# Phenylketonuria Encyclopedia Article

## Phenylketonuria

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# Phenylketonuria

Phenylketonuria (PKU) is an inherited metabolic disorder in which an enzyme (phenylalanine hydroxylase) that is crucial to the appropriate processing of the **amino acid** phenylalanine, is absent or deficient. Normally, phenylalanine is converted to tyrosine in the body. When phenylalanine cannot be broken down, it accumulates in excess quantities throughout the body, causing mental retardation and other neurological complications. Treatment is usually started during babyhood; delaying such treatment results in a significantly lowered intelligence quotient (IQ) by age one. Because tyrosine is involved in the production of melanin (pigment), people with PKU usually have lighter skin and hair than other family members.

PKU is an autosomal recessive disorder, and is caused by mutations in both alleles of the **gene** responsible for phenylalanine hydroxylase, found on **chromosome** 12. Parents, both with the recessive gene **mutation** responsible for PKU, have a 25% chance with each pregnancy of producing a child affected with PKU. In the general population, the incidence of an adult **carrier** of PKU is approximately one in fifty people. PKU occurs in approximately one out of every 10,000 to 15,000 Caucasian or Oriental births in the United States. The incidence of PKU in the African--American population is much less. Treatment involves a strict diet low in phenylalanine (warnings aimed at people with PKU can be found on cans of diet drinks containing the artificial sweetener aspartame, which is made from phenylalanine). **Gene therapy** offers the potential for permanently correcting the enzyme deficiencies of PKU and other **inborn errors of metabolism**. Current research focuses on the use of genetically altered adenoviruses (one of the causes of the common cold) to correct certain other inherited liver--based metabolic diseases. Understanding of the exact mechanisms of the neurological complications associated with PKU are, however, little understood, and knowledge of the precise genetic mutations responsible for PKU have yet to yield significant advances in treatment or prevention of PKU. Because it is vital to begin diet treatment immediately, most nations in the developed world require all that all infants be tested for the disease within the first week of life.