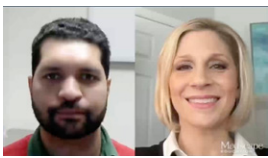


## Rare Disease Day 2024

Icahn School of Medicine at Mount Sinai invited APF Director Nicole Castellano to speak as part of a Continuing Medical Education presentation by Dr. Ibrahim Elsharkawi released on Rare Disease Day! Dr. Elsharkawi is a Medical Biochemical Geneticist physician and Assistant Professor in Genetics and Pediatrics at the Icahn School of Medicine at Mount Sinai. He educated on the Porphyrins and interviewed Nicole, who shared her diagnostic journey. Thank you to the Icahn School of Medicine at Mount Sinai and to Dr. Elsharkawi for shining a light on porphyria for Rare Disease Day 2024! The more awareness created, the more doctors will think to test for Porphyria, which will reduce the current 15-year delay to diagnosis. To watch Dr. Ibrahim and Nicole, please visit this link: <https://youtu.be/LRvqosLHMBQ?si=3nZFAbXPHG8bdFB->



Claire Richmond is a writer and advocate for Rare Diseases, particularly Porphyria. She has Acute Hepatic Porphyria and participates on the APF Member Advisory Board. Claire shared her story on the News Station We Are Iowa and published an article in the Iowa Capital Dispatch titled "Here's What I Learned From

Seven Years with a Rare Disease - Leap Day is also Rare Disease Day." Way to go, Claire - You are such a great advocate for all who suffer from porphyria. Thank you for making a difference!



Aside from the most recent CME course described above for Rare Disease Day, the APF was asked to educate the Fellows at the Swedish Hospital in Seattle. When Nicole's brother passed, Desiree Lyon, the retired Director of the APF, spoke in Nicole's place about her experience with acute porphyria, followed by a Q&A session. The APF is scheduled to participate in other physician education opportunities.

Channel 5 News Story link - <https://www.weareiowa.com/video/news/health/524-2dc01b1f-cb5c-4f7f-adb9-c2cb7f564136>



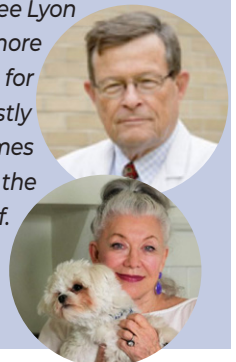
Article Iowa Capital Dispatch link - <https://iowacapitaldispatch.com/2024/02/29/heres-what-ive-learned-from-seven-years-with-rare-disease/>



Thank you! Thank You! Your Christmas and Holiday gifts enabled the APF to distribute more than 3,700 doctor packets to physicians and patients to provide to their doctors. These packets are replete with diagnostic and treatment information on each specific type of porphyria. With your donations, we also were able to create three new porphyria publications which detail subjects like anesthesia in porphyria, pregnancy, initiate a huge National Porphyria Awareness Week campaign, initiate the Center of Excellence program and so much more.

## Continued Congratulations!!

The APF continues to hear from patients and doctors with worldwide retirement well wishes for Desiree Lyon after co-founding the APF and serving for over 40 years. Among the many notes and calls was one from Dr. Karl Anderson, the first APF Scientific Advisory Board Chairman. Upon her retirement, Dr. Karl Anderson wrote, "Congratulations and best wishes to Desiree Lyon (Dee) for her retirement! Dee has done more than anyone, anywhere for patients and for supporting research on porphyrias, mostly through the APF, which she and JY (James Young) founded many years ago. She ran the APF well and has so much to be proud of. Hope we can get together again at some future porphyria meetings or celebrations! Sincerely and all the best!"



## Recordati Invites APF Director to Share Patient Journey

APF Executive Director Nicole Castellano was invited to speak at a Recordati Rare Diseases meeting in Ft. Lauderdale, FL, to explain what living life with AIP is like along with other rare disease patients. France Lebel, Senior Director, Medical Affairs Canada and Medical Affairs Metabolic North America, interviewed Nicole as she shared her personal journey with AIP, the APF's role in her journey, as well as what fueled her to become an advocate for other patients as the new Director of the APF. Members of the audience asked many questions to better understand what it's like to live with AIP. One employee asked Nicole if she could summarize in one sentence advice for other rare disease patients. Nicole answered, "No matter how hard it gets, do not give up!"

