We proudly announce the recipients of the 2021 American Porphyria Foundation President’s Awards. They honor members and partners who have gone above and beyond to support the foundation or members, and who have served to increase awareness of porphyria.

Jennifer Beck (EPP)
Jennifer has been a visible force for the APF and the porphyria community. From participating on the APF Membership Advisory Board and representing APF as keynote speaker at the NORD Summit opening session to her continual presence on social media, she has championed those with porphyria.

Janie and Jerry Williams
Janie and Jerry have been staunch supporters of the APF’s mission for many years. Their support has contributed to the implementation of valuable programs and porphyria awareness. Janie shared that “the foundation has truly been a blessing for both of them!”

Manisha Balwani, M.D.
Dr. Balwani serves on the APF Scientific Advisory Board and is a Professor in the Department of Genetics and Genomic Sciences at the Icahn School of Medicine at Mount Sinai, New York City. In addition, she is an investigator for the NIH-supported Porphyrias Consortium. Dr. Balwani is a tireless supporter of the porphyria community. Her expertise as both a clinician and a researcher have served to advance porphyria patient care and treatment. The APF is very appreciative that Dr. Balwani is beyond generous with her time and talent! Dr. Balwani said, “it’s been the most wonderful partnership with the APF and truly rewarding working for the patient community. I look forward to many more years of collaboration.”

The Shadow Jumpers program was created to help give kids with Erythropoietic Protoporphyria (EPP) and their families a place to learn about this rare disease, read tips and tricks learned over time and to hear from fellow kids. Through spreading awareness, fellow EPP interviews, tips for protecting yourself outside and some insight for parents, we hope all families living with EPP will look at this condition as a challenge they can overcome.

A main goal of Shadow Jumpers is to help kids do the things they have always wanted but were prevented by the sun. Pain combined with the emotional discomfort from having to manage this disease may lead kids to avoid certain activities, relationships, and paths in life. Shadow Jumpers wants to remind kids that they can still follow their dreams!

2021 Find Your Shadow Recipients
We’re happy to announce our family recipients for 2021’s FIND YOUR SHADOW project! This year, we are going to support more families in addition to fulfilling dreams from 2020 delayed due to Covid-19. Shadow Jumpers and the APF can’t wait to share dream details and some amazing stories with you as we finish off the year taking down the sun for these FIND YOUR SHADOW recipients!

The Campbell Family
Deagon (6), Mason (12), Braxton (8), Andrea, Brad

The Tonhaeuser Family
Adeline (4), Cole (19), Sophia (12), Kurt Jr (8), Megan, Kurt

The Zamora Family
Anthony (14), Adrian (17), Keyra (4), Ayan (2), Zhianna (2), Adrian Sr, Delena

The Ludwig Family
Cheyenne (10), Riley (8), Ariana (5), Carolyn, Joshua

The Chadwick Family
Madison (9), Isaac (8), Natassja, David

Left to right; Campbell Family, Cheyenne Ludwig, Madison & Isaac Chadwick, Adeline Tonhaeuser, Zamora Family, Tonhaeuser Family, Adeline, Deagon Campbell, Zamora Family, Ludwig Family, Isaac Campbell, Chadwick Family

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

It’s just around the corner!
HEME BIOSYNTHESIS AND THE PORPHYRIAS PLANNED FOR OCTOBER 2021

Scientific Meeting

This 2.5-day educational symposium will feature international and national experts presenting 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.

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

Register at [www.porphyrissymposium.com](http://www.porphyrissymposium.com)

You are invited

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

OCTOBER 15-17
Hyatt Regency Schaumburg near the Chicago O’Hare Airport

Visit [www.porphyrissymposium.com](http://www.porphyrissymposium.com) for agenda, hotel information

Is your physician interested in learning more about PORPHRYIA?
Share this invitation to register.

Special Patient Day Meeting

Join us for a special Patient Day Meeting during the Heme Biosynthesis Symposium!

With informational sessions and networking opportunities, this in-person and virtual event is a must-attend for all patients globally. This is an opportunity to learn up-to-date information, get all your questions answered, and to make new acquaintances and friends.

Highlights of the Day Include:
» Latest diagnostic & treatments options
» Educational sessions on each of the eight porphyrias
» The importance of research
» Pain management including the use of cannabis
» Psychological issues and quality of life discussions
» “Ask the Porphyrias Experts” open discussion panel

Don’t miss out on this unique opportunity to participate in-person or online – registration is now open!

Register for the Patient Day Event Now at [www.porphyrissymposium.com](http://www.porphyrissymposium.com).

APF’S 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 of 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 Porphryias 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.

