The
British Porphyria Association newsletter

Registered Charity No.1089609

ISSUE 14 • **MAY 2007**

Open Day at Kings College Hospital!

We are pleased that we are having another Porphyria Open Day this year, organised by Dr Marsden. It will be held at **King's College Hospital, Denmark Hill, London SE5 on Saturday 2nd June 2007.** We are starting at 10.30, with talks in the morning. As well as Dr Marsden, we will have talks by Dr Sarkany and Dr Rees. These will be on the various types of porphyria and their testing. Following lunch (free!) we will have a question and answer session, and then a tour of the labs for BPA members. We aim to finish at 4pm.

Previous open days have proved to be very popular, so do come along. If you are hoping to come, please let the BPA know and we will send full details of how to get there.

Contact details for the BPA are on the back.

HELEN GIBBS BALL

May Ball in Memory of Helen Rosemary Gibbs on Monday 28th May at 5pm. The venue will be Moreton Hall, Moreton Morrell, Nr Warwick, Warwickshire. Dress code Lounge suits with a 'pink twist' Tickets are £25 and all proceeds to The Helen Gibbs Fund (The British Porphyria Association)
Ticket enquiries to **allanandjoymay@hotmail.com**

We have recently received a cheque for £1247 for the Helen Gibbs fund, from a charity night organised last summer, by two friends of Helen. The BPA sends many thanks for all they are doing.

Calling all Skin Porphyria members

Do you have problems with fluorescent lights and low energy light bulbs? It has been brought to our attention that some EPP sufferers in particular do find this affects them. If you do too, you may be interested to learn that the government's plan to force us all to use fluorescent bulbs has prompted an online petition. If you disagree with these plans and want to support light sensitive sufferers please sign the petition, at **http://petitions.pm.gov.uk/righttolight**

Thank you!

OPEN COMMITTEE MEETING

If you live in the north of England or Scotland, then a more local event is our open committee meeting in June. It will be in the **City of Durham** on **Saturday June 23rd,** from 2.30pm. Please come along, and meet the committee and others with porphyria.

We will be providing tea, and there will be an evening social event that day as well. Please let us know if you hope to come.

MEET YOUR COMMITTEE...

We are giving you here brief pen portraits of many of the members of the BPA committee. They all give help to the BPA in what spare time they have. Do come and meet us in person some time at one of our meetings.

John Chamberlayne, *Chairman, and manager of web-site.* married to Kirstine, *minutes secretary.*
We are in our early 60s, with two grown-up daughters. Our elder daughter has been in hospital several times in the past 10 years with attacks of Variegate Porphyria. She inherited this from John and his mother. Neither have had severe attacks, so only found we had porphyria after our daughter was diagnosed. We are retired. Prior to that John was in a technical role in a factory making TV tubes and Kirstine was a biology teacher in technical colleges. We live in Durham.

John and Kirstine Chamberlayne are chairman and minutes secretary of the BPA

Alan Molyneux, *Treasurer.*
I am 56yrs old, married to Sharon and have one son, Daniel, 29 yrs old and one daughter, Wendy 27 yrs old. Both Wendy and I have AIP. I have never had any attacks since 1969, Wendy has never had an attack at all. I am a builder, and have been in the trade for 40 yrs. We live in Wigan, Lancashire. Sharon and I come from Bolton, Lancashire originally. I have a very large family, 54 + at the last count. There are a few with AIP.

Karen Harris, *Help-line.*
I am proud to have been involved with the BPA since its inception back in 1998/9. I am a trustee and past Chairman. I now take more of a 'backseat', as I have several commitments which have had to take priority. I currently run the helpline answering calls from patients, family members, doctors, nurses, journalists etc, etc. I have Variegate Porphyria and have had various symptoms on and off over the last 30 years, however, remain fairly healthy most of the time and would be considered as having a 'latent' condition now. (No major active symptoms).

Sarah Pepperdine, *correspondence Secretary.*
I was lucky as I was diagnosed with Erythropoietic Protoporphyria (EPP) quite early on when I was 4 years old. I joined the BPA committee after attending a meeting 3 years ago. I become the secretary a couple of years ago. I didn't understand about Porphyria and all the other types until getting involved with the BPA and up until last year I hadn't ever met another EPP sufferer. It was so good to chat to other people who completely understand your condition, the BPA is great for getting people together who can share their experiences and offer support to each other as well as the medical knowledge. I'm 37, separated with two children, Ryan is 15 and Eloise is 7. As my condition isn't genetically dominant my children luckily haven't inherited EPP.

