



The Inherited Metabolic Disorders News

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Winter 2021 Volume 18 Issue 1

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From the Editor

Hey there everyone, hope you have been keeping well! I can't believe we are coming up on one year since all of this craziness started! Be sure to check out the "What's New" section of the newsletter for important event updates. As always, please send any photos or artwork to meghan.zadorsky@lhsc.on.ca. We love to see what you have been up to and all of your smiling faces, now more than ever!



Meghan Zadorsky

From Dr Chitra Prasad



Dear Friends,
Greetings for the year 2021!

I hope you and your families are safe during these unprecedented times. We have all learnt a lot during the last year dealing with the worldwide Coronavirus pandemic. Our metabolic team and you all have adapted to the virtual care. Our team has seen many of you through OTN and talked to you on the phone. While it is not the same as meeting in person, I am still very grateful for these modern inventions. I am also very thankful to our metabolic team (Dr. Natalya Karp, Rana, Suzanne, Sue, Melanie and Dr. Tony Rupar and his laboratory team) for their hard work and dedication during these times.

Our family story is from Urszula, Dominik, Claire and Helen. Thanks so much for sharing your journey about LCHAD (a very complex long chain fatty acid oxidation disorder). With help of newborn screening the diagnosis has been made early and under Suzanne's guidance we hope for the very best for dear Helen. I am amazed at how much resilience Urszula and family have shown, adapting to a different diet, frequent blood draws and hospital visits. Each one of you show extraordinary courage in the face of unknown.

This brings me to the fact that unfortunately we won't be having our Metabolic Family Workshop this year but let us keep our hope for having it next year. I have been reading and reflecting a lot during the last year and have come across Stoic philosophy. Meghan has done a wonderful job summarizing some of these principles of stoicism for all of us in the current newsletter which are very helpful. Thanks Meghan!

I like this quote by Marcus Aurelius (Stoic philosopher King) (26 April 121 – 17 March 180 AD) Roman emperor

"Have power over your mind — not outside events. Realize this, and you will find strength"

With very best wishes
Chitra Prasad



"Faith is taking the first step even when you don't see the whole staircase."— Martin Luther King Jr.

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Personal Stories



Helen

The way in which we learned about Helen's LCHAD (Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency), was thankfully through the newborn screening test (NBS). And so, from the age of 8 days, Helen has been under the careful care of her medical team – which consists of Dr. Andrea Yu (now in Ottawa) and Dr. Chitra Prasad – pediatric genetics and metabolism, Suzanne Ratko – dietician, Dr. Luis Altamirano-Diaz - pediatric cardiology, and Dr. Inas Makar - pediatric ophthalmology. Through their insightful guidance we have been able to manage and maneuver around the strange dietary obstacles that LCHAD presents its patients.

When people ask us how we reacted to Helen's diagnosis – after all, no one we know has ever heard of LCHADD and how it completely takes over your life management – we smile and say that we just take it day by day. Of course, the initial phone call advising us that Helen's blood sample came back positive for one of the NBS diseases was a complete shock and left us scrambling to find out as much as we could, before even meeting with the medical team. Was that a good idea? Probably not, as the internet does not have too much information about LCHADD, and

what it does have can raise the hair on the back of your neck – like the short life expectancy, childhood blindness, developmental challenges, possible link to SIDS, and general wasting of the little body – and this is all because the body is unable to change some of the fat eaten into energy that the body needs to function. Thankfully, when we did meet with all the doctors, they put our fears at ease and went into extreme detail outlining what the disease is, and how we will adjust Helen's diet so that she has the best chance of survival. What followed next was a battery of blood and urine tests, which would confirm the initial diagnosis; cardio screening to rule out neonatal hypoglycemia and cardiomyopathy; an eye exam that would establish a baseline of Helen's vision; genetic testing; as well as immediate enrollment into the National Food Distribution Centre food supply, which would secure for Helen the formula she will need to consume in order to thrive. The first immediate change that we had to make in Helen's diet was the removal of breast milk – from now on she would drink special formula (Lipistart) designed for fatty acid oxidation disorders with just a splash of breast milk. Through the 90% Lipistart and 10% breast milk combination, we were able to strike a "fat balance", by giving Helen some very long chain fats (from the small amount of breast milk), skip over the ones she couldn't digest (long chain fats) and go right into the ones that her body can work with - medium chain fats. The inability to bond with Helen though breast feeding was a little devastating for me personally as I quite enjoyed nursing our first daughter and was looking forward to doing the same again. What I was able to do instead for Helen, was to provide her with as much milk as possible, before we would have to resort to using over the counter formula instead. And so, with some effort and determination I was able to pump enough milk to last Helen for over 3 months, before we had to introduce regular over the counter formula. Thankfully, Helen took well to being bottle fed and she had no major issues digesting the formulated milk. The challenge was then the timing of Helen's feedings and the maximum allowable fasting – in her case, she was fed every 3 hours around the clock for about 3 months, then 4 hours and very recently she has moved to formula every 5 hours with carefully prepared and measured meals in between.



