**Phenylketonuria (PKU)**

**Summary**

- The newborn screening test, which is available to all newborns, can detect whether a child has phenylketonuria (PKU).
- Children with PKU can grow and develop normally, if they are carefully monitored and given a diet low in phenylalanine.
- Without a special diet, PKU can cause brain damage.

PKU is a genetic disorder that prevents the normal breakdown of a protein found in some foods. Proteins are made up of building blocks called amino acids; in PKU, the body cannot break down the amino acid phenylalanine. Because phenylalanine cannot be broken down (metabolised) normally, it builds up in the blood and tissues.

This build-up prevents the brain from developing properly. Progressive intellectual disability results if PKU is not treated from early infancy. With treatment, children with PKU can grow and develop normally.

**A simple test for newborns can detect PKU**

All newborn babies are tested for PKU through the newborn screening test taken during the first few days of life. About one in 10,000 newborn babies are affected with PKU.

See our video on [newborn bloodspot screening and PKU](betterhealth.vic.gov.au).

**PKU is a genetic disorder**

PKU is a genetic disorder that occurs when a baby has two faulty copies of the PKU gene. Genes are inherited particles found in all cells of our body. They form the blueprints for all growth, development and functions of the body. Each gene is responsible for sending a message that tells the cells how to make a particular product. The PKU gene tells the cell to make an enzyme that breaks down the amino acid phenylalanine. Faults in the genes (mutations) may cause problems in the body because the correct message is not being sent. In PKU, the cells are not making the enzyme that breaks down phenylalanine, so it builds up in the blood and tissues.

There are two copies of every gene in each cell—one copy we get from our mother and the other copy we get from our father. If a person has one normal copy and one faulty copy of the PKU gene, they are called carriers. Carriers of PKU are healthy because the normal copy overrides the faulty gene. This means the cells produce enough enzymes to prevent the build-up of phenylalanine.

When both parents are carriers of the faulty PKU gene, their child will be born with PKU if they receive one copy of the faulty gene from each parent. When both parents are carriers, the possibilities in each pregnancy are:

- 1 in 4 chance of having an affected child
- 2 in 4 chance of having a child that is a carrier
- 1 in 4 chance of having a child that is not a carrier.

**Children with PKU need special diets**

A baby with PKU will need to be on a special milk formula. As the child gets older, they will need a diet that is low in phenylalanine. Regular blood tests are needed to monitor the level of phenylalanine in the blood. This special diet is essential during childhood to prevent damage to the brain while it is still growing. The diet should be continued throughout life to optimise school performance, concentration and the ability to think clearly.

**Where to get help**

- Your doctor

[betterhealth.vic.gov.au](betterhealth.vic.gov.au)
Things to remember

- The newborn screening test, which is available to all newborns, can detect whether a child has PKU.
- Children with PKU can grow and develop normally, if they are carefully monitored and given a diet low in phenylalanine.
- Without a special diet, PKU can cause brain damage.

This page has been produced in consultation with and approved by:

Murdoch Children's Research Institute

Content on this website is provided for information purposes only. Information about a therapy, service, product or treatment does not in any way endorse or support such therapy, service, product or treatment and is not intended to replace advice from your doctor or other registered health professional. The information and materials contained on this website are not intended to constitute a comprehensive guide concerning all aspects of the therapy, product or treatment described on the website. All users are urged to always seek advice from a registered health care professional for diagnosis and answers to their medical questions and to ascertain whether the particular therapy, service, product or treatment described on the website is suitable in their circumstances. The State of Victoria and the Department of Health & Human Services shall not bear any liability for reliance by any user on the materials contained on this website.

For the latest updates and more information, visit www.betterhealth.vic.gov.au