Issue 34 / May 2017

THE
BRITISH PORPHYRIA ASSOCIATION
NEWSLETTER

Registered Charity No. 1089609

# BREXIT AND GENETIC DISORDERS

The BPA recently attended the extremely useful Genetic Disorders Leadership Symposium. One of the more topical and general presentations, by Nisha Taylor from the Association of Medical Research Charities (AMRC) considered how Brexit could impact on UK medical research and patients with genetic disorders.

It was conceded that it is still very early in the process, and there are no definitive answers. However there are significant risks and opportunities in the process. Three key interconnected areas will be affected:

**Funding and collaboration:** The EU is a significant funder of research and science. The UK currently receives the second highest amount of funding from Horizon 2020 – the EU's programme for research and science. There's no plan yet as to how future funding will work, but AMRC will be calling for continued access to EU funding programmes and the closest possible affiliations. AMRC is currently looking into the value of UK research to the EU, and argues that the EU needs the UK, as much of the UK research benefits the EU too, e.g. that done by Cancer Research UK.

Whilst funding is important, collaborative opportunities are just as important. It was noted that although funding may alter, personal networks of collaboration will be unlikely to disintegrate – sometimes people have been collaborating for many years and many condition-specific networks are likely to be maintained.

**People:** Research and science is a very international field – a lot of research in the UK is being led by EU nationals. Brexit uncertainty may cause researchers to rethink whether they want to come to the UK as it could be a more challenging and risky option. On the other hand, the current immigration system restricts access to non-EU students/researchers, therefore there is the potential for a new system to be more inclusive to non-EU nationals.

# THANK YOU AND CONGRATULATIONS

As many of you will have noticed at our last AGM in October, Sue Burrell, our BPA administrator was rather pregnant. We are delighted to announce that January saw the safe arrival of Abigail Penny-Rose to very happy parents, Sue and Ian. Congratulations and best wishes to the whole family.

We are pleased that Liz agreed to offer maternity cover. We are paying her for the 26 hours a month that we were paying Sue.

**A HUGE THANK YOU** also goes to the Porphyria Charitable Trust, including the trustees and all who supported this charity a number of years ago.

A non-charitable trust was initially set up with the aim of helping a young girl with congenital erythropoietic porphyria, a very rare condition involving complete intolerance to visible light and many other health complications.

During the efforts to raise funds, other donations were received that were specified as being for porphyria sufferers generally. As these could not be added to the original trust, the Porphyria Charitable Trust was established. After a long period of inactivity, the trustees decided that the funds were not sufficient alone to be able to provide meaningful support to people with porphyria. With this in mind, they approached the BPA to see whether we would be able to use the funds in accordance with the original aims of their charity – the relief of porphyria sufferers and also research into porphyria.

The BPA felt this was entirely possible as the BPA aims are very similar, and were delighted to accept this offer. A few weeks ago, a tremendous sum of £8,635 was credited to the BPA bank account. We will be looking into the best ways to utilise these funds over the next few months.

The BPA are delighted to announce that Kuehne + Nagel donated a huge £1000 to support the BPA in December 2016. **VERY SPECIAL THANKS** go to Paula Fowler and Penny Ward who both (unbeknown to each other) nominated the BPA to receive Kuehne + Nagel's annual Christmas donation.

# BREXIT AND GENETIC DISORDERS (*Cont.*)

**Regulation:** EU regulations set frameworks for research, including clinical trials, databases, medicines, devices and safety. They make it cost-effective to bring innovative new therapies to rare disease patients, e.g., the authorisation process for drugs for all 27 countries takes place through the EMA (European Medicines Agency) – once we exit, we must leave the EMA. However, AMRC hopes that the relationships and frameworks will remain aligned and compatible and that collaboration will remain. It was noted here that there is the potential opportunity to speed up processes or do things differently, such as diverge in areas of emerging research. A balance will need to be struck between the wish to remain aligned and the potential opportunity to diverge.

In terms of research, the greater numbers of patients in a wider EU population makes clinical trials more effective. Rare diseases research would be difficult in small numbers present in a singular country.

