The Inherited Metabolic Disorders News

Children's Hospital
London Health Sciences Centre

Summer 2021 Volume 18 Issue 2

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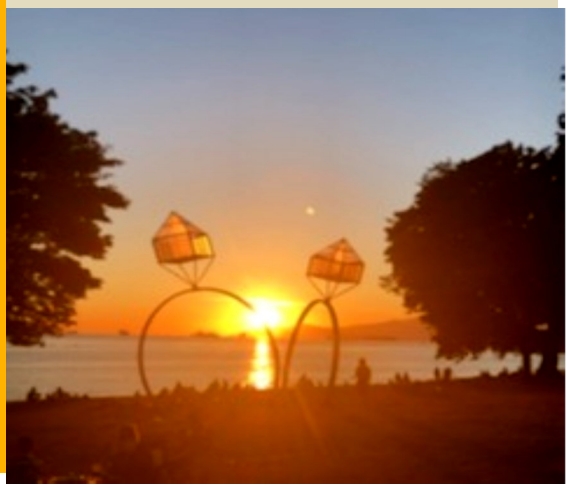
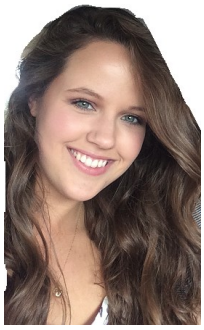
In this Issue



From the Editor



Hey there everyone! Hope you have all been keeping well! It's with mixed emotions that I share that this will be my last newsletter as I will be attending Sarah Lawrence College this fall to begin studying to become a genetic counsellor. I want to thank everyone for sharing their stories, photos, and artwork with me over the last few years; it has been so much fun! I will definitely be keeping an eye on what everyone is up to so please be sure to keep sharing for future issues! All the best!

Meghan Zadorsky

From Dr Chitra Prasad

Dear Friends,
Greetings

I hope that all of you are remaining safe during the pandemic. These are certainly challenging times however we have also seen unprecedented cooperation amongst the scientists all over the world to create the vaccine for COVID-19. I am also thankful for Zoom and other virtual platforms as they have allowed us to stay connected. We are continuing to see patients in person as well as in a virtual manner. I'm truly impressed with our children who have adapted so well to online learning. Let us hope that the pandemic gets resolved and we are able to interact again in person with our friends and families. In this newsletter we bring to you the story of two amazing sisters, Christine and Joan Park. In fact, they were two of the first patients that I saw in the metabolic clinic when I arrived at LHSC in 2003, about 18 years ago. They both are doing amazingly well in their respective fields in spite of having a very challenging metabolic disorder: glycogen storage disorder type 1a. Christine has also undergone liver transplant. Suzanne Ratko, our metabolic dietitian, has done a wonderful job looking after both the sisters. I would also like to congratulate their parents for their constant support and encouragement.

I would like to thank Meghan Zadorsky, our genetics assistant, who will be starting her genetic counselling program in the fall, for her great contributions to the metabolic newsletters and the Metabolic Family Workshop. Unfortunately, we will not be having the Metabolic Family Workshop this year as you're aware but I hope that we all can stay connected through the metabolic newsletters. Please continue to share your stories, your achievements, and your pictures with us.

As many of you might be aware we have all been shaken with the loss of a Muslim family in London Ontario who were unfortunately killed. These events certainly remind us to remain compassionate towards ourselves and others. Our prayers are with the young boy who has survived the attack. In this issue of newsletter, we bring to you some insights about self-compassion by Dr. Kristin Neff.

Wishing you all a great summer and best wishes for healthy times.

Your friend,
Chitra Prasad

"Compassion and tolerance are not a sign of weakness, but a sign of strength."— Dalai Lama

Personal Stories

Christine & Joan

It all started when my parents brought me into the world on January, 31, 1997. They thought they had a healthy baby girl. Little did they know that in a few months, I would begin showing symptoms of a sick child and would be diagnosed with Glycogen Storage Disease Type 1a at 6 months old. This was only the beginning chapter of a life my parents never imagined or prepared for; a life filled with hospital stays, dietary restrictions, and a sea of concerns for the future.

