

IN THIS ISSUE

Announcements | **2**

News | **3**

Cutaneous Porphyria | **4**

Acute Porphyria | **5**

Member Stories | **6**

Advocacy | **7**

DEAR MEMBERS:

We love you and thank you for your kindness. Together we give education, support and hope to a world of porphyria people in need.

FROM APF STAFF:

Nicole Castellano, Imani Thomas, Paxton Cumming, Amy Burke, Yvette Strange, Carol Hughes, and Virginia Salcutan

AND THE APF MEMBER ADVISORY BOARD:

Sean Albright, Tasha Alicea, Steve Ferry, Terri Witter, Gudrun Debes, Mayra Martinez, Keshia Johnson, George Hodder, Claire Richmond, Debra Knapp, Mary Schloetter, Kika Shabot, and Andrew McManamon



Introducing “Dr. DNA” Dr. John Manak

Our newest Scientific Advisory Board member is Dr. John Manak, also known as “Dr. DNA.” He studied oncogenes and obtained his PhD at Columbia University. He trained as a molecular geneticist at Stanford University and used fruit flies to model human disorders, in particular, studying the fly orthologue of the c-Myb protooncogene. More recently, he’s worked on identifying a new function for Dm-Myb in stabilizing Polycomb repressive domains.

One major focus is to model an epilepsy syndrome in prickle-mutant flies that is strikingly similar to the clinical features of humans carrying PRICKLE mutations, including an early-onset, progressive epilepsy

featuring myoclonic and tonic-clonic seizures as well as ataxia. Recent work provided direct genetic evidence that oxidative stress-mediated activation of the brain innate immune system leads to neuronal cell death, which exacerbates the seizure phenotype.

Another focus is identifying rare, high effect size gene variants that cause congenital anomalies. Genomics techniques help identify new genes/genomic rearrangements for spina bifida, bilateral renal agenesis, branchio-oto-renal syndrome, and cleft lip and/or palate. Once relevant genes are identified, vertebrate models functionally validate them.

Dr. Manak teaches at the University of Iowa Departments of Biology and

Pediatrics at the Carver College of Medicine.

In addition to porphyria projects, Dr. Manak is preparing an educational Zoom for APF Members. Stay tuned!



Dr. Manak is a leading expert in genetic research and a new member of the APF Scientific Advisory Board.

ANNOUNCEMENTS

Tucker, APF Calendar Winner

George Hodder's pup, Tucker, won first place at our APF Pet Calendar Contest! Other winning pets featured include Mila (owned by Nicole Castellano), in third place, Armstrong (owned by Lara Quintero) and in fourth place, Memphis (owned by William Brougher and Miller Polly). Other pets will be featured in the calendar.

Tucker's previous owners passed away, and George shared this heartwarming story, "We took Tucker into our home in May of 2019, and he's been a daily comfort. We are honored and blessed to have him. He is a gentle and loving pup and enjoys snuggling up and sitting with any of us on the couch." Tucker was twelve in the photo and sadly will never see the cover. An

aggressive cancer recently took his life. "We'll miss him," George said. Congratulations and condolences to George Hodder and his family.

Pet calendars make fantastic holiday gifts! Get yours starting November 15th at [the APF online store](#).



George Hodder's pup, Tucker, featured on the cover of the 2024 APF pet calendar!



The National Organization of Rare Disorders (NORD) Summit was held in Washington, DC on Oct 15 - 17. Patients, organizations, and stakeholders from around the world gathered to tackle the most pressing issues facing the rare disease community, including the Inflation Reduction Act, gene editing, equitable access to care, and more. [Send us your thoughts](#) on these subjects and we'll add your ideas to the NORD Summit report.

Recommendations from Patient Experience

Updates on diagnosis and treatment of acute hepatic porphyria (AHP) using data from patient interviews are thanks to a collaboration between the Recordati Rare Disease Medical Affairs Metabolic team and a group of North American porphyria experts. One of the most informative issues addressed is that Panhematin should be given as soon as possible at the onset of an attack. In other words: act fast!

Reports of delayed treatment resulted in nerve damage and ongoing chronic pain. Those living with chronic symptoms warned others to quickly address their attack pain, don't stay home and "tough it out."

One woman said, "I regret the many times I stayed home instead of getting treated because the ER is such a terrible experience, or I couldn't

easily get a babysitter, or I was too ill to drive or too ill to even battle my way to the ER. I now suffer so much nerve damage that I wish I had known that nerve damage can occur."

Read [Science Direct article](#) "Acute Hepatic Porphyrias: Recommendations for diagnosis and management with real-world examples." Act fast!

