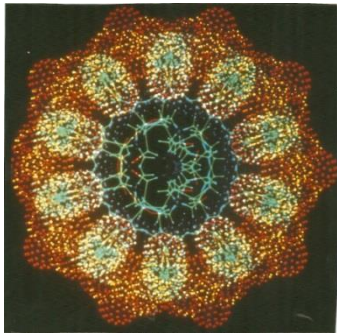


MERRY CHRISTMAS, HAPPY HOLIDAYS, HAPPY HANUKKAH, HAPPY NEW YEAR!



At first glance, the photo on the left appears to be a classic Rose Window from one of the early gothic cathedrals in Europe or a Christmas wreath decorated to the hilt, but it is neither. Instead, it is a cross section of a DNA molecule. Can you see the double helix in the middle? This tiny little DNA holds all the information about YOU: your hair, your height, your eyes, your frame, your mind. The photo has been published in our newsletters and on the APF website for many years, but it deserves to be introduced repeatedly to remind us as the Good Book says that we are "fearfully and wonderfully made." (Psalm 139:14)

This beautiful identifier is what each one of us carries in our bodies, like a serial number. And like a serial number, it marks each and every human being as an individual; special

and unique. Although we have shown this photo on a number of occasions, it is perfect for the holiday season, time of the year, illustrating the uniqueness and beauty in all of us and the wonder and order of life.

Each of you is very special in the world and is very special to us. We have enjoyed meeting many of you and thank you for the gift of your friendship. We wish you a wonderful Christmas, Happy Hanukkah, Happy Holidays and a Blessed and Happy New Year. Know that we are here when you need us and are ready to give you a powerful gift, the gift of information about porphyria. **"Those who know the most, do the best."**

What you can give is yourself. We are on the brink of many discoveries about porphyria, but researchers can't go further without your participation. Please join a study by contacting the APF.

Safe Unsafe Drug List Dr. Peter Tishler, who serves on the APF Scientific Advisory Board, is an esteemed



porphyria expert from Harvard Medical School. He has been overseeing the Safe/Unsafe drug list for many years and periodically asks our members to assist him in updating the list. I hope you have been using the APF Drug Database (<http://APFdrugdatabase.com>), which we established some years ago. I am updating the drug database once again, to add new medications and reinforce or change the safety of existent medications. Thus, I write once again to ask all APF members with an acute porphyria (acute intermittent porphyria (AIP), variegate porphyria (VP), hereditary coproporphyrinuria (HCP), ALA dehydratase deficiency porphyria (ADP) to provide information regarding your medications. We need to discover more medications that are safe and not safe for the acute porphyrias. You may have been prescribed a medication that was not on the list but

worked well for you or may have been prescribed a medication on the list that made you ill. Your comments are important to us.

If you have not received the Evaluation of Medications questionnaire for each medication, please contact the APF, and they will send one to you immediately. If you have received a form, please make a second copy if you want to add more medications to your list. Then please return it to the APF as soon as possible, and they will, in turn, send all of the questionnaires to me. I may contact you to clarify aspects of your report. Your participation is very important. Many thanks, Peter V. Tishler, MD.

U.S. Food and Drug Administration

Protecting and Promoting *Your* Health

YOU CAN MOTIVATE THE FDA TO GRANT SCENESSE ACCELERATED APPROVAL! The mission of the FDA is to protect and promote the public health.

That being said, we understand the role of the FDA to watch for the safety of the drugs they approve. In the 14 year history of the Afamelanotide/Scenesse treatment for EPP, there have been no negative reports.

The FDA is also charged with promoting the public health. This means they have a duty to approve medications that are safe, effective and beneficial to our health. So far, the FDA has not approved Afamelanotide/Scenesse even though it is safe, effective and beneficial to EPP patients.