My name is Christine, and I was born with Glycogen Storage Disease Type 1a. In layman's terms, GSD is a rare genetic disorder caused by a deficiency in one of the enzymes that breaks down glycogen into glucose. This allows for the excessive accumulation of glycogen in the liver and kidneys, and as a result, interferes with the body's ability to process food into energy and maintain normal blood sugar levels. Due to this missing enzyme, those affected by GSD display an enlarged liver as a response to the glycogen build-up, and are prone to hypoglycemia without a controlled diet. As a child, I was an extremely picky eater and I would refuse to eat anything my parents would give me. This was obviously terrible for my GSD management and it would cause me to experience episodes of hypoglycemia. My parents brought me to LHSC in hopes of answers and guidance, and it was through those doors that they met Dr. Chitra Prasad and registered dietician, Suzanne Ratko. I was 4 years old when I was introduced to the metabolic team, and from the day I was placed under the care of Dr. Prasad and Suzanne, our lives were changed for the better. My parents and I were shown that it was possible to live a normal, healthy life while dealing with GSD. To help control my blood sugar levels, I was given a regimen of cornstarch and water as my main "medicine" during the day, and a gastrostomy tube that delivered a feeding formula to keep me stable during the night. I was advised of a diet restrictive of foods that consisted of sugars and lactose to help manage the effects of GSD and subdue further risks associated with the disorder. Although following a controlled diet was difficult at times for me, especially as a growing child, Suzanne always made it manageable by providing me with alternative options and resources to help compensate for the foods I wasn't allowed to have.

It was only two years after coming to Canada and joining the metabolic team at LHSC, when my younger sister, Joan, was born. It was discovered that she too had the same rare condition, Glycogen Storage Disease Type 1a. As overwhelming as this was for my parents; in hindsight, it was truly a blessing in disguise. Since the both of us were diagnosed with the same disorder, we always had one another to lean on and learn from as we navigated through the obstacles that came with having GSD. Joan was also placed under the same treatment plan as myself, and through the metabolic team's diligent efforts, both of us are able to live happy, and relatively healthy, normal lives. We were able to attend school, join extracurricular activities, and go travelling. These were always concerns my parents had for us, as they did not want us to miss out on our childhoods due to our health condition. The metabolic team at LHSC provided ways to conveniently deal with the hindrances associated with GSD.

In my last year of high school, Dr. Prasad and Suzanne were able to transition me from my night time g-tube feeds to Glycosade, a newly marketed formula that mimicked the effects of cornstarch. It was essentially a modified cornstarch that allowed me an 8-hour window of "stabilized" blood sugar levels compared to the 4-hour window of my regular cornstarch. This was a giant progression in my GSD treatment as this option allowed me to have more freedom and normalcy in my life. Thanks to this transition, I was able to move out to Ottawa for university and live there independently with no major issues or hospital visits. However, things took a slight turn for the worse in 2019, when my most recent ultrasound and MRI scans showed significant adenoma growths (benign tumors) on my liver. Although these adenomas were not deemed harmful, it was concerning to Dr. Prasad as the progression of these growths were rapid, and the cancer risks associated with these tumors. I was given two choices at that point: either continue on with my treatment while monitoring the adenoma growth, or sign up as a recipient for a liver transplant. The idea of a transplant was not a new concept to me, but it was always viewed as a last resort as it was such a daunting procedure. However, I decided that a transplant was worth undergoing for my situation, and thus marked the beginning of my transplant journey. Fortunately, I didn't have to wait long for a donor. It was only 6 months of being on the waitlist when I got the call saying I had a matching donor available. I will forever be grateful for the donor family and their selflessness to allow me a second chance at a healthier life. On April 22, 2020, amidst the beginning of the pandemic lockdown, I underwent a successful liver transplant at LHSC University Hospital led by Dr. Andrew Skaro and Dr. Ephraim Tang.

