British Porphyria Association

Education / Support / Research

Registered Charity No. 1089609

The BPA would like to thank Orphan Europe for providing a grant to assist in the funding of this meeting

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Introduction to the British Porphyria Association and the Porphyrias

Liz Gill

Welcome

- Housekeeping and the venue
- BPA bags
- Leaflets and merchandise
- Agenda for the day
  - All together this morning
  - Short comfort break – split into acute (scientific talks) and cutaneous (informal chat)
  - Lunch – split again into acute (informal chat) and cutaneous (scientific talks)
  - Short comfort break – all back together for final talks and Q&A session
  - Informal discussion times

Thank you…

Introduction to the BPA

- Established in 1999, became a registered charity in 2001
- Up to March 2015, run solely by a committee of volunteers
  - Now have a paid administrator for 12 hours a week to support volunteers
- Aim to provide support, education and research
- Currently almost 500 people on database
    - Approx. 10% medical contacts and other organisations
    - 450 individuals and families
- Continuing to grow and evolve

What do we do?

- Hold two events per year for our members
- Publish two newsletters a year and design and publish various patient information leaflets
- Run telephone and email helplines, as well as our own website
- Monitor a Facebook page and have recently started to use Twitter (with not much input as yet)
- Coordinate various fundraising efforts with places in the London Marathon, Ride London and the Great North Run to name just a few
- Continuing with merchandise – to sell and raise awareness and for our fundraisers (incl. specialised sportswear)

What do we do?

- Attend relevant meetings in the UK including BIPNET and NAPS
- Attend meetings internationally – ICPP and IPPN
  - BIPNET – British and Irish Porphyria Network

    a professional forum for specialist clinicians and scientists in the porphyria field, working collaboratively to share expertise and promote best practice in the care of patients with all forms of porphyria.
  - NAPS – National Acute Porphyria Service

    established in April 2012. Funded by the Department of Health to help patients with severe acute porphyria. NAPS centres: Cardiff and London, with Salford and Leeds as regional centres.
  - ICPP – International Conference on Porphyrins and Porphyrias
  - IPPN – International Porphyria Patient Network

- The BPA help support all of the different types of porphyria in different ways.
- As a charity, we:
  - Are very grateful for our member subscriptions
  - Are very grateful for our sponsors who help with events such as this
  - Are very grateful for the fundraising efforts of our members
- We are also very reliant on the skills of our committee to keep moving forward – if you have relevant skills and the time to help, please let us know.

Introducing the porphyrias

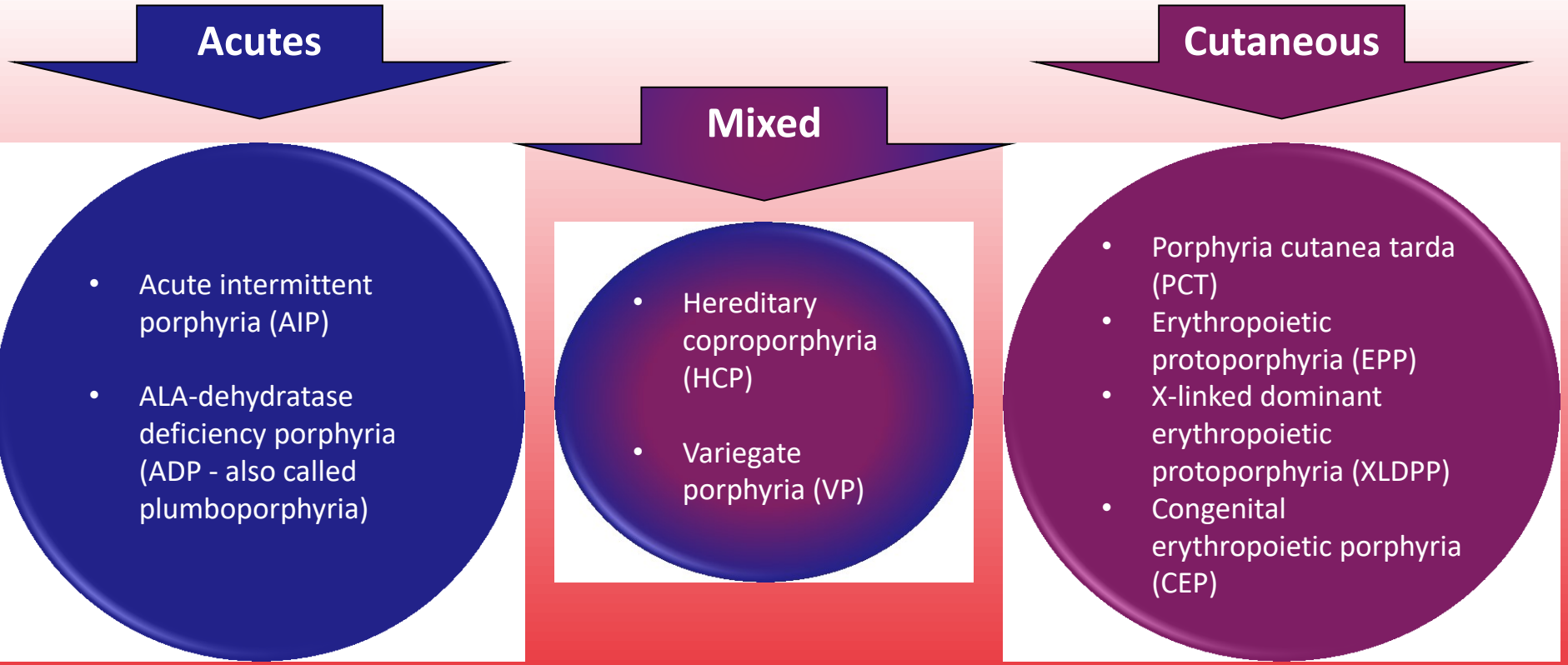
What are the porphyrias?

