The Inherited Metabolic Disorders News

Children's Hospital
London Health Sciences Centre

Summer 2020 Volume 17 Issue 2

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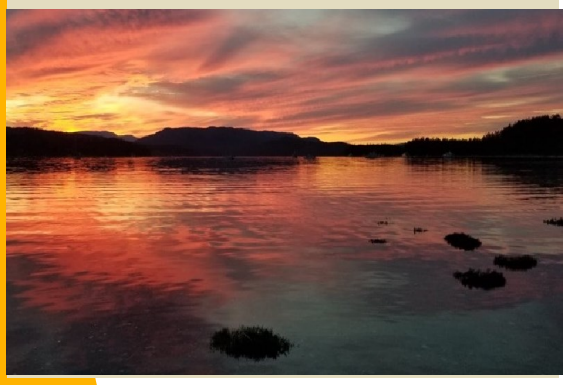
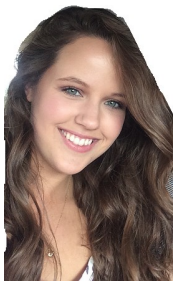
In this Issue



From the Editor



Hey there everyone! What a whirlwind these last few months have been! I hope you all have managed to take care of yourselves and your families, and have begun to adjust to these new times. Dr. Prasad and I are really excited to share with you some thoughts and resources we have both personally turned to in these uncertain times. As always, please send along any photos and contributions to meghan.zadorsky@lhsc.on.ca. Take care everyone!

Meghan Zadorsky

From Dr Chitra Prasad

Dear Friends,
Greetings

I am hoping you all are safe in the COVID-19 times. These are truly unprecedented times. The whole world has come to a standstill because of a tiny virus. As always, I try and think about the messages we have learnt during this period. For me it has been a feeling of gratitude for all the good things in life. In Canada we are blessed with following "discipline" and" guidelines" which is not so easy in some of the other countries. I would encourage each one of you to keep a list of all the lessons that you and your family may have learnt during these unique times. Children look up to parents and can view this time as a time of adventure, family time and togetherness. In this issue of our newsletter Meghan Zadorsky (our genetics assistant) and I have compiled some resources on happiness, gratitude, and mental and emotional health during COVID times. I have myself read these books/blogs and find their messages very timely.

Our Dr. Andrea Yu will be leaving us for her home-town of Ottawa in September. I know many of you have met her. She is a wonderful metabolic physician and has done a great job even in the short time that she has been here. I am going to miss her a lot! I do wish her all the best as she will be closer to her family in Ottawa. I invite all of you to read one of her patient's family's story that they have shared about their experiences with citrin deficiency, in this issue of the newsletter.

These COVID times have changed a lot of ways in which we have been practicing patient care. My colleagues and I have been doing virtual telehealth patient visits with some of you. The experience is novel but it also makes us aware of this technology that can be used for some of the follow up visits in the future.

Hope you all can enjoy the summer, beautiful walks and the lovely greenery each day.

Please take good care of yourselves and your families. Practice social distancing, hand cleaning and also remain connected with your loved ones through new technologies!!!!
With best wishes
Your friend,

Chitra Prasad

"Hope is important because it can make the present moment less difficult to bear. If we believe that tomorrow will be better, we can bear a hardship today." —Thich Nhat Hanh

Personal Stories

Adina

We noticed Adina began to look a little yellow when she was around 4 weeks old. She had been colicky and produced more dirty diapers than I could count in a day which I thought was normal for a newborn. However, as a first-time mom, I didn't know that a baby could become jaundiced weeks after birth and because her skin colour is a little tanned, the yellow tinge was difficult to notice and I was constantly second-guessing my eye-sight. However, when the whites of her eyes began to look murky, we took her into urgent care where we discovered she was jaundiced and we had to take her into pediatric emergency at the London Children's Hospital. Despite her elevated bilirubin levels, Adina was breast-feeding well and slowly but surely, was gaining weight as well as behaving like a normal new born – she just looked a little yellow.

