PHENYLKETONURIA

(PKU)

A LEAFLET FOR PARENTS

WHAT IS PKU?

It is an inherited disorder which prevents the normal breakdown of protein due to an enzyme deficiency. If untreated it can lead to severe mental handicap and learning difficulties.

HOW DID THE DOCTOR FIND OUT?

All new-born babies should have a test for the condition between the 5th and 8th day after birth. The test on your baby was positive and further tests have shown that your child has Phenylketonuria. PKU is not a single condition but a group of disorders of varying severity. About 1 in 10,000 children in the UK are affected and between 60 - 70 new cases are born each year.

HOW IS PKU TREATED?

The treatment is very successful. The food we eat contains protein, which is made up of smaller parts called amino acids. In PKU one of these amino acids, called phenylalanine, cannot be broken down in the body in the usual way. Phenylalanine therefore builds up in the person's blood. Without treatment this leads to impaired brain development. However, this can be prevented by reducing the amount of protein eaten in the diet. Your child will therefore need a special low protein diet. The diet must be low in phenylalanine but otherwise well balanced to allow for growth and good health. Your hospital dietitian will help you to understand about this and explain fully what to do.

WHAT SHOULD PARENTS DO?

Apart from the diet, treat your child like other children, and try not to be over sympathetic or protective. A positive and encouraging attitude by all the family is much more likely to lead to acceptance of the treatment and help the child. The diet may seem complicated at first but the dietitian will give you a lot of support and help when you need it.

WILL YOUR CHILD GROW UP OKAY?

For all children with PKU who are treated from infancy, the answer is yes. Provided your child's PKU is well controlled he/she should be able to lead a normal life. Life expectancy is normal.

However, if dietary control is poor there can be problems with schooling and behaviour. The child may not achieve as much as he/she is capable of if the diet is not well controlled.

IS THE DIET FOR LIFE?

The current recommendations are that it is probably safer to continue with diet life long. For girls with PKU a strict diet is particularly important just before and during pregnancy to avoid damage to the developing baby.

WHAT ABOUT OTHER CHILDREN IN YOUR FAMILY?

Both 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. All babies are routinely screened for PKU.

FURTHER INFORMATION

A more detailed account of the condition is given on our website, the Society helpline and in the booklet 'The Child with Phenylketonuria' which is available from the Society. Frequently Asked Questions are on the NSPKU website.

The Society will help you to meet other parents in the same situation.

It is run by parents and adults with PKU and has:

An Honorary Medical Advisory Panel
Quarterly Magazines
Annual Conference/Holiday Weekend
Regional Day Conferences
Informal Meetings
Cookery Workshops and Demonstrations
Dietary Information Booklet
A series of information leaflets
Posters for clinics, surgeries and promotional events
Member of the European Society for PKU
Other publications are in the course of preparation

BASIC PRINCIPLES

The PKU diet should be supervised by a dietitian.

WHAT CAN MY CHILD EAT?

Most fruit, some vegetables, salads, sugar, jam, syrups and fats such as butter and cooking oil can be used quite freely. There are special manufactured foods available on prescription from the GP for the child with PKU. These are low in protein. These include low protein bread, biscuits, flour, spaghetti and other pasta which can all be taken freely and can be used to provide variety in your child's diet.

WHICH FOODS HAVE TO BE AVOIDED?

Meat, fish, cheese eggs, nuts, milk and soya are rich in protein and therefore phenylalanine so they are not allowed. Ordinary flour, cakes, biscuits and pasta also contain too much protein for the child with PKU.

CAN MY CHILD EAT ANY PROTEIN?

Some foods like potato and cereals contain small amounts of protein and they are given in small measured quantities. These measured foods are spread out between the day's meals and snacks. The blood phenylalanine is measured regularly to monitor the diet.

WHERE WILL MY CHILD GET THE PROTEIN THEY NEED TO GROW NORMALLY?

As high protein foods cannot be eaten by your child these have been replaced by a special protein mixture from which the phenylalanine has been removed. There is a large range of protein substitutes in a variety of forms which are available on prescription and these will provide the essential 'safe' protein for growth and development. Your paediatrician or dietitian will advise you which are most suitable for your child, the amount required and how it should be taken.

The protein substitute is an extremely important part of the diet for the PKU child and it must be taken in divided doses spread out over the day. This helps to keep the phenylalanine levels steady throughout the day.

Produced by the National Society for Phenylketonuria
and its Medical Advisory Panel.

N S P K U

The National Society for Phenylketonuria (United Kingdom) Ltd.

The Society is a registered charity.
It offers support to people with PKU and their families by producing various publications including a quarterly newsletter, organising formal and informal meetings and conferences.

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