At a Glance
Phenylketonuria (PKU)

Deficient enzyme: Phenylalanine hydroxylase (PAH); converts phenylalanine to tyrosine

Enzyme Cofactor: Tetrahydrobiopterin (BH_4)

Toxic Metabolite: Phenylalanine (Phe)

Clinical Presentation, if untreated: irreversible intellectual disabilities, seizures, behavioral abnormalities
eczema, "musty" odor, hypopigmentation (skin, hair, iris)

Goal Treatment Range: Blood Phenylalanine: 120-360 µmol/L (2-6 mg/dL)
(to convert mg/dL to µmol/L multiply by 60)
Blood Tyrosine: normal for lab

Nutrient Needs by Age*

Age	Phe mg/d	Phe mg/kg/d	Tyr mg/d	Protein g/kg/d	Energy
0-3 mo	130-430	25-70	1100-1300	2.5-3.0	Age appropriate DRI
3-6 mo	135-400	20-45	1400-2100	2.0-3.0	
6-9 mo	145-370	15-35	2500-3000	2.0-2.5	
9-12 mo	135-330	10-35	2500-3000	2.0-2.5	
1-4 yrs	200-320	-	2800-3500	1.5-2.1	
>4 yrs to adult	200-1100	-	4000-6000	120-140% DRI	

*SERN/GMDI PKU Management Guidelines; https://southeastgenetics.org/ngp/guidelines.php

Starting a PKU Diet

1. Determine goals for Phe (mg), Protein (g) Tyrosine (mg), Energy (kcal)
   - use 50 mg Phe = 1 g protein to calculate mg of phenylalanine from grams of protein.
2. Calculate amount of whole protein source (breast milk, infant formula, food) needed to meet Phe goal.
3. Calculate amount of medical food needed, in addition to the whole protein source to meet total protein goal.
4. Calculate energy intake from whole protein and medical food sources to ensure total calorie needs are met.
5. Calculate tyrosine intake from whole protein and medical food sources.

Implementing the Simplified PKU Diet*

1. At 4-6 months of age, when solid food is introduced, consider implementing the Simplified PKU Diet.
2. Reduce phe allowance (from whole protein source) by 30% (40% in those with more restrictive Phe allowances).
3. Allow unmeasured intake of "uncounted foods." These are fruits, vegetables, foods with <75 mg Phe/100g and all other foods with <20 mg Phe or <0.4 g protein per serving.
4. Monitor blood Phe weekly for the first 4 weeks, without making diet changes.

*Bernstein LE, et al. Multiclinic Observations on the Simplified Diet in PKU. J Nutr Metab. 2017.

Phenylketonuria (PKU)

Medical Foods for PKU

	Abbott abbottnutrition.com	Cambrooke Cambrooke.com	Mead Johnson hcp.meadjohnson.com	Nutricia NutriciaMetabolics.com	Vitaflo www.vitafloUSA.com
Infant (0-1 yr)	Phenex™-1		Phenyl-Free® 1	PKU Periflex®Early Years	PKU explore™ 5, 10
Toddler & Young Children	Phenex™-1 Phenex™-2	*Glytactin® BetterMilk 15 *Glytactin® RTD 10, 15 *Glytactin®BUILD 10, 20/20 *Glytactin® COMPLETE 10 Bar *Glytactin® RESTORE 10 *Glytactin® RESTORE Powder 5 *Glytactin® SWIRL 15	Phenyl-Free® 1 Phenyl-Free® 2	PKU Periflex® Junior Plus PhenylAde® Essential Drink Mix *PhenylAde® GMP Drink Mix *PhenylAde® GMP Ready *PhenylAde® GMP Mix-In *PhenylAde® GMP Ready *PhenylAde® GMP Ultra	PKU gel™ PKU trio™ PKU express® 15 PKU cooler® 10, 15 *PKU sphere® 15 *PKU sphere® liquid
Children thru Adult	Phenex™-2	*Glytactin® BetterMilk 15, Lite *Glytactin® RTD 10, 15, Lite *Glytactin® BUILD 10, 20/20 *Glytactin® RESTORE 10, Lite *Glytactin® RESTORE Powder 5, Lite 10 *Glytactin® COMPLETE 10 Bar *Glytactin® SWIRL 15	Phenyl-Free® 2 Phenyl-Free® 2HP	Periflex® Advance Periflex® LQ Phenylade® Essential Drink Mix Phenylade® Drink Mix 40, 60 Phenylade® MTE Amino Acid Blend *PhenylAde® GMP Drink Mix *PhenylAde® GMP Ready *PhenylAde® GMP Ultra *PhenylAde® GMP Mix-In PKU Lophlex® LQ & Powder XPhe Maxamum® Phlexy-10® Tablets, Drink Mix **PhenylAde® PheBLOC™ LNAA	PKU express® 15, 20 PKU cooler® 10, 15, 20 PKU Air® 20 *PKU sphere® 15, 20 *PKU sphere® liquid

*A Glycomacropeptide (GMP) product **Product used for Large Neutral Amino Acid therapy

Medical Therapy (www.biomarin.com)

Kuvan (sapropterin dihydrochloride): synthetic form of BH4 (PAH cofactor) Dose: 5-20 mg/kg/d

Palynziq (pegvaliase): phenylalanine ammonia lyase (enzyme substitution for PAH) Dose: 20-60 mg/d

Laboratory Monitoring*

Blood Phenylalanine[1]
Blood Tyrosine[1]
Plasma Amino Acids[2]
Prealbumin[2]
25-OH Vitamin D[2]
CBC[2]
Zinc, Copper[3]
Vitamin B_{12}[3]
Essential Fatty Acids[3]

[1] Weekly in infancy, weekly to monthly thereafter
[2] Every 6-12 months
[3] As indicated

*www.gmdi.org/Resources/Nutrition-Guidelines/Phenylketonuria-PKU