

IN THIS ISSUE

Pharma Friends | **2**

Announcements | **3**

Cutaneous Porphyria | **4**

Acute Porphyria | **5**

Advocacy | **6**

Member Story | **7**

In Memory | **8**



May you and yours be blessed with the spirit of the season, the joy of the festivities, the love of family and friends, the hope of a wonderful future, the peace of a fruitful life and improved health now and in the coming year.

Your friends at the APF—Amy C. Pax, Amy B. Yvette, Carol, Deborah, Christie, Elizabeth, Lelia, Susan, Virginia, and Desiree

A New Memoir About Caregiving for HCP



Matthew Spaur wrote a new memoir about caregiving for his wife with HCP.

Matthew Spaur published a new memoir, "Making a Small Fortune," the story of caring for two family members with hereditary coproporphyrin (HCP) while running a family-owned newspaper. At the start of the 21st century, Matthew

remarried and started a weekly newspaper with his new wife, Connye. Soon, Wall Street's tech stock bubble burst, launching a recession. Then business really took a turn for the worse after 9/11.

Meanwhile, Connye became sickened by a disease that had plagued her years earlier. Initially, she searched for less stressful work, but by the end of the year was on disability. At 35, extreme pain and nausea led to her need for a walker.

After exploring several other theories, a rheumatologist recommended porphyria lab tests. Connye's test results confirmed a HCP. In another six

months, her teenaged son was also diagnosed with HCP.

Matthew found that starting a small business is one of the hardest and most important jobs around—until you become a caregiver for people with porphyria.



Buy a book and donate to the APF! Matthew is donating \$5 for each of the first 200 copies purchased by friends of APF. Simply email a proof of purchase to amy@porphyria-foundation.org.

PHARMA FRIENDS

The relationship between pharmaceutical companies and patient organizations is synergistic. They manufacture the drugs that keep patients alive, and we help educate about the new life changing treatments. Introducing: Pharma Friends! Because we don't always get to meet the people who work behind the scenes.

An Interview with Recordati's France Lebel, M.Sc.

France Lebel is the Director, Medical Affairs Canada and Metabolic North America at Recordati Rare Disease. Her role is to lead the medical teams in Canada and US. "We are responsible for medical education, we answer question about our medicines. We also work hard with the health care community to increase disease awareness that would lead to better patient care."

She believes the best part of her career is when a patient receives a diagnostic that leads to appropriate care and to better quality of life and when patient associations can grow and be successful with our support. "I hope the impact I am making in the porphyria world is to help physicians connect with experts, patients with physicians and that it all contributes to better patient care,"

she said. Her position does not put France in direct contact with patients who are active members of patient associations to discuss educational needs and collaboration. That is why collaboration with the APF is helpful to understand what is important to patients and their doctors.

In fact, France says she has learned via the APF that "patients can and should take a more active role in their own care and that patients can be efficient

partners in their own treatment team. They are smart individuals who know the disease first hand and teach us every day about it."

Family is key in her life. France and husband, David, have been married since 1999. Additions are her younger sister, her grown children and her 8-month old granddaughter Marie-Laurence. Her daughter Emilie, lives in the UK with her partner James and Marie while Louis lives in Montreal and Charles is in Ottawa.

Aside from hard work, France enjoys reading novels and biographies, crafts and painting though she admits she is not very good at her artistic endeavors but does make her own Christmas cards every year. Maybe France will send us example of her cards for us to share. Thank you, France!



France poses with her eight-month old granddaughter Marie—look at that resemblance!

Pharmaceutical and Assistance Programs

PANHEMATIN

Contact Panhematin.com to learn about the Pal Program. Pal provides one-on-one support for patients and their caregivers that may include answering questions, helping with access issues, and connecting resources.

GIVLAARI

Alnylam Assist is an assistance program for acute patients in need of Givlaari treatment. Alnylam Assist offers personalized, comprehensive support for patients throughout the treatment process. Visit AlnylamAssist.com or call 833-256-2748.

SCENESSE

Clinuvel manufactures Scenesse for EPP and has a savings program that may be available to cover out of pocket costs of treatment for commercial, private health insurance, Medicare, and Medicaid. See Scenesse.com to begin the process.

HEALTHWELL FOUNDATION

Healthwell Foundation is a resource that fills the gap by assisting with copays, premiums, deductibles & out-of-pocket expenses when Health Insurance Is Not Enough. Contact healthwellfoundaiton.org or 800-675-8416.

