

# THE BRITISH PORPHYRIA ASSOCIATION NEWSLETTER

Registered Charity No. 1089609



## NEW RESEARCH: WHY DO SOME PATIENTS GET ACUTE ATTACKS AND OTHERS DO NOT?

This question has been troubling us all for a very long time. We know that there are triggers, such as alcohol and drugs, but some people seem to be able to tolerate these triggers while others cannot.

Most acute intermittent porphyria patients who have attacks have a mutation in their acute intermittent porphyria gene but so do some of their relatives who never get any symptoms. So, we are wondering: "Is there a defect in another gene which, together with the one we know about, causes acute attacks?"

Porphyria is caused by a defect in one of the enzymes in the pathway that makes haem. There is an enzyme that controls this pathway in the liver, known as ALAS1, and this seems to be an obvious place to start looking.

We will use two groups of patients.

1. Patients who have been admitted to hospital with a biochemically confirmed acute attack.

2. Relatives of these patients over the age of 45 who have the familial mutation in the acute intermittent porphyria gene but have never had an attack. It has been very difficult to decide who should go into this second group. We anticipate that most people who are going to have an attack will have had one by age 45, although we do know that some, for whatever reason, present later. However if we make the age limit too high we will not have enough patients to test.

The samples will be anonymous and we will test 20 individuals.

The British Porphyria Association has provided us with a grant for this investigation. We will use techniques

called polymerase chain reaction (PCR) and fluorescent sequencing and look for any changes in the gene. Everyone has changes in their genes and the difficult part will be determining if any of the changes will affect the process of making haem. Any potentially significant changes will be investigated as part of a larger future study.

### Wish us luck!

*Sharon Whatley: Chemical Biochemist, University Hospital of Wales*

***"The BPA are delighted to be supporting Sharon Whatley's research which will be conducted at the University Hospital of Wales. We will continue to provide updates on this research, as and when they become available."***

## WITH THANKS

### DANCE PERFORMANCE RAISES £700!

Sarah Chapman and friends at the Valerie Shepherd Dance Academy raised £700 for the BPA by holding a dance performance at the Customs House, South Shields. Both Sarah and her brother, Matthew, suffer from EPP. Over 90 children took part to an enthusiastic audience! The BPA would like to extend a massive thank you to all involved in putting the performance together and helping to continue to raise awareness and funds.



### HELL DOWN SOUTH!

Michael Lake and his friends, Alex Gatty and Stephen Smith, ran the Hellrunner Down South to raise funds for the BPA this year. It is a half-marathon run through mud, bogs, freezing cold chest-deep water and across hills all in January – Brrrrr! The BPA would like to give them a huge thank you for raising a whopping £1,497.



### RIDE LONDON-SURREY 100 – 10 AUGUST 2014

We are pleased to announce that we have filled our two places for the annual Ride London-Surrey 100. Ian Burrell and Dougal Betts will be training hard over the coming months in order to be fully prepared to ride 100 miles in one day on Sunday 10 August 2014. They will also be working hard to generate their sponsorship goal of £1,000 for the BPA. You can follow their training and journey by looking at their JustGiving page: [www.justgiving.com/Ian-and-Dougal](http://www.justgiving.com/Ian-and-Dougal) where you can also sponsor them if you would like.



### IN MEMORY

The BPA would like to thank friends and family members of Mr David Christiansen, who donated £80 in his memory.

### CUSTOM VAN SHOW: WEEKEND 13/14 SEPTEMBER 2014

Ray Lancaster, one of our loyal BPA members and porphyria sufferers, has organised the Revolution Custom & Classic Van Club Show, which will be held in Skegness on the weekend of the 13 and 14 September 2014, to raise awareness and funds for the BPA. Van owners and enthusiasts will be coming from all over the country to show off their vans: self-builds, work vans, pick-ups, works in progress, and characterful wrecks! A rough schedule of events is as follows, more details will be on our website in due course.

#### Saturday:

- Meet up and display vans
- Entertainment and competitions (for adults and kids): blind driving, ball in a bowl, and much more!

- Evening drive through Skegness

#### Sunday:

- Leisure morning until noon
- Final spit 'n' polish before parading vans on the centre field
- At 2pm trophies will be awarded, raffle drawn, etc. before close down/pack up

Thanks go to Ian Clarke from the Leisure Park, who has donated the venue for the event and a raffle prize of one week's camping/caravan pitch with electric hook-up.

The event will be held at the Skegness Water Leisure Park, Lincolnshire, PE25 1JF (Tel: 01754 899400). For an overnight stay at the camping/caravan site, bookings are £18 a pitch or £22 with electric hook-up. Bookings are to be

made directly with the venue, quoting ref: BPA.

The BPA would love members to attend and show their support; if successful, Ray hopes the event will be repeated annually. Additionally, anyone who is interested in helping with the event/a stall can contact Ray (details below) or Liz Gill on 0300 30 200 30. The BPA will be holding a small stall of merchandise and will be available periodically over the weekend to offer support and answer any questions that may arise.