The drug has already been approved in Europe. Our citizens in the US who suffer from EPP are raising funds to travel to Switzerland and pay for the implant. This is outrageous that we must travel at great expense to receive treatment that is available to half of the world. Considering that the drug is approved in Europe, there have been five clinical trials, seven years of experience with the patients taking the drug for compassionate use in Europe, it is even more outrageous. Therefore, the APF is undertaking a major advocacy effort to assure that the FDA will approve Afamelanotide.

If you have EPP or are a family member or friend of an EPP patient, we need your help. Please contact the APF at 866.APF.3635. Your participation is essential for us to be heard. We have had a great start. Every major television network has aired a story on EPP and the success of the Afamelanotide treatment and more to come!!!!

WHAT ONE FAMILY CAN DO Shawn Willis and his family hosted *The Moo've it in the Moonlight Run* on 8.28.15 to benefit the APF. The event, which was held in the Willis' hometown of Burlington, NC, was a tremendous success with over 200 participants who helped raise over \$2,000. Shawn is the ultimate inspirational person. Although he suffers from EPP, Shawn storms full speed ahead in life and experiences the world first hand despite EPP. Shawn tackles the mountains, kayaking in the ocean, working in an orphanage he supports in Africa, volunteering for the EPP clinical trials with Afamelanotide, and now hosting a major fund raising event to benefit the APF and enhance porphyria awareness. Shawn is a terrific husband and father. He runs several Chick-Fil-A restaurants, is active in his community and is a great advocate for porphyria and the APF. Thanks, Shawn Willis. You are a Renaissance Man.



runs several Chick-Fil-A restaurants, is active in his community and is a great advocate for porphyria and the APF. Thanks, Shawn Willis. You are a Renaissance Man.

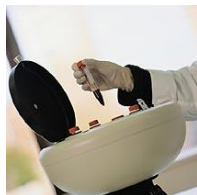
SHADOW RACE REPORT When the Cook family of Vernon, Texas joined the APF, they immediately started helping enhance porphyria awareness. Mom, Lee Ann and Chris Cook, wanted their sons Cason and Caul, to have the best treatment possible for their EPP. They started with the Annual Hat Day and now LeAnn and the family are hosting **The Shadow Race**, a barrel race for EPP and the APF. The event was held at the WCEC Indoor Arena, Vernon TX and had many attendees. **The Shadow Race** T-Shirts were a big hit and the race was a fun, exciting, BIG success. Photos of the event will appear in the next newsletter.



THE BENT RODS BASS CLUB REPORT We would like to thank our member, Victor Mejias, and his fishing club *The Bent Rods Bass Club* for their fundraising tournament benefitting the APF. Victor suffers with EPP and is severely photosensitive...BUT Victor loves to fish so he finds a way to do what he loves. He suggested that his fishing club raise funds for the APF via the annual tournament. The tournament was for people who love fishing, too. We sincerely appreciate your efforts and hope you all caught lots of bass in the process. Editor's note: Victor also serves as an EPP Facebook administrator and as a member of the APF/FDA advisory committee.



RESEARCH is life changing. We now have a government grant to research porphyria, but we cannot conduct research without YOU. World renowned porphyria researchers are conducting research but they cannot continue without YOU. YOU are key to all the discoveries that will change our health now and in the future. There are very important research studies ongoing right now that need research volunteers. You will not be asked to take a drug, rather you will donate your blood and answer a host of pertinent questions. Please join for yourself and your children.



- Everyone can participate in the Longitudinal Study. All you need to do is fill out questionnaires that reveal important facts about porphyria if enough people enter the study.
- Volunteers are needed for the Natural History part of the Alnylam Study to show the potential of their drug to prevent attacks. Fly to a research center and back home with all expenses paid.
- Research volunteers are also needed for the Panhematin study to verify that Panhematin can be used to prevent attacks. Although patients have been given the drug for a long time to prevent attacks, this study will give doctors the evidence to use it for attacks and to prevent attacks.
- Register on the National Porphyria Research Registry: <http://www.rarediseasesnetwork.org/porphyrias/index.htm>.

