



## Canadian PKU & Allied Disorders Inc. Newsletter

September 2011

In This Issue  
Welcome to CanPKU  
PKU Profile  
Hat's Off To...  
Ask Us!  
Travel Scholarship  
CanPKU Growth  
New with Kuvan  
Advocacy 101  
Become a Member!

### Welcome!

CanPKU would like to welcome the following individuals to our organization:



**Jennifer Ballagh** was elected as a new **Director** of CanPKU at the annual general meeting in July, when the Board of Directors was expanded from five to seven members. She has donated her time and professional photography services to CanPKU since 2009 and represented CanPKU at the annual workshop of the London clinic. She is Mom of 4 year old Sydney, CPKU and 9 year old Amelia and lives in London, Ontario.

**Frances Goodfellow** is a Certified Management Accountant (CMA) by

Dear Nicole,

Thanks for taking the time to stay up to date on what's new with CanPKU and PKU in general. I'd like to take a minute to address the notion that CanPKU is 'Kuvan-centric'. Some people perceive us as being puppets of BioMarin, the company that produces Kuvan and I want to assure you that this is not the case.

Yes, BioMarin is our biggest sponsor and without their funding CanPKU would not be able to do all that we do. And yes, we believe that Kuvan is a useful tool for PKU management for some patients and we believe fully that it should be covered for every patient in Canada who can benefit from it.

But we are not focused on only getting Kuvan covered. Some provinces do not provide funding for the necessary low-phe foods (BC, SK & PEI) while others do not even have a metabolic clinic doctor (SK & PEI)! Others only provide formula for children (MB, NL & PEI). We are committed to ensuring that all patients in Canada have access to all the tools in the toolbox so that those living with PKU have the best chance at success. This means fighting for clinic care, low phe food & formula coverage as well as Kuvan. Ensuring that we have access to these tools will help ensure that we have access to future tools such as PEG-PAL and the home phe monitor.

This newsletter is loaded with information about Kuvan because that is what is new and happening now. We will bring you news about other treatments, coverage and care issues as they arise!

Sincerely,  
Nicole Pallone  
Mom of Rosie, 3, CPKU & Carmella, 9 mos  
Vice President, CanPKU

### 2011 Remaining Events:

#### **September 18th: BC PKU Day, Vancouver, BC**

The day will include a walkathon around Langara Golf Course as well as lunch and an education session at the Chan Auditorium at BC Children's Hospital (Child & Family Research Institute).

At the education session you will hear about the very first PKU patient ever treated with a low-phe diet in a presentation by John Adams, CanPKU's President. You will also hear from Dr. Louis Wolf himself, the developer of the first low-phe formula. Each participant will be able to choose 2 of 4 mini workshops to attend and we will wrap up the day with

profession and has signed on as our new **Treasurer**, a great relief to acting Treasurer John Adams. She is mom to Hailey (PKU) and Brandon and lives in Bolton, Ontario.

**Wilma McCormack** resides in Ottawa, and has two children, one with PKU. In addition to being a PKU-mom, Wilma works at Algonquin College in the role of Program Quality Assurance Administrator, and is the new **CanPKU Membership Chair**, relieving Chairman Cristian Baigorria of his double duty.



**Brienna Young** was elected as a new **Director** for CanPKU at the annual general meeting in July. She is mom of 4 year old Hayden, CPKU, and also a full time nursing student. In her spare time (??) she educates other nurses about PKU. You can read more about her in the article about our first Travel Scholarship Award!

**Welcome aboard everyone!**

**PKU Profiles**  
**Niall Quinn**

an expert panel Q & A session. There will also be childcare provided and a silent auction with some great items!

### **October 15th: Quebec PKU Day, Montreal, PQ**

The Canadian PKU and Allied Disorders non-profit will host its third annual Educational Day in Montreal, on Saturday, October 15th from 11:00 am to 3:30 pm, in the Banquet Hall of the Centre communautaire intergénérationnel d'Outremont, at 999 McEachran Avenue, Montreal (Outremont) H2V 3E6. A light lunch buffet will greet participants on their arrival and a supervised daycare will be provided for the little ones.

