



AMERICAN PORPHYRIA FOUNDATION

3rd Quarter, 2021

ANNUAL MEMBERSHIP DONATION

Make it possible for us to carry out our mission. Together, we can do anything.

The American Porphyria Foundation has remained open, productive, and focused on our mission throughout the global pandemic. We have served both patients and physicians with a focus on research, education, awareness, advocacy, and patient support. Patients always come first at the APF – your calls, emails, issues and requests are #1 on our list each day. We ask that you renew your membership so we can continue to put patients, and their families, first. Thank you for your support!



PLEASE DONATE OR RENEW YOUR MEMBERSHIP TODAY!

The suggested annual membership donation is \$35, though we welcome and appreciate every single gift. To donate, please visit www.porphyrifoundation.org, call 866-APF-3635 or mail a check to 4915 St. Elmo Avenue, Suite 200, Bethesda, MD, 20814.

Together we can be a force for improved porphyria healthcare.

SEPTEMBER IS PAIN AWARENESS MONTH!



A common thread across all the porphyrias is PAIN. We have heard AIP – Acute Intermittent Porphyria – renamed to **“Always In Pain”** and Variegate Porphyria to **“Very Painful.”** Whether it is triggered by an acute attack, a cutaneous reaction, sometimes both, or chronic pain that has manifested after years of nerve damage, the pain our patient community experiences is exquisite. Pain in the acute porphyrias is often managed through opioids – the only safe medication that touches the horrific pain – which lands our patients directly in the center of the opioid crisis. The pain

of an EPP reaction doesn't respond to analgesics or medications, leaving time and darkness as respite. Creams and topicals may help the blistering porphyrias, but they don't relieve the fragility that leave patients wincing.

We are not alone in the porphyrias. According to a recent study at NIH, 25.3 million adults in the US suffer with daily pain.

PAIN AWARENESS MONTH, honored each year in September, is a prime opportunity to talk about issues related to pain – and remind us that we have a voice in advocacy and awareness related to pain.

A Pain Management Best Practices Inter-Agency Task Force was established in 2018 by the Department of Health and Human Services to propose updates to best practices and issue recommendations that address gaps or inconsistencies for man-

aging chronic and acute pain. The APF, with the support of several members, was vocal in public comment to ensure that the voice of rare disease – specifically a pharmacogenetic disease where attacks are actually triggered by unsafe medications – was included in the guidance. We are now serving on a committee organized by the U.S. Pain Foundation to help ensure best practices are implemented.

Please join the APF in sharing the **PAIN** that we as a patient community endure, and participate in conversations that help to lift our voices.



INVISIBLE PROJECT – A PUBLICATION OF THE US PAIN FOUNDATION

U.S. Pain Foundation's flagship publication, the INVisible Project, has released a new edition focused on health care disparities related to pain. Here is their latest release: Check out the latest issue of the INVisible Project, a magazine from U.S. Pain that includes profiles of pain warriors and articles with tips and insight! <https://invisibleproject.org/magazines/disparities-edition>.

HEME BIOSYNTHESIS AND THE PORPHYRIAS 2021: CONSENSUS DIAGNOSES, VARIANT DISORDERS, NEW & EMERGING THERAPIES

October 15-17, 2021

Hyatt Regency Schaumburg Near Chicago O'Hare Airport

Scientific Conference [IN-PERSON]

Heme Biosynthesis and the Porphyrins 2021: Consensus Diagnoses, Variant Disorders, New & Emerging Therapies, is to be held October 15-17, 2021, at the Hyatt Regency Schaumburg near the Chicago O'Hare Airport. This 2.5-day educational symposium will feature international and national experts who will present on 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 the acute hepatic and erythropoietic porphyrias. This is a must-attend for any physician interested in the porphyrias...and any patient who is wants to take a deep dive into the science of this group of diseases.

Patient Day [IN-PERSON OR VIRTUAL]

Don't miss out! The first international patient day since 2018 will take place in Schaumburg, IL (just outside Chicago) on Sunday, October 17, 9am-3pm Central. Vaccinated patients are welcome in person – and everyone is welcome over livestream! Registration Now Open!

Meeting Highlights:

- » Expert international faculty
- » Latest clinical & basic research advances
- » Talk tables during breakfast and lunch
- » Separate clinical session for patients
- » Symposium offers up to 25 CME credits

Register now: www.porphyrissymposium.com.