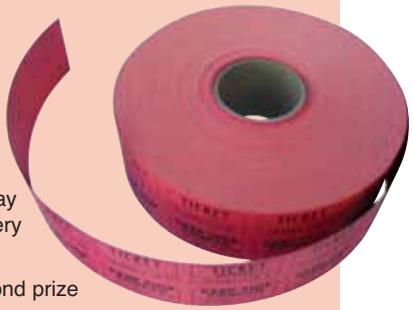
Sarah Pepperdine is correspondence secretary of the BPA

Elizabeth Critchley. I became a trustee for the BPA in 2004 after being involved with the society on a more casual basis since its inception around eight years ago. I suffer from Acute Intermittent Porphyria and have had recurrent attacks for the past 11 years - my first one being at the age of 19. I have regular Haem arginate treatments which help to keep the worst of the attacks at bay and allow me to maintain a relatively normal lifestyle. I try to help out with the BPA wherever possible as I feel it is important to maintain contact with fellow sufferers who often feel isolated and keep up to date with the latest research.

Anne & Keith Newton. We became involved with the BPA in its early days following daughter Elizabeth's diagnosis with AIP. Family tests confirmed that Elizabeth's sister, Suzanne, and brother John, also carry the gene. However, to date, they have been symptom free, helped I am sure by the knowledge that they carry the gene and are proactive in avoiding the triggers. The need for patients and families to become knowledgeable about their condition and have contact with other sufferers, together with up to date information on treatments and research is important. The BPA provides this link and is the reason we first became involved.

Anne has been a Trustee of the Association since it achieved charitable status and carried out the role of Treasurer for two years, stepping down at the AGM last October due to personal and family commitments. However, both Anne and Keith aim to continue to be involved and help out wherever possible to ensure that the BPA continues to flourish.

Alexandra Rose Day Joint Charities Autumn Raffle



Once again we will be taking part in the Alexandra Rose Day joint charities autumn raffle. During the summer we will be sending out books of raffle tickets to each member, which we hope you will be able to sell to family, friends and work colleagues. This is a great way for us to raise extra funds, without the need for us to apply for a lottery license, so we hope that as many of you as possible will support it.

There are a number of prizes, the first prize being £2000 cash, Second prize of £1000 cash, Third prize of £500 cash and several runners up prizes.

Tickets will be 50p each with 5 tickets in a book. The BPA will keep 80% of the cash from our ticket sales, sending 20% to Alexandra Rose Day for administration costs and prizes.

Good luck to everyone who buys a ticket!

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 or write to us (see back page for these).

1st International Meeting of Porphyria Patients, in Rome

26th & 27th October, 2006.

This was run by our Italian counterparts, AMaPo, with the help of Prof.Biolcati, Director of the Rome Centre for Porphyria. There were talks on various aspects of porphyria, very like one of our Open Days, given by experts from several countries, as well as talks from a number of patient groups including our own. The conference lasted a day and a half, so it is only possible to give you a summary. Talks were in English or Italian, and the conference room was provided with simultaneous translation, through portable head-sets. This worked very well most of the time, though one of the translators managed to get on the wrong channel at one point! We were able to ask questions or make comments after every couple of talks, and the breaks and lunch times also gave us valuable time to talk to the experts and to other patient representatives.

Points of interest from the talks.

There is a real need for faster diagnosis, in all countries. Porphyrias are not included in standard lab tests and symptoms are missed. For example, one patient who died of an **acute** porphyria had red urine (ignored), and was given surgery, then psychiatric treatment. She lapsed into a coma and was transferred to ICU who finally diagnosed the porphyria. She was given Normosang, but died. Jasmine (who attended the conference) finally found out she had **EPP** at the age of 26, by searching the internet, then insisting her doctor had the tests done. Others have gone 35 years before diagnosis.

Finding carriers who have no symptoms was also regarded as very important (DNA testing to find the gene mutation, and then testing relatives). Finding the gene mutation is not always possible and success rates vary between countries, which may be caused by technical difficulties or by regional variations in the gene mutations.

With DNA testing, those with an **acute porphyria** would then know the triggers to avoid and have a better chance of staying healthy. This was emphasised by Regina, from Hungary, whose cousin failed to inform her that she had been diagnosed with AIP. As a result Regina had a very severe attack triggered by the wrong drugs, and has attacks about four times a year. Her story also showed how often those with AIP are treated as psychiatric cases. This can also happen to those with EPP, and other porphyrics who have no obvious skin symptoms.

It is also important to find **carriers of non-acute** porphyrias so that they can be advised of the risks to their families.

DNA Testing varies a lot between countries. It is funded in most of the EU, but the USA only has some testing as part of a study.

European Porphyria Initiative is attempting to develop a common approach, with standardised diagnosis and tests for the porphyrias. A standard drugs list is needed for acute porphyrias, with proper assessment of unsafe drugs so the risks are understood when there is no alternative treatment for another condition. If plenty of countries co-operate, there will be enough patients to give good answers. EPNET has asked for a grant to set up a registry of centres for lab work and diagnosis, in which Cardiff, at least, would be included. The hope is to have a Europe-wide organisation for patients.

Acute porphyrias

VP and HCP. High numbers of VP are found in Chile, as well as South Africa, through a similar founder effect (with a different mutation). A few individuals have been found with 2 VP genes (homozygous). All have had skin symptoms, but none have had acute attacks. Active HCP usually causes acute attacks but not skin symptoms, but homozygous individuals get skin symptoms and kidney problems. Very odd!

