



AMERICAN PORPHYRIA FOUNDATION

1st Quarter, 2022

HAPPY BIRTHDAY, APF! CONGRATULATIONS!

We are 40 years old, and we are always up to date!

Forty years ago, James Young and Desiree Lyon founded the APF. Starting with two members and an office on a kitchen table, the APF has grown to several offices around the country and a database of **15,000 members**. We believe the APF has the largest database of patients around the world with the largest database of 5,000 treating physicians and the largest database of international patients. Our mottos, STRONGER TOGETHER and PATIENTS FIRST, have not changed, nor has our mission to enhance patient and physician education and awareness. Foremost is our goal to provide patient support. Still active at the APF, James Young continues as the APF Treasurer and Desiree Lyon as the Global Director. Both continue to be devoted to patient services.



WARREN HUDSON, CHAIRMAN OF APF

I would like to introduce myself—my name is Warren Hudson, and I have been

a member of the American Porphyria Foundation (APF) since 2002. For over a decade, I was an AIP caregiver. I also assisted the APF with legislative and lobbying activity, started the Caregiver's Support Group and volunteered many hours on various projects. In 2010, the APF's Chairman of the Board of Trust-

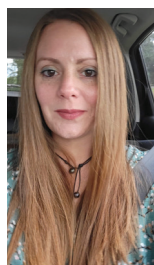
ees asked if I would join the Board. The APF was there when I needed help, and I wanted to do my part to ensure it would continue to be there to support others.

At that time, in an effort to be a better advocate, I did a lot of research to learn more about porphyria. I noticed that there was information on the biochemistry but not on strategies for living with the chronic disorder. Specifically, I saw very little related to pain in the Porphyrias—either from a neuroscience or a psychological/social perspective. My belief and experience is that

chronic and acute pain has the most impact on a patient's daily living and quality of life. Although pain and patient related services are still something I want to pursue, late last year, several former Board Members and foundation leadership asked for me to serve as the Chairman of the Board of Trustees. The APF experienced a lot of growth domestically and internationally so our Board and management wanted a greater focus on governance and the foundation's administrative processes.

Story continues on page 6

A SAD BUT AMAZING BIT OF HISTORY



Ashley Dice

Every day at the APF we hear stories of courage and hope. Today we are sharing another wondrous story.

Over 30 years ago, I asked my sister, Deborah Fedele, to visit a young man, Robert Dice, who was in a hospital suffering from a critical AIP attack. Sadly, unbeknownst to me, he was near death when Deborah arrived. She

comforted him and held his frail hand when he reached out to her. After spending time with him, she left but assured him that our APF members sent him love and prayers. Soon thereafter we learned that he had passed at only 24 years old.

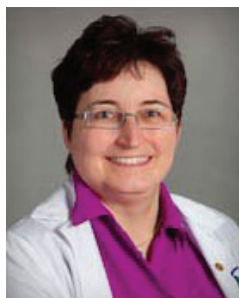
Fast forward thirty years. I held an APF Patient Meeting at my home in Florida. Among the attendees was a lovely young woman whose name sounded familiar. In talking with Ashley Dice, I discovered that her fa-

ther had died of porphyria. On a hunch, I asked where he had died, and it turned out to be the same hospital in Alabama and the same man from long ago. Ashley was a baby when her dad died, so it was a heart-breaking conversation but wonderful that we had found each other.

