Phenylketonuria (PKU)

Phenylketonuria (PKU) is an autosomal recessive genetic disorder characterized by the deficiency of an enzyme necessary for phenylalanine metabolism. The Guthrie Blood Test is a newborn screening tool critical in diagnosing PKU. The condition may present with frequent vomiting, irritability, cognitive impairment, and a distinctive musty odor. Treatment includes maintaining a low-phenylalanine diet by avoiding foods containing aspartame or protein. In combination with the PKU diet, the medication sapropterin (Kuvan) may help improve tolerance of phenylalanine ingestion.



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Cause

Autosomal Recessive Genetic Disorder
Recessive-chocolate
Phenylketonuria is an autosomal recessive genetic disorder characterized by an enzyme deficiency. Children of two heterozygous parents containing the recessive gene have a 25% chance of being affected and a 50% chance of being carriers of the trait. Males and females are affected equally.

Enzyme Deficiency
Enzymes Broken
Phenylketonuria is caused by the deficiency of an enzyme required to metabolize the essential amino acid phenylalanine. Phenylalanine is found in foods containing aspartame and high protein. Accumulation of unmetabolized phenylalanine may result in cognitive impairment.

Assessment

Failure to Thrive
Very Skinny Baby
Since PKU is not inherited, infants may experience failure to thrive since they cannot effectively metabolize the high amounts of phenylalanine present in the mother's blood. Symptoms include low birth weight, an abnormally small head, and frequent vomiting.

Irritability
Irritated
The child with PKU may demonstrate irritable, hyperactive, and erratic behavior. Behavioral, emotional, and social problems may emerge as the child grows into adulthood.

Musty or Mousy Odor
Mustache-mice with Odor
Excessive amounts of phenylalanine cause the patient to have a musty or mousy odor. The distinct odor is evident in the patient's breath, skin, or urine.

Cognitive Impairment
Cogs Impaired
Since the PKU patient lacks the enzyme required to metabolize phenylalanine, the accumulation of the amino acid may lead to cognitive impairment. Problems may include delayed development and intellectual disability. Additional neurological problems include seizures and tremors.

Interventions

Low Phenylalanine Diet
Down-arrow Phoenix-Aladdin with Nutritional-plate
A low-phenylalanine diet helps prevent brain damage leading to intellectual disability. Infants are given special formulas, such as phenex and lofenalac, that are low in phenylalanine while providing essential protein and nutrients for normal growth. As the patient matures, a diet low in protein and aspartame is

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recommended.

Low Protein Foods (Diet)

Down-arrow Mr. Protein

Patients with PKU should consume low protein foods. Natural sources of dietary protein, such as meat, eggs, dairy products, and nuts contain high amounts of phenylalanine, and thus cannot be metabolized.

Avoid Aspartame

Avoid-sign Ass-sweetener

Digested aspartame releases phenylalanine into the bloodstream. Instruct the patient to avoid artificial sweeteners, such as NutraSweet and Equal. The patient should also avoid diet soda and drinks containing aspartame.

Sapropterin (Kuvan)

Soap-rope

Sapropterin (Kuvan), or BH4, is a medication indicated to increase tolerance of phenylalanine ingestion. Since Kuvan is a new drug, the FDA is conducting ongoing studies to evaluate the medication's efficiency. For optimal results, the drug is indicated to be used with the PKU diet.

Considerations

Guthrie Blood Test

Guthrie-gravy Blood Test

Immediate detection and intervention of PKU is critical for preventing major health problems. The Guthrie Blood Test is a newborn screening tool used after the infant is 24 hours old and has ingested dietary protein. If the infant is discharged before 24 hours old, then a second newborn screening test is performed at 1-2 weeks following birth. The blood sample is collected by performing a heel stick.

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