



AMERICAN PORPHYRIA FOUNDATION

4th Quarter, 2021

HAPPY HOLIDAYS

FROM THE AMERICAN PORPHYRIA FOUNDATION

It is our great joy this holiday season to say thank you to you, our wonderful members, and to wish you the very best for 2022. We hope you enjoy a wonderful holiday season and a new year filled with peace, love, and happiness!

– The APF Staff and Board of Trustees

HEME BIOSYNTHESIS AND THE PORPHYRIAS

The Heme Biosynthesis and the Porphyrins 2021: Consensus Diagnoses, Variant Disorders, New & Emerging Therapies, was held October 15-17, 2021, at the Hyatt Regency Schaumburg, IL near the Chicago O'Hare Airport.

This 2.5-day educational symposium featured international and national experts who presented the latest findings on the biology, transport, and regulation of heme biosynthesis; as well as the clinical features, management, and current and emerging treatments for acute hepatic and erythropoietic porphyrias. **Thank you to our amazing sponsors and contributors!**

THANK YOU TO OUR SPONSORS

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GRAND ROUNDS

PORPHYRIA GRAND ROUNDS

The American Porphyria Foundation has implemented a Porphyria Grand Rounds educational program that was created by porphyria experts, bolstered by the voices of patients, and delivered by qualified physicians. We look forward to continuing to execute this series throughout 2022.

If your physician is interested in more information regarding this program, please have them contact the American Porphyria Foundation.

ACG 2021 – LAS VEGAS

The American Porphyria Foundation exhibited at the 86th Annual American College of Gastroenterology (ACG) Scientific Meeting and Post Graduate Course in Las Vegas, Nevada! With the appropriate safety protocols in place, ACG became an excellent opportunity for the APF to connect with the GI physicians and trainees from across the world to spread the word about Porphyria. Edrin Williams, our Director of Patient Services, pictured was in attendance. #ACG2021



ASH 2021 – ATLANTA

The 63rd Annual American Society of Hematology Meeting (ASH) will be held in Atlanta, GA on December 11-14, 2021, at the Georgia World Congress Center. We are looking forward to connecting with hematologist from around the country to further educate them about Porphyria. The American Porphyria Foundation will have an exhibit booth. Stay tuned for details surrounding upcoming conventions that we will be attending in 2022.



MORE EXHIBITS

ACMG 2022
Nashville, TN
March 23-25, 2022

DDW 2022
San Diego, CA
May 21-24, 2022

AALSD 2022
Washington, DC
November 4-7, 2022



PATIENT EDUCATION MEETINGS 2022

For 2021, we opted to keep our patient education and support meetings virtual. This year alone, we have had a total of 14 virtual meetings! As we come into a new year, we are considering returning to in-person meetings. While your safety in our main priority, we feel that you would benefit from interacting with fellow members in the Porphyria community. **We would like to hear from you!** With the appropriate protocols in place, would you be willing to attend an in-person meeting? Please share your thoughts with us via email at info@porphyriafoundation.org



MEDICAL TRANSPORTATION ASSISTANCE FROM MERCY MEDICAL ANGELS

MERCY MEDICAL ANGELS

Mercy Medical Angels have been providing free transportation to medical care since 1972.

On the ground with gas cards, bus, and train tickets, and in the air with trips flown by volunteer pilots and commercial airlines, Mercy Medical Angels is here to ensure that no one in need is denied medical care because they don't have transportation. Apply for assistance today!

MEMBER STORIES NEEDED!



We want to invite you to share your story with the porphyria community. The APF hears regular feedback that the stories listed on our site have helped them feel less isolated and work toward diagnosis. Stories should be about 500 – 2000 words and full of your personality. Make sure to include information about your specific porphyria type, your diagnostic journey, and how this disease has impacted your life. With permission, all stories will be featured on the APF website. We will also feature select stories in our quarterly newsletter. Send your stories via email to info@porphyriafoundation.org with a recent photo and the APF staff will respond with a waiver for permission to share your story on our platforms.



DILANI VITHANAGE – ACUTE INTERMITTENT PORPHYRIA

taking those medicines, I fell while walking. Within 24 hours, I was completely paralyzed and could not swallow fluid or food. I was also barely conscious. I was treated in the Intensive Care Unit of the hospital where I received a diagnosis of Guillain Barre Syndrome following an infection.

