

**PATIENT EDUCATION MEETING**

Thanks to **Ariel Lager** who hosted the Philadelphia Patient Education Meeting and to porphyria expert, **Dr. Manish Thapar**, who made an outstanding presentation on the porphyrias for the attendees and answered their many questions. Dr. Thapar is a professor at Thomas Jefferson University in Philadelphia where his clinical and research interests are in viral hepatitis, liver cancer and transplant, genetic and metabolic liver disease and the porphyrias.

We are very proud that Dr. Thapar is one of the *Protect the Future* doctors. He was mentored in the porphyrias by expert, Dr. Herbert Bonkovsky.

We also thank Erik Sokolowski, Patient Advocacy and Engagement manager of Alnylam Pharmaceuticals, Inc., who spoke about Alnylam's emerging treatment for the acute porphyrias. People also enjoy the friendships they make and often continue to gather outside of the first meeting. It is also an opportunity to share coping mechanisms and hints that help ease pain, nausea, photosensitivity, etc. Patient Education Meetings also provide an excellent means to connect with others with your type of porphyria and to learn more than the basics about the disease.



The next Patient Education Meeting was held in Indianapolis, Indiana and was hosted by **Nichol Kirby**. This meeting will be followed by a meeting in the Chicago area hosted by Ruth Dee Bruno. A meeting is currently being planned in Denver, Colorado and will be hosted by Evelyn Jacobucci. Evelyn and Dee have been APF supporters for many years and are treasured, caring friends to many APF members. Details will appear on the APF website and in the Enews. If you do not receive the weekly Enews, please contact the APF office and send us your email address.

DESIREE WINS RARE IMPACT AWARD

from NORD, National Organization of Rare Disorders. APF Executive Director, Desiree Lyon Howe, was honored for her work for the past 30 plus years in the rare disease community and making a meaningful change in the lives of people with rare diseases, including her involvement in all legislation that affects people with rare diseases since the 1983 Orphan Drug Act. Speaking of Desiree and other awardees, NORD President Peter Saltonstall, said, "People who make a positive impact on the rare disease community go above and beyond, overcoming certain obstacles, to make meaningful change in the lives of others. It is our honor to recognize their work to help the 1 in 10 Americans—most of whom are children—living with rare diseases."



During the pre-ceremony activities, Desiree participated in a media panel where she expanded on the barriers and unique hurdles rare disease patients face to receive appropriate treatment including insurance difficulties and finding a specialist or primary care doctor to treat their disease. She is committed to breaking down these barriers and ensuring access to affordable treatment for every porphyria patient. She worked for 30 years to strengthen the orphan drug development process. Most

recently, she helped gain two upcoming coveted FDA/Patient Focused Drug Development meetings for Erythropoietic Protoporphyrin and Acute Porphyrias.

Desiree oversees the APF efforts to join in mainstream activities involving Rare Diseases Legislation and encourages all APF members to participate in legislative gatherings like Legislative Conferences and Rare Disease Congressional Caucus Briefings. She also has been involved in efforts to ensure increased funding for The National Institutes of Health (NIH) and the NIH Office of Rare Diseases Research (ORDR), the FDA and Rare Disease Research and Training Incentives.

Aside from her responsibilities as Executive Director and Co-founder of the APF, Desiree says her most gratifying experiences have been communicating with patients and their families and teaching them the value of education about their type of porphyria. Her favorite message to patients is, "Those who know the most, do the best."

BRIGITTA OWEN

I used to live in North Wales for nearly 20 years. In 1996 my daughter and I came back home to Switzerland. I was not feeling well and had some numbness in my legs. I stayed home hoping the attack would pass. My daughter was worried and asked to sleep in my bed. In the morning she couldn't wake me and my breathing was labored. She rang my doctor who had me airlifted to the hospital, as we live in the mountains. My doctor took my daughter Heidi in while I was in Intensive Care. Not long ago we spoke about this. I have terrible guilt about putting her through all that but she said that for her all that was normal. Going to school coming home and mum was in hospital again. The worse thing for her was to see that helicopter take off with mummy inside and not knowing will she come back!! After that attack, I was given a port and hematin. I'm very lucky to have a good doctor who goes beyond his duty. The



APF is also fantastic. All these years carrying on and fighting alone and all of the sudden I can share and be understood ...thank you!

