

# THE BRITISH PORPHYRIA ASSOCIATION NEWSLETTER

Registered Charity No. 1089609



## INTERNATIONAL CONGRESS ON PORPHYRINS AND PORPHYRIAS (ICPP)

ICPP is an international meeting held every two years. With over 500 attendees from 59 countries, the ICPP meeting was held in Milan, Italy in early September. It provides an opportunity for doctors, researchers, clinicians and patient groups from around the world to come together to share knowledge, present research and debate best practices. The event started with a Patient Day, then the three-day scientific programme followed.

The event was a global success for many reasons. Before the formal ICPP programme even began, the Global Porphyria Advocacy Coalition (GPAC) held its inaugural meeting at which the organisation was formalised. *See the separate article on GPAC for more information.*

### Updates from the scientific community

On to the knowledge gained from the ICPP. There are lots of different areas for research going forwards. In comparison to previous years, the event felt more focused on the all-encompassing impact of the porphyrias, as well as giving a greater emphasis on mental health needs, which it was acknowledged was due to a

stronger patient voice. This is something we will continue to advocate for.

Notable sessions/discussions included:

- A panel discussion on defining an **acute** attack: this session highlighted that there are differences in approaches to treatment in different countries. The soon-to-be-developed porphyria care guidelines (current EPNET working group project) will take into account all of these different approaches to try to find a consensus-based approach to care.
- Various sessions on outcomes of the Givosiran clinical trial for **acute porphyrias**. For more information, please see [www.alnylam.com](http://www.alnylam.com)
- A presentation on a novel approach that might lead to treatments for **EPP/XLEPP and CEP**. Researchers are looking into ways to inhibit production of a protein called ALAS2, which could reduce the levels of ALA in the bone marrow and protoporphyrin IX in the blood and bone marrow, the elements that cause symptoms for these patients. This is still at a very



early stage of research, but is an interesting concept that we will be keen to see develop as time goes by.

- There was also considerable discussion over gene variants and which ones really cause problems in porphyria. It is now known that very few people with a gene mutation will ever have an attack of **acute porphyria**. It is believed that there are predisposing, modifier or protective

genes that make some become active and others not. An exciting worldwide research project, due to start soon, aims to study non-symptomatic family members (mothers and sisters) of people with active porphyria to try to find out more. Amongst other things, they will do complete gene sequencing to try to find out what gene modifies their gene or protects against having attacks.

# THE BPA – CELEBRATING 20 YEARS!

It's 20 years since the first BPA newsletter was issued in October 1999. Over the years, the charity has grown significantly and we really have come a long way since then, to be a well-recognised porphyria support group working with patients, medical professionals and drug companies.

It was back in 1998 that Karen Harris, one of our founding members, former chairman and current trustee, read a newspaper article that represented the porphyrias in a shocking and negative manner, this prompted her to write to the newspaper and sparked the foundations of the BPA, a charity to help those with porphyria and their families. With the help of Prof Timothy Peters and Prof George Elder (which was very gratefully

received), a small group of patients were brought together in London to discuss how they might be able to work towards a support group. In short, it was from these humble beginnings that the BPA was founded.

The BPA has grown from just a handful of members in 1999 to over 345 today. We would like to take the opportunity to thank everyone who has made donations of time or money over the many years, as it is this dedication and commitment from our members, doctors, pharmaceutical companies and the BPA committee and advisers that have made the BPA what it is today. There are too many names to mention individually, but we would like to say the HUGEST of THANK YOUs to EVERYONE. We really are grateful to you all!

## 20th Anniversary Festival – Manchester – Saturday 12th October 2019

For those of you who may not know, the BPA held a celebratory Festival event in Manchester to commemorate our 20th Anniversary. The day was truly AMAZING and we would like to give a HUGE thank you to all 169 people who attended and got enthusiastically involved in the workshops and interactive sessions – you made the day what it was! We'd also like to say a MASSIVE thank you to all of the speakers, all of the BPA volunteers, the videographers, the STEM ambassadors, the childcare team, the catering and venue staff, and anyone else who helped in the realisation of this fantastic event!

