Issue 50 / **Jan 2026**

THE BRITISH PORPHYRIA ASSOCIATION NEWSLETTER

Registered Charity No. 1089609

EXCITING NEW ERA FOR THE BPA

As we begin the new year, we are excited to share an important step forward in strengthening our organisation for the future. Over the past year, we have reflected on how we can continue to grow, adapt and remain resilient in a changing landscape with several new medications likely to come into the regulatory framework. With a refreshed vision and a clearer organisational direction we hope this will position us to better serve the porphyria community and deliver lasting impact.

To support this next chapter, we are pleased to announce the establishment of a new primary CEO role. **Liz Gill will take on this position**, providing dedicated strategic leadership and ensuring we have the focus and capacity needed to guide the organisation through its next phase of development.

At the same time, **Sue Burrell will transition into the role of Community Engagement Specialist at the BPA.** This new position reflects our commitment to strengthening national and international relationships, amplifying community voices, and ensuring that the people we serve remain at the heart of everything we do.

This will empower Sue to fully realise her role as President of the Global Porphyria Advocacy Coalition (GPAC), enabling her to channel her time, experience, insight, and passion into global leadership and expanded community advocacy. We are excited to support this inspiring and natural next chapter for Sue.

These changes mark an important milestone in our journey toward greater resilience, clarity and impact. We look forward to working together – staff, volunteers, partners and community members – to make this year a very exciting one.

Here's to a strong and inspiring year ahead.

LIGHTS, CAMERA, ACTION!

As you'll read later, our Smiley Awards entry isn't the only film we've been working on – it's just one in a series of porphyria awareness videos that are now available to watch on our **website and YouTube channel**!

The series was created using footage taken by filmmaker Sam Morrison and his colleagues Owen Saddington and Kyle Adams at 2024's Alfresco activity weekend for families affected by EPP. Sam is a BPA member who has EPP himself, so he has been well placed to create a sensitive and empowering portrait of our community through these videos. We hope that they can be a useful resource for helping you communicate your experiences with porphyria to your friends, family, and wider communities, so that they can understand you and your condition better.

All of the videos can be viewed via the QR below or by visiting our website at *porphyria.org.uk/awareness-videos* or our YouTube channel at *youtube.com/@britishporphyriaassociatio7939*.

And a huge thank you to our fantastic video participants for making all of this possible by sharing your porphyria stories!

It's quite sad I guess, if you're seeing all

What are the psychological effects ...

they gave me more antihistamines.

How are people diagnosed with EPP? ...

being diagnosed when I was 32

How are people diagnosed with EPP? ...

We're also engaging them in events like

What is the British Porphyria Association ...

It's a condition which is very hard

How do patients cope with EPP? ...

do stuff, I have to miss out.

What's it like to have an invisible illness? ...

THE GENERATION STUDY

The **Genomics England Generation Study** is a landmark programme that will sequence the genomes of **100,000 newborn babies** in partnership with the NHS. Recruitment began in 2024 and will continue until March 2027, with the aim of understanding how whole genome sequencing can improve the diagnosis and treatment of rare genetic conditions www.genomicsengland.co.uk/initiatives/newborns.

Every year, hundreds of babies are born with rare genetic conditions that are difficult to diagnose. Early detection through genome sequencing could allow timely interventions that transform health outcomes.

What conditions are included?

The study screens for over **200 rare genetic conditions**, chosen because they are serious and can affect health early in life, have treatments or interventions available and are not currently part of standard newborn screening.

Several porphyrias are included:

- **Erythropoietic protoporphyria (EPP) and X-linked protoporphyria (XLP)** – FECH and ALAS2 genes
- **Variegate porphyria (VP)** – PPOX gene
- **Congenital erythropoietic porphyria (CEP)** – UROS gene
- **Hepatoerythropoietic porphyria (HEP)** – UROD gene

AIP is not included because a lot of people have an AIP gene change, but only a very small proportion have AIP symptoms.

How does it work?

The test is free, safe and performed soon after birth alongside routine newborn screening. DNA is collected from a small blood sample. Results are returned to families if a condition is identified, with support provided by NHS specialists. Families are also given clear information about the benefits and limitations of joining the study.

Looking ahead

In June 2025, the NHS announced plans to expand whole genome sequencing to all newborns within the next decade, beginning with a national rollout in 2026. The Generation Study is a crucial step in shaping how this will work in practice, ensuring that families, clinicians and policymakers understand both the opportunities and challenges.

The Generation Study is actively recruiting, testing and shaping the future of newborn screening in the UK. For families affected by rare diseases, this represents a major step toward earlier diagnosis, better care, and more hope for the future.

