



AMERICAN PORPHYRIA FOUNDATION

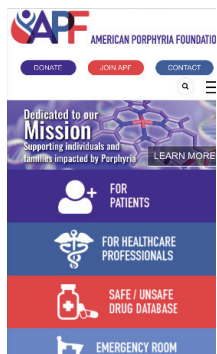
4th Quarter, 2018



*It is our great joy this holiday season to say thank you to you, our wonderful members, and to wish you the very best for 2019. We hope you enjoy a wonderful holiday season and a new year filled with peace and happiness!*

## 2019 PET CALENDAR IS NOW AVAILABLE

The pet calendar contest has been a wonderful way to showcase our beloved pets and have fun with our porphyria community! Dozens of photos of adorable and cherished pets were submitted – and the votes came in for the top featured calendar slots – though each is a winner in our book (and all are included in the calendar!) because they provide the unconditional love and support that you need every day. We are pleased to honor them in the 2019 Pet Calendars, [available for purchase now!](#) Please contact the APF on [866.APF.3635](tel:866.APF.3635) to order.



## WEBSITE LAUNCH AND UPDATE

We are excited to announce that we have launched an updated APF website. It is optimized to offer the patient community and healthcare professionals easy access to critical content – written by renowned porphyria experts- on all the porphyrias. You can access the website from any browser and on any device at [www.porphyrifoundation.org](http://www.porphyrifoundation.org). Stay tuned for updated content, which will be enhanced to reflect the most recent

internationally available information. If you have any suggestions that you would like to see on the site – please submit ideas to [porphyrus@porphyrifoundation.com](mailto:porphyrus@porphyrifoundation.com). Thank you to our members, to our industry partners, and to our brilliant physicians for making this exciting update possible!

## UPDATE FROM ALNYLAM PHARMACEUTICALS



Alnylam Pharmaceuticals announced in mid-October that in consultation with the U.S. Food and Drug Administration (FDA), the company

plans to pursue a full approval based on complete results of the ENVISION Phase 3 study of Givosiran, an investigational RNAi therapeutic targeting aminolevulinic acid synthase 1 (ALAS1) for the treatment of acute hepatic porphyrias, rather than filing based on the interim Phase 3 results. The FDA has also agreed to a rolling submission of a New Drug Application (NDA), which will be initiated in 2018.

*Thank you to all acute porphyria patients who participated in the clinical trials.... you are the medical heroes!*

PHYRIA FOUNDATION



## WELCOME TO THE APF, AUTUMNLEE MEYER – NEW OFFICE MANAGER

Autumnlee joined the APF team in September and has already proven to be a valuable member of our team. Though originally from Wilmington, NC, she has spent the last 4 years in Lynchburg, VA attending Liberty University where she studied Speech Communications. As office manager at the APF, she will assist in the operations of our office in Bethesda, MD by managing special projects, constructing patient and physician packets, as well as answering your

emails and phone calls and much more! Autumnlee is excited to begin this journey of learning more about porphyria and being able to contribute to the foundation's mission. When not at the office she enjoys exploring all that DC has to offer from concerts to the unbelievable restaurants and unique museums.

## EPP CLINICAL TRIAL! PARTICIPANTS NEEDED! DO NOT DELAY!

Be a medical hero. Contact the APF to learn more about being part of this important trial for EPP. **Call 866.APF.3635.**

### PHARMACEUTICAL COMPANY:

Mitsubishi Tanabe

### LENGTH ON TREATMENT:

16 weeks

### TREATMENT:

Oral (pills)

### NUMBER OF APPOINTMENTS:

7-9

### ARRANGEMENTS:

Includes travel/hotel/food

### STIPEND:

Each participant will receive a stipend

### LOCATIONS:

New York City, NY/Winston-Salem, NC/  
Miami, FL/Galveston, TX/Salt Lake City,  
UT/San Francisco, CA

### HOW MANY PARTICIPANTS DO WE NEED:

102 (1 in 4 patients)

### THANK YOU:

To all the medical heroes who are now participating in this trial!