NORD RARE DISEASE EDUCATIONAL SUPPORT FORUM



Are you interested in attending the Porphyrins Symposium this October in Schaumburg, IL? NORD offers reimbursement costs of rare disease conferences and educational programs. Limited financial assistance is offered with the cost of travel to a rare disease educational conference or program and with the costs of lodging should an overnight stay be required in order to attend or participate in a rare disease educational program. How to apply? Contact NORD's Rare Disease Educational Support Programs, Phone: 1-860-556-2208 or Email: rdeducate@rarediseases.org.

FARM STAND FUNDRAISING!



The McKillop family grew a bounty of vegetables in their backyard vegetable farm this summer. From cucumbers, tomatoes and peppers to carrots and string beans, their backyard garden overflowed with produce – and so did their hearts! Organizing a farm stand exchange to raise funds for the APF, Morgan (EPP, age 11) and her family sought to give back to this organization that has offered support and friendship.

Morgan is active in the garden, as she and her parents and siblings tend to the veggies. Saving the hard work for the evening when the sun is low and bearable, their agrarian efforts have supplied three farm stand exchanges. The events have been a raving success, and were even featured in The Suffolk County News. The APF is grateful for the McKillops and others who are digging in to make life better for those living with porphyria! Hip hip HOORAY! Pictured: Colleen (mom) and Morgan; Ben (dad) and Morgan.





disc)medicine

DISC MEDICINE

announced in a recent press release that it has “entered into an exclusive worldwide licensing agreement with F. Hoffmann-LaRoche Ltd for the development and commercialization of bitopertin, an orally administered GlyT1 inhibitor with demonstrated effects on the heme biosynthetic pathway.” They have indicated that it intends to develop this drug as a treatment for hematologic disease, initially the erythropoietic porphyrias. APF will be closely watching this progress to share with our members.

RECORDATI PATIENT SUPPORT PROGRAM LAUNCH!

PANHEMATIN® (HEMIN FOR INJECTION)

Have questions about Panhematin®? Here is another resource for answers. Recordati has launched a new program to further support our patient community. The “PAL”, or Patient Advocacy Liaison, program provides one-on-one support for patients and their caregivers. Support can include answering questions such as details about Panhematin® and how it works and helping you if issues arise when accessing Panhematin®. To access this support, visit <https://www.panhematin.com/support> for the opt-in form. These services are at no cost to the patient. We appreciate the dedication of Recordati as they seek to provide resources that are beneficial to those we serve.



COVID-19 VACCINE STATEMENT UPDATE

The APF office has received many calls and questions regarding the vaccine statement on our website formed by the APF Scientific Advisory Board. According to porphyria experts, please note: Any vaccine can have side effects, and some side effects might possibly make porphyria worse. But vaccines themselves don't exacerbate porphyria. Any drug, including those listed as safe (in acute porphyria), can cause reactions in some people that might then lead to porphyria exacerbations.

COVID-19 VACCINE STATEMENT

Generally, there is no evidence that any vaccines cause particular problems in people with porphyrias. As a general rule, we therefore recommend that patients with any type porphyria receive vaccinations, in accordance with established guidelines, and under the care and follow-up of their personal physicians.

It is the unanimous opinion of the expert physicians of the Porphyrias Consortium that it is important for all porphyria patients to receive the COVID-19 vaccinations, unless their personal physicians have indicated that, in their particular medical situations, not including porphyria, vaccination is not recommended.

COVID-19 FDA & CDC LEADERS DISCUSS THE PANDEMIC

Early summer brought a discussion among healthcare officials entitled “Continuing the Conversation: FDA and CDC Leaders Discuss the COVID-19 Pandemic, Vaccines, and Special Considerations for the Rare Disease Community.” The National Organization for Rare Disorders (NORD), along with The ALS Association, Cystic Fibrosis Foundation and Muscular Dystrophy Association hosted a special webinar featuring leaders from the US Food and Drug Administration (FDA) and the Centers for Disease Control and Prevention (CDC). Topics included: Effectiveness of the vaccines for immunocompromised individuals; Timeline for vaccine authorization for children under the age of 12; and Future needs for booster shots or vaccine passports, and more. Pre-collected questions from the rare community will addressed in the discussion. This and other important conversations about Covid-19 included the rare disease community. Alone we are rare....together we are strong!



