



AMERICAN
PORPHYRIA
FOUNDATION

SPRING 2023 NEWSLETTER

IN THIS ISSUE

- Announcements | 3
- In Memory/In Honor | 4
- Cutaneous Porphyrin | 5
- Advocacy | 6
- Pharma Friends | 6
- Member Stories | 7

Do You Need a Doctor?

One of the biggest problems for patients with porphyria is locating doctors to manage their care. Because porphyria is rare, most doctors have never encountered a case. Now most experts won't take on a patient until they have a confirmed diagnosis. While this is understandable due to

The APF has a database of 5500 physicians treating porphyria.

the increased number of diagnosed patients needing care, it leaves others in the dark.

Patients with or without a diagnosis can call the APF to find a doctor near them. The APF has a database of 5500 physicians treating porphyria.

Patients nationally and internationally share their doctors with the APF. Physicians also contact the APF for educational materials. We also meet treating doctors at major medical

conventions and meetings, and add them to our list. In fact, at the recent Hematology convention, we added 100 physicians. If you need a treating doctor, please contact: 866-APF-3635 or general@porphyriafoundation.org

For international doctors, contact the APF or see this QR code for worldwide porphyria centers.



Elizabeth Buzney, MD New Scientific Advisory Board Member

We are proud to announce that EPP expert, Dr Elizabeth Buzney, has joined the APF SAB. Dr. Buzney graduated from Harvard University and completed her internship in internal medicine at Beth Israel Deaconess Medical Center, Dr Buzney now serves as the Director, Brigham Dermatology Associates at Brigham and Women's Hospital, Outpatient Clinical Director, Department of Dermatology, and Assistant Professor,

Harvard Medical School. She also recently became the President of



EPP expert, Dr Elizabeth Buzney, has joined the APF SAB.

the Photodermatology Society. Her patients are vocal about their love and esteem for Dr. Buzney and have often referred to her practice as "patient-centric." Particularly, that she listens to her patients and acts on their needs, combined with her medical expertise. Her excellent interview on skin cancer can be seen on YouTube. EPP people will be interested to know that Dr. Buzney provides the Scenesse implant. Welcome, Dr. Buzney!

ANNOUNCEMENTS

Nerve Root Pain in Acute Porphyria

Dr. Elena Pischik's PhD research is focused on characterizing porphyric neuropathy. She currently serves as chair of the Department of Neurology, Consultative and Diagnostic Center with Polyclinics in St. Petersburg, Russia. She made an important discovery, which focused on characterizing porphyric neuropathy. Most of her patients started experiencing low back pain before they developed limb pain and/or

weakness. Her research supports the hypothesis of porphyric neuropathy originating as a nerve root problem.

Porphyria expert, Dr. Mohamed Kazamel, looked deeper into the subject of autonomic neuropathy. His findings should enhance awareness and, in turn, put porphyric neuropathy in the forefront for healthcare practitioners when they encounter a porphyria patient with subacute or



Dr. Elena Pischik researches porphyric neuropathy, and discovered it originates as a nerve root problem.

acute limb weakness which occurred after low back pain.

We hope this groundbreaking finding will be an added diagnostic clue for acute porphyria, and will shorten the 15-year diagnostic journey average.

A Win for the Mexico Porphyria Society

Kika Shabot, leader of the Mexican Porphyria Society, spearheaded the effort to gain reimbursement for Normosang/hematin treatments for Mexico patients in acute attacks. Mexico will now pay for treatment for Normosang when people are having acute attacks—hooray!

The patients in Mexico join the APF in thanking Kika and the Sociedad Mexicana Para Porfiria/Mexico Porphyria Society and Recordati Rare Disease for the hard work to bring this lifesaving treatment to those in need. If you are from Mexico and need help, please contact the Sociedad Mexicana Para Porfiria.



Thanks to Kika Shabot, patients in Mexico will now be reimbursed for Normosang/hematin treatments.



Claire Richmond's podcast episode creates awareness of acute porphyria diagnosis.

Porphyria Podcast

At 33, Claire Richmond received an acute porphyria diagnosis after nearly two decades of searching for answers. In January, she appeared on a radio show broadcast to the state of Iowa called *Talk of Iowa*. Her episode "Rare disease advocate waited 19 years for

her diagnosis" is now a podcast and can be found on your favorite podcast app. Give it a listen!

Know someone with a podcast? Consider approaching them to do an episode on porphyria awareness. Let the APF know and we'll promote it!

ANNOUNCEMENTS

40th Anniversary of the Orphan Drug Act

As one of the original pioneers of the Orphan Drug Act (ODA), Desiree Lyon and the APF, we are keen to join in the celebration the 40th Anniversary of the ODA during Rare Disease Week celebrations in Washington, DC, Feb 28 – March 2, 2023.

