

THE BRITISH PORPHYRIA ASSOCIATION NEWSLETTER



Registered Charity No. 1089609



NEW THERAPIES IN THE PORPHYRIAS: UPDATE FROM LUCERNE

The BPA recently attended the biennial International Congress of Porphyrins and Porphyrins, hosted in May this year by the Swiss Society of Clinical Chemistry in Lucerne, Switzerland. The scientific conference was followed by an International Meeting of Porphyria Patients, to which a number of BPA members attended.

The scientific conference gives an opportunity for international porphyria specialists and researchers to present and discuss new developments in the field. It is hugely reassuring to see how much research is taking place, nationally and internationally, into such rare disorders. Some of the more noteworthy research, that should hopefully generate new potential treatments for the management of patients with porphyria, includes the following:

Scenesse® (afamelanotide) for EPP

Scenesse, manufactured by Clinuvel Pharmaceuticals Ltd, is a small implant which stimulates melanocytes to produce and release melanin, the brown pigment in skin known for its photoprotective effect. The results of a Phase III, multi-centre, double-blind, randomised, placebo-controlled, trial were presented in Lucerne. Scenesse resulted in a more than three-fold increase in the time that patients with EPP could spend in direct sunlight without pain. This treatment could improve the quality of life for patients and is currently awaiting European Medicines Agency (EMA) approval for marketing. The BPA has joined with other European patient organisations to explain to the EMA how EPP patients can be very severely affected and that a new treatment would provide huge benefit to their quality of lives.

RNA therapy for porphyria

Pre-clinical data was also presented on the potential use of small interfering RNA (siRNA) therapy for the treatment of porphyria. Alnylam Pharmaceuticals in the USA showed through animal studies that porphyrin precursors ALA and PBG in plasma and ALAS1 mRNA can be suppressed in the livers of mice and rats. The next step is to identify a final development candidate by late 2013 and the aim is to progress to patient studies in 2014.

Gene therapy for AIP

While haem arginate currently exists as a preventative and responsive therapy for acute porphyria, the porphyrias have long been considered for gene therapy which could eliminate/reduce the need for other treatment. >

continued from page 1

The AIPGENE Consortium is a collaborative project that is looking into the use of a viral vector containing the PBG-deaminase gene for the treatment of AIP. Seven patients have been recruited to the Phase I Clinical Trial that will be completed in 2014. While many steps will need to be taken before conclusive evidence of the safety and efficacy of gene therapy is confirmed, it is very exciting to hear about these important advances.

Other interesting areas of study included:

- Susceptibility factors which may cause some people with a porphyria to suffer

more than others

- Clinical complications and ways to alleviate problems
- New clinical advances, methods of diagnosis, genetic mutations, therapies, and molecular studies
- Case reports of unusual/extraordinary events
- The incidence and prevalence of porphyrias in Europe

Patient day

The patient day was an extremely interesting day. There were summary talks on the latest research activities and clinical advances plus time for questions.

The day also provided patients with a chance to network with other patients, patient organisations and scientific experts from around the world.

Attendees were also encouraged to join the new international initiative, Rare Connect: www.rareconnect.org/en/community/porphyria. The Rare Connect community aims to collate international experiences, so that members can learn more about the porphyrias, to gain greater understanding. It also gives opportunities to meet (in a forum/blog setting) with other porphyria patients and with porphyria specialists from across the world.



RIDE LONDON-SURREY 100 – AUGUST 2013

The BPA would like to congratulate Ian Burrell and Glenn Blake for successfully completing the 'Ride London-Surrey 100' on Sunday 4 August 2013. Ian and Glenn completed it much quicker than anticipated and both thoroughly enjoyed being part of such an exciting event in the capital. We would like to thank the 'Norwich Boys' for all their hard work in raising not only awareness, but also over £1,250.00 for the BPA!

PROPOSED PORPHYRIA SPECIALIST REDUNDANCY - UPDATE

As many of you will already be aware, Cambridge University Hospitals NHS Foundation Trust (CUH) recently proposed changes to the porphyria service at Addenbrooke's Hospital, which would entail making Dr Penny Stein redundant.