STRONGER THAN PORPHYRIA

Who is stronger than porphyria? These warriors who gathered mid-summer for fun and friendship. Fast friends Cheyenne, Morgan, and Gia all live with EPP. Together with their fierce moms, they had a blast at an indoor waterpark resort. Take that, sunshine!

Pictured (l-r): Carolyn Ludwig, Cheyenne Ludwig (11), Colleen McKillop, Morgan McKillop (11), Mary Ventrice, Gia Ventrice (9).

STUDY ON EPP (ERYTHROPOIETIC PROTOPORPHYRIA) PREVALENCE

A recent research study co-led by APF **Protect the Future** physician, Amy Dickey, M.D. and David Christiani, M.D. (both hailing from Massachusetts General Hospital) found a higher underdiagnosed rate in EPP patients than originally estimated by the medical community. The study, which was published in *Genetics in Medicine* and conducted on patients in the United Kingdom, found that the prevalence of EPP in patients was up to three times higher than previously thought. It is Dr. Dickey's hope that this study, along with a planned EPP Clinic at Mass General, will help spread awareness and broaden diagnostic and treatment options for future EPP patients.

The Protect the Future campaign was established to attract and train the next generation of doctors and specialists in the field of Porphyria. Over the next decade, we will lose ninety percent of our valued Porphyria experts. These men and women have led Porphyria research, testing and treatment for the past 30 years. Without financial support, we run the risk of losing knowledge of the disease, quality testing, diagnosis, and treatment, and ultimately a cure.



RESEARCH IS THE KEY TO YOUR CURE!

Visit porphyriafoundation.org for more information

POTTY TALK

PURPLE URINE

The terms porphyrin and porphyria are derived from the Greek word porphyrus, meaning purple. Urine from porphyria patients may be dark or reddish in color **due to the presence of excess porphyrins and related substances** and may darken further after exposure to light.

A Spot of Potty Humor



What do you call a dog that you find in your bathroom? A poodle!

Why did Tiger come looking in the bathroom? To find Pooh!

Did you hear about that film called constipated? It never came out!



ACUTE HEPATIC PORPHYRIA AND CONSTIPATION

A recent social media post questioned, "How do y'all deal with constipation?" Though severe diffuse abdominal pain is the cardinal symptom of an acute porphyria attack, that occurs in about 90% of attacks, there are many other non-specific symptoms that also occur. One of them is constipation. As acute porphyria is a neurovisceral disease, constipation is a result of the function of

the autonomic nervous system. Gastrointestinal symptoms are common during an attack and can include nausea, vomiting, constipation or diarrhea, and abdominal swelling (distention). A painful blockage or obstruction (ileus) of part of the small intestines may also occur. Difficulty passing urine (urinary retention) can also occur. According to Dr. Herbert Bonkovsky, a Gastroenterologist and porphyria expert at Wake Forest Baptist in Winston-Salem, NC, "Constipation is a common symptom, especially among women and persons who consume western-type diets, which tend to be lower in fiber. Severe constipation is often present during acute porphyric attacks—about 90% of cases—although occasionally patients will describe diarrhea instead. Constipation and other GI symptoms often are exacerbated by narcotic analgesics, which are best avoided, especially for chronic and ongoing use. If constipation is chronic, patients should best be treated with high fiber intakes; generous daily water intakes [about 3 liters/day]. Natural cathartics include prunes, prune juice, kiwi fruit, among others. Milk of magnesia and MiraLAX are safe and effective, also." Please visit the Diet and Nutrition section on www.porphyriafoundation.org for further guidance.



SOUTH AFRICAN PORPHYRIA FOUNDATION

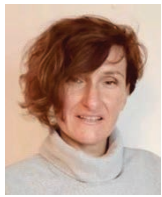
The South African Porphyria Foundation is a newly established volunteer organization created to raise awareness and provide support for patients and their family. The group will focus mainly on Variegate Porphyria, the South African mutation, this being the predominant type of Porphyria, however they will offer support to patients who have many of the other types also found in South Africa. At present, the foundation is funded by the three founding directors of the organization. They are in the process to establish an advisory team and establishing their infrastructure. The APF is proud to support this group as they dedicate themselves to porphyria patients in South Africa.



