

**APF HOSTS PATIENT VOICE MEETING WITH FDA** On March 1, 2017, the American Porphyria

Foundation (APF) hosted a Patient-Focused Drug Development (PFDD) public meeting on the Acute Hepatic Porphyrias. These meetings are a part of the FDA initiative under the fifth authorization of the Prescription Drug User Fee Act to more systematically gather patients' perspectives on their condition and their available therapies. The APF conducted this meeting to enable the FDA to hear face to face from acute porphyria patients, caretakers, and advocates about their experiences. Also, esteemed experts in the field provided an overview of the acute porphyrias, as well as the current and novel therapeutics.

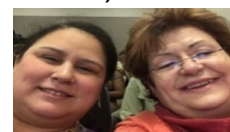
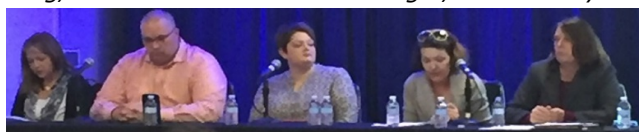
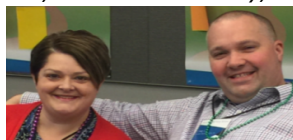
Desiree Lyon Howe, Executive Director of the APF, began the meeting with a hearty welcome to the 100 attendees, including the 40 patients and over 80 people who were attending via the live webcast, industry, FDA representatives and members of the Porphyria Consortium of experts. Next, she introduced Dr. Richard Moscicki, who gave an outstanding presentation on the impact of the Patient Voice in Drug Development.

The following discussions focused on two major features (1) clinical experience and symptoms that are most impactful and (2) patients' perspectives on current treatment and treatment needs. This was accomplished with two panels followed by a facilitated discussion inviting comments from other attendees both in person and via the webcast.

To supplement the input gathered at the meeting, patients and others were encouraged to submit comments on social media and to the APF via telephone and email. Most comments centered on these topics: pain, misdiagnosis, need for treatment to prevent attacks and desire to participate in research. The patient input generated through this Patient-Focused Drug Development meeting and public docket strengthens FDA's understanding of the burden of acute porphyrias and treatments currently used to manage its symptoms. FDA staff will consider carefully this input as it fulfills its role in the drug development process; including advising sponsors on their drug development programs and assessing benefit-risk for products under review for marketing approval. This report may be useful to drug developers as they explore potential areas of unmet need for porphyria attacks and porphyria pain. It may point to the potential need for development and qualification of new outcome measures in clinical trials.

The entire meeting webcast can be viewed in its entirety on the homepage of the APF. Watch the video here: www.porphyrifoundation.org or <https://www.youtube.com/watch?v=urHxVYVAals>. More information on the PFDD initiative can be found at: <http://www.fda.gov/ForIndustry/UserFees/PrescriptionDrugUserFee/ucm326192.htm>.

Row 1: Amy and Craig Chapman, Panel 1 to r Cheryl Martinez, Craig and Amy Chapman, Mary Schloetter, Sharon Dill, Evelyn Jacobucci and Terri Witter. Row 2: Lakeshia Johnson and Diana Sabella, Panel 1 to r Colin McEwen, Michael Boone, Terri Witter, Heather McKinstry, Lisa Kehrberg, Candace Johnson and Ariel Lager, Tara Cantley and Nichol Kirby.



RICHARD A. MOSCICKI, M.D. joined the U.S. Food and Drug Administration's (FDA) Center for Drug Evaluation and Research (CDER), as Deputy Center Director for Science Operations. A nationally recognized expert in clinical research and drug development, Dr. Moscicki brings to the position executive direction of Center operations and leadership in overseeing the development, implementation, and direction of CDER's programs. Before joining CDER, Dr. Moscicki served as senior vice president (SVP), Head of Clinical Development, and chief medical officer at Genzyme Corporation. He joined Genzyme in 1992. Over the past two decades, Dr. Moscicki was responsible for worldwide global regulatory and pharmacovigilance matters, as well as all aspects of clinical research and medical affairs.



