American Porphyria Foundation

What's New at the APF

www.porphyriafoundation.org

Is Your Membership Up to Date? The APF is able to maintain our physician and patient education programs and many other services because of your support. We do not receive government funding to run the APF, rather we receive donations from you, your friends, your family and people interested in the porphyrias. Now we need your support for several programs that are very special.

First, our *Protect the Future* program to train future experts is important to our future health. Without experts, doctors have nowhere to turn for advice and to learn about porphyria. This is a serious problem that we are trying to prevent by training young doctors, but where do we receive funding to do this except for help from our members**.**

Next, we have an enormous physician education program that distributes exceptional educational materials to doctors. Please help us produce these materials.

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American Porphyria Foundation

1st Quarter 2017

INTERNATIONAL CONGRESS OF PORPHYRINS AND PORPHYRIA 2017

BORDEAUX, FRANCE

The International Conference is a gathering of porphyria experts and researchers from around the world and is held every two years. This conference provides them with the opportunity to share their research and discuss the present and future therapies, case studies, etc. The official *Patient Day* is held on **Sunday, June 25, 2017,** followed by Congress Days 1-4, which conclude **Wednesday, June 28, 2017.** *View the agenda below.*

SESSION 1: Fundamentals on Heme Biosynthesis in relation to Porphyrias
SESSION 2: Acute Intermittent Porphyrias
SESSION 3: Bullous Acute and chronic Porphyrias
SESSION 4: Erythropoietic Porphyrias
SESSION 5: Clinical Complications of the Porphyrias
SESSION 6: Clinical evaluation and laboratory investigations in the Porphyrias
SESSION 7: Miscellaneous and Emerging Therapies in the Porphyrias

PLENARY LECTURES Experts from around the world will be presenting lectures on a host of porphyria topics:
PL1: Tissue specific regulation of heme biosynthesis, Barry **PAW** (USA)
PL2: Cellular transport, trafficking, and recycling of heme, Iqbal **HAMZA** (USA)
PL3: Acute Intermittent Porphyria, pathophysiology of acute attacks and recurrency, treatment options, overview and update, Robert J. **DESNICK** (USA)
PL4: Results from Clinical Trial using ALN-AS1, an Investigational RNAi Therapeutic Targeting ALAS1 for the Treatment of Acute Hepatic Porphyrias (ALNYLAM), Eliane SARDH (Sweden) (for EU/USA Porphyria Reference Centers)
PL5: Variegate Porphyria, update on pathophysiology and management of cutaneous symptoms, P. MEISSNER (SA)
PL6: Clinical aspects, pathophysiology and management of PCT, Jorge FRANK (Germany)
PL7: Pharmacological chaperones as a potential therapy for congenital erythropoietic porphyria, Oscar MILLET (SP)
PL8: Splice modulating therapy in EPP, state of the art, Laurent GOUYA (France)
PL9: Afamelanotide for the treatment of protoporphyria-induced phototoxicity, Janneke LANGENDONK (Netherlands)
PL11: Porphyria associated kidney disease, Nicolas PALLET (France)
PL12: Porphyria associated liver disease, Staffan WAHLIN (Sweden)
PL13: The value of long term longitudinal clinical and laboratory observations in porphyria, Karl **ANDERSON** (USA)
PL14: Measurement of therapeutic effect in the porphyrias: Balancing the needs of professionals, patients and the pharmaceutical industry, Elisabeth MINDER (Switzerland)
PL15: Diagnosing and monitoring porphyrias: benefits and disadvantages of centralizing porphyria dedicated laboratory tests, Sverre SANDBERG (Norway)
PL16: EPNET and EMQN EQAS impact on porphyria diagnosis, Sharon WAHTLEY (UK) and A. AARSAND (Norway)
PL17: Gene correction in erythropoietic porphyrias by the CRISPR/Cas9 technology, F. MOREAU-GAUDRY (France)
PL18: Inducing iron deficiency improves erythropoiesis and photosensitivity in CEP, John **PHILLIPS** (USA)
The Social Program begins with the Patient Day gathering, the Welcome Receptions, Tours of the City and ends with a Gala Dinner for all the Conference attendees. See: https://icpp2017.org/

