The National Society for Phenylketonuria (UK) Ltd

News & Views

Website: www.nspku.org

Issue 104

EUROPEAN CONFERENCE REPORT

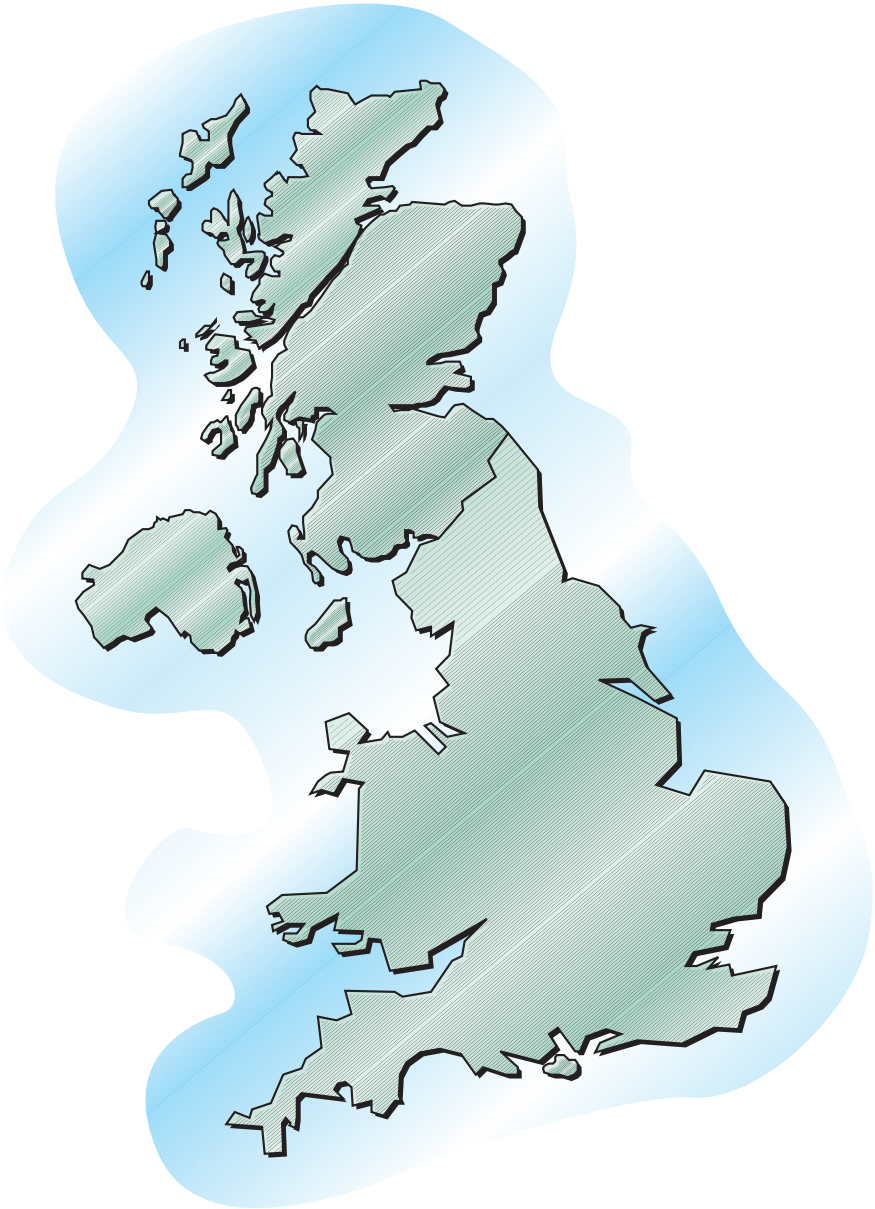
2002

Inside: Dietitian's Report • Maternal PKU • Letters • And much more.....

In Touch

The Council of Management

Telephone Helpline: 0845 603 9136
Address: PO Box 26642, London N14 4ZF
E-Mail: info@nspku.org
Internet: www.nspku.org



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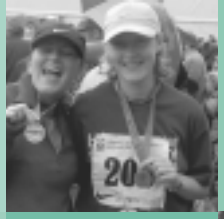
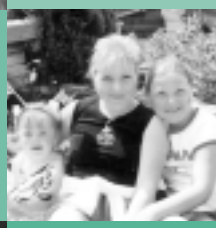

Editors Report...

AH! AH? Oh? Hmmmmmm. Dammit, heeeellllpppp. (English Oxford Dictionary) – ***"to be out of any particular specialist foodstuff just when you need it most and everywhere is more shut than an Afghanistan border"***

You been there? I have.

Christmas was just like this for the Bramley family. Just one week before the great day I found myself peering up into numerous kitchen cupboards with an 8 year-old PKU daughter perched firmly on my shoulder. She was chanting the mantra "I don't like your PKU roasts Dad, I don't like your PKU roasts Dad, pieces of hate pieces of hate etc etc".

To be honest I didn't panic much because I was having roast goose, but there was an inkling or a mild twinge of guilt and a massive punch to the kidneys from Nicola as she made her demands quite clear……"I WANT LASAGNE! And if you don't deliver, you are protein-free TOAST! YOU DIVOT! – (She can be quite scary at full-tilt!)

So there I am. Crumbling in front of the bare cupboards. "What the flip am I gonna do?". The PKU Mafia are on my back and I'm faced with New Year spent at the bottom of a local river in concrete slippers.

DING!!!!!!!!!!!!! IDEA!!!!!!!

I can be quite naughty when it comes to asking favours and won't be backwards at it. But I knew that I wouldn't be disappointed. I phoned Steve Fletcher of Firstplay Dietary Foods and begged on all of the available knees that I had, Pleeeeaaaasseee have you got any PASTA SHEETS PleeeeAAAAASEEEE!! She's GONNA Kill mEEEeee hellp meeee etc etc.

Steve, in his usual style lifted one eyebrow (which is really hard to do over the phone) and stated, "worry not you southern softy, for I shall forward you some of said sheets and you shall have lasagne. Trouble me no more. Southern Softy"

Trembling, I lowered the phone and began the wait.

The package arrived some 4 days later (Steve had obviously read the recipe for "Toasted PKU Dad", which begins….."Leave to sweat for 2 days before posting…")

Not only had he started his production line up to produce some lasagne sheets for Nicola but he had taken the trouble to produce the sheets and press out Christmas trees and Angel shapes for her, for her festive meal at his expense of time and care. He justified the act by nominating the newly pressed sheets as "collectors items."

The moral of the story is this……. The diet takes some doing, the administering of the diet is even harder for the younger ones and even harder for the older ones and even harder on us all. If we don't help each other, it gets even harder. Just one bit of help can make it much easier.

Thanks for the lesson Steve.

Pete Bramley
Editor

NSPKU
The National Society for Phenylketonuria (United Kingdom) Limited

Please address all correspondence to the Editor, *"News & Views"*,
48 Hazeldell, Watton-at-Stone
Hertford SG14 3SN
Tel (01920) 427636
Email: nspku.secretary@ukonline.co.uk

While every care is taken in the preparation of *"News & Views"*, the society cannot be held responsible for any information given or views expressed.

The National Society for Phenylketonuria (United Kingdom) Limited,
PO Box 26642,
London N14 4ZF
Company No. 1256124.
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Email: nspku@ukonline.co.uk
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Treasurer's Report...

AS I write this we are fast approaching Christmas but as you read this it will be a fading memory, so I hope you had a good one and a happy New Year to you all. This will be a busy one for the NSPKU and inevitably for me with the AGM/Study day in March and the ESPKU conference in October.

As we head towards the spring and hopefully some better weather some of you will be beginning your annual round of sponsored runs, swims, hikes, jumps etc etc. Don't forget that I have sponsorship forms and can provide stickers, collecting tubs etc. I am in the process of updating the sponsor forms so some requests from you might "encourage" me to get on with this!!! Once you have completed your event and raised loads of dosh it might be worth checking if your company has a matched giving scheme. A number of employers will match amounts raised for charity either informally or more formally through the matched giving scheme. Contact me for more details.

I have also received news of a new website giving you the opportunity to donate on-line via givenow.org where you can donate to the NSPKU by credit or debit card. You will need to enter the full Charity name (National Society for Phenylketonuria [United Kingdom] Ltd to find us but after that it's quite easy. I will soon be providing a mission statement and some news on our activities for this site. Don't forget to let me know of any other fundraising opportunities you hear about.

Julia Bailey
Treasurer

Donations

to 31st December 2002

(£30.00 and over)

Mr R Masters	100.00
Mrs E Manning	170.00
HSBC Lewisham	47.00
David & Audrey (DP's Financial Services)	142.90
Tracey Wysocki c/o Halley	368.50
Mandy Tonkinson	50.00
Pam Manwaring (via Wendy Cheale)	360.00
Anonymous	100.00
Thorntree Primary School	2,686.35
I & Mrs L Thomas	50.00

Gift Aided

Mr & Mrs Dart	50.00
Barbara Broadbent	200.00

In memory of:

Matthew Bacon	170.00

Chair's Report...

I AM absolutely thrilled to report that sponsorship has been obtained to start the research project to look into the possible benefits of dietary treatment for those previously untreated who have PKU. Well done and thank you to Eleanor and all of those in the dedicated team set up to run the project. Months of hard work have gone into winning this award. Read more about it in Eleanor's report.

The part that the Society is playing in this project is to freely provide Eleanor's services, to make a donation and we will be working closely with the team throughout the research.

AGM 2003

It is our intention that this edition of News & Views reaches you before March 15th, the date set for our AGM. This year we are having a one day event with speakers, small discussion groups and the AGM (12 midday). The day's focus will be teenagers and adults living with PKU but, as usual, will provide information and support for families with newly-diagnosed children. The day, although different in format will provide an opportunity for a get-together and exchange of ideas with others in the same situation as you.

I hope the event will be well attended but don't forget we hope to see a large turnout in October for the exciting ES.PKU Conference!

QUESTIONNAIRES

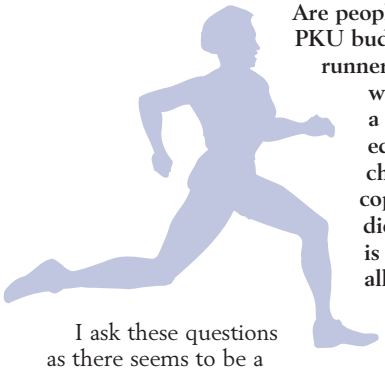
Thank you to all those families who have taken the time to respond to the questionnaire enclosed with the last edition. We are encouraged by the number, but know there are still lots of you out there who have not got round to filling it in.

If, like me, you have mislaid your copy under bunches of holly and piles of Christmas cards then please get in touch with John Skidmore who will gladly supply another. We have already learned a great deal from you and I am confident that your experiences and comments will help the COM improve the support that the society provides.

To help you, we have included a FREE NSPKU pen!! Thanks to John for instigating the questionnaire and working so hard collating all of your replies.

Sara Bartlett
Chair

The London Marathon



Are people connected with PKU budding Marathon runners? Is it because it would appear to be a Marathon in educating your child to accept and cope with the PKU dietary regimen? Or is it because you are all masochists?

I ask these questions as there seems to be a healthy (some would say unhealthy) interest in running in the London Marathon – of course raising hundreds of pounds for the NSPKU in the process.

Many of you who have registered/tried to register for the London Marathon have been told about the Golden Bond Scheme for charities.

This Scheme guarantees the charity in question a number of places for five years. Unfortunately there is a waiting list of 3 to 4 years! The NSPKU is on this waiting list.

So until the NSPKU obtains a place on the Golden Bond Scheme, please register for future London Marathons' in your own name.

A thought !!!!

Is anyone a member of an athletic club who would like to arrange a sponsored Marathon / Half Marathon where the proceeds go to the NSPKU? Would anyone be interested in organising this? How many of you masochists… sorry … members would be interested in running in such an event?

Please reply in writing to our illustrious Editor who has already offered his services as a Starter.

Personally I'm into those sports at the cutting edge such as the World Extreme Ironing Championships ; zorbing ; sail-boarding etc etc … honest!!

Now there ***are*** some ideas for raising money. Any takers??

!!Members' Questionnaire!!

A great response so far to the members questionnaire, ensure you complete your questionnaire if you haven't done so already.

We need to hear from you!

Spring Draw 2003

Firstly a big thank you to the girls at Action Press who filled the last issue of N & V with the Raffle Tickets. It's a soul destroying job.

At the time of writing this article, I have banked over £600 of ticket sales into the Spring Draw Account which is excellent.

Keep up the good work and let's see if we can raise £10,000 this year.

There are plenty of tickets left, so please ask for more if you need any, available from Mr Eric Lange, 27 Western Road, Sutton, Surrey, SM1 2TE.

Remember, the prize for selling the most Raffle Tickets is a free place at the European Conference at Hinckley in October 2003.

KEEP ON SELLING!!

Letters

News & Views, 48 Hazeldell, Watton-at-Stone, Hertford SG14 3SN
Email: nspku.secretary@ukonline.co.uk

The treatment of phenylketonuria varies for each individual patient. No patient should alter their own treatment as a result of reading how another patient manages their diet without first consulting their doctor or dietitian.

Tracy and Claire with their medals

Dear News & Views
Please find enclosed cheque for the NSPKU. Also enclosed is a picture of myself (on the left) and my cousin (Claire Gow) with our medals for completing the Britannic Asset Management Women's 10K Run on Sunday 19 May 2002.

I completed the run in 1:07:22 and raised £368.50 for NSPKU.

Regards
Tracy Wysocki

Dear News & Views

It doesn't all end up in the bin!

The Chartered Institutes of Wastes Management, North East Centre, held a charity raffle in support of the NSPKU at their Christmas Luncheon in December at the Abbey Park Hotel, York. PKU was the chosen charity in support of John Skidmore, the North East's Centre's Honorary Secretary whose youngest daughter Annabelle, aged 3, has PKU.

The Waste Industry (vehicle and equipment manufacturers, academics, public and private sector waste professionals) attended the event from across the North East of England. The raffle raised a tremendous £263.50 for the NSPKU.

John Skidmore

Dr R Davies with Helen Winter who ran the half marathon. Helen is holding Elliot Charlton who has PKU

Dear News & Views
Please find enclosed a cheque for £400.

My son Elliot has PKU and is 17 months old. I got to know Helen Winter, who ran the half marathon and raised this money when I started ordering food for Elliot from my GP where she works as a Pharmacist.

I would like to thank her and all my friends and family who worked so hard to raise the money.

Please find enclosed a photograph of Helen Winter holding my son Elliot. Also in the photograph is Elliot's GP, Dr Robin Davies who along with the rest of the staff at the Northgate Village Surgery, raised £140 towards the £400.

Sincerely
Jacqueline Charlton

Contacts

Hi,
My name is Vida and I am a 21 year old classic PKU. I am currently studying at Queen Margaret University College in Edinburgh for my degree in Dietetics. I will shortly be coming up to the time to prepare my final dissertation and I am starting my preparation now. I am keen to hear from any fellow PKU's who would like to share their experiences with me, so I can gain a well rounded view on 'life with PKU'. I would be very grateful if this could be mentioned in your next 'News & Views' and my e-mail address supplied to any potential respondents.

Many thanks,
Vida Rahmani
diva_la_babe@hotmail.com

Hi
My name is Patrick Taylor. I'm 24 years old with PKU. I would like to make contact with other people preferably from Southampton which is where I live or near by (South of England).

However further away would be satisfactory. Please let me know soon regarding PKU contacts by email.

from
Patrick Taylor
patsstay@hotmail.com

Dietitian's Note:
You might like to contact Jonathan Beaumat or Angela Murphy who are setting up an adult PKU Support Group.

Jonathan: 0781-3438045
Angela: angela.murphy@ucl.ac.uk

They will also be telling us about the support group at the AGM in March. Why not come along?

If you wish to make contact with someone else to share your PKU experiences, write to: News & Views Editor, 48 Hazeldell, Watton-on-Stone, Hertford SG14 3SN.
Or Email: nspku.secretary@ukonline.co.uk

NEW

they're a bit saucy!

TWO GREAT TASTING SAUCY FLAVOURS FOR YOU TO ENJOY

Promin Low Protein Pasta in Sauce Is a brand new fast food product available in two tasty flavours.

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Simply mix with water in a saucepan bring to the boil and then simmer for 10 minutes.

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Promin pasta in sauce will soon become a necessary addition to your kitchen cupboard. Available direct from Firstplay, see the contact details below.

Full analysis available on request.

PROMIN

Firstplay Dietary Foods was formed in 1993 to produce Promin Low Protein products from a small factory in Stockport, where we only produce low protein products.

Promin Pasta and Promin Tri-Colour Pasta are now a well established brand and are available on prescription. Our pasta range also includes Imitation Rice, Pasta meal (available by prescription) and Lasagna Sheets (available by mail order). More recently interaction with PKU patients, parents and Dietitians has resulted in the expansion of the Promin range.

New products have been designed to reflect the needs of a maturing PKU population leading busy hectic lifestyles. Call us for more information or with your dietary suggestions.

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NOW AVAILABLE ON PRESCRIPTION TOGETHER WITH LASAGNE SHEETS & COUS COUS

Please consult your dietitian about how to count these meals in your diet as they contain phenylalanine.

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To order telephone or fax: **0161 474 7576**, E-MAIL: firstplaydf@smartone.co.uk WEB: **www.promin-pku.com**

Maternal PKU

Michayla shares her experience of surviving the PKU diet before and during the pregnancy of her three children...

Michayla with her two girls, Danielle Jayde 10yrs, Chantelle Jordana 2yrs. March 2002

WHEN I was born I was diagnosed with a rare blood disorder know as phenylketonuria (PKU), found when I had my heel prick on the fifth day that all babies have. Due to this finding, I needed to go on a low protein diet and have special low phenylalanine baby milk.

As I got older I could not eat like other children, e.g. no milk, eggs, cheese, fish, meat and chocolate. As a substitute for protein, I had this special drink which wasn't very nice. Also, every month I had a finger prick to keep checks on my blood levels. My brother was diagnosed with this two years later. It was quite hard for my Mum telling us 'we can't have this and can't have that' like all other children. We both didn't understand why.

We both came off the diet when we were 11 years old, and were able to eat normally - no more nasty drink. It was great! We both went out with our Mum for a meal and ate everything we weren't usually allowed.

As the years went on we are both fine but when it came to having a baby myself, I faced the diet again. My brother is fine to go in for a baby with no problems, but for me, it's back on the diet as I carry the baby. I needed to go on the diet before I conceived as my blood level has to be levelled off between 60 and 250umol. I also had to do my blood tests 2/3 times each week, which I didn't like but I knew I had to follow my dietitian's advice.

So, it was back to fruit and vegetables and the drink again. I also had to take glucose powder to make sure I had enough calories each day. It was so hard, but I levelled my blood off quite quickly but it took ages to fall pregnant. As I started to get fed up, I struggled to keep to the diet and finally fell pregnant. I had an 8lb baby girl called Danielle Jayde who is now 10 years old. It was all worth while as she has not got PKU!!

Two years ago I gave the diet another go. Again it was hard going, missing foods that I enjoyed but if this meant I would have a healthy baby, then I would stick it out. I fell pregnant quite quickly this time which made me feel much better and I gave birth to another baby girl 5 weeks early - Chantelle Jordana. I was overwhelmed and again it was worth everything I had to go through - no PKU!!

Finally, I have done it all over again and just had a baby boy 3 weeks early - Jayden Jaymes weighed 7½lbs. Again a healthy baby - no PKU!!

This time I found the diet really easy. My dietitian cannot believe how well I've done to keep to the diet. I am the only one in my area that has gone onto the diet to have children and she says I am a very good example to anyone with PKU who would like to have any children. It is hard, as when you are out and feel hungry, you can just pop into a shop and buy a quick sandwich or sausage roll. On the diet you can't do this. There are plenty of protein free foods, e.g. bread, biscuits, pasta and chocolate but I didn't use any of them. My dietitian couldn't believe I didn't want any of them.

I would like you to print this letter for any other PKU sufferers who might be thinking of having a baby. Just to let them know, yes it is hard to stick to the diet and follow the Dietitians and Doctor's advice, but the end results are brilliant!!!

Michayla Rushby
Age 34

Dietitian's Note: The current advice using present knowledge is that it is recommended to stay on the diet for life if possible. Strict dietary control is essential pre-conception and throughout pregnancy. The chances of a PKU woman having a PKU child is 1:100 so it is relatively low risk. The low protein foods are an important part of the diet to provide energy. Individuals should follow the advice of their own dietitian.

Jayden Jaymes at 8wks

NEW

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at lasta...

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A vacation with PKU!

Travelling with PKU - a parent's experience...

IN JULY 1999 my wife and I were blessed with the birth of our youngest child.

A wonderful daughter Susan who continues to enrich us on a daily basis.

Three weeks later we (as I am sure many others have felt) thought our world had fallen apart when we heard the name Phenylketonuria for the very first time.

Well I am pleased to say that with time and a great deal of help from the many people involved with Susan's care we have slowly learned that our world nor in fact Susan's standard of living had ended. And with a few adjustments we have managed to maintain a fairly normal existence living with P.K.U.

Earlier this year our family decided that we deserved our first ever, foreign vacation and so after the usual decisions as to location and type of holiday required we decided that a trip to the beautiful island of Menorca was just what the doctor ordered.

It was at this point that the P.K.U. decided to rear its head again and my wife and I reverted to a full blown panic attack. How would we feed Susan and still keep on top of her blood levels. What kinds of food would she eat in a foreign country? And how would we get the local people to understand her special needs.

What we decided in the end was that we could trust only ourselves with such important decisions. So like the complete amateurs that we were we ended up packing a complete suitcase of provisions just for Susan. Even down to her own set of scales to weigh all that "funny" foreign food. Apart from the basics such as her Maxamaid drink and a tin of Polycal In case of any illness. We also somehow managed to take with us biscuits, bread and PKU drinks (Duocal). We thought that if the worst happened we would feed her these things with a few fruits and veg cooked and prepared by ourselves.

My wife and I were so afraid wondering had we made the right decision to go abroad where we would be all alone and so far from the reassurance of our fantastic dietitian and doctors who help us constantly and somehow manage to keep us from going insane.

I could not help but wonder how many other families with a P.K.U. child have the same sort of fears. And may have even chosen not to go abroad but rather to stay in the relative safety of England's shores because of this very real fear we have that only at home in our own little spaces can we keep our precious children safe.

Well I have taken it upon myself to write this story to reassure all of those worried mums and dads out there that those fears are groundless. And in no way represent or do justice to the many wonderful people we met on that beautiful island.

In our fortnight on Menorca we were very surprised to find that the local people could not have done more or tried harder to help us. On one occasion in particular we went into a local hotel for a Sunday dinner simply because they served an English meal and we felt that our children may benefit from a meal they have regularly at home.

On entering the restaurant I approached a waiter to ask for his help in catering to Susan's meal. He promptly asked me to wait a moment and ran off into the kitchen with great speed. He returned a moment later with the head chef in tow and carrying a pen and pad. The chef then prompted me to tell him what Susan could eat how much of each item she could have and exactly how we required it cooked and presented.

It should also be noted that for this service we were charged no more money than the normal rate for a Child's meal which was in the region of £1.50. It would be easy to say that this was the exception to the rule over there but it was no more impressive than the way we were treated all over that wonderful place by some very wonderful people.

So should you decide next year to stay in Britain for your family holiday I hope it will be because you love this place and not because you feel afraid to take your child abroad because I would like to think that this story goes in some small way to alleviate those fears.

In closing this story I would like to tell you as long as you take the same sensible precautions that you would take at home and that you carry with you such things as Maxamaid and Polycal as these will be very expensive and difficult to get hold of abroad. Then you have nothing to be afraid of so ask for help if you are not sure and trust that the local people will do all in their power to help you have the same fantastic time that was had by myself and all my family.

All that remains is for me to wish all well for the future and to say that we hope to see you all out by the pools and on the beaches when my family and I take our next holiday.

We are thinking of trying Disneyland U.S.A. next.

All the best.
Fred and Josie.
From Nottingham.

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Special Foods
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International Meeting in Denmark Considers Prese

Dr. Flemming Güttler, from the J.F.K Institute in Glostrup, Denmark, organiser of the Elsinore meeting.

FROM JUNE 27 to 30, 2002, 135 scientists and PKU clinicians from 24 countries attended an exciting international meeting in Elsinore, Denmark.

Dr. Flemming Güttler, a recognised expert in the field of inherited metabolic diseases and molecular genetics organised the meeting.

It came 8 years after a similar meeting in Elsinore, also organised by Dr. Güttler, that celebrated the 60th anniversary of the discovery of PKU.

As a meeting participant on both occasions, I marvelled at how much new research is going on, and how far we have come over the 8 years in deepening our understanding of PKU at a biochemical and genetic level.

PKU in the 21st Century
by Dr. Charles R. Scriver,
Dept. of Human Genetics,
McGill University, Montreal, Quebec

The Danish philosopher Soren Kierkegaard observed that life can only be understood backward but must be lived forward. It is a useful perspective for PKU.

At the beginning of the 20th century, scientists recognised that genetic variation and Mendel's rules of inheritance would explain some of our human biochemical individuality. In 1899, Sir Archibald Garrod gave the name "inborn error of metabolism" to this idea of a genetically caused variation in metabolism. With its discovery in 1934, phenylketonuria was the fifth example of Mendelian inheritance of a metabolic variant; the associated disease in this case (mental retardation) was highly significant and attracted notice.

The cause of the disease lay both in Nature (a genetic mutation) and in Nurture (dietary phenylalanine, the toxic chemical in PKU). Once the Nature-Nurture relationship was surmised, clinicians recognised that nurture could perhaps be altered (with a low phenylalanine diet), metabolism controlled, and thus mental retardation prevented. If this were done right after birth, there was also a rationale for newborn screening (early diagnosis and treatment). PKU thus became the prototype for a treatable genetic disease and for newborn screening. Most significantly, it transformed our views about what could be done to treat "genetic disease." In the latter half of the 20th century, we identified the mechanisms underlying the metabolic abnormality causing elevated blood phenylalanine in PKU: we recognised and characterised the pathways of phenylalanine metabolism; and we identified and characterised the enzyme (phenylalanine hydroxy-lase, or PAH) that is necessary to convert phenylalanine to tyrosine at the head of the major pathway. We also mapped the area harboring the gene that is responsible for making the enzyme, locating it on human chromosome 12. We then cloned its DNA sequence, allowing us to analyse for the genetic mutations that result in PKU.

We soon discovered that many mutations cause phenylketonuria. Currently we know of over 400 mutations causing elevated blood phenylalanine (see http://data.mch.mcgill.ca/pahdb_new/). But in any human population, we found that less than 10 different mutations will account for about

nt Knowledge of PKU and Future challenges

75 percent of the mutations appearing in the population. We also discovered that 3 out of every 4 PKU patients inherit two different mutations from their "silent carrier" parents, instead of two similar genetic mutations.

PKU is an "autosomal recessive" disorder (in Mendelian jargon) which means that the parents who carry the mutation are silent about its effect because the mutant effect is buffered by the rest of the genome. Only when the child inherits two copies, one from each parent, does the PKU disease manifest itself. We now know that different mutations have very different effects on phenylalanine hydroxylase function, with variable metabolic effects and different clinical consequences. Thus every patient has his or her own form of "PKU." The genetic explanation lies in the combination of PAH gene mutations and the background genome of 30,000 other genes and their effects. Each PKU patient is indeed unique!

We know that the PAH enzyme requires a co-factor called tetrahydrobiopterin (BH4) to function. BH4 is the product of a biosynthetic pathway involving several enzymes. The cofactor must be regenerated following the phenylalanine hydroxylation reaction; two enzymes are involved in that process. Mutations affecting synthesis or recycling of BH4 also cause hyperphenyl-aninemia (HPA) in very severe variant forms, which do not respond to dietary restriction of phenylalanine alone. PKU management programs now take into account these "BH4-dependent" forms of HPA, the frequency of which varies among human populations. However, we are identifying new forms of HPA that are "BH4-responsive." They seem to involve one of two response mechanisms: either the high dose of BH4 overcomes "unfriendly binding" of cofactor on the surface of the PAH enzyme; or the BH4 molecule itself stabilizes the mutant PAH enzyme protein and allows it to function.