AMERICAN PORPHYRIA FOUNDATION
Virtual Patient Education Meetings

DIAGNOSIS AND TESTING OF THE PORPHYRIAS

Dr. Karl Anderson, porphyria expert, held a virtual education meeting in early September to discuss diagnosis and testing. The meeting was very popular and Dr. Anderson was generous with his time to respond to patient questions. Stay tuned for future meetings and share meeting requests that you have with us.



PROFESSOR BEATA KIE-WILK MD, PHD

The APF recently hosted a porphyria ZOOM presentation for patients in Poland. We were honored that Professor Beata Kie-Wilk, MD, PhD was our skilled presenter. Professor Kie-Wilk MD, PhD is head of the Laboratory of Rare Metabolic Diseases JUMC Department of Metabolic Diseases at Jagiellonian University Medical College in Kraków, Poland. She began over ten years ago to care for patients with rare metabolic diseases, like lysosomal storage diseases, disorders of amino acid and long-chain fatty acid metabolism. One group of patients had acute intermittent porphyria. In addition to outpatient care, Professor Kie-Wilk offers patients hospitalization in the department in the event of metabolic decompensation, access to modern diagnostics and care by a multidisciplinary team dedicated to patients with rare metabolic diseases. She tries to deepen her medical knowledge and, in cooperation with her patients, looks for new solutions, diagnostic and therapeutic methods. Aside from her esteemed career in science and medicine, she enjoys genetic and epigenetic mechanisms of ultra-rare diseases, history of art, classical music, and sports.




We are honored that Dr. Monroy recently presented an APF ZOOM educational call for Spanish speaking porphyria patients, their families and friends. Dr. Susana Monroy was born and raised in Mexico City. She obtained her Medical Genetics degree from the Instituto Nacional de

Pediatría (National Institute of Pediatrics) in Mexico City, where she also works as a medical researcher. Dr. Monroy has a private practice at the ABC Medical Center and launched the “Porfiria México” website in 2004 to provide Spanish-speaking physicians, porphyria patients, and their families with reliable information and medical advice. One of her main areas of interest is the porphyrias. She has been active in the field since 2000. She was the principal investigator for the Envision and Explore trials. The Instituto Nacional de Pediatría was the only site in Latin America that conducted those studies.

Porphyrias Fund

HealthWell offers up to \$4,000 in financial assistance to help with the cost of insurance premiums or medication copayments associated with the treatment and management of the various forms of Porphyria.

To learn more about the fund, determine eligibility, and apply for assistance, visit www.HealthWellFoundation.org, or call (800) 675-8416 to speak with a HealthWell representative.



HEALTHWELL FOUNDATION®
When health insurance is not enough.™

HEALTHWELL FOUNDATION

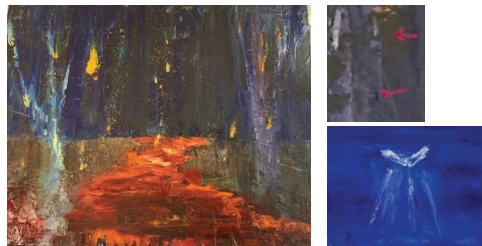
The HealthWell Foundation is a nationally recognized, independent non-profit organization founded in 2003, that has served as a safety net across over 80 disease areas for more than 615,000 underinsured patients by providing access to life-changing medical treatments. Since its inception, HealthWell has provided over \$2.1 billion in grant support to access life-changing medical treatments patients otherwise would not be able to afford. With copays, premiums, deductibles

and out-of-pocket expenses for supplies, supplements, surgeries and more, they offer financial assistance through a number of Disease Funds, including Porphyria. Through its Porphyrias Fund, HealthWell provides up to \$4,000 in copayment or insurance premium assistance for a 12-month grant period to eligible patients living with porphyrias who have annual household incomes up to 400 percent of the federal poverty level. Since launching the fund in 2006, the Foundation has awarded more than \$255,000 in financial assistance to over 80 patients. Applying for assistance takes less than five minutes. To learn more about the fund, eligibility, and application, please visit HealthWell’s Porphyrias Fund page.

