



Ruth Bruno After being diagnosed with AIP, Ruth entered a new program for adults interested in a nursing career. There she discovered her true calling. Read the full story of how she became a nurse and her harrowing illness on the APF website *Member Stories* section. Ruth writes about her experience, *"It was Friday and very cold in Illinois. I didn't have too much pain, just weakness and nausea. Finally that afternoon I was taken by my husband to the local ER. I remember seeing the triage nurse, but then I went into a coma and respiratory paralysis. I was intubated until the hospitalist felt I was stable and removed the tube. Just 30 minutes later, I went into full paralysis and had an emergency tracheotomy and was put on a respirator for 21 days. I had 3 surgeries that night to repair the damage done to my throat and stop the bleeding from the emergency tracheotomy. I remained comatose from January 16th–March 3rd but, the details are too graphic for the story. When I finally started to awaken, I was in such pain that I wanted to die. Now I was dealing with AIP pain, plus pain from all that had happened prior and 40 lbs. of fluid that had to be removed. Waking from the coma brought more pain, both physical and emotional, because now I could not talk and my eyes which bled from the tear ducts and would not close. I also had a feeding tube, could not write, turn myself, stand or walk. Nurses cared for me around the clock in intensive care, working to prevent bed sores and administer medications, and call the doctor if the need arose.*

Now, like many others with porphyria, I know that doctors treating me would NOT call the "Porphyria Experts" for help. My family rallied and they fought for proper care, drugs etc. and brought me through. My oldest son quit his job and brought me home and became my 24/7 nurse. My husband and youngest son were there to help and my oldest learned how to care for my tracheotomy and puree food, etc. The three men in my life gave me my life back. AIP is not something I would wish on anyone, but it has taught me so much. I have the BEST NURSES EVER, my husband and sons. I practiced on them when I was a student and they studied with me as little children and then years later as big strong men they took over my care. They carried me, taught me to walk again, dressed me, prayed for me, read to me and so much more. My message is, 'Caregivers are nurses.' I loved being a nurse and in my heart I always will be a nurse." Read the entire story on the APF website.

Waiting On The FDA Is Not Easy

It has been two years since the Phase II and Phase III Clinical Trials with Afamelanotide for EPP first began. The trials are completed and now it is time for all the data to be assessed and the FDA and the European counterpart, the EMA, to make a decision. Waiting on these decisions is not the hardest part, but it is sure not easy. APF members have done the miraculous, repeatedly. We registered over 180 patients in both trials in only one month—a feat never achieved previously. Our members made such an impression on the FDA that they convinced the FDA to film a porphyria video and to grant Phase III trials, timely. Almost all of the participants stayed in the trials, even when it was clear they had the placebo. This shows great dedication to the research and to all the future EPP sufferers. The participants infused the FDA with hundreds of letters describing their plight and ten EPP people visited the FDA in person to tell their story. THANK YOU!

Lyon's Share *"Every day the phone rings with a new person in need of help. Generally, they have just been diagnosed and want to hear that they are not alone. Some are very scared. A few words from the APF staff or me gives them hope for the future. I remember being so afraid when I had my first attack of AIP. I was in terrible pain and so sick I was sent on an air ambulance to the NIH in Washington, DC. There I was seen by an expert who gave me Hematin therapy every day. Although I was barely able to walk or talk, I sensed I was improving. Several weeks later, I met two people who also had porphyria. When I was finally well enough to roll myself to the medical library, I would read about porphyria and try to put the information in readable form and share it with the two patients. Our subsequent discussion led to the idea of an APF. Now there are over 5000 members and very comprehensive physician and patient educational programs, as well as our PTF program to train future experts. Plus, we have a great support program for patients and physicians who reach out for help with their patients.*

YOU are the reason the APF has been able to assist the thousands of people who reach out to us each year for help. YOU are the reason the APF has been able to educate thousands of doctors every year. The APF is all about YOU. Now the APF needs your help to continue all these valuable programs and services. We need YOUR donations to help us continue the PTF program and expand our physician and patient education programs, as well as our research efforts. Every one of YOUR donations are tax deductible. One dollar is as important as one thousand, because it comes from YOU! Thanks, Your friend, Desiree Lyon Howe"