PATIENT ACCESS TO CARE TOOLKIT

A downloadable *Access to Care Toolkit* is a resource designed to help patients living with Acute Intermittent Porphyria (AIP), Hereditary Coproporphyria (HCP), and Variegate Porphyria (VP) or their caregivers, loved ones and healthcare providers secure access to Panhematin at their preferred health facility. We have recently learned of patients who are being denied this treatment from some hospitals and directed to secure another healthcare provider. If this has happened to you or someone you know or care for, please use these tools to request help from your state and local representatives and health advocacy organizations. We understand the debilitating effects of acute porphyria and hope these resources will help you secure access to Panhematin when you need it most.

The *Toolkit* contains the following materials:
Healthcare Conversation Tracker is a simple form to record your conversations with doctors, insurance agents, etc.
Customizable letter templates to record your details to use for doctors, state departments, insurance, etc.
AIP Access to Care Fact Sheet defines AIP, its symptoms and why it's important for patients to get immediate care
Patient Bill of Rights can be used to support your appeal for access to treatment
This *Toolkit* can be found on the APF website: www.porphyriafoundation.org

APF MEETS WITH FDA FOR ACUTE PORPHYRIAS

The new air in Washington, DC is one of PATIENT INVOLVEMENT in health issues, particularly with drug development. In fact, before a drug is approved, FDA is encouraged to participate in Patient Focused Drug Development Meetings. The **Patient Focused Drug Development** (PFDD) initiative is part of FDA commitments under the fifth authorization of the Prescription Drug User Fee Act (PDUFA V). The patient perspective is critical in helping FDA understand the context in which regulatory decisions are made for new drugs. PFDD meetings give FDA an important opportunity to hear directly from patients, patient advocates, and caretakers about the symptoms that matter most to them, the impact the disease has on patients' daily lives, and patients' experiences with currently available treatments. This input can inform FDA's decisions and oversight both during drug development and during the review of a drug application.

The APF secured one of these coveted meetings for EPP in October of 2016. Now we have also secured a PFDD meeting for the acute porphyrias on March 1, 2017 at the College Park Marriott Hotel & Conference Center in Maryland. Over 70 patients, families, physicians, and industry partners attended this meeting. Participants included experts, Herbert Bonkovsky, MD, John Phillips, PhD, Karl Anderson, MD, Robert Desnick, MD, PhD, and patients Lisa Kehrberg, MD, Ariel Lager, Tara Cantley and husband, Shane, Amy Chapman and husband, Craig, Rose Jeans, Nichol Kirby, Candace Johnson, Lakeshia Johnson, Michael Boone, and wife, Sandra, Terri Witter, Evelyn Jacobucci, Colin McEwen, Mary Schloetter, Tracey Kelly, Lina Rebeiz, Louise Schloesser Braun, and MANY more!

If you missed the webcast, a full recording is available on the APF YouTube Channel at www.youtube.com/porphyriafoundation. The APF is also soliciting your comments. Please send them to the APF and we will share them with the FDA. A *Voice of the Patient Report* will become available after the conclusion of the meeting, along with submitted patient experiences.

NATIONAL PORPHYRIA AWARENESS WEEK (NPAW) April 22-29, 2017

The NPAW is the time each year for **YOU** to bring porphyria awarness to your community. We, at The American Porphyria Foundation, encourage **YOU** to help raise awareness and provide accurate information about porphyria where you live. Over the years, we have made a great impact in physician education. Now we also need to improve porphyria awareness among other medical professionals and the public in general. During National Porphyria Awareness Week (NPAW), participants can choose from a large range of opportunities to be proactive and enhance awareness among your family, friends, and community. Attracting media attention is one major means to accomplish this goal. YOU have a story to tell. Ask your local newspaper or community newsletter to include a story about your porphyria or just write an article on your experience with porphyria and submit it. You can be involved in any way that works with your schedule, resources, community, and interests. Every effort is vital to increase awareness.

