

Welcome to
**33rd
Annual Conference**
2006

Programme Information & Abstracts

The Hinckley Island Hotel

✚ N S P K U ✚

The National Society for Phenylketonuria (United Kingdom) Ltd.

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It's all about you.

Chris Hendriksz, Consultant in Clinical Inherited Metabolic Disorders,
Birmingham Children's Hospital.

The brain is the most important organ affected by PKU but does exercise have a protective effect on the brain? Exercise plays an important role in general health improvement but where do you start, how do you get motivated and how will it influence my levels? What is regarded as exercise and are there any fun ways of doing exercise?

How do I start and is it really a case of "No pain no gain"

These are common questions asked especially by teenagers and I will try to address these and also explore some potential research ideas relating to exercise in PKU.

Unless you make the decision that your health is within your own hands and look at ways to improve all aspects of health you may limit your quality of life.

Be selfish and take control because "it is all about you"!

You, me and exercise needn't seem like a marathon!

Joanna Eardley
Metabolic Dietitian, Evelina Children's Hospital, London

The media remind us daily that childhood obesity is on the increase, is this also happening to children with PKU?

We are reminded constantly that we are not exercising enough and how we could all improve our diets. But how do we go about doing this?

This presentation will set the scene for the talks over the weekend addressing these topics and also give you an insight into my story of the blisters but loosing weight, the freezing cold morning runs but the feeling of elation, the effort but the marathon medals!

Research in PKU: A compilation of studies

MACDONALD A,
Birmingham Children's Hospital, Steelhouse Lane,
Birmingham, UK. B4 6NH

HOME DELIVERY SERVICE FOR ESSENTIAL DIETARY PRODUCTS

The traditional system for acquiring special dietary products (patient prompted prescriptions generated by a GP and dispensed by a chemist) is problematic. A home delivery service for distributing dietary products was investigated. A prospective, controlled, home delivery trial for essential dietary products was conducted in 62 patients with inherited metabolic disorders (50 with PKU) for 12 months. 30 subjects used a monthly home delivery service (Homeward: Nutricia) to receive clinical foods, 32 remained on the traditional system. Each month, the home delivery service checked home stock levels, acquired GP prescriptions, and home delivered products. Products used in standard amounts were included in the home-delivery service i.e. protein substitutes, vitamin/mineral and energy supplements. An independent researcher completed monthly telephone interviews with patients/parents about any prescription issues. It was found that the home delivery service was a more reliable system for dispensing essential clinical feeds than the traditional system and was associated with less prescription errors. Incorrect protein substitute was dispensed once by the home delivery service compared with 12 incidents in the control group; and incorrect flavours of protein substitute were dispensed to the home delivery group once compared with 15 errors via the chemist. The home delivery service delayed delivery of protein substitute in 1 subject on 3 occasions compared with 30 occasions in 15 subjects via the chemist. 11 control subjects had 4 or more prescription problems in one year compared with only one in the home delivery group. All the study group patients/parents said the home delivery service was reliable, convenient and deliveries were usually complete. In conclusion, the long term use of a home delivery service for essential dietary products was safer, effective and more reliable than conventional systems.

DOES READY TO DRINK PROTEIN SUBSTITUTE IMPROVE COMPLIANCE IN PKU?

In PKU, compliance with protein substitute (PS) is an issue in older people. The efficacy of a new ready to drink protein substitute (with vitamins and minerals) was investigated in a 6 week, 3 part, open, randomised controlled study. 25 subjects with PKU with a median age of 30 y were recruited. In parts 1 and 2, subjects were randomised to either the liquid or powder. In part 3, subjects chose either the liquid or powder or a combination of both. All but one subject chose the liquid protein substitute in part 3 as either their sole (68%, n=17) or partial source (28 %, n=7) of protein substitute. The liquid protein substitute helped improve quality of life, reduced self consciousness when taking protein substitute and was convenient. It was also easier to take outside the home and it reduced wastage. In conclusion, the liquid protein substitute was popular with teenagers and adults with PKU. It reduced self-consciousness, improved life quality and compliance.

COOKING 4 FUN

Encouraging an early interest in cooking is one way to help children understand and develop a positive attitude towards their low protein diet. A teaching aid to motivate children to cook with their special products was developed. A group of 19 children all chose at least one favourite low protein dish to prepare and cook using their own instructions, mainly working in their own kitchens. A 2 part DVD (total length 36 minutes), divided into two age groups (children under 10 y; and children 10 y and over) was produced demonstrating 26 low protein recipes. An accompanying recipe book was also produced. This is a practical, fun, teaching aid which should help children develop their low protein cooking skills which has led to many children exploring with low protein cooking.

*These projects were conducted with the help of many others including:
Anne Daly, Nilusha Manj, and Maggie Lilburn.*

Adults with Previously Untreated PKU - Trial Update

Lesley Robertson, Research Dietitian, Charles Dent Metabolic Unit,
National Hospital for Neurology & Neurosurgery, London.

The aim of the trial is to assess the impact of a low phenylalanine diet on adults with previously untreated PKU. Since starting work on the project in July 2003 we have reached our target of recruiting 36 adults. The study is a double blind crossover design, where each of the recruits have a trial of a PKU and a normal diet. This means we will not have any final results until the last recruit has completed the trial in early 2007. From then we will then analyse the data and hopefully be ready to present and publish the full results later that year.

Prior to the full study, a postal survey was undertaken and distributed to relevant healthcare professionals throughout the UK. The initial case-finding questionnaire was followed by a more detailed survey about medical and social care. The first questionnaire established the whereabouts of adults with untreated PKU, the second covered a brief medical history, including current medication, and diet history. The social aspects of care were also noted including mobility, communication skills, dependence on carers and the presence or absence of challenging behaviour.