After admittedly doing some research on Google while waiting for a pediatrician in the emergency room, we thought it was just going to be a normal case of jaundice and that with proper care and nutrition, it would go away. To our surprise, that was not the case. After more blood work and plenty of heel pokes, the nurses and the pediatrician on-call told us they weren't too sure what was causing the spike in her bilirubin levels and booked us an appointment with a liver specialist the following morning.

The next morning, we went for Adina's appointment with Doctor Bax who expressed his concern of a possible liver disease called biliary atresia. If she had this disease, there was a small window to help correct the disease, albeit temporarily, with a surgery, so Adina was admitted almost immediately to do more testing. After being admitted, Doctor Bax connected us with another pediatric liver specialist named Doctor Zizzo who wanted to rule out other liver disorders that may be causing a spike in her bilirubin levels while we waited for a biopsy to be done to know if she had biliary atresia. Thankfully, all other liver disorders were ruled out and the biopsy showed her liver working fine however slightly irritated; but this only brought us further from a diagnosis. During this time, the doctors, nurses and cleaning staff made us feel at home and definitely helped us feel less overwhelmed in seeing our daughter being hooked up to different monitors and wires and going through multiple heel pokes and blood work.

After the liver biopsy revealed nothing out of the ordinary, Doctor Zizzo prescribed multiple medications that would help Adina's liver while we would visit each week for more blood work and check-ups. Eventually, Doctor Zizzo ordered a DNA test to be done and connected us with Doctor Yu, a geneticist who played a big role in Adina's diagnosis. A few weeks after Adina submitted blood work for DNA testing, Doctor Zizzo called us with a potential diagnosis that Doctor Yu discovered. At this point, Adina was nearing 5 months old. At our check-up with Doctor Zizzo and Doctor Yu, we were told that Adina very likely has a metabolic disease called citrullinemia type 2 or in Adina's specific case, citrin deficiency, caused by an autosomal recessive gene that me and her father both have. This type of disease means her body prefers lipid and protein rich foods over high carb or lactose and galactose rich diets. Lactose, galactose and simple carbohydrates cause her bilirubin levels to spike and if left undiagnosed, could potentially lead to dangerous consequences. Individuals with her diagnosis normally gain weight slowly which Adina was, but not dangerously slow. Doctor Yu asked how I felt about eliminating breastmilk from Adina's diet and instead putting her on a soy formula which I gladly accepted – anything to help Adina thrive. We decided to come back for bloodwork after putting Adina on soy formula to see if that would make a difference in her bilirubin levels and miraculously, her levels decreased significantly to within a normal range. A few months later, the DNA test confirmed the citrin deficiency diagnosis. We went back for a check-up and Adina had already gained a significant amount of weight and her blood work came back healthy and normal. We are extremely blessed in that Adina never presented as sick or lethargic, even though at first, she was gaining weight slowly and was cranky – she was still hitting all her milestones. But after switching her to soy milk, she became a different baby. She slept better, played more, didn't produce as many dirty diapers and started gaining weight rapidly.

Adina was at the age to start solids, so we were connected with Suzanne Ratko, a dietician who helps us with Adina's everyday diet as she gets older. Before the pandemic, she started daycare and the food menu honestly overwhelmed me because I couldn't be the one handling her food. However, the daycare gave me the menu, and any questions I'd have about food I'd email Suzanne about and man, we must have emailed her over a dozen times with food-related questions just in the days leading up to Adina starting daycare. Suzanne was a huge help in figuring out her diet and which foods to substitute or replace. Right now, Adina is almost 18 months and loves her chicken and avocado and is as happy and healthy as ever. She's very active and does not know how to sit still. Like most babies, she gets into absolutely everything within her reach. I am very thankful that Adina has a great team at the London Children's Hospital who continue to follow up on Adina frequently and I still reach out to all her doctors with any questions I have.

Adina Ali Ismail born March 10, 2019.
First Hospital stay from approx. May 1st – May 14th 2019.
Second Hospital Stay from approx. June 10-13th 2019.
Official confirmed diagnosis in November 2019.

