

The British Porphyria Association newsletter



Registered Charity No.1089609

ISSUE 17 • NOV 2008



My name is Sandra.
I am 51 years old.

I was diagnosed with
Porphyria Cutanea Tarda (PCT)
in September 2003.

Sandra's Story

I was 45 years old and fit as a fiddle when I noticed that my urine was purple; I've had a hysterectomy so I was baffled and went to my GP.

He tested my urine and said that I had blood, sugar and protein in my urine. He gave me antibiotics for an infection, although I had no pain anywhere. I completed the course but the symptom continued.

One day my urine was deep red and I panicked. I bottled some and took it to the doctor. Again he tested it and gave me more antibiotics. To be honest, although I knew it wasn't right, I let it go for a few weeks.

I also had facial hair growing, which I put down to taking HRT, but I was going about my normal daily life and getting excited about my forthcoming holiday to Tenerife. Around 3 days before my holiday I noticed a blister on my hand. I knew that I hadn't hurt myself but I wasn't worried as it was only a blister.

I boarded the plane and to be honest the blister was annoying and was getting bigger, so I decided to release the fluid with a sterilised needle in the hope that it would heal. While on holiday the blister would not heal and was throbbing; I put it down to the heat and thought it would heal when I got home to Scotland.

Within a couple of weeks of returning both hands were throbbing painfully and clusters of blisters were grouping all over my hands. I returned to my doctor who gave me creams. It got to the stage that when I was driving, the heat coming through the windscreen caused terrible pain in my hands, which were now a mess and my skin had become very fragile. The least knock and the skin ripped. Then marks started to appear on my face and feet; my feet were so swollen I couldn't wear shoes. I was a mess and in pain.

A couple of years down the line I wondered whether there were other people feeling the way I did.

I was tired and didn't feel or look like me. I was feeling old. I was back and forth to my GP, who didn't have any answers either. We were both baffled until one day I said, "Look, don't think I'm daft, but could there be a connection between my urine and this blistering?" Don't ask me what made me think of this but these were the only things occurring that I'd never had before. He said, "Sandra will you leave this with me? I'll take your files home tonight and have a look."

At 9am the next morning he phoned: "Sandra I think you have a metabolic disorder, I've made you an appointment at the hospital for tomorrow morning."

To cut this story short, consultants and doctors came and went looking at my hands checking my urine. "No need for a biopsy", one doctor said, "it's Porphyria Cutanea Tarda, she has milia", pointing at my hands. I was bewildered. What were they talking about? One consultant briefly explained it to me; all I remember him saying was something about porphyrins, ferritin levels and start venesection straight away. This is where they remove 500mls of blood from you and repeat it periodically until the ferritin level decreases.

I was told to stay out of the light and avoid alcohol and any medication. I felt that my life had been dragged from under me.

It's important for me to say that within one year of diagnosis of PCT I was diagnosed with Haemochromatosis, a blood condition that I was born with but had no indication of. This condition produces and stores iron in the blood and it attacks the main organs if not detected; my liver was partly damaged, but has repaired itself through venesection as my ferritin level reduced.

Haemochromatosis is the cause for my PCT which is the only porphyria which is acquired. Family should be tested for the faulty gene which causes Haemochromatosis as this can be passed on through generations.

I was so alone and frightened and had many questions, but then I heard about the BPA. The BPA showed compassion, understanding and empathy, they listened to me and gave me answers to my questions.

A couple of years down the line I wondered whether there were other people feeling the way I did. With help from the BPA I managed to attend an event held in Leeds. It was amazing to see other people with porphyria. I decided to join the committee in order to start up a support group in Scotland.

Special thanks to all of the committee for the work they tirelessly volunteer to do, giving up their own free time.

Sandra McNab.

