



President's Report



By: Lois J. Aitken

In the spirit of charity the Canadian Porphyria Foundation opened 2003 **celebrating 15 years** of organizational development and membership growth. The spirit that created the CPF 15 years ago is the same spirit that guides the Board of Directors and National Coordinators, and inspires the staff and volunteers.

We are a group of ordinary people working together with a fair sense of commitment and a lot of compassion. We are dedicated to our mission. We continue to strive for superior **awareness, support** and **education**. We are working hard to bring hope to people diagnosed with porphyria.

The CPF is unique in Canada because we are the only porphyria organization working to "improve the quality of life for people affected by porphyria". Our mission is enhanced as we develop values that encourage community spirit, partnership building and enhance fundraising.

At the CPF office we are amazed at the increased numbers of diagnosed individuals and families contacting us for porphyria support and educational information. We feel that both modern technology and the influx of South African doctors coming into Canada are contributing to this increase. The South African doctors have studied the porphyrias, recognize the symptoms, and test & diagnose their patients.

Once people are diagnosed with porphyria they contact the Canadian Porphyria Foundation, searching for support and educational information that will help them learn how to better manage and cope with their disease. The educational information on the different types of porphyria that medical professionals have prepared for the CPF is a tremendous help; by acquiring knowledge, people with porphyria learn to prevent many of their acute attacks. The prevention of illness decreases doctor's visits, hospital stays, and emergency entrances which results in enormous cost savings to Health Canada.

One of this year's highlights was the creation of the organization's motto, "**Increasing Knowledge – Improving Health**". With the mission and motto foremost in the minds of the Board of Directors, our work is making a difference in people's lives, nationally and internationally.

It is an honor for me to be a part of this growing organization. As I look back over the past 15 years I am thankful for the commitment and support of so many who care.

National Porphyria Day: June 1st

The CPF National Board of Directors have declared June 1st "National Porphyria Day".

Plans for inauguration celebrations on June 1st, 2005, are underway at the national office for Winnipeg, MB; Halifax, NS; Calgary, AB; Vancouver, BC; Saskatoon, SK; Cornerbrook, NF; Montreal, PQ; Toronto, ON; and Whitehorse, YK.

The kick-off theme for 2005 will focus on "**children with sun-sensitive porphyria**". To brighten the hearts of these children who cannot tolerate sunlight, organized fun events will include wearing sunglasses and hats to prevent acute attacks and provide skin protection. Goofy sunglasses and wacky hats will be welcomed!

In the sun-sensitive porphyrias, the liver can be affected and a liver transplant is occasionally necessary. Not only the rays of the sun can cause extreme pain and severe skin damage to the individual; other factors are ultraviolet and bright lights, operating room lights, and incubator lights.

One newborn baby suffered 3rd degree burns caused by the incubator lights before her doctors and specialists discovered she had Congenital Erythropoietic Protoporphyrria. Other light-sensitive porphyrias are Erythropoietic Protoporphyrria and Porphyria Cutanea Tarda.

Declaring a National Porphyria Day means people all across Canada will celebrate annually the mission of the CPF; "...to improve the quality of life for those people affected by porphyria".

Lumitene & Porphyria

The sun-sensitive porphyrias can very often be treated with Lumitene.

The original treatment formula for this form of porphyria was known as Solatene, a prescription-only beta-carotene capsule that is no longer available. Lumitene has taken its place and is 100% compatible and the identical formulation of Solatene but does not require a prescription in the USA – *although you should certainly discuss this with your doctor.*

Each Lumitene capsule is composed of beadlets containing 30mg beta-carotene. Patients receiving Lumitene should be advised against taking supplementary Vitamin A since Lumitene administration will fulfill normal Vitamin A requirements. You should also be cautioned to continue sun protection and be forewarned that your skin may appear slightly yellow while receiving Lumitene.

Lumitene may be administered either in a single daily dose or in divided doses, preferably with meals. The usual dosage for children under 14 is 30 to 150mg (1-5 capsules) per day. Capsules may be opened and the contents mixed in orange or tomato juice to aid administration. The usual adult dosage is 30 to 300mg (1-10 capsules) per day.

