The Inherited Metabolic Disorders News

To receive the next issue of the newsletter by email, contact maria.bata@lhsc.on.ca

Children's Hospital
London Health Sciences Centre

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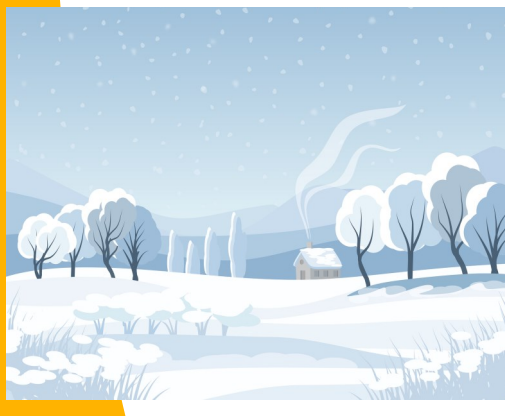
In this Issue



From the Editor

Hello everyone! I am happy to introduce myself as the new editor of the newsletter! It has truly been such a rewarding project to be a part of. I've felt so inspired while compiling the stories, photos, and resources for this season's newsletter, and I hope that you all feel the same sentiment as you read through. The theme for this edition is *mindfulness,* and I hope that we can all carry this theme throughout our winter season. All the best!

Maria Bata

From Dr Chitra Prasad

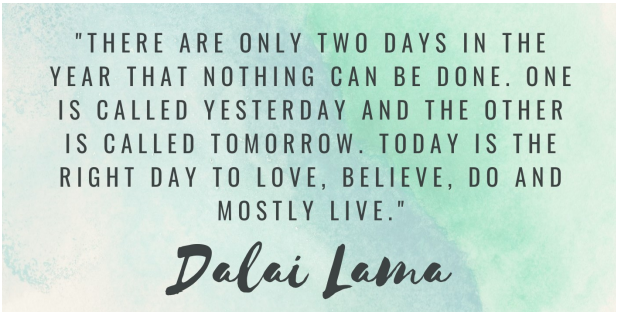
Dear Friends,

Greetings for the new year. I know the pandemic has affected each one of us in many different ways. It has not been easy for the whole world dealing with the losses, health burden, economic issues along with social isolation and rising mental health problems. In spite of all the challenges, we as humans continue to adapt to the newer realities. The human spirit remains resilient. I am grateful that I was able to go to India for a couple of weeks.

In this newsletter you will find an incredible story of Jeannette and her journey with phenylketonuria (PKU). She has made remarkable progress after going back on the PKU diet. Our metabolic team wants to wish Sue McLean (our social worker) the very best for her retirement. Sue helped many of our metabolic patients. We have new members in our metabolic team Carrie Charters (new metabolic social worker), Dr. Mia Sethna (pediatrician) who is helping us with some of our metabolic patients and Maria Bata (our new resource associate and also the editor for our newsletter). I welcome all of them to our metabolic team. With the pandemic, we have not been able to have our regular metabolic family workshops and I know we all miss getting together and learning from one another. Let us hope we can have one next year. I'm happy that we are able to communicate through the metabolic newsletters. Please continue to share your stories, achievements and photos. As I reflect on how best we can all go through these challenging times, I find that being "mindful" certainly brings peace even in the face of most uncertainties. Please find some resources for mindfulness in this newsletter which may be helpful.

My very best wishes for you and your families.

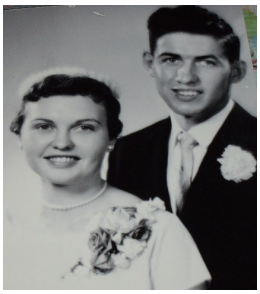
Your friend,
Chitra Prasad



Personal Story: Jeanette's PKU Journey

My name is Jeanette and I was born on May 12, 1967. I am 54 years old, and I was diagnosed with PKU at birth. I am the youngest of five children. In October of 1966, it came to pass that all babies born were to have a screening to see if they had PKU, and my screening was positive. My family had never heard of this before, and the doctors had to keep me in the hospital to get my diet and my phenylalanine levels in the normal range. I was in the hospital for about 3 weeks.

The doctors needed to test the rest of my family to see if my siblings had PKU. They found out my older sister, Margaret, also had PKU. They were concerned about Margaret's progress due the fact she was almost 3 and had been eating regular protein foods. They immediately put her on the PKU diet.



My mom and dad were overwhelmed and scared at the same time. As I was in the hospital with the doctors, my mom was working on my sister's diet. Mom said it was exceedingly difficult because there was very little information when we were growing up. My mom had to measure each one of our foods. Both my sister and I had different levels of phenylamine that we could handle. I was considered a classic PKU, and my sister had a milder case.