Speakers include Dr. Jerry Vockley who will present on the new and emerging treatment options for PKU, and Dr. Christiane Auray-Blais who will share with us her expertise in mass screening for inborn errors of metabolism in the Province, and inform us of the new development in this area, and their impact on PKU. After a Health Break, participants will have the opportunity to ask questions about PKU to a panel of experts comprised of clinicians and patients willing to share their personal or professional experience.

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### **CanPKU Awards First Travel Scholarship!**

This year CanPKU President John Adams and Vice-President Nicole Pallone dedicated to put part of the budget towards travel scholarships. These scholarships help PKU families get to CanPKU events when they otherwise couldn't. We realize that PKU families often live far away from a major city but with such a rare disorder the numbers aren't there to warrant events in more remote areas. We also realize that PKU affects families of all different means and we want everyone to have a chance to attend. The first ever CanPKU Travel Scholarship was awarded to Brienna Young, a BC resident, full-time Nursing student and single mother of her 4 year old PKU son Hayden. With the scholarship and a great deal on airfare, Brienna, Hayden and Brienna's boyfriend Josh were able to fly out to Ontario to attend the Polo-4-Kids event and AGM in Alliston, ON. Brienna had this to say about the experience:

" We live in Kamloops, B.C. which is about 4 hours north-east of Vancouver. Since both Josh and I are full time nursing students, we would not have been able to attend if it wasn't for the generosity of CanPKU.

Hayden was so excited to go on the airplane... Ok ok so was I! We talked about it nonstop for over a week when we found out we got the scholarship. Hayden got to pick out his own blue carryon luggage which had wheels. We stuffed it full of low protein snacks, his scale and formula... and many toy cars. When we got to Toronto we stopped at the Sick Kids Hospital and went to the Specialty Food Shop. It was *huge* compared to our specialty food store at the BC Children's Hospital. There was even a freezer with cold items like CBF cheese slices and tweeks! In Vancouver we only get dry items like pasta, cereals and cookies. I think



## Quinn

Niall was born on a beautiful autumn day in Happy Valley-Goose Bay in Labrador on October 9, 1974 before newborn screening had become routine in the area. At birth he was a beautiful, bright and healthy 9lb 8oz baby. Niall was breastfed and he continued to thrive and progress at an apparently normal rate. However, shortly after he was brought home, Niall began crying almost all of the time. His parents took him back to the hospital several times and each time they were told it was colic. "There were times when my husband left for work in the morning with my rocking Niall," says Niall's mom, Doreen, "and when he came home eight-plus hours later, I was still there."

When her son was 3-4 months old, Doreen noticed that Niall's progress had slowed - in fact behaviours present at two months were no longer present. By the time Niall was six months, the family had moved to Stephenville on the west coast of Newfoundland and Labrador and began taking him to the doctors because of concerns over his development. It was six months later before a doctor finally agreed and referred the family to a visiting pediatrician, who diagnosed Niall with mild Cerebral Palsy, with possible mild mental retardation.

The rest, as they say, is history. Niall experienced a severe "failure to progress", and as he approaches his 37th birthday he has no communications skills and can do little without assistance. He can walk and

the cashier thought we were a little bit crazy because I was taking pictures of Hayden next to the big aisles full of low protein food. But it was the biggest selection I'd ever seen!



It was exhilarating to watch Hayden play with other PKU children for the first time. He understands which foods he can and cannot have, but he is only just beginning to understand why. Hayden was very excited to meet other children who also had to 'weigh their food'. In addition, he was able to try new foods which were made by a fabulous chef, Gina Valente, who is a knowledgeable and talented chef, as well as a mother of a child with PKU. She educated us about the difference between synthetic and natural proteins, the importance of adding DHA and omega fatty acids to his diet, and opened my eyes to experimenting with new ingredients that I would never have thought to use.