HOPE WITHIN PAIN: Living with Porphyria by Andrew Gunter, MD

Dr. Gunter, a friend of the APF, accepted a challenge to paint what he felt after he listened to a video about living with the pain of porphyria. Listening to the words of patients inspired him to represent them on canvas. An underpainting of a descending dove representing hope for all suffering to end. A middle layer of black gesso and painters tape represents the struggles of medical therapy. And last, the outer layer of

oil paints denote facial expressions of pain randomly found within the trees.



THE STAT ACT

is a bipartisan bill that was created with the input of the rare

disease community aimed at improving the development of and access to therapies for the rare disease community. The STAT Act will enact targeted, impactful, and attainable policy reforms at the Food and Drug Administration (FDA) to accelerate the development of therapies across the spectrum of rare diseases and disorders and facilitate patient access to such therapies. Contact your state representatives in Congress to ask for their support in co-sponsoring this important bill! Click here for more information.



THE ASSISTANCE FUND (TAF)

helps patients and families facing high medical out-of-pocket costs by providing financial assistance for copayments, coinsurance, deductibles, and other health-related expenses. Since its founding in 2009, TAF has helped nearly 135,000 children and adults in all 50 states, Washington, DC, and Puerto Rico.

Among TAF’s more than 70 disease programs is the Porphyria Financial Assistance Program. The Porphyria Financial Assistance Program provides financial assistance for out-of-pocket costs associated with prescription drug assistance (copays, deductibles, and coinsurance) on FDA-approved treatment, health insurance premiums, therapy administration costs, disease management (such as prescribing physician copayments), treatment-related travel costs, and genetic testing. To be eligible for assistance, patients must be U.S. citizens or permanent residents, meet certain income requirements, have a diagnosis of the disease named in the disease program, have government or private health insurance, and a prescription for an FDA-approved treatment for the disease. Once a patient is enrolled in a disease program, their coverage lasts the entire calendar year and there is no cap on the amount of assistance in that calendar year. To learn more, visit <https://enroll.tafcares.org/>, or call (855) 927-1624.



MIKE BOONE

CONGRATULATIONS to APF member Mike Boone! Mike recently graduated with an Associates of Applied Science from Concorde College in Aurora, CO. Mike is a true AIP warrior and is now helping others in their battles. In his own words...

In 2008 I was diagnosed with AIP. Before the doctor told me he asked if I had life insurance because I wouldn't be able to get any after he told me. He was right. A few months later I found myself in a funeral home, picking out my casket wondering what the future for my children would be, I was 32. I decided to fight. I continued to work wearing an ambulatory IV pump to work twice a week for

my D10 infusions. I was working construction and people were surprised. As time went by I kept getting worse. It was to the point that I was on pain meds 24/7. Then in August of 2014 I was hospitalized twice and had to quit working. I became nearly bedridden for some time. Panhematin didn't work anymore. Then I got lucky and got into the Givlaari trial. Well, a little lucky, I was the placebo patient for phase 1. It was hell but I kept going. Then I got the real deal and in roughly three months time I was off of my pain medications. I only had occasional breakthrough pain. I kept going and finished the study in May of 2020. I continue to get the drug and have not needed any pain medication at all in well over a year, if not longer than that. Last week I graduate with an AAS degree in respiratory therapy. Porphyria took my life but, I fought back. With Givlaari I was able to start taking my life back. Now, I'm going to help others with their battles. Now, I'm going to help battle COVID.



ANGEL FLIGHTS

The mission of Angel Flight Soars is to help families get to their life-changing medical

treatments, surgeries, and clinical programs as often and as long as necessary, to ensure that no one has to go without medical treatment simply because of travel challenges. Angel Flight Soars coordinates free air transportation with volunteer pilots on private aircraft for medically stable and ambulatory patients. This community service is available to those residing in, traveling to, or traveling through Alabama, Georgia, Mississippi, North Carolina, South Carolina, and Tennessee. If needs extend beyond this service area, Angel Flight Soars partners with sister organizations via the Air Care Alliance, to meet the needs of the community they serve."

Below is a link to their "Angel Flight Real Stories" video: https://www.youtube.com/watch?v=V_yMhBYVMDI&t=3s



The **Rare Disease Clinical Research Network (RDCRN)** is the is an NIH-funded research network of 20 active consortia or research groups—teams of researchers, patients and clinicians—each focused on a group of rare disorders. The network fosters collaborative research among scientists to better understand how particular rare diseases progress and to develop improved approaches for diagnosis and treatment. The **Porphyrias Consortium (PC)** is in one of the most experienced consortia in the network, and the APF is actively involved as a patient advocacy group.