The formula was extremely difficult to drink. It tasted terrible. There was no flavored formula, no pre-made drink boxes, no pouches pre-measured. It came in a can that you had to measure each time. My siblings and I grew up in the 70s as a child. We grew up in the country and on a farm, which made it much easier to get fresh vegetables because we had our own garden. My mom had to measure absolutely everything that we ate. As for my grades in school, they were average, and I never failed a grade. My sister was about 6 months behind her twin brother before she was on the diet. After she was on the PKU diet, she quickly caught up to her twin brother and the rest of the class. My mom was and is an amazing mother. She had to be strong and would educate my family members on how we could not have protein.

My parents would take my sister and I to London Children's Hospital several times to get our blood tested and to see the doctor. The doctors would do several types of tests to see how we were developing mentally and physically. Finally, the doctors developed a blood test where my mom would pick our fingers and use the dot cards so we could mail them to the London Hospital instead of making several trips. I remember getting the finger picks and I would run around the furniture trying to get away from my mom. The pick needles that they would use was a little steel pick with a very sharp triangle edge. They hurt very much. I was going to be glad when I could stop doing them.

The doctors would continually do studies on PKU children. My sister and I were the 26th and 27th children in Ontario to be diagnosed with PKU. The doctors said once we turned 5 years old we could go off the diet. Once we were ready to have children of our own, that would have to go back on the diet in order to get our phenylalanine levels to be safe to have a healthy baby.

I was married in 1990 and we planned to have children so I went back on the PKU diet in January 1993. It only took me four months to get my levels to a safe level to conceive a baby. I got pregnant almost right away. I had my healthy baby boy on January 6, 1994. The diet had changed so much more from the time that we were children to the time we were having our babies. The formula was a bit better. It still tasted terrible and smelled bad. The dietitian I worked with gave me some tips on how to make it easier to drink. I also learned some tips from my older sister when she had her babies. One thing that was a great improvement while having my baby, was that they had some specialty foods available. I was introduced to some low pro pasta and low pro bread. They also had things like pizza and baking mixes to make pizza dough and cupcakes. I would make low protein cookies and pasta salads. It was much more enjoyable to have a variety of food to choose from.

Personal Story - continued

My sister and I had been off the diet for several years. We were done having children and we didn't feel the need to be on the PKU diet. My sister Margaret is 3 years older than myself. When Margaret started going through menopause, my mother and I would notice she was getting a little more forgetful. We figured it was the menopause effects. As a few years went by, her husband and close family members would notice she was getting a little more forgetful than usual. Our family was getting worried she was starting symptoms of dementia. Margaret would say she forgets sometimes but not a lot. She didn't think it was anything to worry about.

Margaret would start to notice a bit of brain fog. She thought it was maybe lack of sleep. We also noticed she was more anxious and more confrontational with me. Over the last couple of years this would get more noticeable. I did some research on the Canpku.org website. I was interested to see if maybe this had anything to do with our PKU. I was noticing some remarkably interesting similarities in adults with PKU and not being on the diet. It explained how over time PKU adults can develop symptoms like brain fog and forgetfulness when not on the diet.

I encouraged my sister to reach out to London Health Sciences to see if they would talk to my sister and see if there is any connection to this brain fog and forgetfulness she is having. Our family was getting concerned she was maybe getting dementia. My sister reached out to the London Metabolic clinic and got an appointment. They told her it was a definite connection with not being on the diet. My sister decided to go back on the PKU diet in September 2019.

Within a few months after going back on the diet, she was a much happier person. She was much more friendly and not as anxious. Her brain fog started to clear up and her forgetfulness was much better. I had my sister back. The sister I knew from our younger years. It was amazing. My mom noticed she would be singing while cleaning house. Margaret would more energy and didn't feel so worn out. Margaret was doing so well on the diet. She was getting her phenylalanine levels on track and she was getting close to 16 grams of protein food per day.

The diet was much improved from even the time we had our babies in the late 80s and early 90s. The formula was so much better now - there were actual flavors, and it was pre-measured. There was so much more low protein food to choose from like premade Cam Burgers, baking mixes that can make chicken patties, hotdog mixes, low protein rice and pasta of course. It was utterly amazing. My sister wanted me to go back on the diet with her. I told her no, she could try it first and see how it goes. I was not wanting to give up my freedom on not having to count my food daily. I was not wanting to give up being able to go to restaurants and not being able to order anything that I wanted. I didn't want to give up my burgers. I didn't want to start the PKU diet again and not be able to stick to it. I did notice great changes in my sister and I was incredibly pleased to get her back on track. She was much healthier for it.