**IF NOT YOU, THEN WHO?**



## 35 YEARS LATER: THE ORPHAN DRUG ACT AND PANHEMATIN®

35 years ago, APF Executive Director, Desiree Lyon Howe helped secure FDA approval for Panhematin®, the first Orphan Drug for an acute hepatic porphyria. In 1983, President Ronald Reagan signed the Orphan Drug Act into law, and the FDA approved Panhematin®. Panhematin® is

the only FDA approved drug therapy for acute porphyria attacks. Recently, Recordati Rare Diseases launched a 350mg dosage strength of Panhematin®. The 350mg dosage strength has been launched to better accommodate the current average weight of patients and the current dosing of Panhematin® in clinical practice. The prescribing instructions have numerous updates including the removal of the boxed warning, addition of clinical studies section, updates to the adverse events section, and changes to dosing and administration. The distribution process for Panhematin® is the same. A patient assistance program was also developed to help patients who experience issues with access to treatment and offers comprehensive assistance for insurance related issues. Please contact the APF office for additional information or visit the Panhematin® website ([www.panhematin.com](http://www.panhematin.com)) for more information.

## PANHEMATIN. ACCESS TO PANHEMATIN® (HEMIN FOR INJECTION)

Many people with acute porphyria are receiving Panhematin® to stop their attacks while others receive it on a weekly basis to prevent attacks. Others are receiving Panhematin® on a weekly or monthly basis to prevent attacks. Panhematin has been an effective treatment for thirty years. However, now in some states, people with Medicare or Medicaid are being denied treatment. The excuses from the hospitals are varied but their validity is questionable. One of the biggest issues is that patients are told that they cannot receive treatment in the hospital and must receive treatment outpatient. This is outrageous as patients are most often extremely ill and need hospitalization and treatment to stop the attack. With the help of Dr. Robert Desnick and Dr. Herbert Bonkovsky, the APF has approached Medicare and Medicaid a number of times to identify the problem to them and solicit a change. We need your help. If you have had any problems receiving treatment, please contact Desiree at the APF at [866.APF.3635](tel:866.APF.3635) or [porphyrus@porphyriafoundation.com](mailto:porphyrus@porphyriafoundation.com).

## JOVANNA WILLIAMS – WHAT ONE WOMAN CAN DO!



“How dare they!” was the first thought of Jovanna Williams, APF member and AIP patient, when she learned that her Medicaid coverage for pain management treatment had been suddenly, and without any prior warning, been dropped. “If they won’t treat me because they know nothing about AIP, they’ll learn really fast!” Jovanna then set off on a 19-day mission to have her opioid pain management treatment approved by Medicaid, her only form of insurance. A vision of her marching down to Governor Parson’s office in the state capital comes to mind when she describes her plan to educate her legislators on her situation and her disease. In reality, she crawled and clawed her way there—stopping four times to gain enough energy, praying to God with every single step that she would make it alive. She made it – and was armed with key evidence. First, a firm letter from her doctor whom Jovanna

had immediately contacted, asking “would you be willing to fight for me and all the people in the state of Missouri?” She also carried impactful APF literature. She was bolstered by her singular goal to be heard. Governor Parson listened and was truly affected by the words on the purple-colored flyer that described porphyria pain as “a thousand flaming swords.” The governor’s office, Representative Vicky Hartzler of the US House of Representatives, Dr. Veronica Anwuri of Saint Luke’s Primary Care in Lee’s Summit MO, the American Porphyria Foundation, and Jovanna worked feverishly together over the next 19 days to educate and advocate for approval of her opioid treatment. Each day without pain medications was pure torture. IT WORKED! Jovanna received back-to-back calls from the pharmacy, from Family Services, from Vicky Hartzler, and – most importantly – from Medicaid, that her appeal had been approved. Her story of persistence and perseverance in the face of overwhelming pain is a lesson in patient advocacy. Jovanna had been diagnosed with AIP fifteen years prior, after undergoing many surgeries and finally whole genome testing to find the cause of her symptoms. Pain management came soon after, and she has been a diligent and active self-advocate and patient advocate since, fostering positive relationships with her physicians. Meeting the APF five years ago was also life-changing for Jovanna. “By finding the APF, I felt validated. It wasn’t only me. Desiree listened to me and took action to protect me in this process.” Her advice? Go big. Find someone in important in your community that can help. Most of all, “don’t sit and complain, do something about it.” She did, and it worked for her and for all porphyria patients in the state of Missouri. “You think I’m done? No way – I’ll do whatever needs to be done so everyone with acute porphyria has access to pain medication!” Go Jovanna!



