Dear Readers,

On behalf of the CanPKU+ team, we wish you a summer filled with sunshine, laughter, and countless moments of joy!

Warmest regards,

Marjorie Guhl
CanPKU+ Newseditor

Inside this issue

CanPKU+ feels it is important to represent our community using REAL faces of those who are affected by each disorder, where we can. If you would like to be featured on our promotional or educational materials, flyers, website and more here is how!

1. Complete a RARE FACES photo release

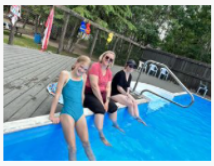
2. Grab a favorite photo of the person with Rare in your household

3. Email them both to info@canpku.org

Then keep your eye out and you may just see yourself featured online or in print on our materials!

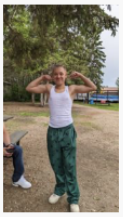










Check out our 2023 Camp MagniPHEque highlight page!

Thank you to all those who attended and to all the volunteers that made it happen!

I had the pleasure of attending the BC PKU day and it was so much fun! It was great to meet other families and learn from the experts!

Check out our events page for photos of our 2023 regional events!

We're on TikTok!

Click here to find us!

Recently received donations:

Stephen Schwindt

Click to donate

We have a new logo!

CanPKU+/CanPCU+ will be our acronym going forward in an effort to be more inclusive of the other Allied Disorders we represent and assist. The move to adding Denim (Jean/Gene) Blue was to better align ourselves with the traditional colours recognized in various other areas of the globe.

CANADIAN PKU AND ALLIED DISORDERS INC.
PCU ET MALADIES APPARENTÉES CANADA INC.

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Parce que la connaissance mène à une meilleure santé

www.CANPKU.org

Another DTC Success Story!

Thank you to Margaret for sharing her story!

Hi everyone!

My name is Margaret, I'm 27 years old and I'm living my best life in Truro, Nova Scotia. I was diagnosed with Classic PKU as a baby and have been a patient at the IWK health centre ever since. I also have 3 siblings all without PKU and have a tolerance of 9g of protein. I work in a nursing home as a caregiver, and love helping others! I was approved for the disability tax credit back in February and this is my story.

The disability tax credit didn't exist when I was first diagnosed with my PKU. I had followed the diet for life treatment plan for all of my childhood. Growing up in the 90's and early 2000's low protein options in grocery stores were almost nonexistent, and coming from a single parent household, specialty foods to order outside of my covered ones weren't always an option, but a luxury.

I had only begun to hear about the DTC in my late teens to early twenties. At that point the success rate was very hit or miss and I was unaware of how little of the process I actually had to do. It wasn't until I met a girl through work who has the disability tax credit for her type one diabetes that I began to hear about the process of applying.

I began to start looking into it, and I was genuinely baffled at how little I had to do. Aside from the one page of the application with my personal info and my spreadsheet, my genetics doctor (shoutout to Dr. Goobie!) handled the rest. I had several zoom meetings with Frances and we finalized my spreadsheet in June 2022 and sent it to my doctor to finish off. I sent it at a really busy time for my clinic, but they managed to finish my application by the end of 2022, and I was approved end of January, with my tax refund coming in February. At a DTC info session I attended virtually, John had told us that the disability tax code had been updated the year prior, and that very few cases with PKU were turned away now due to that amendment.

Being born in 1996, I was approved from the year 2000 until 2027, with my retro pay going back to 2012. I don't think I will ever forget the feeling of opening my bank account to check something and having a mini heart attack when I saw the balance! I will have to reapply for the 2028 tax year, but I saved all my things in a folder to make the process even easier! I took another piece of advice from the DTC info session and went straight to my bank and opened a disability savings plan, and reinvested some money into it, with help from the government it will continue to grow and supplement my retirement income in the future. It was really helpful this year at tax time and greatly changed my tax return.