Liver Meeting

Every November, we host an exhibit booth at the American Association for the Study of Liver Diseases (AASLD) meeting, also known as the "Liver Meeting." Last month in Boston, we met the world's leading experts in hepatology. Next year's Liver Meeting is in San Diego. [Let us know](#) if you'd like to volunteer at the booth!

Is Givlaari® Right for you?

Alnylam Pharmaceuticals has a user-friendly website to understand Givlaari® (givosarin) and how the treatment works to reduce acute porphyria attacks. Visit givlaarihcp.com/how-givlaari-works. For assistance in accessing Givlaari, see alnylamassist.com.

The APF talks to people about Givlaari® every day. Contact us with questions at 866-APF-3635 or general@PorphyriaFoundation.org.



The Spanish Porphyria Meeting

Fide Mirón, President of the Spanish Porphyria Association (AEP), invited Desiree to their recent annual meeting. Desiree began a friendship with the organization's founder, Rosario Cartes, 20 years ago after she read a *Reader's Digest* article about the APF. With Desiree's help, she initiated the AEP in Spain.

Katrina Parra, who leads the AEP social media, also spoke at the meeting, saying she received help from the APF. Katrina became seriously ill and paralyzed, but no doctor could find the cause. Interestingly, an insurance company suggested porphyria, and her true cause of her near-death illness was found at last. Since then, she's used

her experience to educate the world on acute porphyria. Watch Katrina's powerful videos on [Instagram](#) and [TikTok @katriparra](#).

Leading porphyria experts, Dr. Rafael Salamanca, Dr. Antonio Fontanellas and Dr. Oscar Millet spoke on various aspects of porphyria, including Dr. Millet's Congenital Erythropoietic Porphyria (CEP) research. Spanish patients are fortunate to have brilliant doctors caring for them and heading research projects. Dr. Salamanca, who retired years ago, continues to be the beloved "grandfather" of the Spanish porphyria community and Rosario Cartes is the beloved "grandmother."

Next year's International Porphyria Conference will be held in Pamplona,



Desiree Lyon (left) attended the Spanish Porphyria Meeting with AEP Founder and Retired President, Rosario Cartes (right).

Spain and led by Dr. Fontanellas. Community members around the world will attend. We hope you and your doctor will consider going!

Our "Rock"



Long-time APF Employee Carol Hughes.

Carol Hughes is our "rock." In today's vernacular, a "rock" is a person who can be relied upon as stable and reliable. Carol is our APF Chicago Office Administrator and part-time assistant to former President James Young. Her role evolved several decades ago as we grew larger and our needs became more complex.

Carol assumed important duties like payroll, taxes and other financial tasks. She's watched us grow from a small group to the international foundation of today. When asked about the best part of her job, Carol said, "I love my boss. He is like family,"

When she is not working at the APF, Carol is devoted to her church and serves as Treasurer. She loves sewing and gardening. She is currently tending to a litter of foster kittens. She visits her son and two daughters as much as possible.

New Global Foundations

FUNDACIÓN PORFIRIA CHILE

For years, Eileen Hudson maintained a porphyria group in Chile. But now it is official! The Fundación Porfiria Chile is a fully functioning, outstanding porphyria organization. Connect with them on social media [@porfiria_chile](#). Contact Eileen at eileenhud@gmail.com for information. She sends the APF her appreciation for helping bring about this wonderful new organization.

VIVIPORFIRIA

Learn more about a new Italian porphyria organization called VIVIporfiria at viviporfiria.it. Led by Porphyria Researcher Francesca Granata, VIVIporfiria organized an educational session on November 11. If you live in a country without a patient organization, contact the APF and we will help you start one where you live.

The Gift

by Polly Havard



A bone marrow transplant cured Madelyn's EPP.

My daughter, Madelyn, was diagnosed with EPP at four years old. My mom and aunt both have EPP, so we were able to diagnose her at an early age. She has a twin sister without porphyria, so that made the diagnosis even more difficult to navigate. We found a beautiful place to vacation in Boone, NC, allowing Madelyn to play outside despite EPP. She was a determined child, who loved running cross-country. She'd layer up in the heat of a Memphis summer and run, despite the difficulty of having EPP.

At 11, she experienced extreme pains in her abdomen. Thankfully, her pediatrician was proactive and sent us for a liver ultrasound. Her liver and spleen were enlarged. A liver biopsy

revealed she had cirrhosis. Thankfully her liver counts were still strong. We were referred to St. Jude Children's Research Hospital here in Memphis for a bone marrow transplant. St. Jude had never treated a patient with EPP, but they know bone marrow transplants better than any children's hospital in the country. Doctors there worked with Dr. Ashwani Singal, who is on the APF board of experts and at that time, was at the Porphyria Center in Birmingham, Alabama. He helped them understand the EPP side of Madelyn's condition so they could know exactly how to treat her.