## THE OPIOID PROBLEM FOR ACUTE PORPHYRIA

Severe, unremitting abdominal pain occurs in 85-95% of cases during acute porphyric attacks. Pain also can occur in arms, legs, shoulders, etc. Experts advise the use of Panhematin® soon after the attack begins to mitigate the pain and other symptoms. Not only is Panhematin® effective in stopping the symptoms, it also stops or modifies the pain. Since acute porphyria are pharmacogenetic diseases, they can be triggered by many drugs. Thus, it is essential to know which drugs are safe and unsafe. Caution when administering any drug is important, because of the unpredictable individual responses to particular medications. Because opiate analgesics have been highly researched, they are listed as safe on the major porphyria drug databases. This is not meant to infer they are safe for people who have individual allergies to opioid drugs. To check on the safety of a drug for acute porphyrias, please see the APF Drug Database on the APF website: [porphyriafoundation.org](http://porphyriafoundation.org).

One huge problem is that opioids are being targeted by government agencies and use of opioids is being restrained. Opioids are on the safe list for the acute porphyrias while many other typical drugs used for pain management are not safe. Therefore, many people are enduring severe pain without access to pain management.

This is not a new movement, rather it has been escalating over the past six years. The APF has been asking patients to contact their legislators to help gain proper pain management since opioids are being denied. The APF has also provided government testimony about this reprehensible situation. Unfortunately, despite their patients' life of pain and their primary care doctors' orders, many pain management physicians and hospitalists are removing opioid medications from acute porphyria patients' regimen leaving no workable options for pain. In fact, recently Medicare and Medicaid patients in Missouri were denied payment of their opioid prescriptions. Working together with the APF, some patients have gained special access by doggedly contacting Congressmen and government agencies until they provide the needed help.

## CALL TO ACTION – ACUTE PORPHYRIA

The APF has instituted a new direction for the pain program by asking for special access pain management. This special access is provided to cancer patients and should also be given to porphyria patients. If you are suffering chronic pain or pain during attacks and have been denied access to pain medication, please send your experience to the APF as soon as possible. Please contact the APF at [866.APF.3635](tel:866.APF.3635) or [edrinw@porphyriafoundation.org](mailto:edrinw@porphyriafoundation.org).

We also ask that you write a letter to your Congressman. Here is a sample script:

Dear Congressman XXX,

Although we understand that the misuse of and addiction to opioids has become a national crisis affecting public health, as well as social and economic welfare, we implore you to protect citizens who live in intractable pain. Many of these people, like those with acute porphyria, are unable to function without the respite that opioids provide.

The Acute Porphyrias, are group of ultra-rare metabolic diseases which are known to be among the most painful in human kind. Furthermore, the acute porphyrias are pharmacogenetic diseases, meaning there exists a genetic inability to properly metabolize many drugs, thus causing dangerous, life threatening reactions. Opioids are the only group of effective pain management drugs that can be prescribed safely to treat porphyria pain without placing patients' lives in severe danger.

Therefore, it is imperative that porphyria patients, who are compliant with their prescribed instructions, be given a special exclusion to receive opioid prescriptions with the direction of their own physicians who understand the porphyrias.

Sincerely, XXX



## PAIN MANAGEMENT INITIATIVE

Why is it that we hear about pain related to porphyria every single day, but we don't talk about it to the medical community and legislators in an official capacity? Decision-makers need to understand porphyria in order to push for access to effective pain management. **FACT:**

Acute porphyria is a pharmacogenetic disease. Patients CANNOT take specific medications. **FACT:** There is an opioid crisis in the US and legislators are limiting access to opioids for pain. **MYTH:** There is nothing that we can do. We need an exclusion for porphyria for pain management in the war on opioids – and we will work together to fight for it. Stay tuned for a plan of action!