Dr. Moscicki received his medical degree from Northwestern University Medical School and completed his residency in Internal Medicine, followed by a fellowship at Massachusetts General Hospital (MGH) in immunology and immunopathology. He remained on staff at MGH and on the faculty of Harvard Medical School from 1979 until 2013. He is board certified in internal medicine, diagnostic and laboratory immunology, and allergy and immunology. Dr. Moscicki presented at the APF PFDD meeting. He has been widely published on the importance of the FDA to include patient experiences in drug development. His view is that hearing patients' testimonies allows the FDA to better comprehend and appreciate the burden of disease, the current treatments and the risks tolerance.

NATIONAL PORPHYRIA AWARENESS WEEK, APRIL 22-29 The challenge of living with porphyria starts with how little is known about it among friends, family and the medical community. That is why **National Porphyria Awareness Week** was such a success. It provides **YOU** the opportunity and platform to reach out to your community to enhance porphyria awareness. Below are a few of our members and what they did for NPAW. Keep up the efforts all year long! See many of the other activities here: <http://www.porphyrifoundation.com/news>.

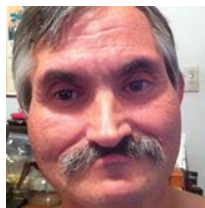
Photos: L to R – Justin Hamilton created a collage, Jake Velazquez hosting a Porphyria Party in Brooklyn, Sean Albright has an APF Racecar for online racing, Suzanne Hagen dyed her hair purple, the Cook Family hosts an annual Barrel Race, APF staff wearing APF gear, Sharon Dill had a TV program on her testimony at FDA, Cassie and Mitchell Tucker designed and sold EPP shirts, Jared Ulmer shared his story with a high school class, and Louise Schlosser educated healthcare professionals at medical centers.



SAVE THE DATE The APF will be hosting a PHYSICIAN EDUCATION MEETING January 12-14, 2018 in Atlanta, Georgia. Physicians from around the country will be attending this prestigious conference. They will have the opportunity to learn from the best because famous experts from the US and Europe will be lecturing on all of the porphyrias. Here is your chance for your doctor to learn more about porphyria and gain the kind of expertise you need for your best healthcare. Please tell your doctor about the meeting and ask them to attend. They will earn CME credits, which are necessary educational credits for your doctor to earn every year.

You can also attend the Patient Sessions of the Conference or register and attend the entire meeting. The Patient Sessions will also be taught by the same experts as the physician conference, but the material will be geared towards laymen. This is an awesome opportunity for patients to learn about your particular type of porphyria. Spread the word. Maybe even host a fundraiser for you and your doctor to attend. *Contact the APF!*

AWARENESS AT ITS HIGHEST Desiree got a call from Eric Lifschitz the day Awareness Week began. He said, I'm making sure my hospital is aware of porphyria. I'm teaching my whole team about porphyria. He did such a good job that when Desiree spoke with the charge nurse at the St. Francis Hospital in Long Island, Cathy Pirolo, she offered one of the greatest compliments a porphyria patient can hear, "Eric Lifschitz teaches the staff here every time he comes in to the hospital. We have learned so much from Eric. He is the expert at living the disease. We appreciate Eric!!!"



Eric also has wonderful things to say about his care team! Plus, Eric takes every opportunity possible to explain AIP and thank the nurses and doctors for the good care he is given. It is a great synergistic relationship that has worked well to assure Eric's good care and better care for other porphyria patients who are admitted to St. Francis Hospital. Thanks Eric for advancing awareness!