This landmark legislation came into being to incentivize the development of treatments and cures for rare disease patients. This law came about by the hard work of pioneering patient advocates, like Desiree and APF members, as well as forward-thinking legislators from both sides of the political aisle. Each of them recognized that manufacturers needed to invest in rare diseases. Because rare disease drugs were considered to be too unprofitable

to pursue, few pharma companies invested in the drugs until the government incentivized them with tax advantages, etc through the ODA.

The bill was signed into law forty years ago on January 4, 1983 by President Ronald Reagan, making rare disease treatments more favorable for pharmaceutical manufacturers. The ODA resulted in the development and approval of approximately 700 rare disease drugs and biologics thus providing treatment for thousands of children and adults suffering with rare disease.

Although the ODA has brought great benefit and hope to the rare community; approximately 95 percent of the more than 10,000 known rare diseases still do not have FDA-

approved therapies. Since this rare disease crisis is not solved, those of us with rare diseases need to continue to try to protect our fellow sufferers by encouraging our legislators to find means to enhance rare disease drug development. Let's use the 40th Anniversary of the ODA to create momentum for more drug development and more advocacy for rare diseases.



Read news coverage on Desiree's Orphan Drug Act's efforts.

Porphyria Awareness Week: Spotlight Porphyria

"Spotlight Porphyria" is the theme of our annual Porphyria Awareness Week (PAW), April 15-23. PAW is the time of year that our members take on awareness activities suggested by the APF or created on their own design. Heightening awareness is of ultimate importance as the public, along with the medical community, have the opportunity to learn about the porphyrias.

In years past, our members have delivered brochures to clinics, hosted presentations on their cases to hospitals, and gotten local news to do a story. Our members have held

motorcycle races, mud wrestling, and midnight runs to enhance porphyria awareness. Your activities are far more effective than those of the APF because YOU have the disease and your story is powerful.

Throughout the year and especially during Porphyria Awareness Week, the APF will provide tools to our members to help our members advance the unique challenges and burdens that people with rare diseases encounter every day.

Don't know where to start? First, please send the APF your story. Nothing, absolutely nothing, moves



people to action more than the testimonies of those facing porphyria challenges head-on. Watch for details on the website, Enews, and social.

ANNOUNCEMENTS

APF Member Testimonial

If you need help with diagnosis, locating a doctor, getting treatment, contact the APF at 866-APF-3635 or general@porphyriafoundation.org. Read how Stephani was helped.

Thank you APF so much for our conversation yesterday. It was a pleasure to speak with you and so informative. I am deeply grateful for all you have done and continue to do to help the porphyria community. All of your persistent advocacy for and professional representation of all of us enabled me to be diagnosed within a few months of onset of symptoms, gave my doctor at the time a course to follow when he was dumbfounded, and provided

a resource for me to learn and understand what was for me an initially very confusing diagnosis. My brother was able to avoid misdiagnosis of MS a few months later for his historic undiagnosed episodes because I had my ER packet to show his doctors. I had the opportunity to share my patient perspective with medical students. All of these things which made our family's Porphyria journey so much less difficult than so many others' experiences were thanks to the groundwork the APF put in place. "Thank you" doesn't come close to expressing the gratitude I have for all you have done as our Champion.

Cheers + Tears

APF member, Terri Witter, suggested we host a Tears and Cheers column. It will feature news from our porphyria community, both cheers and tears.

Cheers: Congratulations Sian Ball, who despite a battle with EPP, ongoing liver damage, aplastic anemia, facial scars and a breast tumor, she pushed on and even graduated 6 months early.

Congratulations to Elizabeth Brounger upon her graduation from the University of Georgia.

Tears: We convey our sympathy to Cathy Bass who recently lost her mother, Betty Wallis.

APF FRIENDS OFTEN GIVE DONATIONS IN MEMORY OR HONOR OF THEIR DEAR LOVED ONES. WE ARE GRATEFUL FOR EACH OF THEM.