Dosage should be adjusted depending on the severity of the symptoms and the response of the patient. Several weeks of therapy are necessary to accumulate enough Lumitene in the skin to have a noticeable effect. Patients

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should be instructed not to increase exposure to sunlight until they appear carotenemic (first seen as yellowness of palms and soles). This usually occurs after 2 to 6 weeks of therapy. Exposure to the sun may then be increased gradually. *The protective effect is not total and each patient should establish his/her own limits of exposure.*

Note:

To receive a shipment of Lumitene in Canada, you (through your doctor) will need to obtain a "Special Access Request Form" from Health Canada. For more information on requesting a Special Access Request Form from Health Canada, call, fax or access their web page:

Call Centre: 613-941-2108

Fax Number: 613-941-3194

Web Page:

<http://www.hc-sc.gc.ca/hpb-dgps/therapeut/htmleng/sap.html>

Equivalent Pharmaceutical Industries Corporation (EPIC) is the exclusive sales agent for Lumitene.

Long Distance Number: 1-800-866-0978

Local Number: 1-516-997-1005

Fax Number: 1-516-997-3660

Web Page: <http://www.epic4health.com>

Yes! I want to help the CPF continue to improve the quality of life for people affected by Porphyria!

We would like to thank all of our readers for their contributions to the Canadian Porphyria Foundation in the past, and would like to remind you that your continued support is needed. Contributions from concerned individuals help to ensure the ongoing success of the foundation's work to provide support, guidance and education for porphyric sufferers. By making a tax-deductible contribution today, you can ensure the continuation of these important efforts. Please call our toll-free number OR complete this form and fax or mail it along with your cheque or credit card information:

Gifts made using a credit card can be made by phone by calling **toll-free, 1-866-476-2801**.

Gifts made using a credit card can also be faxed to **204-476-2800** or mailed to the address below along with the form: —>

**The Canadian Porphyria Foundation
P.O. Box 1206 Neepawa, MB, R0J 1H0
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Gifts made by cheque (made payable to the Canadian Porphyria Foundation) should be mailed directly to the Canadian Porphyria Foundation's national head office.

Lyon's Share



By: *Desiree Lyon*
American Porphyria Foundation Executive Director
Alabama, USA

Many patients call with a lengthy list of symptoms and want to know if they fit a porphyria attack. After explaining that there are different types of porphyria, I then relate the fact that Acute Intermittent Porphyria (AIP), Hereditary Coproporphyrinuria (HCP), and Variegate Porphyria (VP) imitate the symptoms of countless diseases. Since these exhibit the most confusing symptoms, I would like to focus on them.

Severe abdominal pain appears to be the most classic symptom. In fact, some medical professionals have deemed these porphyrias as the “tic-tac-toe” diseases, because some patients have endured so many exploratory surgeries that they have a “tic-tac-toe” pattern on their abdomen. Many patients also experience severe abdominal swelling.

The abdomen is not the only region affected. Back and leg aches are also a major complaint. The increase in Porphobilinogen (PBG) appears to affect nerve endings and cause numerous neurovisceral and neurological symptoms. ALA and PBG must be positive if AIP or HCP are indicated.

Other classic symptoms include increased blood pressure, rapid pulse, constipation, nausea & vomiting, urinary retention, and excessive sweating. Some patients develop peripheral neuropathy and experience muscle weakness ranging from general malaise to paralysis. Respiratory paralysis can occur and is extremely dangerous.

Others experience a loss of feeling in various parts of the body. Depending on the severity of the attack, these symptoms usually subside as the attack diminishes.

Salt and water levels in the body can also be affected depending on the severity of the attack.

BECOME A VOLUNTEER TODAY!

The CPF is always looking for new volunteers to bring exciting ideas to the foundation! Discover the joy of volunteering today by:

Calling: **1-866-476-2801** or
E-mailing: **porphyria@cpf-inc.ca**

Remember, your assistance will help to promote porphyria awareness!



We gratefully accept any & all gift donations. The CPF is a registered charity, which means your donations are recognized with a tax receipt!

Neurological symptoms vary greatly from agitation and confusion to hallucinations. It does not appear, however, that porphyria causes chronic psychiatric problems. The neurological changes generally diminish as the porphyria attack subsides.