- A group of rare metabolic disorders
- Most are inherited due to a faulty gene
- All interfere with the way the body makes haem

Why is haem so important?

- Haem is a vital component in the body
- It is needed to make haemoglobin and important proteins
- For those with porphyria, there is a lack of one of the enzymes (special proteins) needed to produce haem
- During the production of haem, some porphyrin and porphyrin precursors can be overproduced
  - In large quantities these can act as toxins to the body
- The way they build up differs depending on the type of porphyria

The different types of porphyria



The acute porphyrias

- AIP and ADP (as well as VP and HCP)
- Are caused by a build up of ALA and PBG (aminolevulinic acid and porphobilinogens)
- Certain drugs and other factors can trigger attacks
- Attack symptoms can include:
  - Intense pain
  - Nausea, sickness and constipation
  - Low salt levels (and possibly convulsions)
  - Increases in heart rate and blood pressure
  - Muscle weakness

The skin (cutaneous) porphyrias

- EPP, XLEPP, PCT and CEP
- Are caused by a build up of porphyrins in the blood
- These porphyrins are damaging to the skin layers or the blood vessels (depending on the porphyria) as they react to various types of light
- Symptoms differ depending on the type of porphyria

The skin porphyrias (cont..)

- Porphyria cutanea tarda (PCT) is the most common cutaneous porphyria
  - 20% is inherited, 80% triggered by other means
- Congenital erythropoietic porphyria (CEP) is extremely rare
  - It affects many different parts of the body, including the skin, eyes and bone marrow
  - Severe scarring often results due to extreme sensitivity to visible light

Erythropoietic protoporphyria (EPP) and X-linked EPP (XLEPP)

- Characterised by reactions to visible light

  (Blue, violet and other spectrums of light), rather than UVB/UVA which normally causes sunburn

  Some artificial lights can also be problematic in a small number of people

- A phototoxic chemical reaction occurs when the skin is exposed to light:
  - Severe burning, itching, swelling - extremely painful
  - Often there are no visible signs of burning, meaning it is very difficult to diagnose, especially in children

The mixed porphyrias

- VP and HCP
- In addition to experiencing acute symptoms, they can also experience blistering of the skin when exposed to sunlight, resulting in fragile skin
- Need to be aware of safe drugs list and also skin protection elements

Incidence rates in the UK are difficult to determine

- Incidence differs depending on the type of porphyria
- It has been suggested that between 1 in 10,000 and 1 in 75,000 people may have a faulty porphyria gene
- Of these, many will never develop porphyria symptoms

Treatment of the porphyrias

- **The acute porphyrias**
  - There are various therapeutic treatments: but primarily, any triggers should be removed and then expert clinicians will guide patient care depending on the severity of the patient.
  - Options can include:
    - Haem arginate treatment
    - Hormone suppression
    - Pain management

Treatment of the porphyrias

- **The skin porphyrias**
  - Depending on the type, there are various therapies are available through expert clinicians, all with varying degrees of success, including:
    - Light therapy
    - Beta carotene
    - Dundee cream
    - SCENESSE®? Not available on NHS yet …

The future…

… is bright as clinicians and drug companies around the world are continually working to find better treatments/cures for the different porphyrias, incl.

- **Orphan Europe**
- **Clinuvel Pharmaceuticals Ltd**
- **Alnylam Pharmaceuticals**

Future Porphyria Events

- **2018 Open Day – Queen Elizabeth Hospital, Glasgow**

  9th June 2018

- **2018 Autumn Conference & AGM – to be confirmed**

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