After the initial diagnosis, in order to make sure that she was gaining enough weight under the new feeding regime, we would visit our family doctor once a week and check Helen's weight and general development. At first we considered buying a baby scale and simply doing this at home, but I think we would have driven ourselves crazy and likely weighed her after every meal and "heavy" diaper. By doing it once a week, we kept our sanity and enjoyed a tiny break in our daily routine of eat, sleep, change diaper. I am happy and proud to report that Helen has been gaining weight steadily and went from being in the 60th percentile at 6 days old to 87th percentile at 2 months old and she has managed to maintain this position.



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As with any child, there are good days and there are tough days. Through quick intervention of the metabolic team and access to telephone consultation 24/7, we were usually able to manage a lot of common illnesses and keep them at bay – especially by quickly adjusting Helen’s calorie, fluid and fat intake. However, one day in early June 2020, Helen’s fever would just not go down and she started to shake. Consulting with her medical team, a decision was made to rush her to Children’s Hospital and get checked out by the emergency team. Helen’s medical Emergency Protocol was sent down to the admitting station and once we arrived, the nurses and doctors understood – that even though Helen is arriving at the hospital with just a fever and no other signs of illness, that she must be admitted quickly and seen by a doctor – her fasting window was closing on us quickly and she would

need an IV with fluids shortly. What followed was a battery of tests, blood samples, IV’s, x-rays, ECG and even a COVID test. We stayed at the hospital for 5 days while Helen regained her strength and appetite. Unfortunately, being a lively baby even when sick caused Helen’s IVs to sometimes fall out – needing numerous re-attachments. It was heart wrenching to watch her being hooked up to so many machines, and undergo so many tests. In the end, the infection remained a mystery and Helen recovered enough to be allowed to go home.

As Helen’s diet keeps changing, and we slowly introduce solids and more fat from food rather than milk formula, Helen continues to have her blood drawn and tested for enzyme activity. It is a necessary procedure that helps us understand how the fat is being absorbed and to make sure that we aren’t giving her too much. Unfortunately, she has now developed a bit of a fear of needles and reminds me every day that she has “pokies” on her arms. Thankfully we have learned that there is a very special person that can come to the hospital and help Helen deal with her fears. We look forward to meeting with Erika, Childlife Specialist, at our next visit.



In order to keep track of Helen’s daily intake of formula and fat, I have kept an electronic journal – which is shared on a weekly basis with the metabolic team. In one app, I have been tracking Helen’s daily activities – when she has a bottle, a meal, how much she has slept, number of diapers, how often she has vomited and when she had taken any type of medication. The other app – Cronometer – allows me to keep track of the amount of fat that she has consumed from various sources throughout the day. This comes especially handy when you are limited to eating only 6g of fat a day and you have to calculate the amount of fat per gram of food consumed. Thankfully a lot of the food we eat is already loaded into the app, which saves me from having to input what I see on the nutritional label. It also allows me to create my own recipes and by carefully weighing Helen’s meal before and after – I know exactly how much food and fat she has consumed and how much more she needs to eat in a given day. This app has been an absolute blessing as I was dreading the day Helen would start eating normal food and I would have no clue how much fat she had eaten. What also has been great, is the whole family’s adaptation of our new eating regime, and everyone is on board and understands that Helen can’t eat just “anything” – that all food must be carefully considered. As time has progressed, Helen has slowly opened herself up to try new foods and who would have known that *dill pickle soup* would become one of her favorite meals?!

