Phenylketonuria (PKU) and Breastfeeding

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Phenylketonuria (PKU) is a hereditary effect which affects 1 in 12,000 live births. It is inherited as an autosomal recessive trait which means each parent of the PKU child has a normal and an abnormal gene for this disorder. Thus, each sibling of these parents will have a 1 in 4 chance of having PKU. A Newborn Screen is used in the United States to diagnose PKU.

PKU is a deficiency of phenylalanine hydroxylase which is needed for the metabolism of protein. When the phenylalanine is not completely metabolized it accumulates in the blood and tissues, including the brain. If left untreated, it will lead to developmental delay in the first year of life, and progress to severe mental retardation. It can also cause seizures, autistic-like behavior, rashes, uneven pigmentation, and inevitably a reduced life span. For these reasons it is imperative that once PKU is discovered appropriate treatment begin to decrease phenylalanine levels in the infant’s blood.

Breastfed newborns may continue to nurse after PKU has been diagnosed. The PKU newborn may enjoy all the benefits that breastmilk provides if phenylalanine levels are monitored. Breastmilk is actually lower in phenylalanine when compared to commercial formulas. Thus, the newborn can have larger volumes of breastmilk within the constraint of the prescribed phenylalanine limit. The serum concentration should be maintained between 120-360 umol/L. The amount of phenylalanine in the breastmilk is calculated along with the dietary needs of the infant. The amount of breastmilk allowed to maintain serum levels is calculated and the remainder of the infant’s feeding must be a phenylalanine free formula. The process is quite simple if the mother chooses to pump and provide her milk via bottle; however, it becomes much more complex if she chooses “at breast” feedings. In the later case, the intake of the infant must be quantified either by weighing the child before and after feedings, or by estimating volume intake by time at breast. In either case, intake of breastmilk by these means is not definitive; therefore, phenylalanine intake is not exact. In most cases, a mother may choose any of the above methods to control the infant’s PKU diagnosis and promote optimal growth and development of her child.

References:
