# The British Porphyria Association newsletter

Registered Charity No.1089609

ISSUE 13 • **DEC 2006**

# Oh no! Not another conference!

Since the last newsletter came out, John and I (Kirstine Chamberlayne) seem to have spent far too much time at meetings and conferences. However, they were all interesting, for different reasons. I have just noted a few points, or a report would take several newsletters!

**Open Day at Cardiff**, on June 3rd. As you would expect from the programme, it was a very good day. The photos of the effects of the skin porphyrias, shown by Prof. Elder, were a bit of an eye-opener to those of us associated with acute porphyrias. We heard about DNA analysis and then saw the labs where this is done, as well as some of the tests for the type of porphyria – very interesting. Dr. Badminton and Prof. Elder were very helpful answering questions from members.

**Committee meeting**, at Buckden near Huntingdon, 17th June, with a talk afterwards by Prof. Cox from Addenbrookes, on the acute porphyrias. Members were glad to ask questions and share their own experiences with an expert. Prof. Cox is hoping to organise a dinner to raise funds for the BPA, probably in 2007.

**Porphyria Interest Group** on July 4th in Birmingham was for medical specialists and lab staff working with the porphyrias. John advertised our two published leaflets and gave notice of the coming publication of others.

Another speaker was Prof. Deybach from Paris, who is the Director of the European Porphyria Initiative. He gave the results of a trial on Enzyme Replacement Therapy for acute attacks, which proved ineffective, unfortunately. There was also a talk about liver transplants for those having repeated acute attacks which don't respond to haem. This is obviously not the easy option but, if successful, it can be life transforming.

**Climb Conference** on the 30th September, again in Birmingham. This was a 25th Birthday celebration, as well as a look at how things have progressed in the treatment of metabolic diseases. Family support was virtually non-existent 25 years ago (which was why Climb was started) and gigantic strides in genetics and molecular biology make it easier for effective treatments to be developed. Many inherited diseases were covered, many considerably more rare than porphyria. Prof. Cox gave short talks on Enzyme Replacement Therapy, and Gene Therapy. His talk made it clear that testing new treatments can be very dangerous. This makes animal experiments necessary, unless we are prepared to risk what little health an affected person has.

**Genetic Interest Group AGM and Conference** was in central London, on October 17th. (BPA is a member of GIG.) Two speakers were from the Department of Health. One talked about the need for health literacy to help keep NHS costs down, and how the government was trying to get patients more involved. The other talked about extra funding to reduce times for DNA testing (Cardiff was on the list of labs getting extra cash). The final talk was by a Professor involved in training doctors, about getting them to “think genetic”. It was good to know that something is being done about doctors overlooking genetic disorders!

### International Porphyria Patients Conference in Rome!

This deserves more than a few lines, so a report will be in the next newsletter. However, one thing stuck out **– *problems with porphyria are not exclusive to the UK, especially slow diagnosis!***

## BPA's AGM

### in Sidcup, on October 14th

We have a change of treasurer - Anne Newton has stepped down. She organised our first audits, and has kept the committee well informed of the financial situation. Alan Molyneux, the new treasurer, has been a committee member for the last year, and a willing helper.

Also, Karen Harris finally decided she needed a break from running the Helpline. So the number for the Helpline will change and the job will be shared by the committee members, for the time being. We will let you know the new number when it is set up.

Many thanks to Karen and Anne. The rest of the committee remains the same.

After the meeting, we had a talk by Dr. Robert Sarkany (St Thomas' Hospital) on EPP. He caused some amusement with an cartoon showing the effect of a faulty enzyme as a traffic jam! He gave a very clear account of why EPP is rather different from the other skin porphyrias and what can be done to treat it. He was happy to answer questions from members.

*Buffet and general discussion after our AGM on Oct. 14th. Dr. Sarkany (our guest speaker) is on the left at the far end.*

**The BPA committee are very grateful to all the porphyria experts who have given talks for us over the past year. They have all very kindly answered questions, and these have not always been easy.**

***Many thanks to you all.***

# EXPERIENCE OF AIP By Frances Kelton

My AIP occurred about the time I was a young college student studying for a diploma in Applied Biology in the early 1970's.

Feeling ill did not surprise me as I had suffered from severe asthma and severe eczema.

As I was afraid to lose my place at college I soldiered on.

I had been taking Tedral tables for asthma, which contained a barbiturate to counteract an unwanted effect of ephedrine contained in the tablet. Also I had taken a few of my mother's sleeping pills to get some sleep before my exams. These were most probably the two chief precipitating factors of my attack.

Consequently I experienced acute abdominal pain, dark red urine, (it was almost black at one point), and was extremely nervous. The nervousness was so bad I needed pin-drop silence and so was afraid of being admitted to hospital.

My hair was falling out in big bunches. My back and limbs ached. The last symptom to appear was digital extensor paralysis of my fingers except the small finger on my left hand. I was grossly underweight. It was all hell. Doctors were baffled.

Symptoms fluctuated. No one examined me when the abdominal pain was at its worst and I was thrashing about in the bed. I was variously believed to have hysteria, anorexia, appendicitis and even Hodgkin's disease. My appendix was removed and biopsies taken. The results of an EMG indicated a neurological abnormality.

Then my father advised my urine be tested for AIP, as his brother had been extremely ill with this. The first two times the result was negative. Mother pleaded with hospital staff to do the test again. The result of the third test was positive. Mother and I shared a private joke. We would say I had gone 'APE' again when I was ill -'A' standing for asthma, the 'P' for porphyria and the 'E' for eczema.

It took me several years to recover from my attack. Essentially, it cleared on its own, once I knew what substances to avoid and started eating more. Eventually I worked as a medical technician for a few years.

My main health problem since my attack has been severe eczema for which I have had about nine hundred injections ACTH (a powerful hormone) spread over twelve years. These days I have a son at university. My porphyria does not interfere much with my life.

## A Happy Christmas and a Healthy New Year to all our members!

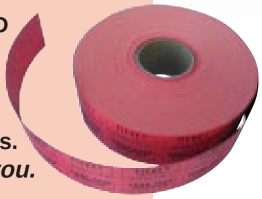
## ALEXANDRA ROSE DAY RAFFLE

Over the summer we sent you all a book of Alexandra Rose Day raffle tickets. We pay them just 20% of the ticket sales, whilst keeping 80% for our funds. Thanks to all of you who bought or sold tickets.

**Sales totalled £532.50. So we paid 20% to ARD (£106.50), and kept for BPA a profit of £426. Many people also sent in donations which totalled £107.50.**

**A grand total of £533.50 was added to our funds.**

***Thankyou.***



**If you move house, or change any other contact details, please let us know.**

With each mailing we send out, we always get a few back with "addressee unknown"!

Let us know by phone or email (see back pages for these)
or write to BPA, 136 Devonshire Rd., Durham City, DH1 2BL.

## MANY THANKS...

To Mr Leigh the head teacher and all the pupils at Lingham Primary School, Town Head Lane, Moreton, Wirral, Merseyside, for their **kind donation of £300. They chose the BPA for their charity for the year 2005 - 2006.**

Thanks also to Hayley Morrison and her daughter, Skye, who has A.I.P., and is a pupil at the school. Skye was presented with the cheque at the end of the summer term.

## LEAFLETS

We are having 3 more leaflets printed. We hope to have them ready to send out with this newsletter.

They cover drugs and porphyria; AIP; and skin porphyria. We will be sending you two of each of the ones that are relevant to your type of porphyria.

Do pass round our leaflets to your medical contacts. The more that know of porphyria the better. If you want more of these leaflets, or the two introductory ones that we sent out earlier in the year, please let us know, and we will be pleased to send you more.

**The leaflets can also be downloaded from our web-site: www.porphyia.org.uk, on the news page.**

## HELP WANTED:

**Can anyone suggest a celebrity patron we might approach, to help with porphyria awareness?**

**We would prefer not to use a politician, as they can provoke an adverse reaction.**

**Any ideas?**

## DATES FOR YOUR DIARY

**February's open committee meeting** is in Reading, probably on 17th (speaker willing!)

**Open Day at Kings College Hospital**, London, Saturday 2nd June.

**June's open committee meeting** is in Durham, probably on 16th (again, speaker willing!)

**AGM in October in Cardiff**, details to be arranged.

# LIVING WITH EPP - Sean's story

**Even today, my memory goes back as far as my first experience with porphyria, as it was a painful one.** I was in Spain, on a family holiday. My parents noticed me fidgeting and hiding in the shade. My legs, face and hands were on fire. The only relief from pain was a temporary one when I was lying in a cold bath. As soon as I came out of the cold water, the pain came back.

I was up and down all night, in and out of the bath. The next day, the swelling of the skin began. My hands, face and legs doubled in size. This was continuous throughout my life – every summer on a couple of occasions, my skin would burst open.

The worst attack was last year when I became 40. I was at the races and it was red hot. I was exposed for about 2 hours. The pain was so intense I had to grind my teeth – it was agony. When I woke up the next day, I resembled elephant man. My eyes were shut and my forehead was really swollen. Under my fingernails were boiling.

Throughout my teenage years were the worst, as when my friends would knock for me, I could never come out. Nobody understood. Although I've come to terms with it, nothing seems to help, just one simple rule. Don't go out in the sun.

## Presents to buy?

If you still have presents to get and are happy to buy on-line, why not use

**www.Buy.at/BPA ?**

**Typically 5% of the purchase price comes to BPA.**

If you have not visited the web site, please take some time to have a look.

