



CanPKU News

Issue No.14

Canadian PKU & Allied Disorders Inc. Newsletter

December 2013

In This Issue

Quebec PKU Event 2013

MSUD Profile

Featured Recipe

Hat's Off To

U of Liverpool Study

Suggestions for 2014?

Advocacy Updates

2014 Event Series

50 Years of Newborn Screening

Become a Member

Dear Reader,

I know I always seem to say that it is hard to believe a full three months has passed since the last newsletter, but, with the holiday season fast approaching, there is no denying the fact that winter is now upon us. It may not be as cold in the Lower Mainland of BC, where I reside, as many other parts of the country, but there is certainly a chill in the air that makes many of us, myself included, look forward to some time off at home over the holidays!

With December halfway over, we wanted to let all the readers know what CanPKU has been working on for the new year for event planning and advocacy work. Our annual provincial event days for 2014, organized for the benefit of everyone in the metabolic community, are now well into the planning stages. Details of scheduled days are below and we hope to see as many participants and attendees at these as possible - they are a wonderful way to meet people and find out all the latest and greatest goings on!

In addition to the year ahead, as always, we have updates on the many things that have happened over the last three months, which we feel to be newsworthy. Not only updates on successful advocacy work, but articles featuring our latest personal profile, recipes, studies and much more! Please enjoy the final issue of 2013 for the CanPKU and Allied Disorders Newsletter.

Wishing you a successful and happy 2014,

Ruth
Editor

Appanah

CanPKU and Allied Disorders Inc. Newsletter

Meet Our Sponsors

Click logos to view their websites!

B:OMARIN[®]

MeadJohnson[™]
Nutrition
NOURISHING THE BEST START IN LIFE

NUTRICIA
Metabolic Nutrition

PATIENT POWER[®]
because knowledge is the best medicine


Specialty Food
SHOP
1-800-737-7976



Innovation in Nutrition

Quebec PKU Event 2013

Quebec PKU Day was held in Sherbrooke, Que., on Sunday June 2nd, 2013. This coincided with the GARROD Symposium, which brought together many health professionals and scientists associated with centres for the management of hereditary metabolic diseases throughout Canada and the US.

Both events took place at the Faculty of Medicine and Health Sciences, Université de Sherbrooke, located in the CHUS Fleurimont Hospital. We are very grateful to Christiane Auray-Blais, head of organization for the Garrod Symposium, and her staff, for all the help we received to host our own event. Christiane even recruited two of her graduate students to man our daycare!

The first speaker was Dr. Desirée A. White, Ph.D., a Professor of Psychology at Washington University in St. Louis. In her Developmental Neuropsychology Research Laboratory she investigates executive abilities in children with disorders that disrupt brain function, with a focus on PKU. In her presentation entitled "PKU and the Brain" she presented - with a healthy dose of humour - an overview of findings from her laboratory related to executive abilities, brain function, and treatment with tetrahydrobiopterin in individuals with PKU.

Next Marie Lefrançois, Dt.P., an experienced nutritionist specializing in metabolic diseases at Montreal Children's Hospital (McGill University

Advocacy Updates and BC Treatment Coverage

I am so excited on so many levels to see BC Health Minister Terry Lake step up and implement funding for the specialty low protein foods that people with PKU and other IEM's (inborn errors of metabolism) require.

The BC Metabolic Nutrition Program will now provide up to \$3,000 per year to each patient as of January 1, 2014. Formulas will be covered as before and are not included in this dollar amount. All items must be ordered through the Special Products Distribution Centre at BC Children's Hospital and they ask you to limit your orders to \$250 monthly.

This is such a huge win: for CanPKU, PKU patients in BC, and myself personally. Back in the summer of 2009, when I wrote my first letter to my local MLA about this issue, I naively thought that just bringing this issue to their attention would solve the problem.

Why wouldn't they do what was right and what almost all other provinces were doing? However, I soon learned that I was facing an uphill battle.

A year later, another CanPKU volunteer, Amanda Cosburn, asked me to help her organize an event in Vancouver. Within a few months I was keen to work more closely with this excellent organization and excitedly took on the role of Vice President. As an added bonus, I now had the voice of CanPKU added to my own in advocating for change in BC.

Still, that didn't seem to be enough.

The government met with us, considered options and repeatedly told us no.

Finally, over the last year, things started to come together. My MLA Bill Bennett became an active supporter of our quest. We were working

Health Centre), spoke on the "Autonomy of the PKU Diet" and strategies that parents can employ to help ensure that teens manage their restricted diets effectively, thereby leading to an adulthood of good compliance.

