THE
BRITISH PORPHYRIA ASSOCIATION
NEWSLETTER

Registered Charity No. 1089609

GENETIC PROFILING

It is now possible to pay genetic testing companies to get all your genes analysed, telling you if you have mutations (changes) in any of them. The BPA has had enquiries from people who assume they have a porphyria, because they have a change in one of the relevant genes. Unfortunately, many people don't realise that most of the changes don't do any harm, since they don't get genetic counselling with the tests (something porphyria experts think is very important).

Specialists spend a lot of time trying to work out if these mutations mean anything or not, and even they do not always know for sure. Our porphyria experts do know many of the genetic changes that cause a porphyria so, once someone has been diagnosed, they may be able to test the genes of relatives to see if they are at risk. (Please see our Testing for Porphyria leaflet.)

However, as we know from porphyria families, many members have mistakes that do not cause any medical problems. When they do cause problems, samples of blood, urine or faeces have to be tested to confirm that the biochemistry matches their symptoms and that the porphyria is responsible for their illness. Genetic tests are unable to provide this information.

To understand a bit about how genes work, it helps to think of them as a kind of bar code, like shops use. Instead of a short list of numbers, the gene code gives a long list of amino acids (small molecules) which can then be joined in a chain to form a protein (a very, very large molecule). A change in a gene may mean that the 'wrong' amino acid is put into the protein chain.

Proteins curl up, almost like a ball of string, and the most important bits (active sites) are usually near the middle. The outer bits may be less important, and a single change there often does no harm. Changes near the active sites are more likely to cause harm.

Genetic profiling is relatively new and may be useful in years to come when we know more. If you wish to help with research which involves (free) profiling, without you getting the results, please do. Studies like this can help in the long term.

Unfortunately, at the moment, the paid-for profiling seems to be causing unnecessary anxiety. So, the BPA would advise people not to use this form of genetic testing.

THANK YOU AND CONGRATULATIONS

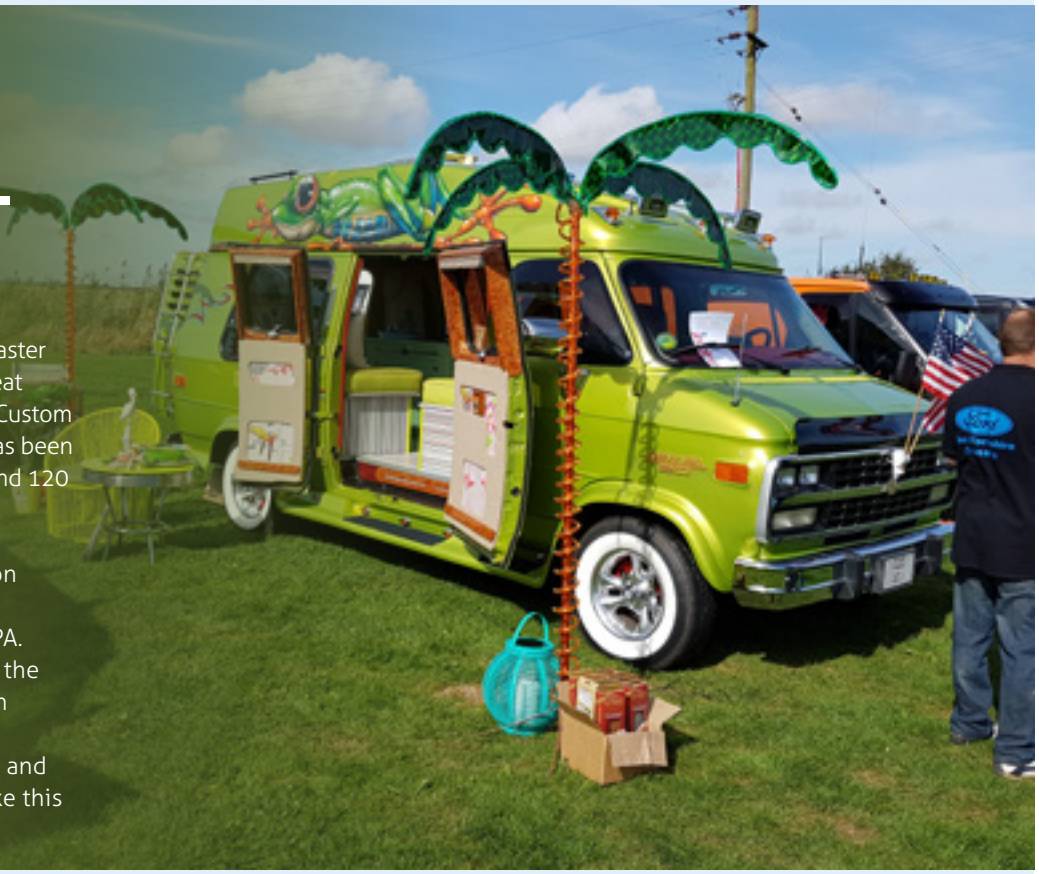
Thank you to anyone who has been involved in fundraising for the BPA and to all those people who make donations too (whether big or small, whether regular or sporadic), without you, the BPA would not be able to provide the support that it does to people with porphyria.

