

# THE BRITISH PORPHYRIA ASSOCIATION NEWSLETTER

Registered Charity No. 1089609



## DISC MEDICINE UPDATE ON BITOPERTIN

New research with initial positive results has been published on Bitopertin, a potential treatment for Erythropoietic Protoporphyrin (EPP).

Disc Medicine presented the preliminary results of their ongoing BEACON Phase II open-label trial at the European Hematology Association Conference in Frankfurt on 9 June 2023. The trial is evaluating bitopertin, an orally administered glycine transporter 1 (GlyT1) inhibitor, in patients with erythropoietic protoporphyria (EPP) and X-linked protoporphyria (XLP).

### Highlights of the initial data presented included:

- Consistent and dose-dependent reductions of protoporphyrin IX (PPIX), the disease-causing metabolite in EPP, were observed in patients treated with bitopertin
- Patients reported significant improvements in sunlight tolerance and measures of quality-of-life
- Bitopertin was well-tolerated, with no meaningful changes in hemoglobin observed

Read the full press release on the **Disc Medicine website:**

<https://ir.discmedicine.com/news-releases/news-release-details/disc-presents-positive-initial-data-phase-2-beacon-trial>. You can also view the data presented at the European Hematology Association Conference in **this PowerPoint presentation:** [http://porphyria.org.uk/wp-content/uploads/2023/06/Disc-Medicine-Initial-BEACON-Data-Webcast-Draft\\_Updated-6.7.23.pdf](http://porphyria.org.uk/wp-content/uploads/2023/06/Disc-Medicine-Initial-BEACON-Data-Webcast-Draft_Updated-6.7.23.pdf)



## UPDATE FROM CLINUVEL ON SCENESSE® FOR EPP PATIENTS IN ENGLAND

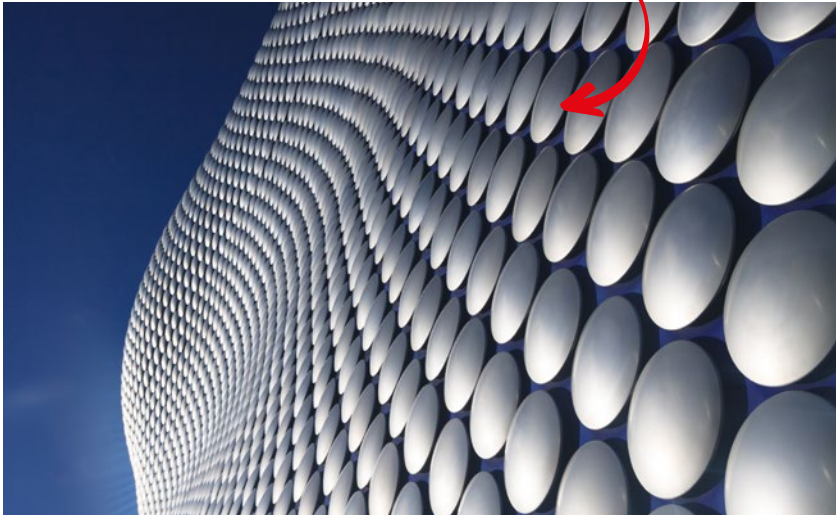
At the BPA, we're immensely disappointed that SCENESSE® is still not available to EPP patients in England. This means that patients have been denied access to what is presently considered as the only effective treatment that is available for their condition.

Please see a statement from CLINUVEL relating to the outcome of the most recent NICE appeal hearing: <https://www.youtube.com/watch?v=5i9YkoVs2RU>. We'll keep you updated of further developments as soon as we know more.

## EVENTS 2023

### BPA RECONNECT

BIRMINGHAM, 3 AND 4 JUNE 2023



Thank you to everyone that helped make the BPA's gatherings in early June at the Tamworth SnowDome (near Birmingham) such a great success. The event was well attended and the feedback was overwhelmingly positive and much enjoyed by all in attendance. We started with a quiz and social evening on the Saturday evening – which we will definitely do again as it provided a wonderful opportunity for families to come together to chat informally and get to know each other.

The Sunday brought together some excellent patient stories from Salman, Kay, Clare and then Victoria and Antony which set the tone for the day. Some excellent discussions followed in the question and answer sessions with the experts – we have transcripts of many of the sessions, so please get in touch with [georgia.newman@porphyria.org.uk](mailto:georgia.newman@porphyria.org.uk) if you would like anything sending.