She entered the hospital on June 18, 2018 and received 11 rounds of chemotherapy in nine days to prep her for new DNA cells. St. Jude found the perfect transplant donor, and she

received his cells on June 28, 2018. After six weeks of isolated hospital stay, she continued recovering from the intensity of chemotherapy for an additional nine months.

During this time, we learned the new DNA cells cured her EPP. Her protoporphyrin counts are in the normal range, and she can enjoy the sun without any pain! Her liver is healing, as well.

Last summer, four years post-transplant, she was able to work as a lifeguard. This summer she is working as a research intern at St. Jude and is hoping to study medicine. We are so thankful for all the doctors who worked so hard to get our daughter healthy. We are also thrilled that there are now several treatment options for our EPP family. [Read full story here.](#)

EPP TRIALS

For information on the Disc or Mitsubishi research trials, please contact the APF 866-APF-3635.

EPP Reading

Read the results of a new study that clearly shows how afamelanotide treatment can dramatically and positively impact the lives of EPP patients. [Read "Into the Light: Afamelanotide and the Treatment of Erythropoietic Protoporphyrin in the United States."](#)

Cyac elle[®] News

Many are asking about Clinuvel's new product Cyac elle[®], the next generation of photoprotection against UVB, HEV and blue light. Cyac elle[®] blocks more than 70% of HEV and blue light, but is only available [online](#) in Europe. Stay tuned to the APF and be the first to hear when it's available for use in the U.S.!

PCT News

A new report studied chronic hepatitis C (CHC) as a risk factor for porphyria cutanea tarda (PCT). The research treated patients living with both conditions with

ledipasvir/sofosbuvirwasis, a drug for the treatment of hepatitis C, for approximately a year. The conclusions were ledipasvir/sofosbuvir, and likely other direct-acting antivirals, is an effective treatment for HCV in the presence of PCT. The drug can lead to clinical remission of PCT without

additional phlebotomy or low-dose hydroxychloroquine treatment.

[Read "Ledipasvir/Sofosbuvir Is Effective as Sole Treatment of Porphyria Cutanea Tarda with Chronic Hepatitis C."](#)

A New Liver for an Ultra Rare Person

When we answered the phone, an excited, familiar voice on the other end announced, "I got a new liver!"

It was our dear friend, Milton Cubas. Milton is one of only nine people in the world with the rare ALAD Porphyria (ADP), and was the first diagnosed in the western hemisphere. Many people think that ADP cannot be diagnosed, but Dr. Karl Anderson and Milton are proof that it is possible!

Milton experienced a miracle. He received a liver from a recently deceased relative after his family learned about the ADP. At the time, Milton was in South America. When



Milton has ADP and recently received a life-saving liver transplant.

he found out, everyone from the pilots, flight attendants, and the airline itself began clearing the way to allow him to return immediately.

Soon, he was back in Miami getting prepared for the transplant.

These days, Milton is feeling better than ever, thanks to his relative. [Read full story here.](#)

ADP is characterized by almost complete deficiency of the enzyme delta-aminolevulinic acid (ALA) dehydratase by the ALAD gene. Deficiency of this enzyme leads to the accumulation of the toxic porphyrin precursor ALA, rather than the usual porphobilinogen (PBG). ADP symptoms can present in childhood and all nine known cases are males. [More info on ADP.](#)

30 Years for an Answer

Testimonial by Sharon Koch

My porphyria started in the mid-1960s when I was in high school. In a haze of confusion, I couldn't remember my class schedule. Doctors patted my head and told my parents not to worry, that I was just being a "normal teenager." I got worse with depression, extreme muscle pain, weakness, low back pain, abdominal pain and no appetite. I lost weight on and off for six years. I'm a medical assistant and I loved my job, but there were days my hands and arms shook so badly that I couldn't do my work. No doctors could identify my problem.

When I had major surgery, the resulting abdominal pain became so severe that for the first time my

doctors became concerned. Maybe it wasn't all in my head. Medication made my pain even worse, so my family and I were scared. I couldn't stand up without help. My doctor thought I had multiple sclerosis, but tests were negative. Again, I was told I was exaggerating!