## INTER-AGENCY TASK FORCE ON PAIN

The APF is involved with an agency task force to ensure that the porphyria community is represented in decisions about federal pain management practices. We made public comments at the opening session – giving the direct message that porphyria patients require serious opioid pain management treatment that should not be lumped in with addiction issues. The Pain Management Best Practices Inter-Agency Task Force was established to propose updates to best practices and issue recommendations that address gaps or inconsistencies for managing chronic and acute pain. The Task Force will also provide the public with an opportunity to comment on any proposed updates and recommendations and develop a strategy for disseminating information about best practices. The Task Force consists of representatives from relevant HHS agencies, the Departments of Veterans Affairs and Defense and the Office of National Drug Control Policy. Non-federal representatives include individuals representing diverse disciplines and views, as well as experts in areas related to pain management, pain advocacy, addiction, recovery, substance use disorders, mental health, minority health and more.



## PROTECT THE FUTURE

The APF *Protect the Future* program addresses the scarcity of physicians and scientists and lack of expertise about the porphyrias among physicians, patients and the general public. The APF *Protect the Future* program provides training for future porphyria experts by using the present experts in the field to mentor them over a three to five-year period. To accomplish this goal, the American Porphyria Foundation was responsible for bringing together six senior porphyria experts and formed the Porphyria Research Consortium of the NIH Rare Disease Clinical Research Network (RDCRN). This esteemed group of experts are located in six Porphyria Centers across the US. There are several objectives of the PTF program: **OBJECTIVE 1.** Train more physicians and scientists to become qualified as experts in the field of the porphyrias and to be the next generation of porphyria experts and scientists. **OBJECTIVE 2.** Provide information and training to primary care and specialist physicians so they can advance knowledge of appropriate diagnosis and clinical management services to a widespread population of patients with multiple types of porphyria. **OBJECTIVE 3.** Involve PTF doctors in the publications, research, abstracts, posters and similar expectation. **OBJECTIVE 4.** Expand the new satellite clinics and the expertise of the PTF trainees/directors. To date, thirty-two young doctors and researchers have been trained in the porphyrias under this program. Interested in supporting the PTF program? Please contact the APF on [866.APF.3635](tel:866.APF.3635) to donate directly to this critical initiative.

## AMY DICKEY, M.D. – PROTECT THE FUTURE PHYSICIAN



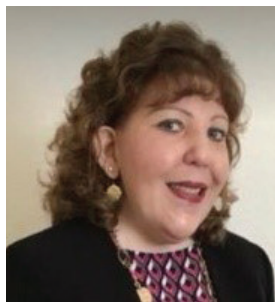
We are thrilled to introduce our new APF *Protect the Future* physician, Dr. Amy Dickey of Massachusetts General Hospital. Dr. Dickey is trained in internal medicine, pulmonary, and critical care. She will be mentored under Porphyria Expert Dr. Karl Anderson, University of Texas Medical Branch along with other mentors and will soon institute a satellite porphyria center at MGH. Dr. Dickey has already had a recent opportunity to educate fellow physicians on the porphyrias. In mid-October, she presented a physician education training session to the internal medicine residents at Massachusetts General Hospital alongside Padmini Pillai, a PhD in biomedical engineering at MIT – who lives with AIP. Please offer Dr. Dickey a warm welcome to the PTF program.

## THE LITTLE IMITATOR

Why is porphyria called, the “little imitator?” This was a term given to the porphyrias by Jan WALDENSTROM in his paper *“The porphyrias as inborn errors of metabolism.”* *Am J Med.* 1957 May;22(5):758–773. Interestingly, syphilis was called the “the great imitator.” The term “the little imitator” stuck and was later used by a number of other experts, including Dr. Herbert Bonkovsky and Dr. Claus Pierach in their publications. One of the reasons for the name is that the clinical presentation can vary widely and has both central nervous symptoms or photosensitivity or both. For example, in porphyria cutanea tarda (PCT) in which the skin changes are quite typical, the acute porphyrias can have cutaneous and neurological symptoms. If severe, immediate laboratory results are important yet not always possible, because tests must be sent to specialized laboratories.