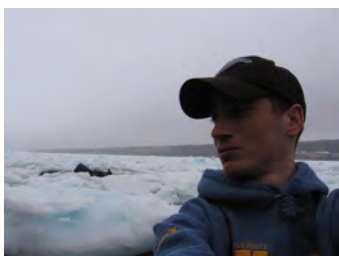
I learned so much more than I thought I would at this event. We learned about Kuvan, PegPal and were lucky enough to hear from Dr. Barbara Burton. I'd seen her presentations on YouTube, so it was very exciting to see her in person! Being a nursing student with a passion for metabolic nursing, I thought I knew all there was to know about PKU. However, there is only so much that a text book or articles can teach you. Hearing from other families and older individuals with PKU really impacted my thinking. One individual described what it felt like when he went off diet, even for a day. He said it was like a "3-5 day mental hangover" which he could not shake off no matter what he did. I also learned that Kuvan didn't just lower phe levels, but it can also help to lift the mental fog and increase clarity and concentration. This was a eureka moment for me because I often witness in Hayden a certain inability to concentrate on the task at hand.

Attending the CanPKU event and meeting other families has drastically changed my perspective of Hayden's PKU. The information I learned from other families cannot be found in a clinical text book, and now I see the knowledge gaps that I had before. I have asked Hayden's physician about starting Kuvan to help with his concentration and behavior issues. In addition, I find myself being more patient with Hayden because I now have a better understanding of his challenges.

feed himself if the food is put on the table for him. He goes out for respite care 35 hours per week but is somewhat agoraphobic and does not take well to other outings. Aside from that, one of us has to be here in the house with him at all times. He is also subject to two types of seizures.

A second son, Michael, was born February 10, 1986 and had none of the same issues.

## Brian Quinn



In a sometimes cruel contrast, the Quinn's third son Brian was also born on a beautiful autumn day (October 7, 1987) and weighed even more than Niall (10 lbs. 5 oz.) and was also bright and beautiful at birth. However, today Brian is an intelligent and personable young man.

Brian was also being breastfed as a newborn when the word came that he had this unpronounceable disorder. "Because Brian's doctor and dietitian were both knowledgeable and progressive, I was able to continue breast feeding once Brian went on formula," says Doreen.

It wasn't until this point, with Niall 13 years old, that he was properly diagnosed with PKU.

As a baby, toddler and child Brian's levels remained low and relatively stable. He progressed well in school and did very well in French, Math, English and Drama. He is very athletic and has played baseball & basketball and loves a good game of tennis. He was also on his junior high cross country running team, and is an accomplished swimmer. Brian progressed through the Canadian Sea Cadet program and continues to work with the program as an officer. Following high school, he successfully

I truly believe that knowledge leads to better health and I couldn't have asked for more knowledge in Alliston!"

We are thrilled that Brienna got so much out of the experience and that we are able to offer travel scholarships for the BC event as well. Unfortunately, sustaining the travel scholarship idea may be difficult. We can't guarantee that we will have funds available next year to do the same but we thought it was worthwhile to see how much of a need there was.

If you are interested in donating funds specific to a National Travel Scholarship Fund, please contact Vice-President and Director Nicole Pallone at [nicole.pallone@canpku.org](mailto:nicole.pallone@canpku.org).

## CanPKU is Growing!

At our first 'annual' AGM (okay, it will be annual from now on!) Vice President Nicole Pallone reported on the growth of CanPKU. Yes, we have more funding but the more telling sign of growth is our increase in membership. Records from the first couple of years are spotty but to the best of our knowledge, the breakdown goes like this:

December 31, 2008: 13

December 31, 2009: 27

December 31, 2010: 44

August 31, 2011: 83

An increase in membership is great news because it boosts our commitment to the PKU community and also makes us more credible in the eyes of the government. Encourage your friends and family to become members!

## What's New with Kuvan?

On April 30, 2010, Health Canada issued a Notice of Compliance to BioMarin Pharmaceutical Inc. for the drug product, Kuvan.