After that, another two recurrent similar episodes happened after taking medicines for abdominal pain and vomiting. During these attacks, the same pediatric physician cared for me. My legs were paralyzed, so I had to use a wheelchair to move and could not go to school for months. As I was still not diagnosed, a lot of investigations were carried out, including EEG, EMG, scan, testing cerebral spinal fluid, blood tests, and urine tests. Some of the investigations were painful and scared me. Finally, I was diagnosed with acute intermittent porphyria (AIP), which is a rare, inherited disease. Since AIP was precipitated by unsafe drugs were given, I was given an unsafe drug list. Gradually, I recovered. Recently, I had an attack, However, due to lack of AIP awareness among medical professionals, I

was treated for Gastritis instead of AIP.

On and off, I experience severe abdominal pain, vomiting, frequent constipation and sometimes back and leg pain. There are times I have leg muscle cramps, too, that are so severe that I want to be alone and relax. It is also upsetting that AIP treatment is not easily available and people are treated with other medicines.

I always look for the positive aspects of life. Despite my condition, now I continue my higher education in Master's in Rehabilitation studies. I decided to select a carrier in physical rehabilitation of patients with physical disabilities, because as a kid, I also had difficulties in walking, which had a negative impact on my childhood education. Therefore, I am determined to support people and children with physical disabilities to make their lives better through my carrier. Now I am a Prosthetist and orthotist and work for a Rheumatology and Rehabilitation hospital, which is the national center for physical rehabilitation of disabilities in my country.

After many years now, I am aware of my disease, and I realized that I must live with the condition. Since I was six years old, I have had severe leg pain, especially in the calf area. Some nights I cried. Some days I sat near the road because I could not walk back home from school due to leg pain. Doctors advised me to take vitamin B. They said leg pain at such a young age can be due to vitamin B deficiency. When I was eleven years old, I had severe abdominal pain and vomiting. The Doctor prescribed medications diagnosing an infection. After



NEW FACEBOOK SUPPORT GROUP CREATED

We are proud to announce that we have helped initiate another FACEBOOK support group. Our friends from India can now have their own means to share with one another. We also located physicians who are willing to help diagnose and treat. Natasha Bimbhat Ahluwalia (bottom) has kindly offered to lead the group. One of our USA members, Gunja Gardharia (top), will assist with the group. If anyone is interested in joining, please PM Global Director, Desiree Lyon, on Facebook or contact the APF.

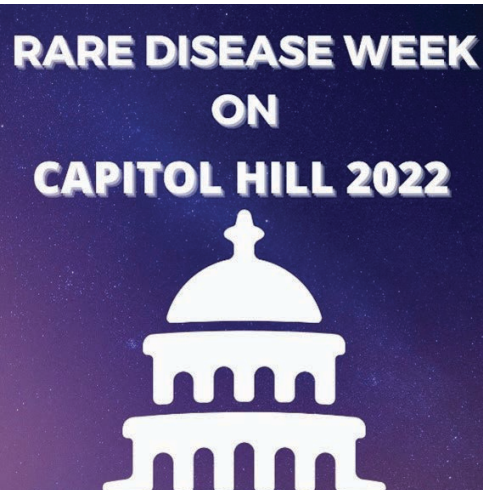


Natahsa writes, I have AIP and was diagnosed about 16 years ago when I had an acute attack in India where I live. I could not eat or drink, vomited like crazy and could not pass stool or urine. The doctors had put me on IV fluids and unknown medications. My stomach was being cleaned through a pipe inserted in my abdomen, and I lost consciousness. After 3 or 4 days of suffering, I was still not diagnosed. The doctors gave up on me and told my family to take me to another hospital as they could no longer help. After lot of research, my family shifted me to Fortis Memorial Hospital, where I was taken into the emergency where an endocrinologist diagnosed me with AIP. All I was told was never to have an empty stomach for too long and to eat a high carbohydrate diet. Within a year of being diagnosed with AIP, I began having seizures, including seizures while sleeping, cooking and even driving. After a lot of searching for safe medications, the Neurologists put me on medicines to try to help me though I still have seizures at time. Now that I am 45, I am starting menopause which brings on other complications like blood pressure. Thus, the doctors are once again trying to find safe medicines. It seems there is a never-ending process of research for Porphyria patients.