The APF will help you accomplish your own activity by providing: porphyria brochures, *Porphyria Live* DVDs, fact sheets, PowerPoint presentations and materials for medical seminars, press releases for local newspapers and television and other suggestions. Even if you cannot organize an event, the materials are invaluable to mental health professionals, colleges, hospitals, employee assistance programs and all types of health organizations.

Tell your story to local media. Help others by spreading your experience. Television, newspapers, and community magazines are looking for people who have undertaken the challenge with a rare illness.

Help others by sharing knowledge about porphyria with your community, including your family members, friends and the physicians in your local hospitals. Suggest that they host a seminar, grand rounds on porphyria or a local meeting where you can hand out materials.

Share your story on social media. It is an easy and effective way of getting porphyria in the public eye.

Assist at medical conventions or health fairs to educate laypersons and physicians on porphyria. Ask your hospital or doctor if there is a local meeting where you can hand out materials or tell your experience. Befriend your physicians, and they will share their newfound knowledge of the disease.

Volunteer your talents or skills to help achieve the educational programs of the APF. You are talented, let's use it for our common good, for example, donate your paintings, sculptures, computer expertise, business acumen, etc. for our fall raffle or to help APF.

Purchase an APF T-Shirt or Cap and wear it. See the website for shirts and other APF products.

Learn how to be an advocate in your daily life and share your knowledge everywhere.

PLEASE REMEMBER TO UPDATE YOUR CONTACT INFORMATION WITH THE APF. NEW INFORMATION ARRIVES WEEKLY VIA ENEWS AND QUARTERLY VIA NEWSLETTER. BE SURE TO UPDATE TODAY!

THE RAREST PORPHYRIA

DID YOU KNOW DELTA-AMINOLEVULINATE DEHYDRATASE; ALAD or ADP is the rarest of all of the porphyrias with only 8 people diagnosed worldwide, mostly in Europe. It is an autosomal recessive disease that is characterized by an almost complete deficiency of the enzyme delta-aminolevulinic acid (ALA) dehydratase. Deficiency of this enzyme leads to the accumulation of the porphyrin precursor ALA, which can potentially result in a variety of symptoms, which vary from one person to another. Affected individuals experience intense and neurovisceral symptoms. Like the other acute porphyrias, affected individuals may experience severe abdominal cramping or pain accompanied by vomiting and constipation and neuropathy. During infancy, gastrointestinal abnormalities may cause an affected child to fail to grow and gain weight as expected. In severe cases, the motor nerves are involved, resulting in loss or partial impairment of the ability to use voluntary muscles. ALAD porphyria can also be associated with psychological changes during an acute attack. In severe cases, loss of contact from reality (psychosis) has been reported.

ALAD porphyria is caused by mutations in the *ALAD* gene. The disease is inherited as an autosomal recessive disorder. This means that both copies of the *ALAD* gene have a mutation. Recessive genetic disorders occur when an individual inherits two copies of an abnormal gene for the same trait, one from each parent. If an individual inherits one normal gene and one gene for the disease, the person will be a carrier for the disease but usually will not show symptoms. The risk for two carrier parents to both pass the altered gene and have an affected child is 25% with each pregnancy. The risk to have a child who is a carrier like the parents is 50% with each pregnancy. The chance for a child to receive normal genes from both parents is 25%. The risk is the same for males and females. Parents who are close relatives (consanguineous) have a higher chance than unrelated parents to both carry the same abnormal gene, which increases the risk to have children with a recessive genetic disorder. The *ALAD* gene contains instructions for creating the enzyme aminolevulinate dehydratase (ALAD), which is necessary for the production of heme, the part of hemoglobin, which is the oxygen-carrying component of red blood cells. Like AIP, VP and HCP, a number of agents can precipitate attacks, including alcohol, certain drugs, physical and psychological stress, infection, reduced caloric intake, dehydration and estrogen and progesterone.