You can learn more, check which hospitals are participating, and register your interest at generationstudy.co.uk.

IN CASE YOU MISSED IT: CYACÊLLE SUNSCREEN SURVEY

Have you received a free bottle of CYACÊLLE sunscreen from the BPA this year? We want to hear from you!

We'd love to be able to report back on the experience of this product for porphyria patients who experience phototoxic/photosensitive reactions, so, with this in mind, we'd greatly appreciate if you would complete our short feedback survey, available at https://forms.office.com/e/YjYj4BmpLJ (a link/QR code was also sent out with your sunscreen).

Results from the survey so far show that 71% of respondents would use CYACÊLLE again, and that it has 3.8 effectiveness on a sunny day and 4.3 effectiveness on a cloudy day (when rated out of 5).

The more feedback we get, the more likely we are to receive more products in the future that we can distribute to the porphyria community – thank you in advance!

If you have already completed the survey, you don't need to do it again.

We also still have a small number of supplies left so the giveaway is still open to anyone affected by the skin porphyrias in the UK and Ireland, even if you've already received some! Whether you're ordering for the first time or coming back for more, complete the order form at peoplesfundraising.com/fundraising/bpa-cyacelle-2025.

NOTE ON EXPIRATION DATE: Clinuvel have confirmed through testing that the sunscreen is safe and effective to use for 12 months after opening, regardless of the expiration date written on the bottle.

CLAIMING BENEFITS AND CONCESSIONS

If symptoms of porphyria are affecting your daily life (including your ability to work), you may be trying to access various disability concessions or tackle the benefits system for the first time.

The below information aims to provide a summary of the types of assistance available and direct you to places/resources that will be able to help you more. The BPA can provide supporting letters for your applications to any of these schemes - just email us at helpline@porphyria.org.uk.

ACCESS CARD

Access Cards offer increased accessibility to thousands of UK attractions, venues, and events for individuals facing challenges due to disabilities, including porphyria.

Anyone who meets the criteria for being a disabled person, as defined by the Equality Act, is eligible for an Access Card. This means that anyone whose life is impacted by EPP, or an acute porphyria (that has a substantial and long-term impact on them), is likely to be eligible. We can help you with your application by providing a supporting letter.

Find out more at *www.accesscard.online*

BLUE BADGE

Blue Badges help people with disabilities or health conditions park closer to their destination.

You can check your eligibility and apply for the Blue Badge scheme at *gov.uk/transport/blue-badges*

NATIONAL DISABILITY CARD

The National Disability Card is an easy, secure and portable way to show others that you have a verified disability and is accepted at 500+ locations across the UK as proof of eligibility for concessions or carer tickets. It also allows you to access online discounts from the scheme's partner brands. Like with the Access Card, anyone with a physical or mental impairment that has a substantial and long-term negative effect on your ability to do normal daily activities is eligible to apply.

Find out more at *disabilityid.co.uk/faq*

PERSONAL INDEPENDENCE PAYMENT/ DISABILITY LIVING ALLOWANCE

Personal Independence Payment (PIP) and Disability Living Allowance (DLA) are government benefits to help with extra living costs if you or your child have a long-term physical or mental health condition or disability, as well as difficulty doing certain everyday tasks or getting around because of the condition. You can get PIP even if you're working, have savings or are getting most other benefits.

Find out more at *www.gov.uk/browse/benefits/disability*

HIDDEN DISABILITIES SUNFLOWER

The Hidden Disabilities Sunflower is a simple tool for you to voluntarily share that you have a hidden disability. Simply by wearing a Sunflower lanyard or badge, you're letting everyone know that you might need extra help, understanding or just more time in shops, at work, on transport or in public spaces. There is no qualifying list of invisible disabilities – you simply choose to wear the Sunflower to indicate that you may need additional support.

Staff in businesses across the UK are trained to recognise the Sunflower lanyard, including in retail, tourism, transport (airports, railway networks, coach and bus services, ferries), education (universities, schools and colleges), healthcare, theme parks, and theatres.

Find out more at *hdsunflower.com/uk*

NHS HELP WITH HEALTH COSTS SCHEME

The NHS Help with Health Costs scheme provides financial support for people in England who struggle to afford medical expenses. It covers costs such as NHS prescriptions, dental treatment, sight tests, glasses, contact lenses, travel to NHS appointments, and wigs or fabric supports, so it may be able to help you with more than just prescriptions. Eligibility depends on factors like income, benefits, and medical conditions.