PS - If something is worrying you, then it's not silly. Go to your GP.

www.porphyrria.org.uk

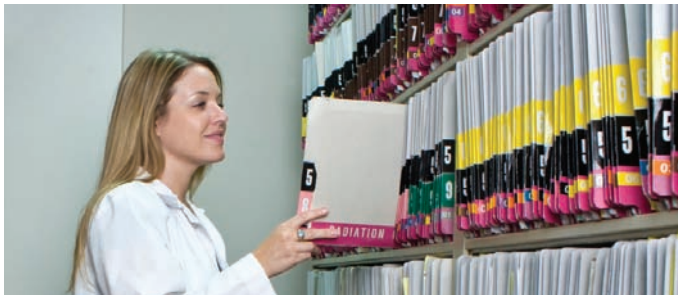


Porphyria Research...

We reported on the research study into AIP that we are supporting, on the front page of our last newsletter.

As well as this, we are glad to report below on two trials taking place on EPP.

The first is taking place in Manchester, and the second in London, but I think those with EPP anywhere in the country can take part.



Pilot investigation into the use of colestyramine as a therapy for patients with erythropoietic protoporphyria (EPP)

Dr Robert Sarkany, Consultant Dermatologist at St Thomas' Hospital in London is looking for EPP sufferers to take part in a research study. To be eligible you would need to be able to travel to London once a month for blood tests, the study will last for 7 months.

What is this study all about and what is the point of it?

In EPP, the pain you experience on exposure to sunlight is caused by a substance in the blood called 'protoporphyrin'. This is a normal body chemical but it is produced in excessive amounts in EPP. At the moment we cannot stop the protoporphyrin being produced in EPP. However, we hope that colestyramine, a medication, may help the body to get rid of protoporphyrin faster than usual, and so reduce the pain on sun exposure. Colestyramine has been used for many years as a treatment for people with high cholesterol.

Since most EPP patients do not get much benefit from the currently available treatments, we think it is important to know definitively whether this treatment is helpful or not to patients. To see whether the colestyramine helps in EPP, we will ask you to have a blood sample taken every month during the study, from which we will measure the amount of protoporphyrin in your blood. Since you will be taking the colestyramine in the middle of the study but not at the beginning or end of it, we will be able to see if the amount of protoporphyrin in your blood is lower when you are on the colestyramine treatment. This 'pilot study' we are asking you to take part in is needed to give us the information we need to get a useful answer ('yes it works' or 'no, it doesn't work') from the full study which will happen afterwards. The measurements of what happens to your blood protoporphyrin level will be used to work out how many patients are needed for the full study which will follow this 'pilot study'. The pilot study is a vital part of the research, without which we won't be able to find out if colestyramine works or not.

The study is organized and funded by the Photobiology Unit, St John's Institute of Dermatology, Guy's and St Thomas' NHS Trust. It does not involve any funding or involvement by any commercial body or pharmaceutical company. The doctors involved are not being paid to do this study.

Further information can be found on the BPA website or from Dr. R. Sarkany, Consultant Dermatologist, Photobiology Unit, St Johns Institute of Dermatology, St Thomas' Hospital, Lambeth Palace Road, London SE1 7EH. Tel: 0207 188 6389



A Study to Evaluate the Safety and Efficacy of CUV1647 Implants in Patients with Erythropoietic Protoporphyrin (EPP).

Clinuvel Pharmaceuticals Limited has a new drug currently in development that aims to help prevent the occurrence and reduce the severity of EPP reactions. The drug has been formulated as an implant (small rod) that is inserted under the skin above the hip, and slowly dissolves over a 2-3 month period.

If you do decide to participate in this study, you will receive six CUV1647 implants over 12 months. You will be required to visit the photobiology unit 15 times over 12 months, where you will be asked some questions about your EPP, have urine and blood taken for testing and undergo some skin measurements. In addition to these visits, a clinic visit is required the day after each implantation procedure, for collection of blood for safety monitoring. We intend to commence this study November 2008. You will be reimbursed for travel expenses.

