



AMERICAN PORPHYRIA FOUNDATION

4th Quarter, 2020

WE WISH YOU A Joyous  
Holiday Season  
COUNTING YOUR MANY Blessings

The year 2020 has required patience and resilience. True to form, the porphyria community and our supporters have met the many challenges with monumental courage. Patients have shifted to telehealth and fought for diagnosis and access to treatment. The APF has moved to providing up-to-date education initiatives virtually. Our dedicated physicians and supportive pharmaceutical partners have reinforced us along the way. Through these renewed approaches to our health and wellness, one thing remains the same – the APF has maintained our relentless focus on supporting you. We will continue to fight for access to care and treatment, provide top-notch

education in a safe, effective manner – all in collaboration with our brilliant, world-renowned porphyria experts. We will continue our advancement of research initiatives while also advocating to our legislative bodies. And we will continue to spearhead awareness efforts across the country and globe.

We are honored to support you, and we are grateful for your gifts of time, talent and treasure to the APF!

We extend our heartfelt best wishes for a safe 2021.

*With Blessings, Your friends at the APF*

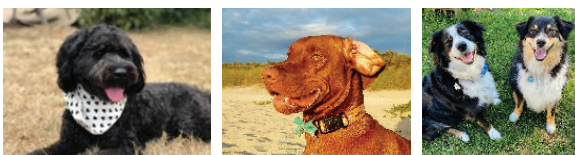


## APF 2021 PET CALENDAR

The APF held a 2021 pet calendar contest throughout October to benefit the critical **Protect the Future** program. We had nearly seventy adorable contestants who received

votes and donations, and who will grace the pages of the 2021 calendar - with the winner taking the front cover. Calendars are available while supplies last on the APF Online Store @ [www.porphyrifoundation.org](http://www.porphyrifoundation.org). We extend our gratitude to APF member NICOLE CASTELLANO, who led this initiative and put the fun in fundraising!

The **Protect the Future** campaign was established to attract and train the next generation of doctors and specialists in the field of Porphyria. Past generations of men and women have led Porphyria research, testing, and treatment for the last 40 years. Without financial support, we run the risk of losing knowledge of the disease, quality testing, diagnosis, and treatment, and ultimately – a cure.



Top three contestants (l-r: Frankie, Laszlo, Oakley & Sage)

## PORPHYRIAS CONSORTIUM – LONGITUDINAL STUDY

You have the power to influence research in the porphyrias by participating in this important study. Participants are needed across all porphyrias. The NIH-supported Porphyrias Consortium (PC) consists of six Porphyria Centers and eight Porphyria Satellite Centers across the USA. One of the CRITICAL studies that the PC implements is the Longitudinal Study (LS). The LS is an observational study to learn more about the natural history in patients with acute and cutaneous porphyria. You can find detailed information here: <https://www.rarediseasesnetwork.org/cms/porphyrias/7201>.

Have you already participated in the Longitudinal Study? If so, it may be time to update your information with the research coordinator at your participating site.

If not, all porphyria patients (adults and children) with a confirmed diagnosis are eligible and encouraged to participate. Please contact Edrin Williams via email at [edrinw@porphyrifoundation.org](mailto:edrinw@porphyrifoundation.org) or via phone at 301-312-8741 to contact the study coordinator at the participating location nearest you.

**“REMEMBER...RESEARCH IS THE KEY TO YOUR CURE.”**



### COVID-19 and PORPHYRIA

Updates and statements from porphyria experts will be listed on [www.porphyrifoundation.org](http://www.porphyrifoundation.org).



## MITSUBISHI TANABE – PHASE 3 CLINICAL TRIAL UPDATE



Thank you to all the EPP and XLP medical heroes that are already participating in this pivotal study! Whether you have received the active drug or the placebo, we appreciate what you are doing to further research and a potential treatment. Though the second half of the study is optional, it is a wonderful opportunity for each participant to experience the active drug. People have varying reasons for participating in research. Some do it for the kids, others for a chance for time in the sun, and still others to give back to their porphyria community. Whatever your reason...you are important! Participants are still needed!