It is important to join the registry so that your name will go directly to the researcher teams. If you need help with the registry, just call the APF and Natalia or Jessica will help you. For details on how you can participate, call the APF at 866.APF.3635 or 713.266.9617. YOU ARE THE KEY TO FINDING NEW TREATMENTS AND A CURE.

AMAZON SMILE You can also support the APF through the AmazonSmile program! Amazon will donate 0.5-0.8% of the price of your eligible purchases to the APF, at no cost to you. Please make us your choice of a charitable organization and support porphyria research while shopping! Please note, the program will only be available to shoppers who visit Amazon via a special web address - [smile.amazon.com](http://smile.amazon.com/ch/36-4401266) - instead of the normal Amazon.com homepage. It is easy and free. AmazonSmile is the same Amazon you know. Same products, same prices, same service. Thank you for supporting us! Please start at this link: <http://smile.amazon.com/ch/36-4401266>. It is very easy to also help the APF fund our PROTECT THE FUTURE program to train future experts.

EMERGENCY ROOM GUIDELINES

The Emergency Room Guidelines for acute porphyrias can be found easily on the APF website: www.APFerguidelines.com. These Emergency Room Guidelines cover essential information for the emergency physician treating a patient in an acute porphyria attack, including common precipitating factors, typical presentation and other diagnostic clues, making the initial diagnosis, common sequelae and best practices for treatment. A **PowerPoint presentation (as PDF)** is also available.

It is a good idea to print the list and take it with you along with your Drug List, your diagnosis and your **Warning Wallet Card**. If you do not have a warning card, contact the APF and request one. If you do not have an APF **Emergency Room Kit**, you can purchase one from the APF. They are very comprehensive and include important educational articles, major medical journal articles, Panhematin brochures, CME course information and other data pertinent to diagnosis and treatment. Contact the APF for details on the Kit and Warning Card.

**EMERGENCY
ROOM**

1. The human porphyrias are clinical disorders reflecting defects in heme biosynthesis.
2. Acute porphyrias cause acute attacks of neurological symptoms that can be life-threatening.
3. Acute attacks are triggered by certain drugs, sex steroid hormones, reduced intake of calories and carbohydrates, alcohol and unknown factors.
4. Many of these factors stimulate heme synthesis in the liver, which in the face of a metabolic enzyme defect, leads to increased production of heme precursors that may be neurotoxic.
5. Delta-aminolevulinic acid (ALA) and porphobilinogen (PBG), are porphyrin precursors and intermediates in the heme biosynthetic pathway.
6. ALA and porphobilinogen (PBG) are almost always elevated in urine during an acute attack.
7. The most common emergency room (ER) clinical presentation is acute abdominal pain. Other features may include seizures, confusion and hallucinations, and a progressive polyaxonal motor neuropathy, which can progress to paralysis and respiratory failure requiring a ventilator.
8. A high index of suspicion in the presence of nonspecific symptoms is important for diagnosis. A family history of porphyria, female sex, onset during the luteal phase of the menstrual cycle, or recent use of a porphyrinogenic drug may be diagnostic clues.
9. A new diagnosis of porphyria as the cause of acute symptoms must be substantiated by finding a substantial increase in urine porphobilinogen (PBG).
10. Treatment should start promptly after the diagnosis is made. Mild attacks are sometimes treated with glucose loading (e.g. 3L of 10% glucose daily by vein).
11. Most acute attacks should be treated with hemin (Panhematin®, Recordati Rare Diseases at: www.recordatirarediseases.com or 866.654.0539) 3-4mg/kg into a large peripheral vein or venous access port daily for 4 days. Reconstituting Panhematin® with human serum albumin rather than sterile water is recommended prior to infusion. This helps prevent phlebitis at the site of intravenous infusion.
12. Hospitalization is usually required for symptomatic treatment of pain, nausea and vomiting, correction of electrolyte imbalance and observation for respiratory impairment in a general medical service or ICU.