A year later, my new donor liver is healthy and functions normally, but I still have Glycogen Storage Disease Type 1a present in my genes and specifically, my kidneys. As a result, I still have to follow the same guidelines that were used to treat my old liver, but with looser restrictions this time. I am grateful to still be cared for and monitored by Dr. Prasad and Suzanne as they have impacted my life in more ways than one could imagine. Without their help and support, my sister and I would not be where we are today. I am now 24 years old, have completed my undergrad, and am currently working from home. By June, my sister Joan will graduate high school and attend university in the fall to study nursing; a field she found passion and interest in once she recognized that her experiences as a patient would enable her to connect with others. This is to say that none of this would be possible without the never-ending support provided by Dr. Prasad, Suzanne Ratko, and the rest of the metabolic team at LHSC. Throughout our GSD journeys, we have come to realize that having a metabolic disorder is not the end-all be-all, and it should not limit one's quality of life. We hope that by sharing our story, it will show other families that there is always a light at the end of the tunnel.

Featured This Issue

Glycogen Storage Disease Type 1a (Von Gierke disease)

Compiled by Dr. Chitra Prasad

Glycogen storage diseases are a group of disorders in which stored glycogen cannot be metabolized into glucose to supply energy and to maintain steady blood glucose levels for the body. Glycogen storage disease type I (GSDI) is characterized by accumulation of excessive glycogen and fat in the liver and kidneys that can result in an enlarged liver and kidneys and growth delay leading to short stature. GSDI is associated with abnormalities (mutations) in the G6PC gene (GSDIA). These mutations, which account for approximately 80% of GSDI, result in enzyme deficiencies that block glycogen breakdown in affected organs causing excess amounts of glycogen and fat accumulation in the body tissues and low levels of circulating glucose in the blood. This type of GSDI is termed glycogen storage disease type Ia. The enzyme deficiency also results in an imbalance or excessive accumulation of other metabolites, especially lactate, uric acid, and fats like lipids and triglycerides.

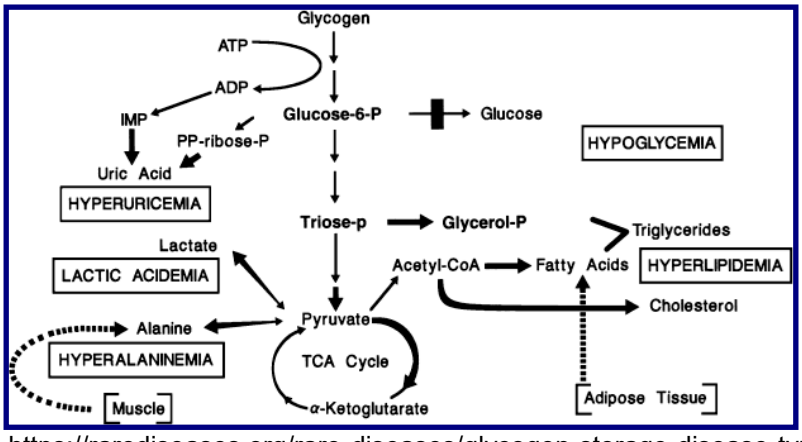
The primary symptom of GSDI in infancy is a low blood sugar level (hypoglycemia). Symptoms of GSDI usually begin at three to four months of age and include enlargement of the liver (hepatomegaly), kidney (nephromegaly), elevated levels of lactate, uric acid and lipids (both total lipids and triglycerides), and possible seizures due to repeated episodes of hypoglycemia. Continued low blood sugar can lead to delayed growth and development and muscle weakness. Affected children who are untreated typically have doll-like faces with fat cheeks, relatively thin extremities, short stature, and protuberant abdomen.

