American Porphyria Foundation

4th Quarter, 2010

Kate, Desiree, Yvette, Elizabeth, Maria, and Ann

We wish you a Merry Christmas, a Blessed Holiday Season and a Healthy, Happy New Year.

You have enabled us to make great strides this past year by providing support for our many patient and physician education and awareness programs and services. One service is our Toll Free number which has been a life-line to our caring, helpful staff. It also has been our pleasure to become acquainted with so many of you and your physicians. We look forward to more new friends and another successful year.

What Kind of Doctor Should I See

Patients and family members often contact the APF wanting to know what type of doctor will know most about their kind of porphyria, and what type of specialist will provide the best care. While hematologists or gastroenterologists are often the go-to doctors for acute porphyria, with dermatologists taking care of patients with cutaneous porphyrias, it's important to be aware that most porphyria patients will be cared for by doctors who have never treated porphyria. And that's fine.

The APF emphasizes that all people needing care should first have a primary care physician who can oversee their care. The good news is that the APF will send your doctor a very comprehensive Physician Information Kit containing guidance from porphyria experts. They will also help facilitate a consult with a porphyria expert if special problems arise. The most important thing for all patients is to have a relationship with a local doctor who will get to know you, learn about porphyria and take care of your health needs. So what kind of doctor should you see if you have porphyria? **A GOOD DOCTOR!**

More EPP Trials

More EPP Clinical trials with Afamelanotide are on the horizon for spring of 2011. If you would like to participate in the trials, even if you were a participant in the recent trials, please contact the APF. If you are willing to join us in our efforts to enlighten the FDA on the need of a treatment for EPP, please contact us.

Merry Sunshine

My name is **Allison Linner** and I am seventeen. I came into this world on a cold, rainy day in early October, but my Dad said that the moment I was born, the sun peeked through the clouds, thus my nickname, "Sunshine."

For some reason, my parents always knew there was something wrong with me. My life was dotted with strange symptoms, including lots of pain and lots of doctors for a girl my age. Each doctor diagnosed me with something else. When I hit puberty, things worsened. I had terrible episodes during which I would be doubled-over in front of the toilet, wanting to be unconscious because of the pain, but unwilling because I knew if I passed out I might choke on my own vomit. The toilet even became my pew. I fought to stay conscious while these other things were occurring. I could feel that my heartbeat was weak and irregular. Another strange symptom to my endless array was extraordinary photosensitivity. When ill, I would lie on my bed with my eyes shut, a wet washcloth over my eyes, the blinds closed tight, and I would still be irritated by light. This is one reason I thought I was just nuts. I even had times when I experienced paranoia and hallucinations. I would cry and pray hard. I wondered how my nickname could ever be Sunshine.

I visited the doctor and questioned him, "In your professional opinion, do you think these symptoms that all of my different doctors have diagnosed over the years, are intertwined in some way?" He said, "No, there is no such disease that has all of your symptoms." But there had to be, I thought." I knew in my heart that each symptom was a puzzle piece and I realized that my family and I would have to play large roles in putting it together.

One day, my Dad and I talked a lot about what was happening. He promised me that we would find out what was wrong since we shared similar symptoms. At the end of our discussion, he jokingly said that maybe because we were so sensitive to light, we were vampires. He also said that there actually was a disease upon which people wrongly said the legends were based. I decided to look it up, just for fun. Reading the list of symptoms was like reading the story of my life. It was actually a very spiritual experience, it felt like reading the scriptures. A few days later, I went to the doctor and asked him if he could order the tests. He said "Sure, why not?" While waiting on the obscure test results, I performed at home the '24-hour urine test.' Within a few hours, my urine was a deep purple-red hue. A few short days later, the tests came back positive for Hereditary Coproporphyria (HCP.)

This is a hard disease to have, there is no doubt about it. I know what pain is. But every day, I thank our Father in Heaven that I finally know what ails me; and that I'm not crazy. I've learned to cope with it daily. When I do have flare-ups, that is probably when I'm closer to God than any other time. It is so humbling. I am actually grateful for my disease because it does so much to compel me to be a better person.

