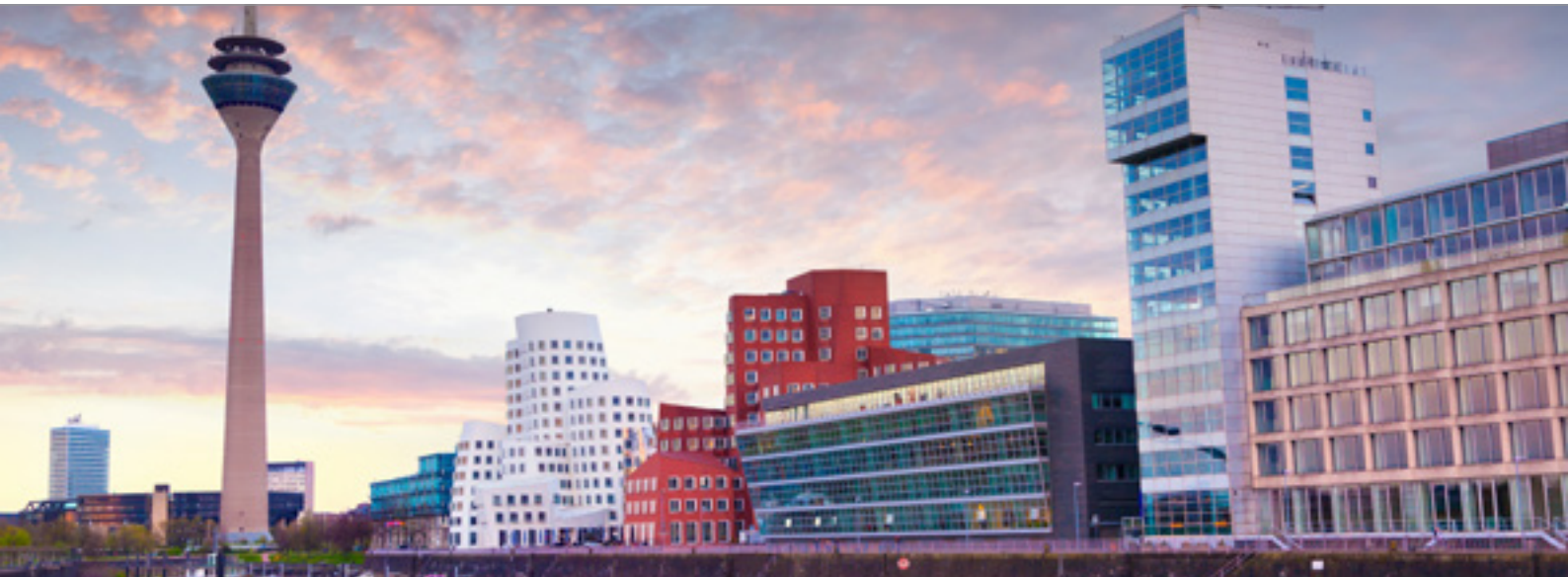


# THE BRITISH PORPHYRIA ASSOCIATION NEWSLETTER

Registered Charity No. 1089609



## INTERNATIONAL PORPHYRIA MEETINGS - GERMANY

The **International Congress on Porphyrins and Porphyrias (ICPP)**, which included an **International Patient Day**, was held in Dusseldorf, Germany between the 13 and 16 September 2016.

The **Patient Day**, was a great success with representatives attending from 18 countries from around the world, including Australia, Brazil, Russia and many European countries, as well as the BPA from the UK. The day was very well organised (with thanks to the German and Dutch patient organisations) and covered many aspects, but its main focus was on providing updates on new therapies in the cutaneous and acute porphyrias. There were many opportunities for informal discussions about how our different patient organisations have been developing over the last couple of years.

**International patient cooperation** – the heads of the international patient groups also met to discuss ways in which we could all work together, rather than simply convening once every two years. The BPA are happy to be part of this movement, as our collective voice for those with porphyria will be stronger when we are united together – we will

keep you updated as things develop. Patient cooperation is likely to be involved in some way with EPNET (the European Porphyria Network), as EPNET is hoping to apply to set up a European Porphyria Registry (EPR), as a European Reference Network. As part of this, they hope to obtain European funding to cover future international conferences and to improve care across Europe.