The latter idea fits into an important new view: namely that 60% of PAH gene mutations produce their effect through "misfolding" of the PAH enzyme protein subunit, leading to its rapid intracellular removal. Anything that would stabilise the protein for a longer life in the cell would improve function and thus be "a treatment." BH4-responsive PKU thus becomes a model for thinking about such a form of therapy for other genetic diseases because 50% of all human mutations probably produce their effects through protein misfolding and instability. One anticipates a growth industry in the invention of soluble chemical "chaperones" to stabilise mutant proteins. When we sequenced the PAH gene we anticipated that mutation analysis would predict phenotype (clinical expression) and thus the intensity of treatment for classical PKU and variant forms of HPA. While there is a broad correlation between mutation type and metabolic effect, the prediction is not perfect. PKU then becomes a reminder that "genotype" (the specific mutation) is not necessarily the "phenotype" (clinical expression) for any Mendelian disorder.

Thus PKU emerges as yet another prototype, showing that Mendelian diseases are indeed complex. Accordingly, scientists interested in PKU are learning on another level what parents and patients have always known: notably, that every patient has his or her own particular form of PKU/hyper-phenylalaninemia. In other words, treat the patient not the genetic mutation!

New guidelines from Europe and a recent NIH consensus conference in the U.S. (see Winter 2001 issue) recommend treatment of PKU as early as possible, with greater stringency as long as possible, perhaps even for a lifetime. The effectiveness of treatment is particularly important for the female patient who wishes to become pregnant and protect the fetus from the intrauterine effects of maternal hyperphenylalaninemia (see Fall 2002 issue). Because of the prospect for prolonged treatment we hope to see treatment modalities improved. This would include improving the properties of existing dietary products; adopting other metabolic approaches to prevent excess plasma phenylalanine from penetrating the brain compartment; and to provide alternative enzyme therapy (for example, there is research underway with the enzyme phenylalanine ammonia lyase to degrade phenylalanine in the intestine before it is absorbed into the body pool).

PKU in the 21st century will continue to illustrate how the intellectual discipline of genetics and the practice of genetics are different, yet also come together. Mendelian genetics was a transforming paradigm to understand many hundreds of human diseases; but a solely Mendelian view is an oversimplification for most genetic problems. PKU has been a prototype for the growth of this knowledge about human genetics. The latter is important not only for people with PKU but also for those with any human genetic disease as simple or as complex as it may be.

PAH Enzyme Structural Studies
by Dr. Heidi Erlandsen, Dr. Scient and Dr. Raymond C. Stevens,
The Scripps Research Institute, La Jolla, California

We know that mutations in the gene that "encodes" for the enzyme phenylalanine hydroxylase (PAH) cause both phenylketonuria (PKU) and hyper-phenylalaninemia (HPA). PAH converts the amino acid phenylalanine from proteins in the diet into another amino-acid, tyrosine. As Dr. Scriver points out, the enzyme needs a cofactor, called tetrahydrobiopterin (BH4), as well as oxygen from the air we breathe. We now know there are more than 400 PKU/HPA mutations in the PAH gene.

Most of these PKU/HPA mutations result in an enzyme with changed chemical and physical properties compared to the normal enzyme (often referred to as "wild-type PAH"). Scientists have studied extensively the effects of these mutations on the PAH enzyme in biochemical systems within a test-tube (also called in vitro systems) by using a cloned and biochemically-produced version of the enzyme.

To help in the interpretation of the mutational studies, our research group first determined a three-dimensional atomic structure of the phenylalanine hydroxylase wild-type enzyme in 1997 *(see figure, 1)*. This structure of the PAH enzyme is a representation of the shape of the human protein. It tells us where the individual atoms of each amino acid that constitutes the protein are located, in three-dimensions. Based on this three-dimensional structure we can identify the sites of the mutations found in PKU/HPA patients, and start to explain why the mutation leads to non-functional protein (and therefore an accumulation of the amino acid L-phenylalanine, acquired through eating proteins in the diet).

The three-dimensional structure of the PAH enzyme shows that the enzyme forms three distinct domains (sections): one regulatory domain of the enzyme (which regulates the phenylalanine to tyrosine conversion), one catalytic domain (responsible for the actual phenylalanine to tyrosine conversion), and finally a tetramerization domain (responsible for putting together 4 molecules of the same kind into a functional large molecule called a "tetramer"). We have identified several three-dimensional structures of phenylalanine hydroxylase since 1997. We now know where the phe to tyrosine conversion occurs, and where the phenylalanine and BH4 is located in the structure. The exciting thing is that we can use this three-dimensional structure to predict the effects of newly discovered mutations on the PAH enzyme.

The future goal for our research is to use the structure to make a form of the PAH enzyme that can be taken orally (before a meal) along with the BH4 cofactor, as an enzyme replacement therapy (along the lines of the Phenylase—phenylalanine ammonia lyase—research mentioned in the Fall 1999 issue). Hopefully, this would mean ultimately that a patient can consume a "normal" diet instead of a low phenylalanine diet.

Investigation of Phe Transport into the Human Brain By Dr. Harald E. Moller, Max Planck Institute of Cognitive Neuroscience, Leipzig, Germany

Magnetic resonance imaging (MRI) is a modern technology based on the extremely weak magnetism of certain atomic nuclei, for example, hydrogen nuclei (commonly referred to as "protons") in water molecules. By utilizing a strong magnet and radio-frequency signals, detailed images from the living human body are recorded entirely non-invasively and with no known risk. Further metabolic information about certain chemicals in different tissues can be obtained with the same equipment and a closely related method called magnetic resonance spectroscopy (MRS). Researchers at Yale University were the first to detect a phenylalanine MRS signal in the brain of animals made hyperphenylalanin-emic. Even

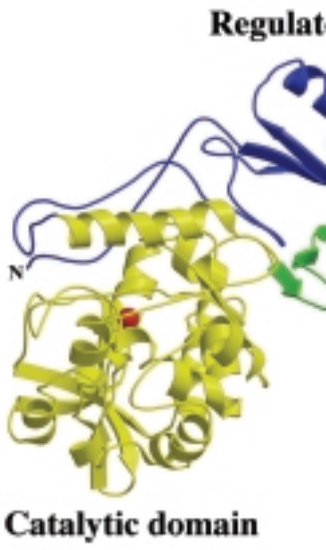

Figure 1.

more interesting, they observed that the amount of phenylalanine in brain differed from the amount in blood.

Our laboratory at the University of Münster, Germany, like a few others, uses MRS to measure brain phe concentrations in patients with PKU. Because PKU can result in neurological and psychological problems for off-diet patients, we are interested in comparing brain phe and occurrence of such problems. Phenylalanine is not produced by the human body. When food is ingested, phe is delivered to the brain by circulating blood. However, like most other chemicals, phe cannot enter the brain cells directly. It must be transported across the "blood-brain barrier" by a specific carrier protein located in the capillary cell membrane. (First indications that the central nervous system is separated from the blood by a barrier of some kind arose about a century ago, when the German immunologist Paul Ehrlich observed that dyes injected into a vein of laboratory animals stained all organs except the brain.)

Phenylalanine shares its blood-brain barrier carrier protein with a number of other amino acids, all of which compete for available carrier sites. This concept might ultimately be put into use to support diet therapy in PKU: by ingesting high amounts of competing amino acids to block carrier sites for phe, brain phe could be reduced despite high blood phe (Editor's note: This is the idea behind the Danish pill, PreKUnil, described in the Winter 2002 issue).

We also study the rate of phe transport across the blood-brain barrier. After the patient takes a certain amount of phe we execute a series of MRS examinations to observe how rapidly it accumulates in the brain and then declines back to normal levels. Recently, we observed abnormally low brain phenylalanine concentrations in spite of high blood levels in three untreated women with classic PKU. Although they had never received any diet, they were almost unaffected clinically and had normal intelligence scores. Our hypothesis is that phe is not transported efficiently into the brain if it cannot bind well to the carrier protein. This would explain abnormally low brain phe levels and might provide some sort of "protection" of the brain against PKU in some individuals. Studies in a larger group of patients indicated that the blood-brain barrier phe transport properties vary to some extent among individuals, and correlate weakly with the patient's clinical status. However, more questions remain to be answered before we can reliably ascertain that individual blood-brain barrier transport is a major factor or only one of subtle influence in typical PKU patients. We believe that other experiments on phe transport and its potential manipulation by therapy are indicated.

Figure 1. Schematic of the "backbone" peptide (protein) structure of the human phenylalanine hydroxylase enzyme. The "curly" regions are caused by the peptide forming a specific structure called alpha-helices. There are 3 regions of the enzyme: the regulatory domain (amino acids number 19-142 of the peptide sequence), catalytic domain (amino acids 143-410 and the tetramerization domain (amino acids 411-452). The active site, where the L-Phenylalanine to L-Tyrosine conversion takes place, has an iron atom which is vital for the enzyme's function (shown as a very small darkened sphere at the center of the catalytic domain).

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Developing an Information Pack fo

Susan Durham-Shearer,
Research Dietitian
University London College Hospital NHS Trust

FOR THE past two years I have been involved in a research project at University College London Hospital, working alongside metabolic consultant Dr Philip Lee and metabolic dietitian Maggie Lilburn. The aim of this project has been to develop a practical information pack for teenagers and adults with PKU. This article is to share some of the results of this work and our plans for making the pack available to other people with PKU.

