WHAT IS PHENYLKETONURIA?

Phenylketonuria (PKU) is an inherited, autosomal recessive disorder, which affects the way proteins are metabolised.

■ It is caused by a defective enzyme, which should convert the amino acid phenylalanine into tyrosine.

■ This results in a build up of phenylalanine, a state called hyperphenylalaninaemia.

■ Hyperphenylalaninaemia affects normal growth and development, especially of brain tissue.

SCREENING

■ The aim of screening is to detect the presence of PKU sufficiently early to enable the initiation of treatment and the prevention of mental retardation and later morbidity.

■ A blood screening test for PKU was developed in the USA by Guthrie and was introduced into the UK in the late 1960s.

■ The test is now offered to all infants between six and 10 days old.

■ Confirmed abnormal results should also be notified to the PKU Register.

■ Some families may require formal genetic counselling and should be referred to a clinical geneticist.

INCIDENCE/PREVALENCE

■ One of the enzyme defects that cause PKU is present in approximately 1/10,000 births in northern Europeans.

■ One person in every 50 of the general UK population carries a mutated copy of the affected gene.

■ Incidence varies according to country and race and is 1/4,500 in Ireland, 1/16,000 in Switzerland, 1/50,000 in US blacks and 1/15,000 in US Caucasians (Hofman et al, 1991).

■ Parents who have a child with PKU have a one in four risk of any subsequent children being affected.

TREATMENT

■ The combination of screening and a low phenylalanine diet has greatly improved the prospects for those with PKU.

■ When diagnosis is confirmed a special low phenylalanine feed is given until levels of phenylalanine fall to within the treatment range (this is higher than the normal reference range). This reduction in level usually takes a few days.

■ Small amounts of breast or formula milk are then introduced to provide a limited amount of phenylalanine.

DIETARY MANAGEMENT

The low phenylalanine diet consists of three parts:

■ A protein substitute, which includes all other amino acids except phenylalanine and an additional vitamins and minerals supplement.

■ A small measured quantity of phenylalanine, from foods such as potatoes, baked beans and breakfast cereals (Cockburn, 1993).

■ Foods low enough in phenylalanine to be permitted in normal quantities (there are only a few, which include most fruit and vegetables, sugar, butter, boiled sweets and some squashes). In addition many specially produced low-protein products such as low protein flour, bread and pasta are available on prescription.

NURSING IMPLICATIONS

■ It is recommended that people with PKU follow the special diet for life.

■ All immunisations can proceed normally.

■ If a child’s phenylalanine control is kept within the acceptable range, growth and development will not be affected. Therefore, extra developmental assessments should not be required.

■ After diagnosis the family may need extra support in coping with shock, anxiety or stress.

■ It is important that dietary advice is consistent. The child’s dietitian should, therefore, be the only person giving advice.

REFERENCES


WEBSITE

National Society for Phenylketonuria: http://web.ukonline.co.uk/nspku

FURTHER READING