Packing up for AASLD

American Academy for the Study of Liver Disease (AASLD) hosts an annual meeting that is attended by hepatologists, nurse practitioners, pharmaceutical companies and health care professionals from around the world. The APF maintains an exhibit booth each year to distribute porphyria information and answer questions about the porphyrias. Gudrun Debes, PhD, a patient, immunologist and patient advocate, volunteered at the APF booth along APF Director, Desiree Lyon. Conventions are a primary venue to provide educational materials.

The APF is exhibiting at the American Society for Hematology in New

Orleans December 10–13th. If you would like to assist us in the booth, contact the APF at 866-APF.3635. Your help is greatly appreciated.



Gudrun, who lives with AIP, and Desiree on their way to work the APF booth at the AASLD Annual Meeting.

APF friends often give donations in memory or honor of their dear loved ones.



IN HONOR

Diana Sabella by George Rusnak
Josephine Dzygala by APF staff

IN MEMORY

Susie Young, beloved wife of APF Trustee, James Young, by Warren Hudson, Ron Polly, Nicole Castellano, Paul Stickler, Lelia Dodson, Desiree Lyon, Amy Chapman, Amy Burke, Pax Cummings, Deborah Fedele, Elizabeth Brougher, Carol Hughes, Yvette Strange, Susan Smith

Michelle Ann Mobley by Family and Desiree Lyon

Lisa McKinney Smith by Lynne Collins, Family and Friends

Vincent Kuklewski by Carole

Woodrow Thompson by Marilyn

Joan Weeks by WC Weeks

Our deepest sympathy to Pharma Friend, France Lebel, for the loss of her father

Ring Physician and Porphyria Expert



Dr. Heflin, porphyria expert, working as Ring Physician for the Nevada Athletic Commission.

You may have seen Dr Joseph Heflin on the ESPN network as a leading Ring Physician for the Nevada Athletic Commission. For the past 30 years, Dr. Heflin has been ring side carefully monitoring the medical safety of the boxers over the boxing matches and now the mixed martial arts matches. He is also charged with stopping the fights when the fighters health is at risk. In fact, the photo reflects Dr. Heflin noting a medical necessity.

You may not be aware that for the past 30 years, Dr. Heflin also is in the porphyria fight. As an internist and emergency room physician, he began his interest in the porphyrias when multiple family members were diagnosed with AIP. Before he became a physician, his relative was

near death in the hospital with an unknown disease. He remained at the bedside night and day until NIH provided a diagnosis of AIP. Thus, began his desire to help patient with porphyria. Dr. Heflin has treated all types of porphyria in his career.

In fact, Dr. Heflin recently provided the APF with a video on the Frequently Asked Questions (FAQ) which is posted on the APF YouTube channel (access using the QR code below).



A Scenesse Success

Katie Huber Welty has erythropoietic protoporphyria (EPP) and has spent her life avoiding the sunlight. Now she is on the Scenesse treatment and proudly posted the following wonderful story on Facebook.

"I spent the morning at the zoo *in flip flops*. Mowed my entire lawn yesterday in my tank top (this is a two hour job in the sun). Last week, I rode roller coasters all day at an amusement park *with no gloves on!*"

At 8 weeks out from her final Scenesse implant for the year, Katie said she knows these moments "are numbered." She's grateful the FDA approved this medication. "My second summer on Scenesse has been nothing short of a dream come true."

If you want to learn about Scenesse for EPP and XLP, or gain access to the Scenesse treatment see www.Scenesse.com.



Thanks to Scenesse, Katie spent a sunny summer day with her family at Star Wars: Galaxy's Edge in Disney World.

A Journey with CEP

KHAWAJA MUHAMMAD ASIF,
lives with CEP



My name is Khawaja Muhammad Asif and I live in Azad Kashmir, Pakistan. I am 26 years old and have congenital erythropoietic porphyria (CEP). It's is an inherited disease. my mother has a history of CEP. My sisters do not have this disease, but my brother does. One of my brothers died of CEP.

For the past four years, I've had hand and skin problems. My finger joints are getting hard; my index fingernails are bending and getting thick. Blisters on the back of the hands, heart rate elevation, breathing problems, and abdominal pain are problematic for me. I live in a village where there is a lot of hard work to

do. I've learned to manage it. The last sunburn attack happened three years ago and took two months to heal. It left scars on my face. I remember four severe sunburn infections in my lifetime.

Burns and blisters caused by the sun heal very slowly, but wounds and blisters caused by friction heal quickly. The skin on the back of my hands is rough, dry and darkening. It becomes totally black and I need to wash it many times a day.