Now fast forward three more years. My family and I were enjoying dinner at a nearby beach restaurant when we began

Story continues on page 4

DR MARTINE EXTERMANN PRESENTS PORPHYRIA GRAND ROUNDS



Recently, Dr Extermann presented a successful Porphyria Grand Rounds at Moffett Cancer Center in Tampa, Florida to an audience of 101 physicians. Dr. Martine Extermann is a highly esteemed Professor of Oncology and Medicine at the University of South Florida and is Pro-gram Leader in the Senior Adult Oncology Program at Moffitt Cancer Center in Tampa Florida. She earned her MD and her medical PhD at the University of Geneva, Switzerland. Aside from her expertise in Oncology, Dr Extermann is also a knowledgeable porphyria expert, whose patients comment that she is compassionate, brilliant and worthy of the honor and respect she receives from her patients and colleagues. We are proud to have Dr. Extermann represent the American Porphyria Foundation. To watch Dr. Extermann's presentation, please see: <https://moffitt.hosted.panopto.com/Panopto/Pages/Viewer.aspx?id=27cbe1b-5ad4-49a1-94f2-ae2301308388>

zoom JOIN OUR APF EDUCATIONAL VIDEO CONFERENCING SERIES



Dr. Sanjay Hapani will be presenting a Zoom call for APF members and friends during Porphyria Awareness Week. Porphyria expert, Dr. Sanjay Hapani, MD is a Hematology Specialist in Oklahoma City at Mercy Hospital, Great Plains Regional Medical Center and Integris Baptist Medical Center. Interestingly, Dr. Hapani was treating porphyria patients when a particular patient received attention from a local news network, KOCO 5. After airing the patient's porphyria story, the show not only led to heightened porphyria awareness, but also led to other porphyria patients reaching out to Dr. Hapani for help. Dr. Hapani's popularity in the porphyria world has grown considerably, with patients commenting on his compassion and brilliance. Dr. Hapani admits that even to him, porphyria can be a mystery.



WATCH FACEBOOK, ENEWS AND THE APF WEBSITE FOR DETAILS OF THE ZOOM CALL WITH DR HAPANI.



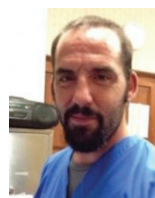
Dr. Silver Samuel, professor of internal medicine and hematology at the University of Michigan Medical School in Ann Arbor. will also present an APF patient ZOOM call in April. Before his retirement, Dr. Silver was highly awarded in the fields of malignant hematology, bleeding disorders, platelet dysfunction, myeloproliferative, and myelodysplastic disorders. Fortunately, Dr. Silver also was lauded as a porphyria expert who was widely sought for his expertise in porphyria by doctors and patients alike. Dr. Silver was a key researcher in the Givlaari trials and has served on the APF Scientific Advisory Board for many years.



ZOOM at 730 PM EST, March 31, 2022 Watch for details on the APF website.

PORPHYRIA CUTANEA TARDA (PCT)

Porphyria Cutanea Tarda (PCT) is the most common of the Porphyrias. PCT is rare and is characterized by painful, blistering skin lesions that develop on sun-exposed skin. Affected skin is fragile and may peel or blister after minor trauma. Liver abnormalities may also occur. PCT is essentially an acquired disease, but some individuals inherit an enzyme deficiency, which leads to the development of "familial PCT." These patients often remain latent and never have symptoms. Acquired factors, such as iron, alcohol, Hepatitis C Virus (HCV), HIV, estrogens (used, for example, in oral contraceptives and prostate cancer treatment) and possibly smoking, combine to cause a deficiency of UROD in the liver. Hemochromatosis, an iron overload disorder, also can pre-dispose individuals to PCT.



Patients John Gladding and Darlene Folkes have very interesting PCT stories. John relates his adventure beginning with acquiring Hepatitis C through a blood transfusion he was given when he was born. After his diagnosis, he was prescribed phlebotomies, the same treatment for two other diseases.



Darlene's PCT journey began at 44 years old. Wanting to get in shape, she began exercising outdoors. The ensuing blisters and prickly bumps on her hands refused to heal and in fact got worse, prompting her doctors to consider a sun allergy. During a visit to the ER, a biopsy indicated PCT. Darlene joined the APF and caught up with all the educational materials about PCT.