Interested in learning more? The APF will share details of the study and connect patients (ages 12-75) with a study site. Contact Edrin Williams, APF Director of Patient Services, [edrinw@porphyriafoundation.org](mailto:edrinw@porphyriafoundation.org) or 866-APF-3635.

New study locations at Porphyria Centers have been added, including:

- » UTMB (Dr. K. Anderson) – Galveston, TX
- » University of Miami (Dr. C. Levy) – Miami, FL
- » Mount Sinai (Dr. M. Balwani) – New York

Patients can also participate at these sites:

- » Wake Forest Baptist (Dr. H. Bonkovsky) – Winston-Salem, NC
- » Dr. B. Freilich – Kansas City, MO


## SCENESSE® APPROVED IN AUSTRALIA!

Clinuvel Pharmaceuticals LTD announced that the Australian Therapeutic Goods Administration (TGA) has approved the registration of SCENESSE® (afamelanotide) for the prevention of phototoxicity in adult patients with erythropoietic protoporphyria. SCENESSE® is the first treatment approved for EPP patients in Australia. *Congratulations to our friends down under!*

## SCENESSE® IN THE US!

If you are pursuing treatment with SCENESSE® next Spring, do not delay starting the insurance process. This season proved that the entire process for reimbursement approval may take several months. Start by heading to [www.scenesse.com](http://www.scenesse.com) and apply to the Savings Program.

## ALNYLAM ACT – UPDATE

**AlnylamAct**  Alnylam sponsors no-charge, third-party genetic testing and counseling for individuals who may carry a gene mutation known to be associated with acute hepatic porphyria (AHP). There have been about 1,000 tests completed to date. The testing has resulted in approximately 10% positivity. If you are seeking genetic testing, please contact the APF for information.

## GIVLAARI® REMINDER!

Patients receiving GIVLAARI® treatment should ensure that injections are received consistently. According to the data resulting from studies, *once-monthly* injections of GIVLAARI® (Givosiran) led to a reduction in attacks. Patients should work with healthcare providers to ensure timely injections.

## FDA IDENTIFIES PANHEMATIN® AS AN “ESSENTIAL MEDICINE”

In early August, an Executive Order titled **Ensuring Essential Medicines, Medical Countermeasures, and Critical Inputs are Made in the United States** was issued. This order directed the FDA to identify a list of essential medicines that are medically necessary to have available at all times in the amount adequate to serve patient needs and in the appropriate dosage forms. **Panhematin® (Hemin for Injection) was listed as one of the 223 drug and biological product essential medicines.** The goal is to secure sufficient and reliable long-term access to these treatments. As we battle the current pandemic, this list of medications will protect against a future public health emergency. In collaboration with the Department of Health and Human Services (HHS), the FDA is also charged with addressing supply chain issues. The list, published October 30, includes medicines most needed in US acute care facilities specializing in short-term treatment for severe injuries or illnesses and urgent medical conditions. Essential medicines needed to meet certain criteria to be listed, such as ones necessary to address life-threatening medical conditions and used to stabilize patients.

## FDA – VOICE OF THE PATIENT

Remember the FDA Patient-Focused Drug Development (PFDD) meeting in White Oak, Maryland in March of 2017? Acute porphyria patients showed up by the dozens and shared their patient experiences living with this devastating disease. The Voice of the Patient (VOP) report was only recently posted on the FDA's *External Resources or Information Related to Patients' Experience* webpage. Listening to the video of the meeting and reading the report is a reminder of the impact of living with AHP.

