UNITED PORPHYRIAS ASSOCIATION
Advancing Awareness, Research & Therapies

Get the latest

Dear Member, here are your porphyria UPdates for January!

THE ORPHAN DRUG ACT AND PORPHYRIA!

The Orphan Drug Act (ODA) was ratified 40 years ago this month! This legislation has had a significant impact on the porphyrias. From its beginnings in 1983 through today, the ODA has been a critical incentive to encourage research and development for rare diseases.

We thank our pharmaceutical partners who are committed to current and emerging treatments in the porphyrias, our expert **Scientific Advisory Board** leadership who champion research and, of course, our porphyria patient community that give of themselves by participating in clinical trials. It is only through this united front that we can advance research and therapeutics!

About the ODA

What is an Orphan Drug?
An investigational therapy for a rare disease that impacts fewer than 200,000 individuals. An orphan drug designation is granted by the FDA Office of Orphan Products and Development.

How does the ODA help?
The ODA encourages increased development of drugs for rare diseases through incentives for pharmaceutical companies including: 7 years of market exclusivity, tax breaks for research and development expenses, and a waiver of FDA User Fees.

How does UPA support the ODA?
The ODA policy has supported significant

Orphan Drug Designations in the Porphyrias

December 2022: DISC MEDICINE (Bitopertin) for the treatment of EPP.

June 2020: MITSUBISHI TANABE (Dersimelagon) for the treatment of cutaneous variants of porphyria.

April 2018: ATLAS MOLECULAR PHARMA SL (Ciclopirox) for the treatment of CEP.

August 2016: ALNYLAM PHARMACEUTICALS (Givosiran) for the treatment of adults with acute hepatic porphyria and became an FDA approved drug (Givlaari) in November 2019.

July 2008: CLINUVEL PHARMACEUTICALS (afamelonotide) to increase pain free light exposure in adult patients with a history of phototoxic reactions from EPP and became an FDA approved Drug (Scenesse) in October 2019.

July 1983: RECORDATI RARE DISEASES (current manufacturer) Panhematin

research in the porphyrias. UPA joins the National Organization for Rare Disorders (NORD) in advocating for federal and state policies that protect and further Orphan Drug Development.

(hemin) was approved by the FDA as the ***very first orphan drug*** to relieve repeated attacks of acute porphyria.

UNITED PORPHYRIAS ASSOCIATION TURNS 1!

We thank each of you for your tremendous support of UPA throughout our first year!

As the **future of porphyria,** UPA will keep patients at the forefront of our work, our fingers on the pulse of treatment development, and will remain hyper focused on advancing awareness, research, and therapies in all porphyrias.

We appreciate and applaud our resilient patient community, visionary Board of Directors, world renowned **Scientific Advisory Board,** and forward-thinking pharmaceutical partners. We are Porphyria Together!

JOIN US FOR A FACEBOOK LIVE SESSION- FEB 18

Looking for an expert to answer your porphyria questions?

Dr. Bruce Wang (Porphyria Expert, USCF and UPA Scientific Advisory Board Member) will be taking your questions live on **Saturday, February 18 at 1 PM ET** on our **Porphyria Together** Facebook group.

Be sure to join the **Porphyria Together group** in advance and we'll see you there!

JOIN GROUP

FACES OF PORPHYRIA

NIH Highlights Porphyria Researcher

The work of neurologist Mohamed Kazamel, MD, of University of Alabama, Birmingham and UPA Scientific Advisory Board, was recently highlighted by the Rare Diseases Clinical Research Network (RDCRN) of NIH.

"The continuous suffering of patients and the presence of large gaps of knowledge in the pathophysiology of porphyric neuropathy both inspired me to seek more answers for these patients and to help my fellow hepatologist, hematologist, and geneticist members of the Porphyrias Consortium."

Check out **this article** where he shares a recent discovery and what he is working on now!

Mohamed Kazamel, MD
UPA Scientific Advisory Board Member

RESEARCH RoundUP

Dosimetry is a Promising Method for Measuring Light Exposure in EPP and Might Help Predict and Prevent EPP Symptoms

Amy Dickey, MD
UPA Scientific Advisory Board Member

UPA Scientific Advisory Board Member Amy Dickey, MD (Mass General Hospital, Boston) led a study using a small wearable device (dosimeter) to measure UV light exposure over time in patients with EPP.

24 EPP patients participated by completing light sensitivity surveys and wearing a UV light monitor for a month; manually reporting how much time they spent outdoors for an additional month; and reporting EPP symptoms throughout.

The study showed that UV light monitoring (dosimetry) consistently performed better than participants' self-reporting of time outdoors. For example, the UV light monitor data but not the self-reported data on outdoor time correlated with participants' self-reported level of light tolerance.

This study suggests wearable UV light monitoring may be a promising method for measuring light sensitivity in EPP and for predicting and preventing EPP symptoms.

Click here to read the publication, and **here** for research that is now using dosimetry!

Thank you to the study participants for making this study possible!

CURRENT RESEARCH OPPORTUNITIES

Participating in porphyria research is an important way that we can make a difference. Research gives us a better understanding of porphyria and is an important part of developing and testing new treatments.

Below are some current research opportunities. You can learn more about these and new opportunities **here** or by contacting **UPA at 1-800-868-1292 or info@porphyria.org.**

ALL PORPHYRIAS

(EPP, XLP, CEP, VP, AIP, HCP, PCT, HEP, ADP)

LONGITUDINAL STUDY

- Long-Term Follow-Up Study to Better Understanding the Natural History of Porphyria.

EPP

DISC MEDICINE - BITOPERTIN

- Phase 2 Clinical Trial for patients with EPP.
- If you'd like more information, please **fill out this short form**

EPP/XLP

CIMETIDINE

- Assessing treatment with Cimetidine in patients ages 15+ with EPP/XLP.
- Note: there is no longer a requirement to experience symptoms within 30-minutes of sunlight exposure.

EPP/XLP Gene Modifier Study

- Study investigating genetic factors associated with differences in light sensitivity among patients with EPP and XLP.

ACUTE HEPATIC PORPHYRIAS

(AIP, VP, HCP)

Medication Study to Inform Drug Safety

- Online observational study to create an updated list of medications that are risky or safe for people with one of the acute porphyrias.

AIP

AIP INTERVIEWS

- 60-Minute interview with porphyria specialists to discuss patient experience.

AIP GENE MODIFIER STUDY

- Study to find other genes that may make a person with AIP more or less likely to develop acute attacks.

Learn more about research opportunities **here**
OR
by contacting UPA at 1-800-868-1292 or info@porphyria.org.

JOIN US FOR NIH RARE DISEASE DAY 2023!

UPA President Kristen Wheeden and Jennifer Beck, an EPP Patient will speak on a *Rare Diseases Therapeutics and the Role of Advocacy and Industry Collaborations* panel during Rare Disease Day at the National Institutes of Health.

This session will explore the spectrum of advocacy–industry collaborations across the life cycle of therapeutic development and share how patient advocacy can play an important role in research efforts. Panelists from patient advocacy and industry will encourage participation in research and inspire hope for future treatments in more rare diseases. View the agenda and register to attend (in person or virtually) **here.**

REGISTER

STAY IN TOUCH!

Add United Porphyrias **INSTA**, **FACEBOOK**, **TWITTER** to your social media feeds.

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