Read John and Darlene's stories in their entirety: <https://porphyriafoundation.org/for-patients/member-stories/porphyria-cutanea-tarda/>

WE FIGHT PORPHYRIA TOGETHER



Congratulations, Jeannie Lodge-Brando from London, Ontario, Canada, who won the 2022 Porphyria Awareness Week Theme Contest. The choice was difficult, because every suggestion was outstanding. In fact, they were all so superb that we hope to use them throughout the year.

Jeannie's description: We Fight Porphyria Together! Hands locked together inside a globe. WE FIGHT PORPHYRIA TOGETHER from all over the world, so we can support each other and educate medical personnel how intense, rare and the need to listen to us and read the material provided. Definitely purple!!!

A LOOK AT PORPHYRIA PAIN

Author, **Claire Richmond**, addresses PAIN, the number one complaint of porphyria patients.



There is a general lack of understanding and awareness when it comes to pain associated with acute porphyria attacks. Porphyria pain is not well defined in research. It's misunderstood by doctors and industry professionals, and it's easy to see why. Pain cannot be objectively measured or seen, and everyone responds differently during an attack. Someone with acute intermittent porphyria (AIP) may be writhing in bed, howling, and clutching a barf bag. Or, if they're like me, they might be curled up in a fetal position, crying silent tears, and trying to keep breathing. For far too long, the community has been focused on finding a cure rather than learning about the pain porphyria patients endure. It's time to get patients and members of the medical community on the same page with porphyria attack pain. It's time to rewrite the definition of porphyria pain, in hopes we can inspire new research.

Descriptions from research

Let's start with assessing what we know from medical literature. Porphyria pain is under researched, and there's a lack of evidence-based recommendations for management and treatment. Digging into published studies, I found only vague descriptions of porphyria attack pain. For example:

An excerpt from the journal *Patient*, referring to acute porphyria, describes "diffuse severe pain affecting the abdomen, back, or limbs."

An excerpt from *The Netherlands Journal of Medicine*, referring to cutaneous porphyria, describes "severe pain or blistering of the light-exposed areas of the skin."

While straightforward, those descriptions were limited by perspective. A doctor or researcher may be able to summarize aspects of physical pain during an attack, but only someone who's lived through attacks can detail the mental and emotional depth of the experience.

Descriptions from patients

Describing pain in a more useful and holistic way seemed like the easiest job for a patient; after all, we are the true experts in

attack pain. So, how do I describe porphyria pain? Here's my definition:

"Porphyria attack pain is like routinely being tortured and betrayed by my own body."

As I am only one person representing one porphyria subtype, I asked others who have porphyria for help in describing attack pain. Some agreed the pain was worse than childbirth, kidney stones, and broken bones. Some called it "torture." Following are selected quotes from some of these conversations:

Emerald, hereditary coproporphyria *Porphyria pain "feels like I'm getting crushed inside a red hot iron maiden that's also being hit by lightning. I often feel traumatized with attacks when I go to an ER and no one knows what porphyria is."*

Betsy, variegate porphyria *"One of the worst attacks I've ever had, the pain intensified until it was like someone had impaled me through my abdomen."*

Jessica, acute intermittent porphyria *"It felt like all my organs were in a vice grip and it was very hard to breathe."*

Robert, acute intermittent porphyria *"The pain begins as a toothache, like pain in my long bones of my arms, legs. It then progresses into severe abdominal pain that feels like I have a red hot poker stuck into my liver from the front and back [that] is being moved all about through my abdominal area."*

Maree, porphyria cutanea tarda *It's "like walking through a fire, and you can't get away."*

Cortney, erythropoietic protoporphyria *"I ran an entire half marathon with a fractured pelvic bone. Prior to the race, and even after, I swore I just had tendinitis because I don't know how to interpret pain levels correctly."*



The difference between descriptions in the literature and those written by people living with porphyria were striking. Accounts from real people made it clear that attack pain is an experience far beyond physical sensation, it's also emotional, traumatic, complicated, and evolving.

As people with porphyria, we serve a critical role in raising awareness. But I also think of the depth we could provide research, too, if we were recognized for our expertise and sought out in designing studies that investigate attack pain. Most of all, I dream of the day porphyria pain is better understood, when patients know how to advocate for the care we need, and physicians associate porphyria attacks with extreme pain.