## TREATMENT TESTIMONIALS

The American Porphyria Foundation (APF) is gathering testimonials from members about their experience with FDA-approved treatments (Panhematin, Scenesse®, GIVLAARI®). If you are

interested in sharing your experience with the APF, please include your response using the link:

<http://survey.constantcontact.com/survey/a07eh5fxy13kb-jishyq/start>

## PANHEMATIN® PREVENTION STUDY

PANHEMATIN®, hemin for injection, is used to halt and prevent acute hepatic porphyria attacks. Panhematin®, the first FDA-approved treatment for acute porphyria, is also often administered as a prophylactic treatment. Dr. Karl Anderson, the Porphyria Expert at the University of Texas Medical Branch in Galveston, Texas, is currently recruiting subjects for this clinical trial. Subjects must be diagnosed with acute hepatic porphyria (AIP, VP, HCP, or ADP), on a current regimen of prophylactic treatment (bi-weekly, weekly, monthly, etc.), and not receiving any other therapies to prevent acute porphyria attacks. Travel, including airfare, ground transportation, meals, and room accommodations, are covered.

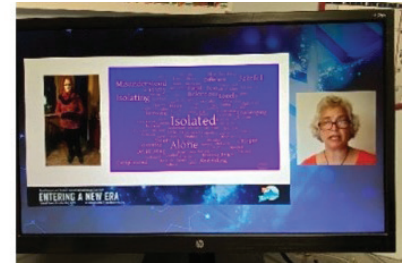
If you currently receive prophylactic heme treatment, you may be eligible. For more information contact Edrin Williams, Director of Patient Services, at [edrinw@porphyriafoundation.org](mailto:edrinw@porphyriafoundation.org) or 301.347.7166. Thank you to all participants!

## NORD VIRTUAL RARE DISEASE SUMMIT 2020



The National Organization for Rare Disorders held its' annual Rare Diseases and Orphan Products Breakthrough Summit, titled Entering a New Era, on October 8 – 9, 2020. The Summit covered the effects of emerging technology in rare disease, with public health and drug pricing being top priorities in government.

We are proud to share that APF member, Jennifer Beck, was a keynote speaker on behalf of the American Porphyria Foundation at the opening NORD Summit session. Jennifer shared her journey to diagnosis with Erythropoietic Protoporphyrin (EPP), the impact of living with EPP, and the transformation she experienced on treatment with SCENESSE. Thank you for your tremendous advocacy effort, Jennifer! Nearly 900 people attended the live programming. NORD attributes the success of the event to the extraordinary speakers. Jennifer, thank you for representing porphyria!



**FLU SHOT** **FLU SEASON IS COMING** .....  
Time to schedule your flu shot! According to Porphyria Experts, the flu shot is safe for all porphyrias.

## NIH COVID-19 SURVEY IN RARE DISEASE



The Rare Disease Clinical Research Network (RDCRN), which supports the Porphyrias Consortium, initiated a COVID-19 Survey related to rare disease. The survey was completed by nearly four thousand rare disease patients, including those with porphyria. Key findings include that the pandemic negatively affected access to regular health care, treatment

for the rare disease, special diet, special treatment and hospitalization, even among those who reported acquiring the infection. Findings also included that the pandemic caused mood changes/anxiety/stress in both the patients and their family members to an extent that required medical attention. The data is being further analyzed to understand the interaction between COVID-19 and rare disease. This survey mirrors the findings of the APF survey initiated last April.

## AWARDS



### ALNYLAM PHARMACEUTICALS RECEIVES INDUSTRY INNOVATION AWARD!

On October 9, NORD presented the 2020 Rare Impact Awards® as an online streaming event. The event celebrated the individuals, groups and companies that made extraordinary strides on behalf of the millions of Americans impacted by rare diseases. We are pleased to share that Alnylam Pharmaceuticals was the recipient of the Industry Innovation Award. Lina Rebeiz, who lives with acute porphyria, shared her personal story living with Acute Intermittent Porphyria. She highlighted her experience living with porphyria and her diagnostic journey. Lina presented the prestigious award to Amy Simon, MD, Vice Present Clinical Development, Alnylam. Dr. Simon expressed her sincere gratitude to NORD and shared the award with the porphyria community, who helped bring treatment to patients around the world.



