THE
BRITISH PORPHYRIA ASSOCIATION
NEWSLETTER

Registered Charity No. 1089609

PORPHYRINS AND PORPHYRIAS CONFERENCE – CARDIFF

MONDAY TO THURSDAY, 10-14 APRIL 2011

The four-day porphyrins and porphyrias conference was held in Cardiff, Wales, between the 10th and 14th April 2011. The conference presented numerous interesting scientific presentations on: the acute porphyrias and their diagnostic matters; the cutaneous porphyrias, including erythropoietic porphyrias and bullous porphyrias; dermatological issues; and, interesting discussions on new developments in treating the various types of porphyria.

Scientific specialists from all around the world travelled to Cardiff to present their research and to facilitate intellectual discussions with other professionals specialising in porphyria.

Three members of the BPA attended the main conference; they were excited about the amount of research that is currently been undertaken both nationally and internationally into the different acute and cutaneous porphyrias.

The winter newsletter will present further summaries and synopses of some of the research presented at the main conference.

Pre Patient Conference Meal – Cardiff –Wednesday 13 April 2011

The BPA organised an informal meal for porphyria patients on the Wednesday evening. A total of 21 people attended from across the UK, with four people attending from the Netherlands and one from Switzerland. The majority of attendees had EPP or were particularly interested in EPP. The meal was a wonderful success which provided opportunities for people to discuss their conditions and treatments in a relaxed but supportive environment.

We plan to arrange other informal meals as we feel they provide a fantastic environment for our BPA members. If you would be interested in helping to arrange a meal or informal gathering near you, please contact us via: events@porphyria.org.uk.

"SCIENTIFIC SPECIALISTS FROM ALL AROUND THE WORLD TRAVELLED TO CARDIFF TO PRESENT THEIR RESEARCH AND TO FACILITATE INTELLECTUAL DISCUSSIONS WITH OTHER PROFESSIONALS SPECIALISING IN PORPHYRIA."

The International Porphyria Patient Day Conference – Cardiff – Thursday 14 April 2011

Over 60 people attended the patient day in Cardiff, the majority of attendees were most interested in EPP, with some AIP, VP, HCP and PCT interests. The day consisted of five clinical presentations into various types of porphyria from a number of international doctors and professors and information from other international patient support groups. We are very grateful for the attendance and support from: Dr Mike Badminton; Professor Jorge Frank; Professor Elisabeth Minder; Professor Alex Anstey; Professor Gloria Gonzalez-Aseguinolaza; Dr Paul Wilson; Professor Richard Hift; Lachlan Hay; Marcello Paglione; John Chamberlayne; Ieda Bussman and Sylvie Le Moal.

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PORPHYRINS AND PORPHYRIAS CONFERENCE – CARDIFF CONTINUED

TWO OF THE MOST INTERESTING PRESENTATIONS ARE REVIEWED BELOW:

Many EPP patients may have already heard of the clinical trials that Clinuvel have been undertaking. The trials are currently in the third phase and so far appear to be delivering positive results. Afamelanotide (trade name SCENESSE®) increases the levels of melanin in the skin and therefore shields against UV radiation (UVR) and the visible light in sunlight that triggers acute photosensitivity. The drug is delivered by a small subcutaneous dissolving implant. Increased pigmentation of the skin appears after two days and lasts up to two months.

Around 100 patients were given either Scenesse or a placebo (12-month period) and were asked to complete a diary of pain/sun exposure. The individual daily pain scores appear to be significantly lower in patients receiving Scenesse compared to those receiving the placebo. The trial results are due to be finalised in the next few months; we will keep you updated on further information.

Another interesting discussion looked into the possibility of gene therapy, currently for AIP, but maybe one day for other acute porphyrias too. This trial of gene therapy would involve the transfer of a gene to liver cells using a modified virus with the aim of transferring a therapeutic benefit. The phase I trials are currently being undertaken in Spain, where clinical safety is being examined on a small group of patients. It is expected to take around three years to establish the safety and efficiency of the procedure, after which the company aims to undertake further trials on a wider number of people, over a number of years. The treatment is far from being confirmed as a viable option and it appears at the moment that considerable risks are involved. However it is very encouraging to see the progress that is being made with regard to novel treatments within the field of porphyria.

We are delighted that our patient conference feedback forms indicate attendees found the content either very good or excellent, and preferred the clinical presentations and information on new treatments that are currently being researched. Unfortunately, the room we hired was not as protected from light as we had been advised, but we have learnt from this and will avoid similar problems at future events.