Just as things were going great, things took a turn for the worst. My sister suddenly passed away in May of 2020. She had a brain aneurysm and was rushed to the hospital. The doctors tried to stop the bleeding but to no avail. We were all in shock. She was doing great, and all was well. Due to the history of our family with brain aneurysms, the rest of my siblings and I got MRIs to see if there was anyone else that had any signs of them.

Personal Story - continued

When it came my turn for my MRI, I didn't have any signs of aneurysms. The doctor told me I did have some white matter spots in my brain and that it could be the start of MS (Multiple Sclerosis). I thought, "Oh no" - I didn't want to believe it. I also suffer from fibromyalgia. Some days it would be very difficult to walk and the pain in my legs can be difficult. I figured my tiredness is from my fibromyalgia. My mom encouraged me to talk to the metabolic clinic in London. When I talked to the doctor and dietitian in London Health Sciences Centre, they told me the white matter is the effects of not being on the diet. I was hesitant to go back on the diet but I decided to give it a try. To my surprise, it was much better than I thought. My levels before the diet were 2057 Phe. Just by cutting back on protein and regular breads and pasta, my levels came down to 1332 Phe. When I started the diet on December 10, 2021. My Phenylalanine count at starting the PKU diet was at 900 Phe. I started off with 8 grams of protein food and 80 grams of formula. Now, as of May 5, 2021 my Phe count is 256 and I am up to 23 grams of protein food. WOW, Amazing!!!

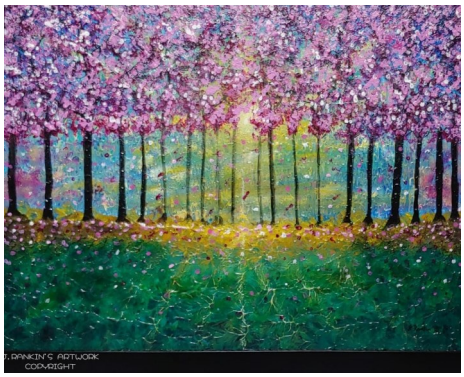
It was difficult the first couple of weeks and was worried I was going to not stick with it. To my amazement, it was much better than I thought because of the many options to choose from and it being so much easier to count my protein food counts per day.

I have noticed, along with my mom and son, that I have more energy and I am much happier. I was anxious before the diet and didn't realize it until lately. To my astonishment, my fibromyalgia is getting slowly better as well. I felt the best when I was on the diet while being pregnant and I am feeling great and healthy once again. I have been working full time for many years, and I am completing an online basic psychology course through my local college. I had also completed a PSW course through Fanshawe College in the early 2000s. I worked in that field for 10 years, but due to a lower back injury, I went into a different direction. I now have been working in the sales field for a major hotel company for the last 9 years and I am considered one of the top sales agents in my field.

I am so glad to hear that the doctors are keeping the PKU patients on the diet for life. Some days can and will be difficult. If we knew all the things about PKU back when I was growing up, I would have been happy to stay on the diet. I never knew being off the diet for a long time and over the years and as you grow older, that you can get the brain fog or forgetfulness, or physical difficulties in walking or other physical difficulties.

For the young ladies that have PKU and are thinking of having a baby, please work with your PKU doctor so you can have a happy healthy baby. I want to take a minute to thank all the staff, doctors and dietitians etc. in the Metabolic Clinic at London Health Sciences in London, ON Canada. They have done a tremendous job in providing the best knowledge and care for my sister Margaret McKellar and myself over the years of our journey. We truly thank you and appreciate everything you do for all your patients.