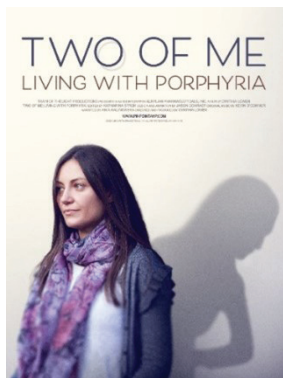
Claire Richmond hopes speaking her truth builds connections and generates hope. At 32, she was diagnosed with AIP after a 19-year search for answers. As severe, unexplained pain became a focal point of life, she exiled her body to protect her mind. Through laughter and challenges, Claire rebuilds the bridge between her physical and mental being.

THANK YOU, *Porphyria News*, for allowing us to reprint Claire's article.

EDITORS NOTE:

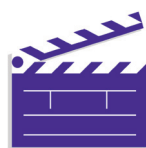
Read a comprehensive review of the current evidence **Mechanisms of Neuronal Damage in Acute Hepatic Porphyrias**, **Andrea Ricci, Elena Di Pierro, Matteo Marcacci, Paolo Ventura** <https://pubmed.ncbi.nlm.nih.gov/34943446/>

TWO OF ME: LIVING WITH PORPHYRIA



Two of Me: Living with Porphyria is a video that chronicles the lives of seven people around the world living with acute hepatic porphyria (AHP). Directed by Emmy-nominated filmmaker Cynthia Lowen and developed in collaboration with the AHP community, the film is a story of human strength, perseverance and survival. Many common themes emerged in their stories, including the challenges in receiving a diagnosis, the immense physical, mental and emotional toll of the disease, as well as its impact on relationships, careers and aspirations. Through compelling personal interviews, **Two of Me: Living with Porphyria** explores these key themes and aims to inspire viewers to “Tell 15” others about the film to generate greater awareness of AHP and help shorten the 15 years it can take to reach an accurate diagnosis. **Two of Me: Living with Porphyria** is available on PinpointAHP.com.

Thank you to Alnylam Pharmaceutical for producing this extraordinary documentary!



Also, view the LIFETIME channel program, the **Balancing Act**, with AIP patient Nicole Castellano and porphyria clinician Dr. Angelica Erwin. It can be seen at: www.porphyrria.com/patient-stories/balancing-act. Enjoy!!!



FACEBOOK NETWORKING

To find support and chat with other porphyria patients and families, join one of the seven APF FACEBOOK groups:

- » Porphyria - American Porphyria Foundation
- » APF PORPHYRIA – Porphyria Research
- » APF PORPHYRIA – Acute Porphyria (AIP, HCP, VP, ADP)
- » APF PORPHYRIA – Porphyria Cutanea Tarda (PCT)
- » APF Porphyria- Congenital Erythropoietic Porphyria
- » APF Porphyria- Erythropoietic Protoporphyrria
- » Porphyria International Support and Education

Continued from page 1

discussing porphyria. My sister said, “Remember when You sent me to see the young man who died in the Tuskegee Hospital?” I promptly shared the amazing story of meeting his daughter, Ashley, and her mother.

As soon as we arrived home from dinner, I got back to work and opened FACEBOOK to answer patient questions. The very first thing I saw was a Facebook notice that it

was Ashley’s birthday! I couldn’t believe the coincidence! I contacted her and wished her a Happy Birthday and shared with her what had happened at dinner. Neither one of us could believe the “God incidence” that had just occurred. Deborah also contacted Ashley to tell her all about her time with her father at the hospital and how heartbroken we all were at his passing.

Then came the next part of the story. Ash-

ley told us that February 28, only two days hence, was the Anniversary of her father’s death. Imagine Robert’s passing anniversary is February 28, RARE DISEASE DAY. Her story is fitting for tomorrow.