When I turned 17 on October 10, my family gave me a card that has a cartoon sun smiling on it and it reads: "Happy, Bright, and Lots of Fun." Inside, each member of my family wrote a little note to me. In my Dad's message he wrote, "You don't need the sun because you carry so much light inside you wherever you go." **I guess I'm still Sunshine after all.** ☺

The Sun and You

You might think it strange to write about photosensitivity and the sun in the wintertime, but information about the sun is never out of sync when it comes to porphyria. Since a photosensitive person can be affected by the sun at any time, it is helpful to learn several facts about what determines the sun's intensity.

As you know, the closer to the equator you are, the more intense the sun's rays and, the higher up you are, the more intense the sun's rays. Your UV exposure increases by 10% for every 3,280 feet in altitude; at 6,000 to 8,000 feet in elevation, you're exposed to 25% more ultraviolet radiation than at sea level. Also, snow is an efficient reflector of sunlight. When skiing or hiking in snow, 80-90% of UV light is reflected at you, dramatically increasing your sun exposure. Grass in comparison reflects only about 3% of sunlight. Water, especially when still, also reflects sunlight. Interestingly, still lakes, can reflect up to 100% of UV light (hence the term mirrored lake), doubling your UV exposure.

Members contact the APF for a Total Block sample.

A Family Affair

What do you do when you have a photosensitive porphyria and have to work in the sun. Ask Arthur Shull who has EPP and grew up on a dairy farm. He faced the sun each morning as he checked on the herd. Dairy farming is his life not just his profession. EPP has made it difficult for him, because he has had to work in the outdoors every day. He learned to keep on his hat and gloves, cover up and use other photo protective measures.

Because he has had to endure severe photosensitivity and wanted to help other people with EPP, Arthur volunteered for the EPP clinical trials with *Afamelanotide* and has been taking the train to and from the Mount Sinai Hospital in New York City.

In fact, the trials have become a family affair! Arthur's brother, Alan, also has EPP and flies to UC San Francisco to participate in the trials. Unlike Arthur, his brother didn't remain on the farm. Instead he moved to Oregon so he could better avoid the damaging sunrays. Like the other volunteers, both men are very important to the results of the trials.

Although some patients can clearly see the benefit of the *Afamelanotide*, Arthur thinks that he received the placebo. Regardless, being a participant is of ultimate importance whether a person receives the "real drug" or the placebo. Since EPP is rare, it takes a commitment from every participant to make the trials a success.

If you have EPP and want to talk with another EPP person, Arthur is an especially friendly person and has offered to talk with other EPP people. And when you have your milk each morning, think of him. Contact the APF for Arthur's number.

Kudos to all the EPP volunteers!!!! We sincerely thank the volunteers for their time and willingness to help people with EPP in the future.

An Okinawa Love Call

Andrea Reyes and her husband, Dario, had already endured a separation when he was assigned to Okinawa, Japan. Andrea tried to join him but found out that her health was in the way!!!! Before the Air Force would allow Andrea to join him, they wanted information about porphyria and assurances that she could receive Panhematin in Okinawa quickly if there was an emergency during her porphyria attacks.

The APF staff responded by sending our comprehensive physician information packet to her doctor. Then we contacted Dave Tworek at the Medical Affairs Division of Lundbeck Pharmaceutical for help with the Panhematin questions. He, too, responded quickly and provided all the emergency and shipment telephone numbers the Air Force required to order Panhematin in an emergency and the procedure for it to arrive timely. In fact, Panhematin can be shipped from a location near the Chicago, O'Hare airport within hours and arrive at US destinations within 24 hours. Fortunately, US military installations around the world are considered US destinations.

Since the APF staff was a big help, they felt part of the excitement when Andrea was finally given permission to join her husband in Okinawa. Not long after she arrived, she sent us a poignant photo of their reunion with a special "thank you." Being a blessing to our members is a joy for each of us.

Interestingly, many doctors and their patients in need of Panhematin are not aware that they can receive the treatment with 24 hours through the Panhematin Emergency Line: **1-800-673-6723**.