The clinical presentation is challenging, especially with the acute patients (acute intermittent porphyria, variegate porphyria, hereditary coproporphyrin and ADP porphyria), during an attack. Most patients (approximately 85-95%) suffer severe abdominal pain. However, the variety of symptoms thereafter is why the “imitator” description has taken hold. Extreme leg, shoulder blade, neck and arm pain, headaches, seizures, nausea, liver pain, chest pain, and a host of other symptoms can occur during an attack of acute porphyria. This diversity of symptoms causes physicians to order a wide range of medical tests. The “little imitator” tag holds true in that a multiplicity of symptoms leads nowhere until the doctor thinks to test for porphyria. Most of the time, doctors have already performed MRIs, Ultrasounds, CT scans, etc, for gastrointestinal symptoms. A majority of the tests are negative because porphyria only is detected in porphyria specific tests. To add to the confusion, it is important to note that no single test covers all the porphyrias. Each type has their own tests and they must be performed in highly specialized laboratories. Now DNA can detect the mutations/types 99% of the time. So even if you have every symptom in the book and yours imitate every known disease, DNA can ferret out these possibilities and help come to a correct diagnosis.

## JESSICA MELTON – PATIENT HIGHLIGHT



My name is Jessica and I have Acute Intermittent Porphyria (AIP). I was unofficially diagnosed at nineteen years old, and then officially diagnosed via DNA testing at the age of twenty-five. By the time I was nineteen, I had already endured over ten surgeries, including several exploratory surgeries to discover the cause of my unexplained abdominal pain. I was lucky that my hematologist quickly started me on Panhematin. When the treatment was finished, I could not believe it, because my pain was gone for the first time in my life! I am now thirty-five and I still receive hematin on a regular basis. Unfortunately, due to the chronic nature of the disease, damage has been done to my nerves and organs. I was diagnosed with gastroparesis, neuropathic pseudo-obstruction, seizures, and Tourette Syndrome. The icing on the cake is an extremely elevated ferritin level. For two years now, I have been getting weekly inpatient 24-hour intravenous treatments \* to strip the iron from my body. Despite the constant uphill battle with this disease, I was able to graduate with my associate degree in special education and my paralegal certificate. My life is certainly not perfect, but one thing is for sure; if I can overcome all of this, I can do pretty much anything I set my mind to. ***Thank you, Jessica, for your courageous fight and determination to not allow Porphyria to overtake your joy for accomplishing your goals. Congratulations to you!***

## 2018 GLOBAL GENES SUMMIT



Edrin Williams, APF Director of Patient Services attended the 2018 Global Genes RARE Patient Advocacy Summit in Irvine, CA. This year marked a Decade of Hope for the Global Genes Team. As a first-time attendee, Edrin attended a freshman orientation to orient new comers on the packed agenda of the days coming ahead. This meeting brought together over 800 patients, advocates, organization leaders, industry and exhibitors. The sessions ranged in discussions on managing the unknown when there is no diagnosis, current legislative activities, understanding who you are as a rare disease patient/patient advocate, understanding the emotional health of rare disease patients and families, health policy, biomarkers and so much more. Aside from meeting people from all walks of life with a RARE disease, Edrin learned from various speakers about their experiences within the rare disease community. One session that really struck Edrin, was the Keynote address led by Mrs. Rachel Callander. Mrs. Callander spoke about how the language that healthcare providers use can often cause confusion, frustration, anger or hurt. As patients/patient advocates we must be willing to challenge our healthcare providers with language regarding living with a rare disease.

To view these sessions, please look on the Global Genes website at [www.globalgenes.org](http://www.globalgenes.org). **SAVE THE DATE 2019: RARE Patient Advocacy Summit to be held in San Diego, CA September 18 – 20, 2019.** Pictured with Edrin is Nicole Boice, Founder of Global Genes. #2018GGSummit #DecadeOfHope



### ATTENTION SHADOW JUMPERS!!!

Make sure to visit the APF website on December 15th to download the 2019 contest application form. One 2019 youth recipient and his/her family will have the opportunity

to experience a trip to Disney complete with a plan to stay safe from the sun.

## PHYSICIAN EDUCATION AT THE AMERICAN COLLEGE OF EMERGENCY PHYSICIANS (ACEP)

Dr. Mark Korson spoke at ACEP this October in San Diego about metabolic diseases, highlighting porphyria. Metabolic disorders often present to the emergency departments, especially those that are treatable. Porphyria figured prominently in his line-up of diseases to be discussed. To make the presentation lively and engaging, he included video clips from the Recordati video set, that feature Desiree Lyon Howe. She was able to virtually teach the crowd with him! Dr. Korson plans on repeating this discussion at a workshop in Boston this December. Dr. Mark S. Korson is board certified as a Clinical Biochemical Geneticist and has extensive experience in the diagnosis and management of children & adults with a wide array of inborn errors of metabolism, specifically mitochondrial & metabolic disorders.