In particular, in this edition of the newsletter, the BPA would like to say a heartfelt THANK YOU to Dave Jones for his spectacular personal donation of £720. It is very much appreciated.

GREAT SOUTH RUN 2016

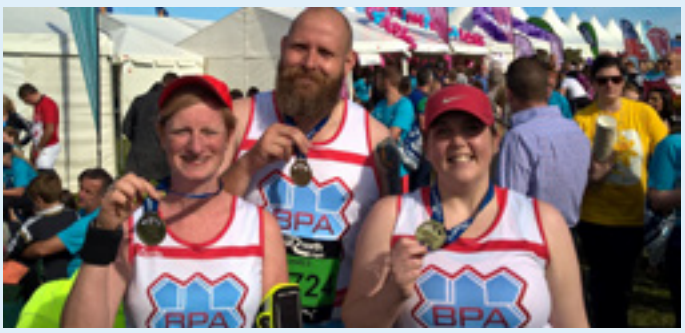
Our second big running event this autumn has been the Great South Run – for the first time we had five places for this event too. Michaela Davey, Simon Pay, Jamie Wentzell and Karen Harris (one of the BPA's trustees and committee member) all managed to run and raised together a great figure of £1076. Again, funds are still trickling in for the Great South Run, so we do not have a final total yet. Unfortunately, Gavin Tearle had to pull out after a last minute torn hamstring injury – ouch! But, he is going to do another event for us soon. Get well soon, Gavin!

CUSTOM VAN AND CAR RALLY – SKEGNESS



For the third year running, Ray Lancaster and Jenny Coombs organised a great event to raise funds for the BPA. The Custom Van and Car Rally held in Skegness has been growing each year and this year around 120 vans and cars attended.

Despite torrential rain and huge hailstones which put a dampener on the Saturday, they still managed to raise a fantastic £449.16 for the BPA. WELL DONE for repeatedly making the tremendous effort that you do each year to raise funds for the BPA and THANK YOU to all of the organisers and attendees who have helped to make this event such a success.



GREAT NORTH RUN 2016

The BPA were delighted to have five people running in the Great North Run this September – Emily Todd, Jim Gill, Debra Baker, Dan Stevenson and Mark Hopkins. On a beautiful and rather warm day, all runners completed the 13.1 miles with great times. The BPA would like to say a huge WELL DONE for all of your hard work and commitment, as well as a massive THANK YOU for managing to raise well over £2570 between you (total not yet finalised – as funds are still coming in).

UPDATE ON ALNYLAM'S ALN-AS1: AN INVESTIGATIONAL TREATMENT FOR THE ACUTE HEPATIC PORPHYRIAS

In September 2016, at an international scientific meeting in Rome, Alnylam presented data on its clinical progress with ALN-AS1. This is a subcutaneously administered (i.e. injected under the skin) RNAi therapy, for the treatment of acute hepatic porphyrias (AHP). It targets aminolevulinic acid synthase 1 (ALAS1) . The full presentation of data can be viewed at: **http://www.alnylam.com/web/assets/SSIEM-2016_AS1_Ph1_Capella_Deck_090716.pdf.**

Parts A and B of the ongoing Phase 1 study, called ALN-AS1-001, used participants that are 'asymptomatic high excreters' (termed ASHE). This demonstrated that single or double doses of ALN-AS1 was generally well tolerated. There were no serious side effects or clinically significant laboratory abnormalities related to the study drug.