Alnylam (al-NIGH-lam) is a biopharmaceutical company developing novel therapeutics based on RNA interference, or RNAi. Alnylam is leading the translation of RNAi as a new class of innovative medicines with a core focus on RNAi

therapeutics toward genetically defined targets for the treatment of serious, life-threatening diseases with limited treatment options for patients and their caregivers. Alnylam is the name of the center star of Orion's belt in the constellation Orion. The star has a luminosity that is 250,000 times greater than the sun, representative of the potential strength that RNAi therapeutics could bring to bear for human health. ALN-AS1 is a subcutaneous RNAi therapeutic targeting aminolevullinate synthase-1 (ALAS-1) in development for the treatment of acute intermittent porphyria (AIP). ALN-AS1 has the potential to be a therapy for the treatment of acute porphyria attacks, as well as a prophylactic approach for the prevention of recurrent attacks. Inhibition of ALAS-1 is known to reduce the accumulation of heme precursors that cause the clinical symptoms of AIP.

In October 2013, William Querbes from Alnylam presented new pre-clinical research at the *Oligonucleotide Therapeutics Society* demonstrating that subcutaneous administration of a GalNAc-siRNA targeting ALAS-1 leads to rapid, dose-dependent, and long-lasting knockdown of ALAS-1 mRNA and complete inhibition of the toxic intermediates that mediate the symptoms and pathology of AIP. Based on their findings, the Alnylam team expects to advance ALN-AS1 into the clinic in 2014. Additionally, they intend to directly commercialize ALN-AS1 around the world. Patient volunteers will be needed for research of their new treatment. If you are interested, contact the APF.



William Querbes



Camp Horizon is a great camp in Millville, Pennsylvania for children with photosensitivity. Dermatologists, who are acutely aware of the need of children with photosensitivity pay for kids to attend the camp through their Dermatology Association. Two of our APF members were selected to attend: Willow Morfka (right) and Alessia Callahan. Willow, who is 11, was diagnosed with Epp in 2012. Prior to that they had been told she had erythromyalgia. She was symptomatic since she was six when she complained about her feet burning. Then all her sun exposed skin began burning with

greater intensity until she couldn't be touched. It was not until Willow's dad discovered a news article about EPP, did he have a clue of her EPP diagnosis. When Willow had her gall bladder removed, she was finally diagnosed with EPP. She was fortunate to attend Camp Horizon where the activities are all for children who are photosensitive. Unlike most camps, the cabins are air conditioned and the activities are all designed around limited light exposure.

Alessia Callahan is 11 and entering 6th grade. She is a free spirited little girl who loves music, plays the violin, field hockey and basketball. Alessia is a good student and a fantastic big sister to Connor, 5, and Bella who is just 1 years old. From early on she had a sun problem. Her dad can remember at 6 months old she cried at the beach. Allergy tests were not productive, so when she was 8 years old, she went to the Children's Hospital of Philadelphia where they said she had Polymorphis Light Eruption. Fortunately, they discovered that my cousin and her granddaughter had EPP and had symptoms identical to Alessia. They then secured an appointment with A.I. Dupont in the Dermatology department where a team of physicians met with them and heard their story. They took pictures and listened and concluded Alessia had EPP but tested her blood to be certain. Within days, the answer was EPP. Since then, Alessia visits the hematology department and gets yearly blood tests. She even took part in a dermatology study at Jefferson Hospital. This is Alessia's first time at Camp Horizon. The family didn't know what to expect but the staff, campers and facility exceeded their expectations! Alessia met Willow and they had a BLAST!!! Another wonderful part of this story is that their two mothers, Jeneen Callahan and Helen Morfka, met on the APF Facebook and discovered upon arriving at camp that the girls were roommates. A God-incidence!!!! If you would like your child to attend Camp Horizon next summer, please see their website for information and an application.



APF SUPPORT One of the main goals of the APF is to provide support networks for patients to meet and share their experiences. Joyce Shelor and her son, Brad Rusher, who has AIP, were thrilled when they met Megan Davenport on the APF AIP FACEBOOK group. The three decided that meeting in person would be enjoyable and helpful to compare notes. Being able to communicate with other people with the same disease is not only a good opportunity to expand your knowledge, but also it is a great way to support one another. If

you have not joined the APF IN TOUCH NETWORK or one of the APF FACEBOOK groups or read the Twitter, E-news and the *Purple Light Blog*, please contact the APF and give us your contact information, including your email. The APF support networks will open up a new world for you. Being able to communicate with others with porphyria can be emotionally healing and very educational, not to mention the emotional support people can give each other.