Recordati manufactures Panhematin®, which treats acute porphyrias and was the first drug to gain Orphan Drug Status. Nicole felt everyone at the meeting was truly interested in what patients go through to better understand the reason behind their hard work. The APF thanks Recordati for the opportunity to spread awareness and for their continuous efforts to understand and support patients!



*France Lebel of Recordati Rare Diseases interviews APF Director Nicole Castellano*

## Eric Lifschitz AIP Warrior

While in Ft. Lauderdale, Nicole was honored to meet one of the first APF members ever, Eric Lifschitz (AIP), his friend Marty, Eric's doctor, and some of his nurses. Nicole noticed everyone's eyes lit up when they saw Eric, and they shared how much they had learned about porphyria from Eric over the years. Marty has known Eric for decades and has learned an incredible amount about porphyria and helps get Eric to his appointments.

Nicole said Eric is one of the strongest people you will ever meet! He was very ill, but he had the most positive outlook. Nicole learned about Eric's journey and how he takes the opportunity to spread awareness and educate about porphyria everywhere he goes. Eric is one of the original Porphyria Warriors! Thank you, Eric, for being such an inspiration to all patients struggling with porphyria and for always making opportunities to generously share your extensive porphyria knowledge!



*In the photo left to right: Nurses Debra and Julie, then Nicole, Eric, Marty, and head nurse Casey*

## Dr. DNA Zoom ~ Good Genes Gone Bad



Because of the great interest in DNA and the expertise of our new Scientific Advisory Board member, Dr. John Manak, the APF recently hosted a momentous DNA Zoom entitled, GOOD GENES GONE BAD. Not only did Dr. Manak present the basics of Genetics 101, but he also gave detailed information on the genetics of the porphyrias, answered questions from

the audience about their specific DNA issues and touched on the conjecture that porphyria courses through the bloodlines of the Hanover, Stuart and Tudor families. We also learned about a "de novo mutation," which is a rare occurrence when the gene

mutation arises despite not being seen in either parent.

Dr. Manak is a born teacher and has the natural ability to decode the complexities of genetics, making it easy to understand. Thank you, Dr. Manak!

The feedback from the Zoom was so positive that the APF and Dr. Manak will schedule future DNA Zooms. Watch our eNews, APF website and our social media channels for details of future Zooms with experts like Dr. Manak - everyone is invited to attend!

For those who missed this extraordinary Zoom, you can watch the recording on our YouTube channel here: <https://youtu.be/QcxO5ZZMtck?si=v3y2TsRgww1T4KsH>



## Jessica's Triumph!!!

After a long and extremely difficult trudge through the medical system, Jessica Melton has triumphed over AIP. Shortly after puberty, Jessica began having bouts of



excruciating abdominal pain along with other severe symptoms. Her parents repeatedly took her to the emergency room for help that did not happen. What complicated her case was that her symptoms disappeared just as mysteriously as they had appeared, making any diagnosis a challenge.

Her first triumph was her AIP diagnosis and subsequent treatments. Despite repeated misdiagnoses, Jessica found a physician who refused to give up until he found the source of her many attacks. He first treated Jessica with a glucose IV, which provided limited success. Then she reached out to the APF where Desiree shared about Panhematin treatment and a wealth of additional information. At 19, she began Panhematin infusions, which kept her attacks at bay.

Her next triumph occurred when she became ill again. As before, she reached out to the APF several years ago and discussed her situation with Desiree, who promptly told her about Givlaari, a new treatment to reduce the number of attacks. Fearful that she couldn't have Panhematin for breakthrough attacks, Desiree assured her that other people on Givlaari were using both. After this assurance, Jessica began her Givlaari injections with great success. Within a short time, she was able to begin working part-time and volunteering at her local Boys and Girls club. Givlaari has reduced the number of attacks and hospitalizations. Fortunately, when she does have attacks, her doctor orders Panhematin, which stops the attack quickly. Jessica is grateful to her parents for their understanding and care, her doctor for his kindness and willingness to learn about AIP and stay the course until she was diagnosed and treated, and to the entire APF staff.