The severity of an attack often depends on the individual's exposure to unsafe drugs, menstrual cycle, fasting, alcohol, infection, or a combination of these factors. Patients with Acute Intermittent Porphyria (AIP) do not exhibit the cutaneous problems associated with VP and HCP.

In VP and HCP, stinging, burning, redness, blistering, and scarring can occur in sun-exposed areas of the skin. The phototoxicity can be extremely severe, and patients in the riskier range must be protected from the light. I have received information from patients who have had to change jobs because of a light problem at work. Others have procured special permission to cover their car windows. One APF volunteer is so photosensitive that she wears a hooded cape when she ventures outside.

Since the range of symptoms is vast, it is important to document an attack. For example, one doesn't want to misdiagnose appendicitis as a porphyria attack or vice versa. Checking the PBG/Aminolevulinic Acid (ALA) levels can help differentiate between the two.

If muscle weakness develops during an attack, complete recovery may take months to several years. An increased pulse rate is also an indicator during an attack. Most patients recover completely from an attack.

Severe attacks are not common now because patients who are diagnosed avoid precipitating factors. Also, most porphyrias are diagnosed before a patient reaches that point.

Please be careful with your information source. Complex questions are not generally answered with simple solutions. The porphyrias are RARE! Thus, there are few physicians with expertise in this group of disorders. SEEK their help!

“Lyon's Share” is reprinted with permission from Desiree Lyon and the American Porphyria Foundation.

Carly Anne's Story: The Girl with the Big Purple Hat



By: Kim Moya
California, USA

I'm only 5 years old and was diagnosed with Erythropoetic Protoporphria (EPP) two years ago. I couldn't get in a hot car without my hands feeling like they were on fire, so my mom and dad thought I was just a cranky child. That changed when I scratched my hands and face until there were scabs left. We went to our wonderful pediatrician, Dr.

Adrienne Tuch, who works with Kaiser Permanente Medical Centre in Northern California. My hands looked odd, so she immediately referred us to Dr. Pham, another wonderful physician. Coincidentally, Dr. Pham only worked in that facility two days a week and was based 50 miles away in a different facility, so for her to have been there that day was a miracle.

Dr. Pham took a look at my face and hands and brought in a big book with a scary picture of a person with a case of EPP worse than mine. Before we left her office, she gave my mom a referral to the genetics department, a prescription to be tested, and an explanation that porphyria was hereditary. The tests were positive for EPP, so my dad and sister were tested too, especially since they both complained of itching and sensitive skin. Sure enough, my dad was diagnosed with a less severe case of EPP than mine. My sister's protoporphyrin levels were so low that they didn't believe she had it, but my mom still wonders since she's extremely sensitive to the sun and will only wear long-sleeved shirts in the hot summer.

Kaiser sent us to Dr. John Epstein at UCSF Medical Centre in San Francisco, who happens to be on the American Porphyria Foundation (APF) medical advisory board. He confirmed the diagnosis but said that beyond beta-carotene/

Lumitene, there wasn't much more that we could do except wear protective clothing and be careful about sun exposure. Nonetheless, mom is an information junkie, so she got online and joined the APF and EPPREF Foundations, printed out all the information she could find, and is still trying to help me. I'm in my second year of preschool, and I wear either a big purple hat or a safari hat every day of the year. My friends remind me if I forget to wear it and, being 5 years old, I still look kind of cute in it! Mom is also even thinking about writing a children's book that describes my condition from a child's point of view. She is also preparing me for a full day of kindergarten next year, including requesting the classroom closest to the playground, so I can go inside during recess when I need to get out of the sun. Plus, I can quickly run my hands under cold water or put them on ice, which works best to relieve the burning on the tops of my hands.

I've tried all kinds of gloves, fans, creams and ice products, including one of my favorites - a chilled and insulated beer stein that I put my hands in while we're driving in the car or at my brother or sister's baseball games. Mom just ordered a solar-powered fan that she saw in an airline magazine that clips onto the car window and keeps the inside of the car cool. She also got a handicap placard and window tinting for our van to block the spectrum of light rays that affect my skin.

I had a really bad attack last 4th of July, even though I was in the shade all day. It was so bad that Dr. Tuch had to prescribe Tylenol with Codeine and Benadryl. It calmed me down enough to get some sleep, but my parents were really worried since they couldn't make me feel comfortable no matter what they did.