MORGAN’S STORY

The mission of Sing Me a Story is to serve deserving families in need through the imaginative stories of their children and the power of music. When a child submits a story through a patient organization, it is given to a songwriter who creates a song out of the child’s words. Morgan McKillop (EPP, Age 11) submitted a story to the APF that she had written while at home from school recovering from a reaction. “Morgan’s Song” was written and recorded by Samara Michael. You can listen here: [http://singmeastory.org/stories/morgan-s-story?song=1427](http://singmeastory.org/stories/morgan-s-story?song=1427)

2021 EPNET WEBINAR SERIES

The European Porphyrias Network has hosted a series of webinars over the last several months featuring porphyria experts, aimed at educating medical professionals and specialists in laboratory medicine. The series addressed the acute porphyrias, the erythropoietic protoporphyrrias, and porphyria cutanea tarda. The final session is on laboratory diagnosis. Our European colleagues have implemented an excellent series available at [https://porphyria.eu/en/content/2021-webinar-series-porphyrias](https://porphyria.eu/en/content/2021-webinar-series-porphyrias).
We are pleased to share that enrollment in the Mitsubishi Tanabe MT-7117 Phase 3 clinical trial to study the investigational drug Dersimelagon in Erythropoietic Protoporphyria (EPP) and X-Linked Porphyria (XLP) is now complete! The study enrolled participants from around the world. A Phase 3 trial is the final clinical trial stage before a potential new drug application is submitted to the FDA.

Thank you to all the research heroes who are participating in this clinical trial! Your community is grateful!

A voluntary open-label extension (OLE) study will be available for participants that complete the ongoing Phase 3 study. An OLE study is a clinical trial that enrolls participants of a previous clinical trial and is designed to gather the long-term safety and tolerability data on an investigational drug. Current Phase 3 trial participants interested in possibly continuing their treatment with dersimelagon (MT-7117) are encouraged to refer to their study site for further information.

If you are currently receiving prophylactic heme treatment and are interested in participating in the ongoing Panhematin® Prevention Study, please contact Edrin Williams, Director of Patient Services at edrinw@porphyriafoundation.org or 301.347.7166.

This research study is important for physicians to better understand the benefits of prophylactic heme treatment. Travel to the study site is included.

Inclusion criteria for the Panhematin® Prevention Study:

- Have well-diagnosed and documented AIP/HCP or VP with lab results;
- Had previous attacks of porphyria with symptoms such as abdominal pain, back or limb pain;
- Currently on a regimen of Panhematin® infusion to prevent acute attacks.

This year’s Recordati Rare Disease/APF Click Campaign was a huge success. For every CLICK, the APF received a $5 donation, up to $5,000 from Recordati Rare Diseases. We are grateful to Recordati for helping us continue our mission to spread porphyria awareness and educate our communities about this rare disease! Thank you to all members, families, physicians, and others who took the time to “click”! Most importantly, thank you for continuing to spread the word about porphyria to your community!
RARE DISEASE WEEK 2021

Rare Disease Week on Capitol Hill is an annual event that connects the rare disease community with legislators and fellow advocates. It is an opportunity to educate and spread awareness while advocating for porphyria. While we had hoped to participate in-person, Rare Disease Week will be going virtual for 2021.

Virtual Rare Disease Week on Capitol Hill 2021 will be held July 14-22 and will include the same opportunities as in-person Rare Disease Week, plus more! Go to www.everylifefoundation.org for more information. The APF is in the process of developing a policy one-sheet on legislative issues important to the porphyrias. We hope you are interested in joining this important event and will be glad to discuss policy issues one-on-one. Call the APF anytime.

RARE DISEASE DAY AT NIH

Each year, NCATS (National Center for Advanced Translation- al Sciences) and the NIH Clinical Center sponsor Rare Disease Day at NIH to raise awareness about rare diseases, those affected, and NIH research initiatives. Rare Disease Day takes place worldwide, on or near the last day of February. This year’s Rare Disease Day took place virtually. The event still allowed participants to raise awareness among policy makers and the general public by sharing the impact of living with a rare disease. Kristen Wheeden, APF Executive Director, moderated an afternoon session regarding the effects COVID-19 has had on rare disease research. It was a well-attended session that discussed the ways in which physicians, researchers, and patient advocacy groups have been required to pivot in the face of the pandemic to continue research efforts. Over 1,000 participants engaged in Rare Disease Day 2021!

PORPHYRIAS CONSORTIUM
Pilot & Feasibility Program

The Porphyria Consortium is providing a novel support mechanism for U.S. trainees, postdoctorates, and junior faculty interested in pursuing advancements in porphyria.

