The Inherited Metabolic Disorders News

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Children's Hospital
London Health Sciences Centre

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From the Editor

Hello everyone! I am happy to introduce myself as the new editor of the newsletter! I would like to thank my colleague and office mate, Maria Bata, for her contributions to the newsletter as our previous editor. It is a pleasure to be involved in this project, it is truly special to our metabolic genetics team and we hope to you as well. I look forward to hearing your feedback. Best wishes! ***Suzanne Nurse***

From Dr Chitra Prasad

Dear Friends,

Greetings. I hope you and your families are doing well. I am pleased to let you know that our metabolic team now has a number of new members. Please check out their details in the newsletter.

In October I was invited to participate as faculty for National American Metabolic Academy by my mentor Dr. Mark Korson. It was a truly wonderful experience. I got to meet many metabolic specialists and trainees from all over the world. I was also honored to meet Dr. Saudubray father of modern metabolic medicine.

The digital technology has truly impacted the whole world. Many of our lectures and education sessions are being held over zoom or Microsoft teams. This has many advantages as we can access and share information over these platforms without having actually to travel. These technologies have also allowed the virtual appointments that we are doing with some of our patients and families.

In this newsletter we share a story by Sonia Rusin-Franke, a very bright sixteen-year-old individual in their own words. Sonia was diagnosed as having Wilson disease after their many encounters with different specialists. This is a rare condition and I'm happy to share that Sonia is doing very well after starting treatment for this condition. Sonia and their parents continue to amaze me and all the other specialists with their depth of knowledge and understanding about the disease.

Human spirit remains resilient in the face of pandemics. Even as the human race struggles with Covid-19 and other infections, we continue to hope for a healthier and better future.

I wish you and your families the very best in 2023.

Your friend
Chitra Prasad

Dr. Chitra Prasad & Dr. Jean-Marie M. Saudubray at the 2022 North American Metabolic Academy

***"Hope is being able to see that there is light despite all of the darkness."* – Desmond Tutu**

Personal Story: Sonia Rusin-Franke

Hi! My name is Sonia, I'm 16 years old, and I live in Stratford, Ontario. I'm agender, which means that I have no gender identity. I use both they/them and he/him pronouns. I like frogs, the colour blue, Shakespearean tragedies, ancient history, and orange soda. I'm an INTJ, a Gemini, and a Ravenclaw. I'm a huge musical theatre nerd—my favourite musicals are *Hamilton* and *Newsies*, and I have a long list of musicals I've not yet seen that I believe I should, which I'm slowly working my way through. I also have Wilson disease.

Wilson disease is a genetic metabolic disorder that means I cannot metabolize copper. The problem is that there is at least a trace amount of copper in all foods. So, the copper from all the food I had eaten for the 15 years of my life before my diagnosis was building up in my brain and corneas. This was unusual in that the most buildup of copper in a Wilson disease patient is typically in the liver, as that is where copper metabolism takes place, for the most part. However, there was almost no damage to my liver, and my symptoms were entirely neurological. Excepting, of course, the tell-tale mark of Wilson disease — Kayser-Fleischer rings. These are rings of copper deposit around the iris, usually only detectable with a slit lamp eye exam, however mine were visible to the naked eye, and obscured the colour of my eyes.

I began exhibiting the first signs of Wilson disease in 2017, when I developed mild sialorrhea (excessive drooling). It was barely noticed by both me and others over the next couple of years, until it worsened noticeably along with the appearance of other neurological symptoms. In the summer of 2019, my parents noticed that my speech had become slurred and hard to understand, and my handwriting small and illegible. Additionally, I had noticed my hands and fingers would shake sometimes, and I had very limited fine motor skills. The most noticeable symptoms were the speech impairment and the sialorrhea, so my parents made an appointment with a speech therapist, who referred me to a myofunctional therapist. I attended monthly appointments from February 2020 to March 2021, and succeeded in ameliorating these problems, however they did not go away.

In November 2020, I attended an appointment at the London Health Sciences Centre to have a neurological exam performed. Around this time, I also developed psychiatric symptoms of depression, anxiety, irritability, and lethargy. The results of the neurological exam raised concerns, and an MRI was scheduled for March 2021. The MRI results were very concerning. The doctors suggested that I be admitted to hospital to undergo additional tests to determine a diagnosis.