In addition, ALN-AS1 administration resulted in rapid, dose-dependent lowering of aminolevulinic acid (ALA) and porphobilinogen (PBG), the toxic haem precursors that are thought to trigger porphyria attacks (see Clinicaltrials.gov: NCT02452372).

Alnylam is currently conducting Part C of the Phase 1 study, in individuals with acute intermittent porphyria (AIP) who experience recurrent porphyria attacks. Alnylam plans to present interim data from Part C in December 2016, with additional study data in 2017.

Alnylam has also completed enrolment in the EXPLORE study with a total of 112 participants. EXPLORE is a global study of individuals with AIP, variegate porphyria, and hereditary coproporphyria who experience recurrent attacks. Analyses are ongoing and will help improve understanding of the effects of the disease and the management of individuals with acute porphyria.

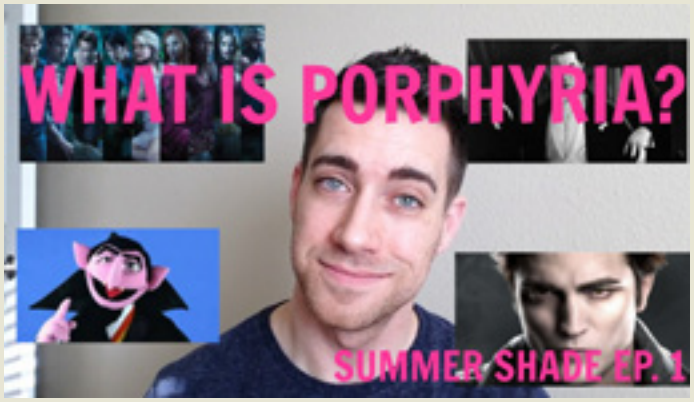
For more information about ALN-AS1, please visit: **http://www.alnylam.com/product-pipeline/porphyria/**. The BPA will continue to provide updates from Alnylam as and when they become available.

EPP VIDEO LOG BY JARED ULMER

If you suffer from EPP, or just want to know more about it, then take a look at the Porphyria J channel on You Tube. Each video is around 5 minutes long and there are 13 episodes which cover an impressive range of topics, including: What is EPP? What causes the reactions? What's it like to live with EPP? What's it like to be in a relationship and bringing up kids when you have EPP? and more... Although the session where Jared experiments with using a sunbed is quite radical (episode 10), this series is one of the most accessible resources we've come across. It's a very positive approach to raising awareness of EPP and great to call on when you can't face trying to describe the condition for the millionth time. If you suffer from EPP, watching the series is a must, it will help make you realise you are most definitely not alone.

Search You Tube for Porphyria J or visit **http://tinyurl.com/jzszl2h**

Full URL **https://www.youtube.com/channel/UC7SYTLc6RSGptlHHQ8Aapmg**



NICE APPROVAL FOR SCENESSE®

As reported in our last newsletter, the BPA have been awaiting news from the NICE (National Institute for Health and Care Excellence) review into whether SCENESSE® would be evaluated under the Highly Specialised Technology (HST) Programme for its introduction in England.

HST evaluations are recommendations on the use of new and existing highly specialised medicines within the NHS. The HST programme only considers drugs for very rare conditions.

NICE has recommended to the UK Department of Health that SCENESSE® is to be evaluated under the Single Technology Appraisal (STA) procedure. This means that the whole process effectively starts again and we are unlikely to hear anything until next year, when we will be invited to take part in another consultation.

In the meantime, Clinuvel are starting training in the specialist centres in the UK, so that as soon as a decision is made they are fully prepared. At present, SCENESSE® has been approved as a standard therapy in the Netherlands and Austria and they have started dosing and collecting data.

We will keep you updated on any further developments and will keep doing all we can to keep the process moving forwards.

BOB'S EPP STORY

Born in 1944, both my parents came from families of seven children; my father, born in 1907 and his younger sister born in 1909 had something wrong with their skin and suffered painful but non-visible burning when exposed to strong sunlight. The older five children had no such problem. My sister, 12 years older than me, had been confirmed as having the same strange affliction when she was young. However, there was no NHS in those days and we were a poor family, living in West Yorkshire. My aunt had no blood descendants of her own.

