NHD
THE DIETITIANS' MAGAZINE

eArticle with CPD
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NUTRICIA Metabolics

Volume 4.19 - October 9th 2014

LIVING WITH PKU: THE TEENAGE YEARS

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Phenylketonuria (PKU) is the most common of inborn errors of metabolism and affects approximately one in every 10,000 births in the UK (one in 4,500 in Northern Ireland) (1). The inability to metabolise the amino acid, phenylalanine, which is caused by the partial or total absence of phenylalanine hydroxylase (PAH) enzyme activity, results in high plasma levels.

A raised plasma level of phenylalanine promotes the formation of neurotoxic substances, phenylpyruvic acid and phenylethylamine. If left untreated, significant cognitive impairment occurs.

A strict low phenylalanine (Phe) diet from birth is the treatment for PKU. The main aim of dietary treatment is to maintain plasma Phe levels within desirable limits (see Table 1). The exclusion of dietary protein sources is advised, replacement through low protein/Phe alternatives and Phe free protein substitutes is a key task for dietitians working with PKU patients. Ensuring nutritional adequacy through calorie, vitamin and mineral supplementation may be required at times. See Table 2 overleaf for the key principals of the low protein/Phe diet.

It is widely accepted now that PKU patients should follow a low protein diet for life (4). Although many PKU patients will be used to this diet from as far back as they can remember; it can be a challenge to stick to it, as they become more independent in their adolescent years. There are the usual pressures of being a teenager as well as adhering to a diet that isn't exactly

Emma has been working as a Paediatric Dietitian for five years and her caseload includes HETF, disability, coeliac disease, cystic fibrosis, PKU and childhood obesity. She has been a local Coeliac UK group organiser for 18 months.

Table 1: Desirable plasma Phe levels (1, 2)

Birth to 5 years	120-360µmol/L
>5 years	120-480µmol/L
>10 years	120-480µmol/L - however, levels up to 700µmol/L are acceptable for this age and into adulthood
Preconception and throughout pregnancy	100-250µmol/L - much tighter control required as Phe levels in the placenta will be much higher and, therefore, highly toxic to the foetus

Table 2: Key principals of the low protein/Phe diet

Dietary element	Examples	Comments
Foods not permitted	Meat, fish, eggs, cheese, nuts, seeds, soya, tofu, flour and flour based products: bread, biscuits, pasta, cakes; chocolate/ confectionary; sweeteners such as aspartame.	Check medications - aspartame is sometimes used in sugar-free drugs. Sucralose and saccharine are safe to be used. Foods containing aspartame will be labelled: 'contains a source of phenylalanine'.
Exchange foods (contain some phenylalanine)	Potatoes and potato products. Some fruits and vegetables, e.g. peas, sweetcorn, beans, banana, figs. Rice, breakfast cereals, dairy foods, e.g. milk, cream	One exchange = 50mg Phe 50mg Phe = 1.0g protein. These foods must be weighed and various weights of each food will yield one exchange, e.g. 25g peas = one exchange but 10g Weetabix is also one exchange. Due to the variation in PAH enzyme activity in patients, tolerance to Phe will vary. Therefore, there is variation between patients in the permitted amount of exchanges in order to maintain safe plasma Phe levels.
Free foods (low Phe foods to be eaten in normal amounts)	Many fruits and vegetables (excl those mentioned above), herbs, spices. Sugar and sugar products. Some cereals, fats and cooking sauces - some restrictions may apply. Rice milk, hemp milk; beverages such as tea, coffee, squashes/fizzy drinks. Alcohol - spirits, wine, ciders contain trace amounts of Phe therefore permitted.	Rice milk is suitable for children over five years of age. Limit to 400ml per day only. Not suitable for pregnancy. When using mixers in alcohol ensure they are aspartame free. Likewise for premixed alcoholic drinks, e.g. alcopops. Lager/beer or cream liquors contain enough Phe for them to have an exchange value.
Prescribed/ specialist low protein foods (exchange-free unless stated)	Bread, pasta, rice, biscuits, flour/bread/cake mixes, 'burger'/'sausage' mixes. Egg, milk and chocolate replacements, readymade snacks.	Prescribed via GP and available through home delivery service provided by the various low protein food/protein substitute manufacturers. Essential for growth and development. Ideally given three times per day at each meal time. Can improve plasma Phe control (3).
Prescribed protein substitutes	A key part of the PKU diet. Provides Phe-free protein replacement. Also contains other essential amino acids, fatty acids, vitamins, minerals and calories.	

For full details of excluded and permitted foods, see the many useful dietary information publications available at: www.nspku.org including NSPKU's Dietary Information for the treatment of Phenylketonuria (2013-2014) <accessed 31/03/14>

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Hannah tells me that she has 'cheat days' where she won't count her exchanges and enjoys more of these foods than she should.

run of the mill, which can make life tricky for these young people. Spending time to equip teenagers with the skills and knowledge that they need to cope with their diet, sometimes helps to make things that bit easier. There are some excellent sources of online information for teenagers which are useful for encouraging independence and confidence with their diet.

Case studies

LEWIS'S STORY

Lewis is 15 years old. He is an active boy, playing football on a weekly basis for a local under 16s team. He is also keen on martial arts, taking several sessions per week to build up his belts. There are no concerns regarding his academic performance at school. His low protein diet has always been well supported by his mum and his Phe levels maintained well within the normal range.