To date, only 157 adults have been identified and completed or partially completed the questionnaires. The majority were born before newborn screening (1969) and are living in community care homes for people with learning disability.

As the birth incidence predicts 70 new cases per year, we found significantly fewer than expected. Those identified had significant psychosocial and medical problems. Following these questionnaires we started the trial assessing the efficacy of a low phenylalanine diet.

Healthy Eating – Am I bovered ?

Barbara Cochrane, Clinical Specialist Dietitian
Royal Hospital for Sick Children, Yorkhill, Glasgow

Healthy eating and the trend towards diet and lifestyle changes have been a major topic in the news, in magazines and on the television. It has been recognized that there is a worrying trend towards obesity in the general population and the associated risks that accompany obesity are also on the increase.

Diabetes in particular is on the increase and we now see 1-2 children a year developing Type 2 Diabetes when as little as 5 years ago there were no children seen with this condition. Diabetes is associated with heart disease, strokes, kidney failure and blindness. These conditions are all going to be increasing in the general population if we do not take the problem of obesity seriously.

Many dietitians, parents, partners and associated professionals look upon diet and PKU with sympathy. General lifestyle, fitness and overall diet are often looked at with tunnel vision. We tend to look at Phenylalanine levels, how the family is coping in general and are reluctant to tackle problems with weight and exercise. It is a mindset we are going to have to change, if we are not going to see the general trends to ill health reflected in the PKU clinic.

Families also need to change their attitude and not be upset when weight becomes a topic of conversation. The attitude of “the diet is very restricted and I am not going to stop the treats” will have to be changed to one which looks at overall healthy eating and accepts that the child or adult with PKU is as likely to develop conditions the same as everyone else and we all have a duty to try and prevent that – or accept the consequences.

PKU management in adolescents and adults

Andrew Morris

Consultant Paediatrician in Metabolic Medicine Royal Manchester
Children's Hospital

Strict control of blood phenylalanine concentrations is essential during infancy and childhood, to prevent learning difficulties. The evidence is less clear for adolescents or adults with PKU.

There is a strong suspicion that adolescents do less well in their exams if their levels are high and we recommend that they should stay on diet at least until they leave school. It may, however, be necessary to discontinue dietary treatment if conflict over the diet is leading to family breakdown or if poor compliance leads to a risk of serious nutritional problems.

In adults, it is still uncertain whether dietary treatment is needed. Some people report poor concentration or irritability when their blood phenylalanine concentrations are high. People with high phenylalanine levels have abnormalities on brain scans but these do not seem to be associated with clinical problems.

Strict dietary treatment is essential during pregnancy in women with PKU. Without this, almost all babies have serious learning difficulties and there is a high risk of malformations (especially affecting the heart).

Everyone with PKU should have specialist follow-up, even if they have discontinued dietary treatment. The aims are

- to support people who remain on diet.

- to avoid nutritional problems in people 'off diet'.

- Many eat little meat or dairy products and they may become vitamin B12 or calcium deficient.

- to help women to start treatment prior to pregnancy.

- to check that PKU has no unrecognised long-term complications.

The PKU Registers

Alison Munro.

Research Nurse. The UK Newborn screening Programme Center.
C/o Executive Offices. Great Ormond Street Hospital. London

The UK Phenylketonuria (PKU) register was set up in 1964 with Medical Research Council funding in order to monitor the long-term health and development outcomes of children who had an early diagnosis of PKU through newborn screening. The register is unique in the world because of its national coverage and long history of data collection

Throughout the years, the register has been used to address many important questions, including:

- 1 The relationship between strict, early treatment and childhood development
- 2 Educational progress in children with PKU
- 3 The increased risk of birth defects in children born to women with PKU who are not on treatment.

The findings have led to improved treatment and follow-up for both children and adults with PKU. All individuals born in the UK between 1964 and 1998 and who received a diagnosis of PKU following newborn screening have been placed on the register. Some individuals born before 1964 who were diagnosed later in life (for example, when they had children) have also been reported to the register.

Later data was reported to the register of virtually all pregnancies and births of women with PKU in the United Kingdom between 1978 and 1997. The physical and intellectual development of the off spring of these women was also recorded.

Data collection for the register carried on continuously until 1994, after which issues around funding caused some disruption. Funding stopped completely in 1998, and as a result, data collection ceased.

In 2002 the UK Newborn Screening Programme Centre (UKNSPC) was set up to develop standards and monitor the quality of newborn blood spot screening in the UK. The UKNSPC now looks after the PKU register. It aims to make sure that the register can be used for research once more for the benefit of individuals with PKU and their families.

The Future

The UKNSPC proposes to contact all individuals on the register to secure their consent for retention and use of existing data for research. Ethical approval is being sought to contact individuals whose data are included in the register. Initial contact will be through clinics, with tracing through the NHS Central Register and subsequent contact through general practitioners for those no longer attending clinics. There are approximately 2700 individuals on the register, most of whom are born between 1964 and 1998. Families identified as a consequence of the current national audit of children born between 1994 and 2005 and who have a positive newborn screening test for PKU will also be contacted. This audit is being carried out to assess the quality of the PKU newborn screening programme.

Updates on this project and the results of any research study based on the PKU register will be made available and published. No individual whose data has been used for research will ever be identified in any reports or publications. All data presented will be completely anonymous.

We are grateful to all the following companies who have supported us during the last year.

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