High lipid levels can lead to the formation of fatty skin growths called xanthomas. Other conditions that can be associated with untreated GSD1 include; osteoporosis, delayed puberty, gout (arthritis caused by accumulation of uric acid), kidney disease, pulmonary hypertension (high blood pressure in the arteries that supply the lungs), hepatic adenoma (benign liver tumors), polycystic ovaries in females, an inflammation of the pancreas (pancreatitis), diarrhea, and changes in brain function due to repeated episodes of hypoglycemia. Impaired platelet function can lead to a bleeding tendency with frequent nose bleeds (epistaxis).

Early diagnosis and effective treatment can result in normal growth and puberty and many affected individuals live into adulthood and enjoy normal life activities. Many female patients have had successful pregnancies and childbirth.

Type I glycogen storage disease is inherited as an autosomal recessive genetic disorder. The risk for two carrier parents to both pass the non-working gene and, therefore, have an affected child is 25% with each pregnancy.

GSDI is treated with a special diet in order to maintain normal glucose levels, prevent hypoglycemia and maximize growth and development. Frequent small servings of carbohydrates must be maintained during the day and night throughout life. Calcium, vitamin D and iron supplements may be recommended to avoid deficits. Frequent feedings of uncooked cornstarch are used to maintain and improve blood levels of glucose. Allopurinol, a drug capable of reducing the level of uric acid in the blood, may be useful to control the symptoms of gout-like arthritis during the adolescent years. Medications may be prescribed to lower lipid levels and prevent and/or treat kidney disease. Liver tumors (adenomas) can be treated with minor surgery or a procedure in which adenomas are ablated using heat and current (radiofrequency ablation). Kidney and/or liver transplantation are sometimes considered if other therapies are unsuccessful or where liver adenomas keep growing.



References: https://rarediseases.org/rare-diseases/glycogen-storage-disease-type-i/

Authors: Deeksha Bali, PhD & Yuan-Tsong Chen, MD, PhD

Suzanne's Corner

Angelo's Favourite Red Cabbage Salad

The entire family can enjoy this colourful and tasty side dish. It is "Angelo Approved".

Entire recipe

Protein (grams)	Fat (grams)	Carbohydrate (grams)	Calories (kcal)
4.0	40	15.7	445

Makes 2-4 servings

Salad

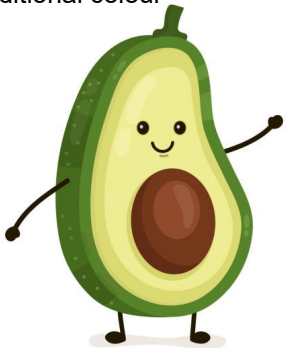
Amount	Ingredient
1 cup	Red cabbage, shredded
½ cup	Carrots, grated
1 tablespoon	Green onions, chopped
1 tablespoon	Pomegranate seeds
½ cup	Avocado, cubed
1 tablespoon	Parsley, chopped
¼ cup	Red pepper, slices

Dressing

Amount	Ingredient
½ teaspoon	Salt
½ teaspoon	Pepper
2 tablespoons	Extra virgin olive oil
1	Lemon, squeezed for juice

Tips

- To reduce total protein, consider reducing or eliminating the avocado. Avocado provides 1.6 grams protein. Subtract the amount from the total protein per recipe
- To reduce the total fat, try reducing the amount of avocado and/or olive oil used. Avocado provides 11.6 grams fat and olive oil 27.4 g fat. Remove from the total fat per recipe
- Those who must follow a very restricted long chain fat diet, remove avocado and olive oil. Try adding a few mL of MCT oil
- Garnish with green beans to add additional colour



Avery's and Damien's Favourites by Chef Dad

Thank you to Nora and Jon for providing these delicious recipes! The entire family will enjoy these recipes!