Thank you also to our financial supporters. In January, we were fortunate to be a successful recipient in Alnylam Pharmaceuticals' Advocacy for Impact competitive grant programme. This meant that an event of this scale really was possible! We would also like to thank Recordati Rare Diseases (previously known as Orphan Europe) and Clinuvel (UK) Ltd for their continued support and grants which helped to support the Festival. Indeed, as Antony Fearn mentioned in his introduction to the festival day, a patient event of this scale dedicated to porphyria has not

been seen anywhere in the world before!

## Plans for the future

The BPA's work streams have evolved over the last 20 years in line with new developments and areas that our members have needed support in.

The BPA's three main aims for support, education and research will remain integral to the charity's mission and we expect to continue evolving as patient needs develop and change.

We hope to continue to develop by focusing on:

- Bringing more volunteers into the charity with specific skills that can help us achieve monumental things!
- Generating increased engagement
- Being more accessible to children and young people
- Encouraging and supporting research
- Finding better treatments for all of the porphyrias
- Maintaining and fostering networks in the UK, Europe and Globally

We really are excited to see what new developments there will be in the porphyria world over the coming years, there are bound to be changes in terms of treatment options and patient care too – it will no doubt be an exciting few years to come!



## FESTIVAL ORGANISING TEAM

On behalf of all those who came to the Festival, we need to give a big thanks to the working group that organised the Festival. They were led by Sue Burrell, and also consisted of Antony

Fearn, Sophia Speakman, Richard Bennett, Liz Gill and John Critchley.

A year ago, they had already worked out the outline of what we would be doing, and had costed it

out – very accurately – so we could apply for grants to cover the costs.

They have continued to work hard at organising all the details, arranging the speakers, and contacting, and

keeping in touch with all the members who came to the meeting. Well done, and thank you for all your hard work.

*John Chamberlayne*  
BPA Chair



## GLOBAL PORPHYRIA ADVOCACY COALITION – GPAC



Left to right: Sue Burrell, Sean Hegarty, Joaquín Montoto and Peder Sorenson

Over the last few years there have been many discussions within the global porphyria patient support community about coming together to form an umbrella organisation which would support and represent all porphyrias across the world. The vision was that this new group would be fully inclusive and collaborative and would represent all of the porphyrias. By supporting each other and promoting the sharing of knowledge and resources between national groups, it will enable each group to be stronger.

Throughout 2019 much work occurred between these groups, via teleconferences

and email, to establish this new umbrella organisation for porphyria patient advocacy groups worldwide. The BPA's Chairman, John Chamberlayne, volunteered to be the interim leader of this new group, known as the GLOBAL PORPHYRIA ADVOCACY COALITION – GPAC. John, in this role, was instrumental within GPAC, he chaired many of the first meetings and was responsible for completing the application to register GPAC within the UK as a Charitable Incorporated Organisation (CIO) with the Charities Commission – along with lots of other paperwork.

During the international porphyria congress in Milan

in September, GPAC held its first face-to-face meeting where the first Executives were elected. We are pleased to announce that the BPA's very own Sue Burrell was elected to be the President of GPAC for a 2-year term. Sean Hegarty from the Porphyria Association Inc. Australia is the Vice President, Joaquín Montoto from Asociación Española de Porfiria (Spain) is the Secretary and Peder Sorenson from Denmark is the Treasurer – CONGRATULATIONS to all of you!

Since its inaugural meeting, GPAC has grown to include 24 member organisations, representing 33 countries

from around the world. GPAC's members will join working groups to focus on the following key areas: access to treatment, support, research, awareness and education.

Ultimately, GPAC aims to connect, support, engage and complement national porphyria patient groups from across the world. Through the provision of a truly global network with a strong unified voice it aims to fully represent and safeguard the interests of all porphyria patients. With this in mind, we will keep you updated on any new developments that may be of interest to the BPA as they develop.