"ATTENDEES FOUND THE CONTENT VERY GOOD OR EXCELLENT"

Feedback also indicated that the talks on EPP and Afamelanotide as a treatment, and the brief presentation from Clinuvel were the most interesting for EPP patients.

We are delighted that all the patients found the clinical talks very interesting, informative and encouraging.

We are due to hold our AGM in Birmingham in September this year and we are holding an Open Day at Hope Hospital in Salford in June 2012, we hope that we will have a large group again so that we can arrange an informal meal and a formal programme with interesting clinical presentations. See the separate article on our **AGM in Birmingham**. Information on the **Open Day** at Salford will be in the next newsletter. ■

CEP STUDY FINDINGS

Many members will be aware that we have helped various researchers with funding to enable them to undertake research into the porphyrias. One such instance was a study undertaken by Ru Katugampola, and various other physicians from around the world, into congenital erythropoietic porphyria (CEP), one of the rarest and most severe porphyrias.

As this is such a rare disorder, data is very scarce regarding clinical traits and the resulting impact on quality of life for its sufferers. This study, therefore, aimed to provide a comprehensive clinical picture of the CEP phenotypes (which are the observable characteristics of an organism, such as structure, biochemical or physiological properties, resulting from the expression of an organism's genes, as well as the influence of environmental factors). It also aimed to examine the impact of CEP and its treatments on the quality of life of affected individuals and their families.

Assessing 27 patients throughout the UK and three European countries (France, Germany and Switzerland), the study observed different extremes of the condition. Patients with certain gene mutations had a late onset of the condition and were able to manage their lives successfully with photoprotection. These patients regarded the impact of CEP on their quality of life to be minimal. However, those who had other gene mutations developed early onset of the condition and their quality of life was severely impaired. They suffered with a wide array of chronic symptoms including severe photosensitivity to all visible light, photophobia, photomutilation, blindness, haemolytic anaemia and osteoporosis, among others.

For some, bone marrow transplantation can be successful; however this treatment is associated with severe complications. Although research is still ongoing, this research highlighted that all patients, including those with milder versions, be regularly monitored for complications and encouraged to protect their skin and eyes at all times from visible light. ■

RARE DISEASES DAY

FEBRUARY 28TH 2011, WESTMINSTER

The Rare Disease Day reception held at Westminster was well attended by representatives of over 100 different conditions, medical advisors and politicians. It is estimated that there are over 6000 rare diseases affecting 3.5 million people in the UK at some point in their lives (1 in 17 people). Collectively, rare diseases are not rare! Speakers included Liz Kendall MP, Mr Julian Huppert MP, Ms Jayne Hughes, Professor Sir Jon Burn, Mr Alastair Ken OBE, and Earl Howe. Liz Kendall read out a statement from Ed Milliband, Leader of the Labour Party.

The aim of the reception was to raise awareness of rare diseases.

- To emphasise rare diseases as a health priority.
- To ensure that by acting simultaneously and collaboratively, nationally and internationally the voice of rare disease patients is heard by more people.
- To highlight the issues affecting people with rare diseases.
- To bring all of the stakeholders involved in rare diseases together. This includes patients, families, carers, policy makers, healthcare providers, clinicians, researchers, health workers, industry and patient organisations.
- To coordinate policy actions for rare diseases at each level, across the UK, at European level and internationally.

RDUK's new report "Improving Lives, Optimising Resources: A vision for the UK Rare Disease Strategy" was launched. The theme of Rare Disease Day 2011 is "Rare but Equal".

In 2011 Rare Disease Day sought to draw attention to:

1. The gaps in health that exist for rare disease patients between and within countries in the EU.
2. The gaps in health that exist for rare disease patients compared to other sectors of the society.

To advocate for:

1. Equal access for rare disease patients to health care and social services.
2. Equal access to basic social rights; health, education, employment, housing.
3. Equal access to orphan drugs and treatments.

Reproduced from literature provided by Rare Disease UK

"IT IS ESTIMATED THAT THERE ARE OVER 6000 RARE DISEASES AFFECTING 3.5 MILLION PEOPLE IN THE UK AT SOME POINT IN THEIR LIVES"

SHOW EPP WHO'S BOSS

By Beth Venis aged 14

I was not diagnosed with EPP until I was 12. Looking back, I did have symptoms that I can remember from aged 6. My feet used to hurt and my hands used to tingle when I was out in the sun, but I didn't think anything of it. I didn't really go out in the sun much as I have red hair and blue eyes. My first big reaction was when my face became swollen aged 7. Mam took me to A&E, but they thought it was an allergic reaction to something I had eaten.