For anyone who has been considering it, I highly recommend it! It's a little bit of work but the results are life changing! CanPKU was amazing and so supportive throughout the whole thing, so don't let fear of denial hold you back! If you're interested in following my journey, I documented my journey on my PKU Instagram

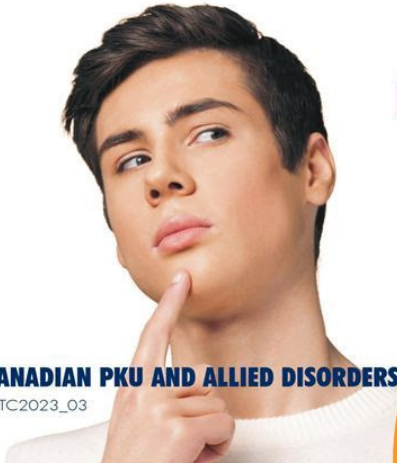
journey, I documented my journey on my PKU Instagram @thatpkugirl

Can’t wait to make some new friends!


TELEPHONE

DISABILITY TAX CREDIT (DTC)
Did you know?

- We have helped +150 adults and families with a child or children living with rare disorders to receive the Disability Tax Credit.
- ***No one who followed our advice has been refused!*** In the early period, there were some rejections - all successfully appealed.
- ***You don't have to be "disabled" to be eligible.*** Most approvals have been for people on a time-consuming therapy, the "medical diet", under clinic guidance, to prevent becoming significantly impaired, mentally or physically.
- In mid-2022, Parliament changed the DTC portion of the Income Tax Act to recognize that ***those on medical formulas and/or medical foods are eligible under "life sustaining therapy".***
- The DTC opens the door for several other benefits and credits, which vary for children or adults.
- The DTC can be made ***retroactive for up to ten years. You may get a bunch of tax rebates!***
- Clinics and other patient groups are asking for our help with DTC applications for other conditions... and we are helping them get approved!



Have you applied for the DTC?
What are you waiting for?
We can help!

L'autre côté français.

www.CanPKU.org

Because Knowledge Leads to Better Health

CANADIAN PKU AND ALLIED DISORDERS INC. PCU ET MALADIES APPARENTÉES CANADA INC.

DTC2023_03

CRÉDIT D'IMPÔT POUR PERSONNES HANDICAPÉES (CIPH)
Le saviez-vous ?

- Nous avons aidé plus de 150 adultes et familles avec un enfant ou des enfants vivant avec la PCU à demander et à recevoir le crédit d'impôt pour personnes handicapées.
- ***Aucune personne ayant suivi nos conseils n'a été refusée!*** La

UPCOMING EVENTS

Please note: If you are unable to register for an event that you believe you should have access to, please email to info@canpku.org and we will help you out!

All buttons are clickable to be taken to the registration page.

Zoom with us!

Watch for upcoming dates

Parents of those with PKU (All ages of PKU)

July 18: 8pm est.

Feature Story

In this issue's feature story we meet the Colter family. I had the privilege of meeting Melanie at the recent CanPKU+ regional event in Vancouver, BC where she shared Masen's Classical Homocystinuria (“HCU”) diagnosis story.

Masen was born on May 25, 2013, in Surrey, British Columbia, Canada, a city just outside of Vancouver, British Columbia. Masen received the standard newborn screening test, and everything came back perfectly. We went home with our happy, and we believed healthy, newborn baby boy.

Masen was the sweetest baby and brought us so much joy and happiness. He ate well, slept well and was such an easy going and happy little guy. We never had any serious worries about his health or well-being.

Just before kindergarten we took Masen for an eye exam and learned that Masen needed glasses. This wasn’t a big concern as it’s quite common and it also runs in our family. Masen rocked his new glasses, and somehow looked even more adorable than we already thought him to be!

Masen thrived in kindergarten, making new friends, and learning new things. However, within these first few years of school it became apparent that he was struggling with learning to read and write. It takes a little extra help and some hard work, but Masen always catches up to where he needs to be when he puts his mind to it. He is a very hardworking kid and does well in school. This didn’t cause any huge concerns or red flags for us.

In February of 2021, when Masen was 7 years old, we had a routine eye doctor’s appointment. We were very lucky to see this doctor on this day because he saw something that triggered something he learned in medical school, something that many others likely would not have thought about or recognized due to how rare it is. We got the ball rolling on what would be months of living in fear with many unknowns, and lots of medical tests. This is where our Classical Homocystinuria (“HCU”) journey began.

Masen was officially diagnosed with HCU on June 25, 2021, at just over 8 years old. Our world was turned upside down as we learned of the health risks and of the tools in which to manage HCU. We continue to work hard daily to overcome the challenges that this diagnosis has brought us.