If you need treatment for any type of porphyria or need an assistance program, ask the American Porphyria Foundation for help. Also, if you are on Givlaari and are having difficulty gaining access to Panhematin treatment, contact the APF at [general@porphyriafoundation.org](mailto:general@porphyriafoundation.org) or Call our Toll Free number 866-APF-3635.

## Thank You, Isabel Allende

The Isabel Allende Foundation has provided an Empathy Grant to support the programs of the APF. Isabel Allende is one of the most esteemed authors globally and the first internationally successful female Latin American author. She has been described as the "most widely read Spanish language writer in the world." Her 26 books have been translated into 30 languages and have sold more than 77 million copies worldwide. One of her books was *Paula*, an account of the passing of her daughter, Paula Frias, at age twenty-nine from acute intermittent porphyria (AIP). It is a powerful yet poignant book that all porphyria patients and families will want to read. Her most recent novel, *The Wind Knows My Name*, at age eighty, is a story of two unforgeable characters who are searching for land and home. Ms. Allende was inducted into the American Academy of Arts and Letters and received Chile's National Literature Prize, as well as the coveted Presidential Medal of Freedom in 2014. We are grateful not only for this year's Empathy Grant and for all of the funding Ms. Allende has provided in the past.



## God, We Need to Have a Conversation

See the poignant interview with APF member, Mayra Martinez, who was the guest on the MY BEST FAIL podcast with Diana Lanham. Mayra details her astounding journey from suffering minor porphyria attacks to paralysis and a near death experience. Her mystery disease was finally identified as acute intermittent porphyria but the journey to that diagnosis was one of dreadful pain and challenging, life-threatening symptoms.



Mayra enjoyed a successful career as a VP of a large electrical contracting company. Then she began having strange symptoms that doctor after doctor could not diagnose. As you watch the rest of the story, many of you will think that Mayra's experience is like viewing your own. <https://www.youtube.com/watch?v=4lti6uP9zpY&t=916s>

## New SAB Member



DR DAVID DIUGUID has been named a new APF Scientific Advisory Board member. Dr. Diuguid came to the attention of the APF when a number of acute porphyria patients described him as “outstanding, exceptional, the best ever doctors” and other accolades. His porphyria patients are devoted to him and frequently comment on his compassion and porphyria expertise.

Diuguid received his undergraduate degree from Harvard College, where he graduated cum laude.

He matriculated at Weill Cornell Medical School and received his medical degree in 1979. He did his residency at Boston University Medical Center and served as Chief Resident. He then completed his fellowship training at Tufts New England Medical Center in Boston and served on the faculty there for three years before transferring to the Hematology and Oncology Service at Columbia New York Presbyterian. Welcome Dr. Diuguid!

## Don't Miss The Upcoming Education Events

In conjunction with Rare Disease Day and the National Porphyria Awareness Week, the APF has numerous patient and physician educational opportunities planned. Several EPP, AHP and PCT ZOOM conferences will be held, the Podcast will be initiated, the mental health ZOOM, and Let's talk Porphyria!

Don't forget --- The APF is also hosting Book Club, Porphyria Partners, and Movie Night, along with our weekly Instagram and Motivational Monday, Warrior Wednesday, Porphyria Pets Friday and Satisfy the Soul Sunday.



Watch for details of the porphyria Centers of Excellence (COE) published on the APF website and eNews. Contact the APF NOW for a treating doctor in your area and a FREE doctor packet!

Did you know we have free warning cards for AHP, EPP, PCT, and CEP? The card is designed to be kept on your person and serves as a medical alert card. Emergency medical professionals can use information on the card as an educational diagnostic aid. The information on the card contains your specific porphyria type, a definition of the porphyria, guidelines for avoiding an attack, and contact information for the APF.