Featured This Issue

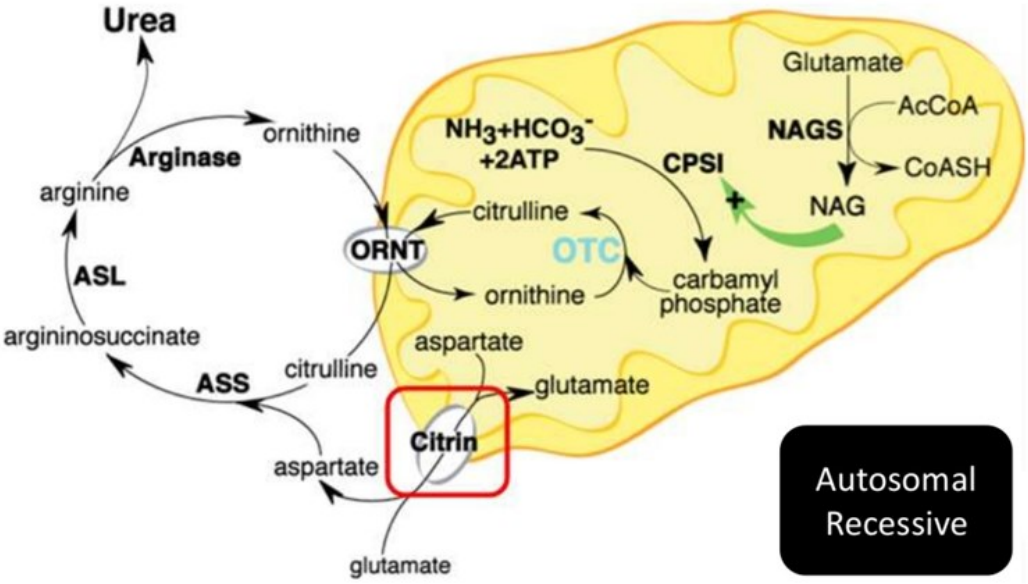
Citrin Deficiency

Compiled by Dr. Andrea Yu

Citrin deficiency is a rare inborn error of metabolism which can present in several different ways depending on how old a person is when they first have symptoms. Citrin is an important protein found in the liver, kidneys and heart whose purpose is to be an aspartate-glutamate carrier. Children who are less than 1 years old can present with neonatal intrahepatic cholestasis caused by citrin deficiency (NICCD). They often have a history of growth restriction and liver problems. They may have difficulty breaking down galactose, which is a sugar found in breast milk and cow's milk-based baby formulas. Children older than 1 year can present with failure to thrive and dyslipidemia caused by citrin deficiency (FTTDCD). Many children develop a protein-rich or lipid-rich food preference and an aversion to carbohydrate-rich foods. If a person does not present until they are an adult, then they could present with recurrent hyperammonemic episodes causing neuropsychiatric symptoms and the condition is called citrullinemia type 2.

Citrin deficiency is inherited in an autosomal recessive manner and occurs when a person has two disease-causing (ie. Pathogenic) mutations in the gene *SLC25A13.* Citrin deficiency was first described in the Japanese population but it is now known that there are affected individuals in many different ethnic groups.

Treatment for citrin deficiency can include changing the diet to reduce lactose and supplementing with medium-chain triglycerides. In children with NICCD, this may be enough to resolve all their symptoms. Adults with citrullinemia type 2 can be more difficult to treat and some severe cases may require a liver transplantation.



Source: https://www.slideshare.net/childrenliverindia/citrin-deficiency

References:

GeneReviews: Citrin Deficiency (https://www.ncbi.nlm.nih.gov/books/NBK1181/)

Current treatment for citrin deficiency during NICCD and adaptation/compensation stages: Strategy to prevent CTLN2 (https://www.sciencedirect.com/science/article/pii/S1096719219303270)

Suzanne's Corner

0% Fat Banana Bread Recipe by *Helen*

Ingredients:

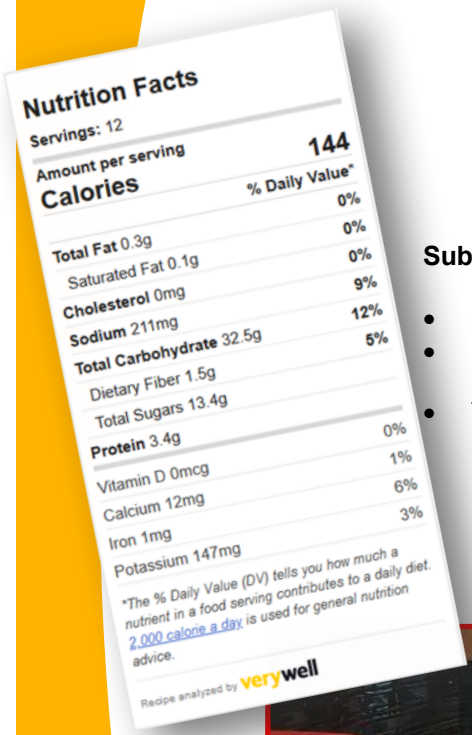
- 1/2 cup unsweetened apple sauce (replaces 1/2 cup butter)
- 1/2 to 1 cup granulated sugar (your preference, I used 1/2 cup)
- 1/3 cup egg whites (replaces 2 whole eggs)
- 1/4 cup skim 0.1% milk
- 2 cups all-purpose flour
- 1 tsp baking soda
- 1/2 tsp salt
- 1 tsp vanilla extract
- 3 ripe bananas
- Ungreased bread pan (I used 11x5) – parchment paper optional

Directions:

1. Preheat oven to 350°F
2. Cream together apple sauce and sugar (adding sugar in gradually)
3. Mix in egg whites
4. Mix in skim milk and vanilla
5. In a separate bowl, combine sifted flour + baking soda + salt. Hand whisk gently together to mix.
6. Add the combined dry ingredients to the wet mixture above
7. Once nicely combined, add in the mashed bananas
8. Bake in preheated oven for 50-55min (unless bananas were used as butter substitute, then 38-41min baking time)

Substitutions:

- Use apple sauce instead of butter. Use in equal amounts.
- Bananas also make a great butter substitute. Use in equal amounts. Reduces baking time by up to 25%!!
- There are a number of whole egg substitutes that you can use:
  - 1/4 cup plain yogurt (low fat) = 1 whole egg
  - 1/4 cup carbonated water = 1 whole egg
  - 1/3 cup (85ml) egg whites = 2 whole eggs (as per guide on egg white carton – Kirkland Signatures)



Nutrition Facts

Servings: 12

Amount per serving	
Calories	144
	% Daily Value*
Total Fat 0.3g	0%
Saturated Fat 0.1g	0%
Cholesterol 0mg	0%
Sodium 211mg	9%
Total Carbohydrate 32.5g	12%
Dietary Fiber 1.5g	5%
Total Sugars 13.4g	
Protein 3.4g	
Vitamin D 0mcg	0%
Calcium 12mg	1%
Iron 1mg	6%
Potassium 147mg	3%

*The % Daily Value (DV) tells you how much a nutrient in a food serving contributes to a daily diet. 2,000 calorie a day is used for general nutrition advice.

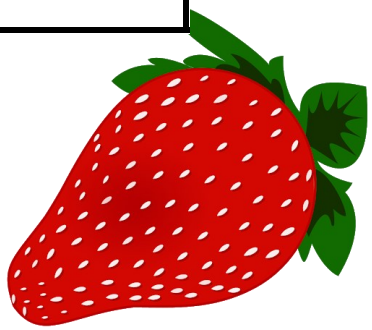
Recipe analyzed by verywell

Strawberry Sorbet: A Summer Suggestion from Kaitlynn

https://www.asweetpeachef.com/strawberry-sorbet/

Makes 6 servings

Prep time	60 minutes
Cook time	0 minutes
Processing	5 minutes
Total time to make	1 hour 5 minutes



Ingredients:

- 3 cups fresh or frozen strawberries
- 2 tablespoons honey (as needed for sweetness)
- 1/4 cup warm water