### DR. MANISHA BALWANI, PORPHYRIA EXPERT, RECEIVES AWARD FOR EXCELLENCE IN PHYSICIAN COMMUNICATION

We are delighted to share that Dr. Manisha Balwani, Professor, Icahn School of Medicine at Mt. Sinai, has received the 2020 Cullman Family Award for Excellence in Physician Communication. This annual award honors Mount Sinai Health System physicians and advanced practice providers who demonstrate exceptional communication in clinical practice. Recipients of this year's award ranked in the top one percent nationally in provider communication for 2019 as measured by the Centers for Medicare and Medicaid Services' Clinician and Group Consumer Assessment of Healthcare Providers and Systems (CG CAHPS) patient experience survey. Congratulations, Dr. Balwani!

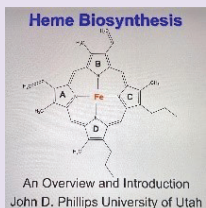
## APF VIRTUAL PATIENT EDUCATION

The daunting effects of the COVID-19 pandemic did not slow down the APF's efforts to provide essential, updated information, services, and patient education meetings. In addition to 7 patient education meetings held virtually, we also had multiple Porphyria Consortium informative sessions via social media. Topics covered included: COVID-19, diet & nutrition, and heme basics. The APF also hosted a summer series of patient-to-patient information seminars via Zoom for each of the specific porphyrias, as well as our pediatric and young adult communities. Stay tuned for even more critical initiatives to take place in the upcoming few months.



### Heme Basics

There is nothing basic about heme! However, Dr. John Phillips (Porphyria Expert, University of Utah) sought to introduce the heme biosynthetic pathway and the porphyrias to a level we can all understand. He shared how it all works and what goes wrong when you have porphyria. He started at the history of the field, educated on the chemistry of the porphyrias, through the molecular biology and gene and targeted therapies. According to Dr. Phillips, there is still much to learn about the porphyrias – and we are glad he is a powerhouse leading the charge. We thank him for sharing his expertise with us.



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### CEP Member Meet-Up

The Congenital Erythropoietic Porphyria (CEP) community had an opportunity to join together for a ZOOM meeting in mid-October. The meeting was prompted when a young patient shared that he

had never met anyone with his disease. We set out to fix that immediately! Dr. R. Desnick, Porphyria Expert, Icahn School of Medicine at Mount Sinai, began the discussion with information from the Porphyrias Consortium and the evolution of knowledge about this disease. We moved into questions and engagement among the group. No one wanted to leave – and we are delighted to host another gathering soon. One day, we hope to meet in person!

### Nutrition and Acute Porphyria

As diet is one of the factors that influences the manifestations of acute porphyria, Dr. H. Bonkovsky (Porphyria Expert, Wake Forest Baptist, Winston-Salem, NC) and Rebecca Mohning, MS (Nutrition/Dietician) presented nutritional guidelines that emphasized a high carbohydrate intake as part of a balanced diet that provides all essential nutrients. The goals of the recommendations are to prevent acute attacks of Porphyria that may be related to diet, avoid deficiencies of nutrients, and maintain a normal body weight. Dr. Bonkovsky led the presentation, with Ms. Mohning providing practical recommendations. There is a comprehensive Diet and Nutrition section on the APF website: <https://porphyriafoundation.org/for-patients/diet-and-nutrition/>.

### Coming Soon via Zoom!

#### GIVLAARI® Experiences and Information, Dec. 9, 6 p.m.

Dr. M. Balwani, Porphyria Expert, Icahn School of Medicine at Mt. Sinai

#### Genetics in the Porphyrias 101, Jan. 28, 7 p.m.

Dr. Hetanshi Naik, Icahn School of Medicine at Mt. Sinai

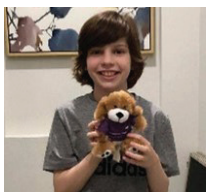
#### Research Opportunities, Feb. 25, 7 p.m.