Editor's note: *Eric is one of Desiree's oldest porphyria friends. Desiree and Eric met as research patients over 30 years ago at the Rockefeller Hospital in NYC with Dr. Karl Anderson.*

RESEARCH VOLUNTEERS ARE MEDICAL HEROS and RESEARCH VOLUNTEERS ARE NEEDED



Research volunteers deserve the title of "Medical Hero." They are not only volunteering for themselves but for you and your family. Michael Boone and Lakeisha Johnson are now good friends. They both volunteered for the Alynlam Study, the Panhematin Study and the Longitudinal Study at the same research institute, the University of Texas Medical Branch Porphyria Center in Galveston, Texas. There are seven more research centers where you can participate. Please join the others and become a Medical Hero, too. It takes little of your time but makes a big impact on your life. In fact, the Longitudinal Study only takes donating your blood and answering questions one time a year for five years. You may have thought it was far more complex. When you do need to do more, all travel is paid for and that includes flights, hotels, meals and you even get DNA testing free for the clinical studies. All you need to do is contact the APF and they will connect you with the nearest research center. *Call the APF today at 866-APF-3635!*

IMPORTANT NEWS!!! NEW PANHEMATIN



There are thousands of patients who have been and continue to be saved by the acute porphyria treatment, PANHEMATIN®. It is the only pharmaceutical treatment to date for the treatment of Acute Intermittent Porphyria, Hereditary Coproporphyrria and Variegate Porphyria.

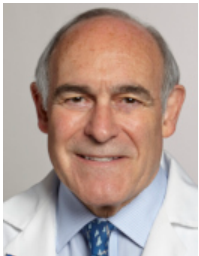
The FDA recently approved Recordati Rare Diseases' (RRD) supplemental Biologics License Application for a new dosage strength of its hemin-based acute porphyria treatment (2016). RRD's new PANHEMATIN® is the first new hemin-based product to receive FDA approval since 1983, when it became the first Orphan Drug. RRD plans to release the new product in July 2017. At launch, RRD plans to enhance its Patient Assistance Program to include a new Co-Pay Assistance Program.

The new PANHEMATIN® 350 mg single vial provides 350 mg dosage strength of hemin, increasing the number of patients potentially eligible for one vial/infusion treatment at recommended dosing. The new PANHEMATIN® 350 mg dosage strength will also benefit from the review of 30+ years of clinical experience since predecessor product launched, resulting in, among other things, significant label changes:

- Elimination of black box warning
- Revised preparation and administration instructions
- Introduction of clinical trials and experience section

The PANHEMATIN® 350 mg dosage strength will be offered at the same price as the predecessor PANHEMATIN® 313 mg product. This is very exciting news. It won't be long till July. Tell your doctors!!!!

ROBERT DESNICK, M.D., PH.D.



Has served on the APF Scientific Advisory Board since the APF was founded. He recently won the prestigious Rare Impact Award, which is NORD's signature event which brings together the entire rare disease community. The Ceremony will be held on Thursday, May 18, 2017 at the Ronald Reagan Building and International Trade Center in Washington, DC. The APF is very proud to have Dr. Desnick as a member of our Scientific Advisory Board for over 30 years. Since he has been a major force in your improved health, we hope you will attend to thank him for his service.

Dr. Desnick deserves this honor. He is a human geneticist whose research accomplishments include significant developments in disease gene discovery, inherited metabolic diseases, and the treatment of genetic diseases, including the development of enzyme replacement therapy for Fabry disease. Dr. Desnick is the Dean for Genetics and Genomics, and Professor and Chairman Emeritus of the Department of Genetics & Genomic Sciences at The Icahn School of Medicine at Mount Sinai in New York City. Additionally, he is Professor of Pediatrics, Professor of Oncological Sciences, and Professor of Obstetrics, Gynecology and Reproductive Science at The Mount Sinai Hospital. He is also the author of more than **600** peer-reviewed articles in scientific journals, **200** book chapters and is the editor of **nine** books. He holds **13** patents and is included in Castle Connolly's lists of Best Doctors in America, Best Doctors in New York and New York Magazine's list of the Best Doctors every year since the inception of the rating. He was elected to the Institute of Medicine in 2004.