After lunch, Dr. John Mitchell, MD, a Researcher in Endocrinology and Biomedical Genetics at McGill University Health Centre, and part of Metabolic Disorders Clinic team at the Montreal Children's Hospital, spoke about the recently published "Guidelines for Treating PKU". It is hoped this work will help reduce excessive variability in clinical practice and be an important reference guide for advocating for funding of necessary treatments.

The day ended with a cooking demo by Geneviève Lafrance, PDt, dietician/nutritionist in Medical Genetics at the CHUS in Sherbrooke. She teamed up with one of her patients, Maxime Olivier, to demonstrate how a low protein meal can be absolutely tasty and creative. They whipped up Grilled Eggplant and Portobello Mushrooms marinated with satay spices, Tian of eggplant, zucchini and tomatoes seasoned with herbes de Provence and Caramelized Pineapple with Vanilla.

As usual, time flew by and the moment had come to say thank you and goodbye to our speakers, our volunteers and all the participants.

Because Knowledge Leads to Better Health!

with a great team of consultants who helped us develop strategies and stay focused. And when MLA Terry Lake - who had already met with Brienna Young and Amanda Cosburn about PKU - was announced as the new Minister of Health last May, we felt a renewed optimism.

I can say unequivocally that the real difference maker was the involvement of other PKU patients and families. We grew to be a solid group of a dozen advocates in BC, all following the CanPKU lead. We met with our respective MLA's, and others such as MLA Steve Thompson also became very active in helping us. We asked our friends and family to write letters and had hundreds sent in. We contacted media, wrote our own letters and got petition signatures. Minister Lake accepted Amanda Cosburn's invitation to the Kamloops venue of our National Walkathon on June 1st and he spoke with a few different PKU families.

Together, our voice was louder. And we were heard!

Yes, there is still work to do in BC. The proposed coverage is not ideal, and we will be working with the BC Ministry of Health to expand the program to include a bigger variety of foods. Kuvan is also something that still needs to be publicly funded so that all PKU patients in BC can reach their best possible outcomes.

But more than ever, I am convinced that we can get the job done, by staying focused and working together. There is work like this to do in every province, so if you want to be a part of these great changes let us know!

Congratulations BC - we did it!!

**Nicole Pallone
VP**

MSUD Profile*

Grayson McGill



At just four days old, Grayson McGill had open-heart surgery to repair a rare heart defect. Four hours after coming out of the OR, his parents were pulled into a boardroom and told that unrelated, but in addition to the heart problem, Grayson also has Maple Syrup Urine Disease.

MSUD is similar to PKU, although the odds are 1 in 180,000. Unlike PKU, there is no 'going off diet', as elevated leucine levels causes brain swelling followed by a coma. Unmonitored MSUD is fatal.

Luckily for his two dads, Grayson has a great temperament, which makes the regular blood work and hospital visits pretty smooth. His restricted diet, although stressful, is very manageable and thankfully he has an agreeable personality.

Grayson is now three, and he attends pre-school three days a week. He loves learning and already knows his uppercase, lowercase and letter sounds.

As a family, they have also committed to giving back. Grayson was the champion child for BC Children's Festival of Trees and he was the cover kid for UBC's Lace Up for Kids fundraiser, which targets research for rare diseases. One of his dads is also a guest speaker at UBC's BioChem 303 class, educating the students on MSUD and what that means for their family life.

2014 Event Series Updates!

We are excited to share with you a few details regarding our event series for next year. Make sure you Save the Date for the event in your area!!

Saturday, April 5, 2014: Vancouver, BC
BC Children's Hospital with keynote speakers Dr. Jerry Vockley and Dr. Denise Ney

Saturday, May 3, 2014: Moncton, NB
Future Inns Hotel and Conference Centre with a presentation from Ryan Clarke from Advocacy Solutions.

Saturday, May 17, 2014: Red Deer, AB
Gaetz Memorial United Church with keynote speakers Dr. Barbara Burton and Dr. Kendra Bjoraker

Sunday, June 1, 2014: Ottawa, ON
Children's Hospital for Eastern Ontario with keynote speakers Dr. Jerry Vockley and Dr. Beth Potter

Saturday, October 18, 2014: Quebec City, QC
Four Points by Sheraton Québec Resort with keynote speaker Dr. Denise Ney

Start thinking now about hosting a Walkathon in your community on Saturday, May 31st to finish May as PKU Awareness Month! This is a simple way to build community and show your support for CanPKU that literally can be organized in just a few hours. Let us know if you are interested in hosting a walk and we will make it easy for you!