By Desiree Lyon

VISIT THE APF STORE!



T-shirts, umbrellas, safe /unsafe druge list, and more!

PATIENTS FURTHERING SCIENCE



Diana Ijames

MITSUBISHI ROUND TABLE APF EPP members, **Lara Romack, Jennifer Beck, Skyler Paris, Danielle Beeman, Diana Ijames, Wesley Scott Conklin and Ryan Anderson** recently participated in a Round Table Discussion with Mitsubishi Tanabe. The EPP people provided feedback on various prototypes of digital solutions, including wearable light sensors and companion smartphone apps. As a potential project, the new technologies will provide automatic alerts about potentially harmful light exposure in conjunction with overall symptom tracking. Mitsubishi wanted to understand more about the challenges of EPP patients and get their perspectives on our proposed digital solutions. The goal of this patient panel was to accelerate the development of an Around-the-Pill Solution that will help keep EPP patients more engaged and on track with their treatment and empower them to manage their disease more effectively. Thanks to Nancy Bayerlein and our EPP friends.



RARE DISEASE DAY

Rare Disease Day was held on February 28th, 2022. There are 7,000 rare diseases that impact more than 300 million people around the world. Thousands of events were held in over 100 countries to celebrate Rare

Disease Day (RDD), including a Global Chain of Lights, like Illumination of Fontaine de la Place Royale in France and Illumination of Estátua D. José I – Terreiro do Paço in Portugal, Illumination of San Francisco City Hall in the USA, as well as art contests, sporting events, fundraising activities, bebinars, Legislative efforts, etc. Thank you, APF members, for making RRD a great success.



**REMEMBER ...
RESEARCH IS THE
KEY TO YOUR CURE!**

Visit porphyriafoundation.org for more information



PLEASE NOTE:

In response to the many questions from patients about treatments and assistance programs, the APF is publishing information to help you gain treatment access. This is not an attempt to promote one treatment over another.

ALNYLAM ASSIST



Alnylam Pharmaceutical offers the Alnylam Assist® program to provide personalized support and assistance to patients who want to gain access to Givlaari, the treatment to reduce attacks of acute porphyrias (aip, hcp, vp and adp). If you need help or want to know more about Givlaari and the assistance program, please call 1-833-256-2748. You can also call the APF for help understanding the Givlaari treatment and be placed in touch with patients who are being treated with Givlaari.

SCENESSE® THE SUN IS OUT !!!



It's time for EPP people to have MORE protection from the intense summer light. SCENESSE®, the only FDA treatment for EPP, is a prescription medication from Clinuvil Pharmaceutical that contains the active substance afamelanotide, which is used to increase tolerance to light in adults with a confirmed diagnosis of erythropoietic protoporphyria (EPP). It is a single implant given subcutaneously every 2 months. Many EPP patients are enjoying life in the outdoors with Scenesse, and so can you! Visit scenesse.com.

Read Jennifer Beck's account of her experience:

When I first heard of Scenesse®, I vowed to do anything possible to get this new treatment. At that point, my reactions were so severe that I was no longer able to be in sunlight or indoor light. My desperation increased as the severity increased. Since it was not approved in the U.S. yet, I began my first trip to Switzerland for my first dose of Scenesse®. The journey was extremely expensive, as was the drug, but I was willing to do whatever it took to find help. The team in Zurich were wonderful and so was the treatment. Since life changed immediately with the first dose, I mortgaged my house to buy freedom from pain and the "imprisonment" inside my home that EPP caused. It was worth every penny as my life was finally changed. Now that Scenesse® SCENESSE is FDA approved, my insurance pays for the treatment. Please go to scenesse.com to gain access and get a list of treating doctors near you. As you can see from the smile on my face, I am enjoying my transformed new life.