The onset of ALAD porphyria is usually during infancy or childhood, but late-onset of the disorder has been reported. The treatment of ALAD porphyria is directed toward the specific symptoms that are present in each individual. Because there have been so few cases of ALAD porphyria, aside from avoiding triggering factors, there is only limited information on treatment for the disorder. Two standard treatments for acute porphyrias, Panhematin and glucose, have not been universally effective in treating ADP. Biochemical test results have an increase of 5'-aminolevulinic acid (ALA) in the liver, other tissues, blood plasma, and urine. Urine coproporphyrin and erythrocyte protoporphyrin are increased. DNA testing is the most effective means for diagnosis.

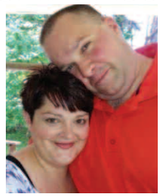
IN MEMORY AND IN HONOR

We send our sincere sympathy to the families and friends who honored their loved ones with their generous gifts to the APF. We join them in thanking you for their donations.

In Memory George J. Rusnak Jr., Deborah A. Hammond, Ariel Lager for **Carol Rusnak,** Gloria R. Sheehan for **Paul Sheehan,** Stephanie Ackerman, Lynn R. Lenhardt, Mary Helen Higgins, Sandra L. Heldt, Jason C.K. Huanio, Leslie A. Salisbury and Barbara E., Steven R. Krikoff for **Holly Salisbury,** Stasia Demichele for **Joe Demichele,** Michael and Carol Farina for **Vincent K. Farina,** Rosemary M. Houlihan for **Branden R. Jackson,** Stephanie E. Rush for **Suzette Frazzini,** Ronald and Norma Brown for **Matthew Brown,** Elizabeth H. Petersen for **Gabrielle Spring Howell,** Joy Campbell, Linda and Bobby Head, David Campbell, Kaye C. Davis for **Norman Campbell,** Bill and Marylou Rickert for **Gina M. Rickert Opperman**, The Pudlicki Family for **Daniel Pudlicki,** Mary Crown for **Mary and Dean,** William Lambert for **Norma K. Lambert**.

In Honor We also thank those who donated in honor of a friend or family member: The Nagin's for **Melissa Nagin,** Patricia Hoover for **Cassandra Hoover,** Steven A. Andrade for **Josephine Dzygala,** Carole F. Gaudette for **Tristen Gaudette,** Linda F. Haynes for **David Milne,** Diana Parrish for **Megan Parrish,** Jennifer R. Ewing, Lisa M. Kancsar, Sharon I. Koch for **Marty Krovetz,** Robert G. Goddu for **Jay Goddu,** Michael Leppert for **Nicole and Craig Leppert,** Dr. Bonnie L. Katz and Dr. Carl L. Tishler for **Dr. Peter Tishler,** Florence Kirshoff for **Ruth Taffet,** David W. Shepherd for **Margaret Shepherd,** Elaine Smuczynski for **Smuczynski Family,** Mary Crown for **Mary and Victoria,** JoLynn Foldesi, Erica Gray, Arlene De La Mora, Sarah and Larry Pritchard, Bill and Nancy Gray, Ruth Wilson, Grayfred Gray, Lori Hanson, Paula Hendrix, Robert Hendrix, Myrna and Donald Cartledge, Mary Frances Donnelly, Sara Elaine and Douglas J. Collier for **Ralph M. Gray,** Connie Helleson for **Jennifer Streeter,** Dale and Renate Moczynski for **Alicia Moczynski,** Michael Pagano for **Andrea M. Pagano Reyes,** David Russell for **Craig and Nicole Leppert,** Cheryl Harriman for **Jeffery Pradovic,** Sharon Dill for **Desiree, Jessica and Amy,** Fred Cerkoney for **Fred, Stephanie and Jason.**

SAN DIEGO PATIENT MEETING



Although Amy Chapman (pictured with husband, Craig) lives in Florida, she hosted the Patient Education Meeting in San Diego recently. Amy has hosted a number of such meetings around the country. Dr. John Phillips made the educational presentation on all of the porphyrias, as well as conducted the *Question and Answer* session of the meeting. Dr. Phillips also has spoken at several patient meeting around the country. These meetings provide YOU with the opportunity to meet other people with different types of porphyria while learning about the disease from an expert. The agenda most often includes a *Question and Answer* session. This is one of the most important parts of the meeting, because most patients have never had the chance to meet a "real" expert who not only knows the answers but who also is one of the researchers who discovered the answers and is continuing to find treatments for us and ultimately a CURE. *Thank you Amy and Dr. Phillips!!!*

We hope to have a meeting each month in 2017, but we can't do it without YOU. Our members host these meetings and the APF sends all the supplies and provides the speakers. If you are interested in hosting a meeting in your area, please contact the APF at 713.266.9617 or porphyrus@porphyriafoundation.com.