AIP. Less than 10% get symptoms, and it may be as little as 2%, as carriers are easily overlooked. Pain was thought to be caused by ALA but, if ALA is taken alone, there is no pain. Haem arginate normally removes pain in 2 to 3 days, with ALA and PBG back to normal. All "acutes" have a risk of liver carcinoma, and should be tested periodically. Tests on cholesterol show no problems. Often the "good" HDLs are raised.

Skin porphyrias

EPP is more common in Japan than Europe, but is unknown in Africa, because the numbers of people with the second (low expression) gene which causes it are high in Japan, but very low in Africa. It has now been shown that a second low-expression gene is also involved in HCP, but not in AIP or VP. (There has been no study of familial PCT, the other dominant gene). However this doesn't rule out the possible influence of other types of gene.

EPP treatment using frequent brief exposure to light to stimulate production of melanin (tanning) is the most successful method, short of excluding light completely. However there is a new drug on test. Liver problems are common, but there is no agreement on the best tests for routine checks, probably urine tests plus ultrasound. It is still not known why some women have no symptoms during pregnancy. One patient reported no problems travelling in India and Egypt, but it got worse in Norway. The answer was not known, but there could be differences in the spectrum. Some people with **EPP** have problems with low energy lights (more UV/purple light).

PCT is the most common porphyria throughout the world. About 25% of them have the familial type, which affects all body cells, not just the liver. HEP is caused by 2 copies of the faulty gene, giving problems earlier in life and sometimes mutilation.

In 1955 in Turkey, a spillage of herbicide caused an outbreak of PCT, so environmental factors are likely to be important in triggering symptoms. About 80% of PCT is associated with Hepatitis B virus. Unfortunately the standard treatment of interferon plus ribavirin cannot be used for porphyria. There are new treatments under test. Vaccines are far more effective, so anyone who could develop liver problems should have them.

Answers to patient's queries

Smoking can increase cirrhosis, so it is not advisable for anyone who could develop liver problems. Anti-inflammatory drugs are also bad for **PCT**, as is getting tired. Someone commented that in Italy, **PCT** is thought to cause tiredness, as well as skin symptoms.

Someone with **EPP** asked about using anti-inflammatory drugs for arthritis and was told it should be safe, but to take regular liver function tests. (Liver problems from EPP can sometimes be treated successfully if caught soon enough.)

This was a very useful conference though the best part, for us, was talking to people from other countries and sharing experiences. At the end, several of us got together and decided we needed an internet forum, to share problems, possible solutions and perhaps co-ordinate action such as publicity. The idea of a National Porphyria Awareness Day seemed to appeal to many people, so we may get other organisations joining us on June 1st.

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.

Web: **www.porphyria.org.uk**

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

Address: **BPA, 136 Devonshire Rd Durham City, DH1 2BL.**

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

Alan's Story

In 1969, when I was 19, I had terrible stomach pains at work. My doctor prescribed laxatives, which were useless. After a week of pains my mother persuaded him to admit me into hospital, as she had had a burst appendix as a child.

In the hospital I was left for a week with the pains until my appendix was finally taken out. But to no avail. The pains continued. At this point I'd been given anaesthetics, antibiotics and even sleeping tablets every night. Slowly I became weaker, until I was unable to move any of my limbs. During this time I was not believed by any of the members of staff, even when I was unable to get up off the floor when I had fallen out of bed.

I even convinced myself that there was something wrong in my mind after they had a psychiatrist come and evaluate me. Suddenly after four weeks my consultant arrived at my bedside and informed me that he wanted to do a lumbar puncture on me, which involved inserting a long needle up my spine to extract fluid. Within two hours I was told that I had Porphyria. I remember a lot of fuss was made to get me ready to be shipped off to Manchester Royal Infirmary, where I stayed for the next five months.

Here I was believed at last, although the damage was already done, regarding all the drugs that I shouldn't have had. As it was early days in the treatment of Porphyria I was willingly used as a guinea pig for various tests to try and understand the condition. Eventually I got stronger, learnt how to walk again, regained a lot of muscle tissue, having gone from eleven down to seven stones, and came home.

Looking back all those years it was the worst episode of my life. I don't blame the first hospital for not knowing about Porphyria, as we know now it's quite a rare condition. It transpired months later that my first consultant just happened to mention my symptoms to a colleague in the hospital canteen over coffee. Thank goodness, this colleague, whom I have always owed my life to, knew of the symptoms of Porphyria.

British Porphyria Association Registered Charity No.1089609

MEMBERSHIP / DONATION FORM & STANDING ORDER

TITLE: NAME:

ADDRESS:

POSTCODE:

EMAIL:

TELEPHONE:

TYPE OF PORPHYRIA:

DATE:

WAYS TO PAY

- ☐ 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 £
- ☐ I would like a receipt

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.

Standing Order BRITISH PORPHYRIA ASSOCIATION
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: TODAY'S DATE:

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

BPA use only: Please pay to British Porphyria Association bank account:
20-43-63

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