### The main ICPP was very interesting and included reports on:

- *New research on long-term use of Scenesse for EPP*: there were reassuringly few adverse effects.
- *Update on gene therapy for AIP*: results were poor, perhaps because it needed to reach more than 1/3 of liver cells.
- *Alnylam's update on a potential treatment for all "acutes"* (see page 4 for a condensed report).
- *Porphyria light factor for EPP*: a chart for the time of year and day, cloud condition, etc., to give some idea of relatively how long it may be possible to stay in the light (particularly for small children). They regarded blue light as worst, but that yellow and red were also

significant, as they penetrate the skin more. At present this is only for Norway, but it is hoped to be extended to other countries.

- *The Norwegian Porphyria Centre (NAPOS) Drug Database*: NAPOS have updated their list based on reactions as well as research on how drugs work. They have now managed to classify over 1,300 drugs; with the newer classifications and re-classifications, a greater proportion are considered safe. The Welsh Medicines Information Centre's (WMIC) SAFE drugs list obtains information from NAPOS for their list.
- *Continuing projects on DNA*: it is now thought that many more people carry the defective gene for AIP – about 100 times those with attacks. "Acutes" who have attacks often belong to known families, suggesting other inherited factors. We expect more analysis of DNA in 2017, as this field is developing so quickly.

We look forward to hearing more of these reports. The next ICPP is due to be held in Bordeaux in June 2017, as soon as we know more details we will keep you updated.

# WITH THANKS

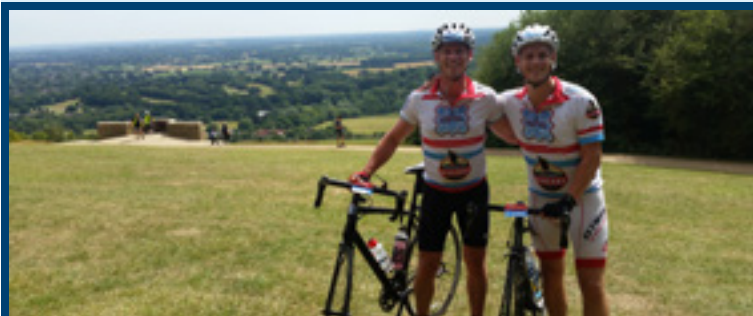
## GREAT NORTH RUN 2015

This is the first year that the BPA have had places in the Great North Run, but we are delighted to say that it was a great success! We would like to say a massive thank you to all our runners, Debra Baker (who raised over £1090), Craig Bell (who raised just

under £300), Emily Todd (who raised over £730 to include a generous donation of £200 from the Kier Group Plc.) and Laura Welch (who raised over £860). You all did amazingly well, despite hotter than expected weather! Between you, you managed to raise

over a whopping £2,975. THANK YOU AGAIN!

We would also like to extend a thank you to the Butterwick Hospice for their hospitality in letting the BPA use their refreshment tent – it was great to have a focal point for runners to meet up.



## RIDE LONDON-SURREY 2015

After our original cyclists for the Ride London event pulled out at the last minute, Dougal Betts and Richard Lee got roped in to helping us out! They were lucky to enjoy a much improved day weather wise after the torrential rain experienced in 2014. THANK YOU for stepping in and raising over a fantastic £790 For the BPA.

## SKEGNESS WEEKENDER

THANK YOU to Ray Lancaster and Jenny Coombs for their great efforts in running the Skegness Custom Van Show again. Despite being plagued by horrendous weather and a lack of volunteers to help put up the marquees in wild winds, they managed to raise a brilliant £332 for the BPA. You deserve a well-earned rest, we think!

## SONG BIRDS

The Parfitt family is continuing with their great fundraising efforts. This year Laura and her friends in the Red Rock singing group have managed to raise £260, after nominating the BPA as their charity of choice this year. MANY THANKS for your continued fundraising!

## IN MEMORY

The BPA would like to offer a sincere THANK YOU to a number of families who have chosen to collect donations in memory of their loved ones, due to the BPA being close to their and their family's hearts.

In memory of Valerie Woodhouse, her daughter Jayne collected a further £100 in memory of her mother who sadly passed away earlier this year.

Audrey Evans also sadly passed away earlier this year. In her memory her family chose to collect donations, raising over £40 for the BPA.

THANK YOU for thinking of the BPA at these difficult times.

## CONTINUED FUNDRAISING...

Much appreciation goes to Claire Creag and her family and friends for their continued efforts in fundraising for the BPA. In addition to selling merchandise for us, Claire has also been involved with roping in the Winton Bowling, Tennis and Social Club, and the Nags Head, both in Eccles, Manchester, into raising funds for us. Fantastic job, **THANK YOU!**

Chris Baker, we would also like to say **THANKS** to you for continuing the mission to raise funds for the light units. Keep it going, you're doing a brilliant job.

