

ipnet

NEWS LETTER

A quarterly newsletter keeping the medical and patient community in touch with Porphyria developments

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FROM IPNET PRESIDENT

Sverre Sandberg



Dear friends and colleagues – Ipnet members,

I am delighted to contribute to the inaugural edition of the Ipnet Newsletter. My sincere gratitude goes to Nica Cappellini and her fellow editors for bringing this initiative to fruition. The International Porphyria Initiative is currently in its initial phase as a global organization. The decision to transition from Epnet to Ipnet was driven by the opportunity to connect with a broader audience and more porphyria centers worldwide. In this new phase, all porphyria centers globally are welcome to apply for full membership in Ipnet, granting them voting rights to influence and prioritize Ipnet's activities. The recent amendments to Ipnet's statutes aim to simplify the process of becoming a full member, and the membership application can be accessed at www.porphyrianet.org. Former "associate" members are encouraged to explore the possibility of upgrading to full membership. This transition holds particular significance as, in early 2024, we will issue a call for nominations for a new Executive Board of Ipnet. Only full members will have the privilege to vote and stand for election to the Board. The unveiling of the new Executive Board will take place during the General Assembly of Ipnet in Pamplona in September 2024, with the board officially commencing its duties in January 2025. We urge all members to actively nominate individuals from various centers to contribute to the Ipnet Board. Ipnet extends its membership options to individuals, corporations, associates, and patient organizations. The organization boasts numerous Working Groups (WGs) and ongoing activities, details of which can be explored at: <https://porphyrianet.org/en/content/ipnet-working-groups>. Noteworthy among these is our dedicated effort to develop evidence-based guidelines for treating and monitoring acute porphyrias. This collaborative endeavor involves a specialized method team in guideline development, and we are pleased to announce that the first paper on "definitions" has already been published.

Additionally, the WG on laboratory diagnostics is preparing a special report on diagnosing and monitoring porphyrias for Clinical Chemistry. The WG-APIS focuses on providing support for acute porphyria to healthcare professionals in regions where specialist care may be limited. The primary objectives include facilitating diagnostic laboratory testing to enhance access to management advice and treatment.



SAVE THE DATE - ICPP 2024 PAMPLONA

Link: <http://ICPP2024.com>

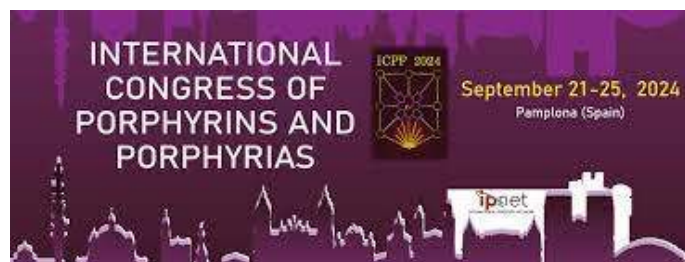
Dear Colleagues, dear friends,

The International Congress of Porphyrins and Porphyrrias will take place on September 21-25, 2024 in Pamplona, Spain, bringing together clinicians, researchers, scholars, patients and their families for three days of networking and learning about the latest research in the porphyria field.

We are preparing an exciting program to learn about and discuss the latest advances in our understanding of the natural history, management and treatment of hepatic and erythropoietic porphyrias.

We will complement the scientific aspects of the event with social activities, including a tour of the historical downtown, vineyards located in a privileged microclimate and some of the best-preserved medieval villages in Spain.

And, "by the way", once in Pamplona don't miss the opportunity to start walking the Way of Saint James, "El Camino"!



See you all in Pamplona!

Best regards from

Sverre Sandberg
Ipnet president

Antonio Fontanellas Roma
President of ICPP 2024

INTERNATIONAL PORPHYRIA NETWORK

An International network of experts working together to improve healthcare services for porphyria patients