In Memory

Maureen Curren for Dr. Peter Tishler
Desiree Lyon for Dr. Joseph Bloomer
Norma and Ronald Brown for Matthew Brown
Kathleen Toelkes for Donna Pagano
Carol and Michael Farina for Vincent Farina
Mary Mistretta for Margaret Jusko Purcell

Mary Mistretta for Ann Jasko
Craig Cloud for his wife Karol Cloud
Trisha Trzybinski for her father, Paul Sheehan
Dianna Poissant for Matthew Cole
Gasper Family for Robert Gasper and Tim Henige
Donald L Johnson for Peggy Louis Johnson

The following people kindly donated for Susie Young: Outback Presents, Susan Berman, Greg Young, Kim and Tom Scholtz, Diana Young, Amanda Cagan, John Valentino, Sharon and Randall Wixen, William Conn, Greg Young, Diana Young, Susan Poll, Philip Young, and Adrienne Young

In Honor

Connie Helleison for Jennifer Streeter
Janet and James Cumming for Desiree Lyon
Colby Rios for Bonnie Wood

Sally Ahner for Ryan Staley
Gary Millett for Diane Young
Greg Young for Diane Young
David Nelson for Marilyn Nelson

Along with our hundreds of individual donors, we also thank the Ruth Lily Foundation, the Isabel Allende Foundation, Robert Waller Trust, Network for Good, Amazon Smile, for their generous donations.

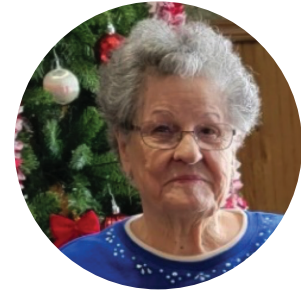


CUTANEOUS PORPHYRIA

Gasper Family Gathering

What started by Rose Gasper as a small family gathering during the Christmas season is now an annual event for 70 family members. Her six children attend with their spouses and kids. Each year they choose a charity to support. This year, they supported the APF because party host, Amy Burke, has Porphyria Cutanea Tarda (PCT). To enhance

awareness, Amy created raffle baskets for the family and added the APF overview brochures and memorabilia to each basket. Amy even made an APF basket filled with APF store items, as well as nuts and candy donated by the APF. The \$250 proceeds from the Gaspers gathering were donated to the APF. Thanks much!



This year's Rose Gasper's family gathering benefited the APF this year.

George Hodder's Story: Finding a Love for Mountain Biking Despite EPP

"Growing up with EPP is hard. In the beginning you don't realize that the reaction you have from the sun is any different than anyone else. Then at some point it hits you that you are different." George Hodder didn't have a name for EPP when he and his siblings suffered with symptoms as children. Over the years, he found a love for mountain biking and adapted his hobby to something he could do outdoors while minimizing symptoms. Now thanks to Scenesse®, he can be outside without worry and competes in mountain bike races along the Upper East Coast.

I am a 53 yr old and currently live in an area between North East and Central Pennsylvania. I grew up in the Pittsburgh suburbs and moved out to where I am now in 2007 for a job. I am the second of 4 kids and 3 of us have EPP. My older sister was the first to present with symptoms in early grade school. She/we used

to call it "the itchies" as kids. My parents, like all other parents without porphyria, had no idea what it was. Around second/third grade, when I started noticing symptoms, my parents first thought I was just copying my sister and/or thought that we just couldn't handle sunburn. Back then, there really wasn't any sun protective clothes, and nobody wore anything like that anyway.

Through the years my mom tried to get us medical help but we would typically be told to try sunscreen or have psychological screenings etc. When our younger brother (the youngest and 7yrs younger than me) started complaining of symptoms my mom pushed harder for help. She was able to get a referral for us to a Dermatologist. (this was back in the HMO days when you couldn't just go to specialists). Our dermatologist was newer and when we presented our symptoms she suspect porphyria

right away. She tested us for it by taking skin core samples and all three of us tested positive. We were so happy to finally have a name for the condition and some hope.

Use the QR code to read George's entire story. See info on APF bicycle jerseys on pg 6.



George Hodder can mountain bike despite EPP, thanks to Scenesse®.



Porphyria Presentation



Dr. Roy E. Smith is kicking off Porphyria Awareness Week with a Zoom presentation on April 15, 2023.

Renowned porphyria expert, Dr Roy E. Smith, is a professor of medicine at the University of Pittsburgh School of Medicine in the Division

of Hematology/Oncology. We are honored to have Dr. Smith start our Porphyria Awareness Week activities with a Zoom presentation for patients on April 15, 2023. During the Zoom call, Dr. Smith will teach on the porphyrias and also hold a question/answer session to familiarize patients and families with their type of porphyria and help them understand the questions they live with each day. Patients and their families and friends are welcome! Details will follow on the APF website, Enews and social media.

New in Store!

Bicycle jerseys inspired by George Hodder are destined for the APF online shop.



PHARMA FRIENDS

Our newest column features friends in the pharmaceutical industry who bring us porphyria treatments. Please meet Cara Hesse, Director of Patient Advocacy and Engagement for Alnylam Pharmaceutical, the manufacturer of Givlaari® (givosiran).