PORPHYRIA IN BERLIN

Another new group world-wide is PORPHYRIA Berlin. Although they are a small group, they are very active and enjoy their twice-a-year meetings. In fact, their leader, Sabine von Wegerer, says their meetings are like a "family reunion." If you would like to know more about the German Porphyria Association, PORPHYRIA Berlin, contact them by email at kontakt@berliner-leberring.de. Read about their group at www.porphyrrie-leberring.de.



RARE DISEASE WEEK 2022

Please mark your calendar for Virtual Rare Disease Week on Capitol Hill February 22nd to March 2nd. (Registration opens December 6th, 2021)

RARE
DISEASE WEEK
ON CAPITOL HILL

Rare Disease Week on Capitol Hill will take place February 22 – March 2, 2022. Sponsored by the EveryLife Foundation, this event promises to reach policymakers as rare disease advocates are educated on legislative issues affecting rare disease with lawmakers.

Rare Disease Day at NIH will occur virtually on Monday, February 28, 2022. Sponsored by the National Center for Advancing Translational Sciences (NCATS) and Clinical Center (CC) at the National Institutes of Health (NIH), Rare Disease Day aims to raise awareness about rare diseases, the people they affect, and NIH research collaborations under way to address scientific challenges and to advance new treatments.



RARE DISEASE DAY®

ARE YOU A CAREGIVER?

Are you a caregiver? If so, you have a story!

We would love to hear about your caregiving experience - what has helped and what has been really hard. The APF invites you to share your story with us so others may find strength and hope in your words. It is a great way to give back to the caregiving community and a powerful way to begin your emotional healing. The only requirement for a story is your honesty, as many find this to be the most cathartic release for both readers and writers. Interested? Email us at info@porphyriafoundation.org.



Influence your members of Congress by asking them to join the Rare Disease Congressional Caucus in support of those living with rare diseases: <https://rareadvocates.org/take-action/?v-src=%2f-campaigns%2f66800%2frespond>

PATIENTS ARE ASKING ...

The APF gathered the most common questions we receive, and expert physicians responded to each. These are available in the Frequently Asked Questions in the Patient Portal on www.porphyriafoundation.org. Bring us your questions and we will seek an expert response.

Here is a great example:



What types of things are helpful or recommended to maintain wellness (diet, medications, supplements, vitamins, etc.)?

For patients with an acute porphyria, it is important to have a healthy, well-balanced diet and avoid fasting or dieting. Excessive carbohydrates are not

recommended routinely. There are no specific recommendations for vitamins or supplements

Patients with cutaneous porphyrias can have low vitamin D levels as they avoid sunlight. We recommend a daily vitamin D supplement in these patients to maintain bone health.



PORPHYRIA AWARENESS WEEK 2022

MARK YOUR CALENDARS! APRIL 10-17, 2022

Share your ideas with us at: info@porphyriafoundation.org

Information to follow soon!



Longitudinal Study – RECRUITING

All Porphyrrias needed to participate! If not you, then who?

The porphyrias are a group of rare metabolic diseases that may present in childhood or adult life and are due to deficiencies of enzymes in the heme biosynthetic pathway. Porphyrrias have various symptoms depending on the type, but these can range from neurological symptoms to sun sensitivity. See the descriptions of each type to get more information. The natural history of these disorders is not well described, and it is not known why some patients are more severe than others. Therefore, the purpose of this long-term follow-up study is to collect a large group of patients with different types of porphyria and to provide a better understanding of the natural history of these disorders. The hope is that this information will help in developing new forms of treatment.

The research aims are:

- » To study the prevalence of specific indicators of disease severity. To study the effects on quality of life and health of various porphyrias.
- » To determine the relationships between disease severity and various biological characteristics, genetic information, and environmental factors.

PROTECT THE FUTURE PROGRAM



Our **Protect the Future** program to train future experts is important. Please consider donating to this program. Yours and your children's future health depends on each of us supporting the training of doctors who will know how to treat us and perform research when our present experts retire.

PANHEMATIN PREVENTION STUDY – UTMB

PANHEMATIN®
(HEMIN FOR INJECTION)

Are you currently using Panhematin® to prevent acute porphyria attacks? Dr. Karl Anderson, esteemed Porphyria Expert at The University of Texas Medical Branch in Galveston, Texas is currently seeking patients for a Panhematin® Prevention study. The purpose of this study is to determine if Panhematin® is effective for prevention of acute attacks of porphyria. We are seeking 10 patients who currently receive prophylactic preventative heme treatment. You may be receiving heme weekly, bi-weekly, or once a month and have successfully prevented attacks. Participation in this study will involve traveling to the study site for participation. Travel to the study site will be included.