Editor's Note: Thank you for sharing your story, Brigitta!

PORPHYRIAS CONSORTIUM

You may have heard us talk about the Porphyrins Consortium before.



The Porphyrins Consortium is composed of porphyria experts that head the six major research sites, as well as three satellite sites, around the US. The staff in each center includes porphyria physicians, researchers, research coordinators, and technical/laboratory staff. Together with the APF, the Porphyrins Consortium enables a large scale collaborative effort to develop new strategies and methods for diagnosis, treatment, and prevention of illness and disability resulting from these rare disorders. The consortium website provides

information to porphyria patients and their families, healthcare professionals, and other interested parties. The website also provides a list of currently active studies, with a description of each study and contact information for research coordinators. Read more about two key members of the Porphyrins Consortium below. Visit this link to learn more about what the Porphyrins Consortium has to offer! <http://www.rarediseasesnetwork.org/cms/porphyrias/>

Hetanshi Naik, MS, CGC completed her Bachelour of Science at McMaster University in Ontario, Canada with honors. After which, she attended the Mount Sinai School of Medicine and graduated with a Master's degree in Genetic Counseling in May of 2010. Since then she has been working at the Mount Sinai Department of Genetics and Genomic Sciences as a genetic counselor and clinical research coordinator, within a multidisciplinary team, that provides comprehensive care for patients with Lysosomal Storage Disorders and the Porphyrins. She is also the Project Manager of the Porphyrins Consortium and is dedicated to advancing research for the Porphyrins. In her free time she loves to bake, hike, roller blade, and read a good book. Many thanks, Hetanshi!!



MOUNT SINAI
SCHOOL OF
MEDICINE

Makiko Yasuda, MD, PhD I was born and raised in Portland, Oregon until my parents moved to Japan when I was 13 years old. After completing medical school and a residency in Pediatrics in Japan, I joined Dr. Desnick's research team at the Icahn School of Medicine at Mount Sinai, NY for what was intended to be a 2 year training program. More than a decade later, I am still here!! I am now a junior faculty at Mount Sinai performing research on the porphyrias, particularly the acute hepatic porphyrias. My research focuses on delineating the mechanism of the acute attacks and exploring new therapies for the acute porphyrias using mouse models. Outside of the laboratory, I enjoy painting and dancing with my 4 year old, snuggling with my newborn son, those very rare date-nights with my husband, cooking, and running.



MOUNT SINAI
SCHOOL OF
MEDICINE

Below is a list of some of Dr. Yasuda's research publications:

- Preclinical Development of a Subcutaneous ALAS1 RNAi Therapeutic for Treatment of Hepatic Porphyrins Using Circulating RNA Quantification.
- Liver Transplantation for Acute Intermittent Porphyria: Biochemical and Pathologic Studies of the Explanted Liver.
- RNAi-mediated silencing of hepatic Alas1 effectively prevents and treats the induced acute attacks in acute intermittent porphyria mice.
- Loss-of-function ferrochelatase and gain-of-function erythroid-specific 5-aminolevulinate synthase mutations causing erythropoietic protoporphyria and x-linked protoporphyria in North American patients reveal novel mutations and a high prevalence of X-linked protoporphyria.
- AAV8-mediated gene therapy prevents induced biochemical attacks of acute intermittent porphyria and improves neuromotor function.

GORDON CONFERENCES The Gordon Research Conferences (GRC) consist of an informal community of experts that was created by scientists to discuss pre-publication research at the forefront of various fields. The Gordon Research Conferences provide an international forum for the presentation and discussion of frontier research in the biological, chemical, and physical sciences, and their related technologies. The APF is a proud supporter and recently assisted with the attendance of *Protect the Future* doctors Jason Marcero, Ph.D., and Bruce Wang, MD.

DID YOU KNOW? The APF manages Facebook Groups! The open group, Porphyria – American Porphyria Foundation, is open to those with porphyria and their loved ones. Beyond the open group, the APF manages closed groups that are specific to people with each type of porphyria. Facebook members have learned a great deal from each other, particularly on the disease specific "closed groups." Hit "Join Group" today to have direct access to APF staff members, as well as to learn from others who share your experiences. We also have twitter and Blog accounts.