### AGM AND VIRTUAL SOCIAL GATHERING – ONLINE

3 OCTOBER 2023

This year we held our AGM virtually, followed by breakout rooms where patients and their families were able to talk with others affected by their type of porphyria. If you would like to read the BPA's Annual Report and AGM slides for 2023, or for previous years, check out our website: <http://porphyria.org.uk/policies-and-procedures/>

## EVENTS IN 2024

We're very excited about a number of porphyria focused events scheduled for 2024 – if you would like to learn more please get in touch, as places are likely to book up fast and in some cases are limited. Contact us now: [helpline@porphyria.org.uk](mailto:helpline@porphyria.org.uk) or 0300 30 200 30 to learn more.

### SAVE THE DATE:

### INTERNATIONAL CONGRESS ON PORPHYRINS AND PORPHYRIA (ICPP)

PAMPLONA, SPAIN  
- 21-25 SEPTEMBER 2024

The BPA will be sending representatives to the congress event in September next year. As always, we will be actively engaged (alongside clinicians, researchers, scholars, patients and their families) over a number of days to build further connections and learn about the latest research in the field of porphyria.







## BPA RECONNECT DUBLIN, IRELAND – SUNDAY 3 MARCH 2024

The BPA are pleased to be returning to the Ashling Hotel, Dublin on Sunday 3 March 2024 from 09:30-17:00 for a much needed patient event in Ireland. This event is being organised with the support of the team at St James's Hospital, Dublin and the British and Irish Porphyrin Network (BIPNET). All are welcome, patients, their families/friends and medical professionals too.

The programme will include doctor and patient talks, question and answer sessions, updates on research and a final social session providing plenty of opportunities to connect and share with other patients.

Register now: <https://buytickets.at/thebritishporphyriaassociation>

## BPA VIRTUAL AGM AND SOCIAL EVENT AUTUMN 2024

The BPA will be holding our AGM virtually in 2024, this will be followed by a social event. Keep an eye out for further communications early next year when we will announce the date and content via email, letter and across our website and social media!

## BPA RECONNECT - ALFRESCO – 12-13 OCTOBER 2024 (ARRIVE 10AM SAT/DEPART 3PM SUN)

### Thorpe Woodlands, Thetford, Norfolk

Following the success of our activity day in October last year, we are delighted to be offering a two-day residential which will take place at **Thorpe Woodlands Adventure Centre** in Thetford Forest, Norfolk. The event will take place on **Saturday 12 and Sunday 13 October 2024** (postponed from Sept 2023 due to unforeseen circumstances – let's just say Sue won't be ice skating again anytime soon). The event is being designed to offer a safe residential environment with numerous outdoor activities for children and young people affected by EPP. Attendees will be allocated to a group with others of a similar age. They will then sleep in dormitories or family pods (depending on the family needs/ages/circumstances).

Family members are not forgotten either. We will be arranging for you to meet with other parents/siblings to build our EPP support community further with plenty of opportunities to share experiences, connect with each other and gain support from the BPA, specialists and a counsellor too. We are finalising details, but there may be the possibility to stay on-site in Pod accommodation (or nearby) too.

All places will be confirmed by the end of 2023, to enable fundraising to support the event and to offer out additional places to our EPP friends from overseas who may wish to join us for this amazing event. If you would like to learn more and book, please get in touch (ASAP) and we will arrange a call to book your space(s); contact Claire/Sue on [Helpline@porphyria.org.uk](mailto:Helpline@porphyria.org.uk).

# GET INVOLVED AND RAISE FUNDS FOR THE BPA...

Are you feeling inspired to fundraise for the BPA and contributing to our mission to support and educate patients, relatives and clinicians about porphyria? If so, here are a few simple ways you can get involved, from anywhere in the country, to raise funds and awareness for porphyria.

## CHRISTMAS FUNDRAISER

There are many ways to get involved – why not share some lovely festive cards and baubles with your friends and family, whilst also supporting the important work that the BPA do. Check out our ebay shop to place your order and get Christmas ready: [https://www.ebay.co.uk/usr/lizgill\\_bpa](https://www.ebay.co.uk/usr/lizgill_bpa) or learn more on our website now: <http://porphyria.org.uk/christmas-fundraiser/>

Christmas cards are priced at £10 for 30 cards (plus p&p) and baubles have been reduced to £16 for a box of 6 (plus p&p). Don't forget, 100% of all profits go to the BPA, so get your orders in ASAP to guarantee delivery in time for Christmas. Last order dates for Christmas delivery is Friday 15 December 2023.



We know it is a little early, but we would like to take this opportunity to THANK YOU for all of your support in 2023 and wish you and your loved ones a very Merry Christmas, from all at the BPA.