Scientific Background

In 1969 routine neonatal screening for phenylketonuria (PKU) was introduced in the UK. Before this time most people with PKU were not diagnosed and remained untreated. This causes mental retardation as the high blood phenylalanine levels damage the infant's brain and central nervous system Paine (1957). We now know that starting a low phenylalanine diet in the first few days of life prevents much of this damage and enables the person to develop normally Sutherland et al (1966). With the start of PKU screening children born in the 1970s have become the "first generation" of early-treated PKU patients to grow up and lead normal lives in the community.

In the early days it was thought safe to stop the diet around 6-8 years of age, and it was unusual for children to remain on diet for longer than this. However, as research continued it became apparent that the children who were taken off diet had lower IQ scores than those who had remained on diet Cabalska et al (1977), Smith et al (1978). It was then decided that children should remain on diet until they were in their teens Azen et al (1991).

Research in the last decade has now shown that raised blood phenylalanine can affect the brain in adults. This has been seen in altered brain scans (Magnetic Resonance Imaging) of people with high blood phenylalanine levels Cleary et al (1995). More studies are needed to fully understand these findings and we are unlikely to understand the long-term effect of high blood phenylalanine in adults for several decades. Until this time doctors have decided that it is safest to recommend adults remain on "diet for life" to hopefully prevent any damage high levels of blood phenylalanine may cause Medical Research Council (1993).

Why is the pack needed?

Because of the relatively recent change in opinion about adults staying on diet, there is a lack of both medical and practical resources to help this group achieve this. Most facilities are still centred around treating children, and the more complex needs of adults have been overlooked.

Some healthcare professionals still see PKU as being a "children's condition" and are not aware of the recommendation for adults to stay on diet. This can create serious obstacles for adults trying to return to, or stay on, the low phenylalanine diet

Good phenylalanine control can only be achieved through good compliance with the diet. Children have their parents to prepare their food for them, do blood tests and motivate them to comply with the diet. Adults must manage their own diet and be responsible for their own PKU management. This can be very difficult when trying to fit the diet around work, social and family commitments, such as eating out, going on holiday and socialising with friends.

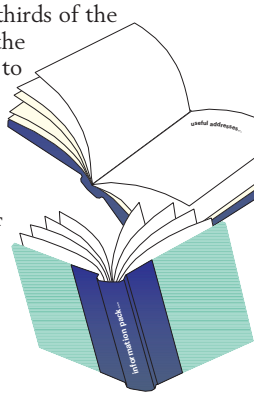
So far little has been written about dealing with these problems and offer practical advice to cope with the diet. To address this need the project at University College London Hospital was set up to develop and audit a resource pack specifically for teenagers and adults with PKU.

How was the pack developed?

The project to develop the pack was started in May 2000. A questionnaire was sent out to 177 early-treated patients in the clinic with classical PKU, either on or off the diet. This questionnaire was used to find out how people were currently managing their diets, their experiences of coping with PKU and what they thought should be included in the resource pack. They were also sent a simple knowledge test to measure how much they knew about PKU and the diet.

72 questionnaires were returned and the results were used to identify what people wanted from the pack, what format it was to be in and which topics to include. The pack was then produced over a twelve-month period with help from members of the University College London Hospital metabolic team and other experts in PKU.

32 patients currently on PKU diet agreed to take part in the audit of the pack. Two thirds of the patients were sent the pack while the other third were not sent the pack to act as a "control group". This was done to see if there was any difference between the two groups which might be due to the pack intervention. After one month all patients were sent a repeat copy of the original questionnaire and knowledge test to see if there had been a change in their dietary management and knowledge about PKU. This was then repeated six months after the intervention.



r Teenagers and Adults with PKU

Results of the project

Much information was gathered from patients describing their experiences with PKU and what they would like to see in the pack. The majority of people said they would like the pack to be a small "filofax style" ring-binder with removable inserts for quick reference. A wide variety of topics were requested, which were incorporated into the pack.

These are listed in the table (below).

Table 1: List of pack contents

- Laminated FACTCARDS explaining key topics in simple language to help assist explaining the diet to people who do not know about PKU
- Understanding PKU and its treatment
- Getting your prescription products
- Recipes, planning meals and cooking tips
- Controlling your weight on diet
- Eating out, alcohol and aspartame
- Work, family and holidays
- Coming off diet
- The future treatment of PKU
- Life stories (accounts written by people with PKU on how they have coped with an aspect on the diet)
- Being on diet in your teens
- Leaving home
- Planning your pregnancy
- Products available on prescription
- Free, forbidden and exchange foods
- Daily PKU diet prescription
- The protein calculator (for working out the protein content of foods)
- Useful addresses

When will the resources be available?

The pack and the other two resources are presently being tested at the University College London Hospital clinic and are available to all PKU patients attending this clinic through the metabolic dietitian. If this trial is successful we hope to make the pack available nation-wide to all metabolic clinics in 2003. When the pack is ready SHS, the company who have sponsored this project and printed the pack, will be liaising with metabolic dietitians around the country to make them aware of this resource. The packs will be made available through your metabolic dietitian who will ensure that your pack contains all the information relevant to your needs.

In addition to the main resource pack two additional resources were also produced in response to the results and comments gathered in the study:

- Overall compliance with blood testing was shown to be poor, therefore a **blood testing folder** designed to hold blood testing equipment was produced to assist patients with organising these items.
- Many patients reported that they had experienced problems when explaining the diet to healthcare professionals who did not understand why they needed the prescription products. In response to this a short booklet **"The Dietary Treatment of Adults with Phenylketonuria (PKU): A Guide for Healthcare Professionals"** was produced, explaining why adult patients are advised to follow a low phenylalanine diet.

Although the audit did not show a statistically significant improvement in patient's PKU knowledge or dietary practices after one month, general feedback about the pack was very positive. The results of the six month audit currently being completed will add to the information already gathered and will show if any longer-term changes have happened as a result of the pack. All the feedback obtained will be used as part of the yearly up-date process of the pack to ensure information is kept current.

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"Hurrah for Supplements!"

I KNOW its generally considered to be a Mothers lot to spend her life nagging at the children (and of course the husband) but adding the daily quota of nags to get protein supplement into my PKU son seemed to make my life just one long nag.

It all started when Louis was about 6 months. He suddenly decided that analogue was not for him. There followed a worrying few weeks while our inexperienced dietitian came up with various ploys to try to encourage him to drink it. After a worrying suggestion, a visit to Isobel Smith at Great Ormond Street and a change of care to St Mary's in Portsmouth Louis was changed onto Maxamaid at about 7 months. This was the start of over 5 years of hell!

Not a big drinker (unlike his Mother) at the best of times, trying to get anywhere near the required quantity of Maxamaid into Louis was a monumental task. The first year, until he was of an age to use other ploys I used to follow him around all day with his cup of Maxamaid persuading him to take sips every now and then. We tried mixing it with juice, squash, fizzy stuff, milkshakes….you name it, it went into his Maxamaid! This was probably the most upsetting stage as the poor boy had no understanding of why his evil Mother was constantly hounding him. The only thing that worked was to deny him any other drink until his Maxamaid was finished. On one occasion I was out for the day with a friend, as Louis had fallen asleep we decided to stop in a café for a cup of tea. While we were there Louis woke and started crying for some of my tea. I refused and offered him his Maxamaid. He became very upset, as did I and through all this I could see a woman and her elderly mother keep looking at me and whispering. I could just tell what they were saying. They were saved from an obscene mouthful by my friend ordering me out of the way while she took over with Louis for a while. The next stage when Louis was about two was giving him a portion after each meal and making him sit at the table until it was finished, we went through every game and bribe in the book. In fact we got so far down the road of bribes that Louis had us wrapped around his little finger. We'd have done anything just to get him to do his Maxamaid and he knew it. Getting back to basics was really hard on him. From doing everything we suddenly did nothing, he just had to sit there until it was gone. One day he sat at the table from breakfast time until lunchtime, had his lunch, went to bed for his sleep then went back to the table until tea time. He then decided to give in and drink it. I dashed off into the lounge a few times that day to have a good cry.

It was not long after this at a conference that other parents suggested that I tried concentrating the Maxamaid so there was a smaller quantity for Louis to drink. It seemed so obvious but it had never occurred to me. Over the next few weeks we gradually reduced the volume from 500ml to about 250ml and started spooning it in. Louis dad invented all sorts of games and life got a bit easier, especially when Auntie Jackie introduced a medicine syringe. However our patient dietitian has had to put up with me sobbing "I can't do this anymore" to her on a number of occasions.

This method lasted for quite some time until Louis was about four when for some reason, I can't remember quite why, we concentrated still further and reduced the Maxamaid to a paste. This was generally better, although we had to make sure Louis didn't swallow too quickly or it all came back again. The worst of this was trying to judge if he was making a fuss because he really did feel as sick as he said or if he was just trying to avoid taking it. I regularly misjudged and the bucket came into play, followed by the fun of trying to judge how much he'd had that day to try to replace what had "resurfaced"!

By the time Louis started school about 6 weeks before his 5th birthday taking Maxamaid was a well established routine. Nagging was very much a part of this (I'm not as good at games as dad!). He even took it to his friend's houses when he went for an after school play, I don't think his friends mum's had to nag.