Support: $30,000 of funding in a single sub-contract to the institution of the applicant.

Please note: Proposed research that falls outside the mission of the Porphyria Consortium will be considered nonresponsive and will not be reviewed. The use of animal models is not permitted.

Program Requirements and Instructions: www.rarediseasesnetwork.org/cms/porphyrias.

PAIN PREVALENCE DATA

A recent study published in the latest issue of the journal Demography highlights the prevalence of chronic pain in the daily lives of individuals. The study suggests “blanket increases across multiple measures, with pain rising in every adult age group, in every demographic group, and at every site of pain for which data exists.” The study has shown that societal pain has incrementally increased from one generation to the next. The publication can be found at: www.news-medical.net/news/20210512/Chronic-pain-is-becoming-more-common-in-the-United-States.aspx

More Interesting Reads on Pain:
- www.painnewsnetwork.org/stories/2021/5/12/americans-reporting-more-pain-than-ever

ABSTRACT HIGHLIGHTING PAIN REDUCTION

An abstract presented at the American Academy of Neurology’s April virtual annual meeting titled “Reduction in Pain During and Between Attacks in Patients with Acute Hepatic Porphyria Treated with Givosiran: A Post-Hoc Analysis of the Phase 3 ENVISION Study” highlighted trial results from the Phase 3 Trial results of patients with acute porphyria taking GIVLAARI®. The results found that after six months patients found reduced pain during and in between acute attacks.

GIVLAARI® (givosiran) is a prescription medicine used to treat acute hepatic porphyria (AHP) in adults. The abstract can be read at: https://index.mirasmart.com/AAN2021/PDFFiles/AAN2021-001463.html.
THE APF ACUTE PORPHYRIA ATTACK TRACKER

Keeping track of your acute porphyria attacks and related symptoms will give you a sense of control and allow your physician to help you manage your attacks over time. Your physician can recognize the timing of your attacks, changes to your body, need for treatment, etc. The APF has developed an interactive tracker to help you keep a file of your acute porphyria attacks, symptoms, treatments, and hospitalizations. The tracker is customizable so you can design it to suit your own needs! Go to: https://porphyriafoundation.org/for-patients/support-assistance/acute-porphyria-attack-tracker to access the tracker.

HEPATOCELLULAR CARCINOMA (HCC) AND ACUTE HEPATIC PORPHYRIA

The expert physicians of the Porphyrias Consortium recommend that all patients with Acute Hepatic Porphyria (AHP), regardless of the severity of symptoms, be screened for liver cancer, sometimes called Hepatocellular Carcinoma or HCC. A study titled Hepatocellular Carcinoma in Acute Hepatic Porphyrias: Results from the Longitudinal Study of the U.S. Porphyrias Consortium (https://aasldpubs.onlinelibrary.wiley.com/doi/epdf/10.1002/hep.31460) appeared in Hepatology, a publication of the American Association for Study of Liver Diseases (AASLD) and was recently published online. According to this study of over 300 patients, “1.5% of patients with AHP had HCC.” Please note – this study was made possible by patient participation in research! Remember … Research is the Key to Your Cure!
LAURA GALLAGHER (EPP)
My Day in the Sun after Scénessé®

I’m not sure if I have words to describe it because I have no idea what it’s going to feel like: To be in the sun without fear, without feeling guarded. To walk a street and instinctively not go to the shady side. To step outside and instantly know if it’s a good or bad day and pay no attention because I’m suddenly now capable. It’ll be a whole new world. A world I’ve never dared to imagine. I have lived a sheltered life, literally.

The freedom of just simply being in sunlight is so foreign to me. I wonder how that’ll feel.

My vision is brilliant! Carefree, happy. Will it be like that? Could it? Will I actually run down the beach with my daughters as we imagine? They are as excited as I am with this possibility. Will I once again disappoint them? Everyone? Will it be enough? Will I be too scared to try? Will I do too much and screw everything up? Is this really a fix to what I’ve been dealing with my entire life? God, I hope so.

I hope I feel sun on my feet, face, hands, and arms and I’m anything but petrified or anxious. I hope that I can stand on the ocean’s edge and watch my feet sink in the sand without concern watching the sun shine down and not be scared that it will hurt me. I pray that I splash with nothing but smiles and laughter as the tide rolls in and maybe even takes me off balance. I hope so much and dare to imagine more. Too much actually. Too much to even consider. I’ve thought about this for so long. I pictured a place where I was that person in only dreams. Is that dream actually going to happen? Carefree and happy in the sun? Seriously?