Continued on the following page

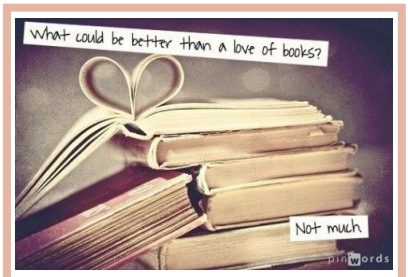
Personal Story - continued

At the time, the suspected condition was a mitochondrial disorder. However, this hypothesis was eliminated after an ophthalmologist at St. Joseph's Hospital, Dr. Sharan, conducted a slit lamp eye exam and determined the presence of Kayser-Fleischer rings in my eyes. From that point on, Wilson disease was the suspected diagnosis, however more tests were conducted to be sure.

During the week, I was subjected to a variety of tests, including neurological examinations, bloodwork, a 24-hour urine test, an abdominal ultrasound, an echocardiogram, an electrocardiogram, a kidney ultrasound, and an audiology exam. It was determined that my heart, kidneys, and liver were perfectly healthy, which is unusual in Wilson disease. The liver is typically the most affected. Despite this, the tests also confirmed my diagnosis of Wilson disease. I was prescribed Trientine to chelate the copper already in my body, and adopted a low-copper diet. Recently, my medication was switched from Trientine to zinc tablets, as the chelation was finished. The zinc tablets prevent copper from being absorbed into my organs and bloodstream. I tried Atropine drops under my tongue to mitigate the sialorrhea, and it was very effective, however it also dried out my mouth and throat so that I had to almost constantly be drinking water, and it was difficult to swallow. I instead received Botox injections into my salivary glands, which has been working very well so far. I continue to attend regular appointments with various doctors to maintain my health and monitor my progress.

While most people might expect such a diagnosis to be upsetting or life-altering, I did not find it to be so in the least. It was quite relieving, in fact, to be able to have an explanation for the steadily increasing difficulty of living normally. The only aspects in which my life is altered in any significant way is with my eating habits, due to the low-copper diet; prohibiting many foods I previously consumed regularly, such as chocolate, nuts, potatoes, kale, and avocado; and my medication, which must be taken between meals with a couple hours in between, preventing me from eating smaller meals with a sprinkling of snacks in between, which had been my preference. However, such difficulties do little to impact my overall quality of life, and I feel as though the benefits of my diagnosis and treatment greatly outweigh the disadvantages it causes.

My plans for the future include studying history at Wilfrid Laurier University, seeing *Hamilton* performed live, writing a novel, and making the most of life! I'm looking forward with an optimistic mindset and holding onto many ambitions in the hopes that at least one of them will carry me far.



I love to read—I'm currently reading *Six of Crows* by Leigh Bardugo—and listen to music—my current favourite song is "Dreams" by The Cranberries.

I also enjoy watching TV—my current favourite shows are *The Owl House, Anne with an E*, and *Stranger Things*.

Featured This Issue

Wilson Disease

Compiled by Dr. Chitra Prasad

Wilson disease is a rare genetic disorder characterized by excess copper stored in various body tissues, particularly the liver, brain, and corneas of the eyes. Our body needs a small amount of copper from food to stay healthy, but without treatment Wilson disease can lead to high copper levels that cause life-threatening organ damage. The disease is progressive and, if left untreated, it may cause liver (hepatic) disease and brain dysfunction. Early diagnosis and treatment may prevent serious long-term disability and life-threatening complications. Treatment is aimed at reducing the amount of copper that has accumulated in the body and maintaining normal copper levels thereafter. Wilson disease affects an estimated 1 out of every 30,000 people. Symptoms of Wilson disease vary a lot from person to person. Wilson disease is present at birth but the symptoms don't appear until copper builds up in the liver, brain, eyes or other organs. People who have Wilson disease typically develop symptoms between ages 5 and 40. However, some people develop symptoms at younger or older ages. A mutation of the *ATP7B* gene causes Wilson disease. This gene is responsible for removing extra copper from our bodies. Diagnosis relies on serum ceruloplasmin which is a protein that carries copper in the bloodstream. People with Wilson disease often have low ceruloplasmin levels. Plasma copper can be higher than normal. Twenty-four hour urine copper is high in patients with Wilson disease. Liver enzymes are high when there's liver damage. Eye examination can give a clue as well about the diagnosis. Treatment for Wilson disease focuses on lowering toxic levels of copper in the body and preventing organ damage. Treatment includes taking medicines that remove copper from the body by using chelating agents, D-penicillamine or trientine. Later taking zinc will prevent intestines from absorbing copper. Treatment is for life. In addition, it is helpful for patients to be eating a diet low in copper.