Our little family has been immensely grateful to the whole Metabolic and Genetics team over at LHSC and Children’s Hospital as well as the cardiac and ophthalmology teams. Suzanne’s guidance in diet modification has been a blessing. Without everyone’s help, Helen would not be where she is today – a silly, bouncy baby who just wants to explore and learn new things every day.

Thank you! Urszula, Dominik, Claire and Helen



Featured This Issue

LCHADD

(Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency)

Compiled by Dr. Chitra Prasad

Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency is a rare metabolic condition that prevents the body from converting certain fats to energy, particularly during periods without food (fasting).

Signs and symptoms of LCHAD deficiency typically appear during infancy or early childhood and can include feeding difficulties, lack of energy (lethargy), low blood sugar (hypoglycemia), weak muscle tone (hypotonia), liver problems, and abnormalities in the light-sensitive tissue at the back of the eye (retina). Later in childhood, people with this condition may experience muscle pain, breakdown of muscle tissue, and a loss of sensation in their arms and legs (peripheral neuropathy). Individuals with LCHAD deficiency are also at risk for serious heart problems, breathing difficulties, coma, and sudden death. Problems related to LCHAD deficiency can be triggered when the body is under stress, for example during periods of fasting, or during illnesses such as viral infections. Thankfully this disorder has now been included in the newborn screening so the management can be started right from first few days of life.

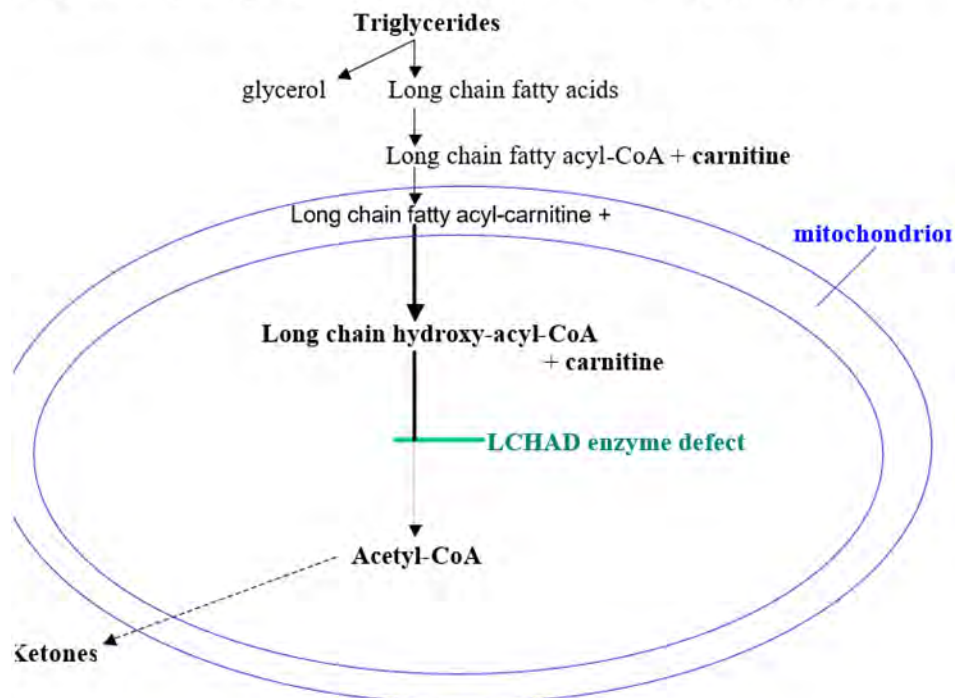
Management of LCHADD is focused on avoiding fasting and minimizing energy production from long-chain fatty acids. It requires a diet low in long-chain fatty acids, supplemented with medium-chain triglyceride oil. This decreases the incidence of hypoketotic hypoglycemia, and improves hypotonia, hepatomegaly, cardiomyopathy, and lactic acidosis. The role of a specialized dietitian like Suzanne Ratko is very essential in the management of this diet.

