast milk or a fixed time limitation of breastfeeding followed by Phe-free medical formula until satiety, and 3) alternate between breastfeeding or expressed breast milk feeding and Phe-free medical formula feedings. For some mothers, breastfeeding occurs only twice a day while others breastfed more frequently such as every other feeding. This difference is due to several factors. Phe concentration significantly decreases over time in breast milk (2806 ± 134 μmol/L at 2 weeks to 2173 ± 87 μmol/L at 8 weeks) (Janas & Picciano, 1986). Also even though PKU is a classic Mendelian disease, there is variable presentation and severity in the clinical severity of PKU related to differing mutations in the gene encoding PAH enzyme resulting in differing degrees of residual PAH activity level (Guldberg et al., 1994). Some infants (classical form) have almost a complete loss of enzyme activity while others have only a partial reduction in the activity of the PAH enzyme. Consequently an infant with a classical form of PKU will be able to breastfeed less often than an infant with a milder form of PKU.

Despite these challenges, mothers have attempted to continue breastfeeding after the diagnosis to provide their infants with the benefits of breast milk and breastfeeding. Nonetheless, studies have revealed that few mothers with a child with PKU persist in breastfeeding. For example an Italian study found that breastfeeding rates were significantly lower in infants with PKU than in the general Italian population (Agostoni et al., 2000). Similarly, a Brazilian study reported that slightly more than half of the mothers were breastfeeding their infants with PKU at 6 months and by 12 months less than a third continued to breastfeed (Kanufre et al., 2007). 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 that significantly few mothers continued to breastfeed after diagnosis (Banta-Wright et al., 2014). Overall, the incidence, prevalence, and duration of breastfeeding when infants have PKU continue to be less than when full-term infants do not have PKU (Agostoni et al., 2000; Banta-Wright et al., 2014; Kanufre et al., 2007; Motzfeldt et al., 1999; Segev et al., 2004). In addition, qualitative analysis of mothers' experiences breastfeeding infants with PKU revealed that breastfeeding an infant with PKU is more work than breastfeeding a healthy term infant who does not have PKU (Banta-Wright, Houck, Kodadek, Steiner, & Knafl, in review). However, there is no information about challenges to breastfeeding an infant with PKU. In order to better understand the issues when breastfeeding an infant with PKU, we asked mothers living in the United States and Canada to identify and describe the challenges to the continuation of breastfeeding after an infant is diagnosed with PKU.

Methods

A quantitative descriptive design was selected for this study. Data were from an international Internet survey conducted to explore mothers' current or retrospective experiences breastfeeding an infant with PKU. A convenience sample of 103 mothers with one child who has PKU participated in the study during a 6-month period from November 2010 to May 2011. Eligibility criteria included having a child with PKU, a maternal age >21 years, the ability to read and write English, and residing in the United States or Canada. The study was approved by the institutional review board of Oregon Health & Science University.

After receiving approval from the institutional review board, the study was announced on the PKU Listserv; the purpose of this Listserv is to support individuals with PKU and their families through sharing their experiences in managing PKU. Subsequently, fourteen other regional and national PKU support groups used the original or modified university approved announcement of the study to communicate its availability to their members. Postings included a brief description of the purpose of the study and the inclusion criteria with the instruction that interested mothers should reply directly to the researcher by email. After being contacted, the research information sheet and instructions for the survey were emailed to mothers. The university's institutional review board waived the requirement for a formal written consent as mothers were informed that survey completion was considered evidence of their agreement to participate. The mean duration for mothers to complete the survey was 19 minutes (SD = 10), and ranged from 7 to 69 minutes. Mothers who completed the survey were sent a $10 (U.S.) electronic gift certificate to a medical foods company which provides low protein items popular with families who have individuals with PKU. Of the 149 women who received the information sheet and instructions for the Internet survey, 119 completed the survey for a return rate of approximately 80%.

The survey contained sections about the demographic characteristics of mothers and their infants and infant feeding history. Both forced-choice and open-ended questions were included. The survey was pilot-tested by six mothers and modified based upon feedback to improve the approach and wording of the questions and forced-choice responses.

After the Internet survey was closed, data were downloaded into an SPSS file, and then compared to the hardcopy of each participant's data with discrepancies resolved accordingly. The survey asked mothers "What challenges did you have while breastfeeding your infant with PKU?" Breastfeeding challenges were defined as any problems or difficulties that mothers self-reported. To develop categories for this question, three more specific questions were asked of the responses: 1) What were the general breastfeeding challenges for mothers breastfeeding an infant with PKU? 2) What were the breastfeeding and PKU challenges for mothers breastfeeding an infant with PKU? and 3) Did some mothers not have challenges breastfeeding an infant with PKU? From the open ended responses in the