AMINO ACID DISORDERS

Phenylketonuria
Hyperphenylalaninemia

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.

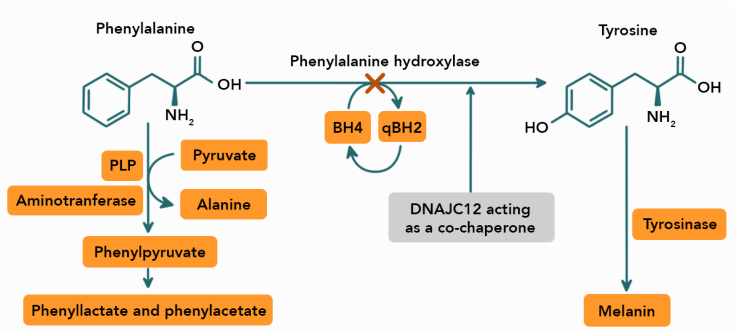
What is the treatment of mild hyperphenylalaninemia?

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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