Overall, it was noted that ministers themselves have no idea of the outcomes, or even in some cases what would be wanted from the outcomes. It is hopeful that organisations such as AMRC can guide the process and help to mould the outcomes.

# UPDATE ON SCENESSE®

We recently reported to members that the progress of SCENESSE® through NICE (National Institute for Health and Care Excellence) had slowed after it was recommended that the drug be assessed under their Single Technology Appraisal (STA) scheme rather than Highly Specialised Technology (HST) process.

This was largely due to NICE believing that more than 1,000 people in the UK were affected by EPP. The BPA, EPP specialists in the UK, and Clinuvel (the drug company who makes SCENESSE®) believed that the figures NICE used in their assessment were incorrect and all appealed this decision.

We have just received the good news that the drug has been reconsidered and it will now be evaluated as a HST. The evaluation is expected to start mid-May 2017 when the BPA will be invited to get involved with the process – although we don't know exactly what that will involve yet. As soon as we know more about how EPP patients can help, we will let you all know. The rapid start date is great news for EPP patients.

In other parts of the UK, the All Wales Medicines Strategy Group and the Scottish Medicines Consortium (who play a similar role to NICE in their respective parts of the UK), have each asked the BPA for input into their decision-making processes regarding SCENESSE®. We have spent a lot of time and enlisted as many patients as possible to ensure that EPP patient needs are fully expressed. Although these processes are still at an early stage and we are unlikely to hear much for a number of months, it is good that there are developments occurring in all parts of Great Britain.

We will keep on bringing you more news as we hear it.

# FUNDRAISING AND AWARENESS

There are lots of events happening this year to raise funds for the BPA and raise awareness of the porphyrias. Some of you will have been involved in International Porphyria Awareness Week (IPAW – 22-29 April 2017). Let us know what you have done and how it went! Updates to follow in our next newsletter.

**Active events:** This year people are taking part in some of the following events: Hampton Court Half Marathon, Total Warrior, Ride London-Surrey 100, Great North Run, Great South Run and Birmingham Marathon to name just a few. Most of our fundraisers have Just Giving pages if you would like to support them. Please go to *www.justgiving.com/britishporphyriaassoc*.

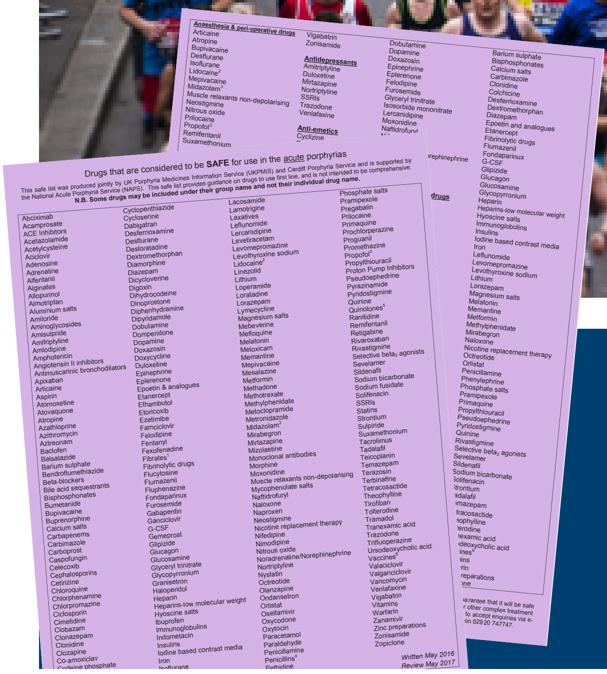
**Would you like to take part in an active challenge? We currently have the following official places available:**

- Birmingham Marathon (Sun 15 Oct 2017): 1 place
- Great North Run (Sun 10 September 2017): 4 places
- Great South Run (Sun 22 October 2017): 5 places

Places are allocated on a first come, first served basis, so please let us know ASAP if you or someone you know is interested on *liz.gill@porphyria.org.uk* and we'll send you more information.