We learned that even though Masen was tested at birth, HCU has a very high rate of false negatives at birth. This is terrifying and life threatening to babies. It’s a wide spectrum of possible outcomes for HCU when it goes untreated. There are many conditions and complications that can arise having to do with the brain, eyes, blood vessels and bones.

Masen had always been a fantastic eater, and essentially overnight all his favorite foods became things he would likely never eat again. While it was extremely difficult to take away his favorite foods, Masen willingly tried the new low protein foods that we were introducing to him, and we celebrated together when he enjoyed something new. He must endure monthly blood draws for us to help keep him healthy. He drinks his formula daily without complaint and has memorized what supplements he must take throughout the day and when. In Spring of 2021, Masen underwent two separate eye surgeries, one on each eye, to repair the damage that had been done from going so many years undiagnosed. As his parents, we could not possibly be prouder of Masen and all that he has overcome.

Today, Masen is an active, energetic, friendly, and resilient little boy, who loves to play sports, watch Pokémon and make everyone laugh. His quick wit and sarcasm are brilliant. He is extraordinary in every way and braver than anyone I know.

While I said that Masen is resilient, we hate that he must be and we long for a day where his daily life will not have to be managed with such strict diet and supplementation to stay healthy. Until then we will continue to celebrate our victories and take this path that we are on one day at a time.

My son and I made these and they were delicious!

A fun summer treat.

Marjorie

-Marjorie
CanPKU+ Newseditor

Recipe from **myfussyeater.com**

Summer Fruit Spring Rolls - My Fussy Eater | Easy Family Recipes

My Fussy Eater
131k followers

Ingredients

Produce

- 1 Apple, medium
- 1 Kiwi fruit
- 1/2 Mango
- 5 Mint, leaves fresh
- 1 Peach, medium
- 1/8 cup Pomegranate arils
- 10 Raspberries
- 4 Strawberries, medium

Condiments

- 2 tbsp Honey
- 1/2 Lime (juice of)

Pasta & Grains

- 10 Paper roll - rice paper

Do you have a favorite recipe?
Send them to us by filling out the form below.
This can be found here on our website.

Recipe name: _____
Serves: _____
Preperation time:

Preparation time: _____

Cooking time: _____

Ingredients

Preparation

Notes

so sweet to bee

ONE!

We want to celebrate with you!

Eat Well, Live Well.
AJINOMOTO

CAMBROOKE

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To receive a first years birthday gift for your little one with Rare
Email to info@canpku.org

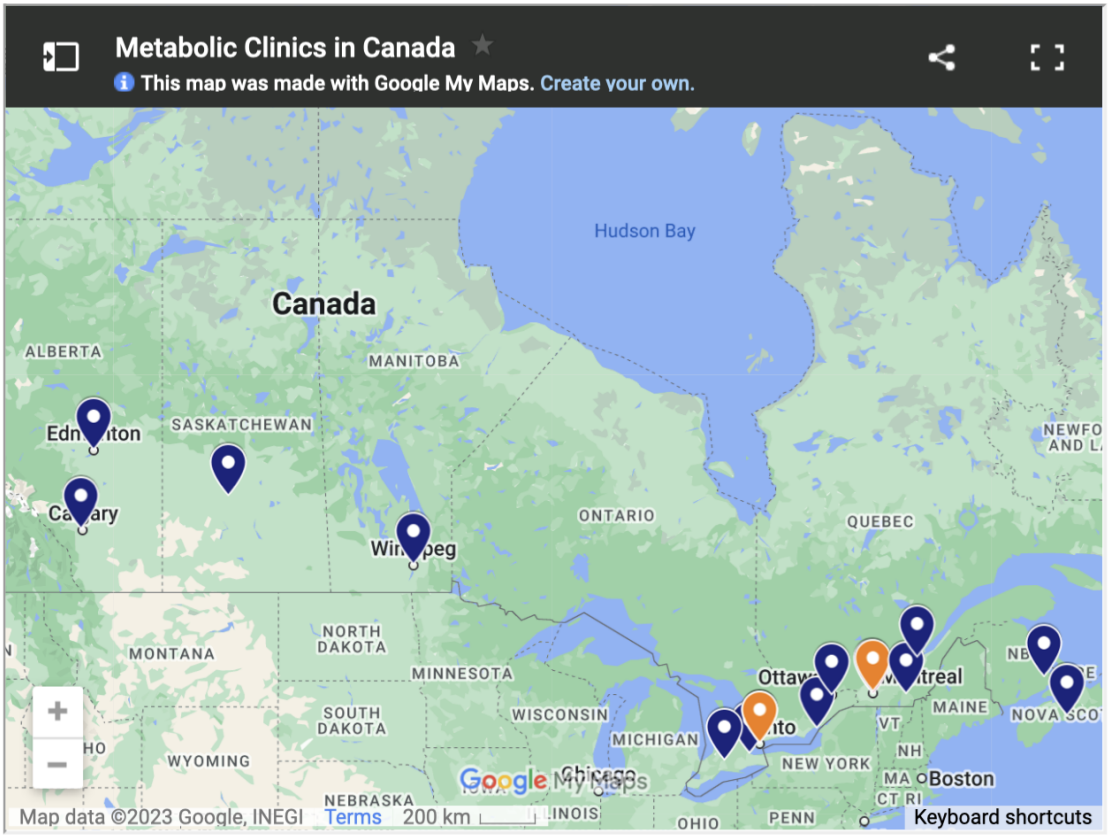
Please be sure to include parents name and email as well as the birthday child's name and birthday.