Contact the APF today for more information and to be connected with a research coordinator nearest you. Email: info@porphyriafoundation.org

THE PORPHYRIAS CONSORTIUM CEP FOCUS GROUP

Researchers in the Porphyrias Consortium are conducting a focus group study for parents of children with Congenital Erythropoietic Porphyria (CEP) and adults with CEP to better understand the experiences of patients and parents, and the issues they face, and to gather more information about CEP.

We will be conducting the focus groups online over Zoom. Each group will be a discussion led by the researchers and will consist of about four to eight participants. You will only need to participate in one focus group, and it is expected to last for about 2 hours.

If you are interested in participating or learning more, please email and we will connect you with the researchers! Contact: hetanshi.naik@mssm.edu.



SCAN THE CODE TO DONATE TODAY

The APF is able to maintain our physician and patient education programs and many other services because of your support. Since we do not receive government funding, we need your support and donations. If you've changed your address or email, we'll also request your new contact information including your email address so that we can send you our monthly Porphyria Post. Update your information

here: <https://porphyriafoundation.org/get-involved/join-apf/>



REMEMBER ...

RESEARCH IS THE KEY TO YOUR CURE!

Visit porphyriafoundation.org for more information





PORPHYRIA WARRIORS

Scan to watch the video.




HOLIDAY GIFTS

ORNAMENTS, SNOW GLOBES, SHIRTS AND SHIRTS, OH MY!

Visit the APF store on our website to find holiday gifts for your loved ones. Visit our website for new merch!

WWW.PORPHYRIAFUNDATION.ORG



A MEDIA SUCCESS

Acute Porphyria patients, Terri Witter (top photo) and Alicia Gayle Bomboy (second down) and world-renowned porphyria expert, Dr. Sylvia Bottomley (bottom photo) recently were featured on a Fox 23 News program about porphyria in Tulsa, OK. It was a very successful porphyria awareness event.



The program featured Terri and Alice as patients suffering with the difficult challenges of acute porphyria, including frequent hospitalizations and unimaginable pain. They conveyed the unique difficulties people with a rare disease encounter, like isolation, inability to locate doctor to treat them and the almost impossible chore of sustaining employment while suffering such frequent hospital trips. The financial hardship brought on by lack of employment and ongoing hospital and other



medical expenses is also unbearable.

Dr. Bottomley adeptly provided the medical expertise needed to convey facts about the acute porphyrias.

Shortly after the television pro-gram, Terri had an attack and was hospitalized. She was able to attach the television program to her medical record, which became very important for the new doctors and nurses. Other patients might also consider using the link or using one of the many videos on the APF YOU TUBE channel or other videos that can be found on our website, porphyriafoundation.org, particularly those in which the experts are teaching. To add to her success, Terri educated the young doctors who were learning via her case. She had the APF send each of them a Doctor packet. Talk about making good of a bad situation!!!

We also encourage you to approach your local media and gain porphyria awareness, just like Terri Witter was able to do.

The APF can send you educational materials that will be very helpful to you. You might also encourage your doctor to participate, too.

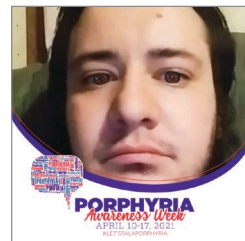
Thank you, Terri, Alicia and Dr. Bottomley.



CONGRATULATIONS, JESSICA MELTON!

Jessica has suffered from AIP since becoming a young adult. Hers was a difficult journey with many hospitalizations and many Panhematin infusions that saved her life. Now Jessica is on Givlaari, which has been a life changer even allowing her to work part-time and reduce pain medication. A longtime AIP sufferer, she was especially proud of being published in the November 2021 issue of *The Legal Secretary*. Her article was "Navigating the Field as a Person with invisible Disabilit." It relates her story about the difficulties of living and working as a paralegal with a disease like porphyria. In 2015, Jessica graduated with her associate degree in Special Education and obtained her paralegal certificate in 2017. Since graduation, she has been working in several different sectors of law.