**Van Show:** For the fourth year running, Ray Lancaster and Jenny Coombs are organising the Revolution Custom & Classic Van Show in Skegness (9-10 September 2017), to raise funds for the BPA. Van owners and enthusiasts will be coming from all over the country to show off their custom vans. There will be entertainment and competitions for adults and kids! Starts at 10am each day.

The event will be held at the Pine Trees Leisure Park, Croft Bank, Skegness Lincolnshire, PE24 4RE (Tel: 01754 762949). *www.pinetreesholidays.co.uk*. Anyone interested in helping with the event/a stall can contact Ray on 07799 392 771 or *magicoazulcc@yahoo.co.uk*. If any business or individual has a raffle prize that they would like to donate, then please let Ray know.



Drugs that are considered to be **SAFE** for use in the acute porphyrias

This safe list was produced jointly by UK Porphyria Medicines Information Service (UKPMIS) and Cardiff Porphyria Service and is supported by the National Acute Porphyria Service (NAPS). This safe list provides guidance on drugs to use first line, and is not intended to be comprehensive.

**N.B. Some drugs may be included under their group name and not their individual drug name.**

Abciximab
Acamprosate
ACE Inhibitors
Acetazolamide
Acetylcysteine
Aciclovir
Adenosine
Adrenaline
Alfentanil
Alginates
Allopurinol
Almotriptan
Aluminium salts
Amiloride
Aminoglycosides
Amisulpride
Amitriptyline
Amlodipine
Amphotericin
Angiotensin II inhibitors
Antimuscarinic bronchodilators
Apixaban
Articaine
Aspirin
Atomoxetine
Atovaquone
Atropine
Azathioprine
Azithromycin
Aztreonam
Baclofen
Balsalazide
Barium sulphate
Bendroflumethiazide
Beta-blockers
Bile acid sequestrants
Bisphosphonates
Bumetanide
Bupivacaine
Buprenorphine
Calcium salts
Carbapenems
Carbimazole
Carboprost
Caspofungin
Celecoxib
Cephalosporins
Cetirizine
Chloroquine
Chlorphenamine
Chlorpromazine
Ciclosporin
Cimetidine
Clobazam
Clonazepam
Clonidine
Clozapine
Co-amoxiclav
Codeine phosphate

Cyclopenthiazide
Cycloserine
Dabigatran
Desferrioxamine
Desflurane
Desloratadine
Dextromethorphan
Diamorphine
Diazepam
Dicycloverine
Digoxin
Dihydrocodeine
Dinoprostone
Diphenhydramine
Dipyridamole
Dobutamine
Domperidone
Dopamine
Doxazosin
Doxycycline
Duloxetine
Epinephrine
Eplerenone
Epoetin & analogues
Etanercept
Ethambutol
Etoricoxib
Ezetimibe
Famciclovir
Felodipine
Fentanyl
Fexofenadine
Fibrates[1]
Fibrinolytic drugs
Flucytosine
Flumazenil
Fluphenazine
Fondaparinux
Furosemide
Gabapentin
Ganciclovir
G-CSF
Gemeprost
Glipizide
Glucagon
Glucosamine
Glyceryl trinitrate
Glycopyrronium
Granisetron
Haloperidol
Heparin
Heparins-low molecular weight
Hyoscine salts
Ibuprofen
Immunoglobulins
Indometacin
Insulins
Iodine based contrast media
Iron
Isoflurane

Lacosamide
Lamotrigine
Laxatives
Leflunomide
Lercanidipine
Levetiracetam
Levomepromazine
Levothyroxine sodium
Lidocaine[2]
Linezolid
Lithium
Loperamide
Loratadine
Lorazepam
Lymecycline
Magnesium salts
Mebeverine
Mefloquine
Melatonin
Meloxicam
Memantine
Mepivacaine
Mesalazine
Metformin
Methadone
Methotrexate
Methylphenidate
Metoclopramide
Metronidazole
Midazolam[3]
Mirabegron
Mirtazapine
Mizolastine
Monoclonal antibodies
Morphine
Moxonidine
Muscle relaxants non-depolarising
Mycophenolate salts
Naftidrofuryl
Naloxone
Naproxen
Neostigmine
Nicotine replacement therapy
Nifedipine
Nimodipine
Nitrous oxide
Noradrenaline/Norephinephrine
Nortriptyline
Nystatin
Octreotide
Olanzapine
Ondansetron
Orlistat
Oseltamivir
Oxycodone
Oxytocin
Paracetamol
Paraldehyde
Penicillamine
Penicillins[4]
Pethidine