And finally, stay tuned for a special event happening this year only, in Summer 2014!

Because Knowledge Leads to Better Health!

Grayson's heart is now fixed and his MSUD management is ongoing. Each and every day, he does something new and amazing and he continues to be a hero to those that love him.

* Please note that we are always looking for children or adult profiles to highlight with each newsletter. If you would like to contribute your or your child's story please contact the editor!

Featured Recipe

Iced Sugar Cookies

These cookies are a staple in our household for everyone. They are a particularly festive treat when decorated with seasonal icing colors!



Cookies:

260 gm wheat starch

1 tsp xanthan gum

50 gm Jello instant pudding (vanilla)

1 tsp baking soda

1/4 tsp salt

1/2 tsp cinnamon

12 tbsp unsalted butter, soft

3/4 cup granulated sugar

1 tbsp brown sugar

25 gm mixed egg

1 1/2 tsp vanilla extract

Icing:

3 cups icing sugar

food coloring of choice

50 Years of Newborn Screening in Canada

2013 marks the 50th anniversary of universal newborn screening in Canada. Screening is a publicly funded health program designed to test infants for a number of conditions that are treatable, but not clinically evident, in the newborn period. In other words, the best mother or father, nurse or doctor do not have a chance to protect a newborn who is at risk of serious harm but is not showing any signs or symptoms. This simple procedure involves taking a blood sample from a newborn via heel prick shortly after birth, usually on the second day of life, using a lancet and blotting the blood collected onto specialized filter papers, which are then sent to designated labs and tested for a number of different conditions. There is also newborn screening for hearing loss and critical congenital heart defects using test devices in the newborn nursery.

Many of these conditions, including PKU, are not immediately apparent at birth but can have devastating effects if not identified and treatment initiated early. Of course most parents of/persons with or health care professionals in the field of metabolic disorders in general, are likely fully aware of the merits of newborn screening. That this process has improved the outcomes of countless babies is indisputable. In fact, newborn screening was named one of the ten greatest public health achievements in the US for the period 2000-2010.

Mass screening itself was first introduced by Prince Edward Island in 1963, and then quickly followed by BC and Quebec in 1964. Phenylketonuria (PKU) was the first test to be developed and was the reason to start newborn screening programs.

Back in 1963, screening was performed using the "Guthrie Test," developed by Dr. Robert Guthrie, whose niece had PKU but was late diagnosed. This was a bacterial inhibition assay that was able to detect the high levels of phenylalanine found in the blood of affected babies. This simple yet effective test was the

We use fruit juice for food coloring. 1 cup of pomegranate juice (12 mg Phe) boiled for approximately 10 minutes until syrupy will make enough pink for a full batch. Let the syrup cool and add it bit by bit to the icing sugar until you get the desired shade of pink/red you want!

Directions:

- 1) Heat oven to 375 C
- 2) Whisk the first four ingredients, set aside.
- 3) Beat the butter and sugars together, then add the egg and vanilla.
- 4) Gradually add the dry ingredients to the mix above until a dough forms.
- 5) Roll out the mix to about 1/2-1/4 inch, transfer to parchment paper and bake for 10-12 minutes.

Phe content: Total recipe = 252 (+ 12 for icing) = 264 mg Phe. Depending on cookie cutter used, divide by this amount. With current used above we made 24 cookies. $264/24$ cookies = 11 mg Phe per cookie.

*Cookie recipe from Cook For Love by Brenda Winiarski (www.cookforlove.org), icing made separately. Note sugar for rolling from the original recipe is omitted because cookies have been iced instead.

Hat's Off To:

The BC Ministry of Health and Minister Terry Lake!

On November 28th, 2013, the BC Ministry of Health announced that they had agreed to fund coverage of low protein medical food for those with a diagnosed and treated inborn error of metabolism disorder (see related article on right).

CanPKU believes there is more work to do in continuing to advocate on behalf of patients with metabolic disorders. The next step in treatment options is coverage for the drug Sapropterin (Kuvan). However, this is truly a great step forward for those patients in BC with an inborn error of metabolism.

Thanks to the Ministry of Health in BC for making this happen.

method that had been used to confirm a diagnosis of PKU for decades. It was not until the advent of tandem mass spectroscopy in the 1990's that this method of testing became obsolete. Dr. Guthrie was also primarily responsible for pioneering the collection of dried blood samples on filter paper as a method of transport for testing, which was critical to implementing screening on a nationwide scale. Newborn screening around the world is still done using similar filter paper.