The authors of the Guidelines are noted porphyria experts: Neville R Pimstone MD, PhD, Director Liver Diseases Greater West Los Angeles Veterans Affairs, Professor Emeritus UC Davis; Karl E. Anderson MD, Professor, Departments of Preventive Medicine and Community Health, Internal Medicine, Pharmacology and Toxicology, Associate Director, General Clinical Research Center, Director, Porphyria Center and Laboratory, University of Texas Medical Branch, Galveston, Texas; Bradley Freilich, MD, Kansas City Gastroenterology and Hepatology, LLC.

What if the doctors will not look at the ER Guidelines or other materials contained in your ER Booklet/Kit? This doesn't happen frequently, but it does occur and when it does happen, it can be disheartening. Here are a few tips to help you gain appropriate attention.

- Make sure your correct diagnosis is included in your packet and try to remember to bring a copy
- Make sure you have major medical journal articles, like the *New England Journal of Medicine* or the *Annals of Internal Medicine*. It is best not to include articles from websites and minor medical journals.
- Have the Safe/Unsafe drug list on the APF website handy.
- Show them your APF *Warning Card*, which lists the URL to *ER Guidelines* and the *Safe-Unsafe Drug List*
- Explain your disease and symptoms as succinctly as possible.

JOIN THE FDA CAMPAIGN



JT von Seggern is a 20 year old college student with Erythropoietic Protoporphyrin (EPP), who also happens to be a competitive, nationally ranked tennis player. Before he discovered he had EPP, JT lived years in pain with no apparent cause. Now the swelling of his hands, forearms, face and feet is so extreme that even the blood vessels in his feet rupture. He also experienced painful scabby sores on his nose, lips, cheeks, neck and ears. He visited numerous dermatologists, and pediatricians without success. Finally, a doctor at Georgetown University Hospital recommended a pediatric dermatologist. By that time his parents had assembled a three ring binder of pictures of all of the episodes, swelling, scabs, scars, etc. His doctor immediately knew JT had EPP and ordered tests to confirm his suspicion. They learned that JT's

sports loving life would be forever altered. At the time, JT was playing both baseball and soccer. Knowing that he could only bear limited exposure to UV light, one of the outdoor sports had to go. JT wore sun protective clothing as much as possible, which allowed him to continue playing soccer. Most other normal childhood activities were challenging at best. Family trips to the beach meant JT had to stay inside until sunset, and his new passion, tennis, had to be played indoors. Class field trips usually ended with JT in pain and missing school for a few days while recovering. While JT handled this burden remarkably well, it has definitely impacted his quality of life and his psyche. All activities that are normally associated with pleasure: picnics, hikes, fishing, parks, sport are a source of pain for JT, but his love of the sport overpowers the pain.

Then the family heard about Scenesse/Afamelanotide and felt the treatment would help JT. He travelled at great expense to Switzerland to receive the implant and, like other EPP patients, and, like others, was able to live in the light pain free. JT and his family will join the APF to strongly advocate for FDA approval.

We need YOU, too. Please join us in this important advocacy effort to gain FDA approval of Afamelanotide. Call the APF 866-APF-3635 for details on what you can do. Thank you!!

EPP AND YOUR WORK



One very essential message we need to send to the FDA is how EPP affects your employment. Nurse Hannah Bailey has a great example for us to give them. If you have one, as well, contact us. Hannah: *Most recently, my career has begun to suffer. While I am still able to satisfactorily perform all of my duties as a RN, I am finding it more and more difficult to recover from working too many twelve-hour shifts in a row under the harsh lighting. I have asked my management to allow me to have a few days break between my shifts. We work three twelve-hour days per week.*