If any business or individual has a raffle prize that they would like to donate, then please let us know.

For more information, or to buy raffle tickets (£2 for a book of five) please contact Ray Lancaster on 07799 392 771 or [magicoazulcc@yahoo.co.uk](mailto:magicoazulcc@yahoo.co.uk).

# FUNDRAISING IN 2015 – WE NEED YOU!

If you have been inspired by any of these fundraising journeys then you may like to consider setting up your own fundraising event to raise funds for the BPA. If you would like to know more, please look at our fundraising pages on our website: [www.porphyria.org.uk/?page\\_id=253](http://www.porphyria.org.uk/?page_id=253).

We are delighted to have secured a further two charity places for the Ride London-Surrey 100 in August 2015. In order to apply for this cycling event, you need to be able to ride 100 miles in less than 9 hours, be confident in riding in a group and be able to raise around £500. If you (and maybe a friend) would be

interested in applying for a BPA charity place for this amazing sporting event in 2015, then please get in touch via: [helpline@porphyria.org.uk](mailto:helpline@porphyria.org.uk) or via: 0300 30 200 30. These places will go on a first come, first served basis, so please get in touch to indicate that you are interested – **Good Luck!**

## EPP: SAM AND ME

I have had EPP, and been light sensitive, for 49 of my 50 years. I tended to 'self-manage' with variable success through much of my life, only achieving a diagnosis when I was 22.

When my children came along: Jo in 1997 and Sam in 2000, I promised that if one or both became light sensitive, I would make the utmost effort to find ways to make life as bearable as possible. Although they both carry the EPP gene, it was Sam, at the age of eight, who suddenly became light sensitive after a school trip, during an unseasonable heat wave. Having being completely distraught at what I had caused, I did 'man up' and start my quest to find the best help for Sam. Needless to say, it hasn't been straightforward, but these are the things I have learned. This is, of course, a very personal view and is looking from a post-diagnostic viewpoint. Also, the aid we have received has been primarily because Sam is a child, I have not tried to apply for support myself – I have a feeling it would be less successful.

A referral to see an occupational therapist (OT) was helpful (a GP or consultant can help with this). The OT came to the house to view Sam in his home environment and led to our County Council (Oxfordshire) paying for our windows to be light proofed with film. Additionally, the OT suggested we apply for Disability Living Allowance (now Personal

Independence Payment or PIP) – it is a dauntingly long form, but we were successful and Sam receives an allowance. It is the lowest level but every little helps – it keeps him in gloves! Within our county, mobility assistance would not be paid for, but we were able to have the car light proofed due to a charitable donation. The BPA does provide grants, but local charities are also worth contacting – often they like to help families in the local community.

The XP Support Group (<http://xpsupportgroup.org.uk>) has a

**AFTER SEVERAL ATTEMPTS, SAM NOW HAS A MUCH-PRIZED BLUE MOBILITY BADGE – THIS IS THANKS TO OUR CONSULTANT, DR SHERU GEORGE WHO HAS A PASSION FOR MAKING DAILY LIFE MANAGEABLE**

wealth of resources for people with light sensitivity. Xeroderma Pigmentosum, although an unrelated condition, also requires vigorous shielding from sunlight and the website has lots of useful information and links.

After several attempts, Sam now has a much-prized blue mobility badge – this is thanks to our consultant, Dr Sheru George who has a passion for making daily life manageable. We did have to go for a lengthy assessment as Sam did not 'fit the boxes'. We had to be shut in a tiny room at the assessment centre because all the light bulbs were open coil, low energy bulbs and Sam was on fire. I think it helped to illustrate

his (our) problems! Having the car light proofed was a key point as it is our 'safe haven' and parking it close to buildings was central to the application.

I have put off talking about Sam's school experiences. I expect people reading this who have children with EPP will guess why! I have provided letters from our GP and Consultant and all the information that I think the school needs and, although the learning support staff work tirelessly to make Sam's school time manageable, he is at a

very large school and ensuring teachers understand his problems and that they follow risk assessments is stressful and challenging. Our sanity has been saved by a number of core staff who work tirelessly to help him and a robust protocol that Sam can revert to. This means he can walk out of a class at any time to keep himself safe. However, after three years it is frustrating that he still has to do this. Any classes with light bulbs that he can't tolerate have been modified, so that he has a safe place to sit – it should work seamlessly but glitches occur. PE is currently impossible.

With respect to treatment, since Sam became light sensitive he has undergone UV

treatment and I now have it too. We are just about to start our sixth year of treatment. I believe it helps me, but it is difficult to judge how successful it is for Sam as his symptoms are still developing and, unfortunately, his skin is deteriorating. I believe the treatment does provide an element of protection. Personally, I have not found sun cream helpful or anaesthetic cream for pain relief, however I know that others find both successful. This is such a personal condition with individual variations. Some years ago, I tried beta-carotene, but turned bright orange and had hideous stomach problems.