Oyster Mushroom Chowder

For the entire recipe which weighs 1820 g:
How Much Phe app
Phenylalanine 743 mg
Protein 22 g
Fat 56.8 g (Note: butter 40.6 g, So Delicious coconut creamer 12 g. Rest of the recipe 4.2 g fat. For those on a very low long chain fat diet, consider replacing the creamer with rice milk and reducing butter)

Step 1- *Mushrooms*

Amount	Ingredients
100 grams	Oyster Mushrooms, chopped
25 grams	Butter (used Earth Balance)
55 grams	White wine
1 gram	Celery seed

Chop mushrooms and sauté in butter
Once mushrooms are browned, add wine and celery seeds
Cool for a minute
Set aside

Step 2- *Cauliflower cream base*

Amount	Ingredients
220 grams	Cauliflower, chopped
25 grams	Butter (used Earth Balance)
¾ cup	So Delicious Coconut creamer

Steam cauliflower until soft
Place cauliflower, butter and coconut creamer in a blender (or may use hand held mixer)
Mix until creamy

Step 3- *Soup Base*

Amount	Ingredients
147 grams	White onion, chopped
147 grams	Celery, chopped
170 grams	Carrots, chopped
316 grams	Potatoes, chopped into small cubes
50 grams	Frozen corn
2 grams	Dried thyme
32 grams	Cornstarch
938 grams	Vegetable broth
118 grams	Rice milk
To taste	Salt and Pepper

Sauté onions, celery and carrots until soft and starting to brown
Add potatoes and toss
Add thyme and cornstarch and mix to coat
Add broth and rice milk
Bring to boil
Add cauliflower cream and mushrooms
Simmer on low and then serve

****Weigh serving and calculate protein or fat content of serving based on information for the entire recipe****

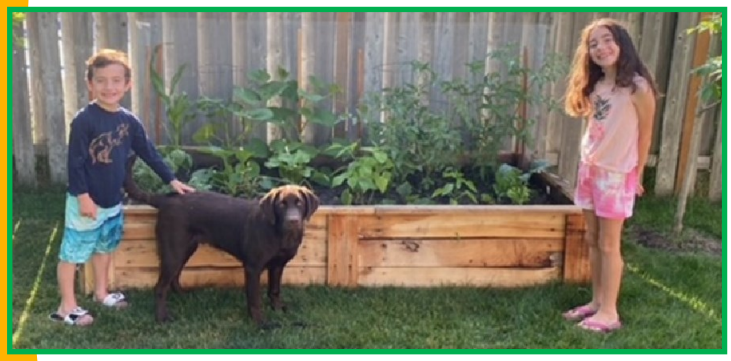
Jackfruit and Pumpkin Indian Spice Chili

For the entire recipe which weighs 967 grams:
How Much Phe app
Phenylalanine 436 mg
Protein 12.8 g
Fat 2.8 g

Amount	Ingredients
190 grams	Onion
15 grams	Garlic
227 grams	Jackfruit in brine, drained
420 grams	Canned tomatoes, diced or crushed
125 grams	Canned pumpkin puree
215 grams	Root beer, regular (not diet)
3 grams	Chili powder
2 grams	Garam masala
1 gram	Turmeric
1 gram	Cumin
To taste	Salt and Pepper

Sauté onions until soft
Add garlic for another 3 minutes or longer
Shred and rinse jackfruit
Add jackfruit to onions and garlic
Add spices to mix and sauté for another 5-10 minutes
Add tomatoes, pumpkin, root beer and stir
Bring to a simmer for 20 minutes, then ready to serve

****Weigh serving and calculate protein or fat content of serving based on information for the entire recipe****



Damien and Avery, PKU along with Jack by their vegetable garden

What's New

Check out the new Congenital Disorders of Glycosylation (CDG) Canada website at http://www.canadacdg.com for information on studies, events, resources, and to sign up for their newsletter!