*Did we miss their birthday? Let us know. All first birthdays celebrated this year will qualify.

Did you know that we now have an interactive map showing all our metabolic clinics as well as patient locations by postal code?

Adults and Children
Adults Only
Children Only

METABOLIC CLINICS IN CANADA:



METABOLIC CLINICS IN CANADA:

encouragement for your fellow PKU ers?
Whatever it may be, if you'd like to share,
please submit to newseditor@canpku.org.

Are you interested in
hosting a FUN-draiser
to support CanPKU?

SPARE FOR RARE

is a fun and exciting bowling fundraiser
(formerly known as PINS 4 PKU)

Contact our Fundraising Chair,
Melissa, at fundraising@canpku.org
for more information

HOW DO I HOST A "SPARE FOR RARE" EVENT?

CONTACT CANPKU

DEVELOP THE CONCEPT

- REACH OUT VIA EMAIL TO OUR FUNDRIASING CHAIR, MELISSA AT FUNDRAISING@CANPKU.ORG FOR MORE INFORMATION AND ASSISTANCE WITH PLANNING THE EVENT

- WILL YOU HAVE A THEME
- WILL THERE BE PRIZES?
- WILL YOU GET THE BOWLING LANES SPONSORED?
- HOW MANY TEAMS/PEOPLE WILL/CAN ATTEND?
- WE CAN HELP!

FIND A VENUE

- FIND A BOWLING CENTER TO HOST YOUR EVENT
- CHOOSE A DATE THAT WORKS FOR YOU AND THE FACILITY
- CHOOSE A TIME FRAME THAT WORKS ON THE CHOSEN DAY

SPREAD THE WORD

- POST ON SOCIAL MEDIA
- HAVE FRIENDS AND FAMILY POST ON SOCIAL MEDIA
- PUT FLYERS UP IN LOCAL BUSINESSES
- POST IN LOCAL SOCIAL MEDIA GROUPS
- WE CAN HELP!

FUNDRAISE

- ASK LOCAL BUSINESSES TO SPONSOR LANES
- HAVE EVENT PARTICIPANTS COLLECT DONATIONS
- YOU COULD CONSIDER HAVING A SILENT AUCTION WITH DONATED PRIZES TO RAISE MONEY AT THE EVENT

THE EVENT

- THE DAY OF THE EVENT IS A TIME FOR FUN!
- REMEMBER TO ENJOY YOURSELF, YOU'VE WORKED HARD ON THIS!
- TAKE PHOTOS AND VIDEOS TO SHARE WITH CANPKU FOR OUR SOCIAL MEDIA PAGES

COMMENT HÉBERGER UN EVÈNEMENT "REPARE POUR RARE"?