The **RDCRN Coalition of Patient Advocacy Groups (CPAG)** promotes collaboration between patient advocacy groups and the RDCRN to facilitate better access to, and earlier benefit from, research on

rare diseases. As the patient advocacy arm of the RDCRN, the RDCRN-CPAG and its members use their position to advance the cause of rare diseases research and improve patient outcomes. The RDCRN-CPAG Steering Committee meets throughout the year to help facilitate the development of activities that will benefit the entire CPAG membership. Those activities include (but are not limited to) webinars, in-person meetings, and one-on-one connections with other CPAG members and RDCRN researchers.

The steering committee of the **RARE DISEASE CLINICAL RESEARCH NETWORK'S (RDCRN) Coalition of Patient Advocacy Groups (CPAG)** has a new leadership team. APF's Executive Director, Kristen Wheeden, will move into the role of Chair and Kristin Anthony, President of the PTEN Foundation (DSC) will be Co-Chair. Congratulations go out to Kristen & Kristin.

In fact, there are multiple committees

PUBLICATION ALERT

Biochemical Diagnosis of Acute Hepatic Porphyria: Updated Expert Recommendations for Primary Care Physicians

ochemical Diagnosis of Acute Hepatic Porphyria: Updated Expert Recommendations for Primary Care Physicians

This publication in the American Journal of the Medical Sciences, led by Dr. Karl Anderson (UTMB) is the outcome of a panel of laboratory scientists and clinical acute hepatic porphyria (AHP) specialists who collaborated to produce recommendations on how to enhance diagnosis of the acute porphyrias in the USA. Though the publication is aimed to reach primary care physicians, it is critical information for all physicians. Doctors who consider porphyria as a potential diagnosis and order the appropriate tests can save patients years in their diagnostic journey. The full text can be accessed here: <https://pubmed.ncbi.nlm.nih.gov/33865828/>

that are engaged to improve rare disease research and advocacy. Members of the Porphyrias Consortium are represented on these committees to promote improved research for the those living with porphyria. The committees are:

- » Career Enhancement Committee – promotes opportunities for early-career researchers.
- » Cross Collaboration Committee – fosters collegiality, exchange and alignment among consortia.
- » Data Standards Committee – defines data standards to improve quality, usability and interoperability.
- » Data Use and Data Sharing Committee – provides guidance on data sharing for the network
- » Diversity – fosters diversity, equity, and inclusion efforts tailored to the unique needs of rare disease.

Rare Disease Week on Capitol Hill is an annual event that connects the rare disease community with their legislators and fellow advocates. This year, it was held in mid-July as a virtual opportunity to educate and spread awareness, while advocating for porphyria. **THANK YOU** to the wonderful advocates who participated and attended educational sessions and met with their members of congress. We'll be 'rare'-ing to get back in-person next year!



NORD: MEDICALERT ASSISTANCE PROGRAM

There are many options for medic alert bracelets or wearables – and the APF will be glad to guide you on wording and access. The National Association for Rare Disorders developed a program to support you. NORD's MedicAlert Assistance Program provides MedicAlert products and services to eligible individuals in the rare disease community. If someone with a rare disease can't speak for themselves in an emergency, MedicAlert can be their voice in providing important and potentially life-saving information. The program provides eligible individuals with a MedicAlert product and 3-years of membership. Contact the APF or NORD for more information.



WELCOME TO WORLD, FRANKLIN ROSS! MEET THE ROSS FAMILY

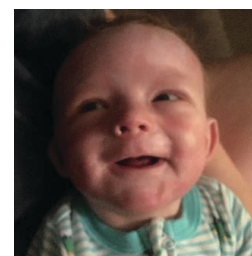
Like all parents, Robert and Kristy Ross of Nashville, TN were overjoyed by the birth of their son, Franklin Thomas Ross, on June 25, 2021. Shortly after his birth, the doctors diagnosed jaundice, which is commonly treated by the use of special lighting. However, in Franklin's case, the lighting didn't help. In fact, it triggered blistering that covered 50% of little Franklin's body, and the doctors noticed red urine in his diaper. They quickly had him transferred to Vanderbilt University Medical Center, where he was diagnosed with Congenital Erythropoietic Porphyria (CEP).