Karli Hedstrom, MPH, Icahn School of Medicine at Mt. Sinai

#### Patient Education Meeting, March 4, 7 p.m.

Sean Quigley, MD, University of Illinois at Chicago

Contact [edrinw@porphyriafoundation.org](mailto:edrinw@porphyriafoundation.org) for information to join!



### ATTACK DOG!

You may have noticed that the APF has a new mascot. Our very own Max, the porphyria attack dog, has won over the hearts of our patient community. The APF recently sent our youngest EPP patient community members their very own attack dog. We received such kind and gracious thanks from many little warriors who found great comfort from the tiny pal. Of course, Max makes a great adult companion as well; and a great holiday gift. To get your own porphyria attack dog, go to the APF Online Store. Pictured: Landon Soares (EPP)



### RARE DISEASE DAY 2021

- » Rare Disease day is celebrated with awareness activities February 28! One in 10 Americans live with a rare disease. That's 30 million Americans who celebrate and are supported this day.
- » Rare Disease Day at NIH will occur virtually on March 1, 2021. 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 Week on Capitol Hill will take place July 19-22, 2021. 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.

**MARK YOUR CALENDARS! ..... APRIL 10-17**

**PORPHYRIA  
AWARENESS WEEK 2021**

We are gearing up to share our plans with you for Porphyria Awareness Week 2021. Do you have plans to raise awareness? Need APF support? Please send ideas to [lany.schneider@porphyriafoundation.org](mailto:lany.schneider@porphyriafoundation.org)

## NEW PUBLICATIONS

### Acute Porphyria and Neuropathy

A new publication by lead author, Dr. Mohamed Kazamel (University of Alabama Birmingham) was released in Current Neurology and Neuroscience Reports.

Porphyria Neuropathy: Pathophysiology, Diagnosis, and Updated Management (Mohamed Kazamel, MD; Robert J. Desnick, MD, PhD; John G. Quigley) reviews the peripheral neurological complications of the acute hepatic porphyrias, as well as recent advances in their pathophysiology and management. Key findings include that neurologists should appreciate the patterns of porphyric neuropathy and long-term outcomes depend on early diagnosis and treatment.

*Dr. Kazamel is a Neurologist and Porphyria Specialist at UAB and is supported by the APF Protect the Future program. Please contact the APF for a PDF of this research.*

### Erythropoietic Protoporphyrin and Underdiagnosis

A publication by lead author Dr. Amy Dickey (Massachusetts General Hospital, Boston, MA) and other esteemed porphyria experts, "Evidence in the UK Biobank for the Underdiagnosis of Erythropoietic Protoporphyrin," has been published in the Genetics in Medicine Journal. Erythropoietic Protoporphyrin (EPP), characterized by painful cutaneous photosensitivity, results from pathogenic variants in ferrochelatase (FECH). For 96% of patients, EPP results from coinheriting a rare pathogenic variant in trans of a common hypomorphic variant c.315-48T>C (minor allele frequency 0.05). The estimated prevalence of EPP derived from the number of diagnosed individuals in Europe is 0.00092%, but this may be conservative due to underdiagnosis. No study has estimated EPP prevalence using large genetic data sets.

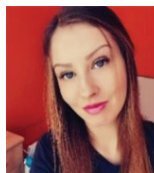
*Dr. Dickey is a Pulmonologist and Porphyria Specialist at MGH and is supported by the APF Protect the Future program. Please contact the APF for a PDF of this research.*

## GLOBAL NEWS

There is much planned on the horizon with our international friends.

On November 9, 2020, the APF hosted a Global ZOOM call for Spanish speaking friends with Kika Shabot, leader of the Mexico Porphyria Society, as moderator and Dr. Daniel Jaramillo as the expert presenter. Eileen Hudson, leader of the Chile Porphyria Society assisted with the planning of this important call.

