Phenylketonuria (PAH)

Phenylketonuria (PKU) is caused by mutations in the PAH gene. This gene encodes for the enzyme phenylalanine hydroxylase, which breaks down the amino acid phenylalanine. The buildup of phenylalanine causes damage to the body, affecting mainly the brain. If left untreated, those with phenylketonuria suffer from intellectual disability, seizures, delayed development, and psychiatric problems. A musty odor from phenylalanine may be evident. Treatment is by a special low phenylalanine diet, which can allow for normal development if strictly adhered to. There are rarer, less damaging forms of the disease, known as non-PKU hyperphenylalaninemia.

Phenylketonuria is found in 1 in 10,000 to 1 in 15,000 new births. Since screening and prompt treatment are almost universal in the USA, the symptoms are very rarely seen. The disease is more common in some ethnic groups, such as Turks (1 in 2,600 births) and Irish (1 in 4,600 births). The condition follows an autosomal recessive pattern, typically requiring both parents to be carriers asymptomatically.

Sources

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