## PURPLE FOR PORPHYRIA

In 2024, the BPA are planning a series of awareness and fundraising **Purple for Porphyria** activities that you can get involved in. To make things easy, we have generated all of the materials you will need to put on a Purple for Porphyria event to raise funds (maybe a "wear purple" donating day, or a purple bake sale).

Why not **SHARE** your own story of porphyria or **SHARE** one of our awareness videos to **Spotlight Porphyria** in your workplace/school/community too. Check out our website for more ideas and information (including letters, posters and donation options) on fundraising for the BPA: <http://porphyria.org.uk/fundraising/>

You could choose any date or you could get involved in an awareness fundraising event when others from the porphyria community will also be getting involved:

- **Rare Disease Day - Thursday 29 February 2024**
- **Porphyria Awareness Week** – Anytime between **11-18 May 2024**
- **Global Porphyria Day – Saturday 18 May 2024**

Help others to **Re-think Rare – Think Porphyria** by organising a **Purple for Porphyria** event to **Spotlight Porphyria**.

Get involved in the **Global Chain of Lights** and organise a local event by getting a landmark 'lit up purple' in the **Light Up for Rare** campaign on **29 February 2024!** Full details on how to contact local landmarks can be found on our website: <http://porphyria.org.uk/fundraising/>



## VOLUNTEERING WITH THE BPA

Do you know of anyone looking for some **voluntary work** as part of their **Duke of Edinburgh** award or for some work experience? If you do, and they are looking for somewhere to volunteer, please send them our way and we can discuss options of how they can get involved! To learn more about DofE volunteering, check out our website: <http://porphyria.org.uk/wp-content/uploads/2023/07/DofE.png>





# ORAN EPP FUNDRAISER EXTRAORDINAIRE

## EMPOWERING PORPHYRIA PATIENTS – EPP FUNDRAISING CAMPAIGN

Can you help the BPA to Empower Porphyria Patients NOW? The BPA have a HUGE fundraising campaign to generate £20,000 to help fund the BPA RECONNECT Alfresco in September 2024: <https://www.justgiving.com/campaign/empoweringporphyriapatients>

All donations and fundraising, no matter how big or small, will help us to raise this staggering amount... below are some easy ideas on how YOU can get involved! The BPA can help support you directly if you would like to fundraise, but don't know where to start. Please get in touch ASAP on 0300 30 200 30 or [helpline@porphyria.org.uk](mailto:helpline@porphyria.org.uk)



**SPORTING  
EVENTS**

Organising your own personal challenge or sporting event is worth the effort and rewards. From epic solo hikes to group cycling events, football tournaments and sports days, there's something for every taste and ability. You could take on a walk, run, cycle or triathlon. Or you might have something more daring in mind!

We coordinate a number of official places each year in some of the most popular sporting events. Register your interest today on [helpline@porphyria.org.uk](mailto:helpline@porphyria.org.uk) or 0300 30 200 30. If an active/endurance event isn't your style, you might have other plans to raise funds. Check out our **Other fundraising ideas** page for more hints and tips on raising funds: <http://porphyria.org.uk/other-fundraising-ideas/>.

### Mission

The Stairway to Heaven walk, to raise £1,000 for the BPA.

### Location

County Fermanagh, the Cuilcagh Mountains

### Date

Sunday 20 August 2023 (chosen as it usually rains in August in Northern Ireland!)

### Challenge

A trail up through the Cuilcagh Mountains, with a height of 666 metres (2,185 ft), which involves completing the 9 miles (15km) round trip before the warmest part of the day

### Team

Oran, who has EPP and 23 of his family members and friends

### Equipment

Sun cream, hats, sun snood and protective gloves, plus the purple 'porphyria' t-shirts

### So how did the challenge go?

(Reported by Breanda, Oran's mum)

"On the weeks leading up to the walk Oran was anxious and was constantly checking the weather on his phone (as he does with school trips and football matches).

On the morning of the walk, Oran was relieved to see the sky was very overcast and he felt he was able to join his friends on the walk.

Even with the morning weather being in Oran's favour, he always has to have his sun cream, hat, sun protective snood and UVA protective gloves with him – as he is always aware the sun could come out at any point (thankfully this did not happen until we were arriving back at the coach, to go home).

Luckily, Oran was well covered and did not have a reaction!

Instead, he had a great day out with his family and friends and did not have to suffer for it later.

We all made it to the top and the view was stunning!"