Wilson disease is a genetic disorder. It is inherited in an autosomal recessive manner where both parents are carriers and have a one in four risk of having an affected child.

References:

https://rarediseases.org/rare-diseases/wilson-disease/
https://my.clevelandclinic.org/health/diseases/5957-wilson-disease

Management of Wilson Disease

Vanessa So, Registered Dietitian

In addition to chelation therapy to lower toxic levels of copper and zinc supplementation to reduce copper absorption, another key method to control copper levels is to limit the amount of copper consumed through food. Small amounts of copper are still essential for the body, even for those with Wilson disease so a "low copper" diet must be followed. A dietitian can help provide guidance on how foods fit in a low copper diet and strategies to meet copper intake recommendations. Foods are categorized into "low", "medium" and "high" copper content per serving to simplify food groups and help with meal planning. The "low" category contains <0.08mg copper per serving and can be consumed freely. Some examples include cucumbers, rice, cheese, eggs, and blueberries. "Medium" category foods contain 0.08-0.20mg per serving and portions must be monitored. This includes foods such as canned tomato products, wheat bread, chocolate milk, beef, and raspberries. The "high" category contains >0.20mg per serving and these foods must be selected less frequently such as sweet potato, peas, soy milk, nuts/seeds and dried fruits. Some tasty examples of a low copper meal include a tomato and arugula omelet with feta cheese, honey soy sauce glazed salmon, and berry peach crisp!

Sonia follows a low copper diet as part of their treatment for Wilson disease. The foods pictured to the right are homemade brioche bread, cream, sugar, eggs, cinnamon stick, and the peels of lemon, orange, and lime, for a Basque version of French toast called torrijas.

Dietitians' Corner
Newsblast

Formula shortages, back orders amid supply chain issues and medical food discontinuation
Unfortunately, issues continue into 2023.

Abbott

- At this time the dietitian still needs to order any Abbott products for you. Maximum of 2 cases per order.
- *Hopefully* by spring, product should be available for you to order
- Please keep track of your supply
- ***Email me your current supply***

Nutricia

- Phenylade 40 Drink Mix (both flavours) is being discontinued.
- XPhe Maxamum pouches will be discontinued. The can format will still be available
- Phenylade Essentials are not being discontinued; however, the pouch format will no longer be produced. Once Phenylade Essentials is available, only the can format may be ordered
- Phenylade Essentials is scheduled to return in all flavours April 2023.

Additional Information from Nutricia:

LOW-PROTEIN FOODS	
Discontinuation date	**Products being discontinued**
While supplies last or March 31, 2023	All Loprofin and Milupa brand products 49608 – **Loprofin Macaroni** 49620 – **Loprofin Lasagne** 49614 – **Loprofin Rice** 49604 – **Loprofin Fusilli (Spirals)** 49610 – **Loprofin Penne** 49616 – **Loprofin Tagliatelle** 49622 – **Loprofin Spaghetti** 49675 – **Loprofin Animal Pasta** 49636 – **Loprofin Baking Mix** 49654 – **Loprofin Cake Mix, Chocolate** 49635 – **Loprofin Loops** 169609 – **Milupa™ lp-fruity, Apple-Banana** 169608 – **Milupa™ lp-fruity, Pear** 53930 – **Milupa™ lp-drink** 33898 – **Milupa™ lp-flakes (Cereal)** 68498 – **Milupa™ lp-ringlets (Cereal)** 

Dietitians' Corner
Newsblast—continued

More Low Protein Diet Updates...

