AMINO ACID DISORDERS

Phenylketonuria
Hyperphenylalaninemia

What is Hyperphenylalaninemia?

Hyperphenylalaninemia is a term that encompasses several disorders that result in increased phenylalanine in the body. In majority of cases, the cause is due to a deficiency of the enzyme phenylalanine hydroxylase. Other cases can be due to defects in the metabolism of the cofactor tetrahydrobiopterin.[1] In the Philippines, all cases of defects in tetrahydrobiopterin have been determined to be due to 6-pyruvoyl tetrahydropterin synthase {6-PTPS) deficiency.

WHAT IS MILD HYPERPHENYLALANINEMIA?

Mild hyperphenylalaninemia is a mild form of phenylketonuria. It is a condition often diagnosed only incidentally during screening for classic phenylketonuria and the increases in phenylalanine levels are insufficiently elevated to cause neurological damage.[2]

CLINICAL MANIFESTATIONS

Children are normal at birth and are often asymptomatic. Regular monitoring of phenylalanine levels should be done.

PATHOPHYSIOLOGY

When the conversion of phenylalanine to tyrosine is blocked, phenylalanine that is not used for protein synthesis accumulates in body fluids or is converted to other metabolites.[1]

Inheritance: autosomal recessive [1•5]

CONFIRMATORY TESTING

Plasma amino acids, urine proteins, DHPR enzyme assay. Further confirmatory testing (i.e. BH4 loading) may be required after referral to a metabolic specialist.

Overview of Disease Management

Mild hyperphenylalaninemia does not require treatment.[5] But regular monitoring of levels until 5 years of age is needed. Should levels exceed 350, consultation with a medical specialist is suggested. Initiation of management should be done in consultation with an attending physician/ metabolic specialist.

Prognosis

Patients affected with mild permanent phenylalaninemia will develop normally without treatment, however, continuous monitoring of levels is recommended. Increasing levels may warrant treatment.

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What is Hyperphenylalaninemia?

Hyperphenylalaninemia is a general term that means that phenylalanine, an amino acid, accumulates in the blood and tissue of the body. This can be detected through newborn screening. The cause of increase of phenylalanine may be due to either a lack of enzyme (chemical scissors) or a lack of the co-factor (a substance needed by the body to allow the enzyme to function properly).

WHAT CAUSES MILD HYPERPHENYLALANINEMIA?

Mild hyperphenylalaninemia is a mild form of phenylketonuria, a condition which causes accumula on of the amino acid phenylalanine in the body due to a slight decrease of the enzyme or chemical scissor known as phenylalanine hydroxylase.

This condition is inherited. The gene is contained in the genetic material that we inherited from our parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with mild hyperphenylalaninemia have one working and one non-working gene coding for mild hyperphenylalaninemia. They do not manifest the disease but can pass them on to their children. They are known as carriers.

If the child inherits the non-working gene from both parents, he or she will have the condition. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

SYMPTOMS OF UNTREATED HYPERPHENYLALANINEMIA

Children with mild hyperphenylalaninemia do not have any symptoms. While the amino acid-phenylalanine is increased in their body, it has been determined that these increases are not harmful to the child. However, monitoring of their blood phenylalanine levels are required.

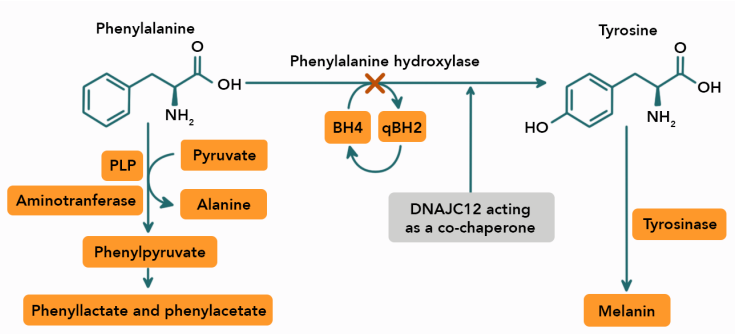
What is the treatment of mild hyperphenylalaninemia?

There is no need to treat mild hyperphenylalaninemia and your child can have a regular diet. However, it is recommended that periodic monitoring of blood phenylalanine levels should be done.

What should I do if my baby is sick?

Since there is no treatment needed for this condition, there are no special recommendations to be done when your child is

Hyperphenylalaninemia patient's breakdown of protein:



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