A BETTER ER VISIT



When a relative was in the hospital, APF member, Colin McEwen, wrote an excellent reminder for porphyria people when they visit the Emergency Room:

This evening's visit was an excellent reminder that maintaining cordial, friendly and firm communication with ER staff, no matter how frustrated you are, will always yield the best results. ERs generally work with tests and exams that can be completed and verified quickly and use treatments only to stabilize patients for discharge or more invasive and extensive care.

The Acute Porphyrias are diseases that have crisis like events. Urine PBG and DNA tests take time, meaning we should be mindful that ERs are not facilities to diagnose porphyria. Few hospitals/ERs keep Panhematin on hand, plus porphyria crisis intervention more often requires multiple, consecutive days, therefore not the best porphyria treatment centers either. I think at times we, especially me tonight, forget that ERs are essentially entrances to hospitals not hospitals themselves. The best outcomes you can hope for from an ER in regards to acute porphyria attack is temporary symptom management and either admission for further treatment or discharge.

The best outcome for suspected, non-confirmed porphyria is specimen collection, temporary symptom relief, and follow-up appointment with a specialist or possible hospitalization. My dad was admitted because of altered mental state and could not be released. The doc ordered the PBG because of family history and we were able to explain how helpful it would be to the hematologists and it would help expedite the process.

In a nutshell, porphies and potential porphies should have tempered expectations for what an ER can and/or will do. They are great for many conditions, but they are not porphyria clinics and should not be thought of as such. Our treatment is just as much (if not more) our responsibility as it is our doctors. Work closely with your primary care physician or porphyria specialist. Have a plan for when attacks occur for weekdays/weekends/holidays, during business hours and after business hours. Keep your ER/educational packet up-to-date and when you travel, make arrangements as if you will have an attack while you are out of town. Lastly, if you are seeking a confirmed diagnosis, contact the APF for information, keep hard copies of all your porphyria tests, keep a journal that includes diet, medications, symptoms (date/times), and questions for your doctor, and always prepare for appointments.

Editor's note: Thanks Colin!!!

SOCIAL MEDIA AND PORPHYRIA

Social media, like Facebook, is not the place for reliable porphyria info on diagnosis and treatment. Many, many people have headed down the wrong road from nonsense medical advice from non-medical people. The APF Facebook groups are monitored for misinformation and only allow scientifically supported information to be distributed.



SHARON KOCH



For me it started in the mid 1960's when I was in high school. My first symptom started with confusion. I couldn't remember my class schedule. Then it started with the doctors. I would be "patted on my head," and they would tell my parents and me that this was just being a teenager and there was nothing to worry about. But I got worse. Depression, extreme muscle pain and weakness, low back pain and abdominal pain, and confusion followed. Also, I had no appetite, losing a large amount of weight in just a couple of weeks. This went on and off for years. There were days that my hands and arms shook so badly that I couldn't work. I had to have major surgery and the abdominal pain that started after that became so severe that for the first time my doctor became concerned. Maybe it wasn't all in my head. The medication that I was given made my pain even worse. Needless to say, my family and I were really scared. At one point my muscle weakness was so severe that I couldn't stand up without someone to help me. My doctor tested me for MS, but the tests were negative and I was told that I was exaggerating my symptoms. It's not normal to have these problems, live in pain and be told "not to worry, all the tests are negative".

These symptoms went on for decades. In the 1990s I was going through a really rough time with muscle pain and weakness, low back pain, abdominal pain and depression. I started with something new, nerve pain and numbness in my thighs and mid back, which continues to this day. I told my doctor about all of this and he said he wanted to do one more test. I didn't want another negative result, so I refused. He finally convinced me and I am really glad that he did. About two weeks later, all of my questions were answered. Finally, a positive!!! I have AIP and it only took about 30 years to get an answer. I had no idea what it meant but I was so happy that it was positive. I know that it sounds strange, but for all of you that have had a similar story, you understand!

The one thing that I feel really bad about is that I passed AIP on to one of my children. At least my daughter doesn't have to go through all that I did. We have an answer to her medical problems.

When my patients used to tell me that something was wrong with them but the doctors couldn't find an answer, I would tell them not to give up. They know their body better than any of the doctors. And I'm living proof!!!

Editor's Note: Great story, Sharon! Visit the Member Stories section of the APF website to read the full version!