This request has been met with considerable pushback. I am already having to wear UV protective gear during my shifts at work, due to the copious amounts of fluorescent lighting used at the hospital. While the ADA allows for this request to be fulfilled, it seems that my management would rather not deal with making necessary adjustments to accommodate my condition, no matter how small those adjustments may be. My story is not unique in that respect. It seems that many employers would rather not accommodate an issue such as photosensitivity, and would rather cast the EPP patient aside as a bad fit for employment, leading to disability compensation that is inadequate and unnecessary, as we are still productive members of society with lots to offer. As my condition continues to worsen, I cannot help but think that a drug, like Afamelanotide, will make a substantial difference for me. If this drug has already provided others with favorable results, allowing people to live more normal lives, neutralizing the damage of the sun for those of us who suffer from EPP, it only makes sense to give us a chance. This chance could provide a higher quality of life and better opportunity for happiness for future generations who would otherwise suffer liver damage and inevitable years of pain, suffering and depression that comes with this rare disease. Hannah is helping the APF with our campaign to gain FDA approval of Afamelanotide.

LANCE JOINS THE FIGHT

Lance Miller also has EPP. He also participated in the Afamelanotide trials and is helping in our effort to advocate for the FDA approval of the drug. Until the trials, Lance said, "Prior to receiving Afamelanotide, I was only able to tolerate the sun for 30 minutes or so. Life was difficult and stressful for me and my family was trying to make accommodations for me. I have two kids that love to be outside and play at the park. I must watch them from a window in my house or my very tinted windows of my vehicle. After I received Afamelanotide, I was working on the family farm for eight hours in the sun in the pastures wearing nothing but a t-shirt and jeans. It allows me to do things I have never been able to do."

Lance is just one person, but his letter to the FDA is powerful. Read the whole letter on the APF website. Your letters and testimony are ESSENTIAL, too. Please contact the APF and join in our advocacy effort to get this drug approved. There is no reason the FDA should be lagging behind Europe. They need to approve it TODAY!

PCT JOHN GLADDING *Back in late 2012, I kept getting these random, tiny blisters. My PCP told me to quit smoking. When that didn't work, she sent me to a dermatologist. I had a blister on my finger, which he removed and biopsied as he suspected I had Porphyria Cutanea Tarda (PCT). He confirmed the PCT and advised me to avoid direct sunlight, and wear at all times Neutrogena sunblock SPF 100+ for sensitive skin, long sleeve t-shirts, gloves, hats, and scarves. In the Texas 100° plus weather it seemed to be difficult. I tried, but the blisters kept coming and increasing in size. I didn't have any health insurance at the time. The dermatologist had told me I had very high iron in my blood and that a scheduled phlebotomy twice a month for 6-8 months would be the cure. Seems easy enough, right? He said he didn't do the phlebotomies and my PCP couldn't arrange them.*



In the meantime I quickly added private insurance coverage at my job at the Alzheimer's and Dementia Assisted Living Community. But the insurance didn't cover the phlebotomies and left me having to pay \$250 twice a month for phlebotomies, which I could not afford. Neither doctor helped me find other options. I ended up moving back to my home state of Michigan. I was nervous of the HIV services in Michigan, but they were amazing. I was also diagnosed with Erythrocythemia, which is a more specific term for saying I have high red blood cell levels. After trying only once a month phlebotomies for all my conditions, I was actually worse. Now we are trying a twice a week regimen I hope will help me improve. I also have Hepatitis C that I acquired through a blood transfusion when I was born in 1979. It is all very complex. Editor's note: If you have a story to tell, please contact the APF: 713.266.9617.