Thank You
Jeanette Rankin



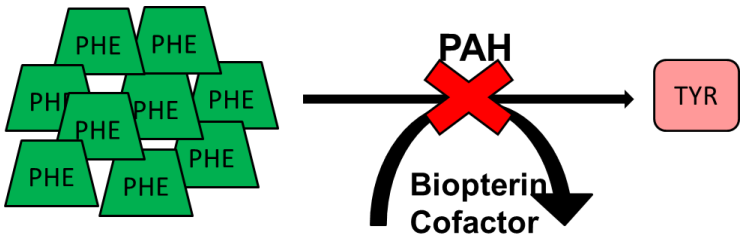
Artwork by Jeanette Rankin

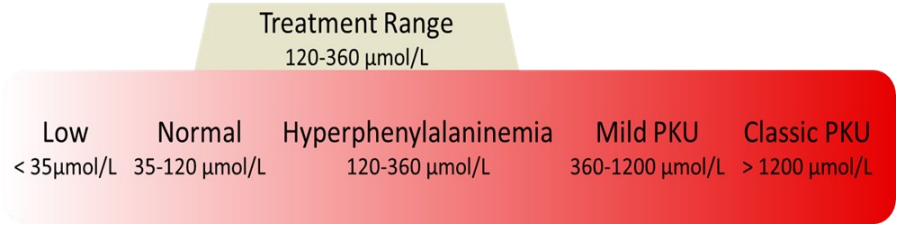
Featured This Issue

Phenylketonuria (PKU): Lessons Learned over the Last 88 Years

Compiled by Dr. Chitra Prasad

Phenylketonuria is a rare genetic disorder with an incidence of approximately 1 in 10,000. Due to a deficiency in the enzyme phenylalanine hydroxylase (PAH), the amino acid phenylalanine (phe) cannot be broken down properly. It was first described by Asbjörn Fölling in 1934 in Norway. PKU is famous in part because it is widely seen as a victory for scientific medicine. If the condition is detected in the newborn period and a specialized diet is started, the severe mental impairment usually caused by PKU is avoided. PKU will always be remembered as the first condition for which newborns were screened. This is thanks to Dr. Bob Guthrie, who nearly 60 years ago made a scientific breakthrough with his inexpensive and accessible screening test.





PKU is a genetic disorder which is inherited in an autosomal recessive manner like many of the other metabolic conditions. There is a 1 in 4 risk of having another affected child when both parents are carriers. Genetic counselling is also an important aspect of management.

Dietary therapy is extremely important for the management of PKU. However, the diet is challenging as many of our patients and families are well aware. Being “on diet” with proper control of phenylalanine level is recommended for life. When Phe isn’t broken down properly, changes in brain chemistry may occur leading to learning and behavior issues, depression, agoraphobia (fear of going outside), panic disorder and multiple other mental concerns. Pregnant women with PKU need to be very careful as the elevated phenylalanine levels can cross across the placenta and affect the baby’s brain chemistry, which can cause mental retardation and heart problems.

Staying on the diet requires motivation and support from the entire family as well as the dietitian and the metabolic clinic. In our clinic we encourage children to start taking part in their own PKU management as early as 3 years old. This allows them independence and also responsibility of managing their PKU. Low protein food workshops, metabolic workshops, sharing of recipes through our newsletters and forming a community definitely makes a difference to the metabolic control. Initially the low-protein diet was only covered for children up to 18 years, however with the patient advocacy now the low-protein diet is covered in Ontario for adults as well. As the diet is changed, there can be deficiencies of trace elements and minerals and that’s why we order specific bloodwork, which monitors these periodically. Neuropsychiatric assessments are performed during school years and bone health is also monitored.

Featured This Issue - continued

Phenylketonuria (PKU): Lessons Learned over the Last 88 Years

Compiled by Dr. Chitra Prasad

Other therapies for PKU are now becoming available. Kuvan (biopterin cofactor) as an oral medication has allowed many patients to introduce/include more foods to their diet. However not all patients are responsive to Kuvan. Large neutral amino acids [LNAAs] are being used in some of the metabolic clinics. Other newer therapies include Palynziq (pegvaliase) (phenylalanine ammonia lyase (PAL), Hepatocyte transplantation, and gene therapy. I'm hopeful that newer therapies will make a positive impact on the patients and their families and make the management of this metabolic disorder easier.

Many of our patients with PKU are leading productive and healthy lives which is certainly a great achievement of modern medicine, successful collaborations of metabolic clinic (particularly dietitian), and most important the patients and their families.

Metabolic Family Workshop Update

Art by Evelyn, hypophosphatasia

Unfortunately as the pandemic continues to persist, we will not be hosting the Metabolic Family Workshop this year. Please stay tuned to future issues of our newsletter to stay in the loop. We can't wait to once again be able to run this event and see everyone once again! Until then, take care!

Art by Noah, PKU

Art by Noah, PKU

Suzanne's Corner

Emmitt's Low Protein Bowl of Yumminess

This recipe is versatile and may be prepared in different ways depending on how much phenylalanine, leucine or protein that you want to use

BE CREATIVE!

Base
Aproten Riso Rice
Country Sunrise Chicken Patty or Mushroom Burger Mix

Toppings
Here are some ideas
Canned corn
Broccoli or cauliflower (steamed)
Shredded low protein cheese
Sweet potatoes (cubed and pan fried)
Peppers
Tomatoes
Carrots (shredded or steamed)
Mushrooms
Avocado

Seasoning/sauce
Another way that you may customize
Chicken consommé seasoning from Cambrooke (mix into rice before other ingredients are added)
Store bought vegetable broth or George Washington seasoning broth (cook the rice in the broth for flavour)
Ketchup/BBQ sauce
Garlic mayonnaise
Salad dressing

Preparation

1. Start with the rice. Emmitt uses 1/3-1/2 cup dry Aproten Riso rice
boil water first, then add the rice stir as soon as added to avoid clumping
follow package for instructions and cooking timie
2. While rice is cooking, make "meat" patty using chicken or mushroom burger mix.
3. Pan-fry the patty
4. Cut cooked patty into bite size pieces
5. Once the rice is done, drain and place in a bowl
6. Add chopped "meat" and add toppings

Nutritional breakdown of this recipe depends on which toppings and seasoning/sauces that you use and the amount of rice and "meat" patty used as the base.