FINALLY DIAGNOSED!! *By John Rockwell Crandall* One of the most satisfying experiences of my life



was being diagnosed with erythropoietic protoporphyria (EPP) in May 2016. Don't get me wrong - I wasn't excited to have the disease, but at last I had the answer to a 45-year-old mystery. My older brother and I were both told we were "allergic to the sun" since the age of three, but we never knew what caused the itching, swelling and burning pain we experienced whenever we spent too much time in the sun. For as long as I can remember, I have been a shy and reserved person. I'm not sure how much the limitations imposed by my sun disorder affected the development of my personality as a child, but I know I was afraid of other people making fun of me when I wore a wide-brimmed hat, long-sleeved shirts and white cotton gloves to protect myself from the sun. The thought of speaking up and telling people about my condition scared me, so I either took part in required outdoors and suffered a painful reaction, or I kept to myself resulting in social isolation. On the rare occasion when I tried to stand up for myself, I was accused of laziness or trying to ruin others' activities. You would think that after years of getting "sunburned" that I would have learned my lesson about putting my skin in danger, but when I was a senior in college, I began an eleven-month study abroad program in Germany and Austria. Four days into the program, I suffered the worst reaction of my life while making the 20-minute walk back and forth between my dorm room and the language institute where I was studying. Three days later, after realizing my dream of living in a



German-speaking country wasn't going to work out, I was on an airplane headed back home. That was a wake-up call that I would need to make major changes in my lifestyle.

Over the years, I wondered what caused my severe reactions to the sun, but no one was able to give me an answer, not even the dermatologist I saw a couple of times during my high school years. In October 2015, however, I found a link to the ABC News story on EPP. As I discovered more sources about the disease, including the NBC Dateline episode "Out of the Shadows" and the website of the American Porphyria Foundation, I grew more convinced that I had EPP. After six months of weaving my way through a chain of physicians, including my primary care physician, a dermatologist, and a hematologist, I was finally able to meet with porphyria expert, Dr. Angelika Erwin, and her assistant, Allison Schreiber, at the Cleveland Clinic. It was a relief to not have to go to an appointment with a stack of papers about EPP and tell the doctor, "I know this is going to sound weird, but I believe I have a rare genetic metabolic blood disorder called EPP, and here are the tests I'd like you to order."

Making contact with other people with EPP through the American Porphyria Foundation's Facebook groups has truly been a blessing for me. Although I wish that none of us had this terrible disease, it is comforting to know other people who understand what I go through on a daily basis, as well as rewarding to be able to provide support for others with EPP. I look forward to the day when Scenesse® is available as a treatment for adult patients of EPP in the United States, and for testing to begin on children so that they can gain access to this treatment.

Editor's Note: Thanks, John! Be sure to visit the Member Stories section of the APF website to read the full story!

WHY JOIN THE APF? Joining the APF is free. However, we can only hope that each one of you volunteer for research, raise awareness, and ask you to please become a member of the APF today. The suggested annual donation is \$35.00/year. If you are able to save \$2.92 a month, that would total \$35.00/year. If you cannot afford a donation, membership is free. We know it's not easy to live with a rare disease, get involved and get the facts, so we have many programs out there for Porphyria: PTF - *Protect the Future* Doctors, Patient Education, Comprehensive Dr. Kits and Patient Kits, etc. Because of your contributions, these things can keep going. If you appreciate what the APF has done for you, please consider giving back.



Donate today: 1-866-APF-3635!

Notice what a few members have to say about why they make an annual membership donation to the APF!

I am proud to say I donate annual dues to the American Porphyria Foundation. There was a point in time when I couldn't afford to give but membership is free to all. I now try to donate dues on my birthday as a gift to me and for everyone else to enjoy. I was diagnosed before the APF really got started. The first couple of years I spent weeks in hospital and university libraries trying to find information on what I was having to live with. So I know what life is like with and without the support and knowledge of the APF. Much prefer life with it. – Terri Witter

I make sure my funds are set aside each year. The APF has sent many Dr. Packets, invites, newsletters, etc. They keep me informed and up to date with research and developments in Porphyria Disease. – Amy Chapman

My family and I donate yearly to the APF because every dollar makes a difference in funding research and spreading porphyria awareness. We are grateful to the APF for all the wonderful things they do and we know that they will only be able to continue their hard work and efforts with the support of its members. – Megan Parrish & Family

Become an APF member and receive the weekly Enews, brochures, newsletters and DVD.