Gary Eyster's Donation Esteemed artist, Gary Eyster, was an APF donor for many years. Thanks to his good friend and APF member, Ralph Gray, Gary also donated his valuable collection of paintings to the APF upon his death. Gary was an innovative, abstract expressionist with studios in Wilmington, North Carolina and York, Pennsylvania. Working until his death in July 2013, Gary never ceased exploring ways to express and fine tune his artistic vision. Gary's training spanned the 1950s and 1960s with traditional art education at schools in Philadelphia, Richmond, London, Paris and Wetzlar, Germany. Striving to break away from habits and routines that can stifle creativity, Gary searched for novel ways to introduce spontaneity and randomness into his artistic process. This led him to paint with such unusual objects as plungers, squeegees, wide mouth jars and squeeze bottles. Gary even created a number of works by applying paint and powdered pigments through layers of water to the canvas below. This unique process also allowed him to pull monotype prints from the surface of the water, creating works that were simultaneously unique and derivative of their larger, canvas progenitors.

Gary's works were shown in some of the most prestigious galleries and shows in the world. He chose the APF to hold his works and use them to help people with porphyria. He also taught young men and women how to achieve the mastery of some of his techniques and was very interested in the *Protect the Future (PTF)* program to train future porphyria experts. He understood the great value of present experts sharing their expertise with young doctors just as he had shared his talent with young artists. The APF is grateful to Gary for his valuable gift. If you are interested in viewing and/or purchasing these works, please see the website, www.garyeyster.com. Your purchase will provide funding for the APF PTF and physician education programs. You can also contact the APF to be placed in contact with Ralph Grey, who can assist you with your questions or purchases.

Keeping You Informed Did you know that the APF has many different avenues for you to keep up to date on what's happening at the APF and in the world of porphyria? Below are a few:

The updated APF Website has news and events bulletins, the acute porphyrias *Safe/Unsafe Drug* database, an extensive *Diet and Nutrition* section, and many *Members Stories* section that is full of patients experiences, a new *Caretaker Support* forum and *Caretaker* stories: porphyriafoundation.com.

The new Caretaker Forum is a means to share with other people who are caring for someone with porphyria. To join the Caretaker support group or receive more information, contact the APF or email us at apfcaregiver@aol.com

The APF Newsletter is mailed to you quarterly. Please join the APF and receive our Award Winning Newsletter.

The fantastic new Mobile Web APP for the acute porphyria *Safe/Unsafe Drug* database was built and provided by one of our APF members: porphyriadrugs.com

The APF Facebook Page can be found at <https://www.facebook.com/AmericanPorphyriaFoundation>

The APF Facebook support groups can be found at <https://www.facebook.com/AmericanPorphyriaFoundation/app/221460014534454>

The APF Purple Light Blog is a great place to learn about porphyria, the experts, and many related subjects. See the "[Purple Light Blog](#)" on the APF website.

Follow the APF on Twitter at [@PorphyriaHelp](https://twitter.com/PorphyriaFoundation)

The APF YouTube channel has some very informative videos, including Member stories:

[Youtube.com/PorphyriaFoundation/](https://www.youtube.com/PorphyriaFoundation/) and become a Member and receive our weekly APF Enews: [Join the APF](#)



Mobile Drug List Have you tried the new Mobile Porphyria Drug List? If not, it is very easy to use and to install on your mobile devices or find on your computer at www.porphyriadrugs.com. Don't forget that the APF drug list is always available, too, and is updated periodically by Dr. Peter Tishler, a Harvard porphyria expert who maintains and updates the site. Your participation in the Drug studies is very important. Advising Dr. Tishler when you have had a good or bad experience with a drug is one of the means that the drug list can be updated or verified.