CONTACTER CANPKU

- CONTACTER PAR E-MAIL NOTRE PRÉSIDENTE DE LA COLLECTE DE FONDS, MELISSA À FUNDRAISING@CANPKU.ORG POUR PLUS D'INFORMATIONS ET DE L'AIDE POUR PLANIFIER L'ÉVÉNEMENT

DÉVELOPPER LE CONCEPT

- AUREZ-VOUS UN THÈME
- Y AURA-T-IL DES PRIX?
- FEREZ-VOUS SPONSORISER LES PISTES DE BOWLING ?
- COMBIEN D'ÉQUIPES/DE PERSONNES PARTICIPERONT/POURRONT PARTICIPER ?
- NOUS POUVONS AIDER!

TROUVER UN LIEU

FAIRE CONNAITRE

- TROUVER UN CENTRE DE BOWLING POUR ACCUEILLIR VOTRE ÉVÉNEMENT
- CHOISISSEZ UNE DATE QUI CONVIENT À VOUS ET À L'ÉTABLISSEMENT
- CHOISISSEZ UNE PLAGE HORAIRE QUI FONCTIONNE LE JOUR CHOISI

- PUBLIER SUR LES RÉSEAUX SOCIAUX
- DEMANDEZ À VOS AMIS ET À VOTRE FAMILLE DE PUBLIER SUR LES RÉSEAUX SOCIAUX
- METTRE DES FLYERS DANS LES COMMERCES LOCAUX
- PUBLIER DANS DES GROUPES DE MÉDIAS SOCIAUX LOCAUX
- NOUS POUVONS AIDER!

LEVER DES FONDS

- DEMANDER AUX ENTREPRISES LOCALES DE PARRAINER DES VOIES
- DEMANDER AUX PARTICIPANTS À L'ÉVÉNEMENT DE RECUEILLIR DES DONS
- VOUS POURRIEZ ENVISAGER D'ORGANISER UN ENCAN SILENCIEUX AVEC DES PRIX DONNÉS POUR AMASSER DES FONDS LORS DE L'ÉVÉNEMENT

L'ÉVÉNEMENT

- LE JOUR DE L'ÉVÉNEMENT EST UN MOMENT DE PLAISIR !
- N'OUBLIEZ PAS DE VOUS AMUSER, VOUS AVEZ TRAVAILLÉ DUR LÀ-DESSUS !
- PRENDRE DES PHOTOS ET DES VIDÉOS À PARTAGER AVEC CANPKU POUR NOS PAGES DE MÉDIAS SOCIAUX

Updates and Articles of Interest

PTC Therapeutics, Inc.
100 Corporate Court
South Plainfield, NJ 07080
www.ptcbio.com

Dear PKU Community,

We are pleased to share the topline results of the global APHENITY clinical trial that evaluated the effects of sepiapterin (an investigational new drug formerly known as PTC923) on the blood phenylalanine levels in children and adults with PKU. We are happy to announce that the trial met the primary endpoint of blood Phe reduction with highly statistically significant and clinically meaningful results.

Sepiapterin demonstrated Phe reduction of 63% in the overall primary analysis population and 69% in the subset of classical PKU patients. In addition, the majority of patients were able to reach target Phe levels in line with U.S. guidelines of <360mmol/L. Sepiapterin was also well-tolerated with no serious adverse events.

APHENITY was a global double-blind, placebo-controlled, registration-directed study which enrolled children and adults with PKU. Participants were randomized to receive sepiapterin or placebo for six weeks with the primary endpoint being reduction in blood phenylalanine levels.

This trial would not have been possible without the support of the entire PKU community, including clinicians, researchers, and most importantly patients and their families. Your support and participation in natural history and clinical trials throughout the years has been, and continues to be, incredibly important in the advancement of treatments for PKU. We thank you for your commitment and sacrifices in making APHENITY a success.

We know families need safe and effective treatments that have the potential to change the course of this rare disease and allow for better quality of life. We believe the APHENITY results position sepiapterin to potentially address the persistent large unmet need for PKU patients. We look forward to meeting with global regulatory authorities to discuss the path to sepiapterin approval and will continue to provide updates on our progress.

Our patient engagement team is available to speak with you and answer questions. They can be reached at 1-833-PTC-HOPE (+1-833-782-4673), or patientengagement@ptcbio.com.

