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PKU DIETETIC MANAGEMENT IN CHILDREN

This article aims to give a brief overview of the dietary management for infants and children with PKU and to highlight some practical issues from our experience. We will also include case studies demonstrating points raised in this article.

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Sharan and Lucy are Paediatric Dietitians who have varied clinical experience and are now specialising in the area of metabolic dietetics. They have worked together for several years.

Phenylketonuria (PKU) is an inborn error of metabolism with an incidence in the UK of one in 10,000. It is caused by an inability to convert the essential amino acid, Phenylalanine (Phe) to Tyrosine, the first step in the breakdown of Phe. In the UK, PKU is diagnosed through newborn screening. It is treated by a diet low in Phe, which should be introduced as soon as possible after diagnosis. Without appropriate treatment the Phe levels in the blood accumulate and can lead to learning difficulties (1). Prompt and effective treatment means that children with PKU can reach their potential and grow up healthily. Following a strict low Phe diet is the only way to achieve acceptable plasma Phe concentrations.

The aim of dietetic treatment is to promote normal growth and development in the child with PKU. This is achieved by restricting Phe intake, supplementing adequately with Phe-free amino acids (protein substitute) and ensuring adequate energy from non-protein sources (2). As the diet is very restrictive, it is also supplemented with a comprehensive range of vitamins, minerals and trace elements. These may be provided by the protein substitute or as a separate supplement.

RESTRICTING PHE INTAKE

Natural sources of dietary protein are severely restricted in order to achieve optimal Phe control yet meet requirements for this essential amino acid. Phe is measured using a system of 'exchanges' where one 'exchange' = 50mg Phe or 1.0g protein. Phe tolerance is variable. Children with moderate or severe PKU usually tolerate 200 to 300mg Phe per day (four to six exchanges).

Supplementing adequately with Phe-free amino acids (protein substitute): protein requirements can only be met by the use of Phe-free protein substitutes. These provide amino acids with a similar profile to dietary protein of high biological value but exclude Phe. The protein substitute will provide the RNI (Recommended Nutrient Intake) for protein. However, the RNI is based on diets providing sufficient high quality dietary protein from animal sources and assumes complete digestibility. When amino acids supply most of the protein intake, requirements may be greater than the RNI for protein (see Table 1).

Table 1: Guidelines for total protein requirements in PKU (3)

Age (years)	Total protein (g/kg)*
0 – 2	3.0
3 – 5	2.5
6 – 10	2.0
11 – 14	1.5
> 14	1.0 (maximum 80g/day)

* Protein equivalent from protein substitute and Phe exchanges (natural protein)

ENSURING ADEQUATE ENERGY FROM NON-PROTEIN SOURCES

Providing adequate energy within the limitations of the Phe restriction necessitates the use of low Phe foods, e.g. fruits, specified vegetables and specially manufactured low protein foods. This requires:

- a careful choice of the specially manufactured low protein foods;
- creative and imaginative use of the fruits and vegetables which are of lowest Phe content;
- use of energy supplements and high energy manufactured foods as appropriate.

Table 2: Useful tools for the weaning home visit

Example nutritional labels
Baby jars
Example weighed portion sizes of foods to be counted
Digital weighing scales
Comprehensive folder of information
Weaning booklets

PRACTICAL ISSUES

Children with PKU represent a variety of challenges throughout childhood to the paediatric dietitian. Here, we summarise some of the issues faced in Sheffield and approaches that have proved successful in tackling them.

STABILISING PHE LEVELS ON DIAGNOSIS

The immediate concern for a baby with newly diagnosed PKU is the raised blood Phe level. The dietitian must intervene immediately by introducing a Phe-free formula to supplement or replace the infant's milk. Pre-treatment blood Phe levels detected by newborn screening can range widely; in our experience from 800 μmol/l to 3,000 μmol/L. The target range is 120 to 360μmol/L (4). Establishing stable levels within this range in the first few weeks of life can be challenging due to a number of factors:

- Difficulty in estimating Phe intakes in breast fed babies
- Fluctuating levels due to periods of rapid and slower growth in newborns
- Parental anxiety surrounding diagnosis

It is important to prepare the family at the first meeting for regular blood tests and changes to their child's dietary regimen until good control is established. A trial and error approach is often necessary, especially with breastfed infants as Phe intakes can only be estimated. Even when the infant's Phe levels are stable within the target range, they are still subject to rise and fall during periods of rapid or slower growth. Blood Phe levels should be monitored once or twice weekly in the first few months of life in order to make appropriate dietary changes. Parents should be reassured that occasional high or low levels will not affect the infant's overall development, but action should be taken to avoid levels outside the target range for prolonged periods (4).

WEANING

Weaning can be a challenging process for parents of children without PKU let alone for those whose child requires a very strict natural protein restriction. It is one of the most complicated stages and it is crucial that parents understand the process in order to maintain control of blood Phe levels.

Weaning education can be broken down into four stages:

1. Starting the weaning process, using natural and synthetic foods with negligible Phe content.
2. Introducing 'exchanges'. Starting to exchange a measured amount of formula/breast milk for a calculated solid exchange.
3. Introducing finger food and extending the range of manufactured low protein products offered. Encouraging a wider variety of exchange foods and low protein foods.
4. More detailed interpretation of nutritional labels and understanding of sauces/condiments allowed within the diet.