EXCITING AFAMELANOTIDE NEWS

Researchers observe that Afamelanotide expands liver protection, as well as photoprotection and reported their findings in the journal, *Therapeutic Advances in Rare Disease*. (2021, Dec Vol. 2: 1-17)

According to Jasmin Barman PhD (photo), who is not only one of the study researchers but is also an EPP person. She and her colleagues, Drs. Anna-Elisabeth Minder, Jasmin Barman-Aksoezen, Mathias Schmid, Elisabeth I. Minder, Henryk Zulewski, Christoph E. Minder and Xiaoye Schneider-Yin, collaborated on this extremely important research. The reason this is so important to EPP people is because in Erythropoietic protoporphyria (EPP) there is an overproduction of protoporphyrin in maturing erythroblasts. Excess protoporphyrin leads to incapacitating phototoxic burns in sunlight exposed skin. The biliary elimination causes cholestatic liver injury in 20% and terminal liver failure in 4% of EPP patients. Thus, the risk of liver injury increases with increasing erythrocyte protoporphyrin concentrations. Afamelanotide was shown to improve sunlight tolerance in EPP and exciting results of expanded liver protection.



PANHEMATIN® ASSISTANCE AND SUPPORT

Patient Advocacy Liaison (PAL) For Individuals Taking Panhematin®
<https://www.panhematin.com/support>

Panhematin® is used to treat acute porphyrias, AIP, HCP, VP, ADP. The PAL provides one-on-one support for patients and their caregivers that may include:

- » Providing education about acute porphyrias and how it can impact the body
- » Answering questions Panhematin®, such as dosing or how Panhematin® works
- » Helping you if issues arise accessing Panhematin®
- » Helping you to connect with resources regarding acute porphyrias

These services are provided at no additional cost to you by Recordati Rare Diseases (RRD), the maker of Panhematin®, to help you manage the Panhematin® therapy. RRD Patient Support Program for Assistance, **Phone: 866-209-7604 Fax: 866-209-7599**. Also available in Spanish.

The reason they looked at me for help is that my career involves consulting on issues related to accounting controls and business processes. I earned an MBA with a concentration in accounting and finance from the University of Texas and hold professional certifications/licenses in Audit, Risk Management and Assurance, and Fraud Examination and others. My thirty-year career focused on auditing, accounting, consulting, and if the APF needed this expertise, I was more than willing to continue volunteering my time. I was unanimously approved in late October, 2021. In less than a month, the APF experienced a lot of change. In the spirit of transparency, I will share with you some of the main events and my view for the future.



Internal Reviews

In this process, in November 2021, I recommended that we hire an outside firm, legal and forensic, to assist us and provide independence in the review process. The board voted to engage this firm and based on the firm's recommendations; we were advised to not make any comments related to the project until they are done. Our intent is to ensure our statements are based on a complete analysis of the facts. When the reviews are complete, we will take the appropriate actions and make the necessary disclosures. I apologize to those demanding immediate answers in response to rumor and outside accusations. If you heard rumors related to "events or actions taken at the APF," this is unfortunate, and I do not understand the motivations behind those spreading rumors and accusations. My focus is on the integrity of the APF and ensuring our business practices comply (in the past and currently) with all applicable laws and adhere to the highest ethical standards. All parties within the APF are working in good faith to build a stronger foundation. Our focus is, as it should be, toward the future helping those in need.



Current Operations

The vacancies at the APF created a need for someone to assist with day-to-day op-

erations. The Board asked Desiree Lyon, our Global Director, to assist us until we identify the right candidate. Her forty years of experience co-founding and running the APF are invaluable, and I have promised her that we are diligently seeking a replacement. I want to also thank our existing and new employees for their assistance and being part of our extraordinary growth, namely the addition of 1,000 new members in the past few months. We have spoken to individuals who are looking forward to joining the APF's Board and Scientific Advisory Board who share our vision for the future. I am excited about what these folks bring to the APF so stay tuned for announcements!



Personnel

Executive Director Kristen Wheeden resigned from the APF in November 2021. When she resigned, she immediately sent a message to around 5,000 recipients announcing her resignation. As she chose the manner and means to address her departure, I directed the staff to not send the information again. I apologize if you were left off her original distribution.