ENJOY!

Suzanne's Corner

Karleigh's Angel Food Cake

Enjoy this delicious low fat dessert made with love by Karleigh's grandmother!

Preheat oven to 350 F (cooking temperature)
Cooking time: 35-40 minutes

Amount	Ingredient
1 ¼ cup	Egg white
1 ½ cup	Sugar, divided
1 cup	Cake flour
1 ¼ teaspoon	Cream of tartar
1 teaspoon	Vanilla
¼ teaspoon	Almond extract
¼ teaspoon	Salt

1. Place egg whites in a large bowl and let stand at room temperature for 30 minutes
2. Place oven rack in lowest position and preheat oven to 350 F
3. Sift ½ cup sugar and all the flour together twice. Set aside.
4. Add cream of tartar, extracts and salt to egg whites
5. Beat on medium speed until soft peaks form
6. Gradually add in remaining sugar, about 2 tablespoons at a time, beating on high speed until stiff peaks form
7. Gradually fold in flour mixture, about ½ cup at a time
8. Gently spoon into an ungreased tube pan. Cut through batter with knife to remove air pockets
9. Bake until lightly brown and entire top appears dry
10. Immediately invert pan, cool completely for 1 h
11. Run a knife around side and centre tube of pan
12. Remove cake to serving plate

Divide cake into 8 slices.

Per slice
5.5 g protein
0.2 g fat
51 g carbohydrate

Karleigh, VLCAD deficiency, making low fat dishes

Staff Updates

Carrie Charters

I'm so grateful for the warm welcome I have received since I started providing social work support to the Metabolics program! My name is Carrie Charters and I am a masters-level social worker stationed predominantly in paediatrics, though very much enjoying working with the adult patients I've had the privilege to spend time with from Metabolics so far. I provide emotional support to patients and families grappling with the identity-shifts and challenges that can come with chronic diagnoses, information and referral to appropriate community-based resources, and advocacy for increased support within the systems you're already connected to. I'm only a phone call away so please don't hesitate to connect – 519-685-8500 x 56149.

Mia Sethna

I am so thrilled to have joined the metabolic team at LHSC! I currently practice general pediatrics in London with a focus on infant feeding and nutrition. However, I have always been interested in genetics and worked in a cytogenetics laboratory before pursuing a career in medicine. I am looking forward to learning from the amazing metabolic team and offering a pediatrician's perspective when caring for our inspiring patients and their families. I live here in London with my husband and our three children who keep us on our toes and make every day an adventure! I hope everyone has a happy, healthy 2022 and I look forward to meeting you!

A big congratulations to Dr. Natalya Karp on her promotion to associate professor! Dr. Karp is such a valuable member of our metabolic team and this is a much deserved milestone.

Congratulations to Sue Maclean on your retirement! Thank you for your dedication over the years, and all the best in your next chapter!

Research and Presentation Updates

Clin Pediatr (Phila) 2021 Nov 18;99228211059668. doi: 10.1177/00099228211059668. An Unexpected Finding of Hepatosplenomegaly in a Pediatric Patient. Hailey C Barootes, **Chitra Prasad**, **C Anthony Rupar** , Dhandapani Ashok. PMID: 34789027 DOI: 10.1177/00099228211059668.

Pediatrics. 2021 Aug;148(2):e2020037747. doi: 10.1542/peds.2020-037747. Core Outcome Sets for Medium-Chain Acyl-CoA Dehydrogenase Deficiency and Phenylketonuria. Pugliese M, Tingley K, Chow A, Pallone N, Smith M, Chakraborty P, Geraghty MT, Irwin JK, Mitchell JJ, Stockler S, Nicholls SG, Offringa M, Rahman A, Tessier LA, Butcher NJ, Iverson R, Lamoureux M, Clifford TJ, Hutton B, Paik K, Tao J, Skidmore B, Coyle D, Duddy K, Dyack S, Greenberg CR, Jain Ghai S, **Karp N**, Korngut L, Kronick J, MacKenzie A, MacKenzie J, Maranda B, Potter M, **Prasad C**, Schulze A, Sparkes R, Taljaard M, Trakadis Y, Walia J, Potter BK; Canadian Inherited Metabolic Diseases Research Network. Epub 2021 Jul 15. PMID: 34266901.
JIMD Rep. 2020 Oct 1;57(1):9-14. doi: 10.1002/jmd2.12171Two cases of carbonic anhydrase VA deficiency-An ultrarare metabolic decompensation syndrome presenting with hyperammonemia, lactic acidosis, ketonuria, and good clinical outcome. Marwaha A, Ibrahim J, Rice T, Hamwi N, **Rupar CA**, Cresswell D, **Prasad C**, Schulze A. eCollection 2021 Jan. PMID: 33473334.