Kuvan is a synthetic formulation of BH4, the co-factor for the enzyme phenylalanine hydroxylase (PAH). PAH hydroxylates Phe through an oxidative reaction to form tyrosine. In patients with PKU, PAH activity is absent or deficient. Treatment with BH4 can activate residual PAH enzyme, improve the oxidative metabolism of Phe, and decrease Phe levels in some patients. You need to talk to your clinician to see if Kuvan<sup>TM</sup> is right for you or your child. The only way to find out if you or your child is a responder to KUVAN is by trying KUVAN.

Some patients respond to Kuvan with a phe reduction, others experience a phe tolerance increase (with a reduction in the amount of medical foods needed) and some experience neuro-cognitive improvements. Everyone is different.

Over 100 patients to date in Canada have been tested for response on Kuvan and BioMarin is actively seeking provincial reimbursement for Kuvan. In the US, over 3000 patients have been exposed to Kuvan. BioMarin is also conducting several clinical trials in Canada with Kuvan.

completed the two-year French Immersion program at Université Ste. Anne in Church Point, Nova Scotia. His ambition is to become a police officer and he is currently working towards that goal. His major PKU issues these days are diet management and a failure to gain weight well. For nearly his whole life, he has been underweight - strong and fit, yes, but very much on the slim side.

The Quinn family is a remarkable example of the difference that newborn screening makes in the lives of those with PKU and the stark contrast of the quality of life between Niall and Brian is testament to the importance of early treatment and diagnosis.

### Hat's Off To...

Carol Hartnett, the head dietitian at BC Children's Hospital who retires at the end of September.

She touched the lives of many with her compassion and her wisdom. She will be sorely missed but we wish her all the best in this new stage of her life!

### ASK US!

"I'm curious about Kuvan for my son but since the Bh4 load when he was a newborn didn't really drop his levels, I don't think he will respond. Is this true?"

**The short answer, as confirmed by Dr. Sylvia Stockler, head of the BC Children's Hospital Metabolic Program, is that no - this is not true. One small dose of the Bh4 isn't a reliable test for the effectiveness of Kuvan. Patients need the proper**

[The Effect of Kuvan on Neurocognitive Function, Blood Phenylalanine Level, Safety, and Pharmacokinetics in Children with PKU \(PKU-015\)](#) is a seven year study designed to evaluate the long term efficacy of Kuvan in preserving neurocognitive function in children with PKU when treatment is initiated at 0-6 years. The PKU-015 study still has a few open spots in the ages 2-4 category. [The Safety and Therapeutic Effects of Sapropterin Dihydrochloride on Neuropsychiatric Symptoms in Phenylketonuria \(PKU\) Patients](#) (PKU-016 trial) is designed to evaluate the therapeutic effects of Kuvan on the symptoms of ADHD and on global function compared to placebo, in subjects with a blood Phe level reduction after treatment, in patients aged 8 years and older. If your centre is not a clinical trial site or you are not eligible for the trials, you might consider the Kuvan Assistance Program.

BioMarin Pharmaceutical (Canada) Inc. is committed to help Canadian Patients obtain access to KUVAN. The Kuvan Assistance Program (KAP) has been established to meet the needs of Canadian patients and health care professionals in optimizing access to KUVAN. The program is sponsored by BioMarin Pharmaceutical (Canada) Inc. and managed through a partner, Innomar Strategies Inc., which provides services offered by the program.

KAP is a private, confidential, and free patient support service for KUVAN patients and their families, caregivers, or health care professionals. It offers assistance with drug plan coverage and reimbursement, enrollment in a Starter Program (a free 30-day starter supply of KUVAN), support in managing KUVAN treatment, and further information on PKU and KUVAN.

Patients must sign a consent form agreeing to be enrolled in the Kuvan Assistance Program. The doctor will complete the form and fax it off.

Once the Kuvan Assistance Program receives the completed Enrollment Form, the Nurse Case Manager will work with the health insurance plan to submit all required paperwork. The Kuvan Assistance Program will provide updates as the process moves forward. To contact the Kuvan Assistance Program, call toll free: 1-877-597-6744, Monday through Friday, 8AM to 8PM EST.