## FIRST PATIENT MEETING IN IRELAND: A GREAT SUCCESS

In June this year, the BPA were delighted to hold the first ever porphyria patient conference in Dublin, Ireland. A total of 44 people attended, mainly from Dublin and the surrounding areas, but a number travelled from Northern Ireland too – it really was wonderful to see so many people! The feedback from the day was very positive and we were delighted to have brought

so many people together – many had never before met others with porphyria.

We are very grateful to Dr Vivion Crowley and Nadia Brazil who helped in the preparations for the day, whilst also speaking alongside Dr Mike Badminton from Cardiff, Dr Vicky McGuire and Nicki Traynor from Dundee and Dr Bob Sarkany from London – all of whom very kindly travelled to Ireland to support this great event.

The BPA would also like to extend a massive thank you to Recordati Rare Diseases (formerly Orphan Europe) who provided a grant which helped with the financing of this meeting.



# THANK YOU

In our 20th year, there has been lots of fundraising and awareness from our supporters. THANK YOU to all of you, whether mentioned below or not. We really appreciate your efforts.

During the autumn, the BPA received a very generous donation of £3,000 from The Lime Tree Society. A huge THANK YOU – this donation will go a long way towards helping to support porphyria patients.

Over the years, the BPA has also been very lucky to have the support of Daniel Wentzell and his team at Coterie Creative. They not only take our newsletter articles, add flair, and convert them into a lovely finalised document for no fee, but this year also covered the significant printing costs for the newsletter too. We are very grateful for Daniel's continued support.

## Sporting events

THANK YOU to all those choosing to take part in sporting events to raise awareness and funds.

In September this year, we had three runners in the Great North Run, David Milne, Sue Hindley and Bryan Chapman. All have relatives with EPP. Together they raised an absolutely fantastic £1,788 for the BPA.

Amy Williams, who has a friend with AIP, chose to run the Cardiff Half Marathon and raised a brilliant £200 to help patients.

Another energetic challenge was completed by Elizabeth Dumbill and 12 others, who completed the Yorkshire Three Peaks, raising a fabulous £390.

## Other events

The children of the Valerie Shepherd Dance Academy have been busy again, in support of members who have EPP, raising a brilliant £42 to help others. THANK YOU to all of you.

Alyx Byrne has continued her awareness campaign on the Isle of Man in support of her niece and nephew who have EPP. Amongst bingo nights and sweets sales, Alyx completed various stages of the

Alyx - Manx Parish Walk in full protective gear



Manx Parish Walk in full photo-protective clothing to raise awareness and funds. Altogether, she has raised a whopping £2,508. THANK YOU Alyx, Natassja and all of your family and friends.

Furthermore, these events encouraged donations from The Zurich Financial Services Ltd, Isle of Man, who sent a cheque for £200.

## GREAT NORTH RUN – 13 SEPTEMBER 2020

The Great North Run will be celebrating its 40th birthday in 2020! The BPA are delighted to have 4 places remaining in what will no doubt be an epic extravaganza of an event.

The successful applicants will need to be able to run the 13.1 miles from Newcastle to South Shields in less than 6 hours, and raise around £300 for the BPA. If you are interested, or would like more information, please contact [sue.burrell@porphyria.org.uk](mailto:sue.burrell@porphyria.org.uk).



David Milne and Sue Hindley



Sarah and Matthew with their Dad, Bryan Chapman



Bryan Chapman

## NEWS: INTERNATIONAL TREATMENTS

Autumn 2019 has been a monumental point in time for new drug development, with two new treatments having been APPROVED for use by porphyria patients in the USA.

Firstly, in October, the US Food and Drugs Administration (FDA) APPROVED Clinuvel's **Scenesse (Afamelanotide)** to treat adult patients with **erythropoietic protoporphyria (EPP)**. This is the first step towards US patients being able to access the treatment. Patient access will take time, as distribution and insurance processes are worked out. The American Porphyria Foundation ([www.porphyrifoundation.org](http://www.porphyrifoundation.org)) and [www.clinuvel.com](http://www.clinuvel.com) will provide more information as it becomes available.

