



## GLOSSARY

### **Amino Acid**

The basic building block of proteins. Phenylalanine is an amino acid. Proteins are broken down into individual amino acids to be absorbed and used by the body.

### **Autosomal Recessive Disorder**

A genetic term that means there is a 25% risk, with each pregnancy, to have an infant with PKU.

### **Biopterin**

A chemical co-factor needed to metabolize phenylalanine. All infants with elevated blood phenylalanine levels will be tested for a rare, potential biopterin deficiency.

### **Carrier**

A person, who does not have PKU, but carries a recessive gene with a normal gene. A carrier is capable of transmitting the disease.

### **Challenge**

A test for tolerance of phenylalanine to determine if your child needs a PKU diet.

### **Chromosome(s)**

Contain the DNA that transmits genetic information. The structure that holds our genes. There are 46 (23 pairs) of chromosomes inside each cell of the body. One member from each pair comes from the mother's egg and the other member of the pair from the father's sperm. The PKU gene is on chromosome 12 (long arm, q; band region 24.1).

### **Enzyme**

A protein made by living cells that helps to cause a chemical change. An enzyme helps the chemical change happen faster. Phenylalanine hydroxylase is an enzyme.

### **Equivalent or Exchange**

A measured amount of food, such as cereal, protein, vegetables, or fruits that is calculated into the PKU diet. An equivalent contains approximately 15 milligrams (mg) of Phenylalanine.

### **Gene(s)**

The basic unit of heredity. Each gene has a certain location on a chromosome. Genes are passed on from the parent to the child. Genes hold a message or instructions about how our body cells should work.

### **Genetics**

Is the study of how similarities are inherited from your parents.

### **Hyperphenylalaninemia**

A long medical word that means higher than normal (hyper), Phenylalanine (PHE) levels, in the blood (emia).



**Inheritance / heredity**

The process by which genes are passed from parents to their children.



**Metabolic Disorder**

A defect in the pathway of phenylalanine metabolism, usually due to a phenylalanine hydroxylase deficiency.

**Metabolic Pathway(s)**

A term used to describe the chemical processes that change proteins, fats, and carbohydrates into smaller units that can be used by the body. The food we eat is digested along “a metabolic pathway” to produce the nutrients our bodies use for energy.



**Metabolism**

The sum of all chemical changes that take place in our body. The chemical changes of food into heat or energy.

**Milligram (mg)**

A unit of weight equal to 1/1000 Th of a gram.



**Phenylalanine (PHE)**

One of the essential amino acids (essential amino acids must be eaten in the diet, non-essential amino acids can be produced by the body). Usually, the excess level of PHE is controlled by being converted to Tyrosine (TYR), another amino acid. In persons with PKU, the PHE builds-up in the blood because the cells can not convert the excess into TYR. The enzyme that converts PHE to TYR is called phenylalanine hydroxylase.

**Phenylketone**

A byproduct of PHE breakdown, which collects in the urine, of persons with untreated PKU.



**Phenylketonuria (PKU)**

A heredity condition caused by the body’s failure to change PHE to TYR because of defect gene that can not produce the enzyme Phenylalanine hydroxylase. The PHE will build-up in the blood and cause brain damage.

**Protein**

An essential ingredient of all living matter. Protein is made out of building blocks called amino acids. Proteins have many uses in the body and are used for building materials, chemical messages, chemical reactions (enzymes), etc.



**Screening**

A heel stick, blood spot on filter paper, which measures phenylalanine levels. All infants will be screened, twice, for PKU.

**Tyrosine (TYR)**

A non-essential amino acid (the body can make this amino acid). The body makes TYR, by changing PHE to TYR. Tyrosine because an essential amino acid (must be eaten in your diet) because the person with PKU can not make TYR.