PORPHYRIA CUTANEA TARDA (PCT) is the most common and also the most readily treated form of



Porphyria. Blisters and crusting of sun-exposed areas of skin are the most prominent features. PCT is caused by a deficiency of the enzyme uroporphyrinogen decarboxylase (UROD) in the liver. PCT is somewhat more common in men than it is in women and usually develops in middle age, hence, the name *tarda* which is Latin for late. Approximately 20% of PCT patients have an inherited (autosomal dominant) deficiency of UROD and are said to have familial (Type II) PCT. At birth UROD is approximately 50% normal in all tissues in these patients. However one or more of the additional causative factors listed below are important in these patients. Type II PCT becomes manifest when the enzyme activity in liver becomes much less than 50% of normal, due to one or more of these additional factors. Patients with familial PCT respond to the same treatments as those who do not have an inherited enzyme deficiency. When UROD does become markedly deficient in the liver, porphyrins accumulate and spill out into the blood. They are then transported to other tissues such

as the skin and are excreted in urine and feces. Porphyrins in the skin absorb light and release this absorbed energy in a manner that leads to generation of reactive forms of oxygen that damage the skin. Therefore, exposure to light leads to skin fragility, blistering and scarring.

Iron has a central role in causing PCT. Liver iron is often increased in PCT. Removal of iron from the body always leads to a remission. Most PCT patients do not have a great excess of iron, and in most cases, removal of only 5-6 pints of blood is needed for successful treatment. **Excess alcohol intake** is very common in PCT. **Hepatitis C** is common in PCT. **Estrogens** are contributing factors, especially in women. **Other factors** are important in some patients.

The most common symptoms of PCT are fragility and blistering of light-exposed areas of the skin—especially the backs of the hands, the lower arms and the face. Patients often report that their skin is unusually fragile, so small bumps or knocks can scrape away the upper layer of the skin or cause a blister. The blisters contain fluid, rupture easily, crust over and then heal slowly. Skin infections, scarring and changes in coloration may result. Small white spots called "milia" are commonly found on the hands and fingers. Another feature which is often seen is excessive growth of facial hair.

The best screening test when PCT is suspected may be a plasma total porphyrin measurement. A normal result excludes active PCT. This test will also detect any other type of Porphyria that is causing skin problems. Further testing is then needed to establish the type of Porphyria. The most widely preferred treatment is repeated phlebotomy (venesection). The objective of the treatment is to decrease the amount of iron in the body to the lower limit of normal. With repeated phlebotomy, the body's iron stores are gradually depleted. This process also removes iron from the liver, and the activity of the UROD is gradually restored. A pint of blood is removed every 1-2 weeks until the ferritin reaches the lower limit of normal. After the plasma porphyrin levels become normal, the patient is able to tolerate sunlight. Low-dose chloroquine is a suitable alternative treatment, especially in patients who cannot tolerate phlebotomies. Chloroquine mobilizes excess porphyrins from the liver. Treatment of Hepatitis C with interferon alpha and ribavirin is available but is often not effective. Treatment of PCT is almost always successful, and the prognosis is usually excellent. The condition is not progressive and seldom disabling. *Please see the APF website PCT section.*

CONGRATULATIONS



Wedding Bells rang for longtime APF member Tracy Yelen and Michael Nudo in Chicago, Illinois. Many of you have seen Tracy on the homepage of the APF website in her outstanding presentation on her experience with Panhematin. Tracy has participated in research for many years and is a great advocate to encourage others to join in the effort. According to Tracy, Michael is all things good, including being a wonderful caretaker when she is ill. This beautiful couple will be added to the APF homepage soon. Take a look. Another young couple married recently. Raine Nallie and Paul Perrault asked friends to donate to their charity of choice instead of gifts and by this kind gesture, a number of their friends donated to the APF. God's Blessings !!

