Phenylketonuria (PKU)

Phenylketonuria or PKU is an autosomal recessive metabolic disorder caused by a mutation in the enzyme phenylalanine hydroxylase. This reaction requires tetrahydrobiopterin as a cofactor and decreased tetrahydrobiopterin can also cause decreased activity of phenylalanine hydroxylase. This enzyme is necessary to metabolize phenylalanine to tyrosine. When the activity of the enzyme is reduced, phenylalanine accumulates and is converted to phenylketones, including phenylacetate, phenylpyruvate, and phenyllactate, that accumulate in the blood and urine. Because tyrosine can no longer be synthesized, it becomes an essential amino acid. Untreated phenylketonuria can lead to mental retardation, growth retardation, seizures, hypopigmentation, and eczema. Phenylketones contain an aromatic amino group, which causes a musty or mousy body odor in individuals, which is characteristic for phenylketonuria. If diagnosed early enough, a newborn can grow with normal brain development by decreasing phenylalanine in the diet. Phenylalanine is contained in aspartame containing products like NutraSweet.



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Pathophysiology

Decreased Phenylalanine Hydroxylase
Down-arrow Phoenix-Aladdin Hydra-lace
This enzyme is necessary to metabolize phenylalanine to tyrosine. When the activity of the enzyme is reduced, phenylalanine accumulates and is converted to phenylketones, including phenylacetate, phenylpyruvate, and phenyllactate, that accumulate in the blood and urine.

Decreased Tetrahydrobiopterin Cofactor
Down-arrow BH4 Tetris-hydra
Phenylalanine hydroxylase requires tetrahydrobiopterin as a cofactor and decreased tetrahydrobiopterin can cause decreased activity of phenylalanine hydroxylase.

Autosomal Recessive
Recessive-chocolate
This disease is inherited in an autosomal recessive fashion.

Tyrosine Becomes Essential
Essential Tires
Because tyrosine can no longer be synthesized, it becomes an essential amino acid.

Phenylalanine Found in Nutrasweet
Nutrasweet packet
If diagnosed early enough, a newborn can grow with normal brain development by decreasing phenylalanine in the diet. Phenylalanine is contained in aspartame containing products like NutraSweet.

Symptoms

Musty or Mousy Body Odor
Mustache-mice with Odor
Phenylketones contain an aromatic amino group, which causes a musty or mousy body odor in individuals, which is characteristic for phenylketonuria.

Growth Retardation
C-clamp
Infants with phenylketonuria fail to grow at an appropriate rate and also fail to attain early developmental milestones.

Seizures
Caesar
Seizures are transient episodes of abnormal excessive neuronal activity in the brain. Seizures can be seen in individuals with phenylketonuria.

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Intellectual Disability

Tar Covered Book

Infants with untreated phenylketonuria can display severely impaired cognitive functioning with deficits in two or more adaptive behaviors.

Hypopigmentation

Hippo-pig with Hypopigmentation

Hypopigmentation, or loss of pigmentation in the skin, is commonly observed in individuals with phenylketonuria due to decrease in the amino acid tyrosine, which is used by melanocytes to make melanin.

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