The disorder is inherited in autosomal recessive manner where the parents have a 25% risk of having another child with LCHADD. Genetic counselling is also an important part of the management.

References: <https://rarediseases.info.nih.gov/diseases/6867/lchad-deficiency>

Metabolic Pathway

Long chain hydroxy-acyl Co-A dehydrogenase deficiency (LCHADD)



Suzanne's Corner



Karson's Favourite Eggplant Dip

This recipe has appeared in a previous newsletter. It is delicious, flavourful and very easy to make. Great to use as a veggie dip or to spread on crackers

A second version is included for our families that have to watch their fat intake. The protein content is higher.

Ingredients

2 cups (1 medium = about 550 g)	Eggplant
6 cloves	Garlic
½ teaspoon	Salt
½ cup	Miracle Whip****
½ cup	Onion, chopped
1	Lemon



*** For low fat version, replace ½ cup Miracle Whip with ½ cup fat free plain yogurt (such as Greek yogurt, plain)

Preparation

- Preheat oven to 400 degrees
- Cut the eggplant in half and bake for 30 minutes or until tender
- Allow eggplant to cool slightly for about 15 minutes, then peel off the skin; discard the skin
- Place the eggplant and the remaining ingredients in a blender or food processor
- Whip until fully blended
- Season as necessary

The recipe indicates that it makes 16 servings with each serving as 28 g or 2 Tablespoons

	Protein	Fat
<u>Entire recipe</u>		
With Miracle Whip	8.8 g	17.2 g
With fat free yogurt, Greek	19.4 g	2.1 g
<u>Per 28 g or 2 Tablespoons</u>		
With Miracle Whip	0.55 g	1.1 g
With fat free yogurt, Greek	1.2 g	0.1 g
<u>Per 1 teaspoon</u>		
With Miracle Whip	0.1 g	0.2 g
With fat free yogurt, Greek	0.2 g	0.02 g



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Emmitt's Mug Cake

This is a very delicious low protein addition to any meal or snack.

4 Tablespoons	Cambrooke Mixquick
1 Tablespoon	Brown sugar
3 Tablespoons	Silk Almond Original beverage
1 Tablespoon	Oil
¼ Teaspoon	Cinnamon
1 Teaspoon	Vanilla



- Spray or grease your mug so that the cake does not stick
- Add all the dry ingredients first, then add the wet. Mix until combined
- Microwave on high for 1 minute
- Enjoy the cake right out of the mug with a spoon

Emmitt has a few suggestions:

- Add your favourite fresh or frozen fruit right before cooking
- Sometimes the cake is a little “raw” on top. If you do not like that, just microwave for another 20-30 seconds. “I like it gooey on top.”

1 mug	0.2 g protein	12 mg phenylalanine
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Find this recipe and more at <https://cookforlove.org/>

ROLL-OUT SUGAR COOKIES

by Brenda Winiarski



3 c (390g) Wheat Starch
3/4 tsp (2g) Xanthan Gum
1/2 tsp (3g) Salt
1 tsp (5g) Baking Powder
50g Vanilla Pudding, Instant, dry mix only

8 TBSP (112g) Butter, regular or unsalted
4 TBSP (48g) Vegetable Shortening (Crisco, Fluffo, etc.)
2 1/8 TBSP (30g) Cream Cheese
2 tsp (10g) Vanilla Extract
3/4 c (150g) Sugar, White Granulated
1/2 egg(s) (25g) Egg, large, lightly beaten
2 TBSP (30g) Water



NUTRITION INFO

Servings: 38

Serving size: 1 cookie

PER SERV	PRO	EXCH
PHE 10	361	
PRO 0.2	6.7	
EXCH 0.6	24.1	

INSTRUCTIONS

Step 1: In a medium sized bowl, combine wheat starch, xanthan gum, baking powder, salt and pudding mix, set aside.

Step 2: In the bowl of standing mixer fitted with paddle attachment, add the butter, cream cheese, and shortening. Mix until combined, about 30 seconds. Add the sugar, creaming until light and fluffy. Add the egg, water and vanilla extract. Add the dry ingredients and mix until the mixture forms a dough.