New hope dawned when other products were made available. I made the mistake when the PKU gel was launched of treating it as some sort of saviour, that Louis would love it and our problems would be over. He was very willing to try it but didn't like it so it was back to my old enemy Maxamaid. I had wised up a bit by the time Minaphlex arrived. Louis again was willing to try it, said it tasted better but didn't want to change from Maxamaid, (what can I say he's a man!). These trials did have one unexpected benefit though, they made him accept the Maxamaid more and if (or should I say when) he made a fuss I could always suggest he tried something else. He was also of an age by now to reason with.

My biggest heartache through these years was always that I was forcing my child to swallow something which, lets be honest is absolutely foul. Through all his tantrums and tears I had a huge amount of sympathy for him but what good was that to him? How do you make a young child understand it's for their own good. Mike and I had started to watch the adults and older children with their tablets with envious eyes. We were almost wishing the years away. Then came our miracle. The scene; a campsite in Cambridgeshire in a tent with dear old N&V ed. Pete Bramley (not a pleasant thought I know!) and his family. Their daughter Nicola was taking her Aminogram tablets when someone suddenly said "Would Louis like to try one?". Being an adventurous type he said yes. We armed him with a huge glass of juice and surrounded him in case he choked (have you seen the size of those things?). One minute it was there and the next it wasn't, it hadn't even touched the sides.

The rest is history, a visit to an amazed dietician and a few phone calls later Louis was sent a supply of the brand new Phlexy 10 tablets. Within a week he was swallowing three at a time and now three months later he can do six. He started off with Phlexy vits as a vitamin supplement which wasn't particularly pleasant but now he's taking vitamin tablets with a rather nice orange flavoured calcium solution. I still have to nag a fair bit but at least I know it doesn't taste awful. Louis at just turned six is the youngest person our dietitian knows to go on to these tablets and after his experience I believe she is going to recommend it to other's who have problems with their supplements at an earlier age than any of us thought possible.

Now I only have to get him to eat!!

Julia Bailey

A Close Shave in France!

An alternative approach to raise funds for NSPKU...

IT WAS a gorgeous August morning, and Julia, Louis, Meryn and I had arrived in South Brittany the night previous, after a fairly tiring car journey from Cherbourg.

Yes, you guessed we were just starting our summer holiday, which I always think is a fantastic feeling. Recuperation and total relaxation from the day to day rat race that we find ourselves embroiled in each day.

We all sat at the breakfast table eating just outside our mobile home enjoying the sunrays and French ambience, when suddenly, I realised what day it was. It was Saturday and this was a significant day in my holiday. Why, I can hear you all ask? Well, this was to be the day when I had agreed to have my head shaved for the NSPKU, to raise funds. Enjoyment soon became panic and panic was reality. It seemed such a good idea at the time but by the end of today I would be hairless which was a very scary thought!. The morning soon passed by.

The sign above the door in Vannes read "Didier DANO Coiffure Hommes", which I can assure you is not translated as "head butchery for men". This was certainly one family outing not to be forgotten. As I stepped inside the hairdressers, I was greeted by a lady smiling (did she know what was going through my mind? – of course not, how could she possibly know?) offering a very friendly bonjour. After all the niceties were over, I was eventually shown to the chair where the act would take place. The stylist then asked me in French what hairstyle I wanted. I retorted very quickly with "no style", I am having my head shaved completely. She looked slightly perplexed wearing a gentle smile; was this English humour at its ridiculous best? Eventually, I managed to convince her. It would have been a perfect opportunity to call the whole stunt off, but alas it was too late. Her razor glided gently over my whole head and as I looked in the mirror it seemed like balls of hair were just falling around me. They were! Julia, Louis and Meryn had positioned themselves carefully, so that they all had a first class view, and amidst all the general frivolity and giggling at my expense, managed to catch some of these moments on film. Even the stylist began to join in and celebrate my misfortune. After a short time, I was finally shaved and the need to continually run my hands over my head became a very regular occurrence. My son Louis (aged 6 years) is PKU, which is why I had decided to have my head shaved in France (I had thought that the French Kings' connection, made it much more personal to our family) although I think Louis was actually quite relieved that we were not in England at the time, as I am sure he would have been embarrassed. We all left.

It was now late afternoon, and we decided to go food shopping after the Coiffure encounter. All I kept thinking in my own mind was were people looking at me because I had no hair now! Isn't it sometimes frightening how your mind runs away with you?

Anyway, for the next 11 days I holidayed with my new look and actually, it was not as bad, by any stretch of the imagination as I had first imagined.

So there you have it….. "Hair Today Gone Tomorrow". No, but seriously, I guess what I learnt from the whole experience is that sometimes, when you first think about something that seems difficult to carry through, it is quite often the reverse. Also and more importantly, raising funds for such a worthwhile cause – The NSPKU is so gratifying.

Finally, I must end by saying a very big thankyou to all of you who were kind enough to sponsor the Head Shave event. It is much appreciated.

Mike Bailey

LONDON AND HOME COUNTIES SUPPORT GROUP

Roller-Skating Event - ***amendment!!***

Sunday 16th March 2003 (not Saturday), from 12–2 pm at Roller City in Welwyn Garden City, Herts. Directions will be forwarded to you, Private hire to PKU L&HC, Resident DJ, FREE Skate Hire or you can bring your own. Food and drinks provided (PKU and non-PKU). If you would like to attend and have not yet replied, please contact:

Sandra Bramley,
48 Hazeldell,
Watton-at-Stone,
Herts, SG14 3SN
(Telephone 01920 427636)

SHS recipes

Cherry Tomato Tarts with Minted Mayonnaise

Ingredients

- 150g (6oz) Loprofin Low Protein Mix
- 75g (3oz) economy butter
- 2.5mlsp (½ tsp) dried basil
- Pinch of salt

Filling

- 75g (3oz) onion, finely chopped
- 1 clove garlic, crushed
- 1 x 15mlsp (1tbsp) olive oil
- 300g (12oz) cherry tomatoes
- 2 x 15mlsp (2tbsp) sundried tomato puree

Garnish

- 3 x 15mlsp (3tbsp) mayonnaise *
- 2.5mlsp (½tsp) mint sauce
- 4 sprigs of fresh basil

Makes 4 tarts
4 x 15cm (5inch) loose bottomed tart tins
Oven temperature: 200°C/400°F/Gas Mark 6

Method

1. Place the Loprofin Mix in a large bowl, rub in the butter until the texture resembles coarse breadcrumbs. Stir in the dried basil and salt.
2. Gradually add sufficient water to give a soft manageable dough, lightly knead for 30 seconds on a surface dusted with Loprofin Mix until the dough is smooth.
3. Roll out the dough and use to line 4 x 15cm (5inch) tart tins.
4. Heat the olive oil and stir fry the onion and garlic for 3 minutes, over a moderate heat, until transparent.
5. Halve the tomatoes.
6. Stir together the tomato puree and onions, then stir in the tomatoes, reheat, stirring gently, for 1 minute.
7. Divide the tomato filling between the pastry cases, arranging the tomatoes decoratively.
8. Bake in a preheated oven for 15-20 minutes, until the pastry is just starting to colour and the tomatoes are bubbling.
9. Combine the mayonnaise and mint sauce. Serve the tarts warm or cold, garnished with a spoonful of Minted Mayonnaise and sprig of fresh basil

Tip

* Check NSPKU Handbook for suitable brand

Vanilla Biscuit Cake

Ingredients

- 1 Low Protein Toffee Flavoured **Duobar**
- 65g (2½ oz) economy butter
- 50g (2oz) caster sugar
- 50g (2oz) Golden syrup
- 2 x 15mlsp (2tbsp) Low Protein **Snopro** Drink
- 1 packet **Loprofin** Low Protein Sweet Biscuits, crushed
- 100g (4oz) cake crumbs from a Low Protein cake or muffin
- 75g (3oz) luxury mixed dried fruit

Topping

- 2 Natural Flavoured Low Protein **Duobars**, melted
- 8 **Loprofin** Low Protein Vanilla Flavoured Wafer Biscuits, crushed

Cuts into16 wedges
17.5cm (7 inch) round sandwich tin, greased and base lined

Method

1. Break the Toffee **Duobar** into pieces and place in a saucepan, with the butter, caster sugar, golden syrup and **Snopro**. Place the pan over a moderate heat, stir occasionally until the ingredients have melted.
2. Increase the heat and bring to the boil. Reduce the heat slightly and cook at a 'rolling' boil for 1 minute.
3. Remove the pan from the heat and stir in the biscuits, cake crumbs and fruit, until thoroughly combined. Transfer the mixture to the prepared cake tin.
4. Press the mixture firmly over the base of the tin and level the surface.
5. Combine the melted natural **Duobars** and crushed Wafer Biscuits and spread evenly over the mixture in the cake tin, chill until set.
6. Cut into small wedges to serve.

Tip

Use a potato masher to make light work of pressing the mixture evenly and firmly into the cake tin.
It is easier to cut the cake if it is allowed to return to room temperature, prior to cutting.

Dietitian's Report

DIFFICULTY IN OBTAINING PRECRIPTIONS

Some of you have been in touch with me about the difficulties you have had in obtaining your prescriptions from the chemist. You may have been told that a certain item is out of stock or discontinued or that it is unobtainable. In order for me to investigate any problems which you have I need to know the following:

1. ***Name of product and manufacturer***
2. ***Name of Chemist, telephone number and name of shop***
3. ***Town or city where this Chemist is situated***
4. ***What the difficulty is i.e. told the product is discontinued***

I can then get in touch with the manufacturers and they can investigate the problem for you. The companies have been very good about this and several problems have been resolved.