J Investig Med High Impact Case Rep. 2021 Jan-Dec;9:23247096211022484. Abetalipoproteinemia Due to a Novel Splicing Variant in MTTP in 3 Siblings. Vlasschaert C, McIntyre AD, Thomson LA, Kennedy BA, **Ratko S**, **Prasad C**, Hegele RA. doi: 10.1177/23247096211022484. PMID: 34078172.

Patient. 2021 Jul 20:1-15. doi: 10.1007/s40271-021-00538-8. Family Experiences with Care for Children with Inherited Metabolic Diseases in Canada: A Cross-Sectional Survey. Chow AJ, Pugliese M, Tessier LA, Chakraborty P, Iverson R, Coyle D, Kronick JB, Wilson K, Hayeems R, Al-Hertani W, Inbar-Feigenberg M, Jain-Ghai S, Laberge AM, Little J, Mitchell JJ, **Prasad C**, Siriwardena K, Sparkes R, Speechley KN, Stockler S, Trakadis Y, Walia JS, Wilson BJ, Potter BK. Online ahead of print. PMID: 34282509.

Poster Presentation Garrod 2021. Proactive Management of Carbonic Anhydrase (CA-VA) deficiency (an under recognized cause of neonatal hyperammonemia) in a known affected newborn. **Chitra Prasad**, **Natalya Karp**, **Suzanne Ratko**, **Melanie Napier** and **Charles A Rupar**.

Poster Presentation Garrod 2021. Inborn Errors of Metabolism Presenting as Movement Disorders in Children. Asuri N Prasad, CA Rupar, Chitra Prasad.

Poster Presentation Garrod 2021.The Canadian Inherited Metabolic Disease Research Network: A population-based cohort of Canadian children diagnosed with rare inherited metabolic disorder. K Tingley, M Pugliese, M Lamoureux, BK Potter, MT Geraghty, JB Kronick, D Coyle, K Wilson, V Austin, C Brunel-Guitton, D Buhas, AKJ Chan, S Dyack, A Feigenbaum, S Goobie, C Rockman-Greenberg, S Jain Ghai, M Inbar-Feigenberg, **N Karp**, M Kozenko, E Langley, M Lines, J Little, J MacKenzie, B Maranda, S Mercimek-Andrews, A Mhanni, G Mitchell, JJ Mitchell, L Nagy, A Pender, M Potter, **C Prasad**, **S Ratko**, R Salvarinova, A Schulze, K Siriwardena, N Sondheimer, R Sparkes, S Stockler-Ipsiroglu, Y Trakadis, L Turner, C Van Karnebeek, H Vallance, A Vandersteen,J Walia, BJ Wilson, AC Yu and P Chakraborty.

Poster Presentation Garrod 2021. A qualitative analysis of caregiver experiences with care for children with inherited metabolic diseases in Canada. Andrea Chow, Michael Pugliese, Laure Tessier, Pranesh Chakraborty, Doug Coyle, Jonathan B. Kronick, Kumanan Wilson, Robin Hayeems, Walla Al-Hertani, Michal Inbar-Feigenberg, Shailly Jain-Ghai, Anne-Marie Laberge, Julian Little, John J. Mitchell, **Chitra Prasad**, Komudi Siriwardena, Rebecca Sparkes, Kathy N. Speechley, Sylvia Stockler, Kylie Tingley, Yannis Trakadis, Jagdeep Walia, Brenda J. Wilson, and Beth K. Potter.

Poster Presentation Garrod 2021. Assessing the value of metabolic chart data for capturing core outcomes for paediatric medium-chain acyl-CoA dehydrogenase (MCAD) deficiency. Ryan Iverson, Pranesh Chakraborty, Monica Taljaard, Michael Pugliese, Kylie Tingley, Doug Coyle, Jonathan B. Kronick, Kumanan Wilson, Valerie Austin, Catherine Brunel-Guitton, Daniela Buhas, Alicia K. J. Chan, Sarah Dyack, Annette Feigenbaum, Michael T. Geraghty, Sharan Goobie, Cheryl R. Greenberg, Shailly Jain Ghai, Michal Inbar-Feigenberg, **Natalya Karp**, Mariya Kozenko, Erica Langley, Matthew Lines, Julian Little, Jennifer MacKenzie, Bruno Maranda, Saadet Mercimek-Andrews, Aizeddin Mhanni, John J. Mitchell, Laura Nagy, Amy Pender, Murray Potter, **Chitra Prasad**, **Suzanne Ratko**, Ramona Salvarinova, Andreas Schulze, Komudi Siriwardena, Neal Sondheimer, Rebecca Sparkes, Sylvia Stockler-Ipsiroglu, Kendra Tapscott, Yannis Trakadis, Lesley Turner, Clara Van Karnebeek, Anthony Vandersteen, Jagdeep Walia, Brenda J. Wilson, Andrea C. Yu, and Beth K. Potter on behalf of the Canadian Inherited Metabolic Diseases Research Network.