I have lived this way for so long. Guarded, sheltered, hidden. Will I be able to embrace the realization that I’m suddenly now okay? Will I succumb to everything my mind has trained me to avoid my entire life?

I’m super scared, super excited. Oh my gosh! Is this really happening? Going to be possible? Do I dare to picture me, ME, anywhere other than in a cabana, under a tree, indoors watching from a window? Am I actually going to be able to be with everyone else?

So many emotions. So many concerns. I hope it’s all I imagine. I hope I’m capable of embracing it. Knowing no other way for so long has had an effect on everything I am. I’m not sure if I will be able to accept that the sun is now suddenly okay and no longer something that’s going to cause me days of pain. An enemy to avoid and now, for the first time, no longer. I’m scared. I’m excited. Totally overwhelmed. But mostly, I’m looking forward to just “being.” It’s finally here, all I’ve imagined my entire life. My time is literally just weeks away.

LAURA GALLAGHER RECEIVED HER FIRST SCÉNESSÉ® IMPLANT THIS MONTH!

EPP MAMA WARRIOR
Inbal ben Shalom – A Force of Nature and an amazing APF Connection

Inbal, a native Israeli, had never heard of porphyria nor had any family members exhibit symptoms. When her daughter, Maya, began exhibiting a burning sensation on her skin, Inbal began a diligent, six-year search with countless doctors to find the source of Maya’s horrendous pain, itching, swelling and sleepless nights. Unfortunately, neither Inbal nor the doctors connected the sun with Maya’s terrible illness. Despite the outward signs, some doctors even intimated Maya’s problem was primarily psychological – the plight of many porphyria patients. They ultimately decided Maya had allergies and began treatment, but Maya continued to suffer.

Inbal was desperate to help her child, yet the symptoms continued and the wait to see the top geneticist proved impossible, so they were put with a young intern. This was the fortuitous event of their lives. The intern had learned of an episode of “House” that paralleled the child’s symptoms and suggested testing Maya. When the results for EPP were returned it was a joyful time for Inbal and Maya – as they had an answer at last. Even more miraculous, the geneticist admitted later that she never would have tested for porphyria had she seen the child.

In her valiant search for treatment, Inbal found that there were many benefits in Israel for children with rare conditions, though they were not known to the public. Inbal has since heightened awareness of these subsidies for other EPP children and those with rare diseases. Israel provides regular medical benefits, gives Maya a caretaker at school and a therapist to help her cope with her EPP. She also was able to acquire therapy for Maya, as well as discounts for electricity, and a host of other opportunities to make life easier for Maya. Inbal subsequently met with Israel’s Health Minister and was able to have porphyria deemed a disability “category” to assure access to certain benefits.

Inbal began receiving invitations to speak about EPP at medical conferences and creating videos on the disorder. She is also a primary contact for diagnosed patients with EPP. Ironically, years after the “House” episode, when Desiree Lyon, APF Global Director, and Inbal connected, Desiree shared her responsibility for the story line about porphyria. Both were teary to think that they were connected years before they ever met.

Inbal and Desiree will be collaborating on more ways to advance porphyria awareness in Israel and make life there less challenging for patients. We sincerely appreciate Inbal and welcome her into the APF Global Program as a Global Ambassador.

PORPHYRIA NURSE ADVOCATE VIDEO ON YOUTUBE!

The Porphyria Nurse Advocate (PNA) team created an awareness week video intended to share information with other nursing professionals. The video and initiative titled “Tell 3 More” encouraged nurses to watch the video and share with three other colleagues. The video begins with Chanan Stauffer, NP at Mt. Sinai in NYC. Chanan, under the expertise of porphyria expert Dr. Manisha Balwani, shared detailed information about the porphyrias. Her introduction was followed by other PNA members sharing details and encouraging others to learn and go to www.porphyriafoundation.org for detailed information. Well done, PNA! Please go the the APF channel on YouTube to see this video and many others!
POPHYRIA CUTANEA TARDA — AWARENESS WEEK STORY!

Chrisy Dirnbeck, R.N.

In preparation for Porphyria Awareness Week, I added the PAW frame to my profile picture on Facebook. Within minutes, my aunt that I was very close to during my childhood commented on my post. This aunt on my mother's side was aware that her mother and one of her sisters, not my mother, had the diagnosis of Porphyria Cutanea Tarda. She was not aware that I share the same diagnosis with them. I was diagnosed with Type II Familial Porphyria Cutanea Tarda in 1991, just after completing college and beginning my career as a High Risk Labor & Delivery nurse. My other aunt that shared the same diagnosis as me did not have any children. In my aunt's mind, the porphyria had stopped with them and had not been passed on to anyone else. No one else in the rest of our family was ever diagnosed or had shown symptoms of the disease until myself, and my aunt was not aware. This information became very important to her because her daughter was pregnant and expecting her first child in September. After we messaged back and forth just to catch up, I proceeded to share any and all of my medical documents with her. I even had my grandmother's original documents from her initial diagnosis in 1977. She was sorry to hear that I shared in the same diagnosis but grateful that she now knew and had all of our medical documents, including my genetic testing and thorough work-up from Mayo Clinic in 2019.