VERY IMPORTANT: PLEASE UPDATE YOUR ADDRESS AND EMAIL.

New PTF Doctor

We are proud to announce that Dr. Sahil Mittal has joined APF's Protect the Future (PTF), our program to train the next generation of porphyria experts. Dr. Mittal will be training with Dr. Karl Anderson at the University of Texas Medical Center in Galveston, Texas and MD Anderson Cancer Center. Dr. Mittal graduated from medical school from one of the most prestigious schools in Patiala, India. He was a fellow in Gastroenterology at UTMB and did his postgraduate studies at Institute of Medical Education and Research in Chandigarh, India. He is a member of the American College of Gastroenterology, American Society for Gastrointestinal and Endoscopy, American Gastroenterological Association and American Porphyria Foundation. Dr. Mittal has a great interest in the porphyrias and will be working with Dr. Anderson on the upcoming Consortium research projects, including the Longitudinal Study and clinical research. He has been awarded the Presidential poster award by ACG in 2007, travel scholarship by AGA for methodology reserach conference 2007. He is also part of the Translational Reserach Scholars Program (TRSP) at UTMB. Dr. Mittal is married and has a one-and-a-half year old and enjoys living in Galveston.

Support Meetings

APF members, Amy Chapman and Marlene Brezee hosted a support meeting in Michigan and Cherry and Thom Schillinger hosted one in California. Dr Peter MacField of Infusion Associates spoke at the Michigan meeting. Having an educated physician attend or present at the meetings is an added bonus. Although the APF still maintains the IN TOUCH network, we do not host support meetings. However, APF members host their own meetings and develop their own agendas. We will, however, notify our members that there will be a meeting in their area or will post a notice if you want to host a meeting. People with porphyria enjoy sharing and learning together, so over the years, we have seen many friendships begin at such support meetings.

You can communicate with other people with porphyria through the APF IN TOUCH network. All you have to do is call the APF for a permission form. Just sign it and return it and you will receive a list of all of the other people who want to be IN TOUCH. In fact, we will be sending out an updated list in a few weeks, so sign up in time to receive it. You can call Yvette and tell her you want to be part of the IN TOUCH network and she will send you the forms to return signed.

Many Thanks

to Amy and Craig Chapman for flying to Houston and donating their time and expertise to help us upgrade our computers and programs. Volunteers are what make the APF such a great organization.

Medical Conventions

APF members, Michael Drew (l), Charles Johnson (r) and EPP expert, Dr. Michelene Mathews Roth, manned the APF booth at the recent Liver Meeting in Boston. Over 8000 liver doctors and nurses gathered at the **American Association for the Study of Liver Diseases (AASLD)** and the APF was there with our educational materials to help update them on the disease. Last year, porphyria expert and board member, Dr. Joseph Bloomer, won the prestigious President's Award for Liver Disease. We are still bragging on Dr. Bloomer for this outstanding achievement and proud that he has been one of the porphyria experts on the APF Scientific Board for over 25 years.

This convention was followed by the **American Society of Hematology (ASH)** December 4-7 in Orlando. Volunteers: Claire and Robert Sadowniczak, and Shaaron Sellars.

A Belly Laugh

When I opened up the email from APF member, Kim, I was aghast. She informed us at the APF that the telephone number we had on the APF website for Quest Diagnostics was not correct. Instead, it was the number of a pornographic site. I couldn't believe my ears. "Surely not," I thought. I hurried to the APF website and found the number we had listed and indeed, it was true!!!! As bad as the error was, our whole office began howling with laugher—what I call belly gushers.

We did not know Quest changed their number, but the new one is 1-800-643-4652.

What Makes Them Different

H. Lundbeck Pharmaceutical is a very large pharma company based in Denmark with a unique and impressive ownership structure that is very different from most corporations. It is so unique that we felt our members would better appreciate the company and their 430 US employees. It is worthy to note that 70% of H. Lundbeck is owned by a foundation that contributes to healthcare research. The foundation was created in 1954 by the widow of founder, Hans Lundbeck and the financial support for research/natural sciences is through grants and fellowship opportunities. The Foundation aims to grant awards that make a difference, with an emphasis on central nervous system (CNS) like the acute porphyrias. Their 2009 contribution to research was a hefty $58 million and in the same year invested 23-24% of their total revenue in research and development along with their foundation investments in R&D.