Over the past few years our members held meetings or fundraising events in almost every state, including Florida, California, New York, Maryland, Texas, Alabama, Missouri, Illinois, North Carolina, Massachusetts, Washington, Oklahoma, Tennessee, Nevada and Colorado. Join families like the Jacobucci family (photo left) who hosted a meeting in Denver. It is one of the most enjoyable meetings you will ever attend and you can learn about porphyria, too. It is also not difficult to host a meeting. The APF helps a lot. We send out a nice invitation to all the patients who live near you. We also send the pamphlets, the DVD and arrange for an expert either to be there or to make a presentation via conferencing into the home or venue. Contact Edrin at the APF today!

ROBERT DOYLE

The first time I got overexposed was on a trip to Florida at age five. My reaction was so severe that my knuckles broke open because they had swelled so badly. The ER doctors said I had "sun poisoning." I went to many doctors of different specialties, including allergists. They all said I was allergic to the sun, and I would outgrow it. The doctor ordered a test and almost 20 years ago, at the age of 38, I finally received a diagnosis of EPP. It took me 33 years to get a diagnosis.

At the age of 54, I noticed that I was getting weaker and weaker. I coughed a lot and had pain in my chest. They ran a blood test and said, "No, you've had a major coronary!" On Thursday, November 10, 2016, my surgeon told me I didn't have a major coronary, but my aortic valve was only functioning at 10% of the normal capacity, and my heart was so overworked trying to push so much blood through such a small opening that my blood levels showed that I had had a major coronary. He then told me that I must have surgery now, or I will die, plain and simple.

While completing pre-operation procedures, I finally got the attention of the anesthesiologist, who decided to have a little chat with me. He informed me that he thought I had AIP and that the operating rooms would not be a problem, but because I have EPP, the surgery needed to be postponed in order to get the appropriate filters for the lights in the operating room. This hospital, which is one of the major hospitals in the county, did not have the appropriate filters for the lights on hand, so they had to be ordered.

My main goal in telling my story is to inspire others with any of the porphyrias to make sure that their hospital has the necessary tools, specifically filters for the operating room lights, available for them in case of an emergency. Speaking with the anesthesiologist a week later, he informed me that if I hadn't alarmed his attention, that I could be dead! In short, the operating room lights could have damaged my heart. What if I had needed an emergency operation done? What if my heart couldn't continue to beat, because it was so debilitated? Please, please talk to your doctor! Have him/her check to see if these filters are available and are at your local hospital, now; YOU just might need them! You never know what is going to happen.

Visit the Member Stories section of the APF Website to read the full version!

JESSICA IS OFF TO MEDICAL SCHOOL

If you have had any dealings with the APF, you will most assuredly have met Jessica Hungate, Patient Services Director. We are sad Jessica is leaving but overjoyed that she has been accepted into medical school in El Paso, Texas. Jessica says that her experience at the APF was key to her acceptance. She also expressed that she is grateful for all the patients have taught her about being a good doctor and for having the experience of meeting and working with the greatest porphyria experts in the world. Desiree Lyon, Executive Director, said, "There are not enough superlatives to describe Jessica, nor enough words of sadness to say how she will be missed, nor enough blessings to heap on her for her goodness." *Congratulations!!!!*

PROTECT THE FUTURE

We are very proud to have Dr. Bruce Wang as one of our *Protect the Future* (PTF) doctors. Dr. Wang has been involved in the PTF program since the beginning and is now one of the best experts in the country. He is a brilliant clinician who works with Dr. Montgomery Bissell at the University of California, San Francisco Porphyria Center. Dr. Wang has participated in the many porphyria research projects of the Porphyria Consortium and has also participated in their publications in major medical journals and textbook chapters.