Directions:

1. If strawberries are not frozen, freeze the strawberries until completely solid. It may take 1-2 hours or leave them in the freezer overnight.
2. Place frozen strawberries into the bowl of a food processor or heavy-duty blender
3. Blend until smooth
4. Add warm water and press with a spatula, as needed
5. Taste when smooth and add honey as needed for sweetness
6. Eat immediately for a softer texture or transfer into a freezer-safe container and freeze for 1-2 hours until firm

Nutrition*:* Amount per 0.5 cup serving

Calories	**68 kcal**
Fat	**1 g**
Carbohydrate	**17 g**
Fibre	**2 g**
Sugar	**14 g**
Protein	**1 g**

Kaitlynn and family celebrating PKU awareness month (May)

Making changes during the pandemic: dried blood collection for PKU patients

Emily

Kaitlynn

Emmitt

For our patients with PKU, part of their management requires them to have regular blood work. Many would go to their local hospital or lab to have their blood drawn and then the blood would be sent to London for analysis in Dr. Rupar's lab. However, early on during the pandemic visits to lab test centres were not always possible or preferred for families. Because blood work monitoring was still necessary, a majority of our patients switched to doing dried blood spot collection from home. They would poke their finger and collect blood on a Spot Blood Card made of filter paper. The card would be mailed to Ottawa for analysis because our lab was not set up to do filter paper analysis. As the pandemic continued, Dr. Rupar set up his lab to do the analysis. And as an added bonus, the lab would be able to analyze samples from patients with Maple Syrup Urine Disease (MSUD) which Ottawa could not do. Hooray! There have been tears, frustration, victories and bravery by our patients and their families. Here are the faces of some of our heroes!

Emily

Stathy

Jack

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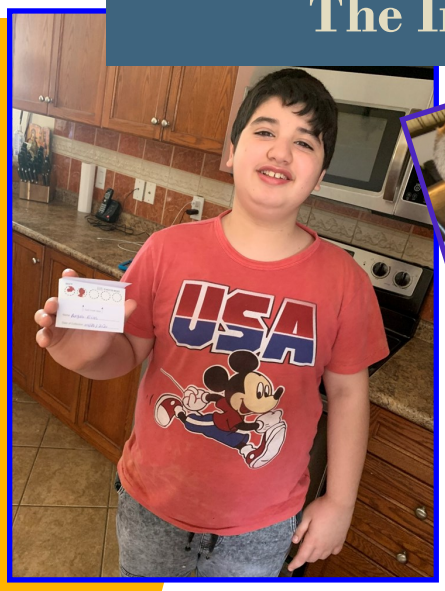
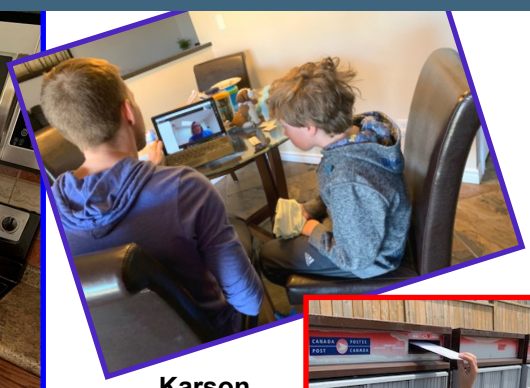


Angelo



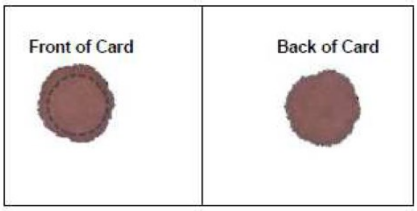
Karson

Abbas

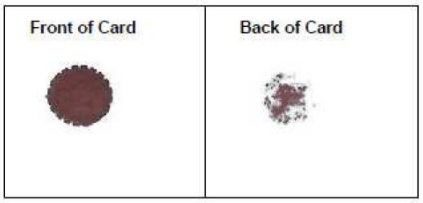
Margie

There have been hiccups along the way. Lancets did not always work. The mail: one sample sent by express post took 23 days to travel from Sault Ste Marie to Ottawa... Poor samples were sent.
To the right are picture samples from a Spot Blood Card showing a satisfactory and unsatisfactory sample. Too little or too much blood may lead to an inaccurate result.