Phosphate salts
Pramipexole
Pregabalin
Prilocaine
Primaquine
Prochlorperazine
Proguanil
Promethazine
Propofol[3]
Propylthiouracil
Proton Pump Inhibitors
Pseudoephedrine
Pyrazinamide
Pyridostigmine
Quinine
Quinolones[5]
Ranitidine
Remifentanil
Retigabine
Rivaroxaban
Rivastigmine
Selective beta$_2$ agonists
Sevelamer
Sildenafil
Sodium bicarbonate
Sodium fusidate
Solifenacin
SSRIs
Statins
Strontium
Sulpiride
Suxamethonium
Tacrolimus
Tadalafil
Teicoplanin
Temazepam
Terazosin
Terbinafine
Tetracosactide
Theophylline
Tirofiban
Tolterodine
Tramadol
Tranexamic acid
Trazodone
Trifluoperazine
Ursodeoxycholic acid
Vaccines[6]
Valaciclovir
Valganciclovir
Vancomycin
Venlafaxine
Vigabatrin
Vitamins
Warfarin
Zanamivir
Zinc preparations
Zonisamide
Zopiclone

*Written May 2016*
*Review May 2017*

**Anaesthesia & peri-operative drugs**
Articaine
Atropine
Bupivacaine
Desflurane
Isoflurane
Lidocaine[2]
Mepivacaine
Midazolam[3]
Muscle relaxants non-depolarising
Neostigmine
Nitrous oxide
Prilocaine
Propofol[3]
Remifentanil
Suxamethonium

Vigabatrin
Zonisamide

**Antidepressants**
Amitriptyline
Duloxetine
Mirtazapine
Nortriptyline
SSRIs
Trazodone
Venlafaxine

**Anti-emetics**
Cyclizine

Dobutamine
Dopamine
Doxazosin
Epinephrine
Eplerenone
Felodipine
Furosemide
Glyceryl trinitrate
Isosorbide mononitrate
Lercanidipine
Moxonidine
Naftidrofuryl
rephinephrine

**drugs**

Barium sulphate
Bisphosphonates
Calcium salts
Carbimazole
Clonidine
Colchicine
Desferrioxamine
Dextromethorphan
Diazepam
Epoetin and analogues
Etanercept
Fibrinolytic drugs
Flumazenil
Fondaparinux
G-CSF
Glipizide
Glucagon
Glucosamine
Glycopyrronium
Heparin
Heparins-low molecular weight
Hyoscine salts
Immunoglobulins
Insulins
Iodine based contrast media
Iron
Leflunomide
Levomepromazine
Levothyroxine sodium
Lithium
Lorazepam
Magnesium salts
Melatonin
Memantine
Metformin
Methylphenidate
Mirabegron
Naloxone
Nicotine replacement therapy
Octreotide
Orlistat
Penicillamine
Phenylephrine
Phosphate salts
Pramipexole
Primaquine
Propylthiouracil
Pseudoephedrine
Pyridostigmine
Quinine
Rivastigmine
Selective beta$_2$ agonists
Sevelamer
Sildenafil
Sodium bicarbonate
Solifenacin
Strontium
adalafil
emazepam
tracosactide
eophylline
terodine
nexamic acid
odeoxycholic acid
cines[6]
mins
rin
reparations
one

uarantee that it will be safe
r other complex treatment
to accept enquiries via e-
on 02920 747747.

# ACUTE PORPHYRIAS: SAFE DRUGS LIST

The SAFE drugs list produced by the UK Porphyria Medicines Information Service (UKPMIS) is updated each year. It is important that acute patients keep an up-to-date copy with them. Please contact 02920 743877 or 02920 742251 to be added to the UKPMIS database to receive your new copy each year. The 2017 version will come out in May and will be lilac.

