

Family Friendly Factsheet

PKU



PKU is a rare inherited disorder which prevents the body from breaking down the essential amino acid phenylalanine. As an essential amino acid, phenylalanine must be sourced from the diet in order to create vital proteins for growth. Although essential, an excess of phenylalanine can cause serious health problems. PKU sufferers have a deficiency of the enzyme phenylalanine hydroxylase which stops the conversion of phenylalanine to tyrosine, a standard reaction in non-sufferers.

The lack of phenylalanine hydroxylase creates a buildup of phenylalanine in the blood, where it is converted to phenylketone which is detected in the urine and used to diagnose the condition. Infants may display early symptoms such as vomiting, eczema, irritability, a mousy odour to the urine, increased muscle tone and more active muscle reflexes. If left untreated, PKU can create major health problems such as mental difficulties, brain damage, seizures, microcephaly and decreased body growth. Babies are screened for PKU shortly after birth using the heel-prick test and the test is then repeated after approximately two weeks to detect any signs of the condition which were initially overlooked.

Sufferers must follow strict rules in order to manage the levels of phenylalanine in their blood by following a special diet. In some cases medicine is also used to control the disorder, by keeping blood levels between 2 and 10 mg/dl. As phenylalanine is an amino acid, many foods which are high in protein contain a source of phenylalanine; including meat, fish, eggs, cheese and other dairy foods. Starchy foods must also be restricted and many diet drinks which contain the sweetener aspartame must be avoided. As this strict diet restricts the intake of

important food groups, the diet must be supplemented with vitamins, minerals and an artificial protein to ensure that the body receives all of the essential nutrients and that growth is not restricted.

Although the condition is treatable, there is no known cure, meaning that a low-phenylalanine diet is a lifelong requirement. At a certain age, a 'challenge' can be introduced to determine whether the sufferer still requires a low-phenylalanine diet or whether the form of PKU from which they are suffering is variant or transient. Although unlikely, there is a possibility that a low-phenylalanine diet is no longer required. However, despite this, most professionals recommend that the sufferer continues to follow a low-phenylalanine diet to encourage maximal development. The treatment of the condition is assisted by a specialist team of dietitians, doctors, biochemists, specialist nurses, psychologists and social workers, all of whom provide help and support to the sufferer to control their condition.

Recent research has brought the hope that enzyme or gene therapy could be used to treat the condition in the future, making it possible for sufferers to consume a more varied diet. In enzyme therapy, the sufferer is given an enzyme supplement, which may stimulate the breakdown of phenylalanine, reducing the levels present in the blood and therefore lowering the risk of serious damage occurring. In gene therapy, the section of DNA which determines the occurrence of PKU is altered and the expression of the condition is suppressed. These treatments are still in the stages of development and further research is underway to determine the role of these treatments in genetic disorders such as PKU.