Phenylketonuria (PKU)

Phenylketonuria is a genetic disorder that is characterized by a person's inability to utilize the essential amino acid, phenylalanine. A person with PKU lacks the enzyme phenylalanine hydroxylase, which is necessary to convert phenylalanine to another amino acid tyrosine. Essential amino acids can only be obtained from the food we eat and they are building blocks of body protein. When phenylalanine is not broken down, it accumulates in the blood and body tissues. Chronically high levels of phenylalanine can cause significant brain damage.

Symptoms

- Vomiting, irritability, eczema and mousy odor to the urine.
- Increased muscle tone and more active muscle tendon reflexes
- Small head, prominent cheek and upper jawbones, widely spaced teeth, decreased body growth
- Intellectual disabilities and seizures

Dietary Recommendations

- Avoid high protein foods such as: meat, fish, poultry, eggs, cheese, milk, dried beans, and peas
- Measured amounts of cereals, starches, fruits, vegetables, and milk substitutes
- Avoid food and beverages containing the artificial sweetener aspartame, such as diet soda.
- To meet individual protein needs a phenylalanine free amino acid formula is given to all age groups