Phenylketonuria Encyclopedia Article

Phenylketonuria

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Phenylketonuria

Phenylketonuria (PKU) is an inherited disorder in which an enzyme (usually phenylalanine hydroxylase) crucial to the appropriate processing of the amino acid, phenylalanine is totally absent or drastically deficient. The result is that phenylalanine cannot be broken down, and it accumulates in large quantities throughout the body. Normally, phenylalanine is converted to tyrosine. Because tyrosine is involved in the production of melanin (pigment), people with PKU usually have lighter skin and hair than other family members. Without treatment, phenylalanine accumulation in the brain causes severe mental retardation. Treatment is usually started during babyhood; delaying such treatment results in a significantly lowered IQ by age one. Treatment involves a diet low in phenylalanine (look for warnings aimed at people with PKU on cans of diet drinks containing the artificial sweetener aspartame, which is made from phenylalanine). PKU strikes about one out of every 20,000 newborns. Because it is so important to start treatment immediately, many states require that all infants be tested for the disease within the first week of life.