He completed his medical education at the University of California, San Francisco School of Medicine in 2007. He went on to complete his Residency here in 2009, and his Fellowship in 2013. Dr. Wang is a valued member of the *Protect the Future* program and we look forward to working with him for many years to come.

Some of his porphyria publications include:

- Gou EW, Balwani M, Bissell DM, Bloomer JR, Bonkovsky HL, Desnick RJ, Naik H, Phillips JD, Singal AK, Wang B, Keel S, Anderson KE. Pitfalls in Erythrocyte Protoporphyrin Measurement for Diagnosis and Monitoring of Protoporphyrias. Clin Chem. 2015 Dec; 61(12):1453-6.
- Wang B, Zhao L, Fish M, Logan CY, Nusse R. Self-renewing diploid Axin2(+) cells fuel homeostatic renewal of the liver. Nature. 2015 Aug 13; 524(7564):180-5.
- Bissell DM, Wang B. Acute Hepatic Porphyria. J Clin Transl Hepatol. 2015 Mar; 3(1):17-26.

For those of you who are new and don't know about our PTF program, it is our most important program. Without porphyria experts and their expertise in this group of diseases, expertise would be lost as each of the experts approach retirement. Patients and doctors would remain in the dark about the porphyrias. Therefore, we train young physicians as the next generation of porphyria experts. The training takes place at Porphyria Centers under the guidance of the present Porphyria Consortium of experts. The funding to train these young experts, like Dr. Wang, comes from you, the people whose lives are affected now and in the future.

Contact the APF to make your donation today!

(Pictured above is the Wang family, Tina, Bruce, Teddy and Andy.)

ALNYLAM

invited Amy Chapman, Rose Jeans and Colin McEwen to present their experiences with acute porphyria to their entire company so that the employees better understand the porphyrias. It is important for people to know why they are working hard to develop a safe drug for a group of patients. These three people did a brilliant job sharing their experience with porphyria at a companywide meeting. While Amy Chapman flew from Boston to San Diego the next day to facilitate a Patient Education Meeting, Rose and Colin filmed an outstanding video on their porphyria experience.

To understand the treatment that Alnylam is developing for the acute porphyrias, see:
https://www.youtube.com/watch?v=Z6QmBG9vm9A&feature=youtu.be

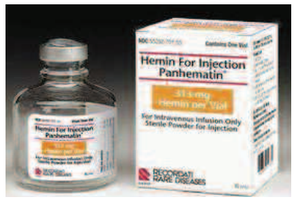
COLOMBIAN FOUNDATION for PORPHYRIA

The Colombian Foundation for Porphyria is one of many groups around the world that the APF has helped become a thriving organization. We thank Dr. Andrade for her message from our Colombian friends.

I am Dr. Marceliana Avila Andrade. I am a medical specialist in physical medicine and rehabilitation, and also a patient with Acute Intermittent Porphyria. In addition, I serve as the President of the Colombian Foundation for Porphyria. Our most recent meeting was attended by 13 patients from Bogota. In Colombia we have about 250 patients in the database of the foundation from different cities of the country. Next year we plan to resume meetings with doctors and specialists to heighten awareness of the porphyrias, as well as patient meetings. We will carry out the genetic study of the patients of the foundation to determine the type of porphyria and to look for the passive carriers in each family member besides to identify if there are mutations common in our population. Our new website page is under construction but the address is www.funcolpor.org.

Best regards,
Dr. Marceliana Avila Andrade, President

PANHEMATIN®



To date, there are **6000-7000** rare diseases that have been identified, however, there are only **400** plus drugs to treat rare diseases. Unlike most rare diseases, we have a treatment for the acute porphyrias. Panhematin was the first Orphan Drug and has been saving lives now since 1983 when the Orphan Drug Act was signed into law by President Ronald Reagan. Without Panhematin, many people with acute porphyrias would not be alive today. At the APF, we receive many questions about Panhematin from newly diagnosed patients. One question often asked is when to start the treatment. Although patients improve with treatment, an attack can still be life threatening. Respiratory insufficiency may require use of an artificial respirator. Therefore, experts agree that porphyria attacks should be treated as early and effectively as possible.