OFF TO THE FDA Hooray!!! We have been requesting face to face meetings with the FDA and have finally secured two *Patient Focused Drug Development* (PFDD) meetings, one for EPP on October 24, 2016 and the other for the Acute Porphyrias on March 1, 2017. These are very coveted meetings with only a few granted each year. Fortunately, we have been able to gain TWO of these meetings.

What are PFDD meetings and why is it important to attend? PFDD meetings give the 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 the FDA's decisions and oversight both during drug development and review of a marketing application. This initiative has the potential to change product development in fundamental ways.

The difference in the EPP and Acute meetings, other than the type of porphyria, is that the EPP meeting is arranged by the FDA and will be held at the FDA White Oak campus on October 24, 2016 (see below for details on how to register). If you cannot attend in person, you may join via webcast. This is your chance to tell the FDA about EPP and how much a treatment is needed. For those of you who participated in the Afamelanotide trials, please register and let the FDA know about your experience on Afamelanotide.

The Acute Porphyrias PFDD meeting will be attended by FDA representatives, but it is sponsored by the APF and will be held near the FDA on March 1, 2017. Details will follow on the APF website and Enews.

INVITATION TO THE EPP MEETING IN DC Dear EPP persons, family and friends, On behalf of the Food and Drug Administration (FDA), we invite you to an upcoming public workshop on Erythropoietic Protoporphyrin (EPP) to be held October 24, 2016 from 10am-4pm (EDT) at the FDA White Oak Campus in Silver Spring, Maryland. Specific details are outlined. The public workshop is intended to discuss how best to facilitate and expedite the development of safe and effective drug therapies to treat signs and symptoms related to EPP. FDA will provide information for and gain perspective from patients and patient advocacy organizations, health care providers, academic experts, and industry on disease symptoms and its impact on patients' daily life, patient experience with current treatment regimens for EPP, and various aspects of clinical development of products intended to treat EPP.

We're asking you to help make this workshop a success by attending and asking other patients, patient representatives, health care providers, academic experts and industry to participate either in-person or through the live webcast. Note to attend the meeting, you must register with the FDA. To learn more about the workshop and register to attend, please see the websites below: **Registration:** <https://eppscientificworkshop.eventbrite.com>

Workshop website: <http://www.fda.gov/Drugs/NewsEvents/ucm501389.htm>

We look forward to this exciting workshop and hope to see you there. If you have any questions, please feel free to contact us at meghana.chalasani@fda.hhs.gov.

Once you register, you will receive the following notice about your testimony:

Hello, Thank you for your interest in FDA's public workshop on Erythropoietic Protoporphyrin (EPP), and for indicating your interest in being a part of the patient panel. To be considered for the panel, please email us your comments to the discussion questions below. The deadline to submit comments is October 10th, 2016. You will be notified of your panel status at least a week in advance of the meeting. Please let us know if you have any further questions.

Discussion Questions

1. Of all the symptoms that you experience because of your condition, which 1-3 symptoms have the most significant impact on your daily life? (Examples may include itching, burning, pain, scarring, etc.)
2. Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of your condition? (Examples of activities may include daily hygiene, work and school performance, participation in sports or social activities, etc.)
3. How have your condition and its symptoms changed over time?
4. What are you currently doing to manage your condition or its symptoms? (Examples may include prescription medicines, phototherapy, over-the-counter products, and other therapies including non-drug therapies such as limiting exposure to sun, diet modification, etc.)
5. What specific symptoms do your therapies address?
6. How has your treatment regimen changed over time, and why?
7. How well does your current treatment regimen control your condition?
8. Would you define your condition today as being well managed?
9. Assuming there is no complete cure for your condition, what specific things would you look for in an ideal treatment for your condition?
10. What would you consider to be a meaningful improvement in your condition (for example specific symptom improvements) that a treatment could provide?

