PKU Infant Case Studies

Using the new PKU Periflex Early Years for dietary management.

Here we present three infant males diagnosed with phenylketonuria (PKU) through newborn screening and confirmatory testing. They were all born full term after uncomplicated pregnancies. The patients presented here were the first in their families to be diagnosed with PKU.

Patient 1 History

4-month old male referred for management of PKU at 8 days of age. Blood phenylalanine (Phe) was 1432 µmol/L and tyrosine was 38 µmol/L on confirmatory testing at 6 days of age. At the initial clinic visit, he was taking either 2-3 fl oz Similac® Advance or expressed breast milk approximately every 3 hours.

Nutritional Management

Patient 1 was started on a 72-hour washout period without any dietary Phe, during which he consumed only PKU Periflex® Early Years formula *ad lib* mixed to 20 calories/oz. The family requested to use standard infant formula rather than continue expressing breast milk. After the washout period, he was started on PKU Periflex Early Years with Similac Advance infant formula providing 45 mg Phe/kg, 2.7 g total protein/kg, and 119 calories/kg. Phe intake per kg increased during the first two months of life (52-54 mg/kg) and decreased at 2.5 months of age (37 mg Phe/kg). Total protein intake ranged from 2.5-2.8 g/kg between 1 and <3 months of age. At 4 months of age, he remained at 37 mg Phe/kg, 2.4 g total protein/kg, and 94 calories/kg.

Results

The introduction of PKU Periflex Early Years at 8 days of age was tolerated well. The family denied constipation, diarrhea, and increased gassiness with introduction of PKU Periflex Early Years. His average weight gain was above the reference range at 38 g/day from 0-4 months of age. At 4 months of age his length and head circumference were appropriate and proportional. Plasma amino acids collected at clinic visits showed normal levels of all amino acids except phenylalanine. Average blood Phe concentrations were 223 µmol/L (target range 120-360 µmol/L) and tyrosine concentrations were normal, 68 µmol/L.

Patient 2 History

4.5-month old male referred for management of PKU at 7 days of age. Blood Phe was 2279 µmol/L and tyrosine was 76 µmol/L on confirmatory testing at 7 days of age. At the initial clinic visit, he was breastfeeding approximately every 2 hours.

Nutritional Management

Patient 2 was started on a 96-hour washout period without any dietary Phe, during which he consumed only Periflex® Infant formula *ad lib* mixed to 20 calories/oz. After the washout period, he continued on Periflex Infant mixed to 20 calories/oz and alternated with breastfeeding. Nutrient intake estimated to be 30 mg Phe/kg, 2.7 g total protein/kg, and 115 calories/kg. A new PKU infant formula had been brought to market and

its introduction was delayed due to pharmacy availability. At 50 days of age, PKU Periflex Early Years was introduced at a 50:50 ratio with Periflex Infant. The family reported increased gassiness and constipation with the change. PKU Periflex Early Years was reduced to provide 20% of total medical food and symptoms improved. From 2-4 months of age, PKU Periflex Early Years increased to provide 60% of total medical food without any additional symptoms. At 4.5 months of age, his Phe intake was approximately 33 mg/kg with 2 g total protein/kg and 94 calories/kg.

Results

Patient 2 required a slower transition to PKU Periflex Early Years to avoid gastrointestinal symptoms. After slowing the transition, PKU Periflex Early Years was tolerated without further problems. His plasma amino acids and prealbumin indicated adequate total protein intake. His average blood Phe concentrations were 231 µmol/L (target range 120-360 µmol/L) and tyrosine concentrations were 74 µmol/L. His weight gain was above the reference range with appropriate height and head circumference.

Patient 3 History

2-month old male referred for management of PKU at 7 days of age. Blood Phe was 938 µmol/L and tyrosine was 26 µmol/L on confirmatory testing at 6 days of age. At the initial clinic visit, he was taking 2 fl oz Enfamil® Newborn every 2-3 hours.

Nutritional Management

Patient 3 was started on a 72-hour washout period without any dietary Phe, during which he consumed only PKU Periflex Early Years formula *ad lib* mixed to 20 calories/oz. After the washout period, he was started on PKU Periflex Early Years with Enfamil® Newborn infant formula providing 45 mg Phe/kg, 2.9 g total protein/kg, and 116 calories/kg. At 19 and 47 days of age Phe intake was increased to 50 mg/kg and 75 mg/kg, respectively based on blood Phe levels. Total protein intake remained between 2.9-3 g/kg with approximately 120 calories/kg between 1 and 2 months of age.

Results

PKU management with Enfamil Newborn and PKU Periflex Early Years was well tolerated and resulted in normal growth and appropriate metabolic control. His average growth rate was 30 grams/day for the first two months of life. His height and head circumference were appropriate for his weight. Plasma amino acid results were normal. His average phenylalanine concentrations were 132 µmol/L and tyrosine concentrations were 61 µmol/L.

This case report* is provided by Krista Viau, PhD, RDN

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