Congratulations! We love babies at the APF and are happy to announce the birth of little Elisabeth "Ella" Maria, who was born 9/18/13 and congratulate her parents, Dr. Angelika Ludtke Erwin, MD PhD and her husband, Phillip. Dr. Erwin is one of our esteemed *Protect the Future* doctors. She was based at Mount Sinai in New York City and has recently moved to Cleveland Clinic. Recently, Dr. Erwin was one of the authors of the GENE Review article on "Congenital Erythropoietic Porphyria." She will also be participating in the exciting upcoming Porphyria Outreach Clinics that we will institute next year. You will hear more about her in the March newsletter. Right now, her hands are full with baby "Ella," her new home and her new work. **Congratulations!**



Janet Collins "I didn't develop PCT symptoms, such as red urine and blisters on the back of my hands, until after menopause. I had a forced menopause from my second chemotherapy treatment for breast cancer. About six year later, I had the red urine, blisters on the back of my hands and terrible tingling in my hands, knees, feet and arms and very fragile skin. A nephrologist administered the 24 hour urine test in a dark bottle in May of 2010, which confirmed I had a full blown case Porphyria Cutanea Tarda (PCT). My brother had the same problem as me, including the bad blisters on the back of his hands. But for seven years, he was told that he was allergic to blood worms. My brother owned a bait and tackle shop in NY. He began getting blisters at 55 years old in the summer, so he believed that the bloodworms were the culprits. Then, the blisters appeared on my hands at 55 years of age, as well. Then I had the same symptoms. Thank goodness for my good nephrologist who diagnosed me. I immediately called my brother and told him, 'I don't think you're allergic to blood worms, I believe you have what I have - PCT.' My brother and I finally have an answer - PCT."



You Are The Medical Heros There has never been a time in the history of porphyria that there has been so much opportunity to participate in research. Research Volunteers are the real Medical Heros, bringing their gift of time and energy to help them and future generations of people with porphyria have a CURE. Right now we have many research projects ongoing that require only that you answer questions and provide your blood. It is not necessary to travel to the research center with most of these studies. The time involved is very manageable and the other requirements are few. However, the

impact is monumental. These studies have been designed by the best experts worldwide, namely, members of the *Porphyrias Research Consortium* (PC). The PC created a large scale collaborative effort to develop new strategies and methods for diagnosis, treatment, and prevention of porphyria illness and disability. You have the opportunity to participate as a research volunteer and change the world of porphyria now and for the future. The first step is to register on the free *Porphyria Contact Registry*. This way the research team will have your contact information on your type of porphyria.

<https://rarediseasesnetwork.epi.usf.edu/porphyrias/register/index.htm>

If you know you want to participate in the studies below or need more information, contact Elizabeth at the APF, and she will place you with the Research Coordinator at the closest center or to the research project of your choice. You do not need to travel to the centers to participate in each research project. A member of the Research team or the Research Coordinator also will contact you and help you become a Research Volunteer. For further information, contact APF at [866 APF3635](tel:866-APF3635). The following are a few of the research projects for you:

- ❖ The *Longitudinal Study* only requires that you donate your blood and answer a host of questions. This study will take place over several years and will enable scientists to gain a better understanding of the porphyrias.
- ❖ Another study focuses on the family with participants from diagnosed members with symptoms and those who are not diagnosed and/or those who have no symptoms. The answers that can be gained from this important study can lead to major treatment advances.
- ❖ An important study using Panhematin® is also in need of patient volunteers. It is important to have predictable, acute attacks. You will receive the Panhematin® without cost to you during this trial.

In Memory and In Honor The APF is grateful to the following people who chose to honor their loved one with a gift to the APF. These gifts are used to enhance the APF educational programs, including the physician education program. *Thank you for your kindness.*

* *In Memory:*

Diane Levere for *Dr. Richard D. Levere*
 Kathleen Toelkes for *Donna Pagano*
 Lisa Kancsar for *Stephen Kancsar*
 Ada Trilling for *Jo Matranga*
 Barbara Smith for *Kenneth Smith*
 Dr. James and Carolyn Atkins for *Martha Atkins*
 Gary Horn for *Sandra Horn*

* *In Honor:*

Desiree Lyon Howe for *Dr. Richard Howe*
 Desiree Lyon Howe for *Ellane Heflin*
 Desiree Lyon Howe for *Dr. Karl Anderson*
 Charlotte Beck for *Cheryl Sundblom*
 Terri Witter for *Dee Bruno and Jeanie Lindsay*
 Diana L. Young for *Tracy Yelen and Dan Yelen*

Survey on Acute Porphyrias Recently, the APF distributed a survey about YOUR treatment for Acute Porphyria. If you have an acute porphyria (AIP,HCP,VP) and have not received the survey, please contact Yvette at the APF ASAP, and she will send one to you, pronto, 866.APF.3635. This is essential to develop improved physician educational publications and programs and to enhance the APF media, research and awareness programs.