Phenylketonuria (PKU)

- Phenylketonuria (also called PKU) is an inherited (passed from parent to child) condition that occurs when the body is unable to break down an amino acid called phenylalanine. Amino acids are the “building blocks” that the body uses to make proteins.

- When phenylalanine cannot be removed from the body, it builds up in the body and causes health problems including:
  - Brain damage
  - Developmental delay (failure to meet developmental milestones on time)
  - Mental retardation
  - Mousy or musty-smelling urine
  - Problems with movement
  - Seizures
  - Skin problems, such as eczema (itchy, scaly skin)

- There is no cure for PKU. However, a special diet low in phenylalanine can help prevent the health problems associated with PKU.

- For more information about PKU, please click on one of the links below.
  - Medline Plus Medical Encyclopedia
  - National Library of Medicine Genetics Home Reference