At 18 months old I contracted "bovine tuberculosis" from unpasteurised milk, and was brought up as "a sickly child". The industry in that area, textiles, engineering and chemicals, produced a permanent fog over the valley where I lived, limiting the sun for much of the year, but by the time I was five, I was found to have the same poor quality skin. After the formation of the NHS, we had a GP who turned out to be a really good heart specialist, but not so well up on skin! My childhood memories include the physical suffering of sun exposure when "playing out" (a strange concept where young children were allowed to be outdoors without adult supervision for long unconstructive periods, mixing sport with hide and seek and Cowboys and Indians) along with the mental stress of being different from the other kids. The Clean Air Act, which reduced the fog, only made my life more difficult.

Secondary school with the requirement to be outdoors at lunch and play time, along with 90-minute cricket in the summer, created more problems in explaining something I didn't understand myself.

Starting work as a student apprentice engineer was an indoors affair so no great problems, but late teens and connecting with girls on sunny days added to life's little difficulties. At 21, I graduated into external technical sales, with low spec company cars having no type of sun filter. When I married, my in-laws were very much into the outdoor life, and my mother-in-law never got over the possibility that her descendants might be "frightened" of the sun like me.

We moved to Manchester for my work and one day made an unplanned trip to the seaside with friends. I spent too long unprotected on the beach and got badly burnt. Back at our friend's house that evening, half a bottle of whiskey taken to numb the pain only made it worse. Having seen my condition, the friends took me to the Manchester and Salford Hospital for Skin Diseases where after several months of testing I was told that I had a type of porphyria. In 1973, I was sent for a week at Homerton Hospital in Hackney, where they tested various frequencies and powers of light on small sections of my back and confirmed, with great satisfaction, that I had Erythropoietic Protoporphyria. I was given the impression that nothing could be done about it, so just keep out of the sun. It was explained that it could be passed on from one generation to the next. Luckily, neither of my sons or my grandchildren seem affected; nor do my sister's children.

During the 1980s, I trialled beta-carotene for nearly three years, and displayed an amazing golden Hollywood glow. Eventually my optician detected related deposits in the back of my eyes and the testing was stopped. Sun lamp treatment around that time didn't seem to achieve much benefit.

Bringing up children who need plenty of time in the sun was difficult for me and involved my very patient wife playing with them while I sat in the shade.

Living in Bedford since 2007, I found that Addenbrooke's Hospital, Cambridge, had a team doing work on porphyria and have been treated there since in the hope of helping future generations.

I only discovered the BPA two years ago, and it's great to know that so many people are interested in helping us.

IMPORTANT DATES FOR 2017

BORDEAUX – SUNDAY 25TH JUNE 2017: INTERNATIONAL PORPHYRIA PATIENT MEETING

The International Porphyria Patient Meeting will be held in the beautiful city of Bordeaux, France. The French patient organisation is working very hard to develop an interesting conference for both the acute and cutaneous porphyrias. Further details will be announced in our Spring newsletter; but, if you may be interested in attending, please keep an eye on our website or Facebook page, as we will post updates once available.

CARDIFF – SATURDAY 7 OCTOBER 2017: BPA AUTUMN CONFERENCE AND AGM

Our Autumn Conference and AGM will be held in the historic town of Cardiff, with the support of Dr Mike Badminton and his team at the University Hospital of Wales. The exact location is still to be confirmed, as is the programme, but if you and your family may be interested in attending, please let us know: helpline@porphyria.org.uk or 0300 30 200 30. Further details will again be provided in our Spring newsletter, but do add the date to your diary now.

PORPHYRIA AWARENESS WEEK

After discussions with the American Porphyria Foundation (APF) and the International Porphyria Patients Network (IPPN), we are joining them in holding an International Porphyria Awareness Week (IPAW) from 17 to 23 April 2017. We are planning on suggesting a number of fundraising and awareness initiatives that you might like to become involved in. More details of these will be circulated in early 2017 via our website and Facebook page, as they become available.

We feel this will help strengthen the impact that we can have, not only on a local level, but on a global level too. If you have any ideas for awareness, publicity or fundraising that you would like to explore with the BPA, please do not hesitate to get in touch.