You can check your eligibility and apply at *www.nhsbsa.nhs.uk/nhs-help-health-costs*

LONDON PATIENT DAY

On Saturday 8 November 2025 we were delighted to welcome patients, their families/friends, and medical professionals to our BPA Connect Patient Day in Stratford, east London.

At our patient days we are always honoured to give porphyria patients a platform to share their personal stories. This time we heard from VP patient Selina on how she manages her symptoms alongside her love of outdoor activities, AIP patient Emma on how crucial her family's advocacy was in her diagnosis journey, and 17-year-old EPP patient Ava told us all about her exciting plans to study archaeology at university! (You can read more about Emma's experience in our latest blog post at *porphyria.org.uk/every-cloud*.)

Our patient experts then joined Q&A panels alongside skin porphyrias specialist Dr Bob Sarkany and acute porphyrias specialist Dr Penny Stein, both of whom are long-time BPA patrons and advisors. We also heard from Dr Pete Schmidt, Chief Medical Officer at Portal Therapeutics, who travelled all the way from California to provide an update on the latest research and development of a promising new medication for EPP.

We finished off the day with a lively escape game activity where excellent teamwork was displayed all around as we put our heads together to decipher the puzzles and clues and successfully catch an MI5 spy!

We are so grateful to all our speakers for sharing their individual expertise and unique perspectives, whether gained from lived experience or medical training. It all contributes to a rich wealth of knowledge that will continue to benefit the whole porphyria community.

If you attended the event, we'd love to hear your thoughts on the day and shaping future events via this short feedback survey: *https://forms.office.com/e/HR1fxzMj6v*.

We'd still like to hear from you even if you registered for a place but were not able to join on the day itself in the end.

We are not collecting personal data and the information you provide will remain anonymous, but your feedback will really help us to plan more opportunities to connect with the porphyria community in the future - thank you in advance!

Photo: chrisdorney - stock.adobe.com

WE NEED YOUR VOTES!

We are delighted to share that we are entrants in the **Smiley Charity Film Awards** – and we need YOUR vote to win!

It only takes 10 seconds and could help us win vital recognition for the porphyrias.

Every vote helps us to raise awareness of porphyria and supports our mission to provide essential support and community to families affected.

So vote TODAY at *smileycharityfilmawards.com/films/empowering-people-with-porphyria* and don't forget to share with your friends and family, too!

Thank you to our participants for sharing their stories, and thank you to Sam Morrison, Owen Saddington, and Kyle Adams for putting together this powerful short film. Thank you to our sponsors for making the Alfresco event happen.

OTHER WAYS TO SUPPORT THE BPA

Donating via standing order

A quick and easy way to support our work is to set up a **monthly standing order** to the charity. A standing order is a regular payment that you can set up to pay other people, organisations or transfer to your other bank accounts. These **regular payments**, however small, are hugely valuable because they give us the stability to plan for long-term projects and allocate our resources more efficiently.

You can amend the amount of the standing order or cancel it as and when you like.

How will my monthly donation help?
Donating little and often means the BPA can make an even bigger impact. A small amount over the course of a year can make a tangible difference in the lives of families affected by porphyria across the UK and Ireland.

To put into context:

- £6 could cover the annual cost of the BPA newsletter (two issues) for one person
- £15 could enable an initial consultation to assess patient/family needs
- £20 could enable patient support for a week via our email and telephone helplines
- £50 could pay for patient travel to a clinic appointment or BPA event
- £50 could pay for a half-day adventurous/challenge activities at an Alfresco activity weekend for young people with EPP
- £180 could cover complete costs for a young person with EPP to attend an Alfresco activity weekend (accommodation, catering and two days of activities)

Start your monthly charity donation now

Set up a standing order using the below bank details today – and don't forget to sign up for Gift Aid to boost your donation! Please use your name and postcode as a payment reference. Sort code: 20-43-63 Account No. 7099 6904

Visit *porphyria.org.uk/donate* **for more information.**

THANK YOU

We want to take a moment to celebrate our amazing community members who have gone above and beyond to make a difference recently through their incredible fundraising efforts. Since our last newsletter in June, our fantastic supporters have raised a combined total of more than **£3,000** for the BPA!

Every milestone reached and every cake sold has helped support our mission, and we couldn't be more grateful. Here's a huge shoutout to some recent fundraisers.

Half marathons were all the rage this season and we were lucky enough to have had five athletic members of Team BPA representing us in two separate corners of the country as the leaves began to turn – **three** in the Great North Run in Newcastle in September and **two** in the Great Eastern Run in Peterborough in October. Thank you to Maisie Hughes, Chloe Bullivant, Emily Todd, and mother and son duo April and Adam Fidler!