These symptoms went on for decades. In mid-1990, I began experiencing nerve pain and numbness in my thighs and mid-back. The numbness continues to this day (editor's note: see Recommendations from Patient Experience, page 2). When I told my doctor, he wanted to test for porphyria. Finally, a test was positive for AIP. It only took 30 years to get an answer! It sounds strange to be happy about positive results, but for all of you



Sharon Koch has AIP, but her diagnostic journey lasted an agonizing 30 years.

with similar stories, you understand!

I feel bad I passed AIP to one of my children. At least my daughter won't have to go through all I did. Whenever someone tells me doctors can't find an answer, I say: "Don't give up! I'm living proof." [Read full story here.](#)



WE DO **NOT** SHARE INFORMATION ABOUT MEMBERS WITHOUT THEIR PERMISSION.
WE DO **NOT** SHARE MEMBERS' NAMES TO PHYSICIANS WITHOUT THEIR PERMISSION.

My Acute Porphyria Story



Melissa has AIP and a rescue Chihuahua named Simon.

Testimonial by Melissa Bueto

It all started my junior year of college. I was chronically anemic, fatigued, and depressed. No one could find anything wrong, so I chalked it up to poor diet. Six years later, I had my first attack. The ER doctor said I had a big “gas bubble” and sent me home with magnesium citrate. Obviously, to them I was being a big baby.

The next attack occurred a month after my daughter’s birth. I thought

it must be my gallbladder, even though all tests came back negative. Attacks continued every few months for the next several years. No one ever had any idea what was causing the mysterious episodes. In 2021, I had gastric sleeve surgery, and then attacks really ramped up. Sometimes my arms and legs quit working, sometimes I struggled to breathe, and sometimes I would nearly pass out.

Each attack is different, but the one constant symptom is debilitating pain. It’s worse than a C-section without anesthesia. My blood pressure increases dramatically, my pulse rises above 150, and I hyperventilate with major anxiety.

Fortunately, I met a new primary care physician who listened to me and said, “Let’s test for porphyria.” In his 25 years of practice, he’d seen one other case. Sure enough, I had AIP. To

have a name put to this misery was a weight lifted off of my shoulders.

I recently trained my dog Simon to call out for help should I lose consciousness in an attack. What good could a little chihuahua do in an emergency? Have you ever heard an incessantly yapping dog? Having him gives me peace of mind. Simon’s even tuning in to when the attacks begin building. For now, all we can do is take each day as it comes. Repeat after me....”right foot, left foot, right foot, left foot.” [Read full story here.](#)

Get Social with the APF

Every day, find new content on our Instagram, X (formerly Twitter) and Facebook accounts. The APF hosts seven Facebook groups, with a total of 10,000 followers. Everyone is welcome to our free educational sessions and virtual meet-up events, like a monthly book club.

If your life’s been impacted by this disease and you’re inspired to share, contact our social media coordinator, Amy Burke, at AmyB@PorphyriaFoundation.org. We’ll archive your story on the APF website as well. In case you missed them, here are some recent posts.

MOTIVATIONAL MONDAY



Cheers & Tears is our special space to share what’s happening in the lives of our members. Let us laugh and cry with you! Contact the APF with your news and send a photo when possible.

CHEERS

Congratulations to **Rebecca Boone**, daughter of APF member, **Mike Boone**, from Brush, Colorado. Rebecca is the 2023 Rodeo Queen!



Rebecca Boone was crowned the 2023 Rodeo Queen! She is the Daughter of APF Member Mike Boone.

Bravo to Rebecca, and to her father, who despite his porphyria, helps with the APF Facebook and educates health providers.

Cheers to **Terri Witter** for making it home from a rehabilitation facility after two years in and out of full-time care. Fun fact: the Cheers & Tears column was Terri’s idea!

TEARS

Our deepest sympathy to the family of **Paolo Castellano**, who passed away from a tragic accident. Paolo was the brother of APF Director Nicole Castellano. He was a long-time supporter of the APF.

ADVOCACY

WARRIOR WEDNESDAY

Testimonial by Elle Rivers

If I hear the word “inconclusive” one more time, I might scream. It started with exhaustion, bone-tired exhaustion. Then the pain began. I went to the doctor, and they ordered tests. We thought it was Lyme disease, lupus, or mono. Then it was cancer. Then it was in my head.

I signed up for therapy and begged my therapist to fix my brain because I didn't want to wake up in pain anymore. But it wasn't in my head.