Research and Presentation Updates

Poster Presentation Garrod 2021. Age-specific Trends in Blood Phenylalanine Levels as an Indicator of Metabolic Control Associated with Pediatric PAH Deficiency Management in Canada. Nataliya Yuskiv , Beth K. Potter, Sylvia Stockler-Ipsiroglu, Michael Pugliese, Kylie Tingley, Monica Lamoureux, Michael T. Geraghty, Jonathan B. Kronick, Doug Coyle, Kumanan Wilson, Valerie Austin, Catherine Brunel-Guitton, Daniela Buhas, Alicia K. J. Chan, Sarah Dyack, Annette Feigenbaum, Alette Giezen, Sharan Goobie, Cheryl R. Greenberg, Shailly Jain Ghai, Michal Inbar-Feigenberg, **Natalya Karp**, Mariya Kozenko, Erica Langley, Matthew Lines , Julian Little, Jennifer MacKenzie, Bruno Maranda, Saadet Mercimek-Andrews, Connie Mohan, Aizeddin Mhanni, Grant Mitchell, John J. Mitchell, Laura Nagy, **Melanie Napier**, Amy Pender, Murray Potter, **Chitra Prasad**, **Suzanne Ratko**, Ramona Salvarinova, Andreas Schulze, Komudi Siriwardena, Neal Sondheimer, Rebecca Sparkes, Yannis Trakadis, Lesley Turner, Clara Van Karnebeek, Hilary Vallance, Anthony Vandersteen, Jagdeep Walia, Ashley Wilson, Brenda J. Wilson, Andrea C. Yu, and Pranesh Chakraborty on behalf of the Canadian Inherited Metabolic Diseases Research Network.

Poster Japanese Neurology Society. Long-term Follow-up of Primary Neurotransmitter Disorders- Single Centre Experience (2004-2021). Shyann Hang, **Chitra Prasad**, **C Anthony Rupar**, **Asuri N Prasad**

Oral presentation Clinical excellence in managing urea cycle disorders. “Neonatal Hyperammonemia- A good news story! **Chitra Prasad**, **Natalya Karp**, **Suzanne Ratko**, **Melanie Napier**, **Tony Rupar**

A Virtual Education/Chat Session for Parents and Primary Caregivers

Join us for an interactive Chat session on Shared Decision Making

Parents of children with complex medical needs are faced with many challenging medical decisions. This session will provide tools, strategies and information to help parents and primary caregivers meet the challenges of decision making for their child. The session will cover:

SDM Team Members :
Bonnie Wooten, Decision Coach
Jennifer Banting, Family Resource Facilitator
Carrie Charters, Paediatric Social Worker
Rhonda Teichrob, Social Worker TVCC

Upcoming Virtual Chat Sessions:

Date : January 27,2022
Time: 4:00- 5:00 pm
Link: **https://bit.ly/3eeytYn**

Date: February 12, 2022
Time: 4:00 – 5:00 pm
Link:**https://bit.ly/33Exnmr**

Date : Mar 3, 2022
Time: 4:00pm – 5:00pm
Link: **https://bit.ly/3yRFmbn**

Our Stars

Karson, PKU

Helen, LCHAD deficiency

Emily M, PKU

Ryder, Quorra, Logan, Abetalipoproteinemia, and Jenny

Margie, PKU

Emily T, PKU

Our Stars - Happy Holidays!

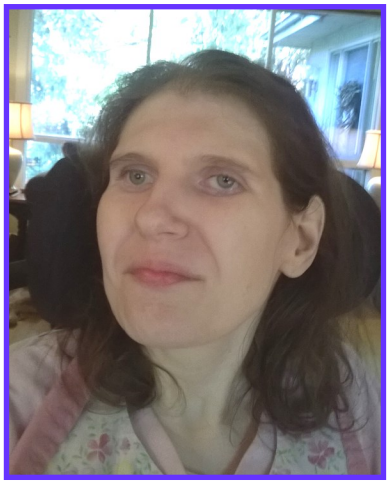
Irelyn, MCAD deficiency

Zayla, PKU

Luke (GSD IX), Everett, and Drew (GSD IX)