Metabolic Family Workshop

Unfortunately as the pandemic persists, we will not be able to host the Metabolic Family Workshop this year. As soon as it is safe to do so, we look forward to welcoming everyone back for this exciting event. Stay tuned to future issues of the newsletter for the latest updates.

Students

Congratulations to Tristan (right) and Prashanth (left)! Tristan will begin her Master's of Genetic Counselling degree at University of Toronto in the fall and Prashanth will begin his medical school training at Queen's University.

Sam

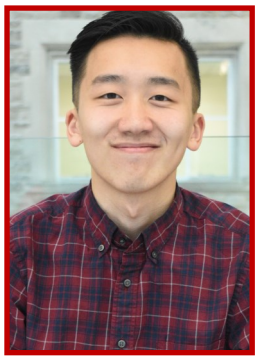
Project title: Late-Onset Ornithine Transcarbamylase Deficiency: Biochemical and Clinical Consequences of a Rare Regulatory Region Mutation

My project focuses on highlighting the biochemical and clinical consequences of a relatively unknown variant in an upstream regulatory region of the ornithine transcarbamylase (OTC) gene. The aim is to summarize various biochemical measurements and clinical symptoms to inform the scientific community about this particular, relatively unknown variant.

Derek

Project title: Culturally sensitive provision of rapid biochemical and molecular diagnosis at birth in children at risk for metabolic disorders in Southwest Ontario.

Children with severe metabolic disorders often present before the results of newborn screening are available and typically experience complicated and lengthy hospitalizations. Families who have already had a child with a severe metabolic disorder may opt for prenatal diagnosis for identification of subsequently affected offspring, but this may not be desired for religious or other reasons. Therefore, the aim of this project is to implement rapid biochemical and molecular testing at birth for families who have chosen not to have prenatal diagnosis and to evaluate its impact on the prognosis and outcome of patients with severe metabolic disorders.



Steven

Project title: Phenotypic variability and impact of newborn screening on patients with severe Biotinidase deficiency Y210C in Southwestern Ontario

The goal of the project is to characterize the phenotypic variability of patients with severe Biotinidase deficiency and to compare patients who were diagnosed early versus those who were diagnosed later on. This comparison will help guide an evaluation of the impact of newborn screening on prognosis and outcomes for Biotinidase deficiency.

Shyann

Project title: Long term follow up of neurotransmitter disorders in the neurometabolic clinic at London Health Sciences Center: Approach to diagnosis, management and neurological follow up (2004 -2021)

Over the last 17 years the neurometabolic clinic at London Health Sciences Center has followed patients with neurotransmitter disorders such as 6-Pyruvoyltetrahydropterin Synthase (6 PTPS) deficiency, Succinic Semialdehyde Dehydrogenase (SSADH) deficiency, Aldehyde dehydrogenase 7 A1 (ALDH7A1) deficiency, Non ketotic hyperglycinemia, and Methylenetetrahydrofolate Reductase (MTHFR) deficiency. The aim of this research project will be to examine how these conditions present in clinical practice, investigative tools utilized in the diagnosis of these disorders, as well as the outcomes of the selected conditions in response to early diagnosis and treatment.