Step 3: Divide the dough in half, placing each half in a gallon sized Ziploc bag. With the top unsealed, lightly roll the dough with a rolling pin to 1 inch thick. Refrigerate until they begin to firm up, 20 to 30 minutes. (Can be refrigerated up to 3 days or frozen up to a month; defrost in refrigerator before using.)

Step 4: Adjust oven rack to middle position; heat oven to 350 degrees. Roll out 1 dough disk to even 1/4-inch thickness between 2 large sheets parchment paper; slide rolled dough on parchment onto baking sheet and chill until firm, about 10 minutes. Meanwhile, repeat with second disk.

Step 5: Using cookie cutters, cut shapes from one piece of dough and bake on parchment-lined baking sheet in 350-degree oven until light golden brown, 10 to 12 minutes.

Step 6: Remove from oven and transfer (on the parchment paper) to a rack for 5 minutes. Remove from parchment and allow to cool completely.

Step 7: Decorate with Royal Icing, if desired.

NOTES

If the phe content is a little too high for you, replace the egg with egg replacer & replace the cream cheese and vanilla pudding with 2 tablespoons shortening, a full package of cheesecake flavored instant pudding and reduce the sugar by 1/4 cup. The cookie will be a little more fragile, so roll it to 1/4 inch instead of 1/2 inch and cook an extra 2 minutes.

This recipe is copyright 2021 CookForLove.org and may not be reproduced or distributed without express permission from Cook For Love and National PKU News.



Daphne, PKU



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Resources

Shared by Childlife Specialist, Erika Clements

Coronavirus: How to Support Your Child

Review each step and ask your child the following questions.



Wonders & Worries

Professional support for children
through a parent's illness.

We will, together.

C **Calmly** adjust to new information and lifestyle changes. The calmer you are, the calmer your child will be.
How has your daily routine changed, and how do you feel about those changes?

O **Observe** where, and how often, your child receives information about coronavirus.
What have you heard about coronavirus?

V **Validate** your child's feelings and concerns.
How do you feel about the information you've heard?

Many people feel the same way.

I **Identify** the facts and address any questions and misinformation.
What questions do you have about coronavirus?

D **Discuss** what we can control, including washing hands, practicing social distancing, and staying home.
What are some things we can control during this time?

1 Stay up-to-date with 1 reliable source, such as the CDC. Limit exposure to the news and social media.
What helps you feel relaxed?

9 As a family, express gratitude for 9 different things each day to enhance positive thinking and improve mental wellness.
What are you grateful for? Let's pick 9 things together!

What's New

Postponed!

Metabolic Family Workshop

With everyone's health as our number one concern, we have made the difficult decision to postpone the Metabolic Family Workshop that was scheduled for May 14, 2021. We cannot wait to see everyone as soon as the pandemic situation allows. We will continue to provide updates in the upcoming issues of the Inherited Metabolic Disorders News, so keep posted!

Virtual Low Protein Pantry

On Saturday, November 28th, 2020, we held our first virtual low protein pantry presented by Anthony from Innomar. It was great to "see" everyone so engaged and interested in learning more about low protein foods available at local grocery stores. Thank you to Innomar, Biomarin and our families for making this event so successful.



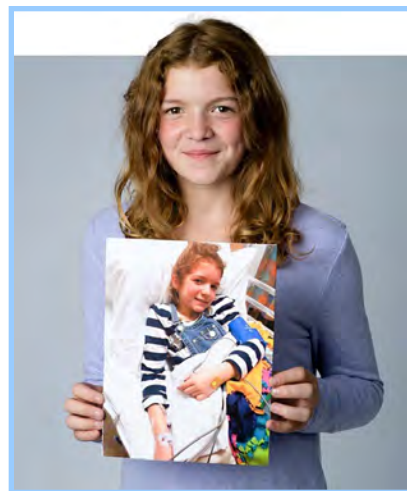
Teaghan

Children's Miracle Network Champion Child 2021

Being Champion Child has been an amazing experience, and I am both honoured and excited to be able to continue on to be the Champion Child for 2021. It has been a lot of fun doing videos, phone interviews and Zoom calls. My experience as a Champion Child is obviously different than my predecessors as my family and I didn't get to go on the annual Florida trip to meet the other Champion Children and their families, and I haven't been able to tour the Walmarts to share my story; but my experience has been very special and truly

memorable. It has been great meeting amazing people and making friendships that will last a lifetime. It is hard to put into words just how much the experience as a Champion Child has changed my life and has brought me out of my shell. I am excited to see what the next year brings.

