

uniformly over the blot paper.

Because of the possibility of variant forms of hyperphenylalaninemia, PKU cofactor variant screen should be performed in all children diagnosed with PKU. A major concern is that a significant number of infants are not rescreened for PKU after early discharge and are at risk for a missed or delayed diagnosis. Give special consideration to screening infants born at home who have no hospital contact and infants adopted internationally.

Therapeutic Management*

Treatment of PKU involves restricting phenylalanine in the diet. Because the genetic enzyme is intracellular, systemic administration of phenylalanine hydroxylase is of no value. Phenylalanine cannot be eliminated because it is an essential amino acid in tissue growth. Therefore, dietary management must meet two criteria: (1) meet the child's nutritional need for optimum growth and (2) maintain phenylalanine levels within a safe range (2 to 6 mg/dl in neonates and children up to 12 years old, and 2 to 10 mg/dl through adolescence) ([Soltanizadeh and Mirmoghtadaie, 2014](#)).

Professionals agree that infants with PKU who have blood phenylalanine levels higher than 10 mg/dl should be started on treatment to establish metabolic control as soon as possible, ideally by 7 to 10 days of age ([Kaye, Committee on Genetics, Accurso, et al, 2006](#)). The daily amounts of phenylalanine are individualized for each child and require frequent changes on the basis of appetite, growth and development, and blood phenylalanine and tyrosine levels.

Because all natural food proteins contain phenylalanine and will be limited, the diet must be supplemented with a specially prepared phenylalanine-free formula (e.g., Phenex-1 for infants or Phenex-2 for children and adults).^{*} The phenylalanine-free formula is an amino acid–modified formula essential in the low phenylalanine diet to provide the appropriate protein, vitamins, minerals, and calories for optimal growth and development. Because tyrosine becomes an essential amino acid, the phenylalanine-free formula supplies an adequate amount, but in some cases, additional supplementation may be needed. The phenylalanine-free amino acid–modified formula for infants has all the nutrients necessary for adequate infant growth. Because of the