Social media has often been criticized but in this case, I am grateful that the connection was made. If you are willing to be open and share your diagnosis, it can have an amazing impact in someone’s life by giving them an opportunity for an early diagnosis! As we all know, an early diagnosis saves a person years of pain and frustration. Porphyria Awareness Week and Facebook for the win!

APF MEMBER FEATURED IN ART EXHIBIT

APF Shadow Jumper 2019 winner and EPP member Morgan McKillop has earned the very special honor of being one of the latest portrait representatives in the Beyond the Diagnosis art exhibit. Morgan's beautiful portrait, a powerful symbolism of life in the shadows, will join over 100 other rare disease portraits in the exhibit – to be seen by thousands globally!

The focus of Beyond the Diagnosis is on the orphan diseases patient community. With the help of artists donating their skills, the exhibit reaching the goal is to put “a face to all 7,000 orphan diseases.” The exhibit travels to medical schools, research institutes and hospitals globally, enticing the medical community to look “beyond the diagnosis to the patient.” The museum tour was amazing. The exhibit was at the NIH, FDA, and several other Rare Disease Day events and was seen by thousands of people from around the world. They are already booking the tour for venues for the rest of 2021. The tour is now live on the Artfully Rare website for everyone to watch. Jorgy did an amazing job with the tour and we are so happy. Visit out website at www.artfullyrare.org and scroll to the bottom for the tour.

INTRODUCING JESSICA HUNGATE ... MEDICAL DOCTOR!

Jessica, former APF staff member, remains a wonderful friend to the APF. We are very proud to share that she is now a medical school graduate! Pictured: Jessica and Desiree Lyon

The APF has been members in good standing of NORD – the National Organization for Rare Disorders since it was developed in the early 1980s.

For many years, people with rare diseases walked alone. Patients and their families coped with daunting medical and financial issues with few resources and no one to guide the way. Then, a small group of patient advocates formed a coalition to unify this community and mobilize support to pass the Orphan Drug Act. In 1983, the coalition became NORD, the National Organization for Rare Disorders. For more than 30 years, NORD has led the way in voicing the needs of the rare disease community, driving supportive policies, advancing medical research, and providing patient and family services for those who need them most.

IN MEMORY & IN HONOR

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

IN MEMORY:

Iany McDonald-Schneider for Carol’s Baby, Bailey; Gary Horn for Sandra Horn; Donald John for Peggy Lewis Johnson; Aunt Bernice, Mr. & Mrs. Thomas Barletta, Cheryl Manzo, Shelley McGinley for Alice Kuznicki; Kathleen Kelahan for Joan McLean; Cyndi Nottage for Natalie Nottage; Susan Nuckols for Camden Nuckols; Michael Bonenfant, Rob, Frances & Elizabeth Daniels, Mary Giudici, KaraRose Gotta, MaryAnne Richardson, Steven Tarolli, Christopher Varley for Kelli Quenzer; George Rusnak for Carol Rusnak; James Witter for Barbara Witter.

IN HONOR:

Paula Andrews for Hattie & Clint Stuhshatz; Anonymous for Amanda Boston’s niece – the artist!; Jesse Lewis Jr., Fran Gandaarillas for Morgan McKillop; Joan Friedman for Debbie Redfield; David & Candy Secrest for Grace Secrest Shield; Erin Strunk for Michael Strunk; Linda Snow Griffin, PhD. for Dr. Carl Tishler; Kevin Vogelpohl & Carmen Ortiz for Tyler Vogelpohl; Carole Aitchison, Andrea Bournias, Mollie Darby, Billy Finn, Sharon Wheeden for Brady Wheeden & the Wheeden Family; Paige Ernst for Braden Whiteside in her residency. We know there are big things in her future!

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To Your Cure

Your help is needed to educate physicians and patients and to support research, becoming a member of the American Porphyria Foundation or making a tax-deductible contribution today. Your support and donations enable the APF to maintain our physician and patient education programs and many other services because of your support. Also, your new contact information is needed to send our weekly Porphyria Post and to ensure you receive all updates and information from the APF. If you have a new address or email, please update your contact information.

What’s New at the APF

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