Finally, a word about my personal nemesis: holidays. Sam and I are light sensitive, but Jo, who is 16, is not and is a sporty sun worshipper. What we have worked out so far is that camping doesn't work, but cruises do – lots of sun and deck activities for Jo and lots of below deck activities for Sam (ice rink, films, clubs, gymnasium), oh and cocktails for me! We have been very lucky to have one such holiday, but I don't think another one is on the cards due to cost. We would love to camp as we are festival freaks. Every year we try camping at a very local folk festival, but always with disastrous results. The reality is that tents equal light. If anyone with EPP has found a really protective tent, do let us know!

# 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.porphyrria.org.uk](http://www.porphyrria.org.uk)  
email: [helpline@porphyrria.org.uk](mailto:helpline@porphyrria.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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## BPA EVENTS 2014

The BPA are pleased to announce that we have two events scheduled this year.

On **Saturday 7 June 2014** we will be holding our **Open Day** at the University Hospital of Wales, Cardiff. The meeting is free to attend and will be held in the Sports and Social Club of the Cardiff Medical Centre, University Hospital of Wales, Cardiff, from 10am to 4pm. The programme will start with registration and refreshments at 10am, for a 10.30am start, and will finish with refreshments at around 4pm. Lunch will also be provided. There will be various talks from patients, a clinical psychologist, talks on acute and cutaneous porphyrias from porphyria specialists, as well as a talk on some new research. There will also be a question and answer session, and time for informal chats with other

porphyria patients and their families. A virtual tour of the laboratories, where the tests for porphyria are conducted, will also be given, as well as a real tour for anyone wishing to attend.

Our **BPA Autumn Conference and AGM** will be held at Salford Royal Hospital, Salford on **Saturday 6 September 2014** between 1.30 and 5pm. We are currently planning this event with the help of Professor Stewart and her team in Salford. As in previous years, we will start with a brief AGM and there will then be a number of clinical talks and a patient talk about living with porphyria.

**If you would like to attend either event, please contact us for more information and directions.**

**BPA Open Day**  
Saturday 7 June 2014  
University Hospital of Wales, Cardiff

**BPA Autumn Conference and AGM**  
Saturday 6 September 2014  
Salford Royal Hospital, Salford

THE BRITISH PORPHYRIA ASSOCIATION Registered Charity No. 1089609

## MEMBERSHIP / DONATION FORM & STANDING ORDER



Title \_\_\_\_\_ Name \_\_\_\_\_  
Address \_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_  
Postcode \_\_\_\_\_  
Email \_\_\_\_\_  
Telephone \_\_\_\_\_  
Type of Porphyria \_\_\_\_\_  
Date \_\_\_\_\_

### WAYS TO PAY

- I would like to pay my annual membership fee of £15  
 I would like to make a donation of:  
 £10  £15  £20  £25  £50 other £ \_\_\_\_\_  
 I would like to set up a standing order (please fill in the form opposite)  
 I have set up a standing order using my internet banking\*  
 I enclose a cheque made payable to the 'British Porphyria Association' for £ \_\_\_\_\_  
 I have made a payment using [www.justgiving.com](http://www.justgiving.com)  
 I would like a receipt

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

By making a donation to the British Porphyria Association you will know that you are making a vital contribution to our work. Simply making a small donation will help us raise awareness, man our helplines or support research.

### Do you pay UK tax?

If you pay UK tax, the BPA can reclaim 25p of tax on every £1 you give. This does not cost you anything and does not affect your personal tax position. Simply sign and date the Gift Aid declaration.

### Standing Order British Porphyria Association

**Please send this form to:** The Treasurer, British Porphyria Association, Rothlea House, 7-8 Quarry Lane, Butterknowle, Bishop Auckland, County Durham, DL13 5LL  
A monthly standing order or any donation you can give will make a difference.

Bank / Building Society name \_\_\_\_\_

Branch address \_\_\_\_\_

Postcode \_\_\_\_\_

Please pay the British Porphyria Association the sum of £ \_\_\_\_\_  
each month / quarter / year (delete as appropriate) from my account until further notice.

Account name(s) \_\_\_\_\_

Sort code \_\_\_\_\_ Account No. \_\_\_\_\_

Starting on\* (Date) \_\_\_\_\_

\*This date must be more than one month after today's date

Signed \_\_\_\_\_ Today's date \_\_\_\_\_

This cancels all existing standing orders to the British Porphyria Association  
(please tick)  yes  no  not applicable

Please pay to British Porphyria Association bank account:  
Sort code: 20-43-63 Account No:7099 6904

### Gift aid declaration

I am a UK tax payer and would like the British Porphyria Association (Registered Charity no. 1089609) to treat all donations that I have made in the last six years and all future donations that I make from the date of this declaration as Gift Aid donations, until I notify you otherwise. I understand I must pay an amount of income tax and/or capital gains tax equal to the tax reclaimed on my donations. (I will advise the BPA if my tax status, name or address changes).

Date \_\_\_\_\_ Signature \_\_\_\_\_