The Chapman family are also continuing their fundraising efforts, recently raising around £150 for the BPA – **THANK YOU** very much and keep up the great work.





# LORRAINE'S EPP STORY

My name is Lorraine and I suffer from EPP. At 43 I have learnt to live with my condition, as best I can, but I am one of those people who pushes themselves to the limit, sometimes I get away with it ... other times I don't!

It was in January that I decided to put myself forward to do the London Marathon to raise funds for the BPA. I was excited yet scared but welcomed the challenge!

Training went well initially. I love running so it was just a case of trying to increase the distance. I tore my rotator cuff somehow and this hindered me slightly and I had a few close calls with the odd icy patch on the pavements but I carried on!

It started to get harder in March when yes, you've guessed it ... that "sunshine" started to get a bit stronger. I tried to go out early mornings or just before sunset.

Then I had had a Sunday out with the children wearing flip flops ... yes you can say it ... silly me! My hands and face were also sore, but my feet were so sore. It was the first proper bit of sunshine I had had in the year I guess. My feet were burning and the nerve pain meant that there was no way I could wear socks let alone shoes. If I even attempted it, it felt like the shoes were knives cutting in to my burning skin.

It took about 5 to 6 days before I could get any shoes back on, and I felt so tired and a bit down as that sunshine was getting in the way of life yet again! Once it had passed I was very very careful. After all ... I had 3 weeks until I had to complete a marathon!

Training was hard after that, especially as I had the added complication of a spell of sunny weather and trying to fit the runs in around 4 children – this meant

that the light made things quite tough!

Race day arrived and it was the best weather that I could have asked for really. There was a lot of cloud coverage and it was dry!

Now, all I can say is that it was one of the best experiences of my life! I knew I wouldn't make it in a good time as I had damaged my calf muscle so it was all taped up and I was instructed by a physio to stop if it hurt! Well, it did hurt badly at about mile 14 so I stopped and walked oh, and danced and then walked and then jogged when I could! It ended up taking 5 hours and 59 minutes but do you know what ... I didn't stop smiling that whole time! I had a mild reaction on my hands but the blisters and the general aching of my body soon made the EPP pain disappear!!!

Would I do it again? Yes ... of course I would!!!

## ALNYLAM REPORTS INTERIM CLINICAL DATA ON INVESTIGATIONAL TREATMENT FOR ACUTE HEPATIC PORPHYRIAS

At the International Congress of Porphyrins and Porphyrins held this past September in Germany, Alnylam reported initial clinical results for ALN-AS1, an investigational RNAi therapy, targeting aminolevulinic acid synthase 1 (ALAS1) for the treatment of the acute hepatic porphyrias.

This Phase 1 study is being performed in 16 asymptomatic "high excretor" (ASHE) patients, who carry the genetic mutation of acute intermittent porphyria (AIP) and have elevated levels of aminolevulinic acid (ALA) and porphobilinogen (PBG), the toxic haem intermediates that are the cause of porphyria attacks.

ALN-AS1 was found to be generally well tolerated with no clinically significant drug-related adverse events to date. 19 adverse events were reported to be mild to moderate in severity and occurred in 12 ALN-AS1-



treated patients. Study results also showed that a single subcutaneous dose (i.e. under the skin) of ALN-AS1 resulted in up to 82% lowering of urinary ALA and up to 93% lowering of urinary PBG that lasted out to 42 days. Alnylam plans to advance the study to a small group of AIP patients experiencing multiple recurrent attacks in early 2016.

Additionally, at the congress, Alnylam reported initial data from EXPLORE, a multinational "natural history" study of AIP, variegate porphyria, and hereditary coproporphyrin patients with recurrent attacks. Results from 68 patients enrolled thus far demonstrated that, on average, these patients are experiencing 10 porphyria

attacks each year. In addition, approximately 50% of all the patients reported chronic symptoms between attacks. This study is also capturing ALA and PBG levels in the urine and blood of patients both while asymptomatic and during attacks, which may help further the understanding of the acute hepatic porphyrias.



