The Management of Breast Feeding Among Infants with Phenylketonuria

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Summary: Treatment for phenylketonuria (PKU) involves using low phenylalanine or phenylalanine-free formulas and supplementation with sufficient phenylalanine for normal growth and development. Eighteen infants with phenylketonuria who received breast milk as their primary phenylalanine source were compared with ten other infants with PKU who received their phenylalanine primarily from infant formulas. There were no significant differences between breast-fed and formula-fed infants for serum phenylalanine, serum tyrosine, length, weight, head circumference, haematocrit, haemoglobin, serum iron, total iron binding capacity, percentage iron saturation, ferritin, plasma zinc and total calorie intake. Breast-fed infants did show lower mean corpuscular volume at 3 months and 6 months of age. Breast-fed infants had lower phenylalanine intake at 2, 4, 5 and 6 months of age. Breast-fed infants at 1, 2, 3, 4, 5 and 6 months of age had lower protein intake. Breast feeding may be continued in the newly diagnosed phenylketonuric infant without any apparent adverse nutritional consequences.

INTRODUCTION

Traditionally a positive newborn screen for phenylketonuria has led to the initiation of dietary treatment using a phenylalanine-free or low-phenylalanine formula. The special formula was supplemented with a cow’s milk-based formula to provide sufficient phenylalanine for adequate growth and development (Acosta and Wenz, 1977). This management strategy has precluded breast feeding the infant with phenylketonuria. However, human milk offers a number of advantages when used as the primary source of supplemental phenylalanine to the infant with phenylketonuria. These include the benefits of breast milk for any infant (Task Force on the Promotion of Breast Feeding, 1982; Position of the American Dietetic Association, 1986). In addition, mature breast milk is lower in phenylalanine,
The protein content of mature breast milk, 0.8–0.9 g/dl (Lonnerdal et al., 1976a, b) is lower than that of cow’s milk, 3.3 g/dl (Posati and Orr, 1976). There is also a lower casein content in breast milk, 2.5 g/L, than cow’s milk, 27.3 g/L (Lonnerdal et al., 1976a). When the percentage of protein from the whey fraction is compared, breast milk, 70%, is higher than cow’s milk, 20% (Lonnerdal et al., 1976a).

In addition to the nutritional and health benefits to the child with phenylketonuria who is breast-fed, there is also a decrease in emotional stress to the mother who had planned to breast feed her infant. While parents of the newly diagnosed infant with phenylketonuria are already experiencing a loss, abrupt weaning of the infant produces additional emotional stress for the mother (Ernest et al., 1979).

This study was undertaken as a clinical trial to determine the feasibility of breast feeding the infant with phenylketonuria. Our early experience and recommendations have been summarized (Ernest et al., 1979), and we have briefly reported a portion of the data previously (McCabe and McCabe, 1986).

**MATERIALS AND METHODS**

During the period 1977–1984, 28 infants with positive newborn screens for phenylketonuria and elevated quantitative serum phenylalanine determinations (McCaman and Robins, 1962) required dietary management at the University of Colorado Health Sciences Center. One group of 18 infants was managed by a phenylalanine restricted formula and supplemental breast feeding and another group of ten infants was managed by supplementation with infant formula. In the breast-fed group, 16 infants were weaned at ages ranging from 1.3 to 25.5 months (mean of 8.9 ± 7.3 months). Two infants were still breast feeding at the conclusion of the study. The mean daily intake of breast milk during the first 6 months of life ranged from a low of 362 ml/day (± 141, n = 13) during the first month of life, to a high of 464 ml/day (± 203, n = 9) during the fourth month of life. An adequate intake of breast milk is required to provide sufficient amounts of dietary phenylalanine. The amount of breast milk consumed was determined by weighing the infant before and after breast feeding. Parents used one of the following scales: Health-O-Meter (Continental Scale Corp., Model 322); Detecto Scale (Model 2501); or Sartorius Computerized Electronic Scale. Each scale was calibrated once a day by the parent using a standard weight.

Since the dietary management of these infants was initiated as they entered the study, frequent determination of serum phenylalanine concentrations was required to assess metabolic control. The blood specimens were more frequent during periods of illness or following a high phenylalanine level. Blood specimens were obtained at least 2 hours postprandially. Parents mailed blood specimens to the laboratory or brought their infant to the laboratory to have the blood drawn there.