Some days, I felt fine—great even—but it never lasted. My skin prickled in the sun, popping hives around my neck if I spent more than twenty minutes outside without sunscreen and shade. I stopped eating because everything tasted like ash. My eyes turned yellow. My urine was dark. The hours before an attack peaked, a white-hot fire would ignite inside my bones, and I knew it wouldn't be long until I felt the familiar, intimate fingers of a ghostly hand slither beneath my skin and squeeze my liver until I couldn't breathe. I would vomit bile and silently sob in the bathroom at work afraid to let everyone know how close I felt to death... and how I often wished it would come. The tests continued to come back “inconclusive.”

In 1997, my father died. He turned yellow, the same shade as the legal pads he used to write his sermons on. They said it was liver failure, but weren't sure why. His tests were also inconclusive. They treated him for hepatitis he didn't have. He endured X-rays, biopsies and CT scans. He was told his blood tests were fine. His liver was fine. He was fine. But he wasn't fine. When he died, I was nine.

The first hematologist I visited suggested porphyria. She did one

test, and it came back inconclusive. Shrugging her shoulders, she said, “It's not a no, but it's not a yes,” and referred me to more specialists.

My liver enzymes were elevated and the lymph node in my neck stuck out. I went on antibiotics, got a biopsy, and still more tests. My arms turned purple from the needle sticks, so they stuck my hands. For months, I bled into tubes and peed into cups and received abnormal, but inconclusive, results. I remember my husband yelling into the phone as I threw up in the bathroom again, “Can you rule it out?” He demanded, “Can you tell me it's NOT porphyria? Because we just want her to feel better. Tell me it's not so we can move on!” They kept saying we needed more tests.

I took leave from my job, then we drained our bank account, savings, and daughter's college fund to pay for lab work. We fought with phlebotomists that exposed my samples to light, their carelessness costing us hundreds of dollars. We quickly ran out of resources and I had to get back to work. We were out of resources. We were out of options.

Then the positive DNA and biochemical results came in. Finally, a test that WAS conclusive. I have HCP. It's something I'll live with for the rest of my life. We can treat it, but we can't cure it, at least not yet. Thanks to my doctors, my family, my friends and everyone I know from the APF. I see you. I hear you. And I am you. [Read full story here.](#)



APF friends often give donations in memory or honor of their dear loved ones. We are grateful for each of them.

IN MEMORY

Desiree Lyon for her father, **Joseph Heflin**, one of the first ten original members of the APF

Desiree Lyon for **Alda Falchetto Isella**, the mother of Rocco Falchetto, the President of the International Porphyria Network and an EPP patient, who successfully advocated to bring Scenesse® to European patients.

Glenn & Carole Kuklewski for **Vince Kuklewski**

Gudrun Debes for **Anna Capri Meyerhoff**

George Rusnak for **Carol Rusnak**

Judy Jollay for **Patricia Ann Scott**

Karen & Don Anderson for **Paolo Castellano**

Lynn Heise for **Paolo Castellano**
Council for Adult & Experiential Learning and Hilary Cooperman for **Paolo Castellano**

Laurel Adams for **Paolo Castellano**

Ken & Pam Kuznicki for **Paolo Castellano**

Ann & Bill Graves for **Paolo Castellano**

Nancy & Jesse Cabral for **Paolo Castellano**

Luciano Castellano for **Paolo Castellano**

Cory Stansbury for **Paolo Castellano**

James & Lucy Buck for **Paolo Castellano**

Thomas Dionese for **Paolo Castellano**

IN HONOR

Desiree Lyon for new APF Director **Nicole Castellano**, and APF Board of Trustee Members, President **Paul Stickler, Diana Ijames, Warren Hudson, Ron Polly, & James Young**

Sharon Koch for **Jagger & Jake Liquori**

Charles Davis for **Ginger Davis**

Carolyn Cafasso for **Adam Cafasso**

Karen Eubanks for **Desiree Lyon**

Ruth Edwards for **Desiree Lyon**

What's New?

Check out www.PorphyrriaFoundation.org

The information contained on the APF website or newsletter is provided for general information only. The APF does not give medical advice or engage in the practice of medicine. The APF under no circumstances recommends particular treatments for specific individuals, and in all cases recommends a consult with personal physician or local treatment center before pursuing any course of treatment.

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Why donate to the APF?

We've put patients first for over 40 years. APF staff work on nights and weekends to help people worldwide find answers to their painful symptoms.

Your donation helps us provide doctor packets filled with pertinent information for newly diagnosed, add to a growing database of 6,000 treating physicians worldwide, support a physician education program, maintain a database of safe and unsafe drugs and more!

Every donation is tax deductible and incredibly appreciated. The APF does not receive government funding.

Donate on our website
24 hours a day,
7 days a week.



THANK
YOU!

UPDATED CONTACT INFORMATION?

Contact 866-APF-3635 or
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