Contact the APF to get yours now!  
Email: [general@porphyrifoundation.org](mailto:general@porphyrifoundation.org) or call 866-APF-3635

## Liver Meeting

APF Director Nicole and APF President Paul Stickler, manned the APF exhibit booth at the annual meeting of the American Association for the Study of Liver Disease (AASLD) at the Boston Hynes Convention Center. The Liver Meeting is the leading gathering of thousands of scientists, and healthcare professionals committed to preventing and curing liver disease, as well as fostering research that leads to improved treatment options for millions of liver disease patients. The Liver Meeting aims to highlight cutting-edge research, emerging clinical trends and hot topics of interest to the hepatology community.

The APF has maintained an exhibit booth at the Liver Meeting for over 30 years, and we will be there for the 2024 meeting in San Diego! If you live nearby and would like to help man the exhibit booth, we need YOU!!! Not only will you be educating doctors, but you will have the opportunity to learn, too.



## Tragic PCT History

Did you know that in 1955-1959 Hexachlorobenzene (HCB) was sprayed in Turkey causing an outbreak of Porphyria Cutanea Tarda? The sprayed seeds were used for bread production causing an epidemic of porphyria cutanea tarda (PCT). 3000-4000 people were affected with a 10 percent mortality rate. The first case of PCT appeared in the Urfa, Province of Turkey. HCB was sprayed to get rid of a wheat fungus. Unfortunately, it may have treated the fungus, but it caused PCT and other diseases.



In 1955 Dr. Cihad Cam, who was Director of the skin clinic in Diyarbakir in Eastern Turkey, found that many children had sores and blisters on their faces and the back of their hands. These children had dark pigmented skin and hair on their faces. Also, their urine was a reddish-brown color. When Dr. Cam examined the urine in ultraviolet light using a Wood's filter, it gave a brilliant red fluorescence. He realized then that these children had Porphyria Cutanea Tarda (PCT). Prior to 1955, he had never seen children with PCT, but in that year and in each subsequent year, he saw many hundreds of affected children.

This epidemic aroused great interest and concern in Turkey. Children were admitted for further study under the care of Dr. Joseph Wray at the Hacettepe Children's Hospital in Ankara. Professor Cecil Watson of Minneapolis, renowned for his porphyria research, also sent one of his esteemed research assistants to assist in the biochemical investigation of the children. Famous American porphyria expert, Dr. Rudi Schmid, also visited Turkey and reported on the epidemic and the role of Hexachlorobenzene.

Dr. Cam, who had taken the dietary history of hundreds of children with porphyria, made a startling discovery. The farmers in the Eastern part of Turkey were extremely poor and their staple diet was bread. Their poverty prevented them from obtaining good wheat crops, so they ate the wheat grown with seed treated with a fungicide. Dr. Cam found the affected children had been eating bread made from the wheat which had been given to the farmers for planting; he quite rightly suspected that the seed wheat was re-sponsible for the epidemic.

Interestingly, twenty-five years later, 161 volunteers were studied and all of them continued to have porphyrin abnormalities, including those exposed as children. The children born to mothers with porphyria 25 years ago all died secondary to maternal milk and transplacental transfer of HCB. Current lactation specimens of porphyria patients show high HCB levels, up to 3.12 ppm, but the infant offspring appear normal, thus illustrating the power of chemicals.

## APF Podcast



The APF has had many firsts over its over 40-year history, and we are ecstatic to announce our new podcast, the first-ever Porphyria podcast, RARELY DISCUSSED! The podcast will focus on Porphyria and Rare Disease. We are most grateful to Recordati Rare Disease for sponsoring this

new and exciting podcast!

Why a podcast? The APF has enjoyed extraordinary growth over its 40-year history, especially over the past year to a present membership of 18,000 patients, families, physicians, friends, industry partners, and persons interested in porphyria. Along with our expansive growth, we have had a major increase in requests for additional educational programs, specifically those that are available on demand, such as the videos available on our YouTube channel <https://www.youtube.com/@PorphyriaFoundation>. Podcasts are particularly helpful for Porphyria people whose illness forces them to spend time alone and reach for podcasts which are the most popular platforms for education and entertainment today. The APF will feature educational programs for patients and physicians, porphyria experts, caretakers, Primary Care Physicians, Emergency Room Physicians, and psychologists.