Secondly, on 20 November 2019 the US FDA APPROVED (via their Priority Review process) Alnylam Pharmaceutical's **Givaaarli (Givosiran)** for adult patients with **acute hepatic porphyria**. Again, this is just the first step towards US patients being able to access the preventative siRNA treatment that has been undergoing trials around the world. Again, patient access will take time, however, this is a positive step for US patients. For more information, please see: [www.alnylam.com](http://www.alnylam.com)

It is important to point out the difference between approval of a treatment and the treatment being funded by national health organisations and health insurance schemes. Approval means the drug is deemed safe and effective in the treatment of a disease. It is then down to national funding bodies to decide whether the drug is deemed cost effective enough for treatment to be funded.

We look on excitedly and will provide reports if these APPROVAL decisions provide any impact on regulatory bodies in Europe and the UK.

## NEWS: INTERNATIONAL DRUG TRIALS

As always, we are keenly watching worldwide developments in potential treatments for all porphyrias.

One such company that we are keeping a watchful eye on is Mitsubishi Tanabe Pharma Corporation who recently announced the successful completion of their US trial into MT-7117. This study, called ENDEAVOR, investigated a potential oral treatment option for **EPP (erythropoietic protoporphyria)**. Adopting a new means of determining efficacy, the study met its primary endpoint of change from baseline in average daily time (in minutes) to first prodromal symptoms associated with sunlight exposure. MT-7117 was generally well tolerated with an acceptable safety profile. Prodromal symptoms are the ones that you get before the main problem develops. With EPP they are often a tingling warning you that you need to avoid any more light exposure.

It is expected that further trials will take place in the near future. It is hoped that the study might be widened to involve locations in Europe. To learn more, please see the press release: [www.mt-pharma.co.jp/e/release/nr/2019/pdf/e\\_MTPC191111.pdf](http://www.mt-pharma.co.jp/e/release/nr/2019/pdf/e_MTPC191111.pdf)



## NEW WORKING GROUPS

To help us achieve our aims, we want to encourage new volunteers to get involved in specific working groups. Operating alongside day-to-day existing committee activities these working groups will focus on specific projects, so that we are really able to utilise specific skills. With this in mind, we are very excited to welcome *Victoria Harrold* on board for *Social Media* and *Natassja Chadwick* and *Alicia Cawthorne* for *Fundraising and Awareness*. We really do look forward to seeing the impact that Victoria, Natassja and Alicia will have on our work. We'd love to hear from anyone else who would like to be involved in getting our working groups started.



## SAVE THE DATE: 19 SEPT 2020

The BPA are pleased to announce that in 2020 we will be holding our Annual Conference on Saturday 19 September 2020.

The exact location is still to be decided – as we are currently looking for suitable venues – but we are delighted to have secured the help of Dr Bob Sarkany from London and therefore expect that the venue will be within easy reach of the capital.

The event planning group are still working on ideas for the day, but we expect it to be full of engaging and interactive content, aimed at all of the porphyrias and all ages. We also plan to build in dedicated socialising time too, with the possibility of activities, as these elements were rated as excellent at the Festival event in Manchester.

We very much hope to see you in September 2020. Registration will be essential as spaces are limited. If you would be interested in attending, please forward the names of anyone wishing to come with you to [sue.burrell@porphyria.org.uk](mailto:sue.burrell@porphyria.org.uk) or call 0300 30 200 30.

Look out for updates and more details via social media and in our Spring newsletter!

# 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.**

Chairman: John Chamberlayne  
Vice-chairman: Liz Gill  
Treasurer: Anne Newton  
Secretary: Richard Bennett  
Administrators: Sue Burrell and Liz Gill  
Patrons: Prof George Elder, Dr Mike Badminton,  
Prof Felicity Stewart, Dr Penny Stein,  
Prof David Rees, Dr Bob Sarkany

Viewpoints 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.

