

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](#)