Dr. Desnick received his Ph.D. in genetics from the University of Minnesota Graduate School and his M.D. from the University of Minnesota Medical School. He is an elected member of the Society for Pediatric Research, the American Pediatric Society, the American Society for Clinical Investigation, and the Association of American Physicians. Also, he is an elected Fellow of the American Academy for the Advancement of Science and an elected member of the Institute of Medicine of the National Academy of Sciences. His research awards include the E.H. Ahrens, Jr. Award for Research from the Association for Patient-Oriented Research and the Award for Excellence in Clinical Research from the National Center for Research Resources from the National Institutes of Health. Dr. Desnick is a past director of the American Board of Medical Genetics, a Founding Diplomat of the American College of Medical Genetics, a past member of the board of directors of the American College of Medical Genetics Foundation, a founder and past-president of the Association of Professors of Human and Medical Genetics, past chair of the Association of American Medical Colleges (AAMC), past member of the AAMC Board of Directors and past chair of the AAMC Council of Academic Societies.

When asked about his award, he said "It's very satisfying when your research efforts lead to new diagnostics or treatment for patients suffering from rare diseases." *Thank you for your work, Dr. Desnick!*

**RESEARCH IS THE KEY TO YOUR CURE.
VOLUNTEER AND BE PART OF THE TREATMENT AND THE CURE!**

ALE SAYS IT BEST IN POETRY



Ale Lezcano is one of our APF Facebook members who shares a great deal of excellent life tips for those with acute porphyria. She is from Asuncion, Paraguay where there are no porphyria experts. Ale is a very brave woman who struggles without the kind of treatment we have here in the USA. For Ale, it is sometimes difficult to talk about the pain of an acute porphyria attack, because it is not pain people can understand unless they have experienced it.

*I feel the sickening
poison
Moving through*

*me, in me, with me
I feel the horrible pain
Moving to my mouth
Yet I can't not scream
My skin turns into the battle field
My body against my body
I always lose*

*Yet my skin won't let you know
I feel the judging eye
Lays on me
Sometimes harder than the tears
my eyes need to wipe out
Because you can't be like that
You can't stay like that
You can't live like that
But you haven't yet spent one
Hour, one minute*

*one second in my shoes
No, in my skin
You haven't felt the fire
You haven't lost your battled body
to
a battle with your own body
Yet, I still... can't scream*

KRISTEN WHEEDEN



If you have EPP or have been involved with EPP advocacy, you will surely have heard the name, Kristen Wheeden. She has been involved with the APF and our EPP efforts since 2008 when we first approached the FDA about Afamelanotide. Kristen has been an adjunct legislation arm for the APF as she has for years kindly attended the Washington, DC meeting for rare diseases. Kristen also was responsible for the *NBC Dateline* program last year on EPP. That program started a big flurry of 20 other national programs on television. Now Kristen is joining us as our APF Director of Development. Kristen comes with lots of experience. After earning her Bachelor and Master of Business Administration, she became active in health care administration. She and her family live in the DC area, which is perfect for the APF and her development work.

The APF has been in the middle of rare disease legislation since rare diseases came to the forefront over thirty years ago. The result of our involvement was that Panhematin® was approved by the FDA as the first Orphan Drug. The first approval was an awesome undertaking and phenomenal end to lots of hard work. Kristen has much training for her new APF position, not just with her present work, but also in her extra endeavors:

American Porphyria Foundation: EPP Advisory Committee, **Since 2015**

Wyngate Village: Board of Directors (Treasurer), **Since 2013**

Foundations, Inc.: Served on Board of Directors for Psychiatric Adult Day Care Facility, **2010-2013**

Montgomery Hospice: Volunteer, patient visits and bereavement training in high schools, **Since 2009**

Bradley Hills Presbyterian Church: Deacon (Congregational compassion and hospitality outreach), **2013-2016**