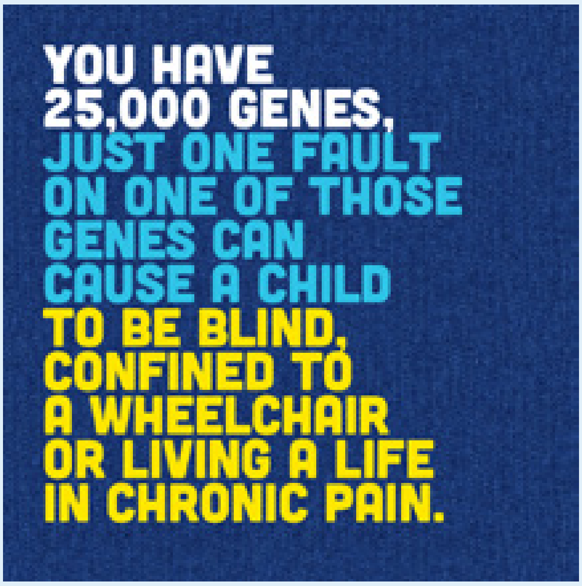
# JEANS FOR GENES DAY

**Friday 22 September 2017**

Lots of rare disease charities are joining together in the hope that increased numbers can make us all more effective. Jeans for Genes is something many of you will have heard of before, but now they are inviting smaller charities to help share the brand and raise awareness together.

A Jeans for Genes Day is a great way to bring a whole school or workplace together and raise money for Genetic Disorders UK and the BPA at the same time. The day invites people to leave their suits and uniforms at home and wear their jeans to work or school in return for a £1 donation which will help change lives.

Go to *www.jeansforgenesday.org* to sign up for a free fundraising pack. Don't forget to choose the British Porphyria Association as your affiliated genetic disorders charity, so that we get 50% of any income raised.



# UPDATE ON ALNYLAM

Alnylam Pharmaceuticals have now renamed their potential drug for treating recurring **acute attacks** of porphyria as **Givosiran** (it was known as ALN-AS1 before). The clinical trials in Phase 1 are looking promising. In Parts A & B, the drug was given to people who carry the gene and don't have attacks, but have high levels of ALA and PBG (known as "asymptomatic high excreters"). Givosiran reduced their levels of ALA and PBG, which is what is needed.

Part C gave two different doses to two groups of symptomatic AIP patients (those having frequent attacks). It reduced the annualised attack rate compared to the run-in period. The results for a third group aren't yet complete. The drug seems to cause no serious side effects, and people generally tolerate it well.

Alnylam announced in March 2017 that the European Medicines Agency (EMA) has granted access to its Priority Medicines (PRIME) scheme for Givosiran. The purpose of the PRIME initiative is to bring treatments to patients faster, for diseases where there is an unmet medical need and where early clinical data is showing potential benefits to patients. The results from the ongoing Phase 1 study formed the basis of the application for PRIME.

The study is continuing and as more results emerge, we will keep members posted. As with all research, even when a drug has shown potential, the processes to get a drug to the market assessment stage are lengthy and require strict adherence to regulations (for safety reasons). Givosiran is still at a very early stage of clinical development. Nonetheless, Alnylam are hoping to advance the medicine into a Phase 3 trial in late 2017.

They are also starting some market research, being done via a company called Cello Health Insights, to learn more about how people with acute porphyria are affected. For more information, please see *www.alnylam.com*.

# UPCOMING EVENTS

## INTERNATIONAL PORPHYRIA PATIENT CONFERENCE

**Bordeaux, France**
**Sunday 25th June 2017**

The event is being organised by the French patient support group, *Association Française des Malades Atteints de Porphyries.* The whole day will be presented in English as well as French. Previous events (held every two years in different countries) have been excellent and extremely useful. There will be talks by porphyria experts and patients, and a chance to meet both from other countries. The day is free to attend, but advance booking/registration is compulsory. Please see *https://icpp2017.org/registration-patient-day/*. If you would like to go, please register as soon as possible.