PORPHYRIA, A Lyon's Share of Trouble an Update

Porphyria: A Lyon's Share of Trouble an Update



If you read the first edition of Desiree's book, you will want to read what has happened over the past years since it was first published. In this remarkable book, Desiree offers a captivating, information rich discussion of the porphyrias, as well as her own case and those of many other patients whom she has helped over the past thirty-five years as co-founder and Executive Director of the American Porphyria Foundation. Desiree embraces the challenges of porphyria with courage and humor and writes about these experiences in an open and honest dialogue. After thirty-five years as a medical writer, she has delineated the new and exciting developments in the treatments of all the porphyrias, offering explanations about this complicated group of diseases in a readable manner. She also tells about the doctors who have dedicated their lives to porphyria and gained "world expert" status. She donated both books to the

APF to help support the APF educational programs. The book can be purchased via AMAZON or on the the APF website to read on your Kindle. If you don't have Kindle, you can use a Kindle software to read on your PC or tablet. Order now for only \$5.99 at: <http://www.amazon.com/dp/B014WZX26M>

MEDIA AND VIDEOS



If you have not been privy to all of the Media that has been on TV in the past six months, people helped us gain TV awareness, and you have missed a lot of great educational and interesting programs about porphyria. You can find a list of television that has been on in the past on the APF website. Programs like *House*, *ER*, *Medical Mysteries*, *Discovery Channel*, *CSI*, *Doc Martin*, *Greys Anatomy*, and many, many more. The newest documentary on EPP is well timed as we press forward to have FDA approval of Afamelanotide. See it at: <http://www.newschannel5.com/news/medical-news/drug-helping-patients-with-epp>.

Please share this video with as many people and in as many places as possible to help heighten porphyria awareness. You may have been able to watch all the programs and documentaries we listed in the last newsletter, like *NBC Dateline*, *ABC Nightline*, *Fox News*, *ABC Good Morning America*, *Telemundo* and many more.

The newest media on Acute Porphyrias is a PBS documentary about AIP which features Dr. Lisa Kehrberg and porphyria expert, Dr. Karl Anderson and Dr. Joseph Bloomer and others. It can be viewed at:

<http://www.explorationhealth.tv/shows/701b.html>

If you are willing to participate in media appearances, it is important to have your story on the Members Stories Section on the APF website. Reporters peruse these rare disease websites and choose people from them to feature. Email your story to porphyrus@porphyriafoundation.org.

The APF also has a new video section to add to the member stories. To kick off our new Video section, take a look at the website and watch Amanda Boston tell how she was misdiagnosed, ended up in a coma in a near death situation, was finally diagnosed with Variegated Porphyria (VP) and was given Panhematin which saved her life. She tells about becoming a research patient, getting involved in the APF and what life is like now. It is a great way to learn about porphyria and share with someone who understands you. Please make your own video and send it to the APF. Make sure that it is medically accurate. You can call Desiree to work with you if you need assistance. Many of you will relate to Amanda's tragic suffering and triumph after her diagnosis. You can also watch Tracy Yelen Nudo, Amy Chapman, Dr. Lisa Kehrberg and Desiree as they, too, tell about their lives pre and post diagnosis with acute porphyria. But we also need stories and videos of people with PCT, EPP, CEP and other acute porphyrias. See Amanda's story: https://www.youtube.com/watch?time_continue=1&v=zmM3HSH8dDk

Or go the APF website or APF YouTube channel and watch them all. We want to encourage each of you to video and write your story.

NATIONAL PORPHYRIA AWARENESS WEEK WILL BE APRIL 16-23, 2016. GET READY!!!!!!

PROTECT THE FUTURE This is the most important program we support!!! The reason is that the present experts are approaching retirement. Some have already retired but are remaining to complete the research projects that are so essential to our health. The least we can do is to support the program to train young doctors as future experts. Training these young doctors is a very expensive endeavour. We need your support to either donate to the program or to host a fundraiser for the program. We have trained twenty young doctors with more to come. These young doctors are included in all the research projects, the publications Please help. We do not want the expertise gained to be lost.

23andMe Renowned geneticist and porphyria expert, Dr. Robert Desnick, has kindly answered our questions regarding diagnosing porphyria via 23andMe. He states, *It has come to our attention that some Porphyria patients have sent their DNA to 23andMe or to other commercial companies and have received results suggesting that they have "DNA confirmed Porphyria."* We are concerned that results from companies other than DNA testing laboratories that have experience in diagnosing porphyrias may provide patients with misleading information.