PATIENTS CORNER

My name is Sue Burrell and I am the current President of the Global Porphyria Advocacy Coalition (GPAC). I would like to introduce you to GPAC, who we are and the work we are doing. We are an umbrella organisation formed of a connected network of over 20 patient advocacy groups (PAGs) focused on porphyria, from countries around the world. We share likeminded aspirations for porphyria patients and realised that we would be stronger if we worked together. We became a recognised charity in 2020, following our first official gathering at the ICPP in Milan in 2019. Despite being a fledgling organisation ourselves, we are formed of Member PAGs with decades of experience amongst us, with excellent links with many of the national Ipnet recognised Porphyria Expert Clinical Centres (PECC). Within our mission to provide a unified and collaborative international voice for porphyria patients, we aim to complement the existing national organisations' work and welcome/support new groups as they emerge and grow into fully fledged PAGs that are linked to Ipnet, the PECCs and expert physician networks. We strive to ensure aligned goals and support the excellent evidence-based community that we are part of, and work to be transparent in our working practices. We are fully inclusive and equally support all of the porphyrias, promoting knowledge and resource sharing among groups. Ultimately, our approach hopes to support patients/caregivers, physicians, researchers and regulatory bodies to help provide a much-needed patient voice (especially in areas where this needs support) – the porphyria community is therefore stronger as a result of working together! Our hopes extend to fostering an environment whereby the global patient porphyria community is a force for collaboration in advocacy for international projects that will benefit the collective porphyria community. Our website details a list of the patient groups/countries that are engaged in and part of the work we do: <https://www.gpac-porphyria.org/members>



IPNET WORKING GROUPS

Ipnet has several active working groups: see details at: <https://porphyrianeet.org/en/content/ipnet-working-groups>. Any person of IPNET full member group can apply to be part of a specific working group contacting the chair of the WG and submitting the motivations and competences to be admitted. The chairs of each working group will report their ongoing activities at the international Congress during the general assembly. The Ipnet executive board may decide to stop a WG in case the program of the WG is completed or when no further progress is being made. Over the last 2 years the PECC WG has been active in endorsing Porphyria Expert Clinical Centers (PECC).

PORPHYRIA EXPERT CLINICAL CENTERS (PECC)

Three types of centers were envisaged: cutaneous PECC, acute PECC and “full” PECC (covering cutaneous and acute). The criteria that were decided on, reflect experience (duration of clinical care for porphyria patients, but also the size of the cohort of patients that is followed), the presence of multidisciplinary care and continuity of care, the availability of the necessary diagnostic and treatment modalities for the different types of porphyria, contribution to scientific progress in the field of porphyria and the endorsement by the hospital management as well as by a relevant patient organisation. The procedure is overseen by a newly created Ipnet Working Group (PECC WG). Several centers have already been endorsed (see [list of endorsed centers here](#)). The procedure remains open all year round.





ACUTE PORPHYRIA INTERNATIONAL SUPPORT GROUP (WG-APIS)

The Acute Porphyria International Support Group (APIS) will offer help to healthcare professionals looking after patients with suspected acute porphyria in countries where porphyria laboratory testing and specialist care are not available. Support will include arranging for biological samples to be sent to a specialist porphyria laboratory in Europe for diagnostic testing without charge. Referring clinicians will be able to contact APIS by completing a webform on the IPNET website. We are developing the necessary processes and hope to be ready to accept referrals by Spring of 2024.

GUIDELINES FOR MONITORING AND TREATMENT OF PORPHYRIA



EPNET/IPNET identified the need of producing “evidence based “ guidelines for the diagnosis and treatment of acute and chronic porphyrias. After critical evaluation the EpNet board found the opportunity to engage a team of expert in producing guidelines lead by Dr Reem Mustafa. EPNET signed an agreement with Dr Mustafa for the first round dedicated to Acute Porphyria.

Publication

Key terms and definitions in acute porphyrias: Results of an international Delphi consensus led by the European porphyria network.

[Penelope E Stein 1](#), [Yonatan Edel 2](#), [Razan Mansour 3](#), [Reem A Mustafa 34](#), [Sverre Sandberg 567](#); [Members of the Acute Porphyria Expert Panel](#) J Inherit Metab Dis2023 Jul;46(4):662-674

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SCIENTIFIC UPDATE ON PORPHYRIAS

181 publications (103 full texts) have been published in 2023. The most relevant are summarised below