Dr. Lisa Kehrberg *"Very soon after diagnosis, a PICC line was placed and treatment with Panhematin began. I felt so much better after only a couple of infusions. I was close to death by the time I was diagnosed and certainly would not be alive today had I not met my internist and had I not received Panhematin timely."*

Charles Johnson *"... the frequency of the attacks rendered me unable to keep a schedule and the pain left me unable to work. Now that I am on Panhematin every two weeks and the attacks have been under control, I am hopeful that I can return to work soon."*

Cheryl Black-Blair *"The procedure itself is similar to IV glucose treatment—and no more uncomfortable. I chatted on the phone through my Panhematin treatment, and by the time I fell asleep I felt a definite lessening of the pain."*

Amy Chapman *"I was a new person. Panhematin works so well for me!"*

Desiree Lyon *"Once a month, I would have a critical attack which put me in the ICU. Finally, I was treated with hemin therapy, which later became Panhematin. Panhematin saved my life over and over."*

Judy Snyder *"I have not had to be hospitalized for an attack in a long time! The Panhematin treatment works for me. I don't believe I would be here today if it weren't for this drug."*

Steve Stevens *"My hematologist suggested that I have a regimen of monthly Panhematin treatments. I am 55 and have learned not to take life for granted. I know there is no cure for VP, but I'm not letting it dictate my life."*

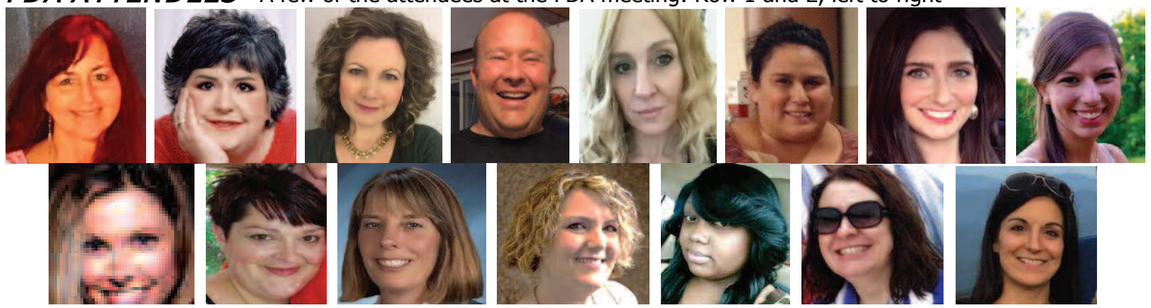
Amanda Boston *"I had the most amazing privilege of participating in the '7203: A double-blind, randomized, placebo-controlled, parallel group trial on the efficacy and safety of Panhematin™ in the treatment of acute attacks of porphyria' research study."* Editor's note: Panhematin saved Amanda's life when she was in a coma and paralyzed.

APF WELCOMES EDRIN WILLIAMS

After completing his Master of Health Services Administration, Edrin Williams relocated to Houston, TX from Ridgeland, MS to take on the position Director of Development for the APF. Aside from assisting with the daily operations of the APF office, Edrin will also head the APF fundraising projects and new educational programs. Edrin represented the APF and all porphyria patients at Rare Disease Week in Washington, DC and facilitated APF involvement in legislation that affects rare diseases. As a former healthcare provider, Edrin is highly experienced with patients and the impact a patient organization can make in a patient's life and on their hope for the future. During his spare time, Edrin enjoys travel and spending time with family and friends. When you contact the APF, give him a hearty welcome.

FDA ATTENDEES

A few of the attendees at the FDA meeting: Row 1 and 2, left to right



1. Louise Braun, Terri Witter, Lisa Kehrberg, Michael Boone, Tara Cantley, Evelyn Jacobucci, Lina Rebeiz, Candace Johnson
2. Ariel Lager, Amy Chapman, Sharon Dill, Mary Schloetter, Lakeshia Johnson, Rose Jeans, Danielle Frazzini Tatarka

WHY SO MANY GIRLS? ACUTE PORPHYRIAS ARE 50/50 BUT WOMEN GET MORE ATTACKS DUE TO THE MENSES.