Satisfactory Sample



Unsatisfactory Sample



Noah

Kasper AKA
Iron Man

Postponed!

16th Annual Low Protein Cooking Demonstration

Unfortunately due to the COVID-19 pandemic, the low protein cooking demonstration that was set to be held on July 18, 2020 will be postponed until 2021. Everyone's health and safety is our number one priority so we look forward to seeing you all at this event next year when it is safe to do so! Details will be shared in the upcoming issues of the newsletter.

Metabolic Family Workshop Coming May 14, 2021!

We will continue to monitor the COVID-19 situation and will make changes to the event as necessary. Keep an eye out for updates in the upcoming issues of The Inherited Metabolic Disorders News.

What's New

From Dr. Andrea Yu

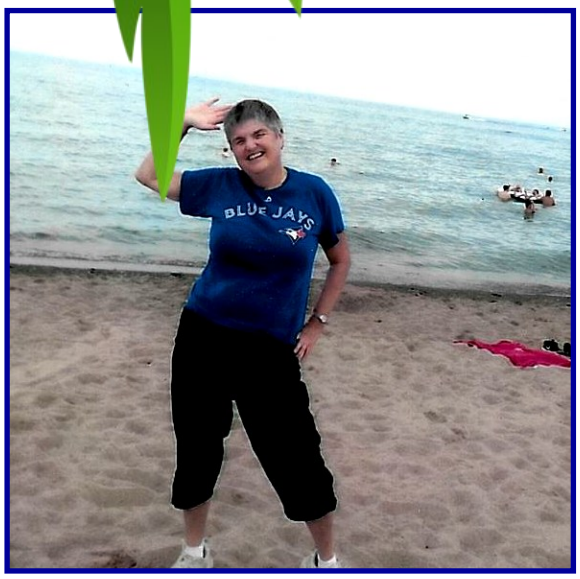
I will be leaving London this fall to return to Ottawa and start a new practice as a Metabolic Physician at CHEO (Children's Hospital of Eastern Ontario). It has been a privilege to serve as a physician to so many of you during my time in London. You have welcomed me and allowed me to be an important part of your families' lives and I have been honoured by the trust that you have shown by sharing your struggles and successes with me. Although I am excited to return to my home town and be with my family and friends again, I know that I will miss London dearly and will always remember my time here. I look forward to following you and your families' milestones and achievements in our Metabolic Newsletters. Best wishes to all of you!

Our Stars

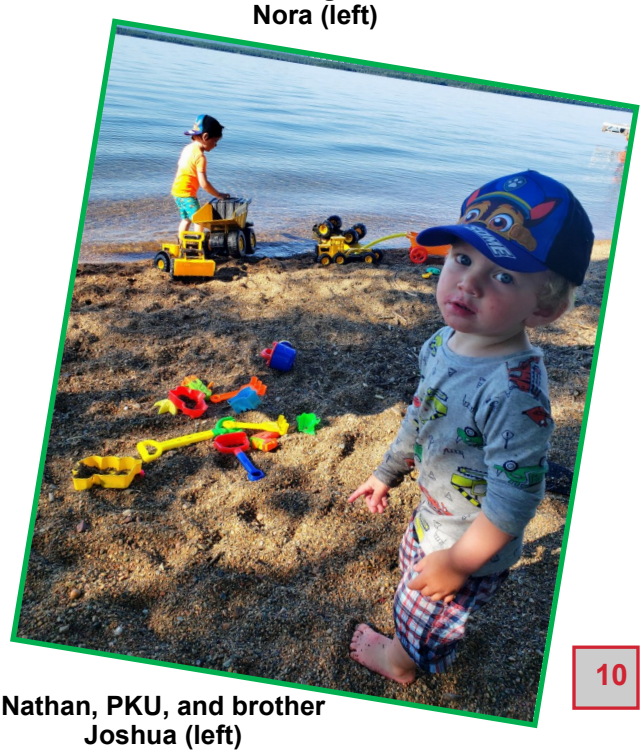
Anik, carbonic anhydrase VA deficiency

Quorra, abetalipoproteinemia, loves helping out in the kitchen!