- 1.Evidence-based consensus guidelines for the diagnosis and management of erythropoietic protoporphyria and X-linked protoporphyria.** Dickey AK, Naik H, Keel SB, Levy C, Beaven SW, Elmariah SB, Erwin AL, Goddu RJ, Hedstrom K, Leaf RK, Kazamel M, Mazepa M, Philpotts LL, Quigley J, Raef H, Rudnick SR, Saberi B, Thapar M, Ungar J, Wang B, Balwani M; Porphyrias Consortium of the Rare Diseases Clinical Research Network. *J Am Acad Dermatol.* 2023 Dec;89(6):1227-1237.
- 2.The Hepatic Porphyrias: Revealing the Complexities of a Rare Disease.** Balogun O, Nejak-Bowen K. *Semin Liver Dis.* 2023 Nov 16
- 3.The alpha-lipoic acid improves glucose metabolism and hyperinsulinemia in Acute Intermittent Porphyria: a nutritional concept for the management of rare disorders.** Longo M, Paolini E, Meroni M, Jericó D, Córdoba KM, Battistin M, Gatti S, Di Pierro E, Fontanellas A, Dongiovanni P. *Cell Mol Gastroenterol Hepatol.* 2023 Nov 16:S2352-345X(23)00202-3. Online ahead of print.
- 4.RNA interference therapy in acute hepatic porphyrias.** Yasuda M, Keel S, Balwani M. *Blood.* 2023 Nov 9;142(19):1589-1599.
- 5.Porphyrin precursors and risk of primary liver cancer in acute intermittent porphyria: A case-control study of 188 patients.** Lissing M, Wester A, Vassiliou D, Floderus Y, Harper P, Sardh E, Wahlin S. *J Inherit Metab Dis.* 2023 Nov;46(6):1186-1194.
- 6.Acute hepatic porphyrias: Recommendations for diagnosis and management with real-world examples.** Moghe A, Dickey A, Erwin A, Leaf RK, O'Brien A, Quigley JG, Thapar M, Anderson KE. *Mol Genet Metab.* 2023 Nov;140(3):107670.
- 7.Efficacy and safety of givosiran for acute hepatic porphyria: Final results of the randomized phase III ENVISION trial.** Kuter DJ, Bonkovsky HL, Monroy S, Ross G, Guillén-Navarro E, Cappellini MD, Minder AE, Hother-Nielsen O, Ventura P, Jia G, Sweetser MT, Thapar M; ENVISION Investigators. *J Hepatol.* 2023 Nov;79(5):1150-1158.
- 8.Erythropoietic Protoporphyria and Afamelanotide: A Patient's Perspective.** O'Reilly M, McGuire VA, Dawe RS. *Clin Exp Dermatol.* 2023 Oct 13:llad346
- 9.Mitochondrial DNA Copy Number Drives the Penetrance of Acute Intermittent Porphyria.** Di Pierro E, Perrone M, Franco M, Granata F, Duca L, Lattuada D, De Luca G, Graziadei G. *Life (Basel).* 2023 Sep 15;13(9):1923
- 10.Update on the Porphyrias.** Dickey AK, Leaf RK, Balwani M. *Annu Rev Med.* 2023 Aug 4. doi: 10.1146/annurev-med-042921-123602. Online ahead of print
- 11.Protoporphyrin IX-induced phototoxicity: Mechanisms and therapeutics.** Hussain Z, Qi Q, Zhu J, Anderson KE, Ma X. *Pharmacol Ther.* 2023 Aug;248:108487.
- 12.Maternal and fetal outcomes in acute hepatic porphyria: A Swedish National Cohort Study.** Mantel Å, Vassiliou D, Lissing M, Stephansson O, Wahlin S, Sardh E. *J Inherit Metab Dis.* 2023 Jul;46(4):675-686.
- 13.Evaluating the Efficacy of a Small Interfering Ribonucleic Acid Molecule, Givosiran, in Treating Acute Intermittent Porphyria: A Systematic Review.** Patel P, Midha S, Shukla S, Dhamija D, Bello AO, Khan S. *Cureus.* 2023 Jun 18;15(6):e40585
- 14.Dersimelagon in Erythropoietic Protoporphyrias.** Manisha Balwani, Herbert L Bonkovsky, Cynthia Levy, Karl E Anderson, D Montgomery Bissell, Charles Parker, Fumihiko Takahashi, Robert J Desnick, Kirstine Belongie; Endeavor Investigators. *N Eng J Med* 2023 Apr 13;388(15):1376-1385
- 15.Dersimelagon in Erythropoietic Protoporphyrias.** Granata F, Dechant C, Falchetto R. *N Engl J Med.* 2023 Jun 29;388(26):2491-2492