

## Phenylketonuria (PKU)

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.

### 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
- Genetic Health Services Victoria Tel. (03) 8341 6201

### 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:**

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