Today, each province and territory in Canada has their own government-run newborn screening program, testing for between 11 and 38 conditions - all of them include Phenylketonuria (PKU).

For more information on newborn screening and source information please [click here](#).

Specialty Products Added Items (Ontario)

Did you know? The Specialty Food Shop located in Toronto at Sick Kids Hospital recently added over a dozen new covered formulas including ready to drink products.

For a link to the latest list of covered items please [click here!](#)

Cambrooke Foods is Now Cambrooke Therapeutics!

On December 4th, 2013, Cambrooke Foods officially announced that they have changed their name to Cambrooke Therapeutics as part of the expanding focus of the company on therapeutic nutritional products in the metabolic market.

Originally founded in 2000 by David and Lynn Paoella, parents of children with PKU, the

*For more details on the subsidy please [click here](#) and a link to Special Products at BC Childrens Hospital please [click here](#).

University Of Liverpool Study - Participants Sought

Researchers from the University of Liverpool are conducting a survey about knowledge, attitudes, and perceptions of individuals with a history of genetic diseases to personalized medicine. The results of the survey will contribute to a better understanding of the perceptions and attitudes that those with a family history of a genetic disorder may have towards personalized medicine and personal health in Canada. The study aims to identify the barriers faced by these same individuals which impedes their participation in programs and related research. Findings from the study may be useful in developing recommendations regarding adoption and implementation of Personalized Medicine in routine clinical practice in Canada.

Researchers are actively looking for participants who would only be required to complete a voluntary survey approximately 7 to 12 minutes long. The survey is completely anonymous and no information regarding participants' contact info is required.

Below is the link to the survey questionnaire which also includes a Participant Information Sheet as well as a Consent to Participate form. If you are interested, please visit the link to complete the survey questions.

<https://www.surveymonkey.com/s/QGMLYZT>

Note that this study has been approved by the International Online Research Ethics Committee at the University of Liverpool.

For any questions or concerns please contact Nahya Khaddoujawada via e-mail ([click here](#)) or phone: +(613)277824

Ideas or Suggestions for 2014?

Massachusetts company has grown to be a leader in the market.

With the name change, the company has also launched a new website to reflect this. [Click here](#) to visit.

For the press release on this topic [click here](#).

Become a Member!

New members will receive one free issue of National PKU News, courtesy of Virginia Schuett and all members now receive a 5% discount on all Country Sunrise products ordered from PKU Perspectives! Other benefits include discounts on CanPKU event registration fees, eligibility for CanPKU Travel Scholarships and voting rights at our Annual General Meeting (General Members only).

To become a member go to:

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

Please ensure that you choose the appropriate category when you next renew your membership!

- **General Member (GM):** any person diagnosed with PKU or other allied disorder; any person related to a person with PKU or other allied disorder and whose life is directly affected by the diagnosed disorder (such as a parent, grandparent, legal guardian, sibling, spouse or child). Only General Members 18 years of age and older having voting rights.
- **Supportive Member (SM):** any person who supports the vision and mission of CanPKU and is interested in advancing its objectives. Supportive Members have NO voting rights.
- **Professional Member (PM):** any professional (doctors, researchers, nurses, nutritionists, among others) related, interested and/or working on PKU and other genetic and/or

CanPKU always encourages feedback from our readers and members. We would like to hear from you if you have any suggestions or topics that you would like to hear about or see featured in any of the 2014 events that are currently being planned.

Please feel free to email [\(click here\)](#) and let us know!

Receiving This Newsletter Twice...?

Did you receive this newsletter twice? That means we have two email addresses for you!

Please email us at the address below and let us know which of your email addresses is the best one to use!

newseditor@canpku.org.

Quick Links...

[Our Website](#)

[Our Previous Newsletters](#)

[Our Facebook Page](#)

metabolic disorders. Professional Members have NO voting rights.

- **Corporate Member (CM):** any corporation interested in helping and supporting CanPKU. Corporate Members have NO voting rights.

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.

Join Our Mailing List!

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.

Sincerely,
Canadian PKU & Allied Disorders Inc.



Because Knowledge Leads to Better Health

www.canpku.org | info@canpku.org
Suite 180 - 260 Adelaide Street East, Toronto, ON M5A1N1
416-207-0064 or 1-877-226-7581 | fax 1-877-789-2462