Bennett and Senna, PKU

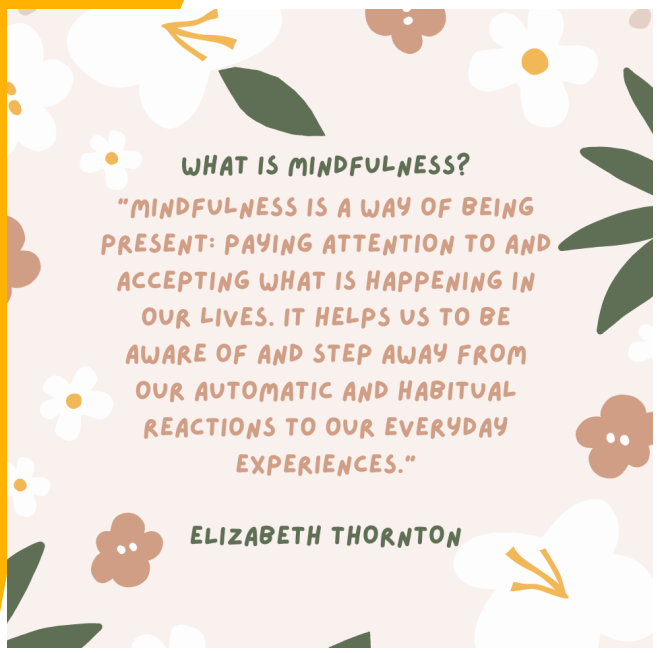
In Remembrance: Tara Cecile (Leukodystrophy)

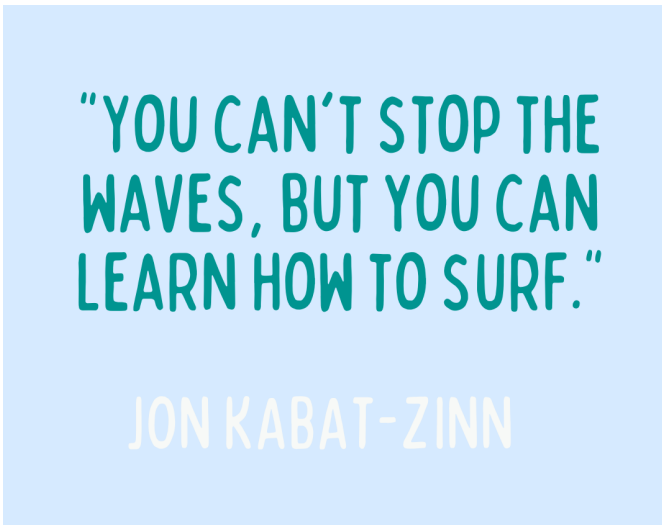


"On the day our precious Tara went to heaven, she took a piece of our hearts with her, but left behind 38 years of everlasting memories. As we reflect on her life, we are continuously reminded about the treasure and gift she was to our family. We will miss her and love her always ♥ (purple was Tara's favourite colour)"

The Cecile Family 💜

Mindfulness Strategies





Jon Kabat-Zinn's work as a scientist, writer, and meditation teacher is focused on mindfulness and introducing this concept and its importance to the mainstream world. We encourage you to visit **www.mindfulnesscds.com** for more about his work and to find resources about mindfulness.

Family Friendly Mindfulness Activities

Source: www.theottoolbox.com/fun-mindfulness-activities

Mindful breathing
Breathe in and out through your nose, feeling the flow of the breath.

Body scan
Lay on your back. Tense up your muscles for 10-15 seconds and then release and relax. Reflect on how your body feels.

Visualization or Guided Imagery
Find a guided imagery script online, slowly read it to your child/family member as they visualize the image in their minds. They can then draw what they visualized and keep this image.

Take a walk
Get some fresh air! Listen to the sounds: birds chirping, wind blowing. Notice the scents and scenery as you are out and about.

Stretching/Yoga
Dim the lights, play calming music, and do some stretches, being aware of how your body feels.

How to Make a Donation

Donated funds are used for future Metabolic Family Workshop Days as well as further teaching and education. If you wish to make a donation, please do so on **The Children's Health Foundation website:** www.childhealth.ca

Ways to Give / Under Giving Options: donate now / Select: Make a Donation or Join Caring Heart Monthly Giving / Follow the prompts and it will give an online form with a comment box in the payment area that you can type in and instruct the funds go to the ***Inherited Metabolic Disorders Program.***

If you would like to donate by phone with your credit card, please call 519.432.8564 or toll-free at 1.888.834.2496, Monday to Friday, 9 am to 5 pm.

Your donation is tax deductible, and an income tax receipt will be mailed to you for donations of $20 or more.
Thank you!

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