## EPP PATIENTS: ARE YOU AFFECTED BY THE LIGHT FROM YOUR PHONE?

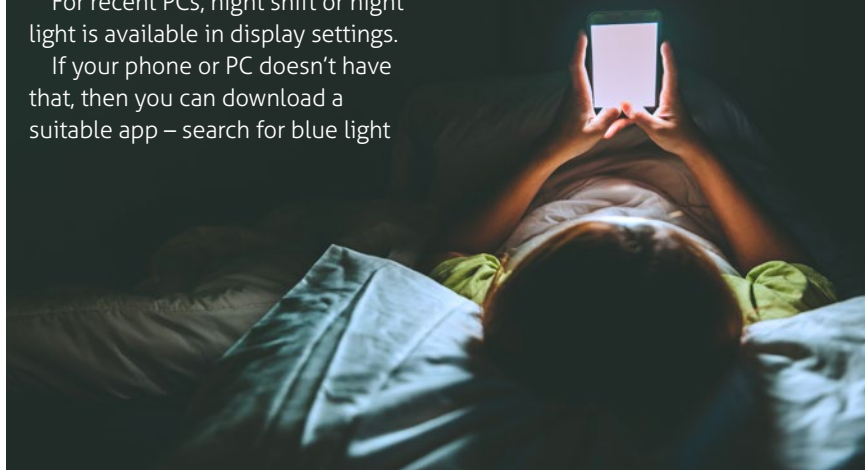
If your EPP is sufficiently severe to be affected by the light from your phone or computer screen, it is possible to reduce the amount of blue light that your device gives off. Some phones have a setting to do that in settings / display called blue light filter, or twilight on Android, night shift on iPhones, or night light on Windows phones.

For recent PCs, night shift or night light is available in display settings.

If your phone or PC doesn't have that, then you can download a suitable app – search for blue light

filter in your app store (play store, app store or Windows marketplace) or Google.

All of these have been designed to help with getting to sleep, but should be just as useful if you need to reduce the blue light emitted to help reduce symptoms of EPP.



## THE BRITISH PORPHYRIA ASSOCIATION Registered Charity No. 1089609

### MEMBERSHIP / DONATION FORM



#### MEMBER DETAILS

Please complete (\*required field) and return to:  
**BPA Treasurer, Rothlea House, 7-8 Quarry Lane, Butterknowle, Bishop Auckland, DL13 5LL or email it to [treasurer@porphyria.org.uk](mailto:treasurer@porphyria.org.uk)**

Title \* Full name \*

Address \*

Postcode \*

Landline Mobile

Email

Type of porphyria

#### Are you a (please tick):

☐ Patient ☐ Relative ☐ Medical professional ☐ Supporter/donor ☐ Other

#### What information are you happy to receive? Tick all boxes that apply:

☐ All BPA communications ☐ Newsletters  
☐ Patient events/information ☐ Fundraising/awareness events

#### How would you like us to keep in touch with you? Tick all boxes that apply:

☐ Post ☐ Email ☐ Telephone ☐ Text message

#### DONATIONS

By making a donation to the British Porphyria Association you will know that you are making a vital contribution to our work. A suggested donation of £15 per year will help us raise awareness, man our helplines or support research.

#### I would like to make a donation of:

☐ £10 ☐ £15 ☐ £20 ☐ £25 ☐ £50 ☐ other £.....

#### WAYS TO DONATE:

- ☐ Cheque: made payable to the British Porphyria Association.  
☐ Online banking (one-off payment or regular standing order):  
BPA bank account: Sort code: 20-43-63, Account no. 7099 6904  
Please use your name and postcode as the payment reference.  
☐ Credit card: via Just Giving ([www.justgiving.com/britishporphyriaassoc](http://www.justgiving.com/britishporphyriaassoc)).  
☐ If you would prefer to complete a standing order form, please call us on 0300 30 200 30 and we will arrange to send one to you.

☐ I would like a receipt

#### 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.

#### GIFT AID DECLARATION

I am a UK taxpayer 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. 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