He takes 14 exchanges per day and 50g protein equivalent from his ready-made protein substitute drinks. However, Lewis has had a few raised Phe levels recently and we discussed these at his latest outpatient appointment. He explained that he had been 'playing around' with his diet and wanted to see what happened if he altered (increased) the amount of exchanges he was permitted. He admitted that he had eaten five to six more exchanges per day, "just to see what happened". We discussed how he felt whilst doing this. He explained that he didn't feel any different; however, Mum did not concur. She reported that Lewis seemed tired and 'moody'. She had noticed that he was less able to concentrate; he seemed 'vague' at times.

A further concern Mum had was that Lewis sometimes forgets to take is final dose of protein substitute. She prompts him to do so, but this can create friction, where Lewis feels his Mum is nagging him. We discussed how Lewis feels about having PKU and his diet. He tells me that it doesn't bother him most of the time; that he doesn't really think about it. However, it does annoy him when he has to explain to new people what it is and why he has to follow the diet. He also says that he genuinely forgets to take his protein substitute because he is busy with after school clubs/sports. We have discussed ways for him to remember to include his final dose. Lewis has agreed to take it before bed when he has completed his daily sports activities, instead of trying to fit it in at teatime. He has also agreed to stick to his recommended amount of exchanges in future.

HANNAH'S STORY

Hannah is 17 years old. She achieved above average grades in her GCSEs and is currently studying animal care at Sixth Form College. She hopes to become a veterinary assistant. She has been well supported by her mum and grandmother. They have both spent time with Hannah, encouraging her to develop her low protein cooking skills, ability to read labels and calculate exchanges.

She takes on average five to seven exchanges per day and requires 50g protein equivalent

from a gel type protein substitute, which she doesn't always take. Her Phe levels have been around 800-900μmol/L since she was about 14 years of age.

Hannah tells me that she has 'cheat days' where she won't count her exchanges and enjoys more of these foods than she should. She reports no adverse effects on her concentration or cognition, but still feels able to concentrate at college and feels well in herself.

During her recent dietetic review, she did express her concerns about PKU and pregnancy. She understands that a much tighter plasma Phe control is required pre- and during pregnancy. Although Hannah isn't planning to become pregnant, she does have a steady boyfriend and she is sexually active. We therefore discussed suitable contraceptive methods and she has since had a contraceptive implant. We have discussed the importance of taking her protein substitutes as prescribed too. Hannah has been reluctant to take them for many years, explaining that they make her feel nauseous. We have tried numerous products, none of which she has taken to.

During her annual review blood tests, Hannah was found to be iron deficient, but her bone profile was satisfactory. She has reluctantly been taking her iron supplementation.

USEFUL ONLINE RESOURCES FOR TEENAGERS WITH PKU.

NSPKU website and publications: www.nspku.org

- PKU - A Leaflet for Teenagers (2001)
- Pregnancy in women with Phenylketonuria (PKU) (2005)
- Advice about alcohol for people with Phenylketonuria (2006)

Plus a good range of dietary information – exchange foods lists, prescription items and recipes. Also available: booklets on prescriptions, travel, work/info for employers.

PKU Connect (Nutricia) www.pkuconnect.co.uk
The website covers many useful topics including: healthy eating, travelling, becoming more independent, alcohol. It also has a forum for members to discuss various PKU related issues. Also links with the Nutricia/SHS PKU friends Facebook page.

Drug company BioMarin - Kuvan: www.kuvan.com
(Kuvan is a medication available as treatment for some suitable PKU patients.) Offers a range of information on diet and PKU, including 'Low Phe for life', which includes useful information on recognising the signs of raised Phe levels. Cooking, eating-on-the-go, alcohol, travelling and pregnancy are all covered too.

References:
1 The National Society for Phenylketonuria (NSPKU) (2004). 'Management of PKU - A consensus document for the diagnosis and management of children, adolescents and adults with phenylketonuria', NSPKU, UK. Available at: www.nspku.org <accessed 31/03/14>
2 NSPKU (2005). 'Pregnancy in women with Phenylketonuria'. Available at: www.nspku.org <accessed 31/03/14>
3 MacDonald A (1999). The Dietary Management of Phenylketonuria, Doctor of Philosophy thesis, The University of Birmingham, Birmingham. Available at: www.etheses.bham.ac.uk <accessed 31/03/14>
4 Medical Research Council Working Party on Phenylketonuria (1993). Recommendations on the dietary management of phenylketonuria. Arch Dis Child, 68 426-27

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Questions relating to: *Living with PKU: the teenage years*
Type your answers below and then **print for your records** or print and complete answers by hand.

Q.1	What is Phenylketonuria (PKU)?
A	
Q.2	What are the risks if PKU is left untreated?
A	
Q.3	Describe the treatment for PKU.
A	
Q.4	Outline some of the challenges for dietitians treating PKU patients.
A	
Q.5	Explain the desirable plasma phenylalanine (Phe) levels from preconception to 10 years.
A	
Q.6	What is an 'exchange' and how do exchanges work in the PKU diet?
A	
Q.7	What food groups can be prescribed by GPs and what are their benefits for a PKU patient?
A	
Q.8	What are the implications of PKU during pregnancy?
A	
Q.9	From the case studies in the article, describe some of the issues that teenagers with PKU may have to deal with.
A	

Please type additional notes here . . .