Although Cara grew up in Bowling Green, Ohio, with a cornfield in her backyard, her dream was to travel the world and learn about other cultures. Her dream came true when she spent six years working in South America (Guyana) and West Africa (Ghana). The first nine years of her career were spent focused on supporting countries going through a democratic transition. Cara says, "I monitored elections and worked to get more women involved in politics. I worked with the media, political parties, parliaments and activists.

It was awe-inspiring to see people walking 20 miles to get to a ballot box to cast their first vote ever!"

The second part of her career was focused on global health. where she says, " I was a global health lobbyist in Congress. Part of my role involved bringing Members of Congress to countries, like Ethiopia, to educate



This month's "Pharma Friend" is Cara Hesse, the Director of Patient Advocacy and Engagement for Alnylam Pharmaceuticals, the manufacturer of Givlaari® (givosiran).

them on how US funding for global health programs was spent."

The common thread throughout her career was partnering with small, local non-profit organizations to help them fulfill their mission and telling the individual story to spark action, including peace in the country, HIV or rare disease assistance. In fact, Cara has had ten years of experience in the rare disease community.

Cara says, "I believe we all have a fundamental right to good health, no matter where we live. That has been my passion." Outside of work, her passions are gardening and sailing.

The APF Director comments, "When I met Cara, I was immediately struck by her sincerity, knowledge, and tranquil demeanor. She listens intently when we bring issues to her attention and responds quickly. Thank you, Cara."

MEMBER STORIES

APF Member Stories: Nicole & Eric

NICOLE CASTELLANO

Nicole is a powerful communicator and is very experienced in the media field. She has been featured in a major television program to promote porphyria awareness, hosted zoom calls to promote APF activities and will be teaching patients how to gain media attention in their local areas. Nicole's been volunteering for the APF for over five years, including heading fundraising activities and translating for us. She speaks Spanish and Italian, which has been significant to our global program.

My name is Nicole Castellano. Before my diagnosis, people described me as a firecracker. I was a pilot, a world-



Nicole Castellano's story and advocacy impacts countless in the porphyria community and beyond.

class skating athlete, a business owner, and an outgoing person who embraced life. I had just graduated from college and started my own business. Things were going great!

Then suddenly, this excruciating abdominal pain came crashing down laying me flat on the ground. I had to crawl to the side of the road, collapsing into a field. The pain was so bad I literally thought, "This is it; I'm dying."

I spent the next 12+ years in and out of hospitals seeing gynecologists,

gastroenterologists, endocrinologists, neurologists, pulmonologists, and pain specialists. I had nine abdominal surgeries, including an unnecessary hysterectomy, which dashed my dreams of having children. Nothing changed, and the pain and weakness continued.

I finally met with a doctor who suspected porphyria. He ordered a urine test and confirmed the diagnosis. Finally, I had an answer!

After being in the dark for years, there was finally a path back to the light. The APF was a great resource to medical information, doctors and to connect with others with porphyria, and that has been very healing.



See *The Balancing Act* TV show here.



Read Nicole's entire story here.

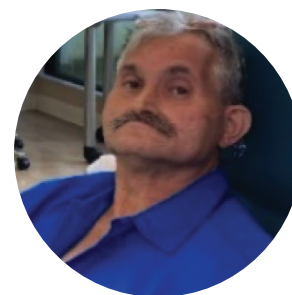
ERIC LIPSCHITZ

Eric is one the first APF members! Since its inception over 40 years ago, he's been active. Eric suffers from AIP. He's endured many critical acute attacks and subsequent hospitalizations, and has survived them all. In fact, every time he's hospitalized for yet another attack, he reminds APF staff that he's a survivor and will keep on fighting. Eric

epitomizes our "Spotlight Porphyria" Awareness Week theme by taking every hospital visit and clinic infusion trip as an opportunity to teach his nurses and doctors about porphyria. Eric lives in South Florida and is a Facebook member.

If you live nearby and want a porphyria friend, Eric is always willing to share his experience with others. If you would like to communicate with

Eric or other patients who live near you, please contact the APF.



Eric Lipschitz was one of the APF's first members, 40 years ago!

What's New?

Check out www.PorphyrriaFoundation.org

The information contained on the APF website or newsletter is provided for 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 consult with personal physician or local treatment center before pursuing any course of treatment.

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Donate to the APF

Donate on our website 24 hours a day,
7 days a week. Thank you!



**THANK
YOU!**

The APF does not receive government funding. Your contributions help educate physicians and patients with life-saving information about the porphyrias. Donations are tax deductible. Become an APF member today!

UPDATED CONTACT INFORMATION?

Contact 866-APF-3635 or
general@porphyriafoundation.org.

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