JESSI BRILL

lives in Northern Virginia. I was diagnosed with EPP about 5 years ago, but my first episode was when I was 18 months old. As, I'm sure, many of you can relate, I ran the gauntlet growing up trying to discover what was wrong with me. After being told that it was anything from having inactive sweat glands that caused swelling to it was all in my head. I stopped going to see doctors until I was 25 when I found the APF. It was after that when my parents were watching "Mystery Diagnosis" and saw the preview of the next episode of the little boy who was having reactions to the sun. They looked at each other after only watching the preview and said "That's Jess".

After making the 7 hr drive to Charlotte, NC, I met with Dr. Bonkovsky who tested me and all the years of no answers narrowed down to a 1 hr meeting with him telling me that I'm not crazy and there is something named EPP that was causing this pain and episodes. I only have my reactions in the warmer months. I have never had a reaction during the winter. And although I love the winter, I have never been skiing or snowboarding in my life so no extended trips outside. Dr. Bonkovsky thinks that this is the reason I've never had a reaction during the winter.

I have a 1 year old little girl named Lexi. I pray she doesn't have to go through what I did my entire life. I remember all too well what it was like growing up being the kid who couldn't go out and play. I don't want that for her even though what I know now would make things completely different for her, still.

We went on our first family vacation this past summer and, of course, it was to the beach. I lasted 2 days with limited time in the sun before I couldn't do it anymore. The rest of my family was there so they took her out for me but it doesn't stop the emotions or tears from getting overwhelming when you know you can't do stuff with your child because of EPP.

Editor's Note: EPP is a family disease. Everyone in the family must change their lives when one person has EPP. Clinuvel has developed a thrilling new treatment that enables EPP to be in the light. It is a revolutionary treatment but there is no FDA approval yet. Please continue to approach the FDA to approve Afamelanotide treatment immediately.

RARE DISEASE DAY

On 28 February 2017**,** the tenth edition of Rare Disease Day saw thousands of people from all over the world came together to advocate for more research on rare diseases. Over the last few decades, funds dedicated to rare disease research have increased. But it can't stop there. Rare Disease Day 2017 provided an opportunity to call upon researchers, universities, students, companies, policy makers and clinicians to conduct more research and to make them aware of the importance of research for the rare disease community. The APF participated in Rare Disease Day. Edrin Williams represented the APF during the Capitol Hill project to visit key Congressmen's offices to request greater funding for rare disease research.

Patient involvement in research has resulted in more research, which is better targeted to the needs of patients. Patients no longer solely reap the benefits of research; they are empowered and valued partners from the beginning to the end of the research process. Patients:

- **Advocate for research** on a specific disease or across diseases. They know where research is needed and work to influence research bodies and companies to prioritize these areas in their research.
- **Fund research.** Individuals or patient organizations, such as the APF, raise money for clinical trials or research projects, on their own or in partnership with private funding initiatives.
- **Partner in research projects** and are included in the governance of research.
- **Participate as subjects in clinical trials** and also in the design of clinical trials. They, therefore, help to ensure that the development of a medicine takes into account their real needs, so that the patient perspective is not overlooked.

DON'T FORGET TO DONATE

THE APF MAINTAINS EXTENSIVE PATIENT AND PHYSICIAN EDUCATION PROGRAMS WITH COMPREHENSIVE PUBLICATIONS AND CURRICULA, AS WELL AS WEEKLY ENEWS, QUARTERLY NEWSLETTERS, SUPPORT SYSTEMS, SOCIAL MEDIA NETWORKS, MAJOR MEDIA CAMPAIGNS, RESEARCH PROJECTS, BROCHURES FOR EACH PORPHYRIA, APF PRODUCTS AND MANY OTHER SERVICES TO ASSIST PATIENTS, FAMILIES, CARETAKERS AND DOCTORS.

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