We went on holiday to Spain when I was 11, it was August and boiling hot. My hands began to tingle and then the pain started. It is hard to explain, but feels a bit like your skin is on fire. There was nothing to see on my hands at first, but after a couple of days the swelling started. I was in agony and kept trying to cool my hands down with cold water and ice. I couldn't bear anyone to touch them as the pain was so bad. Mam tried to get medical advice, but they gave me steroid cream and said that I was allergic to the sun cream. When Mam rubbed it on my hands I screamed as the pain was terrible. We stayed in for the rest of the holiday as I couldn't bear to be outside. I couldn't wait to get home.

When I came home my hands stopped hurting so much after a couple of days, but the skin was dry and cracked, this took weeks to get better. I was told that it was an allergic reaction, and I hoped it would never happen again.

The year after, I was in the garden in May when the tingling started again on my hands; Mam took me to the doctors and although he had no idea what was wrong, he referred me to a dermatologist. I was very lucky, as the dermatologist had seen one case of EPP before and was 99% certain that this is what I had. She told Mam to cancel our holiday to Florida and sent me for blood tests. Six weeks later I was diagnosed with EPP.

I loved being outside, and my hobbies were horse riding, running and being out in the country with my dogs. I was so sad because I was told that I would not be able to go out in the sun, but I loved being outdoors.

I have been very fortunate to meet someone with EPP who lives close by who has inspired me to show EPP who's boss. He is called John Clark and he still goes outside in the sun, and runs ultra marathons, walks in the lakes, skis and lots of other mad things.

I went on my first skiing holiday this year, John helped me with my clothing to make sure I would not be ill, as the sun reflects off the snow and can make people with my condition very poorly. I learned to ski, had a fabulous time and because of John's help I did not get any reaction.

Outdoor lifestyle

I have also discovered climbing, although it is indoors I like to think that I would be able to do this outside. This year I am going to run my first race, with the help of John, who has said he would run with me. I still go horse riding, as I can wear my hat, riding gloves and jodhpurs so I am covered up and look like every other horse rider.

When I first got diagnosed with EPP I felt very sad and thought that my life would change forever. My life has changed forever, but it is better now. I would not like people to feel sorry for me, as EPP has introduced me to John and he has taught me that EPP is part of me and I should never feel sad about that.

The future for me will be bright, I will be spending it outdoors, riding, running, climbing and walking with my dogs, whether the sun is shining or not. I am determined to run my first ultra marathon at 18, and I will do it. If I do get ill I will just go home and think of all the fun that I have had that day and not regret one minute of it. We all get ill sometimes, so how am I any different to anyone else? There are many people in the world who have conditions much worse than me. I sometimes get a bit down and sad, but Mam and my little sister, Cait, won't let me be sad for long.

Mam and John have made a big difference to my life. I don't tell many people about my EPP, as they don't understand. The people important to me do understand and that's all that matters. ■

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.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.

Chairman: John Chamberlayne
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FACEBOOK

Just to remind those of you who use the Social Network site, Facebook, that the BPA has its very own site. Even if you are not already on Facebook, if you have access to the internet, it is worth joining just so that you can access the BPA site. All you need to do is type: 'British Porphyria Association' into the search box on Facebook.

The best thing about being on the BPA Facebook site is that you can discuss issues, ideas and learn lots about all the different types of Porphyria. There have been discussions on the best types of sun cream to use for EPP sufferers, discussion on different types of drugs for those with acute porphyrias and loads more.

BPA AUTUMN CONFERENCE AND AGM

This year we are holding our autumn conference and AGM in **Birmingham** on **Saturday, September 10th, at 2pm.**

It is at Shirley Methodist Church, about 3 miles south of the centre of Birmingham, on the A34. There is a car park behind, and there are buses which go past the church.

As in previous years, we are starting with a brief AGM, and then having talks relating to porphyria, and the experience of living with porphyrias.
If you are interested in coming, please let us know in one of the usual ways (letter, phone, email [events@porphyria.org.uk] or online). We will be having refreshments after the talks, but would you also be interested in getting together for a meal, probably in the evening after the conference?

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

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Date

WAYS TO PAY

☐ I would like to pay my annual membership fee of £15

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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. The Government will pay to the BPA an additional 3p on every £1 you give between 6 April 2008 and 5 April 2011. 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, 11 Blakehill Terrace, Undercliffe, Bradford, West Yorks, BD2 3JS.
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:
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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