### RESULT

**Oran and his family & friends have raised over a phenomenal £4,750 for the BPA!**





**UKPMIS**  
UK Porphyria Medicines  
Information Service

# USING HORMONE REPLACEMENT THERAPY (HRT) IN PORPHYRIA

*An update from UK Porphyria Medicines Information Service*

## Part 1 – HRT and Porphyria Cutanea Tarda (PCT)

In this and upcoming newsletters we are going to be talking about the use of hormone replacement therapy in those with porphyria. This month we're discussing the use of hormone replacement therapy (HRT) in porphyria cutanea tarda (PCT).

### HRT and PCT

HRT is most commonly used to treat menopause symptoms. As you approach menopause, the hormones oestrogen and progesterone fall to low levels. HRT simply replaces these hormones. PCT is the most common type of porphyria. It is usually first noticed due to the skin lesions that can appear.

### PCT in Remission

Treatment of PCT is often very successful and can result in remission of the condition. Remission means that there are no symptoms of PCT. When PCT is in remission, there are no restrictions on the medications that can be taken. Although, drinking alcohol should be avoided as this can cause PCT to stop being in remission.

### Active PCT

There is a risk that PCT may relapse and symptoms may reappear, causing it to become active. PCT is classed as active when at least one of the following is present:

- skin lesions
- increased amounts of porphyrins in the urine or blood

If you have PCT and it is active, you should avoid oestrogen if possible. This is because it may cause your PCT symptoms to get worse. Usually, oestrogen is a part of HRT. It can also be found in some oral contraceptives.



Sometimes, HRT treatment is essential. If you do require HRT treatment and you have PCT, make sure that you are taking the lowest possible dose of oestrogen to manage your symptoms. If you are concerned, contact your doctor for more advice.

**The information here is not exhaustive and is specifically for patients with PCT.** For more information about HRT treatment and PCT speak to your healthcare provider or call us on 029 2184 2251.

In our next newsletter edition, we will be releasing a further article on HRT and the acute porphyrias. In the meantime, for more information on acute porphyria and SAFE medications, visit [www.wmic.wales.nhs.uk/specialist-services/drugs-in-porphyria/](http://www.wmic.wales.nhs.uk/specialist-services/drugs-in-porphyria/) or call us on 029 2184 2251.

Are you still receiving your annual SAFE drugs list? If not, please call us on 029 2184 2251.



# I HAVE EPP... CAN I GO ON HOLIDAY TO DISNEY WORLD?

Interview by Claire (an EPP patient)

I knew my mouth had dropped open and I was seemingly incapable of closing it... we had been speaking to Sarah, the mum of Lizzie, a young EPP patient and she had just told us she was in the middle of preparing for her family's holiday to **FLORIDA**... yes **FLORIDA** the sunny and hot place in the sticky out bit of America famously inhabited by a Mouse with big ears and his various chums...

After I had managed to form words, I managed to ask, "how is that possible for someone with EPP?"

So, after Sarah and her family had returned, I bombarded her with loads of questions...

## How did you know you could go to Florida with minimal risk for Lizzie?

It was a risk, we had been once before Lizzie was diagnosed, this wasn't a particularly successful trip in the beginning, as she had a reaction on the first day, (although we didn't know what was wrong with her at the time). I then took the precaution of going to the shopping Mall and stocked up on long sleeved tops. After that we had a reasonable time, but it was more luck than anything, as we were out mostly on the cloudy afternoons and into the evening.

Once it was becoming more apparent that she was going to be diagnosed with EPP, I planned our next trip. I knew it would be

ok as **Florida's climate** although hot is very tropical and **CLOUDY\*** I knew we could work with this with a lot more confidence than planning a trip to Europe in the summer.

## What did you need to specifically consider when booking a holiday to Florida and Disney?

The **climate**, a European summer pool holiday to somewhere like Spain / Greece is not suitable for us, even with all the precautions, most hotel pools close at 6pm and there is no cloud cover.

Pools, Lizzie is a lover of the water and to have a **pool that was accessible in the evening and partly shaded** was the one thing that was non-negotiable to us to have a good holiday. Our resort had 2 pools, a small one that was **shaded by high buildings until midday** and a large one that was unusable in the day for us but **from 6pm was shaded and then stayed open until 10pm**. She could swim without worrying and that was wonderful to see.

**Indoor activities to do in the day**, bowling, cinema, go karting, museums, so on very sunny afternoons we could keep occupied.

Lizzie and **anyone with EPP is entitled to a Disability access pass** for the Disney theme parks and there is a similar scheme for the other parks. They stress

that this is not a queue jump, but you simply go to the ride, and **they give you a return time**, so you aren't queuing up as long. We found the process seamless at Disney and I didn't need to show any proof to them and just explained our situation and it was added to our digital tickets, I had a letter this year from Lizzie's consultant though in case I was asked for any proof. To be honest I would say over 50% of the ride queues are indoors and air conditioned.