## EPP FEATURED IN THE JOURNAL OF PEDIATRICS



Dr. Manisha Balwani, porphyria expert at Mt. Sinai in New York City, led the research on this publication that was recently featured in the Journal of Pediatrics. This article is a case report and short survey which shows that the diagnostic delay in Erythropoietic Protoporphyrria is over a decade. The survey, in collaboration with the APF, received a great response in a short period of time. Thank you to all EPP patients and caregivers that submitted information to this valuable research. Hopefully, this prominent article will help EPP be recognized by more pediatricians.

<https://www.sciencedirect.com/science/article/pii/S0022347618307704?dgcid=author>

## THANK YOU TO THE APF!

“For the first time in my life I was hospitalized with EPP on September 10, 2018. The teams at both St. Annes E.D. and OSU Wexner Medical Center in Columbus, Ohio were very obliging to the circumstances. I was only in for 2 days and only really awake for maybe a few hours. They paid very close attention to the fact that I need to be careful with anything that affects the liver and that the lights needed to be on at a minimum. Side note: Being a country girl and a football fan, having a view of the horse shoe stadium from the hospital at night was pretty awesome. Another first was the feeling of validation from the medical teams. The team of hematologists where concerned that I might have had a different form of porphyria than EPP. Before leaving the hospital, they set me up with a hematologist that specializes in porphyria. I have to say, if it wasn't for joining this foundation, this trip to the hospital could've been a lot different. In learning what I have from the APF about something I have lived with my whole life and never grasped, being able to go in there and explain my disease made me feel confident in getting the help I needed. One doctor in the ER ask me if I was formally diagnosed. My reply was “I was three. One of my very first memories as a child is getting my blood drawn for testing.” That is something you don't forget. A huge thank you to the American Porphyria Foundation for existing. It's made a huge impact in my life already and I am sincerely looking forward to the patient meeting September 29th in Cincinnati.” *Mandy Kaiser, EPP*

## NEW JERSEY ELEMENTARY SCHOOL SUPPORTS EPP WITH A WALK-A-THON!



Mary Ventrice, mom to Gia (Age 6), worked with their public school system in New Jersey to arrange a walk-a-thon to support EPP and the APF. The walk took place on Monday, October 8th. The APF sent fact sheets to hand out and the Ventrice family was decked out in EPP gear and Shadow Jumper bracelets. Most important, young Gia was supported by

her family and friends! **NEED SOME FLYERS OR FACT SHEETS FOR AN EVENT? CONTACT THE APF AND WE ARE HAPPY TO ASSIST!**



## WHY BECOME A BONE MARROW DONOR? BONE MARROW TRANSPLANTS ... EXPLAINED



Healthy marrow and blood cells are needed to live. When disease affects marrow so that it cannot function properly, a marrow or cord blood transplant could be the best treatment option, and for some patients, offers

the only potential cure. Bone marrow transplant are being done for erythropoietic protoporphyria. What occurs in a bone marrow transplant is an amazing undertaking. A person with healthy blood forming cells is chosen to be the donor for an EPP person. These cells can be collected from the patient for infusion. If the patient's own cells are given, this is called an autologous transplant. An allogeneic transplant occurs when cells are donated by an adult or from public cord blood. Many times, family members donate the blood for the procedure. Since seventy percent of people do not match the patient, doctors often use other cells that do match outside the family. That is why it is important for all who can to become a bone marrow donor. This enables more people to have a chance at a better life. Once the bone marrow is collected, the patients receive high doses chemotherapy cells into their bloodstream. patients receive the healthy cells like receiving medication through an IV. Once the cells are infused, they begin to grow and make more healthy white and red blood cells and platelets. Some people have few problems with a bone marrow transplant, but others can develop serious complications. *Therefore, a decision to have a bone marrow transplant should not be taken lightly.*

