

Chapter 16: Glossary



Term	Definition
Blood Phe monitoring	Individuals with PKU must monitor their blood Phe levels to ensure that their levels remain in a healthy range. Blood samples are taken at home and at the PKU clinic to be analyzed, with the food record to help the PKU clinic dietitian adjust the PKU diet appropriately.
Classical PKU	The most common form of PKU. It results when there are two severe mutations of the PAH gene and as a result there is little or no PAH enzyme activity to convert phenylalanine to tyrosine. These are the most severely affected patients. Phe levels are above 20 mg/dl (>1200 μ mol/L).
Consensus Statement	When related to the NIH, a consensus statement is developed by a panel of experts in the field that presents a “best practices” for treating the disease or disorder being discussed. See http://consensus.nih.gov/ABOUTCDP.html for more information.
Food Record/Diet Record	A record of what foods have been consumed with date and time consumed and amount.
Food Reference Guide	A book that shows how much protein common foods contain, which helps you figure out and plan how much protein you will eat at each meal or snack. It also lists “free foods”, i.e. foods that contain little or no protein, which do not need to be counted. The dietitian at your PKU clinic will give you a reference guide and help you use it.
Glycomacropeptide (GMP)	A whey-based protein produced when making cheese. It is the only known dietary protein that contains a minimal amount of PHE. Foods made with GMP provide an alternative to the amino acid medical foods currently required in the PKU diet that some find to be more palatable than other medical foods.
Home Blood Phe Monitor	A monitor that works similarly to a diabetes monitor, the Home Blood Phe Monitor is currently under development by BioMarin Pharmaceutical Inc., Novato, C., This would allow PKU patients to monitor their Phe levels at home.
Hyperphenylalaneia (HyperPhe)	Associated with high Phe levels, but potentially not high enough to require treatment. Phe level in blood: less than 6 mg/dl (<360 μ mol/L)

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KUVAN® (sapropterin dihydrochloride)	A FDA approved medication that is used as a treatment method for some patients with PKU. KUVAN works by helping the phenylalanine hydroxylase (PAH) enzyme work more effectively to break down PHE in the body.
Large Neutral Amino Acid (LNAA)	LNAA is a treatment option that is mostly used for adults who have difficulty in maintaining the recommended PHE levels. LNAA is considered a medical food product, and comes in a powder or pill form containing essential amino acids (not including PHE) and large neutral amino acids. LNAA contains amino acids similar to those that are found in PKU formulas, but in more concentrated amounts.
Maternal PKU Syndrome	A syndrome that affects babies born to women with PKU whose Phe levels were not well controlled during pregnancy. Symptoms can include heart problems, slow development, small head size and brain damage.
Medical food	Medical Foods include both medical formula and foods modified to be low in protein. The medical formula provides all of the essential amino acids found in protein (except for Phe), as well as tyrosine, vitamins, minerals and trace elements that your body needs that most people who do not have PKU get from their diet. The foods modified to be low in protein provide an essential energy source and satiety with less than 1 gram of protein.
Moderate/Mild PKU	Is associated with elevated Phe levels, but not as high as for those patients with Classical PKU. Phe level in blood: Above 6 mg/dl but less than 20 mg/dl (360- 1200µmol/L).
National Institutes of Health (NIH)	The United States' medical research organization.
Newborn Screening	All babies born in the United States are tested for many diseases and disorders (including phenylketonuria) approximately 24 hours after birth through the state newborn screening test. A blood sample is taken from a needle prick on a baby's heel, and the blood is analyzed in a laboratory.
Phenylalanine (Phe)	An amino acid found in protein that PKU patients cannot break down.
Phenylalanine hydroxylase (PAH)	The enzyme in the liver that is deficient in PKU.

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Phenylketonuria (PKU)	An inherited genetic disorder that affects approximately one in 14,000 babies born in the United States. A person with PKU does not produce enough of an enzyme in their liver called phenylalanine hydroxylase (PAH), which is needed to process the amino acid, phenylalanine (Phe).
PKU Clinic	A clinic that has a group of healthcare professionals trained to support individuals with PKU. Team members often include geneticists, dietitians, genetic counselors, social workers, nurses and sometimes psychologists.
PKU Diet/Low Protein Diet	The PKU diet consists of low protein foods such as fruits, vegetables, fats, sugars, special low protein food such as low protein pasta and medical formula.
Therapeutic Liver Repopulation	Aims to replace the cells that are deficient in PAH (PAH negative cells) with cells that are not PAH-deficient (PAH positive cells). PAH positive cells will then restore function of the liver, theoretically curing PKU.