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PHENYLKETONURIA: AN INBORN ERROR OF METABOLISM

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Abstract*: Phenylketonuria (PKU) is an inherited amino acid metabolic disorder that causes an increase in the blood of a chemical known as phenylalanine. Phenylalanine comes from a person's diet and is used by the body to make proteins. Phenylalanine is found in all food proteins and in some artificial sweeteners. It is an inherited condition caused by a defect in the PAH gene. Both parets must pass on a defective version of the PAH gene for their child to inherit the disorder. Mental deficiency is the most important clinical feature, but convulsion, tremor, rhythmic rocking of the body also occurs. It can be easily detected with a simple blood test. Without dietary treatment, phenylalanine can build up to harmful levels in the body, causing mental retardation and other serious problems. The best treatment for PKU is a diet containing phenylalanine about 30-50 mg/kg body weight and special formula that's often known as Lofenalac. When baby is old enough to eat solid foods high protein foods such as meat, cheese, poultry, eggs and milk are not permitted. Instead the diet is supplemented with specially formulated low protein wheat based products, including bread, flour, biscuits and pasta .This diet is very effective at lowering level of phenylalanine and allows normal growth and development of the child.*

Keywords: *PKU, metabolic disorders, low- protein diet, inborn errors, phenylalanine, PAH gene.*

Introduction: Phenylketonuria is a hereditary condition characterized by mental retardation and the presence of phenylpyruvic acid in the urine. Several cases have been reported from all parts of the world since the first cases were described by Folling in 1934. Phenylketonuria (PKU) is due to a genetic deficiency of phenylalanine hydroxylase of an enzyme essential for the conversion of phenylalanine to tyrosine. In consequence, the blood in particular and also the cerebrospinal fluid contain amount of phenylalanine and its pyruvate, lactate and acetate derivatives greatly in excess of normal.[1] Body needs tyrosine to create neurotransmitters, such as epinephrine, nor epinephrine, and dopamine. PKU is caused by a defect in the gene that helps to create phenylalanine hydroxylase. When this enzyme is missing, the body is unable to break down phenylalanine. This causes a buildup of phenylalanine in the body. Early diagnosis and treatment can help relieve symptoms of PKU and prevent brain damage. [2]

Causes–PKU is an inherited condition caused by a defect in the PAH gene. The PAH gene helps create phenylalanine hydroxylase, the enzyme responsible for breaking down phenylalanine. A dangerous buildup of phenylalanine can occur when someone eats high-protein foods, such as eggs and meat. Both parents must pass on a defective version of the PAH gene for their child to inherit the disorder. If just one parent passes on an altered gene, the child won't have any symptoms, but they'll be a carrier of the gene. [2]

Clinical Feature: Phenylalanine plays a role in the body's production of melanin, the pigment responsible for skin and hair color. Therefore, infants with the condition often have lighter skin, hair, and eyes than brothers or sisters without the disease. Other symptoms may include: 1. Delayed mental and social skills. 2. Head size much smaller than normal. 3. I.Q. of 30 or less. 4. Hyperactivity. 5. Jerking movements of the arms or legs. 6. Mental disability. 7. Seizures. 8. Skin lesions such as dryness, dermatitis. 9. Tremors. 10. Eczema. 11. Convulsion [3,1]

Diagnosis: PKU can be easily detected with a simple blood test. All states in the United States require a PKU screening test for all newborns as part of the newborn screening panel. The test is

generally done by taking a few drops of blood from the baby before the baby leaves the hospital. If the screening test is positive, further blood and urine tests are required to confirm the diagnosis. Presence of phenylpyruvic acid in urine and increase in phenylalanine levels in the plasma. Genetic testing is also done. [3]

Treatment

Diet: Elevation of serum phenylalanine can be effectively controlled by reducing the content of this amino acid in the diet. Complete elimination is not indicated, since phenylalanine is an essential amino acid and sufficient amounts must be supplied to ensure normal physical growth. **[1]** The daily intake of phenylalanine of a child is reduced from the normal level of about 100 mg/kg body weight to about 30-50 mg/kg body weight. If the level exceeds 10mg/100 ml symptoms of Phenylketonuria may appear. Low phenylalanine protein hydrolysates are being marketed by some firm in western countries. These preparations when supplemented with carbohydrates, fats, vitamins, iron and other minerals meet the nutritional requirements of children. Such diets are effective in bringing down the plasma phenylalanine levels to the normal range. If the treatment is started shortly after birth the therapy is effective in bringing about normal development. It is, however, less effective in older infants and does not effect on children over 6 years as the damage to the brain has already occurred and cannot be corrected. **[1]**

Infants with PKU can't be fed breast milk and must consume a special formula that's often known as Lofenalac. When baby is old enough to eat solid foods, it'll need to avoid letting them eat foods high in protein. These include: Eggs, Cheese, Nuts, Milk, Beans, Chicken, and Beef **[2]**

Basic Principles of the Diet for PKU

- Treatment consists of a diet containing only the amount of phenylalanine which is essential for growth and development.
- Meat, fish, cheese, eggs and nuts are rich in protein and therefore phenylalanine so they are not allowed.
- Other foods which contain some protein such as potato, milk and cereals are given in small measured quantities so that the blood phenylalanine, which is measured regularly, is kept within safe limits. These measured foods are spread out between the day's meals. The quantities allowed will vary from person to person.
- Most fruits, some vegetables and salads can be taken in normal quantities but avoid excess use. Sugar, jam, syrups, and fats such as butter, lard and cooking oil can be used freely. There are many manufactured foods which are low in protein and are available on prescription for the person with PKU, including low protein bread, biscuits, flour, spaghetti and other pasta which can all be taken freely and can be used to provide variety in the diet.
- As high protein foods cannot be eaten these have to be replaced by a special protein mixture from which the phenylalanine has been removed. These protein substitutes are all available on prescription and will provide the essential 'safe' protein for growth and development. It is crucial that these are taken regularly with meals and evenly spread out over the day.
- Vitamins and minerals should also be supplemented into the diet. Some protein substitutes include these, but if they are not then vitamin and mineral supplements are available on prescription.
- Specially formulated low protein wheat based products are available, including bread, flour, biscuits and pasta. Some of these are available on prescription; others can be purchased through pharmacy or at specialist stores. **[4]**

Medication: The United States Food and Drug Administration (FDA) recently approved sapropterin (Kuvan) for the treatment of PKU. Sapropterin helps lower phenylalanine levels. This medication must be used in combination with a special PKU meal plan. However, it doesn't work for everyone with PKU. It's most effective in children with mild cases of PKU. [2]

Conclusion: Thus it can be said that Phenylketonuria is an inborn error of amino acid metabolic disorder due to genetic deficiency characterized by mental retardation caused by absence of the enzyme phenylalanine hydroxylase. It can be cure by early diagnosis and proper dietary treatment.

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