

Welcome to  
**28th  
Annual Conference**  
of NSPKU

**Programme Information & Abstracts**

**Study Design**

The subjects were studied on a protein substitute (UPS) and a combination of UPS and AAT. The subjects took the UPS for 12 weeks and then took the combination for 12 weeks. They took at least 50% of their protein requirements from the amino acid tablets (AAT) from UPS. The order they did this in was randomised. Some took UPS first and others took a combination of UPS and AAT first. Blood phenylalanine measurements were taken every 2 weeks and all the blood amino acids were measured at the beginning, at 12 weeks and at the end of the study. All patients kept a diary of all protein substitute taken.

**Results**

Seventy per cent (14/20) preferred the AAT over the UPS, with compliance and acceptability (p < 0.05) significantly better with AAT. Ninety per cent of subjects took the AAT as preferred over the UPS. In particular, the AAT was preferred over the UPS in 14/20 subjects.

**2nd – 4th March 2001**

**Grand Hotel Margate**

**News  
Views**

**NSPKU**

The National Society for Phenylketonuria (United Kingdom) Ltd.

NSPKU, PO Box 26642, London N14 4ZF

## **PKU Gel: a new protein substitute**

A MacDonald, G Rylance, D Asplin, SK Hall,  
Birmingham Children's Hospital

Traditional protein substitutes are given as a drink in PKU. However, they are bitter tasting, are given in large volumes, and smell bad. In some centres it is now common practice to give protein substitutes as a paste. However, if this is done with current protein substitute powders, they tend to taste strong, are runny and sticky and produce inconsistent textures. A new protein substitute is now available called PKU gel. It contains starch and is designed to be given as a paste or gel. One 20 g sachet provides 8.2 g and it contains vitamins and minerals.

### **Study**

The efficacy and safety of PKU gel was studied in 9 children with PKU. They were all on a strict low phenylalanine diet. They were aged from 1 to 10 years. They took either XP Maxamaid or Phlexy 10 mix before they started the trial. The new PKU gel was taken for 8 weeks, and a variety of tests were taken at the beginning and end of the study to check quality of diet, nutritional biochemistry, blood phenylalanine control and growth of the children.

### **Results**

*Dosage of protein substitute:* The children took a smaller, daily quantity of PKU gel than their previous protein substitute. With the PKU gel they took a median dose of 110g daily (range 60g to 140g). The previous median dose of protein substitute (+/- vitamins and minerals) was 160 g daily. Seven children took the new protein substitute as a paste; 2 continued to take PKU gel as a drink.

*Quality of diet and nutritional biochemistry:* The intake of all nutrients exceeded recommended daily amounts. No abnormal blood vitamin or mineral levels were seen.

*Blood phenylalanine levels:* The new PKU gel did not affect blood phenylalanine levels. The mean plasma phenylalanine level was 311 mmol/l before starting the PKU gel and during the 8 weeks of the trial was 318 mmol/l.

*Growth:* This was satisfactory for all children.

*Acceptability:* Most of the children described the texture, taste, and volume of PKU gel as good. No child wanted to return to their previous protein substitute.

*Advantages described by parents:* included no weighing of powder, no need to add vitamins and minerals, good consistency, acceptable taste, easy and quick to prepare, smaller volume and not so sticky as other protein substitutes.

### **Conclusion**

PKU gel is a safe, popular and efficacious protein substitute in PKU.

## **OUTWARD BOUND WEEKENDS**

### **A stepping stone towards greater independence.**

Eleanor Weetch NSPKU Dietitian Advisor

A traditional method of teaching children about their diets is in a hospital or clinic setting on a one to one basis with the dietitian. Advice is frequently given to the mother about cooking, shopping, travel and organisation. The importance of the diet in order to realise the full potential of the child is emphasised and this can result in stress, anxiety and reluctance on behalf of the parents to allow normal independence. Self esteem can be low and the diet itself can be blamed for the restraints many feel in their teenage and young adult lives.

The NSPKU started to run outward bound weekends two years' ago, to help give pre-adolescent children confidence in themselves and their abilities and a chance to manage their diet on their own, but with help if required. Dietitians and a metabolic specialist doctor volunteer to help and this is reassurance for parents to gain their confidence too.

The weekend runs from Friday afternoon to Sunday lunchtime in the summer and there have been two weekends in Derbyshire, one in the Lake District and a day's outward bound in the southwest. The children go mountaineering, abseiling, canoeing and raft building as well as undertaking other activities such as archery, assault course, problem solving and murder mystery crimes. The diet is all provided by the centre, except the protein substitute and vitamins and minerals which the children are expected to make up themselves. They also take care of their exchanges under a watchful eye or two!

The majority really enjoyed the weekend and many of the children went on to do things they would not have had the courage to do before because of lack of confidence. They learnt to work together, help one another and have fun in the beautiful Derbyshire countryside. All went home tired but much more re-assured of their own abilities.

