Need to talk? Call us 0845 2412173 Monday to Friday 9am to 5pm

Climb™

Supporting those affected by Inherited Metabolic Disorders

Phenylketonuria

Phenylketonuria is a disorder of amino acid metabolism. Amino Acids are the "building blocks" of protein. The body breaks down some amino acids. In this disorder there is a deficiency or absence of the liver enzyme phenylalanine hydroxylase. This enzyme is needed to be able to breakdown the amino acid, phenylalanine. This causes a build up of phenylalanine in the brain, which can, with time, cause profound mental deterioration. In most developed countries phenylketonuria (PKU) is tested for at birth.

If a person has been diagnosed with Phenylketonuria at birth then symptoms should generally not occur as a result of a prescribed dietary treatment. If this condition is not diagnosed through a routine neonatal screening programme, symptoms appear during the first few weeks of life. As babies, those affected may have some form of developmental delay and by age one are not reaching their expected developmental milestones. Symptoms include vomiting, irritability, being abnormally drowsy and lethargic, as well as feeding difficulties. Individuals may also have an unusual musty odour. As the phenylalanine levels increase a rash that is similar to eczema that causes itching, redness and blistering can develop. There can be jerky muscle movements, hypertonicity, hyperactivity, ataxia, and seizures.

Synonyms

Alternative names for this condition are:

- Classical Phenylketonuria
- Hyperphenylalanemia
- Phenylalanine Hydroxylase Deficiency
- Phenylalaninemia
- PKU

Further information about this condition is available from Climb.

Disclaimer

Please read our disclaimer and information on data protection.

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