TRIAL OF DIET FOR PREVIOUSLY UNTREATED PKU PEOPLE

This is just to keep you up to date on what is happening about the trial. We shall soon hear from Wellcome if we have been successful in obtaining a grant from them to run this trial. We remain hopeful but if we are rejected then there is another card up our sleeves. We have also applied to the Community Fund which is the charity arm of the lottery. They have a sum of money which is available for research into learning disabililty and for which we believe our work would qualify for very well. The Community Fund is now assessing our application, but it takes time and we will not hear the outcome until March. The assessors have been in touch with us for a detailed telephone interview as well as having our written application form. I will keep everyone informed of the outcomes of these applications as soon as I hear anything more.

PRESCRIPTIONS

There is an up to date list of all foods and drinks which are available on prescription in the Dietary Information Booklet 2002/2003.

All of these products have been recognised as important items in the diet by the ACBS (Advisory Committee on Borderline Substances) and are paid for by the Government for the person with PKU.

The vast majority of GPs prescribe these items as required but I have heard from some people that they find it very difficult to obtain sufficient quantities of foods and for some if a new item is added to the list of foods prescribed another item is crossed off. We do have a leaflet which has been written for GPs which briefly explains the importance of the prescribable foods.

This can be obtained from our publications officer, Joanne Clough, details below.

If problems do become difficult for you to manage a letter from your consultant or dietitian at the hospital which explains that these items are not luxuries but essential sources of calories and variety can help.

Joanne Clough
nspku.publications@nspku.org

WHAT IS THE PHENYLALANINE CONTENT OF GELATINE AND CELLULOSE COATED CAPSULES?

The carer of a patient with phenylketonuria recently asked me this question. The best way of saving time and getting all the information you need is to ask somebody who has some expertise on the subject. I rang SHS as they have a product development department which has looked at many ways of presentation of many types of medication and who would know the importance of the phenylalanine content of casings.

Soft gelatine capsules are usually used to encapsulate liquid or oil medications. They contain typically 4 mg of phenylalanine per capsule. Small quantities of these capsules would not need to be counted in the diet but larger numbers could provide a significant quantity of phenylalanine. Your doctor or dietitian would be able to advise you further about this.

Cellulose is usually used to encapsulate powder products. It makes a hard outer shell and contains very small quantities of phenylalanine. A typical example would be the Phlexy –10 capsules containing amino acids. The phenylalanine content is less than 0.2mg of phenylalanine per 20 capsules. This is a negligible amount.

My thanks must go to Helen Stanton, clinical nutrition specialist at SHS, who provided me with this information.

OVER THE COUNTER MEDICATION FOR CHILDREN

I have been looking at over the counter medicines which do not contain aspartame and are therefore suitable as far as phenylketonuria is concerned. However, you should also ask your pharmacist if you are unsure and of course if there is another medical condition which needs to be considered.

FOR COUGHS

Benylin
Benylin Children's Chesty Coughs Age 1-12 years
Benylin Chesty Coughs non- drowsy Age 6-12 years

Tixylix
Tixylix Baby syrup
Tixylix Chesty Cough

Venos
Venos Honey and Lemon Tickly Coughs from 1 year

Meltus
Meltus Cough Linctus Baby Formula
Meltus Junior

Karvol
Karvol Decongestant Tablets

Streppies
Streppies Sugar Free Children's pastilles Age 4-12 years

TEETHING GELS

Dentinox sugar free
Calgel sugar free
Bonjela ordinary and sugar free

COLIC

Dentinox Colic Drops
Infacol
Woodwards Gripe Water
Nurse Harvey Gripe Water

PAIN AND FEVER RELIEF

Calpol
Calpol 3 month plus sugar free suspension
Calpol 6 plus sugar free suspension

Medinol
Medinol under 6 sugar free
Medinol over 6 sugar free

Numark
Ibruprofen suspension sugar free

TOOTHPASTE

Some of you may have noticed that the artificial sweetener saccharine is used in some flavoured toothpaste. I have been asked if aspartame would ever be used as a sweetener.
The answer is that aspartame would not be used in toothpaste because it does not work in alkaline substances. Toothpaste is an alkaline substance. Therefore people with phenylketonuria need not worry about aspartame being present in toothpaste.

NB: All medication will have the ingredients list with the patient information. If aspartame were present it would be listed.

It is only the artificial sweetener aspartame which contains phenylalanine.

Other artificial sweeteners such as Acesulfame K, sorbitol and saccharine do not contain phenylalanine.

!WELLCOME NEWS!

STOP PRESS! STOP PRESS!

I am delighted to report that our application to Wellcome for funding of the trial to study the effectiveness of low phenylalanine diet for adults with previously untreated phenylketonuria has been successful.

We heard this news just before Christmas. The steering committee is meeting in early January to begin work on recruiting professionals to run the trial and organise the timetable of work. The trial will run for three years and is a huge amount of work but we know that it is very worthwhile. It is such good news for everyone involved; the Society, the steering group who have spent so much time and effort in obtaining this grant and most importantly for those people and their families born before screening who were not treated and for whom this work may mean an improved quality of life.

I will keep readers informed of the progress throughout the year. What a wonderful start to 2003!

PACKED MEALS

Q. Do you take a packed meal to school or work or when away from home for an outing?

A. If the answer to this question is 'yes' then what do you take?

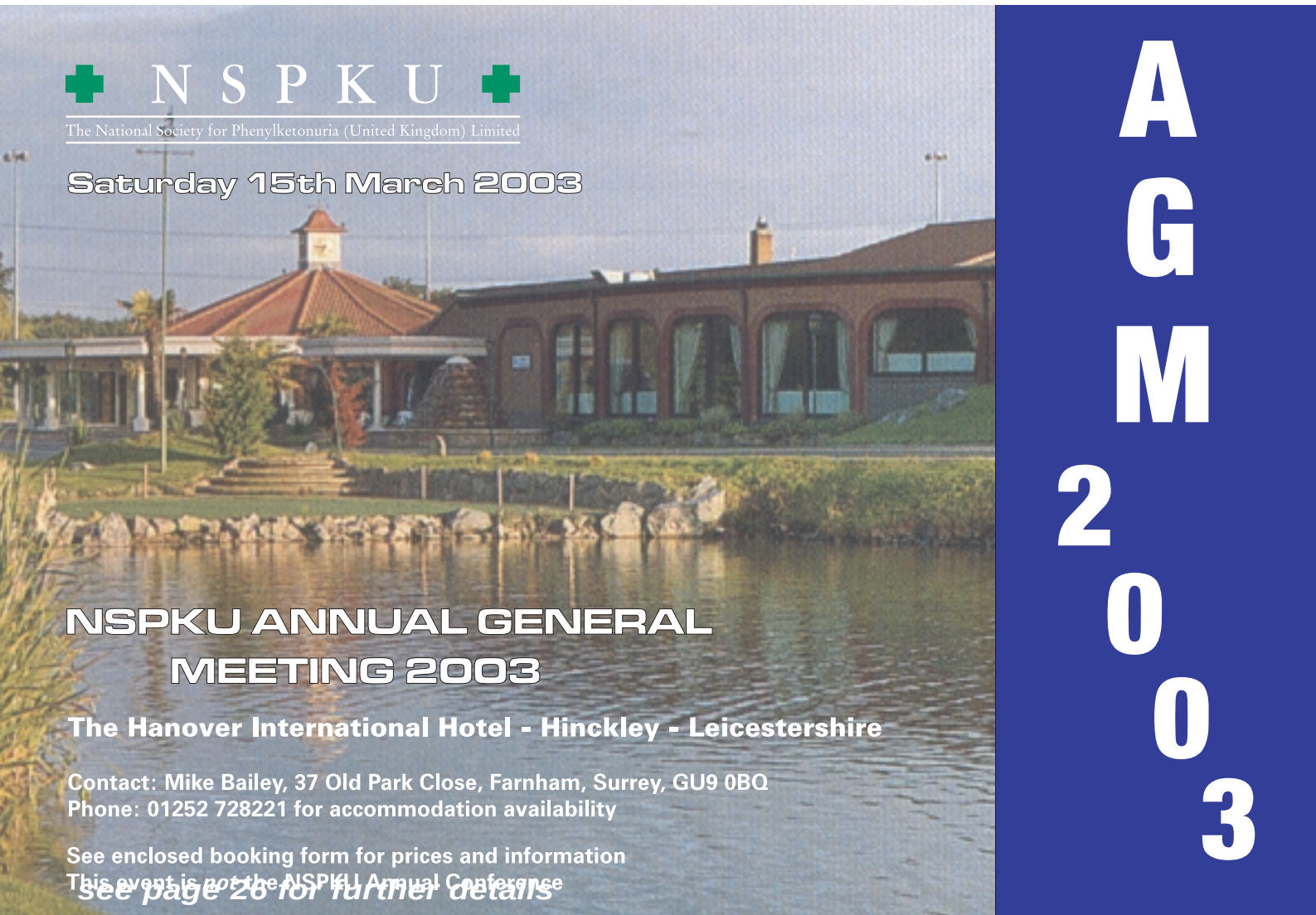
What are your favourite things to pack and what do you find really useful?
Have you got any hints and tips about a packed meal?
Are there any special things you make or bake for the packed meal?

I would love to hear from ANYONE who can help me with this. The reason is we are getting quite low on our publication 'Packed meals for PKUs' and need to up-date it. The best people to help me with this are yourselves so please don't be shy and think that your ideas are not important. They are to me and to others who need some inspiration, and we all need that from time to time!

Please ring, write, telephone or email at the contact details at the front of the magazine.

Thank you for your help and I'll warn the postman that there's going to be an increase in my mail!