The APF Podcast will be a 30-minute, bi-monthly audio and video broadcast. Special educational programs will be produced, including Diagnosis, Treatment explanations and guidelines, Primary Care Physicians and Expert Interviews, Research Phases, Glossary, Process, Parameters, Managing Daily Life Tips, Psychologist Interviews, Patient Resources and Patient Interviews, Media Opportunities, and the POR-FIRE-IA Children's program, to name a few.

THE APF PODCAST TEAM Our greatest asset is our Podcast team, Andrew McManamon, AIP patient, and Nicole Castellano, APF Executive Director, who will produce the Podcast together. Andrew is a realtor and hails from Brighton, Michigan, where he had his first critical AIP attack. After prompt treatment, Andrew recovered and has become an active advocate for the APF. His desire to assist other patients enduring the horrendous pain and sickness he endured has motivated Andrew to use his talents as a podcaster and producer to help the patients gain a better experience with porphyria. One of Andrew's videos got over 46,000 views in a week on TikTok. To view, click here <https://www.tiktok.com/@andrewmcmamonrealtor/video/7221640709462412587>.

## IN REMEMBRANCE

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

### IN MEMORY OF

In Memory Of Paolo Castellano

Kendra Vittorini, Adele Castellano, Eunice Richmond, Elaine Herman, Thomas Koral, Susan & Mark Markese, James & Lucy Buck

In Memory Of Connor Andrew Roy

Shannon Seneca, Elizabeth Roy, Laura & Andrea Braun, Michael, Mark & Joan French, Richard & Peggy Rockwell

In Memory Of Mary Hargett Crown

Mary Crown

In Memory Of Perry Elizabeth Pearce Benton

Douglas Benton

In Memory Of Marvin David Mathia Sr.

Keith Henry, Tracy Chase, Jay and Sandi Sanders, Mark & Shanna Mathia

In Memory Of Carol Rusnak

George Rusnak Jr.

In Loving Memory Of Vincent Farina

Michael & Carol Farina

In Memory Of Daniel Pudlicki And Bob & Virginia Waters

Jeff And Sandy Pudlicki

In Memory Of Matthew Cole

Dianna Poissant

In Memory Of Donna Pagano

Kathleen Toelkes

In Memory Of Jane Bingham Wiggins

Beth Jarvis, JMP, Jenna And Bryan Morgan, Justin Weimer, Joye And Kerry Nelson, Mckay Burton & Thurman (Dee Ann Wright), Mark & Susanne (Lyle Wiggins), Ron & Rebecca Wheelwright, Lee & Pam Crittenden, Kevin & Marilyn Wheelwright, Lorraine, Judd, Stewart And Emme Houser, Gray Robinson With Sas Institute Inc

In Memory Of Doris And Stanley Stavis

Nanette Stavis

In Memory Of Carol Ann (Shirey) Murphy

Joe & Brenda Deluca, Brandon Murphy

In Memory Of Susan Godsted Young

Sharon Bigler

### IN HONOR OF

Stephen Kowalski In Honor Of  
Lisa Coccimiglio

Jeffrey Sereno In Honor Of  
Deborah Sereno

Dianne Fletcher In Honor Of  
Darlene Bishop

Lisa Wohltmann In Honor Of  
Ryan Zila &

Toma Reichman In Honor Of  
Danielle Ovidia

Robert Sullivan In Honor Of  
Quinlan Sullivan

George Rusnak Jr. In Honor Of  
Diana Sabella

Bonnie Niglio In Honor Of  
Desiree Lyon

Sheryl & Alex Mclellan In Honor Of  
Sophia And Moriah Mclellan

Elizabeth Dwyer In Honor Of  
Desiree Lyon

Elaine Sasso In Honor Of  
Elizabeth Britton Meloni

Jim & Janet Cumming In Honor Of  
Desiree Lyon

Mary Ventrice In Honor Of  
Gia Ventrice

Jeff And Sandy Pudlicki In Honor Of  
Aaron Pudlicki

Grayfred Gray In Honor Of  
Ralph Medlin Gray

Anike Binder In Honor Of  
Friederike Binder

## APF STORE

*The APF Store has a few Pet Calendars remaining. You can take advantage of the sale.*