## BONE MARROW TRANSPLANT SURVIVOR SKYLER PARIS

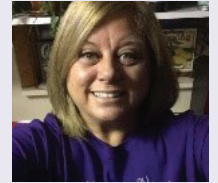
Skyler, who underwent bone marrow transplant in September, continues to recover at Cincinnati Children's Hospital. This mighty warrior is supported by so many who wear their t-shirts with pride and love for Skyler! #teamskyler



Madelyn Havard



Detorie Walker



Ginger Zona



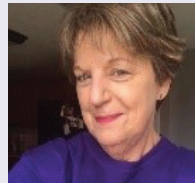
Jennifer Beck & husband



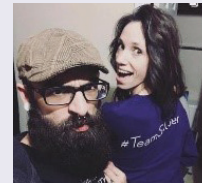
Audrey Schering



Cassie Tucker



Tina Marie Heller

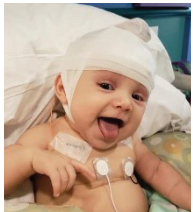


Marc Thompson & Jess



The Herman Family

## THE JOURNEY TO A DIAGNOSIS FOR HARMONI GRACE by Amanda Geer



In October 2017, I found out we were pregnant with our first child and we could not have been more excited. At 38 weeks, our sweet little girl Harmoni Grace was born. A first sign that something was wrong was when the nurse saw what they called a brick dust diaper. We soon learned she was jaundiced, so was sent to the NICU. After two very long days, the doctors could not find anything

wrong with Harmoni's urine. As excited as we were to bring our baby girl home we knew something wasn't right. For two whole months we spent almost every day in Valley Children's Hospital trying to get answers. Every time we took her in for vomiting, the red diaper, lethargy, and not eating, they sent us home saying it was colic. That answer was never good enough for me. As a mother I knew something just wasn't right. Finally, we were admitted due to her hemoglobin being extremely low and a blood transfusion. I can remember that day as a nightmare. On the fifth day the team of doctors came in, sat us down to let us know they might have an answer to all this madness going on with our

sweet girl. They informed us they thought that Harmoni had Porphyria. We met with a hematologist and a genetics doctor and had TONS of testing. We were finally discharged when Harmoni's hemoglobin was at a safe level. We were advised her levels would have to be checked every two weeks to make sure she wouldn't need another transfusion. On September 11th, it was confirmed she has Congenital Erythropoietic Porphyria. Now, we are now working with UCSF to meet with a specialist. We will do everything in our power as her parents to make sure she feels no different than others. At only three months old, Harmoni Grace is the strongest little girl I have ever met. We refuse to let this kick her butt, so we will always make sure she is Porphyria strong.

*Thank you for sharing through a parent's eye how special and wonderful your baby Harmoni is to you. Harmoni you are very special to us and how wonderful it is to have such loving and caring parents fighting for you. To learn more about Congenital Erythropoietic Porphyria (CEP) please view our website [www.porphyrifoundation.org](http://www.porphyrifoundation.org).*



## THE SHADOW RACE 2018

The Cook family has done it again! LeeAnn and family held a successful Shadow Race in Texas on October 27th. The barrel race raised awareness about Porphyria through an amazing community event. We are grateful for your support. Photos are of the winners!



## PATIENT EDUCATION MEETING IN CINCINNATI, OH



The APF held a successful and informative patient education meeting on September 29th at Cincinnati Children's

Hospital. Thank you to Brandy Smith for securing the location and working with the hospital staff to organize the event, and Audrey Schering for working by her side to help promote participation. Dr. Karl Anderson attended virtually to present on the porphyrias and lead a Q&A session. We will have a dozen more meetings across the country within the year! We hope you will come to a session near you. Are you interested in hosting a meeting? Please contact [edrinw@porphyriafoundation.org](mailto:edrinw@porphyriafoundation.org).