### Advocacy 101

Advocacy is verbal support or argument for the purpose of instigating action. It is not activism; it is grounded in logical, rational thought. There are differences between awareness, education, and advocacy - only advocacy is about tangible change.

In each newsletter we will provide simple tasks that everyone can do to further the cause, and tips on how to be an effective advocate.

Previous suggestions included writing a letter to the editor of your local newspaper and also writing a letter to your local MP. Now it is time to pull out the big guns: start writing letters to your provincial government Minister of Health. Keep writing until you get a response!

Tips to make it effective include making it personal and keeping it simple. Tell your PKU story: how it has affected you and your loved ones as well as what needs to be done to make life better for you and yours. Explain a little bit about what PKU is, but keep it simple. Make sure your letter is specific to what needs to be done in your province. (For

dosage by weight for at least a month before an assessment can be made about whether or not a worthwhile response has occurred.

Additional information was provided by Elaina Jurecki, the Nutritionist for Biomarin

"Some patients may have a mutation at the binding site of the PAH enzyme where BH4 connects to activate the enzyme, and when a greater amount of the BH4 is available to saturate the binding site, it can immediately increase the activity of the enzyme. Others could have a mutation in the structure of the PAH enzyme. When you provide BH4, it seems to help stabilize the folding structure of the enzyme allowing it to achieve a configuration that looks more like the normal PAH enzyme. When the PAH enzyme 'looks more normal', your body is less likely to degrade the enzyme, allowing it to be around longer to oxidize more phe to tyrosine. This process tends to take more time where you will see a gradual lowering of blood phe."

Do you have a PKU related question that you are having trouble finding the answer to? Send an email to [tonicole.pallone@canpku.org](mailto:tonicole.pallone@canpku.org) with "Ask Us" as the subject: we'll do our best to find the answer!

information on this, email us or check out the 'resources' section on our website for a summary of what is covered in each province.)

**Have you already written those letters? If you have a child with PKU in school, contact the administration to see if you (or us!) can be part of a professional development day and educate the staff about PKU. These teachers will know what to expect if your child is in their class and will prepare them for anyone else they come in contact with who has PKU. With any luck, they will educate others!**

As with any of your advocating efforts, we're here to help. Feel free to call or email us for assistance in this regard.

*Please note: CanPKU fully supports each of its members being personal advocates; however, any communications that show affiliation with CanPKU including but not limited to the use of the CanPKU logo, CanPKU letterhead or your title as a CanPKU member means that the communication must be pre-approved by the CanPKU President or Vice-President.*

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## Become a Member!

The more members we have the greater our voice is when advocating for better treatment coverage and care.

New members will receive one free issue of National PKU News, courtesy of Virginia Schuett!

By becoming a member you are showing your support for CanPKU to accomplish its goals, which include:

- Creating awareness about PKU and other inherited metabolic disorders;
- Providing a supportive community for those living with PKU and other inherited metabolic disorders;
- Increasing opportunities for PKU families and others to attend educational and networking events;
- To improve the lives of people living with PKU and other inherited metabolic disorders;
- Promoting and supporting research; and
- Advocating for increased treatment coverage across Canada.

Becoming a member includes the following benefits to you:

- Direct contact from CanPKU regarding newsletters and event invitations;
- Direct access to new information regarding treatments, research and Provincial/Territorial advocacy campaigns;
- Support from other individuals and families who understand;
- Reduced registration fees for all CanPKU events;
- Priority access to travel bursaries for CanPKU events, when available;
- Opportunities to volunteer and make a difference;
- Tips on advocacy and creating awareness in your community; and
- Voting rights at Annual General Meetings and Special Meetings.

To become a member go to <http://www.canpku.org/become-a-member>.

Visit Our  
Sponsors



Dear Reader;

Please feel free to forward this newsletter on to your contacts. CanPKU is always looking for volunteers, members and corporate sponsors. For more information about our organization and other PKU resources, please visit our website at [www.canpku.org](http://www.canpku.org).

Sincerely,  
Nicole Pallone, Vice President

Canadian PKU & Allied Disorders Inc.



Because Knowledge Leads to Better Health

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