Sincerely,

Matthew B. Klein, M.D., M.S., FACS
Chief Executive Officer
PTC Therapeutics

PTC Therapeutics Announces Virtual Presentation on Phenylketonuria (PKU) and Sepiapterin

Wednesday, July 19th
12 to 1:30 p.m. ET.

Article Link

News Release

Synlogic Granted Fast Track Designation from FDA for labafenogene marselecobac (SYNB1934) for Treatment of Phenylketonuria

Article Link

Canadian News

English Article: https://theconversation.com/canadian-science-pioneers-role-in-the-human-genome-project-shows-why-its-crucial-to-fund-research-204340

French Article: https://theconversation.com/le-role-des-scientifiques-canadiens-dans-le-projet-du-genome-humain-montre-pourquoi-il-est-crucial-de-financer-la-recherche-204503

Waiting for new drugs for rare disorders in Canada – part 1, regulatory approval: Nigel Rawson and John Adams for Inside Policy

Article Link

CanPKU's Drive to Thrive Committee

As mentioned earlier this year, CanPKU is committed to helping those who were previously misguided (with information that at the time was believed to be correct) that it would be ok to come off their medical diet. This may have been as the medical community believed that the brain had developed enough and would not incur further brain damage by lifting the protein restriction. It may be due to the fact some provinces restricted coverage of therapeutic products (and some still do) so it was impossible to maintain the previous "diet" therapy. Lastly, it may be due to the fact the adult opted for their own reasons to come off "diet" and stop therapy.

We now know that this is not the best course of action. We owe it to those who were previously diagnosed, but are now lost to treatment to help them return to clinic and resume treatments (which have dramatically improved since their time on therapy).

To do this, we need your help. We are looking to form a committee that will help locate and support these individuals. You do not have to be an adult with PKU, just a passion to help. We hope to have representatives from many provinces.

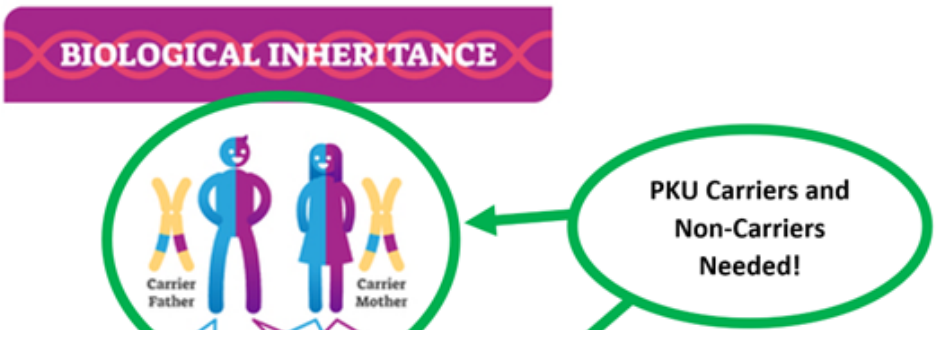
Are you up for it? (Can you join both committees mentioned in this newsletter - Absolutely!) Email to Tanya.Chute@canpku.org to sign up!

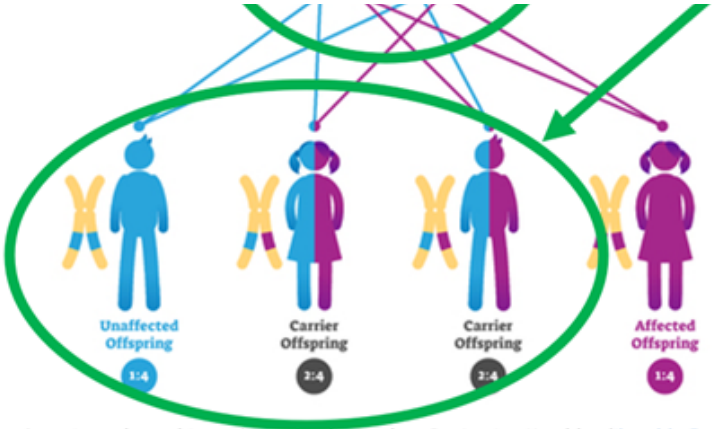
Email Tanya

Eat Well, Live Well.