## SULFA VS SULFUR Containing Amino Acids and Essential Amino Acids

There is a lot of confusion and misinformation about Sulphur, a naturally occurring element and sulfa (sulfonamides), a constituent of some antibiotics and other medications which are very unsafe. Hopefully, you will see the difference in the two and will not be concerned about Sulphur. Our normal diets contain proteins, which are essential to normal growth and health. Proteins are found in both vegetable and animal sources of foods. The building blocks of proteins are called amino acids. Some of these amino acids, such as methionine and cysteine, contain SULPHUR. Such amino acids are not the same thing as "sulfa drugs." SULPHUR is not contraindicated for patients with acute porphyria. Methionine is one of the nine essential amino acids histidine, isoleucine, leucine, lysine, methionine, phenylalanine, threonine, tryptophan, and valine. If adequate amounts of these nine amino acids are not consumed regularly, deficiencies will develop that can lead to malnutrition and disease. Because humans are unable to make these amino acids, they must ingest them regularly to maintain adequate levels. This will permit our bodies to manufacture the hundreds of thousands of proteins that are essential for good health.

Some people have thought that Sulphur is unsafe for porphyria with some Cytochrome p 450 involvement. The opposite is true. In fact, Sulphur is involved in Cytochrome p 450 enzyme, that detoxifies foods and drugs. During the Cytochrome p 450 process to eliminate toxins from the body, the toxin must be liquified to excrete in kidneys. This step involves the creation of a Sulphur and hydrogen bond. So, Sulphur is an important part of the detoxifying process!

## FOOD FACT There is no convincing clinical or scientific evidence that any particular foods (with the exception of alcoholic beverages, as described above) are capable of triggering or worsening acute porphyric attacks.

There are, however, some foods that have been shown to contain chemical substances that, in large amounts, can up-regulate hepatic ALA synthase 1. Such foods include charcoal-broiled meats, cabbage, and Brussels sprouts. The amounts of such foods that would need to be eaten to produce induction of hepatic ALA synthase 1 have not been carefully studied but are probably far above the amounts that would be eaten as part of reasonable, well balanced diets. None of these foods needs to be avoided completely by persons with acute porphyria, unless they have true allergies to them, which are very uncommon. **Moderation in all things is the best course of action.** There are lists of suggested meal plans for persons with acute porphyria who are of normal weight and with normal daily needs for energy. Please see the Diet and Nutrition section at [www.porphyriafoundation.org](http://www.porphyriafoundation.org).

### The Porphyrias Consortium physicians extend best wishes for a happy and healthy new year!

Dr. Karl Anderson, Dr. Manisha Balwani, Dr. Montgomery Bissell, Dr. Joseph Bloomer, Dr. Herbert Bonkovsky, Dr. Robert Desnick, Dr. John Phillips, Dr. Sioban Keel, Dr. Cynthia Levy, Dr. Charles Parker & Dr. Bruce Wang

### HAVE YOU SEEN ALL THE PORPHYRIA VIDEOS AVAILABLE ON THE APF WEBSITE?

<https://porphyriafoundation.org/patients/media/porphyria-videos>

## IN MEMORY & IN HONOR



We thank the families and friends who honored their loved ones with a generous gift to the APF.

### IN MEMORY:

Davida Hansen for **Richard McArthur**; CarolAnn and Paul Stickler for **Dick Howe**; Diane Levere for **Dr. Richard M. Levere**; Sophie Foucault for **Fredrick Michael Cerkony**; Donna Payton for **Dusty Parker**; Zila Reichman for **Tomy Reichman**; Susan Cerkoney, Michelle Stauff, Karen O'Leary, James Stewart for **Fred Cerkoney**; Christine Baer for **Richard M. Chiles**; Kathleen Giacobbe for **John H. Giacobbe**; Stephanie Rushwin for **Suzette Frazzini**; Anita Nagler for **Astairre Dever**; Desiree Lyon for **Luca Nudo**.

We also thank those who honored a friend or family with a generous donation to the APF.

### IN HONOR:

Anne Johnson for **Candace Johnson**; Rodney and Cynthia McCabe for **Nicholas Guanciale**; Laurie Erwin for **Nicholas Guanciale**; James and Barbara Witter for **Terri Witter**; Christin Steiskal for **Tracy Nudo**; James Peetz for **Desiree Lyon**; Rachel Wise for **Jere and Pauline Wise**; Kathleen Venter for **Patricia Wright**; Carol Gaudette for **Tristen Gaudette**.

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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.com](http://www.porphyrifoundation.com)



**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 our weekly Porphyria Post.

**Our *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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