## **Treating Previously Untreated Adults with Phenylketonuria**

Dr Philip Lee

Charles Dent Metabolic Unit, National Hospital  
for Neurology & Neurosurgery London.

Before universal screening for phenylketonuria (PKU) was introduced in the United Kingdom in 1969/70, children were only diagnosed because developmental delay, a mousey smell, and/or eczema alerted doctors to the possibility of PKU. It is estimated that ~ 2000 such individuals were born between 1930 and 1970. Many went undiagnosed, or even if diagnosed, it was often felt that treatment would not improve the brain damage that had occurred. Adults with late diagnosed, never-treated PKU may have problems with learning capacity, aggressive, self-injurious behaviour, epileptic fits and eczema. They are very dependent on others for their care and require a number of medications to control their challenging behaviour. The overall cost of their care is very high

In recent years, a number of late diagnosed PKU adults have been placed on phenylalanine-restricted diets in the hope that this will improve the quality of their lives. It has been calculated that the cost of caring for these individuals could be reduced by about £20,000 per year per patient. The diet has generally been started due to the well-motivated wishes of their carers and/or parents. The results of these attempts have been reported to be variably successful. A review of dietary treatment in 35 late diagnosed PKU patients showed that 54% had much improved, 23% had moderate improvement, 14% changed little and 9% deteriorated. In all of these cases the people providing the diets and observing the responses were aware of what was being done. The potential for biased reporting and the so-called 'placebo' response exists.

To see whether lowering blood phenylalanine in people with late diagnosed PKU improves their behavioural disturbances and quality of life, a randomised, placebo-controlled trial is proposed. This talk will discuss the rationale and current plans for this trial and its relevance for all adults with PKU.

## **PKU – The Insiders Story**

by Amanda Dart

An opportunity to listen to an adult PKU talk about what it **really** is like to live with PKU!

The trials and tribulations of growing up with PKU in a normal family, coping with everyday things and dealing with the not so ordinary situations life throws at us all.

The dilemmas faced on going out with friends, coping with diet at college and whilst working as a member of the teaching profession.

## **PREGNANCY AND PHENYLKETONURIA**

### **My personal experience**

Maria Whitehouse

Having discussed the issues with Dr Lee, our consultant, I started a very strict low protein diet on April 12th 1999 with a view to trying for a baby. I had previously stopped diet aged 16 and had been on normal diet ever since.

I was admitted to the Middlesex Hospital for one week where I was taught the diet and found it was very different from my childhood experience.

I was started on eight exchanges and my levels dropped from 1400µmols/l to 700µmols/l in only 5 days. It was explained to me that the optimum level for conception is between 100 – 250µmols/l.

The fun really started when I got home and started doing the diet for myself. It was a feat of organisation and both my husband and Maggie, my dietitian, were amazing sources of support. My levels were sometimes erratic so the amount of exchanges allowed ranged from about four to eight in the initial stages.

After one month we had permission to try for a baby and on 17th June, 1999 we found out we were pregnant! My levels were a bit high (400) so my exchanges were reduced to 2 immediately and my levels plummeted to within the optimum range. I often felt nauseous and had difficulty keeping the weight on and life was pretty emotional at this stage. At 10 weeks, however, I had a scan and everything was normal.

Throughout the pregnancy I was gradually allowed more exchanges. By the end I was on 30 exchanges.

Jacob, a perfectly normal little boy, weighing 7lb 7ozs was born on February 25th 2000 and is non-PKU.

## **LIVING WITH PKU**

John McKenzie and Mum

When John was born it was such a wonderful thing to happen to me, here we are with a lovely baby and wondering what will be in store for us as a family. However what happened was not what I expected, John being diagnosis with Phenylketonuria or PKU. Pkwaht I had never heard of this nor could I pronounce it.

This was the most devastating news I could have heard, my beautiful baby boy had brain damage or could have brain damage. Here I was frightened angry and wondering what I did in my pregnancy to give him this. I just could not think how I would ever cope with this, the diet keeping his blood levels to the Doctors requirement.

I felt so helpless and lonely as I knew no one with PKU. I went through all the stages of shock, fear denial, why me to acceptance. When I came to accepting that John has PKU I decided that being PKU was not going to stop John living life to the full.

Which I am glad to say he has, as he did not miss out on anything such as going to nursery club, scouts and going to camp. He also went on school outings, had school dinners and thought, as did his peers that they were horrid. We also had lots of wonderful holidays both in this country and abroad.

I feel that John having PKU has not stopped have a life with fun and adventure.



We are grateful to  
UCB for their help in producing this booklet



The National Society for Phenylketonuria (United Kingdom) Ltd.

NSPKU, PO Box 26642, London N14 4ZF

Helpline 0845 6039136

Charity No. 273670

Email: [nspku@ukonline.co.uk](mailto:nspku@ukonline.co.uk)

Company No. 1256124

© Copyright NSPKU

<http://web.ukonline.co.uk/nspku>

February 2001/300