



## INFORMATION FOR PARENTS/CARERS PHENYLKETONURIA - (PKU)

**This fact sheet has been put together to answer some questions you may have after talking to your PKU clinician and before attending your appointment.**

### What is Phenylketonuria (PKU)?

PKU is an inherited disorder which prevents the normal use of protein food, and causes changes in the body chemistry which without treatment can lead to severe mental retardation.

### How is PKU detected?

All newborn babies are screened for several treatable disorders by a blood test taken at about three days of age. Phenylketonuria (PKU) is one of them. About 1 in 10,000 – 14,000 Australians has PKU, and between 20-25 babies are diagnosed with PKU each year, of which about 8 – 10 babies are diagnosed in New South Wales and the Australian Capital Territory.

### How does PKU cause problems?

PKU is a condition in which the body is unable to break down **one** of the protein building blocks from food. These building blocks are called amino acids and **one of them is phenylalanine**. In PKU the phenylalanine cannot be processed (metabolised) normally and builds up in the blood and tissues. The high phenylalanine level can prevent the brain from developing as it should. Progressive mental retardation results **if** the condition is not treated in early infancy.

### Why did my child get PKU?

PKU is an inherited disorder, which occurs when both parents have a mutation (mistake) copy of the PKU gene and pass it on to their baby. Genes are the particles of heredity which occur in all cells in the body, and form the “blue-prints” for all the bodily processes and traits such as hair colour, blood group and so on.

A parent who has one copy of a ‘PKU’ gene and one copy of a normal gene is a carrier of PKU but is **perfectly healthy**. When both parents are carriers there is a one in four chance **in each pregnancy** that their baby will inherit a ‘PKU’ gene from each parent, and so be born with PKU.

This type of inheritance is called autosomal recessive inheritance, and is explained more fully in the PKU handbook you will receive when you come to the hospital. Most families cannot ‘trace’ PKU in any of their relatives.

There is nothing the parents of a child with PKU could have done to prevent their child having PKU.

**Is my child already damaged (before the diet was started)?**

Your child is **not** damaged at all as the diagnosis of PKU is made early in the newborn period. Your baby would have had normal phenylalanine levels while in utero. This is because the mother's metabolism can clear phenylalanine before it accumulates. Only after birth does the phenylalanine level rise.

**Will my child grow normally?**

Children with PKU treated from early infancy grow and develop quite normally providing the treatment is followed. A special low phenylalanine diet is given, and regular blood tests to measure the phenylalanine level are sent to the laboratory.

**The diet for PKU is essential in childhood to prevent damage to the growing brain.**

Continuing the diet during adolescence and adult life gives the best outcome in school performance, concentration and ability to think clearly. Apart from needing a diet, children with PKU should be treated exactly as other children. They are neither more nor less likely to get colds and coughs or other illness. Routine immunisations should be given at the usual times. Most medicines can be given safely, but **check** with your PKU clinic doctor.

**When your baby is small, the diet is relatively easy, and allows you to breast or bottle feed plus give the special protein supplement the dietitian will tell you about when you come to the hospital.**

If your child is diagnosed with PKU, the family may be admitted to hospital for two nights so that confirmatory tests can be carried out and the PKU team can meet with you and discuss the disorder carefully so you fully understand it. The team consists of your clinician, dietitian, social worker, clinical nurse consultant for metabolic disorders and the laboratory staff .

**What about other children in the family**

Both the mother and father of a child with PKU 'carry' the disorder, even though they are perfectly healthy. In each pregnancy there is a 1 in 4 chance that the baby will be affected.

*\* Information obtained from the PKU Handbook 1996. Prepared by the Dietitians Working Party of the Australasian Society for Inborn Errors of Metabolism*

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