Dr. Julian Andres Rameriz was the expert presenter for a second Spanish speaking ZOOM call with the many people in Columbia who suffer from porphyria. These calls were a resounding success and were a great means to learn while meeting other people with porphyria all over Latin America. Patients and doctors from Chile, Mexico, Columbia, Argentina, Spain, and other Central American countries attended the call.



The APF is also assisting patients in Poland in their attempt to gain access to the Givlaari treatment. Patrycja Tykowska (pictured left), who is heading the Polish patient group, has been diligently collecting patients who are having frequent attacks and are in great need to reduce the hospital stays. Attacks are gruesome no matter where you live in the world. Porphyria pain speaks its own language, and all patients understand it and therefore, understand each other.

Another exciting global collaboration is our upcoming ZOOM CEP call. Fide Miron (pictured right), President of the Spanish Porphyria Society, has CEP and will be assisting with the agenda of the call and hopefully will also be presenting. Fide is a courageous advocate for all porphyrias and has been a mainstay in the porphyria community in Europe. Now others around the world will have a chance to meet this wonderful woman. If you would like to participate in the CEP International call, please contact Desiree at the APF.



Next steps will be in India and South Africa!

## 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.porphyrifoundation.org](http://www.porphyrifoundation.org). Bring us your questions and we will seek an expert response. Here is a great example:

### What is "latent" porphyria? Does this mean I will never have symptoms?

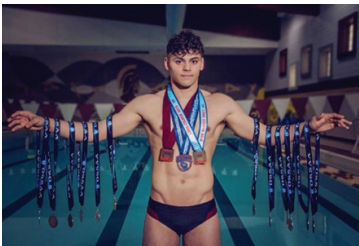
Individuals with a disease-causing mutation without symptoms have "latent" acute porphyria. However, this does not mean that such an individual will never have symptoms. Genetic factors (the presence of a porphyria-causing gene mutation) are not the only factors involved. Exposure to environmental factors, such as medications, can greatly influence whether an individual with a mutation in a porphyria-causing gene has symptoms. This is why it is important that all family members of individuals diagnosed with acute porphyria be tested whether they have symptoms or not, and that all individuals who have a confirmed diagnosis of acute porphyria be educated about and follow the recommended precautionary and preventive measures. The diagnosis of porphyria is always important medical information, even when there are no symptoms. It may, for example, influence the choice of drugs to treat other conditions, the choice of anesthesia for surgery, or dietary recommendations.



## WEDDED BLISS

Paxton Cummings (APF Assistant) married Holland Harries on the beautiful beach of Santa Rosa Beach, Florida on October 24, 2020. We congratulate Pax and his beautiful bride. The wedding was officiated by Desiree Lyon, APF Global Director.

## JAZZ WHITLEY



Heroes come in all ages and sizes. APF's latest hero comes

in the form of a young man who has not let EPP deter him. Jazz Whitley is an elite swimmer, who has committed to Alfred State College in northern New York. This is the time of year when many young people choose their colleges based on many things: academics, social life, future goals and, like Jazz, sports. However, Jazz had to add another consideration to his college choice list – the sun. Though it is clear, by looking at his medal collection, that Alfred College had an easy decision to make; the decision on Jazz's part was more complex.

As is true for most of his young life, Jazz has had to make decisions that most young people do not. Since the age of five he has had to navigate the many "whys" of his condition. Understanding why him, why it took eleven years to diagnose his EPP, why his body attacks him, why the sun on a beautiful summer's day at the lake could harm him, why he had to learn to live with the stigma of hiding his condition from his friends, why – at such a young age he has faced debilitating pain and sleepless nights when "every flare up felt like I was burning alive; like molten lava being poured on my skin", why he needs the darkest tinting allowed by law on his car, and why he has to shadow jump and cover-up head to toe. Though there may have been many times Jazz has wondered "why" he has never given up. He has become a powerful competitive swimmer with an offer from Speedo USA to join their #SpeedoSquad to help promote their swimming lineup. With the help from his loving family, Jazz has learned to turn his whys into why nots and is not limited by his EPP; but unlimited by the possibilities that his talent and determination have given him. Jazz best sums it up in his own words: "One good thing about being an elite swimmer with EPP is that I might be a positive role model for younger kids with EPP. By sharing my story, I hope they will see that if I can achieve my goals while living with EPP, they can too."