(An extra special shoutout to Maisie who was able to secure match funding from her employer, boosting her total donations by **£200** at no extra expense to her or her supporters! Find out how quick and easy match funding can be at porphyria.org.uk/match-funding.)

Together these Team BPA superstars racked up **£2,318** over 65.5 combined miles – the immense physical challenge of a half marathon (13.1 miles!) is no small feat so you should be very proud! Your endurance, dedication, and community spirit truly inspire us all. Every penny raised will go directly towards supporting our projects and services.

The **UK Porphyria Medicines Information Service (UKPMIS)** team hosted a bake sale to highlight their new office location and donated the proceeds to the BPA as a cause close to their hearts. Thank you to **Anna Burgess and whole team** at UKPMIS and the Welsh Medicines Advice Service for serving up a delicious donation of **£105** – maybe our readers will be inspired to start their own Great British Porphyria Association Bake Off!

Anna Parfitt held a dress-down Friday at her workplace in aid of the BPA and raised **£72**! For tips and tricks on holding a fundraiser at your workplace or community centre visit porphyria.org.uk/wear-purple.

We're also deeply grateful to the Gentle family and their community, who chose to donate to the BPA in memory of **Janet Gentle**. This was Janet's wish, after her son Mark found support through the BPA, and we are honoured to have been part of her legacy. Their generosity in raising almost £600 means so much to everyone we support.

Thank you to all our incredible fundraisers for their time, energy, compassion and generosity. Because of you, we're able to make a difference in the lives of those we support. We're proud to have such passionate and dedicated people in our community.

If you've been inspired by the fundraisers in this article, visit porphyria.org.uk/fundraising to find out how you can get involved.

EVENTS IN 2026

We're very excited about a number of porphyria focused events scheduled for 2026 – if you would like to learn more please get in touch: helpline@porphyria.org.uk or 0300 30 200 30.
Register: Please register for all events here: https://buytickets.at/thebritishporphyriaassociation

BPA CONNECT (in person and online)

Saturday 7 March 2026
Venue Crowne Plaza Hotel, Newcastle upon Tyne, NE1 3SA
Time 13:00-18:00

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

Full programme TBC – for now please register your interest and save the date!

Please note: we have introduced a small £5 registration fee per person to reduce the costs incurred when people register but are unable to attend. We're happy to provide a refund for any cancellations made by 28 February 2025. Thank you for your understanding.

Accessibility: We are committed to ensuring our events remain inclusive and accessible to all. If the £5 fee would be a barrier for you, please don't hesitate to contact us. We also offer a limited number of travel bursaries for those who would otherwise be unable to attend. Please contact *helpline@porphyria.org.uk* to discuss further.

Register: Please register your attendance here: https://buytickets.at/thebritishporphyriaassociation

BPA CONNECT Alfresco Residential

16-18 October 2026
Venue: Willersley Castle, Matlock DE4 5JY
Arrival: Evening Friday 16 October or early Sat 17 October 2026
Departure: Early afternoon, Sunday 18 October 2026

GLOBAL COLLABORATION AT INTERNATIONAL PORPHYRIAS SYMPOSIUM

The BPA team recently attended the International Porphyrias Symposium in Arlington, Virginia. It is an event that gathers patient advocates, scientists, clinicians, researchers and industry leaders from the United States and around the world to focus on advancing understanding and treatment of the porphyrias.

The conference opened with a foundational course on haem biosynthesis and the mechanisms underlying the porphyrias, setting the stage for deeper exploration throughout the event. Sessions explored each type of porphyria, highlighting both scientific progress and the lived experiences of patients. The patient journeys were emotive, and powerfully illustrated the real-world impact of our rare conditions, highlighting the urgency for continued research and patient advocacy.

A key theme was the evolving recognition of chronic (ongoing) symptoms in **acute porphyria**. Presentations on neurological manifestations, including autonomic and peripheral neuropathy, revealed that the clinical world is only just beginning to grasp the full extent of there long-term effects, which are really only just becoming apparent with effective treatments being available to separate out acute and chronic symptoms. Other sessions explored liver injury, reproductive health, and experimental research into histamine pathways, including the potential repurposing of antihistamines to treat liver and skin damage—an exciting early-stage concept.

One standout afternoon focused on **Congenital Erythropoietic Porphyria (CEP)**, one of the rarest and most severe forms. Patient interviews were paired with promising research into *Ciclopirox (Atlas Pharmaceuticals)*, which may reduce toxic protoporphyrins by up to 40%. This treatment is currently in Phase II trials and offers a glimmer of hope for those affected.