The Future

In the last newsletter, we announced our "Patient's First" initiative. This is really taking the APF back to its roots. When I became involved, the APF was about helping people and addressing their individual needs. The manner with which we accomplish this may change over time, but the focus should be consistent. I spoke with several patients after I accepted this role and was informed that many felt the APF had strayed away from a "people focused" organization to one that was concerned with drug studies and research. Do not misunderstand, research and studies are critical and all of us want to see continued improvements/advances in treatment options. I am excited that in just the last few years, more new treatments became available for multiple porphyrias. I am proud of the APF's involvement in making this happen. The APF worked with the researchers to identify study participants and reached out to physicians and patient groups all over the world. We also assisted with the difficult

FDA approval process so more options would be available to physicians and their patients. If groups reach out looking for the APF to assist in research efforts, we will continue to provide appropriate assistance where it is needed. When I say "People Focused," I mean that at our core, we are a patient organization. No matter what we do, it has to be with clear benefits for, and in consideration of, our members/patients. If our members feel distanced from the APF, no matter our intentions, the APF is not doing something correctly. I discussed this with many of you as well as other board members and stakeholders in the community. In response, I propose some changes that I hope will align us to our patient focused goals and address these concerns. Some of the highlights are as follows:



Organizational Changes

- 1) Identifying a new Executive Director who clearly understands and believes in our "Patients First" mission,
- 2) Having a patient voice on the Board of Trustees,
- 3) Creating a program for structured volunteer participation,
- 4) Improving Social Media,
- 5) Enhancing Communication,



Initiatives/Patient Resources

- 6) Establish pain programs,
- 7) Develop resources related to addressing insurance problems, applying for SSDI, Medicare/Medicaid and patient financial assistance,
- 8) Assist with resources related specifically to diagnosis.

I was not able to speak to every patient and caregiver, but those with whom I did speak, I appreciate all the feedback. I hope you all are excited about the direction of the APF and I look forward to reporting to you all on our successes!

THE DYNAMIC DUO

THANKS TO AMY CHAPMAN and AMY BURKE FOR THEIR DAILY EFFORTS TO SERVE THE PORPHYRIA SOCIAL MEDICA COMMUNITY. If you need assistance, reach out to them on Facebook or contact the APF at 866 APF-3635.

UPCOMING EXCITING NEW APF SCIENTIFIC ADVISORY BOARD MEMBERS

The APF has added psychologist and pain expert, Dr. Judith Miller, and Emergency Room specialist, Dr. Brad Lee, as well as four more porphyria experts for our Scientific Advisory Board. WATCH THE NEXT NEWSLETTER, WHICH WILL FEATURE ALL OF THEM. We are sincerely grateful for all of our scientists and clinicians!

A LITTLE BIT OF HISTORY

As long as there has been human life, there has been porphyria. Porphyria has continued to mutate throughout ensuing generations. The name “porphyria” is rather recent. For centuries it was known as blood/liver disease as the abdominal pain was thought to be from the liver. The term porphyria is derived from the Greek πορφύρα, porphura, meaning “purple pigment.” The name is likely a reference to the purple discoloration of feces and urine when exposed to light in patients during an attack. Although original descriptions are attributed to Hippocrates, the disease was first explained biochemically by Felix Hoppe-Seyler in 1871 and acute porphyrias were described by Dutch physician Barend Stokvis in 1889.



Hoppe-Seyler

In 1844 Gerardus Johannes Mulder determined the chemical composition of this purplish, iron-free substance, which he named “hematin.” He also illustrated that hematin took up oxygen. In 1867 J.L.W. Thudichum described the beautiful spectrum and fluorescence of these red porphyrins after he published his analysis of urine.