We wish Jazz the very best as he embarks on the next phase of his life.



## ACG AWARD-WINNING ABSTRACT

Congratulations to Dr. Herbert Bonkovsky (Porphyria Expert, Wake Forest University, NC Baptist Medical Center, Winston-Salem, NC) and his outstanding team for winning the Presidential Poster Award in the Liver category at The American College of Gastroenterology Virtual Annual Scientific Meeting. The study titled **Clinical Outcomes in Patients With Acute Hepatic Porphyria Treated With Givosiran Who Stopped Hemin Prophylaxis at Study Entry: A Post Hoc Analysis of Data From the Phase 3 ENVISION Study Through Month 12** is available here: <https://eventscribe.com/2020/ACGPosters/PosterSpeakers.asp?goToLetter=B&pfp=PosterPresenter>.

**Introduction:** Acute Hepatic Porphyria (AHP) is a family of rare genetic diseases due to enzyme defects in heme synthesis in the liver. Accumulation of toxic heme intermediates ALA and PBG may result in neurovisceral attacks and chronic manifestations. Intravenous (IV) hemin is approved to treat acute attacks and is sometimes used off-label prophylactically. In the ENVISION study in AHP patients, Givosiran<sup>®</sup>, an RNAi therapeutic, reduced the composite porphyria annualized porphyria attack rate (AAR) vs. placebo (pbo). A post-hoc analysis was conducted to evaluate outcomes in AHP patients with or without prior hemin prophylaxis prior to screening.

Pictured l-r: Sean Rudnick, MD (Sub-PI), Dee Faust (Project Manager), Herbert Bonkovsky, MD (PI), Michelle Moore, PA-C (Sub-PI), Tyler Bush (Clinical Studies Coordinator)



## THE IMPACT OF AN ACUTE ATTACK

Eryn Sallee (AIP) recently shared a post that highlighted the affect that an acute porphyria attack can wreak on cognitive function. Eryn shared that she was very sick before receiving treatment and was surprised that she wrote these words! Initially feeling that she should remove them, she opted to keep her words public as a testimony to the impact of an attack. "Exceptional human beings wrote me the most uplifting, powerful notes ever - and helped me swim through the toxins to see that I was still there, that my light was still shining."

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*No end in sight hospital beds scared beyond belief sick so suddenly pulled away from my babies alone so alone body is just a shell just a shell nothing is working walking eating talking feeling safe it is all broken genes broken*

*I don't know how I get through.this evil this torture this hell I can't remember did I did I last time, and the time before? and just last week I was laughing joking just yesterday I was reading to them*

*how is it so*

*I am.in here somewhere but my legs aren't working my thoughts a cloud my plans can never be plans my very cells are only parts of what they should be.poison in my veins and the medicine will stop it, slow it right but I'm*

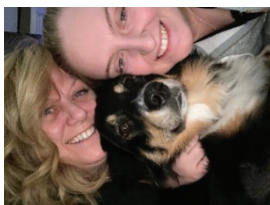
*trapped and it goes on and on no end in sight this bed these nurses always the same furrowed brow always the 'sorry, sorry'*

*But I'm sorry I'm sorry to my babies I miss you you are everything I'll find you*



CONTACT THE APF TODAY TO HAVE A  
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## GROWING UP IMPACTED BY EPP

Ginger Zona recently had an eye-opening experience when her daughter, Jayden, penned a beautiful essay about the impact on her life as the daughter of a parent with EPP. Here are highlights from Jayden's essay for her Reading Writing & Research I class.