If you would like, you can follow my story on Instagram and Facebook by searching "Tenacious_Teag".



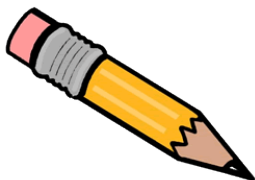
Students



A very special congratulations to Nitya (a student of Dr. Rupar and Dr. Prasad) on completing her Master's degree!

MSc. project title: **Effects of increased sulfatide in Schwann and Mesenchymal stromal cells in metachromatic leukodystrophy.**

Supervisor: Dr. Tony Rupar



Sam (right; student working with Dr. Prasad and Dr. Rupar on late onset OTC deficiency (urea cycle disorder)

Prashanth (left; student working with Dr. Prasad, Dr Rupar, Dr Narayan Prasad and Dr. Siu on Leigh disease in the Mennonite population)



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Our Talented Artists and Stars



Roqaya, PKU



Aaliyah, Gaucher



Karson, PKU



Danny, hyperphenylalaninemia



Elvina, MCADD



Ellen, MCADD



Jordynn, MTHFR deficiency



Eileen, MCADD



Emmitt, PKU

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Inspirations

Stoic Philosophy

Being deep into the second wave of the COVID-19 pandemic, it is easy to get overwhelmed with all of the impacts it has had on our lives. While it is sometimes easier to look at all the pandemic has taken from us and all of the suffering it has caused, it certainly doesn't make us feel better dwelling on the negatives. Stoic philosophy is a way of living that can make us more resilient and happier, and can especially help us reframe this pandemic as an opportunity instead of a just a tragedy. Stoicism focuses on four main virtues that we can try to practice in our daily lives to help us through not only this crazy time, but all challenging situations in our lives and to use them as opportunities for growth.

Courage

“Sometimes even to live is an act of courage.”

— Seneca

Hardships and obstacles like this crazy pandemic can be scary, especially as many things have become uncertain. Stoic philosophy challenges us to use difficult situations as an opportunity to see just what we are capable of handling.



Temperance

“We are what we repeatedly do, therefore excellence is not an act but a habit.”— Aristotle

Temperance refers to moderation or self-restraint. It focuses on the idea of living a balanced life; not doing too little or too much in any aspect. While this can feel like an abstract thought, in terms of daily life, this virtue promotes that we have the power to make big changes in our lives by simply making habits of little things.

Justice

“Live your life in truth and justice, tolerant of those who are neither true nor just.”— Marcus Aurelius

Justice is said to be the most important virtue to the stoics because it was felt that justice influences all other virtues. This virtue focuses on doing the right thing and believing that an individual can make a difference. Thinking about the pandemic, this virtue especially rings true as we have all been called to make responsible choices to help with the fight against COVID-19. While it will take a collective effort to overcome our situation, that collective effort begins with each and every one of us as individuals.



Wisdom

“You cannot learn that which you think you already know.”— Epictetus

The final virtue of wisdom encourages us to always being open to learning. It is said that we have two eyes, two ears, and one mouth so that we should be observing and listening twice as much as we should be speaking. It is valuable to be able to look at every experience, good or bad, as an opportunity to learn and grow.

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“A hero is an ordinary individual who finds the strength to persevere and endure in spite of overwhelming obstacles.”

– Christopher Reeve



Serenity Prayer
Reinhold Niebuhr

GOD,
grant me the
Serenity
to accept the things
I cannot CHANGE;
Courage
to CHANGE
the things I can;
and
Wisdom
to know the DIFFERENCE.

“Never believe that a few caring people can’t change the world. For indeed, that’s all who ever have.”

– Margaret Mead



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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