You can go direct to www.buy.at/BPA or via our web-site www.porphyia.org.uk from the bottom of the front page.

HMV have recently joined buy.at, and give some excellent offers. There are still famous names like M & S, Sky, John Lewis, Littlewoods, Early Learning Centre and Amazon.

**Elane Retford and Andrew MacDonalds got married on 14th October.**

Elaine's mother has AIP, and is one of our members. They asked that guests donate to charity instead of buying wedding presents, **The BPA received £50 as a result of this.**

A wonderful day was had by all who attended, the weather was kind and although it was a double wedding as Elane's friend Sarah Snook married Henk Post, the ceremonies and merriment didn't falter!!

# HELPLINE

We are considering moving our help-line number to a free-phone number, but for the time being it remains as

**T: 01474 369 231**

Would members please note that our helpline is not manned at specific times, due to work and family commitments.

When it is not manned, an answer machine will be in operation.

Please leave name, phone number, day and time of message and someone will call you back as soon as possible.

**The BPA Newsletter is published by the BPA twice yearly.**

**Chairman:** John Chamberlayne
**Treasurer:** Alan Molyneux
**Secretary:** Sarah Pepperdine
**Patrons:** Prof.Timothy Cox, Prof.George Elder

www.porphyria.org.uk
E-mail: helpline@porphyria.org.uk

*View points and opinions contained in this newsletter are reproduced in good faith and do not necessarily reflect the judgement of the British Porphyria Association or its patrons*

# Trust Fund for Helen Gibbs.

**Helen Gibbs died on 11th July 2006 aged 26 in the intensive care unit of the Queen Elizabeth Hospital Birmingham.**

She had been diagnosed as having acute intermittent porphyria at the age of 16 and had had numerous and regular stays in hospital. She had fought back three times after becoming paralysed.

She was a strong and determined person and despite her illness managed to attain a degree in nursing and after training was a staff nurse in the cardio thoracic intensive care unit of the Coventry Hospital.

After a long period in intensive care in 2005 and suffering paralysis she managed to go home for about 2 weeks before having yet another attack. She again became very ill and in April 2006 she underwent a liver transplant but never recovered from this and died from multiple organ failure and sepsis in July. Throughout her life Helen was always positive and outgoing, and had a love of living that was infectious. All those that came into contact with her found her an inspiration and loved her dearly.

The BPA have sent our sincere condolences to her family and friends. During Helen's funeral over £1500 was collected for the BPA. The family wish to raise further funds in her name, and we are setting up a special Helen Gibbs fund within the BPA.

**If you belong to a social group why not hold a fund raising event in aid of the British Porphyria Association. We will be able to help you by providing flyers, sponsor forms etc. Call our help line for assistance on 01474 369231.**

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**British Porphyria Association** Registered Charity No.1089609

## MEMBERSHIP / DONATION FORM & STANDING ORDER

By making a donation to the British Porphyria Association you will know that you are making a vital contribution our work. Simply making a small donation will help us raise awareness, expand our help line or support our research.

**DO YOU PAY UK TAX?**
If you do, you can add 28% to the value of your gift to the British Porphyria Association at no extra cost to yourself. Simply sign and date the Gift Aid declaration and for every £1 you give we can ask the Inland revenue to give us an extra 28p.

TITLE: NAME:
ADDRESS:

POSTCODE:
EMAIL:
TELEPHONE:
TYPE OF PORPHYRIA:
DATE:

☐ I would like to pay my annual membership fee of £10
☐ I would like to make a donation of:
☐ £10 ☐ £20 ☐ £30 ☐ £40 ☐ £50 ☐ other £
☐ I would like to pay by Standing Order (please fill in the form opposite)
☐ I enclose a cheque made payable to the 'British Porphyria Association' for £

**Please send this form to:** The Treasurer, British Porphyria Association, 599 Bolton Road, Aspull, Wigan, Lancs, WN2 1PZ.

A monthly standing order or any donation you can give will make a difference.

BANK/BUILDING SOCIETY NAME:
BRANCH ADDRESS:
POSTCODE:

Please pay the British Porphyria Association the sum of £
each month/quarter/year (delete as appropriate) from my account
until further notice.

ACCOUNT NAME(S):
SORT CODE: ACCOUNT NO:
STARTING ON* (DATE):
*This date must be more than one month after today's date
SIGNED: TODAYS DATE:

This cancels all existing Standing Orders to the British Porphyria Association
(please tick) ☐ yes ☐ no ☐ not applicable

**GIFT AID DECLARATION**
I am a UK Tax payer and would like the British Porphyria Association (Registered Charity No.1089609) to treat all donations I have made since 6th April 2000, and any future donations, as Gift Aid donations until I notify you otherwise. (we can reclaim the tax on your donations at no cost to yourself)

DATE: SIGNATURE:

DESIGNED BY: KAW Design. www.kawdesign.net