EVENTS IN 2016:

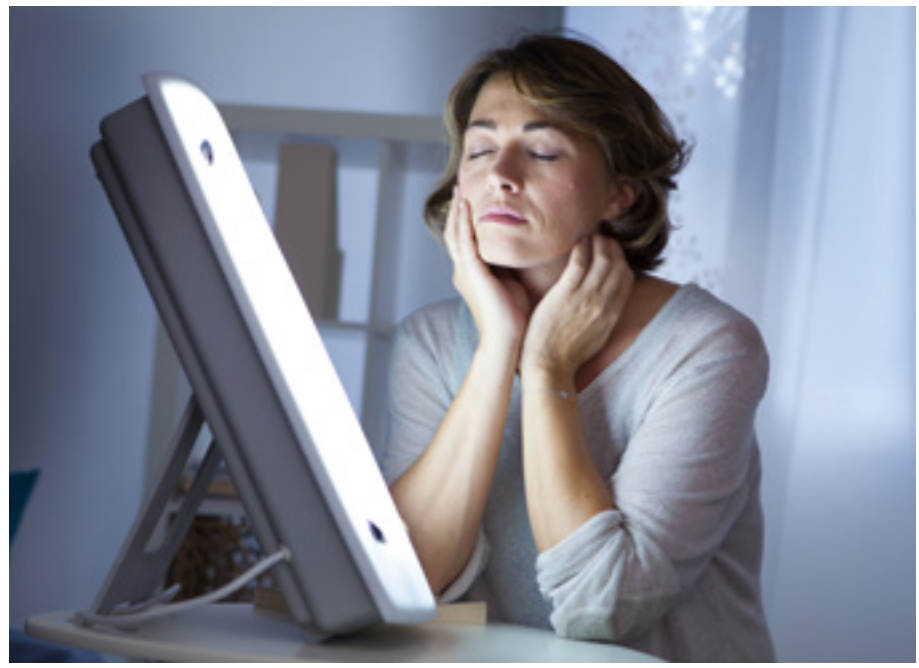
The BPA are pleased to announce that our two member events that were held in 2016 were very well received, as being informative, enjoyable and a great opportunity to meet others with porphyria. Orphan Europe provided a grant to help with these meetings which we are extremely grateful for. These funds helped significantly with the catering and room booking costs incurred on the day.

In July, we held our Open Day at Salford Royal Hospital and were delighted to have 45 people attend. We would like to thank all of the speakers: specifically, Professor Stewart and Professor Rhodes, and their teams, along with Dr Stein from NAPS, who all contributed to making the day a great success.

At the beginning of October, we held our Autumn Conference and AGM in Peterborough and were thrilled to have 60 people in attendance. Again, we are extremely grateful to all of the speakers/ organisers who helped to make the day so successful. Dr Stein and her team from Kings/NAPS were pivotal in providing content and support for the day, along with attendance from Alnylam, Clinuvel, BUPA and the Photobiology Unit in Salford.

We obtained some very positive feedback which we plan to integrate into our future events.

LIGHT PROTECTION FUND



As many of you may be aware, a research fund was set up a couple of years ago to raise monies towards home phototherapy units for a research project taking place in Cardiff. One unit was initially donated from the BPA's general funds and further funds were raised and saved, in the hope of purchasing others, using a specific 'Light Boxes' account. The home phototherapy units were to be used on EPP patients in a research project that aimed to improve access to phototherapy in areas where hospital units were inaccessible or had limited opening hours.

After some time, it has become evident that, unless used in photobiology departments that treat many different photosensitive ailments at different points during the year, these machines are not being used to their best potential. Furthermore, there are only a few departments across the UK that are able to take part in such projects, leading to a significant postcode lottery on whether EPP patients are able to gain access to home phototherapy.

With this in mind, the BPA decided that more people with photosensitive porphyrias would be able to benefit from this fund if it was opened it up to be a 'Light Protection Fund'. BPA members voted in agreement with this proposal when consulted at this year's AGM in Peterborough.

So, we are delighted to announce that we have funds available for light protection projects for members with photosensitivity. For example, window films on cars and homes can significantly improve the quality of life for those with EPP. If you would be interested in applying for a grant to help with the cost of installing a light protection measure, please do let us know on helpline@porphyria.org.uk or 0300 30 200 30.

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

The BPA Newsletter is published by the BPA twice yearly.

NAPS PATIENT SURVEY COMING SOON

Later this year the National Acute Porphyria Service (NAPS) will be sending out a patient survey to all of their patients. The aim is to gain an overall picture of how the service is running so that the service can be then improved for all NAPS patients. The NAPS team would greatly appreciate people taking the time to complete the survey and they welcome all comments and suggestions, whether good or bad.

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