Based on that, in 1871 Hoppe-Seyler crys-

tallized hematin and described its spectrum. He then demonstrated that the crystalline form differed from one animal species to another. Using his own newly constructed gas pump, he found that oxygen formed a loose, dissociable compound with hemoglobin, which he called “oxyhemoglobin.” He renamed the iron-free hematin ‘hematoPorphyrin.’ The German physician is also known for his work toward establishing physiological chemistry (biochemistry) as an academic discipline. He was the first to obtain lecithin in a pure form and introduced the word “proteid” (now protein). Additional contributions included metabolic studies and researches on chlorophyll and blood, and the abovementioned hemoglobin, which he obtained in crystalline form.

In 1874 Dr. J.H. Schultz first described a case of a 33-year-old male weaver who suffered from skin sensitivity, an enlarged spleen and reddish urine from infancy. He called the condition pemphigus leprosus. His was most likely the first description of protoporphyria. Schultz was later credited with giving the disease its name.

In 1880 MacMunn described the dark reddish urine of a patient with symptoms of an attack of acute porphyria.

Shortly after sulphonal was introduced as a hypnotic drug, Joseph Stokvis had a pa-

tient who, after taking the drug, excreted the tell-tale dark reddish urine typical of porphyria. The elderly woman then became paralyzed and died. Stokvis deduced that the pigment in her urine was the hematoporphyrin. Based on that experience, in 1889 B. J. Stokvis published the first case and clinical description of acute hepatic porphyria.



Stokvis

CEP porphyria was identified in the year 1923. In 1930 Hans Fischer, the Nobel laureate, described heme as the compound that makes blood red and grass green. By 1937 Dr. Waldenstrom in Sweden published his findings. For a time AIP was known as Swedish porphyria, or Waldenstrom’s porphyria. In the ‘60s porphyria research began in earnest in Europe and the US. The big break came when scientists were able to recognize ALA and PBG.



Stokvis

By the 1960s, all known types of porphyria had been identified and environmental factors were shown to affect the disease course. Research in the 1980s and 1990s led to the identification of the molecular defects in each type of porphyria. Currently, scientists have focused on gene therapy as a treatment for porphyria.

IN MEMORY & IN HONOR



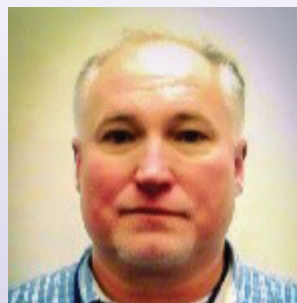
Members honor their loved ones with donations to the APF. We empathize with members who have lost loved ones.

IN MEMORY:

Maureen Curran for **Dr. Peter Tishler**, Desiree Lyon for **Hunt Norment**, Gary Quenzer for his beloved wife, **Rosalinda Quenzer**.

IN HONOR:

Johnathan Turell for **Ryan Turell**, Paula Hendrix for **Ralph Gray**, Laura McQuade for **Andrew Mc Quade**.



We are also sad to report that our database manager, **Hunt Norment**, passed away recently from COVID. Hunt was a greatly loved member of the APF staff and an APF volunteer for almost thirty years. Hunt designed and maintained our 14, 000 member database and was an essential part of our foundation.

What's New at the APF www.porphyrifoundation.com

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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Is Your Membership and Contact Info Up to Date? The APF is able to maintain our physician and patient education programs and many other services because of your support. Since we do not receive government funding, we need your support and donations. We also need your new contact information if you have a new address or email. Be sure to send us your email address so you can receive our weekly Porphyria Post.

IMPORTANT NOTICE: If you sent mail to the APF, and it has not been returned to you, please contact the APF at general@porphyrifoundation.org or call 866 APF 3635. Please address all mail to American Porphyria Foundation, 3475 Valley Road NW, Atlanta, GA 30305.

DON'T FORGET TO DONATE. YOUR HELP IS NEEDED TO EDUCATE PHYSICIANS AND PATIENTS AND SUPPORT RESEARCH - THE KEY TO YOUR CURE!!!




**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, Suite 200
Bethesda, MD 20814
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✉ www.porphyrifoundation.org

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