Leonidas and Eleandra, MCADD, along with sister Nora (left)



Margie, PKU



Nathan, PKU, and brother Joshua (left)

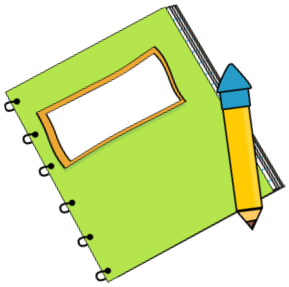
Inspirations

Five Small Changes to Increase Happiness From Shawn Achor's TED Talk: "The Happy Secret to Better Work"

Shawn Achor is a researcher who's work focuses on happiness and positive psychology. Positive psychology is the study of what allows individuals to be resilient and to thrive. Within positive psychology is the idea that the lens with which you view the world shapes your reality. In other words, your mind-set and approach to life has more influence over the way you experience things in life than what is actually happening in your life. This finding is great news because it means we can actively work to change our lens to a more positive one, which can increase happiness and in turn, increase success and productivity. Below are five small changes Shawn shares in his TED Talk that we can try to incorporate in our daily lives to help us rewire our brains and shift our lens to a more positive one to be able to capitalize on The Happiness Advantage– the positive outcomes of living a happier life.

Check out Shawn's TED Talk here:
https://www.ted.com/talks/shawn_achor_the_happy_secret_to_better_work

Gratitude– write down three good things each day and your brain will start to look for positives in the world instead of negatives



Journaling– if you journal about one positive thing each day, your brain gets to relive it

Exercise– teaches the brain that behaviour matters

Meditation– allows the brain to slow down and helps us focus on the task at hand

Random Acts of Kindness– allows you to identify positives in others and spread happiness

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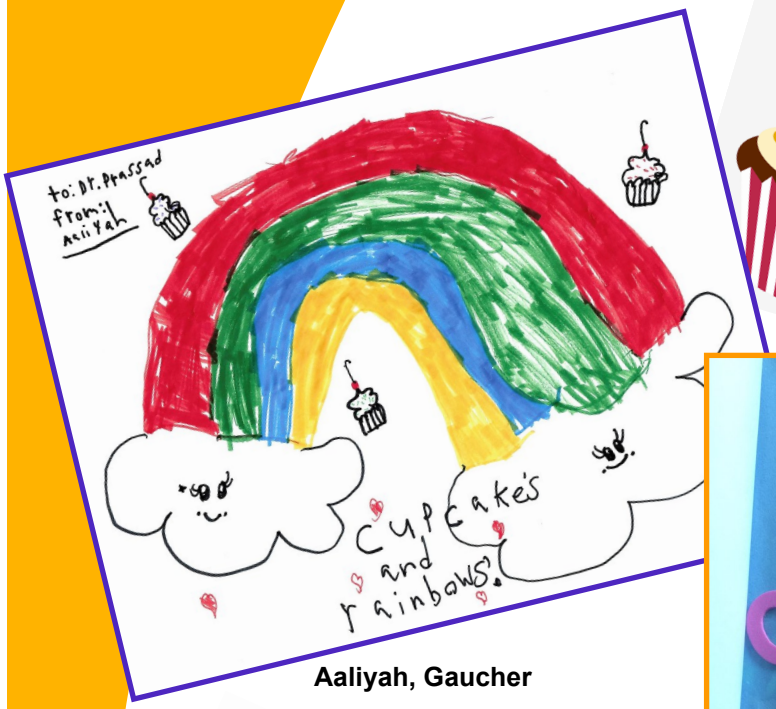
Our Talented Artists



Aaliyah, Gaucher

Noah, PKU

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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!