The **EPP and XLEPP** sessions brought fresh and exciting insights. They also revisited a long-standing hypothesis relating to cimetidine (a medication with contrasting beliefs about its potential for effectiveness in porphyria). A placebo-controlled trial on cimetidine showed it to be ineffective in reducing protoporphyrin IX (PPIX) or alleviating symptoms. However, discussions around genetic testing highlighted its growing importance—particularly in distinguishing XLEPP from EPP, as iron treatment may benefit XLEPP patients but can cause problems in EPP.

Mitsubishi Tanabe Pharma Company shared long-term data from their Phase III trial into dersimelagon, showing minimal adverse events over three years—mainly hyperpigmentation (extra skin colour) and nausea. Patient-reported outcomes showed significant improvement, and the final trial results are expected soon. Early data revealed a one-third reduction in phototoxic reactions.

After showing positive results over recent months, *Disc Medicine* is actively recruiting for its Phase III trial into bitopertin in the US, with UK and European sites to be announced soon.

Meanwhile, *Portal Therapeutics* presented positive midline data from its Phase II trial of PORT 77, showing PPIX reductions of 55% at lower doses and 75% at higher doses over just four days—regardless of initial PPIX levels. A global observational study is set to launch soon, paving the way for Phase III development.

Refer to our *past newsletter* (Issue 49) for more on how each of these therapeutics work.

Overall, the conference left attendees feeling optimistic. With diverse research avenues being explored and patient voices at the forefront, the future of porphyria care looks brighter than ever.

Members of the global porphyria patient advocacy community.

Sue, Liz and Vicky with their poster presented at the conference 'Empowering young people with EPP and XLP to CONNECT, UNDERSTAND, TAKE CONTROL and SHARE'

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.

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 team and porphyria specialists too.

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 us on *helpline@porphyria.org.uk*.

Peer support sessions

The Peer Support Programme offers porphyria patients, as well as their family and friends, a safe and welcoming online space where you can join others with porphyria to chat about challenges and gain tips on things that have worked for others, etc. The programme aims to help people living with porphyria to **understand** their condition, **connect** with others, build the confidence to **take control**, and **share** their story.

Please register for 2026 dates:
https://buytickets.at/thebritishporphyriaassociation

- Mon 9 February, 18:00-19:00
- Mon 20 April, 18:00-19:00
- Mon 8 June, 18:00-19:00
- Mon 7 September, 18:00-19:00

Save the Date: 27-30 September 2026

International Congress on Porphyrins and Porphyria (ICPP) –

Istanbul, Turkey

The BPA will be sending representatives to the congress event in September 2026. As always, we will be actively engaged (alongside clinicians, researchers, patients and their families) over a number of days to build further connections and learn about the latest research in the field of porphyria.

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*
email: *helpline@porphyria.org.uk*
address: BPA, 4 Festival Villas, Woodland,
County Durham DL13 5RG

The BPA Newsletter is published by the BPA twice yearly.

Chair: Dr Vicky McGuire
Treasurer: Claire Jarvis
Secretary: Victoria Harrold
CEO: Liz Gill
Community Engagement Specialist: Sue Burrell
Fundraising & Communications Officer: Georgia Newman
Patrons: Prof George Elder, Prof Mike Badminton, Prof Felicity Stewart, Dr Penny Stein, Prof David Rees, Dr Bob Sarkany, Dr Vicky McGuire

HELP US STAY IN TOUCH AND SUPPORT YOU BETTER

We're currently developing a new database system to help us improve the way we support our members. Our existing system is now outdated, and this update will allow us to communicate more effectively and better understand how we can help you.

We'd be grateful if you could take a few minutes to add your details to the new system using the following link: *http://porphyria.org.uk/new-member-2025/*

This will ensure we have your most accurate contact information and preferences.

Your information will be stored securely and used only in line with our data protection and privacy policies.

As part of this work, we may also follow up with some members by phone to learn more about how we can better support you.

Thank you for helping us build a stronger, more connected community.

THE BRITISH PORPHYRIA ASSOCIATION Registered Charity No. 1089609
MEMBERSHIP / DONATION FORM

MEMBER DETAILS

Please complete (*required field) and return to:
BPA Treasurer, 69 Cannell Road, Loddon, Norwich, Norfolk, NR14 6TP
or email it to *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
☐ Patient events/information
☐ Newsletters
☐ 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*).

☐ 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 ______________