When we got a list of sunscreens with ingredients like Titanium Dioxide and Zinc that might help, we promptly went to the Clinique counter and bought \$50 worth of them. Mom and dad do everything to make me feel more comfortable because when I'm uncomfortable, everybody in the family is affected.

We haven't been to Disneyland, the beach, or any place where the sun shines regularly since I've been diagnosed; we always try to stay inside during the afternoon and most of the day during the hotter months. We've even been talking about moving to Oregon or Washington State, because there's more rain and less sun. After all, we live in Sunnyvale, California, which explains the weather year round!

My liver numbers are stabilizing so I see Dr. Abramson less frequently now. He is our liver specialist, who happened to work in his early years as a doctor with Dr. Montgomery Bissell from the APF board. I am also anaemic, but after taking iron for six months with no change in my iron levels, it isn't bad enough to worry about. I do get tired a lot, but that could be from being in the sunlight for more than an hour. My family and I LOVE it when it rains for days on end. I especially like it because my hands have time to heal. When it's gloomy

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Drugs & Porphyria



*By: Professor Michael R. Moore
Queensland, Australia*

From the outset of the understanding of the diseases that are called the porphyrias it was recognised that drugs and chemicals could have a pronounced effect on the acute forms of these diseases. Exposure to the sedative sulphonal provided the first publication of an attack of Acute Intermittent Porphyrin by Stockvis in 1889. Thereafter, numerous others were able to demonstrate that a range of pharmaceuticals and chemicals have pronounced impacts upon haem biosynthesis and hence, upon porphyria. It is for that reason that the acute porphyrias are known as toxagenetic diseases - diseases with a genetic basis which show an idiosyncratic reaction to a range of drugs and chemicals. It should be emphasised that this impact upon acute porphyria is not mirrored by a similar impact of the effects of the same compounds in the non-acute porphyrias, Porphyrin Cutanea Tarda, Erythropoietic Protoporphyrin, and Congenital Porphyrin. None of these demonstrate the neuropsychiatric features of acute porphyria. However, similar drugs and chemicals might increase synthesis of porphyrins in these conditions, but the life threatening acute attack will not be seen as a consequence of drug utilisation in the non-acute porphyrias. Pathologists state that the relationship to the impact of drugs refers exclusively to Acute Intermittent Porphyrin, Hereditary Coproporphyrin, and Variegated Porphyrin.

The following are three ways in which one can identify drugs as inducers of the acute attack:

- Clinical evidence in patients with acute Porphyrin. This includes anecdotal self-reporting by patients.
- Studies of drug impacts in all culture systems. This includes titration of effect against known inducers.
- Assessment of effects in whole animals. In some cases this can include priming of the animals with a known porphyrinogen like phenobarbitone.

Of these the **“gold standard” is clinical evidence**, but even that suffers from patient variation. One drug that can cause a life threatening attack in one patient may be tolerated by another. This variability in human response clearly complicates our capacity to identify safe or dangerous drugs.

The history of development of the drug list that we all commonly use at the present time probably dates back to the work done by Lennart Wetterberg of Sweden in the 1970's. By 1976 he had prepared listings of drugs thought to be unsafe in porphyria, which brought together a number of different centers and their knowledge of the likely consequences of the uses of these compounds. This level of international cooperation continued with the publication in 1979, subsequent to the conference in Buenos Aires, of an international list of drugs thought to be safe and unsafe in acute porphyria; this subsequently led to the formation of the Committee On Review of Porphyrinogenesis (CORP).

Special thanks to the following organizations for generously providing financial assistance to the CPF:

- ⇒ Manitoba Community Services Council
- ⇒ Power Corporation of Canada
- ⇒ Neepawa & District United Way

International collaboration continues to the present with the most recent collaborative processes being coordinated by Professor Stig Thunell in Stockholm. The results of these deliberations are dependent upon experimental, clinical and anecdotal evidence. This is largely because there is a considerable subjective element in recommendations made with respect to these pharmaceuticals. It is not uncommon for those with expertise in this area to disagree with some of the recommendations contained in the lists. For this reason these are usually presented as recommendations, leaving the responsibility for final decisions on drug use on the shoulders of both patient and physician. One of the difficulties that the authors of the list has to face is the numbers of new drugs introduced annually to the pharmacopoeia combined with the relative paucity of evidence regarding the effects in human patients afflicted by a relatively uncommon disease.