A POEM FROM VICTOR LA FAE



Victor has suffered from HCP for many years, including many hospitalizations. He also is very active in our APF social media and has befriended many other patients. He has not only shared his poetry, he is also sharing the five novels he is writing with the porphyria community.

P-lease, Lord, let me live today.
O-n my own terms in my own way.
R-eally can't take this pain.
P-lease just give me peace.
H-ealth and strength.
Y-ou are all that can get me through.
R-ight now I don't know what I can do.
I-just want to disappear.
A-ll that's left is this single prayer.

PATIENTS FIRST CAMPAIGN

The APF has initiated a PATIENTS FIRST Program to return to our original mission of PATIENTS FIRST. Sometimes when foundations grow as large as the APF, they often focus on programs that leave the patient feeling as if they are on the outside looking in and not belonging to a community of others in a like situation. served directly. To illustrate, when Richard Howe was asked to present at a major medical conference like the one we held recently, he realized that his was the last presentation on the program. When he stood at the podium, he remarked, "As you can see, PATIENTS COME LAST." He certainly received every-one's attention with that remark!!!

We at the APF want to make sure that we place patients first in all of our efforts, but we need YOU to help us identify how we can improve our original mission to serve PATIENTS FIRST. Please email your suggestions to lyonapf@aol.com or call the APF office at **866.APF-3635**. We are here to serve you.

ARCHIVED PDF NEWSLETTERS

We have heard from many APF members that have enjoyed the APF quarterly newsletter over the years. In fact, the first newsletter was published in 1984. "Remember... Research is the Key to Your Cure" has been included in every single newsletter since the first! An archive of all printed publications is kept in the APF office, but you can access the materials on www.porphyrifoundation.org under "News" in the menu, which includes issues since 2005. Take a few moments to read about the research, physician efforts, conferences, meetings, and so much over the years. It is also a powerful reminder of how far we have come in research and treatment in the porphyrias!



APF hits 14,000 members! Let's keep growing! How can you help? Please send the names of your doctors so we can educate them more fully about your type of porphyria! Email info@porphyrifoundation.org.

CALLING ALL WRITERS

If you have a story, an inspiration, a memory, or experience to share, the APF welcomes your contribution to our quarterly newsletters. Patients learn so much from one another – and we welcome your thoughts and writing talent! Email info@porphyrifoundation.org to initiate.



THANKS
Thank you

As we launch into 2022, we want to take a moment to thank all our donors who supported our mission in 2021. Your generosity helped the APF make great strides in research and patient advocacy. We would also like to thank the generous employers of our donors who matched their employees' donations. Many employers make dollar for dollar matches. If you would like to help the APF cause and make your donation go even further, please consider checking with your employer to see if they have a matching program.

GET TREATMENT HELP

Do you have questions about FDA approved treatments for Porphyria? Please email or contact the APF today. We can also help you navigate Patient Assistance Programs if you are having trouble affording your medication or insurance premiums. We are here to answer any questions you might have. Call today: **866.APF.3635**



IS YOUR CONTACT INFO UP TO DATE?

The APF is able to maintain our physician and patient education programs and many other services because of your support. Since we do not receive government funding, we need your support and donations. If you've changed your address or email, we'll also request your new contact information including your email address so that we can send you our monthly Porphyria Post. Update your information here: <https://porphyrifoundation.org/get-involved/join-apf>



WE DID IT!

Thank you all for your incredible support! By taking the time to leave us a review with Great Nonprofits you've helped us earn top-rated status! We are so thankful for you!

The information contained on the American Porphyria Foundation (APF) Web site or in the APF newsletter is provided for your general information only.

The APF does not give medical advice or engage in the practice of medicine. The APF under no circumstances recommends particular treatments for specific individuals, and in all cases recommends that you consult your physician or local treatment center before pursuing any course of treatment.

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Our very own APF store is your perfect solution for last minute gifts and stocking stuffers. Amazon Smile is another great shopping option, and the company donates to American Porphyria Foundation. Simply click and choose the APF at <https://smile.amazon.com> or scan the code, and donations will automatically go to the APF at no cost to you!



SUPPORT THE APF TODAY

Your help is needed to educate physicians and patients and to support research. Become a member of the American Porphyria Foundation or make a tax-deductible contribution today.

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