Eleanor Weetch
Society Dietitian



see page 26 for further details

Fate Special Foods recipes

FATE PAN BREAD

Hello,
This recipe is more of a recipe suggestion really, and came about because of a recent family holiday to Grand Canaria. Travelling very heavily, weighed down with more Low Protein food than clothes, we stayed in self-catering accommodation.

I did not want to spend a lot of time in the kitchen on holiday, but also didn't want the girls to live on potatoes and salad for a fortnight! So we planned to eat in about half of the holiday.

Our main priority was the Fate All-Purpose Mix, the trusted bread maker and the necessary electric socket adaptor, so that we could make a fresh loaf every day. As you know, with making Fate bread in a machine, there is always some bread mixture over, with which I had planned to bake rolls, or make pittas, or even pizzas.

Well, some self-catering apartments have good cooking facilities, and others not so good. Guess which category ours came under? With just two small electric hotplates, and a fridge with an ice box that didn't freeze well, all my ideas of rolls, pittas and pizzas soon disappeared.

So, what to do with the left over bread mixture after putting the bread maker on?
Take a frying pan, a saucepan lid and a good helping of sunshine...

TO MAKE FATE PAN BREAD...

- **Just make the Fate recipe for machine bread and put it to bake.**
- **Then, take half of the remaining mixture, form it into a kind of half frying pan shape, just as you would a bread roll.**
- **Then do the same with the rest.**
- **Put both pieces into the pan, (as it was a non-stick pan, I didn't grease it), cover with a lid.**
- **Place into a sunny spot to prove till about double in size. (Depending on where you are in the world, this could take 5 minutes or 5 hours, but in our case it was 10 minutes.**
- **Then, put it onto a very, very low heat, still with the lid on the pan.**
- **Leave for 5 minutes, then take a wide spatula and turn each piece over.**
- **Replace the lid, and leave for about another 5 minutes to cook through.**

I'm not kidding – they were lovely! (Come to think of it, probably not the first time, because I'd put them on too high a heat and they were cooked too much), but after that they were fantastic. They were very light, soft and I thought, a cross between a bread roll and an old fashioned English muffin.

This recipe may be useful when cooking facilities are not at their best, or for families who like camping. I imagine it could be done easily on a camping gas stove. A great way to always have fresh low protein bread.

Don't forget pan bread can be made anytime or anywhere when you don't have a proper oven or bread maker machine. Just make the Fate bread recipe in the usual way and cook it in a pan.

All it took was a couple of minutes to get ready. A few times I didn't even prove them straight away. If we were going out, I put the pan with the bread and the lid into the fridge until I came back later that day.

Don't forget, details of making Fate bread in a machine are in each pack of Fate Low Protein All-Purpose Mix, available on prescription.

If you would like more details about cooking with bread machines, contact FATE SPECIAL FOODS help-line on 01215 224434. I will be happy to help you.

Eileen Green
Fate Special Foods

STOP PRESS – IMPORTANT NOTICE FROM THE NSPKU 2003 CONFERENCE ORGANISER

Please note that the NSPKU are hosting 2 separate events in 2003, which is different in format to our normal NSPKU Annual Conference.

The Annual General Meeting (AGM)

The Annual General Meeting (AGM) takes place on (Saturday) 15th March 2003, for one day only at the Hanover International Hotel in Hinckley, Leicester. Full booking details are shown in this edition of News & Views.

The European Society of Phenylketonuria (ESPKU)

The European Society of Phenylketonuria (ESPKU) will take place on (Friday) 31st October through to (Sunday) 2nd November 2003 also at the Hanover International Hotel in Hickley, Leicester.

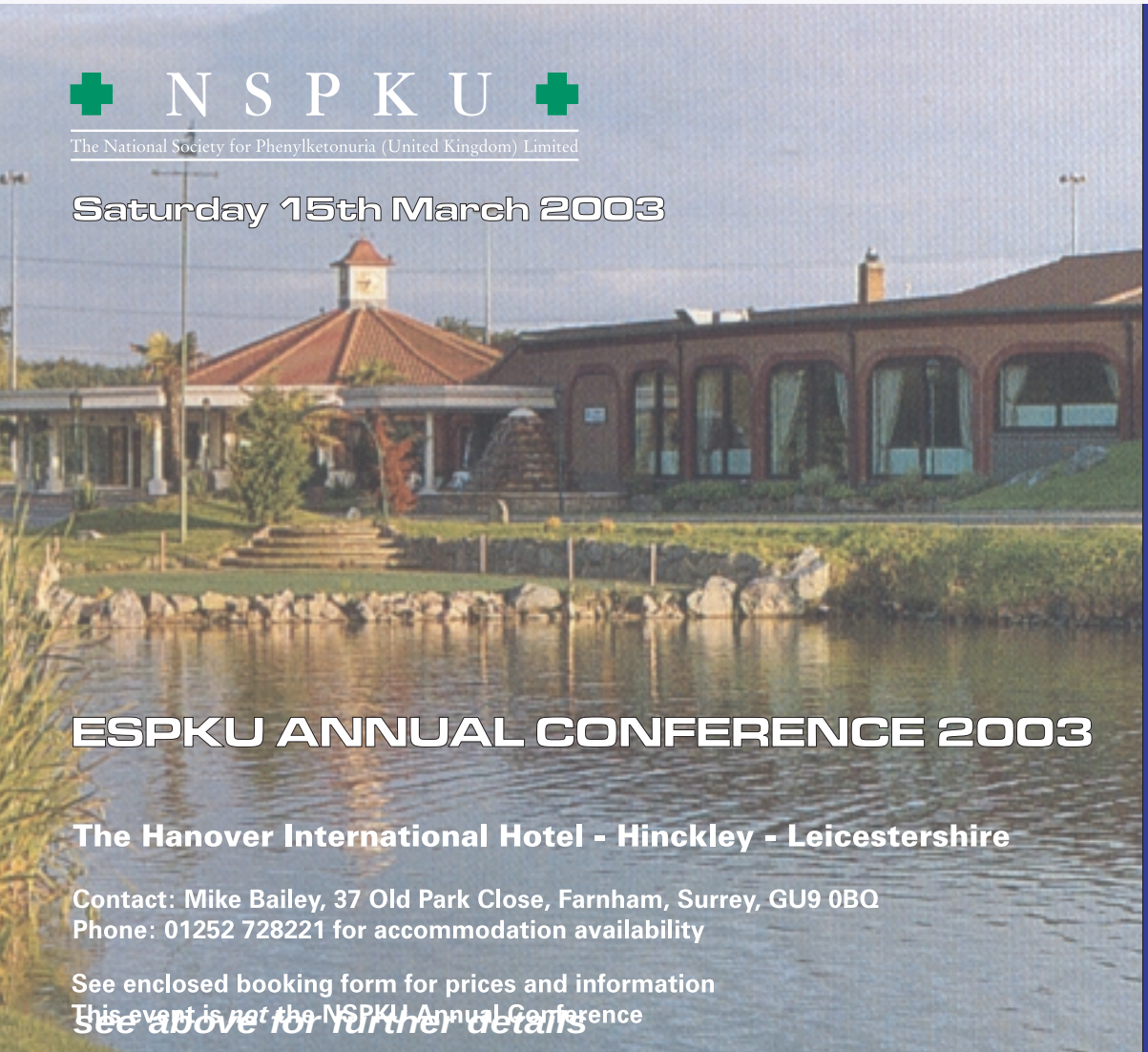
This event is very similar in structure to our normal NSPKU Annual Conference and will attract European PKU families and medical professionals.

The conference will start on Friday morning and finish Sunday lunchtime.

Full booking details will follow in the next edition of News & Views. Please do make a diary note of the dates.

Should anyone still have any questions relating to either event then please do telephone Mike Bailey on (01252) 728221.

See you all there.



ESPKU CONFERENCE 2003

WANTED

YOUR SOCIETY NEEDS

!YOU!

NSPKU Direct has been temporarily suspended due to lack of resources to manage the tasks involved. CAN YOU HELP????????

As a voluntary organisation it is sometimes hard to juggle working life, family life and the organising of an ever-growing dependent organisation. We care about the quality of service delivery that we currently offer to our membership and are ever-mindful of the costs on time and finances that this imposes. Sometimes we can't "keep all of the balls in the air". It just isn't possible.
This is where you might fit in. Merchandising is a valuable source of income for the Society. Finances that we require to deliver the level of service that we do. You will need a couple of hours of free-time every week, be contactable by phone and ideally e-mail (not essential) and have plenty of space (a spare room) to store merchandise. You must be physically able to move light to heavy loads around (or have someone who can do this for you). You should be able to keep a log book of all sales and be able to monitor stocks and the need to re-plenish them when required. The Society will re-imburse you with incurred expenses and will guide you through the initial months of start-up. If you have an interest in this voluntary position please contact Mrs Sara Bartlett on 01476 860379 for further details.

Variety for Life from SHS

Biscuits
Breakfast Cereals
Breads
Egg Replacers
Milk Replacers
Mixes
Pasta

SHS continues to combine innovation with the latest research to design state of the art formulae for the nutritional management of PKU.

The SHS Loprofin and Juvela Low Protein product range offers a choice of tasty, convenient phenylalanine free protein foods and snacks designed for ease of use to people on a PKU diet.

- **Comprehensive product range**
- **Continued excellence in customer service**
- **40 years of experience in diet and PKU**

Contact the SHS Advice Line on 0151 228 1992
or visit our website at www.shsweb.co.uk

SHS Loprofin Low Protein

Juvela Low Protein

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