**For further details please contact:
Clinical Research Nurse
Margaret Brownrigg
Tel: 0161 206 0457.**

FUNDRAISING...

Calling All Children, Grandchildren, Nieces & Nephews

Does the school that your children, grandchildren, nieces and nephews go to, raise funds for charitable purposes?

If they do, please approach them to see if they would be able to conduct a 'non uniform day' for the BPA.

Children are encouraged to pay between 50p and £1 to be able to wear their own clothes to school instead of their usual school uniform. Some schools may be more open to this idea if it is suggested that they keep some of the funds for their Parent Teachers Association.

This is a fantastic way to raise funds for the BPA, as well as increasing awareness of porphyria. It is very easy to administer, as each teacher can collect for their own class and one member of office staff can collate the coins and then send us a cheque.

We can supply printed information for the school about porphyria, which they can send home to parents explaining where the funds are going.

Please let us know if you will be organising something of this nature, especially if you would like us to provide some literature.



Great South Run - Portsmouth 26th Oct 2008

Thanks to everyone who sponsored Karen and Graham Harris for the Great South Run. They have raised over £250 for the BPA.

Karen says: "We had a great race and despite having to 'weave' our way past and through thousands of slower people, in the drizzle and wind.

We both finished in under 1 hour 27 minutes, with Karen in 3,668th place out of 20,000 runners, and Graham in 3,527th place. We were delighted to have reduced our times by more than 6 minutes over last year, and are already talking about entering again next year.

"Is there anyone who would like to join us? If you or someone you know could be capable of completing a 10 mile run, please do not hesitate to contact me for details of applying for a place, and training advice."

EBAY for Charity



Do you use EBAY to sell your unwanted items or to buy things at bargain prices?

If your answer is YES then PLEASE sell your items through the new Ebay for Charity website. You can access it by doing a simple Google search – ebay for charity - or use the following URL <http://pages.ebay.co.uk/ebayforcharity/>.

The site is easy to negotiate with a one, two, three step by step guide. You can donate as little as £1 (minimum donation) up to the full 100% of the sale price. You can still put on reserves, photos and all the usual information. To find the British Porphyria Association in the select a charity option, just put in BPA and it will find us and bring up our full details. You then need to click on select.

Once your item has been sold, you do not have to do anything. Ebay automatically pays the commission to the BPA through Missionfish.

So come on folks, have a good look in your cupboards and lofts to find anything that you think you could sell, raising some cash for you and helping us raise some funds, to enable us to continue to support patients, families, education and research.



BPA MEETINGS

Open Day at Leeds, and AGM & Conference at Liverpool.

We had a very good Open Day in Morley, Leeds, organised by Keith Allen, Consultant Clinical Scientist at Leeds.

Talks and questions covered the full range of porphyrias, treatments available and under test, and lab tests (what they can and can't do). They then kindly showed us around their labs where they do the porphyria testing.

Food and drink for the day were given by Orphan Europe, ordered by Sarah Elgott (who did the same again for us at the AGM). Many thanks to Keith and Sara for their help, and to the medical and lab staff who gave talks and conducted us round the labs. It was an ideal day for those with skin sensitivity, as it was raining!

Our AGM was less well supported by members. The AGM itself was very short, as usual, since there was no change in committee.

Then Sara Elgott showed a DVD produced by Orphan, explaining the use of haem arginate. Dr Helen Murphy up-dated us on the AIP study we are supporting; and Dr Felicity Stewart gave us a brief description of the porphyria service at Hope Hospital in Salford.

Dr Stewart was interested to find out what we wanted from the porphyria service, so we then each had to write two post-its, one with a good experience with treatment, and one with a bad experience. We stuck these on the front of the lectern, so everyone else could read them, and then had a discussion on what we wanted a porphyria service to do for us. Dr Stewart got a list of about ten items to take back and think over. It was obvious that many people still have problems with GPs and local hospital staff, especially with them misunderstanding the effects of a porphyria.