## BPA AUTUMN CONFERENCE

**Holiday Inn – CARDIFF North (by M4 Jct. 32), Merthyr Road, Tongwynlais, Cardiff, CF15 7LH**
**Saturday 7th October (09.30 – 16.15)**

The programme is yet to be finalised, but will include both doctor and patient talks (as well as our brief AGM). There will be updates from Clinuvel and Alnylam (Clinuvel have developed a treatment for EPP and Alnylam are developing one for acute porphyrias), as well as patient experience talks on both acute and skin porphyrias. There will be plenty of opportunities to speak with other patients and members.

The **Autumn Conference is free to attend, but booking is essential.** If you would be interested in attending, please forward the names of anyone wishing to come with you to *liz.gill@porphyria.org.uk* or call 0300 30 200 30 to confirm your attendance. We will send further details out a short while before the event.

If you wish to stay at the hotel before or after the event, the BPA have been able to secure preferential rates of £75 per room/night for bed and breakfast; a number of rooms have been provisionally allocated for the Friday and Saturday, these will be held until 1st September – they are available on a first come, first served basis. Please contact the hotel direct on 02920 522262, and ask for Gemma. The booking code is BPA071017. Any rooms booked after that date will be subject to the best available rates. **Please note that these rooms are payable at the time of booking and are non-refundable.**

We very much hope to see you at one of these events.

***Travel bursary:*** *The BPA do have some funds to help support our members to attend meetings. 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).*

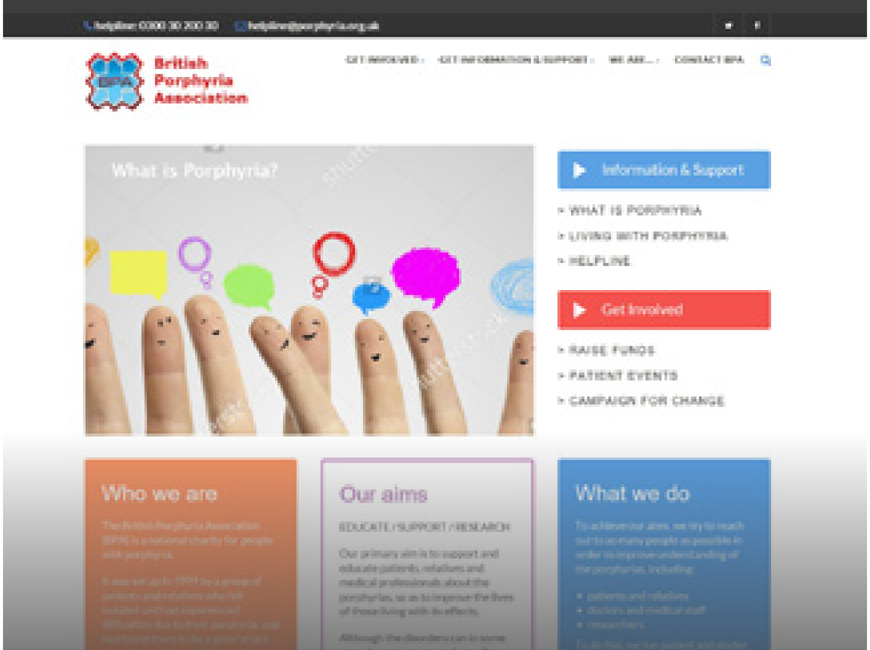
# HELPLINE
## 0300 30 200 30

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*
email: *helpline@porphyria.org.uk*
address: BPA, 136 Devonshire Rd
Durham City, DH1 2BL



# LAUNCH OF OUR NEW WEBSITE

**We're excited to announce that our newly refreshed website is now live!**

Our old website was designed a number of years ago and was not very mobile friendly. In addition to now being easier to use on small screens, the updated site includes changes to navigation. We've also made some small but significant changes to the structure and content. We'll be continuing to update the existing content, so keep coming back to see more changes as they happen: www.porphyria.org.uk.

## 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*
☐ 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