AMY CHAPMAN & CPAG



On May 6, 2016, I represented the APF at the CPAG (Coalition of Patient Advocacy Groups) Meeting in Bethesda, MD in association with the RDCRN (Rare Disease Clinical Research Network). The Porphyrrias Research Consortium is part of the RDCRN. It was a pleasure to be able to attend this event. The CPAG vision was clear from the start. Working together, patient advocacy groups and researchers can make faster progress toward new treatment options and cures, which can improve the lives of all persons and families that are affected by a rare disease. So it was fitting that the American Porphyria Foundation was there! Attending and presenting were doctors, NIH Directors, FDA representatives and patients just like me. I had a voice but questioned how could I use my voice and presence at this meeting? I am a shy person but curious, so when the meeting started, we all were able to introduce ourselves stating who we were, why we were there and who we represented. From the moment I spoke, I was so happy to be listened to by a group of people who had come together for the same reason ~ Rare Diseases!! What a great feeling! Chuck Mohan introduced the Office of Rare Diseases Research (ORDR). I made a wonderful friend, Rashmi Gopal-Srivastava, MSC, PhD, who is the Program Director of ORDR. Although she spoke on many complex topics, she made them all easy to understand.

Also, I had the opportunity to speak with Callyn A Kirk, MSPH, Health Institute, University of Florida. She gave a wonderful presentation on why all people with a rare disease should sign up for RESEARCH! She said many who have a rare disease are shy and afraid to get involved. But we at the APF need each and every one of you to please sign up for research, gain exposure, stand up for yourself and volunteer a bit of your time so that we can all live and have a better quality of life. Callyn asked, "How hard is it to spend 10 minutes of your life to answer a few questions, to see if you qualify!" The Longitudinal Study with the Porphyrrias Consortium only requires your answering questions about your disease. They need volunteers. Thank each one of you who have participated in research.

Bottom line is WE NEED YOU ALL TO GET INVOLVED! PLEASE ask how you can help: 1-866-APF-3635!

We also learned about grants for rare diseases and how little funding rare diseases really get. We also looked at the plan for the next five years of patient engagement and funding. What does this mean for the Porphyrrias? We shall see. I really enjoyed learning and meeting other patients, experts, the RDCRN, NIH and all the members of CPAG.

RESEARCH VOLUNTEER

Merideth McGinley (2nd from left) *Today was my last day participating as a research patient for the Porphyria Center at Mt. Sinai. For the past year, I have worked with a great group of doctors and genetic counselors as they collect data to move a new treatment onto the next phase. Every six months I would go to the hospital where the research team gives me a physical, collects urine and blood samples and I fill out multiple surveys. Every three months, I overnighted samples and filled out additional surveys. This treatment being researched may provide long term solutions to combat the effects of porphyria, and I played my part in this discovery!!! You can join, too. Just contact the APF!*



THE ORPHAN DRUG ACT



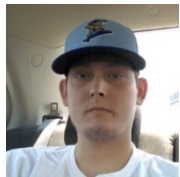
was passed on January 4, 1983 to stimulate the development of drugs and biological products for the treatment of rare diseases. A disease is considered rare or 'orphan' when the prevalence in the United States is less than 200,000 or of low prevalence (less than 5 per 10,000 in the community). The disease is termed 'orphan' because the pharmaceutical industry historically has not had an interest in developing treatments for these small patient populations. Approximately 7,000 rare diseases have been identified. 10-20 million Americans suffer from rare diseases, 50% of which are pediatric patients.

Drug development is a lengthy and expensive process for drug companies, especially those that sell orphan drugs. In fact, drug companies often incur a loss from the development of orphan drugs because of the small patient-population in which orphan drugs are indicated. The Orphan Drug Act allows the federal government to provide incentives to drug manufactures in order to optimize profits from sales of the drug and increase drug manufacturers' willingness and motivation to develop orphan drugs.

Television historian Hal Erickson credits two episodes of the television series Quincy, M.E. for helping the ODA pass in the USA: "Seldom Silent, Never Heard" (1981) and "Give Me Your Weak" (1982). The show's star, Jack Klugman, even testified before Congress concerning the orphan drug issue.

Drug companies nearly universally believe the ODA to be a success. Partly as a result of the 1983 US Orphan Drug Act, Japan adopted it in 1993, as did the European Union in 2000. From the passage of the ODA in 1983 until May 2010, the FDA approved 353 orphan drugs and granted orphan designations to 2,116 compounds. In contrast, prior to 1983, fewer than ten such products came to market. As of 2010, 200 of the roughly 7,000 official orphan diseases have become treatable.