Unfortunately, we don't have room for full reports on the talks. If you would like more details, please look on our web-site, or contact us for a paper copy.

AGM & Conference, 2009

Next year we are holding an Autumn conference in central London, in October 2009.

We are hoping to have talks by three medical experts in porphyria. They are Drs David Rees, Joanne Marsden and Bob Sarkany

We have heard all three before, and are sure they will give useful and informative talks.



HELPLINE

01474 369 231

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 name, phone number, day and time of message and someone will call you back as soon as possible.

Web: www.porphyrria.org.uk

E-mail: helpline@porphyrria.org.uk

**Address: BPA, 136 Devonshire Rd
Durham City, DH1 2BL.**

The BPA Newsletter is published by the BPA twice yearly.

Chairman: John Chamberlayne

Treasurer: Alan Molyneux

Secretary: Sarah Pepperdine

Patrons: Prof. Timothy Cox, Prof. George Elder
Dr. Mike Badminton

View points and opinions contained in this newsletter are reproduced in good faith and do not necessarily reflect the judgement of the British Porphyria Association or its patrons.

Donation to BPA

Brian and Doreen Easey's daughter in law has AIP and is in hospital at Addenbrooke's in Cambridge. She was diagnosed following a very severe attack during pregnancy, which left her paralysed and requiring ventilation for 3 months. She is now recovering, though she will require many more weeks in hospital. Fortunately her baby daughter was born safely, but it has been an extremely difficult time for her and her family.

Brian and Doreen have collected £200 at a party for their 50th Wedding Anniversary. They wish to donate it to a charity related to porphyria, and are kindly giving it to the BPA.

I am sure we all sympathize with their daughter in law, and we wish her a steady and thorough recovery. We are most grateful for the donation from Brian and Doreen.

The students of year 7, Castle Rushen High School, on the Isle of Man, had a Sponsored walk. One of their students suffers from porphyria, so they kindly gave a donation of £100 to the BPA.

Scottish Local Group

One of our committee is setting up a local BPA group in the Glasgow area.

If you have porphyria and wish to join the group, please contact us, and we will ask Sandra to contact you.

We will see how this group goes and then consider setting up other local groups.

EPP on-line forum

Our Secretary, Sarah, is looking to set up an on-line forum for EPP sufferers to exchange stories, helpful hints and tips etc.

Please keep a look out on the BPA website for more information! www.porphyrria.org.uk

Another member who has had EPP all his life recommends the discussion forum that has been created by Clinuvel. He tells us that it is a well presented-discussion forum that enables you to access information and compare notes on how best to deal with EPP. Go to: <http://photoprotection.clinuvel.com/forum>, and select Erythropoietic Protoporphyrria.



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 £10
- I would like to make a donation of:
 £10 £20 £30 £40 £50 other £ _____
- I would like to pay by Standing Order (please fill in the form opposite)
- I enclose a cheque made payable to the 'British Porphyria Association' for £ _____
- I would like a receipt

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, expand our help line or support our research.

DO YOU PAY UK TAX?

If you do, you can add 28% to the value of your gift to the British Porphyria Association at no extra cost to yourself. Simply sign and date the Gift Aid declaration and for every £1 you give we can ask the Inland revenue to give us an extra 28p.

Standing Order BRITISH PORPHYRIA ASSOCIATION

Please send this form to: The Treasurer, British Porphyria Association, 599 Bolton Road, Aspull, Wigan, Lancs, WN2 1PZ.

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

BPA use only: Please pay to British Porphyria Association bank account:

20-43-63 _____

GIFT AID DECLARATION

I am a UK Tax payer and would like the British Porphyria Association (Registered Charity No.1089609) to treat all donations I have made since 6th April 2000, and any future donations, as Gift Aid donations until I notify you otherwise. (We can reclaim the tax on your donations at no cost to yourself.)

DATE: _____ SIGNATURE: _____