Dr Stein treats some of the worst affected acute porphyria patients in the UK and is a vital part of the National Acute Porphyria Service (NAPS), which started last year. Dr Stein has considerable experience of dealing with the most severe cases, and has been providing excellent care for them. Very few specialists in the UK have that kind of experience. The BPA feels strongly that this

goes against all of the original aims of setting up NAPS and have been in correspondence with Addenbrooke's for a number of months, explaining our concerns about the proposed change. The service at Addenbrooke's is currently undergoing an external review and Dr Stein's contract has been extended until 31 December 2013.

We will keep you updated as to how this campaign is going.

If you would like to discuss this issue further, or feel you can help in any way, please do not hesitate to contact us. Our helpline number is 0300 30 200 30, or you can contact our email helpline via helpline@porphyria.org.uk.

WE NEED YOU! NEW BPA CHARITY PLACES IN SPORTING EVENTS IN 2014

The BPA are pleased to announce that we have secured two charity places in the **Ride London-Surrey 100**, to be held in London on Sunday 10 August 2014. In order to apply you need to be able to ride 100 miles in less than 9 hours, be confident in riding in a group and be able to raise around £500.00.

If you would be interested in competing in this amazing

sporting event (or any other event) on behalf of the BPA in 2014, please get in touch via: helpline@porphyria.org.uk or 0300 30 200 30. These places will go on a first come, first served basis so please get in touch as soon as possible – Good Luck!

For more information on the Prudential Ride London event, please see: www.prudentialridelondon.co.uk.



SPONSORED WALK

ALTHOUGH IT WAS HARD WORK, IT WAS ESSENTIAL THAT WE WERE ALL GOING TO BE FIT ENOUGH FOR THE CHALLENGE, AND IT CERTAINLY PAID OFF

On 18 May 2013 four of us set out at 5.30am, from Lewes in Sussex, to walk 26 miles in one day. Our destination was Beachy Head. It was an extremely challenging route taking us over the South Downs with the final seven miles over the Seven Sisters. Despite forecasts of rain, we were lucky to have clear, if somewhat crisp, skies on the day.

We started training at the end of February, battling through some dire weather, including hailstones, snow and frost. On one of our early training walks Wendy was bitten by a dog which had been left to run loose by its owners – despite this she continued with the training walk. Each week we increased our mileage and pushed our speeds. We had to deal with

blisters, twisted ankles, cattle and horses. Although it was hard work, it was essential that we were all going to be fit enough for the challenge, and it certainly paid off.

We completed the walk in a respectable 8 hours 40 minutes, coming 13th, 14th, 15th, and 16th out of 75 people. Friends, family, neighbours and work colleagues were generous sponsors and we raised around £1760 for the BPA.

For the four of us, Wendy, Candy, Sian and myself, it was a wonderful challenge and we created great bonds and a real sense of achievement.

The BPA would like to congratulate Karen, Wendy, Candy and Sian for all their hard work in their training, the event and in raising a significant amount for the BPA – thank you ladies!



KIM'S STORY (ACUTE INTERMITTENT PORPHYRIA – AIP)

I was diagnosed with AIP in 2002 and suffered terribly, resulting in constant attacks and lots of haem arginate treatments. My first attack left me unable to walk, talk or do anything for myself, as the nerve damage and muscle loss was so bad. I slowly recovered, but still had constant attacks and had one approximately every two to three months for the next three years. They then got more frequent and for the next few years, I had an attack every month or two lasting about two weeks.

All of these attacks resulted in hospital admissions to control them and for pain control. I was referred to Prof Cox at Addenbrooke's Hospital, who had lots of suggestions which my local hospital followed. The next year was even worse, where I rotated around being in

hospital for two weeks and then home for about two weeks. I kept visiting the porphyria clinic in Cambridge as an outpatient, as my local hospital was struggling to help me.