In practice, we have found that a weaning home visit is an effective way of imparting the advice. The parents feel more comfortable in their home environment and there is less pressure to rush through the information. We generally cover at least stages 1 to 3 during this visit as most parents are keen to have the overall picture. In order to aid comprehension, a variety of tools are used (Table 2).

The weaning tool developed and available from NSPKU is a useful resource to base the weaning visit on. It is important to follow up this visit with regular parental contact to provide reassurance and further advice, as queries often arise when the theory is put into practice.

DIFFICULTY MOVING ONTO NEW PROTEIN SUBSTITUTES

The range of protein substitutes available are designed to suit protein and micronutrient requirements for different age groups. Children with PKU

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need to change their protein substitute at least twice through childhood; at around one year and sometime between the ages of three and 10. Problems with tolerance can occur when the child moves onto a new protein substitute. There is often a change in consistency, stronger taste and children at these stages of life are often selective eaters.

During recent years, the protein substitute manufacturers have made efforts to improve product variety and flavours. Dietitians have helped to influence the choices these companies make and contribute to improved product tolerance. It is important that the dietitian keeps up to date with new products on the market which may be suitable for their patients. Other tips to improve tolerance include concentrating the Phe-free infant formula in preparation for the next stage product and encouraging patients to try an exposure-based approach when trialling a new product (5).

The number and volume of protein substitutes can usually be reduced as the child gets older with careful choice from the lower volume, higher protein equivalent products available to the older age groups.

The following cases illustrate how there can be considerable heterogeneity between individuals with PKU, even within the same family. The eventual Phe tolerance of the brothers in these case studies is significantly different despite their similar upbringing and dietary management. Their experience also highlights some of the dietary challenges that may present in practice.

Case Studies

Brother A (born November 1998)	Brother B (born November 2002)
Early Phe control Day 6 Guthrie result: 580µmol/L Peak Level: 1,250µmol/L Time taken to achieve stability: seven weeks	**Early Phe control** Day 3 Guthrie result: 770µmol/L Peak Level: 1,460µmol/L Time taken to achieve stability: two weeks
Early feeding Breastfed three days then changed to SMA Gold Started Weaning: 17 weeks Taking full exchange foods: seven months	**Early feeding** SMA Gold Started Weaning: 17 weeks Taking full exchange foods: seven months
Control Phe levels in range most of the time High levels due to: acute illness Occasional refusal to take prescribed volume of protein substitute	**Control** Phe levels in range much of the time High levels due to: acute illness Prolonged period of refusal to take prescribed volume of protein substitute
Range of exchange foods allowance At eight months - four At nine months - seven At two years - eight	**Range of exchange foods allowance** At eight months - five At one year - six At 13-16 months - four to seven At two to three years – three to five
Current exchange foods allowance and protein intake from protein substitute Since the age of four, nine exchanges 41g protein from protein substitute Total intake 1.2g/kg	**Current exchange foods allowance and protein intake from protein substitute** Since the age of four, three exchanges 60g protein from protein substitute Total intake 1.4g/kg
Growth Weight generally progressed along 9th to 25th centile with occasional dips Height very consistent between 0.4th to 2nd centile	**Growth** Weight consistent between 91st and 98th centile Height consistent on the 50th centile
Issues Difficulty moving onto next stage protein substitute during early childhood Prolonged use of feeding bottles Selective eating leading to battles over eating Behaviour problems Inadequate amount of protein substitute	**Issues** Dislike of flavoured protein substitutes, limited availability at time of need for next stage Swapping lunch box foods with friends

References
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2 'Management of PKU', NSPKU February (2004). A consensus document for the diagnosis and management of children, adolescents and adults with Phenylketonuria
3 Vanessa Shaw and Margaret Lawson (2007). Guidelines for total protein requirements in PKU and other amino acid disorders. Clinical Paediatric Dietetics, 3rd Edition, P 316
4 Medical Research Council Working party of Phenylketonuria. Recommendations on the dietary management of Phenylketonuria. Arch. Dis. Child. 1998 78:122-126
5 Van Rijn et al (2012): New Taste adaptation in IMD patients. DMIMD Plenary Session Abstracts April 2012

Sources of detailed practical information
1 Clinical Paediatric Dietetics, 3rd Edition. Vanessa Shaw & Margaret Lawson. 2007 p309-332
2 NSPKU website www.nspku.org and publications

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Questions relating to: *PKU dietetic management in children.*
Type your answers below and then **print for your records.** Alternatively print and complete answers by hand.

Q.1	What is the cause of PKU?
A	
Q.2	What is the aim of dietetic treatment and how is it achieved?
A	
Q.3	What is the Phe tolerance in children?
A	
Q.4	How is Phe measured when restricting intake in children?
A	
Q.5	How are protein requirements met?
A	
Q.6	What are the difficulties in establishing stable Phe levels in newborns diagnosed with PKU?
A	
Q.7	Outline the four weaning stages for infants with PKU.
A	
Q.8	Describe at least three useful dietetic tools for a home visit.
A	
Q.9	When do children with PKU need to move to a new protein substitute and what problems can occur?
A	

Please type additional notes here . . .