Editor's note: *As you can see, Kristen has an advocate's heart. Helping others is in her soul. She is also the mom of three boys one of whom, Brady, has EPP. Kristen's husband, Mike, is also helpful with her advocacy efforts, including porphyria. We welcome Kristen to the APF staff and her family to the big APF family. We are excited that Kristen will help guide the future of the APF and our members.*

PROFILE IN COURAGE



All people with porphyria, automatically deserve the title of a *Profile in Courage*. We will be featuring one of you in each newsletter. One of the most courageous is young Jessica Betteridge, who has spent the better of the last THREE years in the hospital in Melbourne, Australia with critical HCP attacks, yet she continues working on her Ph.D. and her position as the new President of Porphyria Association Inc. (Australia). While extremely ill, she earned her Bachelor of Construction Management and graduated with Honours. In addition to this degree, she recently graduated at the top of her class with the Bachelor of Environments majoring in Architecture also with an additional research honors year at one of the recognized top ten Architecture Schools in the world. She is now working as a Graduate Architect. After another year of experience, as well as

postgraduate training and the successful completion of her board exams, Jessica will become a Registered Australian Architect. In addition to this, she is now doing postgraduate research at the University of Melbourne. Jessica also is fulfilling her goal to bring improved treatment and enhanced education and awareness to Australian patients and the medical community. Trying to handle all of her goals was quite a feat for any normal person... but Jessica is not normal. She is exceptional. She may be very ill, but she is using her skills as an Architect and her passion for making a difference through her own experience as a patient to improve the hospital experience through good architecture. Jessica is a loving, smart, fun, compassionate woman who deserves to be our first *Profile in Courage*.

REMINDERS: The APF has the following items to help improve treatment for Acute Porphyrias

**** Free ACCESS TO CARE TOOLKIT** - This downloadable *Access to Care Toolkit* is a resource designed to help patients living with an Acute Porphyria or their caregivers, loved ones and healthcare providers secure access to Panhematin at their preferred health facility. The Toolkit can be easily accessed and downloaded on the APF Homepage. 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 the Acute Porphyrias and we hope these resources will help you secure access to Panhematin when you need it most.



Effective Management of Acute Intermittent Porphyria: Healthcare Conversation Tracker

Instructions
It is helpful to keep detailed records of your conversations and interactions with doctors, nurses, counselors, and any other healthcare staff. It is also important to keep track of your conversations with insurance providers. This information can support your appeals for access to care and treatment and should be provided with your letter.
Be sure to take notes on all your healthcare visits including the date, time, and outcomes, including any incidents or access to care issues. Also note any phone conversations. Be as specific and accurate as possible. It may help to write down notes immediately after or during the conversation, if possible. This will make it easier to remember. Also note conversations with your insurance provider and include the name of the representative. If you have to leave a telephone message, it is helpful to record the date, time, and person's name.
Use the form below to detail your issues with obtaining access to care and treatment for your condition, or reimbursement. Print multiple copies and record each visit to prove all you make. If you are unable to take notes, ask a relative, friend or caregiver if they can help. Submit a copy of your form along with other documents to support your claims. Remember to be thorough and capture as many details as possible.

The toolkit contains the following materials:
Healthcare Conversation Tracker Record your conversations with your doctors, nurses, insurance providers, etc.
Customizable letter templates Download and insert your personal details to send to hospitals, insurance, etc.
AIP, HCP, VP Access to Care Fact Sheet Reports medical facts and relates why immediate care is necessary.
Patient Bill of Rights Print and insert in the Kit as an easy assessment of your rights as a patient.

**** Free WARNING WALLET CARD** This conveniently sized card fits in your wallet and can be easily identified by Emergency Technicians. It contains the APF website URL, the Safe and Unsafe Drug List URL, and an identifier stating WARNING, THE PATIENT HAS AN ACUTE PORPHYRIA. This simple but bright colored card could save your life. Also having a MedicAlert bracelet is also very important to your health. If you don't have the Wallet Card, contact the APF immediately and ask for the card to be sent to you.