**Also ... Check out the store for many other items from clothing to accessories, including all the new T-shirts and sweat-shirts. Watch for the upcoming National Porphyria Awareness Week shirts, etc. We will be publishing notices of all the items available soon.**

See the following at the store and so much more at:  
<https://porphyriafoundation.org/apf-store/>



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## TEARS & CHEERS

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*Our column to share the happy and sad times in our community.*



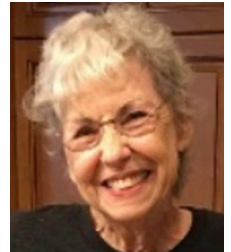
**CHEERS...** Megan Parrish and family are excited about the Baby's arrival. Megan has been an extremely helpful porphyria advocate having shared her story locally and nationally via print media and television. We are all excited about her great news. You can find Megan's story on the APF Website and in the Two of Me documentary. Congratulations!!

**CHEERS...** too, to the APF for now reaching a membership milestone of 18,000 members. What is great about the APF is that despite the large membership, we remain a small, tightknit family of friends.

**TEARS...** Connor Andrew Roy, Age 16, of Hallstead, Pennsylvania passed away recently after complications following a bone marrow transplant or CEP at the Children's Hospital of Philadelphia for CEP. Since his world was primarily out of the sunlight, his biggest passion was creating YouTube videos surrounding the video games he loved to play. In lieu of flowers, the family asked that donations be made to the American Porphyria Foundation.



Ruth "Dee" Bruno was a member of the APF since its inception. She was an RN who used her vast medical knowledge to help others understand porphyria. She gave them hope for a brighter future. We love Ruth and will miss her.



Jane Wiggins passed recently. She was a long time APF member and a help to all. Jane was a gifted vocalist, even recording two albums. She also loved playing the bassoon and acting in local theater.



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## Early Research Patient Passes Away



Our deepest sympathy to the family of Marvin Mathia, who was one of the early NIH porphyria research patients and one of the earliest APF members. Marvin was the patient of the famous Drs. Donald Tschudy and Richard Magnussen at the National Institutes of Health in the early 1970s where he

participated in their porphyria research for four months. He was a pioneer in the study of Acute Intermittent Porphyria and faced many challenges. His wife Marie was by his side forming a formidable team during the many crises.

Also, Marvin had a letter in his porphyria file from the APF dated 1984 thanking him for becoming a Support Leader. Today we have a similar program called Porphyria Partners, which provides a means for patients who are isolated, lonely or need support to help one another. This program was a Godsend to people during Covid. You can become a Support Leader for the Porphyria Partners program. If you would like to help others who need support, you can do as Marvin did by calling the APF at 866-APF-3635 or email [general@porphyriafoundation.org](mailto:general@porphyriafoundation.org).

At the APF, all research volunteers are known as MEDICAL HEROS. Giving your time and energy enables others, as well as yourself, to benefit from research. Each research patient deserves our heartfelt thanks. Marvin was a research volunteer before the APF existed. We are grateful to all who participated in research and by so doing have helped greatly expand the knowledge of all porphyrias, plus, the new treatments, diagnostic understanding, pain understanding and so much more.

## What's New?

Check out [www.PorphyrriaFoundation.org](http://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 that consult with personal physician or local treatment center before pursuing any course of treatment.

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## Donate to the APF

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



The APF does not receive government funding. Your contributions help educate physicians and patients with life-saving information about the porphyrias. Donations are tax deductible. Become an APF member today!

## UPDATED CONTACT INFORMATION?

Contact 866-APF-3635 or  
[general@porphyriafoundation.org](mailto:general@porphyriafoundation.org).

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