In this brief note I am not attempting to delve into all of the factors that contribute to drug porphyrinogenicity - that is, the capacity of pharmaceuticals to induce haem synthesis and consequently induce acute attacks of porphyria. These are some pointers to compounds that might be likely to be inducers such as those metabolised by the haem-requiring cytochrome p450 enzyme systems. In these, the increase in requirements for the cytochrome p450 increases demand for haem and consequentially induces the biosynthetic pathway. However, we are generally unable to predict whether or not a compound might be porphyrinogenic. Within classes of pharmaceuticals like the barbitates - we are able to predict that all will be inducers, but even there the degree of induction is difficult to predict.

A critical component of our evaluation of drugs is the collection of anecdotal evidence from patients. This is usually quite difficult because the effects in the individual patient who is often taking more than one drug are difficult to dissect. None the less, as bodies of evidence accumulate with large numbers of anecdotal reports, it is possible, on the basis of commonality of information, to supplement the experimental evidence and provide useful recommendations on the safety of drugs. Frequently, the effects of drugs can be additive. This means that where people are receiving prescriptions for drugs for a series of indications, the individual modest effects of these drugs upon humans may be cumulative. It has been seen to be particularly true in the treatment of epilepsy. One cannot neglect to note the substances that are not generally thought of as drugs contain within them the capacity to modify in haem synthesis. This means that those who smoke, who consume alcohol, or who use herbal medicines, may

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Drugs & Porphyria

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be contributing to the effect of pharmaceuticals taken at the same time.

In the development of collections of anecdotal material it is essential that note is taken on the type of porphyria involved and whether or not an accurate diagnosis of porphyria has been made. Information on safe drug use in Porphyria Cutanea Tarda does not inform us of anything in relationship to any of the acute porphyrias. Equally, persons with some features of acute porphyria, such as abdominal pain, need appropriate biochemical diagnosis of the porphyria before any evidence can be given to the report of adverse effects. Even those with well-documented acute porphyria can suffer from other conditions unrelated to porphyria such as abdominal pain from a gastrointestinal infection.

Finally, there are circumstances where there is an inevitable need to use a drug, likely to be dangerous in porphyria, to treat a pre-existing medical emergency. In these circumstances one has to examine the balance of probabilities. Balance the fact that there may be a pressing need to treat that other condition for which the consequences are dire against the possibility that the induction of an acute attack of porphyria might occur. The common situation in which this occurs is chemotherapy of cancer in which drug cocktails are commonly used, some or all of which are likely to be in uses of instances and that would be likely to induce haem synthesis. The medical indications for treatment of cancer are usually more pressing than the possibility that the acute attack might occur. In these circumstances the patient and their physician must jointly make the decision on how to proceed with treatment.

For those requiring access to drug lists there are a number of websites that you will find useful. Such as those published by:

Ourselves at: <http://www.entox.uq.edu.au/porphyria>.

From the unit in the University of Cape Town at:

<http://www.uct.ac.za/depts/porphyria>.

And from the unit in Paris at: www.porphyrries.com.fr.

Thank You

The CPF would like to give a special thanks to all of our nation-wide ticket sellers. Your volunteer work has helped increase awareness and funding. Thank you for your efforts and much appreciated support!

Carly Anne's Story

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around here, I also don't scratch my hands as much as I do on sunny days. Instead of having gashes that don't heal well, my hands are fine.

My mom says that my form of porphyria is much milder than many of the other kinds. We're grateful that it's only affecting my hands and face and that **doctors are working on a cure**. Then we could live normally and enjoy sunny California.

The year I was diagnosed with EPP, we exchanged Christmas gifts with other EPPers (that's what my mom calls us)! It meant so much to not only email, but to feel so close to others suffering from the same illness. There is definitely safety in numbers and being able to chat with others has made all the difference to my family since I was diagnosed. We are always looking for other people (especially children) with all forms of porphyria, and would love to hear from you. Feel free to email me and my mom at abdesign@aol.com. We'll be sure to write ASAP!

"Carly Anne's Story" is reprinted with permission from the Moya family and the American Porphyria Foundation.

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