**** Free EMERGENCY ROOM GUIDELINES** cover essential information for the emergency physician treating a patient in an acute porphyria attack, including common precipitating factors, typical presentation and other diagnostic clues, making the initial diagnosis, common sequelae and best practices for treatment. A PDF PowerPoint presentation for instruction is also available. The ER guidelines are written by porphyria experts.

**** Free PRIMARY CARE DOCTOR PACKET** is a collection of articles published in major medical journals and written by renowned porphyria experts focused on the treatment and diagnosis of acute porphyrias and also includes information for EPP. The Kit includes a place for your diagnostic test results. Having your test results with you is of utmost importance lest you be subjected to retesting. Since it took years for some to be diagnosed, going through more diagnostic testing is tiresome and the last thing people want to do again.



**** Free PORPHYRIA LIVE DVD** View world famous experts, patients with all types of porphyrias, family members/caretakers, laboratory staff, researchers, etc., present on the porphyrias. Join the experts in their hospitals and laboratories as they conduct research in their research labs and diagnostic labs. The Mount Sinai DNA lab in New York City is featured as is the Porphyria Laboratory at the University of Texas Medical Center where biochemical testing is done. When this DVD was filmed, Karen Eubanks was in the hospital for treatment. She allowed the crew to watch as she received Panhematin treatment. We meet other patients who share their lives and tell how they cope with a challenging porphyria. Watch patients as they have treatments and their experiences with their different types of porphyria. Enjoy and learn with this free DVD.



**** \$30 EMERGENCY ROOM KIT** This kit contains information that an Emergency Room doctor would need to care for you during an acute porphyria crisis. The official binder is filled with info that helps identify you as a patient with an acute porphyria. It also contains an article on Pain to help emergency room personnel understand the extreme intensity of a porphyria attack and the associated pain. Although sometimes doctors won't even look at the ER kits or packets, most of the time, they are glad to receive helpful information that is not just something people printed from the web.



**** \$30 EPP KIT IS ALSO AVAILABLE**

**** T-SHIRTS (LONG AND SHORT SLEEVE), BOOKS, BANDANAS, BASEBALL CAPS, FRIDGE MAGNETS, WRISTBANDS, PAMPHLETS, KING GEORGE LETTERS, ETC. ARE ALSO AVAILABLE FOR PURCHASE.**
Submit orders to Amy Chapman via email: amy.apf@gmail.com.

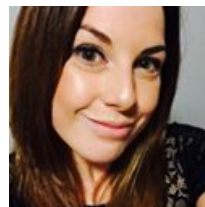
HEATHER MCKINSTRY – LIVER TRANSPLANT



I was diagnosed with AIP at 16 years old. Even with a family history, the doctor refused to test me for Porphyrria. For two years after I was, I had attacks monthly or two times every month. The doctors tried numerous ways to prevent the attacks and eventually we tried weekly infusions of Panhematin®. I received the weekly infusions for over 12 years. Each year my health continued to decline. My quality of life was extremely poor. I had more pain and more attacks, there were fewer good days. My doctors recommended a liver transplant. It was a struggle to find the right transplant team, and submit numerous appeals to my health insurance to approve the surgery. On October 31, 2015 I received a split liver transplant. I had numerous complications following the procedure and the 6 months after. But since then I've gotten progressively better. I'm now a year and half post my transplant with no Porphyrria symptoms, and I'm doing great!

EPP FRIENDS Most people look forward to the warmth and sunshine of summer but not EPP people. They suffer from the light and must keep the sun and the subsequent pain at bay. Read a few of their experiences.