The skin of sun-exposed areas is weak and thin. The skin on the lower portion of my nose and face goes back to normal when I avoid going into sunlight.

I avoid direct contact of eyes to sunlight. Unfortunately, I have lost most of my lower eyelashes which have made my eyes vulnerable to dust infections. I cannot watch TV and work on my laptop late at night.

I've worked as a computer lecturer for three years in a government college. I have an hour walk to work each day in the sun. I never took any medications, just used sun block, which is not effective.

There are many cases of CEP in our region. In our village, one family including brothers and sisters are equally affected in a very bad way. You will see in my pictures I've managed well.

PBG Rapid Test

Because of the number of requests about the new Teco tests, the APF is repeating Teco information.

Teco Diagnostics has developed a urine rapid test to aid in the screening for the acute porphyrias, AIP, HCP, and VP.

Teco leader, Aquil Merchant, explained, "It can take an average of 15 years to get a diagnosis for this disease, which from a patient's perspective, can equate to a lot of unnecessary suffering."

The test is currently available For Professional Use Only and can be purchased directly from Teco Diagnostics. Information to order: 1-800-222-9880, tecodiagnosics.com.

In Memory: Susan Young



Susan Young was the beloved wife of APF co-founder, James Young.

Susan Young, the beloved wife of APF co-founder and president for 40 years, died at home on November 10, 2022. Those of us who knew Susie mourn her loss and join all of you in sending our deepest condolences to James. Susie was an independent spirit and an inspiration for the APF. Thank you for the donations made in her memory and for the many kind expressions of sympathy that have poured into the APF.

REMINDER

Since the incidence of hepatocellular carcinoma (HCC) is higher in acute porphyria, remember to have a

liver ultrasound at least annually. Regardless of attacks, patients with AIP should be screened at age 50.

Eryn's Road to an Acute Porphyria Diagnosis

Eryn was in the prime of her life when porphyria turned it upside down. Walking with her daughters one day, she suddenly felt ill. She went to a nearby emergency room, but was released with no diagnosis. Sadly, within a few weeks Eryn was ill again. Because her unique symptoms worsened quickly and she still had no medical answers, doctors did not hesitate to tell her the disease was in her "head." They were wrong.

Eryn was debilitated with nausea, inability to walk, seizures, and psychiatric issues without explanation. The symptoms grew, as did her outlook on life. She endured 60-plus trips to the ER and even more doctor visits. Finally, she decided to go to

one last appointment with a new dermatologist, who changed her life.

Upon examination, the doctor noticed one tiny, but recognizable bump on her little toe. He told Eryn it was a "shot in the dark," but could reveal an answer. He ran acute porphyria screening tests and was aghast when the results were positive!

Early in her disease awareness, she remembers being wheeled into the hospital. "I was on the phone with Desiree at the APF, who was calming me, educating me, encouraging me, praying for me, telling me I was not alone. I clung to that hope!"

With the APF as a resource and with doctors who cared about her, she began taking Panhematin. She then

discussed Givlaari with her doctors and together they decided to try it. It took over a year for her to level off, but Eryn is now doing well using both treatments as needed. Eryn says, "Panhematin saved my life and Givlaari keeps my life going better!"



Erin fought for her AHP diagnosis, but with help from the APF is finding her life again.

Clinical Research and You

What is a clinical trial? It is a research study in which human volunteers answer specific health questions, including testing a new treatment. Carefully conducted clinical trials are the fastest and safest way to find treatments that work in people and ways to improve health.

Why participate in a clinical trial?

Participants play a more active role in their own health care, gain access to new research treatments before they are widely available, and help others by contributing to medical research.

What happens during a clinical trial?

Trial teams include doctors and nurses as well as social workers. They check the health of the participant at the beginning of the trial, give specific

instructions for participating in the trial, monitor the participant carefully during the trial, and check after the trial is completed. Some clinical trials involve more tests and doctor visits than the participant would normally have for an illness or condition. For all types of trials, the participant works with a research team.

What are the benefits of participating in a clinical trial?

Clinical trials that are well-designed and well-executed are the best approach for eligible participants to play an active role in YOUR own health care. YOU gain access to new research treatments before they are widely available. You can help others by contributing to medical research.

Bitopertin Research for EPP

AURORA study designed to evaluate bitopertin as a potential disease-modifying treatment for adults with EPP in the U.S.

The Aurora study is designed to assess changes in protoporphyrin IX levels, safety, tolerability, photosensitivity and other measures in a double-blind, placebo-controlled setting.