## Do you take any special clothes / sun block?

Surprisingly I used next to no sun block on Lizzie this year, I used the La Roche Posay 400 on the parts of her the face which weren't covered by her hat but that was it.

We had **thin gloves, long sleeved tops with thumbholes, loose pants, and an array of caps and bucket hats**. A new product we tried this year was **sleeves from Coolibar**, this allowed her to wear her short-sleeved tops as well without a base layer underneath. I had been looking for **swimming costumes** all year for Lizzie I finally found one she would wear **with thumbholes** in, so she was able to swim without gloves on as well.

## What was the best thing?

The **accessibility of the pools in the evening**, our hotel was not unique in the pools being open late, most of the large resort hotels have the pools open until 10/11pm. Also going over to the beach and getting 2 hours in the sea around sunset.

One of Lizzies biggest wishes was to visit a water park with her brothers, we found that **Typhoon Lagoon**

**did evening sessions from 6-11pm**. Most of the **theme parks are open late in the summer**, you don't need to go early, what works for us as a family is to go at around 2/3pm which sounds crazy as that's the hottest time of the day, but the parks only get cooler and quieter from this point on as many families who have gotten there for opening are leaving at this point.

We even managed a **daytime session at Discovery Cove**, which is another water park, we had some **cool swim leggings** that I found online which **covered the tops of her feet to compliment her swimsuit that has thumbholes**, and she found her love of the beach this year, with some sunset bodyboarding. She had some bad experiences at the beach in her younger years and had developed anxiety about going anywhere near the sea and sand, so this was a particular highlight.

When we were there Florida was having a heatwave and it was extremely hot, but Lizzie had no adverse reactions, so it was a success.

## What's your advice for EPP patients and their families who may after reading this consider a trip to Disney?

**Prep, prep, prep**. Email the hotels, check the opening times of the theme-parks and the pools, research things to do in the daytime, find out what activities are happening in the evening.

So, as I have EPP and as the nearest I have got to Disney is reflected in this picture (circa 1974 (which explains the tragic socks!)) ...can I go on holiday to Disneyworld in Florida... and yes, with a lot of planning, the right clothes (which it seems have advanced somewhat since the 1974 version of a bucket hat...) and at the right time of year... it seems... **I can!**



\*[NB Sarah didn't shout this I just thought it was vital to highlight this for anyone with EPP!!!!]

# 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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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.

## THANK YOU + WELL DONE

The BPA would like to give thanks to lots of people who have been involved in raising funds, in different ways, over the last few months! Firstly, HUGE thanks go out to the Carlin family, Oran and his Mum and Dad (Oran and Breanda) and their family and friends who raised over £4,750 for the BPA. This was a massive undertaking, the planning and commitment of 23 people to fundraise, raise awareness and be involved is so valued by all at the BPA.

We had a wonderful team involved in the Great North Run (GNR) in September, travelling all the way from Norwich, Norfolk. THANK YOU Ashleigh, Gemma and David for the dedication to train and raise funds for the BPA. WELL DONE and CONGRATULATIONS on collectively raising over £1,550.

THANK YOU for a £100 anonymous donation, a £50 donation from Mr Biddulph and a donation of £50 from Ms Bryant in the summer. We'd also like to THANK our regular donors for your ongoing support.

THANK YOU to all of those who donate via the PayPal Checkout process. And, belated Happy Birthday THANK YOU to Hannah and Michelle (and their family and friends) who utilised birthday fundraisers via PayPal – collectively raising nearly £100.

THANK YOU to Karen Harris and her local Brue Valley Rotary Club (based in Somerset) who donated £60 earlier this year from doing an awareness talk on the BPA and porphyria.

We received our last payment of nearly £60 from Amazon Smile in May – THANK YOU to all of you who have used this platform over the last few years.

Despite no longer being available, you can still help raise funds for the BPA when buying from Amazon and other UK retailers by switching to **Easy Fundraising** which allows the BPA to earn 'cashback' on all your purchases. **THANK YOU to those who already use this platform and why not check out this easy way to raise funds for the BPA:**

[www.easyfundraising.org.uk](http://www.easyfundraising.org.uk)

## 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 want to Gift Aid my donation and any donations I make in the future or have made in the past 4 years to the British Porphyria Association (Registered Charity No. 1089609). I am a UK taxpayer and understand that if I pay less Income Tax and/or Capital Gains Tax than the amount of Gift Aid claimed on all my donations in that tax year it is my responsibility to pay any difference. (I will advise the BPA if my tax status, name or address changes.)

DATE

SIGNATURE