2010 APF Corporate Award

The APF will present the 2010 Corporate Award to Lundbeck, Inc. for their interest in the porphyrias. The award will be presented to Mr. Joe Nolan, Chief Operation Officer of Lundbeck, Inc. which is the US division of the company H. Lundbeck, which is based in Deerfield, Illinois. H. Lundbeck is a global pharmaceutical company established to improve the lives of patients suffering from complex CNS disorders and rare diseases for which few, if any, effective treatments are available.

With the corporate headquarters in Copenhagen, Denmark, they have an international presence with pharmaceuticals in more than 90 markets and 5,900 employees in 56 countries. In the case of Panhematin, we are fortunate that their focus is on specialty therapies to address unmet medical needs. Without them, there is a potential loss of the treatment. Thus, we are sincerely grateful for their steadfast commitment to ensuring access to Panhematin.

We thank Mr. Nolan and the many terrific people who work at Lundbeck, Inc. We appreciate their generosity, which has helped heighten our many programs and services for education and awareness of the porphyrias. We also appreciate their willingness to build relationships with porfessional and advocacy communities in order to better determine needs and to generate solutions for severely ill patient populations, like the porphyrias. Lundbeck, Inc is indeed fulfilling their mission for acute porphyria patients by continuing to make Panhematin available. Over the years, Panhematin has saved countless lives of acute porphyria patients.

Conference Call : Dr. Karl Anderson

Thank you to porphyria expert, Dr. Karl Anderson, for devoting his entire Saturday, November 6 to host the APF "Conference Call With An Expert." Dr. Anderson spent the first 1 hour 30 minutes with acute porphyria patients and the next 1 hour 30 minutes with EPP and PCT patients making presentations on the diseases followed by question/answer sessions.

We also thank, EPP expert, Dr. Michelene Mathews Roth, for attending the call and lending her expertise in EPP. We are fortunate as porphyria patients to have access to both world renowned experts. Dr. Anderson, who is based at the University of Texas Medical Branch and Dr. Roth, who is at Harvard, have both served on the APF Scientific Advisory Board for almost three decades and have participated in a number of these conference calls.

The APF has supported many of these telephone conference calls in the past and will continue the service in the future. Like the other calls, this one was a resounding success and one of the best means for individuals who have never had the chance to talk with an expert to have that opportunity. Participants have told us that they especially enjoyed being able to ask questions they have wanted to ask an expert.

Dr. Anderson treats and consults with patients and doctors and is considered one of the most renowned experts in the world. To learn about these conference calls and how to attend, watch your ENEWS. If you have not registered for the ENEWS, contact the APF and give them your email address.

Thank You Alan Hunter For New Porphyria Cutanea Tarda Brochure

We thank Alan Hunter for supporting the new PCT brochure. Alan is a long time member of the APF and has been very helpful to the APF.

Porphyria Cutanea Tarda (PCT) is the most common and most easily treated porphyria. It occurs worldwide in all ethnic groups and in both sexes. Susceptibility to PCT is often inherited in which case the PCT is known as "familial" (type II). There is also a condition known as acquired PCT, which may occur in individuals with a genetic predisposition. There are very few people who are homozygous PCT, where both parents have the disorder. Those that are have a very severe form of the disorder known as Hepatoerythropoietic Porphyria (HEP).

PCT generally begins in mid-adult life after exposure to certain chemicals that increase the production of porphyrins in the liver. These include alcohol, estrogen, iron overload, ethanol, polychlorinated aromatic hydrocarbons like the dioxins and, hepatitis and human immunodeficiency virus. In PCT, porphyrins accumulate in the liver and are transported to the skin by the blood plasma. When porphyrins are exposed to light and oxygen, they are incited by the light. Fragility of sun-exposed skin occurs, leading to lesions typically on hands and forearms and occasionally on face or feet. Patients also may develop sores, blisters, and tiny cysts on the sun-exposed areas and can sunburn easily, have easily traumatized skin, develop mottled brown patches around the eyes, have increased facial hair and occasionally develop hardened skin on the neck, face and/or chest, as well as crusting and scarring of the skin. PCT can also cause liver damage, liver cancer and cirrhosis of the liver. PCT has been associated with the development of hepatocellular carcinoma.