To learn more about the “Study of Bitopertin to Evaluate the Safety, Tolerability, Efficacy, and PPIX Concentrations in Participants With EPP,” call the APF, 866 APF-3635.

With Medical Research, You Can Be a Hero

If you have porphyria, please consider participating in a clinical trial to evaluate a new treatment or gain more understanding of your disease. Research offers YOU a chance to improve your and your family’s life. Meghan Rohn is a medical hero and has been a research volunteer.

Meghan was fortunate to have been diagnosed with EPP at age three by a dermatologist. Sadly, she had to remain inside much of her life to quell the terrible sun reactions and missed playing outside with her peers. When she ventured outside, she was dressed in the usual long sleeves, hat, gloves etc that was appropriate in the winter

but difficult in the summer heat. Kids taunted her by not speaking or playing with her, as she might be contagious. The EPP was also hard for her family to plan a vacation or handle everyday life without considering Meghan’s condition. Photosensitivity had to be in their plans. Even her younger sister often stayed at home with Megan so she wouldn’t be alone.

Things improved when she began to have high school and college friends who formed a bond with Meghan and helped her cope with the EPP. Meghan now benefits from treatment because she participated in research!

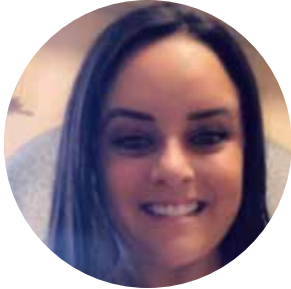
I am constantly monitoring my surroundings. Sun reflects off water, buildings, cement, driveways, glass, etc. I ask people to switch seats with me in the car to be on the less sunny side. Some people think I am seeking attention or acting crazy when I am protecting myself.



MEGHAN ROHN, lives with EPP

A Life-Changing Port

By Arielle Dimino



Arielle lives with VP and recently confronted her fears to get a port placed, which has made getting treatment easier.

Where to begin? I was diagnosed with varigate porphyria (VP) in July of 2019. Prior to being diagnosed, I went through four months of being in and out of the hospital with the most excruciating stomach pain and debilitating nausea I have ever experienced. I was beginning to feel hopeless every time I would leave the hospital without answers. After countless tests, procedures, hospital stays and misdiagnoses from other hospitals, Bryn Mawr Hospital was able to diagnose me through a twenty-four-hour urine test.

After months of being poked, my veins started to collapse, and scar tissue began to form. I had PICC line after PICC line and finally my Hematologist spoke to me about

getting a port. There was no question in my mind that this is what I needed to do. I scheduled my port placement for August 16th. To say I was petrified would be an understatement. I tossed and turned all night the night before and felt like I could get sick at any moment. I went in the morning not knowing what to expect. They did blood work and I changed into my robe. I was told that I would be having a conscious sedation and that made me even more nervous. I have never been a fan of needles, so that scared me too. They finally wheeled my back to the operating room and started prepping me. I'm not going to lie, it's an eerie feeling being awake in the operating room.

The next thing I knew, I felt a few numbing needles in my chest and neck and was given some medication to help me relax. The procedure lasted about 30–40 minutes and was so easy! I made myself overly nervous for nothing. The pain after the procedure was minimal. It felt like I had a stiff neck from sleeping wrong for a few days. I was on the beach the next day enjoying the sun and listening to the waves crash. I had to

stay under the umbrella because of the dissolvable, external sutures and glue, but it was so worth it. I have a very low pain tolerance and was concerned I was going to be in a lot of pain following this procedure, but I was pleasantly surprised. I have a little scar on my chest about an inch big from the procedure.

Getting a port was the best decision I could've made. I currently go twice a week for Panhematin treatments. Having a port has been life changing for me. Although I still feel a little pinch at each treatment, it is nowhere near as invasive as before. The first time using the port was intimidating and scary, but now I know what to expect. It's a piece of cake! I have developed such an incredible relationship with my nurse and am so comfortable with her. I'm thankful that I was given this life changing procedure and that I'm able to get my medication so easily. If you are contemplating getting a port, please do your research to understand the benefits. It was the best choice I could've made as I move forward with the management of this disease. And to think I used to be afraid of needles.

The Woeful Tale of Urine Testing

One of the most frequent complaints from people living with acute porphyria is about testing the urine for PBG, ALA and porphyrin analysis.

What should be a simple task can turn into a diagnostic nightmare! The most common complaint is that urine is left in the light for long periods of time before testing. It is

not uncommon for patients to be told to submit other samples, while continuing to suffer, waiting answers. Contact the APF for assistance with testing at 866-APF-3635.

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!



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?

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