SEAN ALBRIGHT



Sean has a big passion for automobile racing. Sean has Variegate Porphyria, which is one of the Acute Porphyrias with photosensitive symptoms, and has combined his love of racing with VP. Sean has entered into the world of iRacing, an online racing game that is set up to simulate all aspects of Professional Racing. Sean is using his passion to help create more awareness by teaming up with the APF and displaying them on his cars. We are very excited for this opportunity to reach more people about Porphyria and to cheer on one of our own members. Here is Sean's story:



I started having symptoms, such as chest pain, stomach, nausea, and skin issues, when I was young. I was severely hindered because I couldn't do the things normal kids my age did. I started going to different doctors and hospitals to get some sort of diagnosis or help in any form in Kissimmee, FL. Everyone at the ER would tell me it was all in my head and send me home. Around the age of 16, I got significantly worse and was only able to leave the house for school. I moved to West Melbourne, FL at age 16 and found Dr. Lisa Duhaime. She ran tests and kept a standing order



for blood work during times when my symptoms occurred and discovered I had porphyria. At first there was confusion as to whether my skin symptoms were caused by eczema or Porphyria Cutanea Tarda. I was getting treatments every 9 months or so, by this point. I traveled to Mayo Clinic and learned my skin issues were related to VP. My treatments increased in frequency, but I still tried to do normal things like ice skating, playing and watching baseball, etc. I missed over 100 days of school during my senior year of high school, but still graduated with honors. Now I

go for treatment of Panhematin, D5W, or steroids, but the staff is wonderful at Dr. Duhaime's office. They do a great job of keeping me out of the hospital and will fit me in anytime I need it.

I like to go to hockey games, baseball games, and pro-wrestling matches. I always go camping in January for the 24 hours of Daytona event. It's one of the few times each year that I feel 100% normal. I enjoy iracing and have three championships in four years of racing. I like spreading porphyria awareness any way I can through my iracing and frequently tout the APF on forums and other social media outlets. It is hard to see people have porphyria and go undiagnosed for so long. I meet so many doctors that have never even heard of it. I didn't have anyone to talk to until I found the APF. It has helped me feel not so alone. My coping tools, such as racing, are great because during those four hours, nothing else matters besides the car and the track. My best friend/co-owner of my virtual race team is extremely supportive. A support system is one of the most important things. It's helpful to have others to relate to. Without this, it's much harder. Now I try to keep busy and go in for my treatments when needed.



Editor's Note: Thank you for supporting the APF, Sean! Good luck!

AUSTRALIAN PORPHYRIA ASSOCIATION VISITS THE APF



I am Jessica Betteridge, the newly appointed President of the Porphyria Association (Australia). For the past two weeks, I have had the absolute pleasure of working with the American Porphyria Foundation. As our Australian Association is going through a period of change, the insight that the APF has given me will be invaluable for the future. I have really enjoyed seeing the day to day activity and the excellent work that the whole team is doing and the way in which they are an amazing asset to the porphyria community here in the United States, as well as internationally. It was really good to meet with a few patients in the US and hear some of their stories and share my own. A highlight was travelling to UTMB in Galveston to meet with Dr. Karl Anderson and hear all about the groundbreaking, important research that is being conducted there. I would like to thank the whole APF team and in particular Desiree Lyon for accommodating me. It has been a really beneficial trip.

THE UPCOMING PATIENT MEETINGS WILL BE HELD IN SANTA ROSA BEACH, CHICAGO, DENVER, AND MORE!

IN MEMORY AND HONOR

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



In Memory Thank you for choosing to honor a life by making a gift to the APF to help others with porphyria: Marlene Brezee for **Truman Terpenning**; Donald Johnson for **Peggy Johnson**; The McEnroe Family for **Mary McEnroe**; The Fedele, Heflin and Howe families for **Spring Howell**.

In Honor We also thank those who donated in honor of a friend or family member: Sharon Koch for **Matt** and **Wendi Koch**; Elaine E. Sasso for **Elizabeth Britton**.



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The APF does not give medical advice or engage in the practice of medicine. The APF under no circumstances recommends particular treatments for specific individuals, and in all cases recommends that you consult your physician or local treatment center before pursuing any course of treatment.

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What's New at the APF
www.porphyrifoundation.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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