The urine is likely to be reddish or brownish and will glow pink under a fluorescent light. When a skin biopsy is performed, characteristic changes are seen which differentiates PCT from other blistering diseases. PCT is diagnosed by testing the blood plasma, urine, and stool for porphyrins. When 24-hour urine and fecal quantitative porphyrin profiles are performed, there are elevated levels of uroporphyrins in the urine and in the stool, coproporphyrins and uroporphyrins.

The primary treatment is phlebotomy, which involves removing a pint of blood every 1-2 weeks to reduce the amount of iron in the body. Since iron inhibits the deficient enzyme, removing iron via a phlebotomy lowers the porphyrin levels in the liver and plasma. The skin usually becomes normal after five or six phlebotomies. Chloroquine or hydroxychloroquine, which remove excess porphyrins from the liver, can also be prescribed. A person with PCT should also avoid alcohol, iron, estrogens and other factors that cause symptoms. It is important to be careful in the sun and to use appropriate sun protective clothing and opaque sunscreen The APF has samples of the sunscreen, TOTAL BLOCK and a Sun Precautions, clothing catalog for members upon request.

In Memory

We are saddened to hear of the passing of our dear friends. Some of their loved ones have chosen to honor a life by making a gift to the APF. We sincerely appreciate their thoughtfulness and desire to help others with the disease. Please join us in thanking:

Fred Mead for *Rita Mead*; Carl Dean Hughes for *Lillian Hughes*; Maria L. Hollcroft for *John Cicchirillo*; James E. Arzouman for *Susan Arzouman*; Dr. Susan Engel-Arieli for *Tal Dziengiel*; Mary Puccia for *Mary B. Crown*; Donald L. Johnson for *Peggy Lewis Johnson*; Sharon M. Doty for *Robert E. Doty*; Kathleen Toelkes for *Donna Pagano*; Victoria Gehm for *Judith Marie Coley*; The Hobby Family for *Samuel Prosser*; Christine Witherspoon for *Abigail Smelser*; Richard A. Tancreto for *Frank Nicchio*; Annie Depukat for *Patricia Depukat*; Vincent Shell for *Dorothy Shell*.

In Honor

The holidays are a wonderful time to help us advance our mission. Your gifts in honor of the people you appreciate help us continue our educational work and foster research efforts and the search for a cure. Thank you.

Chris Denton for *Bill Carriker*; Kathleen A. Shiel, Eric S. Gray for *Ralph Gray*; Rachelle Brower for *Sarah Kennington*; Elaine E. Sasso for *Elizabeth Britton*; Linda Nagin for *Melissa Nagin*; Joan Steelhammer for *Jann Steelhammer*; John Burkett for *Mira Geffner* and *Edward Geffner*. Desiree Lyon for *James* and *Susie Young, Bill* and *Irene McCutcheon* and *all of the Scientific Advisory Board of the APF.*

Rupa Gill: A Poignant and Courageous Story of CEP

For seventeen years I have lived with CEP (Congenital Erythropoietic Porphyria). CEP is an extremely rare porphyria, which causes excessive facial hair and paper-thin skin. Growing up was very difficult, because I didn't understand what was wrong with me. My parents and I went from doctor to doctor trying to figure out why my skin was ripping so easily, why my face was getting hairy, and why I got sunburned so easily. Time and time again we were left clueless.