AJINOMOTO

CAMBROOKE™





Questions about this study can be directed to: Dr. Justine Keathley (jkeathle@uoguelph.ca). This project has been reviewed by the Research Ethics Board for compliance with federal guidelines for research involving human participants (REB#23-02-009).

Image credit: https://www.123rf.com/clipart-vector/genetic_mutation.html

Let's hear from you!

with a genetic condition

For more information: tasha.wainstein@bcchr.ca

We would like to invite your child (10-19 years old) to participate in a study that aims to explore the experiences of adolescents with genetic conditions. Even if genetic testing did not identify a gene change which explains their medical problems, we are still interested in hearing about their experiences. Participation in this study is voluntary and would involve a ~1 hour virtual interview. During the interview, we will ask some questions about their experiences of living with a genetic condition. They will also be asked to complete three short questionnaires online. We hope this study will help us to design the best possible clinical services for adolescents who have genetic conditions. Your child will receive $20 for sharing their time and expertise on this important study. For additional information about the study, please contact Tasha Wainstein who will be happy to answer any questions you or your child may have (tasha.wainstein@bcchr.ca or 604-875-2000 (ext 6333)).

We believe that the insights of adolescents from your organization will be extremely valuable. If you have any questions about the study, please feel free to contact me (Carly.pistawka1@bcchr.ca) or Tasha (tasha.wainstein@bcchr.ca).

Thank you for your consideration

If you or your caregivers would like more info:
https://tinyurl.com/363t23pu
or contact: tasha.wainstein@bcchr.ca

Will You Share Your Story?

We are looking for **people living with PKU** to share their story. How does PKU affect your life and what does it mean if potential new treatment options become available? We'd like to hear from everyone, but especially from **adults who are struggling to meet treatment targets (Phe > 600 umol/L or 10 mg/dL).**

If you are willing to **share your story**, please email Lori Gorski ***lori.mckenna.gorski@synlogictx.com*** to set up a time to speak at your convenience.

Your story may be shared outside of Synlogic – but only with your approval.

We are so grateful to you, and to the devoted leaders of CanPKU, for their continued collaboration and guidance.

synlogic

RESOURCES

CanPKU has a number of resources which can be downloaded or printed at

www.canpku.org/Downloadable-educational-resources

Getting the most from the CanPKU website and emails!

Did you know you can get tailored emails and event invitations (like a cafe chat between folks like you! That could be a parent of a PKUer, Child, Teen or Adult with PKU!)

To do this we need some help with updating your profile.

Find out how to do this with information found at

www.canpku.org/Update-your-Profile and www.canpku.org/how-to-use-the-website

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Our sponsors are listed in the order that they began to support CanPKU's efforts. This order does not represent the dollars they may contribute.

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Eat Well, Live Well. Aji AJINOMOTO

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RESEARCH • PROGRESS • HOPE

Sponsorship in Kind. These companies provide assistance other than financial.

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deliciously dairy free™

CanPKU News

Canadian PKU and Allied Disorders Inc. is a non-profit association of volunteers, first organized in the Spring of 2008 based in Toronto, Ontario. We are dedicated to providing accurate news, information and support to families and professionals dealing with phenylketonuria and similar, rare, inherited metabolic disorders.

Our mission is to improve the lives of people with PKU and allied disorders and the lives of their families. By allied disorders, we mean other rare, inherited metabolic disorders also detected by the modern miracle of newborn screening.

La « Canadian PKU and Allied Disorders Inc. » est une association à but non lucratif de bénévoles créée au printemps 2008 à Toronto, en Ontario. Nous nous engageons à fournir des nouvelles, des informations et un soutien précis aux familles et aux professionnels travaillant avec la phénylcétonurie et des troubles métaboliques héréditaires rares similaires.

Notre mission est d'améliorer la vie des personnes atteintes de PCU et de troubles apparentés ainsi que la vie de leurs familles. Par troubles apparentés, nous entendons d'autres troubles métaboliques héréditaires rares, également détectés par le miracle moderne du dépistage néonatal.

"Education is the passport to the future, for tomorrow belongs to those who prepare for it today."

—Malcolm X

Privacy Statement:

Please note that CanPKU maintains member/participant/registration information for its own use and does not rent, sell or otherwise provide any identifying information to outsiders.

www.canpku.org

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