Louise Coomber I had my first symptoms of EPP at 18 months old but didn't get a diagnosis until the age of 31! I



was repeatedly told by doctors through my teenage years to "just stay out of the sun." We just don't have widespread knowledge of EPP within our health system in the UK. I eventually became obsessed with finding out what was wrong with me, so I went to my doctors with a stack of information about EPP, and finally found my answers. It was the most incredible feeling after all those years of feeling different but not knowing why. Also meeting and talking to other people who have EPP is an incredible feeling. I have never had a reaction in the winter, with the exception of skiing in Italy, which I put down to the intensity of the sun at altitude. I'm lucky in the sense that in England we only have 5-6 months of warm enough weather to affect me. I also have a daughter who is 9 and doesn't have EPP, which makes it a struggle in the summer. I suffer guilt when I'm forced to say 'Mummy just can't do it.' We tend to holiday outside of season and take advantage of our beautiful coastline. Shaking the feeling of guilt is a struggle. My family is amazing, and my daughter is really lovely and protective now that she understands my EPP. It gets easier in time, and we try to do loads of outdoor activities outside of the summer months.

Liz Cooke I've had EPP since I was a small child but was only diagnosed last year at 48. It is a relief to find out my diagnosis and find others who have it, too. I have two kids, now 13 and 10 years old, who are surfers. They are all obsessed with the beach, and we live by the sea. I'm in the UK and have no reaction in the winter months, but I am very careful between April and October. My partner takes them to the beach if it's full hot sunshine or in the middle of the day. But we can all go if it's mornings or evenings and I wear gloves, long sleeves, and a hat. Basically I cover up, and avoid 11am-3pm sunshine, which is often best for the kids anyway. I can't really go in the sea in the sunshine (obviously) but can sit on the beach under an umbrella and take a big scarf to cover up. I just never do all day there, but again a couple of hours is often better for kids anyway. Also I love to be out in late afternoons and sunsets. We adjust our timings to suit me, and if they go and I can't, I am just happy that they are having fun. We even go camping to Southern France, which is great as the pine forest next to the beaches gives loads of shade. My kids haven't missed out at all, although I've had some terrible reactions at times that set me back, but we can find loads of ways to share and adapt. Wishing all you EPP people lots of luck and happy days.

Nicole Clarke I was diagnosed at a young age. As a child, I would push myself to do lots of stuff I shouldn't have.



We took lots of holidays on and around the water. I always suffered horrific reactions that would have me climbing the walls in agony. But I would still go back for more the next time. I have a seven year old daughter, and since having her, I have been a lot better at restricting what I do. Mostly, I try to recognize the warning signs of when I need to get in the shade and stay there! I do get angry with myself and frustrated, especially in summer when my daughter wants to do things like go to the beach. We live in New Zealand, where there's water everywhere, and I simply can't go. I am very fortunate to have a family that takes her to do those things and frequently takes her for holidays during the summer months that I cannot handle. Now she's older and understands why I can't do these things, and she is always the best at looking after me if I do happen to have a reaction!

Summer has arrived and we still do not have approval for Afamelanotide/Scenesse in the US. We urge you all to contact your congressmen and write letters to the FDA. Send us your written letters, addressed to Dr. Kendall Marcus, and the APF will do the rest. Join us in demanding this life-changing treatment and make your voice heard! *We will NOT give up!*

DON'T FORGET – 2017 INTERNATIONAL CONGRESS OF PORPHYRINS AND PORPHYRIA IN BORDEAUX, FRANCE

This event takes place at the Bordeaux Palais de la Bourse.



Sunday, June 25th, 2017 is *Patient Day/Congress Day 1*.

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. Register today!

ALNYLAM STUDY



Alnylam presented interim results from their ongoing Phase 1 clinical trial with ALN-AS1, an investigational RNAi therapeutic targeting aminolevulinic acid synthase 1 (ALAS1) for the treatment of acute hepatic porphyrias. New data were from Parts A and B of the Phase 1 study, conducted in asymptomatic "high excretor" (ASHE) subjects. These subjects have a mutation in the porphobilinogen deaminase (PBGD) gene as found in acute intermittent porphyria (AIP), and have elevated levels of aminolevulinic acid (ALA) and porphobilinogen (PBG), the toxic heme synthesis intermediates that mediate porphyria attacks.