The year after, I was in hospital for 10 months, my porphyria was at its worst. I was in constant pain and a little crazy due to the high levels of porphyrins. I ended up being admitted to Addenbrooke's to see if they could help me, and as my porphyria was completely uncontrollable and was affecting my life so significantly (especially with regard to my two young children), a liver transplant was suggested as a last resort. The porphyria team decided that I could go on the transplant waiting list. I returned to my local hospital

where they continued treating my attacks and trying to control my pain the best they could. Then, in August 2011, everything got worse, my porphyrin levels were so high and my pain could not be controlled. I went blind and started having fits due to a rare complication. This resulted in me being in intensive care where the doctors did not expect me to survive. I pulled through, although I was told that the fits were caused by my brain swelling and it would most likely happen again – they gave me a few months to live.

The specialists asked for me to be transferred back to Addenbrooke's. Whilst there, I was pushed up the transplant list. I got a liver the next day and had my transplant in September 2011.

The day after my transplant, my porphyrin levels dropped from 504 to 0.2, which is normal for people without porphyria. I haven't had an attack since, which is fabulous and my children love having their mummy at home. Obviously, I still have AIP, as it is in my genes, but I won't suffer from it any more. I still get porphyria type pains when I am ill with anything, but it's just because the nerves are so accustomed to the pain that my brain is sending the wrong messages. I'm left with a lot of muscle weakness and nerve pain, but overall life is now good.

Liver transplant is only needed in rare circumstances, but can transform the lives of those with the very worst AIP and where other treatments have failed to make a difference.

HELPLINE

0300 30 200 30

Would members please note that our helpline is only manned at specific times, due to work and family commitments.

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

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

web: www.porphyrria.org.uk
email: helpline@porphyrria.org.uk
address: BPA, 136 Devonshire Rd
Durham City, DH1 2BL

The BPA Newsletter is published by the BPA twice yearly.

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BPA EVENTS

2013

In addition to the BPA attending the international patient day in May this year, we held two further patient events in London and Dundee.

On **Saturday 8 June**, the BPA held their annual **Open Day at King's College Hospital in London**. Feedback from this event was very positive and indicated that the content was interesting, informative and helpful. We had a good turn out and people seemed to like the balance between clinical and patient experience talks, as well as tours around the laboratories. We also received suggestions for future talks on pain management and diet, which we hope to incorporate into the events which are planned for next year.

Finally, the BPA held its **Autumn Conference and AGM at Ninewell's**

Hospital in Dundee on Saturday

21 September 2013. This event incorporated a brief AGM, clinical and patient experience talks and a tour of the laboratories. People seemed to appreciate the balance between these talks, but in the future we hope to allow more time for structured question and answer sessions.

2014

The BPA will be holding similar events in 2014. **Our Open Day will be held in Cardiff on 7 June 2014**, while our Autumn Conference and AGM will be held in Manchester (possibly Saturday 6 September 2014) – more details will be supplied early in 2014. If you are interested in attending either of these events please do let us know and we can then send you information closer to the time.



THE BRITISH PORPHYRIA ASSOCIATION Registered Charity No. 1089609

MEMBERSHIP / DONATION FORM & STANDING ORDER



Title Name
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Date

WAYS TO PAY

- I would like to pay my annual membership fee of £15
 I would like to make a donation of:
 £10 £15 £20 £25 £50 other £
 I would like to set up a standing order (please fill in the form opposite)
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 I enclose a cheque made payable to the 'British Porphyria Association' for £
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By making a donation to the British Porphyria Association you will know that you are making a vital contribution to our work. Simply making a small donation will help us raise awareness, man our helplines or support research.

Do you pay UK tax?

If you pay UK tax, the BPA can reclaim 25p of tax on every £1 you give. This does not cost you anything and does not affect your personal tax position. Simply sign and date the Gift Aid declaration.

Standing Order British Porphyria Association

Please send this form to: The Treasurer, British Porphyria Association, Rothlea House, 7-8 Quarry Lane, Butterknowle, Bishop Auckland, County Durham, DL13 5LL
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

Please pay to British Porphyria Association bank account:
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Gift aid declaration

I am a UK tax payer and would like the British Porphyria Association (Registered Charity no. 1089609) to treat all donations that I have made in the last six years and all future donations that I make from the date of this declaration as Gift Aid donations, until I notify you otherwise. I understand I must pay an amount of income tax and/or capital gains tax equal to the tax reclaimed on my donations. (I will advise the BPA if my tax status, name or address changes).

Date Signature