A major issue with DNA testing is whether a gene alteration (variant or mutation) is pathogenic (disease-causing) or benign (a change in the gene that does not cause or make one at-risk for the disease). For example, 23andMe does NOT do gene sequencing, but does determine if you have various gene alterations in the porphyria genes, all 57 of which are benign, and are not disease-causing but occur in a particular gene in which other lesions are in fact disease-causing. The benign lesions usually are identified by an "#rs" number. These benign changes are quite common but may lead a patient to believe that he/she has one or more porphyrias. They do not affect the heme biosynthetic enzymes, as they are not pathologic lesions. Of the gene lesions that cause disease, over 98% would be identified by gene sequencing as is done for all the Porphyrias at the Mount Sinai Laboratory and four Porphyrias at the Mayo Laboratory.

The Mount Sinai Genetic Testing Laboratory provides Porphyria DNA testing for all eight Porphyrias and has a full-time Porphyria Genetic Counselor, Dana Doheny, MS, who is available to assist in arranging testing and interpreting the results. Typical time from receipt of sample to result is about two weeks. If the patient has "biochemical-positive" results and a DNA alteration cannot be found, there is a 1-2% chance that the patient has a "cryptic" mutation or a large deletion in the porphyria gene that is difficult to find by sequencing. The Mount Sinai Laboratory will do additional analyses to find the Porphyria gene lesion, if the patient has one.

Mayo Laboratories test for only three of the four Acute Porphyrias and for Erythropoietic Protoporphyrin. To our knowledge they provide sequence results.

PATIENT MEETING was an exceptional event that was an important part of the International Porphyria Congress. The meeting gave people the opportunity to meet others with porphyria, and learn about each type of porphyria. The day-long event began with introductions of all the directors of the many porphyria organizations worldwide. They shared what was happening in their societies. Desiree spoke about the APF and showed photos of some of the significant programs and activities. Presentations on all the porphyrias were given by world experts, including US doctors. It was a wonderful day of learning in a cheerful and relaxed environment. Seeing old friends and making new ones was the best part of the day. (L-R Rocco Falchetto, PhD, Jasmine Barmin, PhD and Elisabeth Minder, MD).



IN MEMORY We are saddened to hear of the passing of dear family members and friends. Some of you have chosen to honor a life by making a gift to the APF to help others with porphyria. Please thank: Lisa Perry, Donna Melander, Russ and Helen Hawkiss, Linda Dunn, Sharon and Edward Mincher, Sierra Pacific Foundation, Linda and Galen Roberts, Nancy and Donald Quintrell for **Daniel J Pudlicki**; Desiree Lyon, Jessica Hungate, Natalia Sturza, Rob Saupe, Amy Chapman, Amy Gasper-Burke, Amanda Boston, Katie Fabian, Pierre Mouldoux and Andrew Turell for **Lawrence B Smith**; Desiree Lyon Howe, Sarah Bucic, Judith A Reynolds, Fay Cruchley for **Jackie Nye**; Diane L Levere for **Dr. Richard D Levere**; Julia Elliott, Erinn Garcia, Cari and Dennis Turner, Allergy Asthma Immunology of Rochester for **Charlotte Szuminski**.



IN HONOR We also thank those who donated to the APF in honor of a friend or family member: Tom Sodeika for **James V Young**; Kimberly Wood, Ann Reynolds for **Shawn Willis**; Meaghan Meachem for **Raine Nallie and Paul Perrault**; Bonnie Schnell, Greg Young, Jeryn Siasu for **Tracy Yelen Nudo**; Greg Young for **Susan Young and Tracy Yelen**; Sheree and David Linker for **Melissa Nagin**; William Dreskin for **Ryan Turell**; Pamela Nadeau, Lauren Fontaine for **Katrina Lambert**.