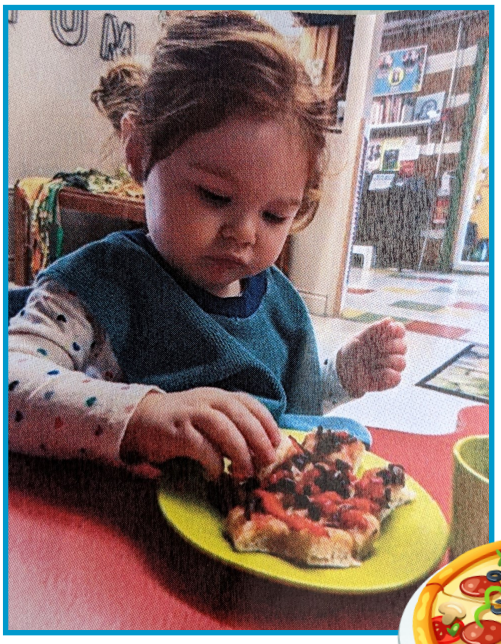
Photo Gallery

Kennady, MMA

Helen, LCHAD deficiency

Angelo, PKU

Martin, Hypophosphatasia, with baby sister Susana

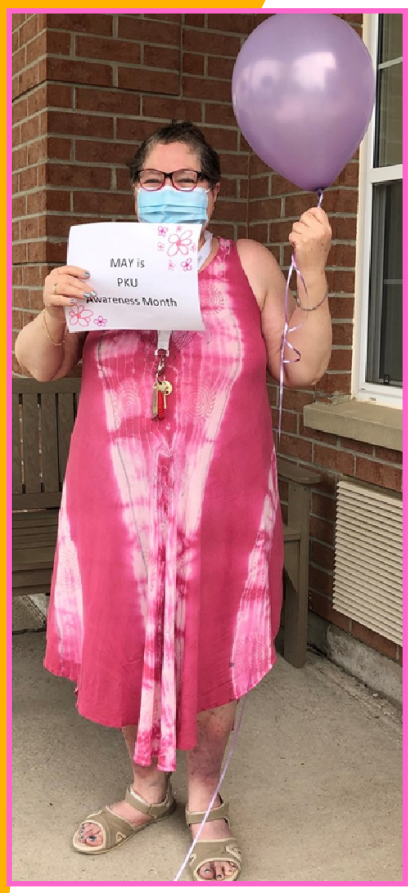


Daphne, PKU

Karson, PKU

Adina, citrin deficiency

Anik and new brother Laekh, carbonic anhydrase VA deficiency, with mom and dad



Carol, PKU

Jack, PKU

Emily, PKU

Emily, PKU

Kaitlynn, all dressed up for PKU awareness month (May)!

To make delicious sugar cookies like Kaitlynn, check out this link: http://www.medicalfood.com/Recipes/Desserts--Snacks/Low-Protein-Holiday-Sugar-Cookies/#.YLLH7fc4aaM

Margie, PKU

Inspirations

Self-Compassion
Dr. Kristin Neff
https://self-compassion.org/

Self-compassion asks us to respond to our own suffering, failures, and imperfections in a kind, loving, and non-judgmental way. It is understanding that all of these things are a part of our human experience and that we must honor and accept our humanness. Sometimes it is easier to be compassionate towards others than it is to ourselves. However, just as others are worthy of love, kindness, understanding, and forgiveness, it is important to show that same acceptance and compassion to ourselves too.

Three Elements of Self-Compassion

Self-kindness vs. Self-judgement
Be warm and understanding towards yourself when you suffer or fail. Imperfection, failing, and hardships are inevitable so it is important to accept them as they come and to be patient and kind towards yourself as you work through them.

"If your compassion does not include yourself, it is incomplete."— Buddha

Common Humanity vs. Isolation
When things don't go your way, it is easy to feel isolated, as if you are the only one experiencing suffering, Recognize that suffering and personal inadequacies are part of our shared human experience and not something that happens to you alone.

"What we don't need in the midst of struggle is shame for being human."— Brené Brown

Mindfulness vs. Over-identification
It is important to take a balanced approach to negative emotions so they are not suppressed or overexaggerated. In relating our personal experiences to others, we are able to see a bigger perspective and gain a better understanding of our own hardships. Mindfulness is a non-judgmental mind state and involves simply observing our emotions and experiences, instead of ignoring them, judging them, or trying to change them.

**"Mindfulness is the aware, balanced acceptance of the present experience."
— Sylvia Boorstein**

Our Talented Artists

Abigail, hyperphenylalaninemia

Annie, MSUD

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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