Results showed that ALN-AS1 administration resulted in rapid, dose-dependent, and durable silencing of liver ALAS1 mRNA in both Part A (N=20) and Part B (N=8) of the trial. In addition, data showed rapid and dose-dependent lowering of ALA and PBG of up to 95%. Reductions in ALA and PBG were highly durable, with effects lasting for over ten months after a single dose. ALN-AS1 continued to be generally well tolerated in ASHE subjects as of the data transfer date. There were three serious adverse events (SAEs) that were all deemed to be unlikely related to study drug. With the exception of one AE that was severe and unrelated to study drug, all other AEs were mild or moderate in severity, most commonly including abdominal pain, diarrhea, hypoesthesia, nasopharyngitis, pruritis, and rash. There were no clinically significant changes in vital signs, electrocardiograms, clinical laboratory parameters, or physical examination. We look forward to the continued advancement of ALN-AS1, which we believe has the potential to be a transformative therapy for patients with acute hepatic porphyrias suffering from recurrent attacks, a group of ultra-rare orphan diseases with enormous unmet medical need.

See: <http://www.alnylam.com/capella/presentations/interim-results-ongoing-aln-as1-phase-1/>

APF PFDD WHITE PAPER

Read the White Paper detailing the FDA Patient Focused Drug Development meeting on the Acute Porphyrias and the ensuing public comments. Although the video is available to watch the meeting as it occurred, the White Paper also includes public responses and the **Report Overview and Key Themes**. This report summarizes the input provided by patients and patient representatives at the meeting and throughout the webcast. To the extent possible, the terms used in this report to describe specific symptoms, impacts, and treatment experiences reflect words used by the personal testimonies given by the patient attendees. Key themes emerged during the Patient Testimonies: Patients struggled with attacks, which caused severe, debilitating symptoms, including extreme neuropathic pain, nausea, weakness to paralysis, confusion, hyponatremia and fatigue. Over time, these symptoms became chronic in many patients. Those with photosensitivity experienced burning and blistering of the skin. Most patients were limited in obtaining adequate pain relief for their intractable pain.

Read the entire White Paper here:

<http://www.porphyrifoundation.com/sites/default/files/APFPFDDMeetingMarch1%2C2017-TheVoiceofthePatient%20%281%29.pdf>

IN MEMORY AND IN HONOR

Robert Waller, author of the blockbuster book and movie, *Bridges of Madison County*, died recently. Robert was a porphyria sufferer and helped the APF in our efforts to gain FDA approval of Scenese. Our condolences to his family and to all of the families whose memory we honor.



In Memory Donald and Linda Johnson for **Peggy Ann Johnson**; Carol E Kuklewski for **Vince Kuklewski**; Marielaina Phelan, Mary Lawrence, Judith Teague, Ralph Bassignani, Soo D'Agnese, Holly and Bill Wheelan, Donna and Brend Pfeiffer, James Ringer, Nicole Hoefler, Marie Saade, Robert Nahm, Robert Finnis, Jana Korytova, Jo-Ann Ullrich, Deborah Sinnott, Thomas Wilkinson, Ralph and Cynthia Bassignani for **Thomas Dominick Russo**; Daryl B Brezee, and Desiree Lyon Howe for **Marlene Brezee**; Stephanie J Adler for **Melinda M Marcalo**; Fred and Carol Ennis for **Holly Salisbury**; Bill and Mary Rickert for **Gina Marie Opperman**; and Kay C Davis for **Norman Clark Campbell**.



In Honor We also thank those who honored a friend or family with a generous donation to the APF: George J Rusnak, Jr. for **Diana L Sabella**; Sara and Douglas Collier, Paula Hendrix for **Ralph Gray**; Jere C Wise for **Rachel E Wise**; and Yu Pan for **Jason Barrett**.



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What's New at the APF

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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 the ENEWS.

Protect the Future program to train future experts is important. Please consider making a donation to this program. Yours and your children's future health depends on each of us supporting the training of doctors who will know how to treat us and perform research when our present experts retire.

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

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