## EVENTS IN 2015:

Our annual **Open Day** was held in Leeds on Friday 29 May 2015. We are delighted to report that it was a great success. We had more than 65 people attend, with an equal split of acute and EPP patients. The day was organised with the help of Dr Stein and was held at one of Leeds University's campus' which worked well for the day. We had a great variety of speakers, including talks from the porphyria testing lab in Leeds, Arc Window Films, representatives from Alnylam Pharmaceuticals and Clinuvel Pharmaceuticals Ltd, as well as two patient experience talks and some very interesting informal discussions and a very informative Q&A session – we are very grateful for all of the speakers and attendees who all contributed to making the day a wonderful success. We also received some great suggestions for future meetings.

On Saturday 24 October we held our **Autumn Conference and AGM** in London at one of Guys and St Thomas' teaching campus'. We are very grateful

to Dr Sarkany who helped to organise the day. In response to feedback, we extended the timings to incorporate a slightly longer day, we also grouped sessions together to allow a specific cutaneous section in the morning and then a more acute section in the afternoon. During these times, people were given the opportunity to use the breakout room for informal discussion instead of attending talks that may not have been of the most relevance to them. The content of the day mirrored the talks in Leeds – all of which were just a little shorter. We are very grateful for the updates from Alnylam Pharmaceuticals and Clinuvel Pharmaceuticals Ltd, and to Dr Sarkany, Dr Stein and Dr Marsden, as well as to Arc Window Films and the three patient experience talks – thank you all for contributing to making the day another great success. We were delighted to have increased attendance again as well as wonderful feedback and suggestions that we plan to integrate into future events.

### QUESTION AND ANSWER: VITAMIN D

Our question and answer session in London raised the problem of Vitamin D deficiency. Most of our Vitamin D is made in skin exposed to daylight. Anyone unable to get outside much, because of poor health or a skin porphyria such as EPP, could be short of Vitamin D. This can cause a variety of problems, including osteoporosis (fragile bones). Your GP can check your Vitamin D levels with a simple blood test and, if needed, recommend a suitable supplement. It is not a good idea to take Vitamin D without tests, since high doses can be dangerous.

## LOOKING FORWARD TO 2016:

Our first face-to-face committee meeting of the year will be held at South Milford WI Hall, LS25 5AF on 5th March 2016 at 12.00. Our committee meetings are open and anyone is welcome to join us.

We are starting to plan our events for 2016 and are pleased to be able to announce that:

Our **Open Day for 2016** will be held at Salford Royal Hospital with the assistance of Professor Stewart. It is provisionally booked for 8th July 2016. We are excited to announce that we plan to trial a slightly different structure for the meeting. We will start the morning and afternoon sessions with some structured talks, but we also plan to have some "table" style discussion groups to focus on different aspects, such as issues affecting children

and transition stages for the different porphyrias – more details will be announced early next year.

Our **AGM and Autumn Conference for 2016** – will be held in Peterborough on either 1st or 8th October, with the support of Dr Stein and Simon Guppy from Kings College Hospital London and their outreach work. The programme content and venue are still to be confirmed, but we hope to be within 5 miles of the centre of Peterborough.

**Please let us know if you would be interested in attending any of our 2016 events** as, based on the popularity of our 2015 events, we expect places to fill quickly. We will send further information about the respective event once it is available.



**Please note:** The BPA do have some funds to help support our members to attend meetings – **but, as this funding has been provided to help those who really need it, the pot available is limited!** If you would like to apply for funding please get in touch (funding available up to: £50.00 for travel and £50.00 for hotels).

# 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.porphyria.org.uk](http://www.porphyria.org.uk)

email: [helpline@porphyria.org.uk](mailto:helpline@porphyria.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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Vice-chairman: Liz Gill

Treasurer: Anne Newton

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Prof. Timothy Cox

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## ACUTE CARE IN SCOTLAND

A meeting was held in Glasgow, in October, to discuss access to porphyria services in Scotland, including issues concerning referrals, testing and onward care. The meeting was well attended by doctors from Scotland, NAPS doctors (from England and Wales), biochemists, geneticists, NHS Scotland commissioners and the BPA.

The current situation was highlighted, and best practice

strategies and pathways were discussed. The BPA were delighted to be involved in putting forward the patient perspective, by demonstrating concerns received from our members.

Over the coming months more work will go into establishing effective networks in Scotland. We look forward to providing further updates as soon as they are available.

THE 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 £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)
- I have set up a standing order using my internet banking\*
- I enclose a cheque made payable to the 'British Porphyria Association' for £ \_\_\_\_\_
- I have made a payment using [www.justgiving.com](http://www.justgiving.com)
- I would like a receipt

\*please use your name and postcode as reference for an online payment

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:  
Sort code: 20-43-63 Account No:7099 6904

### 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 \_\_\_\_\_