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RESEARCH

Commitment to Breastfeeding in the Context of Phenylketonuria

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ABSTRACT

Objective: To describe the meaning and importance of breastfeeding to mothers of infants with phenylketonuria (PKU).

Design: Qualitative description.

Setting: Mothers from the United States and Canada were recruited from the PKU Listserv and interviewed by telephone.

Participants: Ten breastfeeding mothers with infants who had PKU and were younger than age 36 months.

Methods: Mothers' thoughts, decisions, and experiences of breastfeeding their infants with PKU were collected through telephone interviews. Interviews were transcribed verbatim, and data were analyzed using thematic descriptive analysis in the context of PKU.

Results: Participants felt that that breastfeeding an infant with PKU was the healthiest choice and was therefore worth the labor. These women believed that this was what a loving mother would choose. As they continued to breastfeed their infants after diagnosis, the views of the participants changed. Initially they saw PKU as a disorder and felt that their infants were ill; later they felt that their infants were healthy in spite of PKU. *Normal* could mean a breastfeeding infant with PKU.

Conclusion: Findings demonstrate the importance mothers attribute to breastfeeding and their willingness to invest considerable effort to breastfeed. Health care providers working with these mothers should help them strategize for success.

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Phenylketonuria (PKU) is an autosomal recessive condition related to the metabolism of amino acid. This condition results from mutations in the phenylalanine hydroxylase enzyme (PAH), which is required to metabolize phenylalanine (Phe) to tyrosine (Tyr) in the liver (Blau, van Spronsen, & Levy, 2010). As a result of the defective PAH enzyme, Phe and its metabolites accumulate in the blood and eventually the brain and can cause progressive cognitive-neurologic damage in the infant.

The prevalence of PKU varies worldwide but is present in every racial and ethnic group. The greatest prevalence of PKU is in Ireland with one case per 4,500 live births (Zschocke, Mallory, Eiken, & Nevin, 1997). Turkey follows closely with one case per 5,000 live births (Tuncbilek & Ozguc, 2007). Finland has the lowest prevalence in Europe with less than one case per 100,000 births (Guldberg, Henriksen, Sipila, Guttler, & de la Chapelle, 1995). In Latin America, the prevalence of PKU varies from one case per 25,000 births in Brazil to one case per 52,000 births in Cuba (Borrajo, 2007). In comparison, the prevalence of PKU in Asia varies from one case per 15,000 to one case per 100,500 births in different regions of China (Jiang et al., 2003; Zhan, Qin, & Zhao, 2009) to one case per 327,000 births in Thailand (Pangkanon, Charoensiriwatana, Janejai, Boonwanich, & Chaisomchit, 2009).

In the United States, the prevalence of PKU is approximately one case in 20,0000 live births but varies from state to state due to the racial and ethnic makeup of the population (National Institutes of Health Consensus Development Panel, 2001; National Newborn Screening and Genetics Resource Center, 2009). The prevalence in Iowa is consistent with the national reported prevalence of approximately one case per 20,000 births (National Newborn Screening and Genetics Resource Center, 2009), whereas in Louisiana, the prevalence is approximately one case per 10,000 live births

in the White population and one case per 76,000 live births in the non-White population (Louisana Office of Public Health, 2000). Overall, approximately 400 infants are born each year who are positive for PKU in the United States.

In the United States newborn screening for PKU is performed via tandem mass spectrometry on dried blood spots. If a newborn has an abnormally elevated level of Phe (>190 μmol/L) with an increased Phe-to-Tyr (ratio >3) on the newborn screen, this is a presumptive positive result, and follow-up diagnostic testing and assessment at a pediatric metabolic clinic is needed as soon as possible (Northwest Regional Newborn Screening Program, 2010). Follow-up Phe testing is needed to measure Phe concentrations in the blood with positive diagnostic results revealing increased Phe concentrations (>120 μmol/L) (Donlon, Levy, & Scriver, 2014).

Because the advent of newborn screening, infants with PKU can be diagnosed before an insidious neurocognitive insult occurs. During the 1960s and the early days of screening, the standard of care for infants diagnosed with PKU was to wean them immediately from breastfeeding and to institute feedings of low-Phe medical formula in conjunction with standard commercial infant formula with the goal to maintain appropriate Phe levels (120–360 μmol/L) (Blau et al., 2010; Demirkol et al., 2001; Kanufre et al., 2007). This management plan was believed to be the only effective way to allow for precise titration and measurement of Phe to protect neurological and cognitive development of infants with PKU. As researchers demonstrated that breastfeeding and human breast milk were viable options to the original practice of exclusive formula feeding, more mothers attempted to continue breastfeeding after diagnosis (Cornejo et al., 2003; Ernest, McCabe, Neifert, & O'Flynn, 1980; Greve, Wheeler, Green-Burgeson, & Zorn, 1994; Kanufre et al., 2007; McCabe et al., 1989; Motzfeldt, Lilje, & Nylander, 1999).

Although many mothers successfully continue to breastfeed, many mothers decide not to continue to breastfeed after the diagnosis of PKU (Banta-Wright, Press, Knafl, Steiner, & Houck, 2014). In addition, investigators have consistently reported that few mothers of infants with PKU persist in breastfeeding beyond an initial period and breastfeed for a shorter duration than mothers of other healthy term infants (Agostoni, Verduci, Fiori, Riva, & Giovannini, 2000; Banta-Wright et al., 2014; Banta-Wright, Shelton, Lowe, Knafl, & Houck, 2012; Cornejo et al., 2003; Demirkol et al., 2001; Huner & Demirkol, 1996; Kanufre et al., 2007; Motzfeldt et al., 1999; Segev, Abraham, Anikster, & Schwartz, 2004; van Rijn et al., 2003).

Breastfeeding infants with phenylketonuria is challenging in part because it is virtually impossible to precisely determine phenylalanine intake.

The National Institutes of Health (2000) consensus statement on PKU encouraged breastfeeding in conjunction with the use of Phe-free medical formula. In an international survey, MacDonald and colleagues (2006) examined the feeding recommendations of health care providers in various metabolic centers in Europe, South America, United States, Australia, and New Zealand. They found no universal approach to feeding infants with PKU. Treatment management varied among countries and between metabolic centers within countries. However, there was consensus regarding the recommendation of Phe-free medical formula in combination with breastfeeding. In 2011, the National PKU Alliance published "My PKU Binder," a comprehensive resource for parents and individuals with PKU that briefly mentions that breast milk contains less Phe than standard commercial infant formula. This binder, a trusted resource for mothers learning about PKU, provides much needed general information about PKU but lacks specific guidelines for successfully breastfeeding infants with PKU (e.g., guidance on maintaining a breast milk supply when having to alter breast milk intake to maintain Phe levels within the desired range).

Although PKU is a classic Mendelian disease, it has variable clinical presentation and severity due to the different mutations in the gene-encoding PAH enzyme. The combinations of the different mutations result in varying degrees of residual PAH activity level (Guldberg et al., 1994). The residual PAH enzyme activity reflects Phe tolerance. Phenylketonuria is classified based upon the severity of the elevated Phe levels. The normal range of blood Phe concentrations is 50 to 110 μmol/L (Donlon et al., 2014). There are four different phenotypes: classical PKU, moderate PKU, mild PKU, and mild hyperphenylalaninemia (Blau, Hennermann, Langenbeck, & Lichter-Konecki, 2011). Yet classification during the newborn period is difficult to determine as Phe concentrations have not had time to reach their peak values, and treatment is initiated before

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