CEP is an ultra-rare, inherited metabolic disorder resulting from the deficient function of the enzyme uroporphyrinogen III cosynthase (UROS), the fourth enzyme in the heme biosynthetic pathway. Due to the impaired function of this enzyme, excessive amounts of porphyrins accumulate, particularly in the bone marrow, plasma, red blood cells, urine, teeth, and bones. The major symptom of this disorder is hypersensitivity to sunlight and some types of artificial light, such as fluorescent bulbs.

As Robert and Kristy absorbed the diagnosis, they turned to physicians and the APF. Armed with information, they knew their first job as parents was to protect their newborn from sunlight and artificial light. The APF provided the Ross family with information, warning cards, lightbulbs, and connection to porphyria experts, and fellow CEP families. Franklin was discharged and went home on August 6th.

Word of the diagnosis quickly spread in their community and Robert and Kristy were stunned by the support they received. In particular, Solar Solutions in Franklin, TN delivered a huge surprise: they immediately tinted the family car, but also another car and their home—all free of charge! The owner's heartfelt message was a gift in itself. "I know this has to be a really scary time for y'all and we want to help you and your family start this journey and help protect Franklin...your son is going to be a great young man and will change the world one day!"

Robert describes Franklin as "pretty darn great!" and adds, "He's a really sweet boy...opinionated when he doesn't want to eat and mimics his mom's oohs and ahs! His scars are steadily improving." He also shared that there have been so many that have helped in this journey so far. "The APF has been wonderful. They taught us how to protect him. Connecting us with Dr. Desnick made us feel more in control." Robert and Kristy found an unexpected source of support in the insurance company's case manager, and family has been ever-present. Franklin's grandmother, Sheri, has been invaluable, loving and caring for Franklin while Kristy teaches third grade full-time, and Robert works for FedEx and attends Middle Tennessee State University for Mechanical Engineering. We welcome the Ross family to the porphyria community...we will be here to love and support your family!



LONGITUDINAL STUDY:

This study includes ALL porphyrias.
There are nearly 1,000 patients

involved to date...are you one of them?

The natural history of the porphyrias is not well described and it is unknown

why some patients are more severe than others. This long-term, follow-up study is to collect a large group of patients with different types of porphyria to provide a better understanding of the natural history of these disorders to develop new forms of treatment. Goals include: to study the prevalence of specific indicators of disease

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

For information, contact Edrin Williams at edrinw@porphyriafoundation.org.

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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APF Staff Changes



WELCOME, MEGAN GEORGE! As our Advocacy and Program Director, Megan will manage APF office initiatives and additional projects and programs. Megan comes to the APF with previous non-profit rare disease experience at the Healthwell Foundation. She connected with Edrin Williams, Director of Patient Services, at an industry conference in 2019 when they hosted neighboring exhibit booths. Megan is already hard at work advocated for the porphyrias. We are blessed to have her join our team. She can be reached on Megan@porphyriafoundation.org.



GILLIAN WAGNER – OUR WONDERFUL INTERN! Gillian Wagner, a rising sophomore at Loyola University of Chicago and a resident of Arlington, VA, joined the APF as our Summer Intern. Gillian was a welcome addition at the office as she worked on mailings, managed database entries, and did general office work. She also helped to organize the upcoming Porphyrias Symposium in Schaumburg, IL.

We were blessed to have her wit, work ethic, and dedication to the porphyrias this year. We wish you the best as you pursue your degree and hope to see you in 2022, Gillian!

THANK YOU, IANY! After two years as our intrepid Office Manager, Iany Schneider has moved on to another opportunity in Montgomery County, Maryland. Iany was a treasure to the APF. Not only did she organize us all, but she showered kindness and respect on all who contacted the APF. As many of you know, she became fast friends with staff, patients, physicians, and vendors alike. We wish her the best in her new role. We are confident that she will remain out there spreading awareness about porphyria!



**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.

📍 4915 St. Elmo Ave, Suite 200
Bethesda, MD 20814
☎ 866.APF.3635
✉ www.porphyriafoundation.org

**HEME BIOSYNTHESIS AND THE PORPHYRIAS 2021:
CONSENSUS DIAGNOSES, VARIANT DISORDERS, NEW &
EMERGING THERAPIES**
OCTOBER 15-17 | Visit www.porphyriasymposium.com

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