Embracing the Shadows My life growing up was very different. Most children can wake up with a desire to play outside. On hot sunny days, they can go out and swim with the company of their parents, ride bikes, or play outside. On nice summer days, when children want to be pushed on a swing and have fun, they can have their parents push them. I could not have any of this. My mom

has a rare disease called Erythropoietic Protoporphyrria. Symptoms of this disease include excruciating pain, swelling, itching, burning, redness, crying, screaming, fidgeting, which all appear minutes after sun exposure.

There are physical and mental limitations that come along with this disease. My mom cannot cannot leave the house without her floppy black hat, with a face shield and sunglasses on her face, while wearing a long black sleeve shirt, with a black sweat-shirt over that, blocking out as much sun as possible. This brings attention to us. People look at us differently.

My mom wanted me to have a normal childhood. She wanted nothing more than to have the experiences a mother does. But to give me a childhood, she often sent me with my aunt. She was the one to play with me outside, experiencing it all. This

has been so hard on my mom.

During my freshman year of high school, there was a meeting with the FDA in Washington. The room was full of patients of all ages. We listened to their personal stories about how they grew up, and how they adapted to their insane worlds. We then listened to experts from all over the world speak about EPP.

This disease is devastating for not only my mother but our entire family. As a family, we manage the chaos that comes with the disease. We embrace the shadows during the day and welcome the darkness the night brings. We are a family who gets excited when it rains, our entire lifestyle revolves around EPP. For now, we will take what we can get and hope that with research and advocacy treatment will be available for all EPP patients all over the world.



## A SCENESSE® SUCCESS STORY

Darrell Klampe, one of the APF's oldest porphyria patients featured in our third quarter newsletter, has updated us on his success with Scenesse. Mr. Klampe, who has EPP, has had multiple treatments of the breakthrough drug. Stating that, although it took a while, he has started to see positive results and is very pleased. Mr. Klampe, who was born in North Dakota, had his first serious symptoms of porphyria only twelve years ago. He was forced to leave Arizona, where he was living at the time, due to his reactions. Despite this late onset, he recalls that his mother had a blood disorder resulting in an excess of iron in her bloodstream. Mr. Klampe now lives in the Pacific

Northwest – though he says he wishes he lived in Alaska, where he has traveled to on a few occasions. It is his hope that sharing his success story will help others suffering from EPP symptoms who are looking for relief. We wish the very best to Mr. Klampe for his continued success!

## IN MEMORY & IN HONOR



We thank the families and friends who memorialized their loved ones with a generous gift to the APF.

### IN MEMORY:

Sally & Bennie Bowden, John & Sallie Hammond, Rick & Heidi Myers for **Jerry Viands**; Susan Cerkoney for **Fred Cerkoney**; Donald Johnson for **Peggy Ann Lewis Johnson**; Lisa Kancsar for **Stephen Kancsar**; Mary & Frank Olsen for **Lori Elynn Stewart**.

### IN HONOR:

Steven Andrade for **Josephine Dzygala**; Amy Chapman for **Christie Brewer**; Sharon Dill for **APF Volunteers**; Anonymous for **Luca Fierro**; Eric Gray for **Ralph M. Gray**; Sharon Koch for **Matt & Wendi Koch**; Michael & Tracy Leppert for **Craig & Nicole Leppert**; Daniel Magaskie for **Nicole Magaskie**; Elaine Sasso for **Elizabeth Melani**; David Stamborg for **Melissa Nagin**; Amy Chapman for **Michelle Rogers**; George Rusnak, Jr for **Diana Sabella**; Rachel Wise for **Jere & Pauline Wise**.



## IN MEMORY

Robert "Bobby" Norment, an early supporter of the APF, was surrounded by his family when he passed on October 17, 2020. Bobby and his wife, Jean, provided financial and emotional support from day one of the foundation. Their son, Hunt, has provided technological expertise for our website, computer programs and database. Together, son, Davis Norment and the family founded a new type of educational adventure at their college, CreaED. The APF used some of the principals initiated in their experience-based learning system. We send our thanks and sympathy to the Norment family.

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> and donations will automatically go to the APF at no cost to you!



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### 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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