After years of unanswered questions, at age six, a dermatologist mentioned Porphyria, specifically Cutanea Tarda but it was not until we reached Dr. Karl Anderson that we found the real diagnosis. He told us I had CEP but that the dermatologist who said it was Cutanea Tarda may have not been very familiar with the specific Porphyrias and that it is very difficult to decipher between the many types. He also said that although there is no cure for CEP, I can help my skin by eating healthy, staying out of the sun, and applying a lot of sun block when I go out. He led us to the American Porphyria Foundation website, and the resources and information we got were more than ever before. Every time even the least detail or advance in research came along, it was on the website. Personally, I felt better knowing that I'm not alone. There are a handful of people who share my same illness.

Even though it took a while, I accepted the CEP and became a much stronger individual. Most importantly, I felt blessed. I felt blessed that I had friends and a family who accepted the "hairy" me, no questions asked. It was this loving and supportive environment that got me through the darkest of days, the days when people would call me a monkey or ask me if I was a boy. If it weren't for this love, I would have succumbed to the disease, and I knew I didn't want that to happen. I immersed myself in activities such as playing soccer, studying hard, spending time with family and friends, and when I started to believe that I wasn't different, everything just fell into place.

Now, realizing how lucky I was to have such support, I want to be the support system for others who have CEP and think that they're alone, because the truth is they are never alone. If they open up their heart and let people help them and help themselves by accepting the disease, they can overcome the disease, so much so that it no longer will be a "disease" in their eyes. I don't want anyone to feel hopeless like my parents and I felt, running from doctor to doctor, when I was young. It is my time to help.

Editor's note: Rupa will be a leader in our new CEP support efforts. She has had many skin grafts and related surgeries and can still write such an article of hope. Rupa's story will appear in its entirety on the APF website.

Attention CEP Families

The APF would like to facilitate the formation of a CEP group. We are also applying for a grant to help support CEP research and to help us host a CEP Family meeting with porphyria experts. We will need family photos and letters from families describing your experiences with CEP. Please contact the APF if you are interested in participating in the CEP Family Group or helping with the research project.

This disease is extremely rare and is autosomal recessive. It is also known as "Gunther's Disease." The deficient enzyme is uroporphyrinogen III cosynthase (or uroporphyrinogen III synthase). As is characteristic of the erythropoietic porphyrias, symptoms begin during infancy. Sometimes CEP is recognized as a cause of anemia in a fetus before birth. In less severe cases symptoms may begin during adult life. Porphyrins are markedly increased in bone marrow, red blood cells, plasma, urine and feces. Porphyrins are also deposited in the teeth and bones.

Skin photosensitivity may be extreme, and can lead to blistering, severe scarring and increased hair growth. Bacteria may infect the damaged skin. Facial features and fingers may be lost through phototoxic damage as well as infection. Red blood cells have a shortened life-span, and anemia often results. Synthesis of heme and hemoglobin are actually increased to compensate for the shortened red blood cell survival.

The APF hopes to help with research funding as there is very little research on CEP.

National Porphyria Registry

Your participation in the The National Porphyria Registry is extremely important to porphyria research. The Registry is NOT the same thing as joining the APF, instead, it is a site which goes directly to the porphyria Consortium of experts.

We need your participation. First, the porphyria researchers will be collecting information from EPP patients, so if you have EPP, please go to the APF website and click on the Porphyria Registry and follow the instructions.

The researchers will continue with other porphyrias, so if you have registered and not heard from them yet, you will in time. Participating in research does not mean you have to take an experimental drug, rather, you can answer questions about porphyria that are very important for research projects and to determine the incidence of porphyria in the US.

The information contained on the American Porphyria Foundation (APF) Web site or in the APF newsletter is provided for your general information only.

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.

What's Important For You

Remember, if you purchase the DVD *Porphyria Live*, we will send your doctor one **FREE**.

Don't forget there are outstanding resources on the APF website: www.porphyriafoundation.com

Purchase the Primary Care/ER Kit for the Acute Porphyrias and EPP, the *Porphyria Live* DVD, and Desiree Lyon's book: *Porphyria A Lyon's Share of Trouble*. All proceeds go to the APF. You can purchase these items via the APF website or contact the APF.

The APF is conducting a survey to discover information about treatment issues for acute porphyrias. If you did not receive the survey, please contact the APF. If you did, please return it immediately. Your cooperation is greatly needed.

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