Replacement for medical food covered by the inherited metabolic disease (IMD) program

Discontinued product	"Replacement" product covered by IMD program
Loprofin Pasta	Aproten Pasta: ditalini, fusilli, spaghetti, penne, anellini, rigatoni, fettucine, tagliatelle
Loprofin Rice	Aproten Rice (chicchi) Cambrooke Short Grain Japanese Rice DS Spanish Rice DS Imitation Rice
Loprofin Baking mix	Taste Connections – Low protein multi baking mix Cambrooke – Wel-made baking mix DS Bread Machine baking mix
Loprofin Cake mix	No direct option May find recipes using baking mixes (above) and/or Taste Connections – low protein Versa mix
Milupa LP fruity cereal	Cambrooke Creamy Hot Cereal Cinnamon or Vanilla Promin Pastameal (porridge)
Milupa LP loops Milupa LP flakes Loprofin LP loops	No direct option Contact your dietitian for non-covered options
Milupa LP drink	No direct option Contact your dietitian for non-covered options

The IMD program has approved the following low protein foods as of February 21, 2023 for any patient registered and requiring a low protein diet

Foods	
Promin Lasagne	Promin Muffin Mix - Cranberry
Promin Ribbed Macaroni	Promin Muffin Mix - Cinnamon
Promin Small Shells	Cambrooke Sugar Cookie
Promin Short Elbows	Cambrooke Chewy Fudge Brownie
Promin Alphabet Pasta	Taste Connection Wheat Starch
Promin Tri-Colour Spirals	Cambrooke Wheat Starch
Cambrooke Aprotide Pasta Stellini	Aproten Flour Mix (Farina)
Cambrooke Aprotide Pasta Spaghetti	Dietary Specialties low protein baking mix
Cambrooke Aprotide Pasta Fusili	Promin Hot Breakfast Orginal
Promin Imitation Rice/Orzo	Promin Hot Breakfast Apple/Cinnamon
Promin Cous Cous	Promin Hot Breakfast Banana
Aproten Riso Rice Mix	Cambrooke Mal-o-Meal: Frosted flakes
PKU Perspectives (Country Sunrise) Apple Cinnamon Muffin Mix	Cambrooke Mal-o-Meal: Coco Roos
PKU Perspectives (Country Sunrise) Orange Cranberry Muffin Mix	Promin Scrambled Egg Mix
Promin Muffin Mix - Chocolate	Cambrooke Chicken Flavoured Consomme and seasoning
Promin Muffin Mix - Banana	
Promin Muffin Mix - Apple Cinnamon	

Dietitians' Corner
Recipes

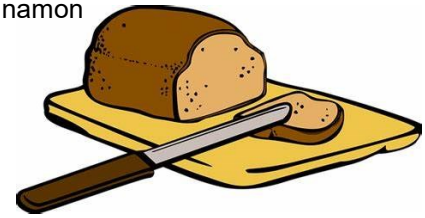
Almost No Fat Banana Bread

A favourite of Karleigh's, VLCAD deficiency. From allrecipes.com
Cook Time: 55 minutes Pan: 8x4 inch loaf Servings: 12

Number of servings	Fat per serving	Protein per serving	Carbohydrate per serving
12	0.5 g	2.6 g	33 g

Ingredients

- cooking spray
- 1.5 cups all purpose flour
- 3/4 cup white sugar
- 1 1/4 teaspoons baking powder
- 1/2 teaspoon baking soda
- 1/2 teaspoon ground cinnamon
- 1 cup banana, mashed
- 1/4 cup applesauce
- 2 egg whites



Directions

- Preheat the oven to 350 °F (175 °C). Lightly grease an 8x4 inch loaf pan
- Stir flour, sugar, baking powder, baking soda and cinnamon together in a large bowl
- Add banana, applesauce, and egg whites; stir just until combined. Pour batter into the prepared pan
- Bake in preheated oven until a toothpick inserted into the centre comes out clean, 50 to 55 minutes
- Turn out onto a wire rack and allow to cool before slicing

Enjoy this low fat treat.
Suzanne

Maple Fudge—A Canadian Treat

This delicious treat is from Céline Mercier Bilodeau and available on www.lowprorecipes.com.
Thank you, Genevieve Lafrance, Registered Dietitian for including this recipe on your website!

	Protein (g)	Phenylalanine (mg)	Leucine (mg)	Fat (g)
Entire recipe	0.3	0	0	1.73
Per serving	0	0	0	0

Number of servings: 36 squares *Serving size:* 1 square

Ingredients

- 540 mL (1 can) maple syrup

Preparation

- Place maple syrup in a large pot
- Place candy thermometer in the pot
- Boil the maple syrup until it reaches 238 °F (114 °C). DO NOT STIR
- Remove from heat and place pan in cold water. Let cool until syrup reaches 125 °F (50 °C)
- Once the temperature is reached, mix with an electric mixer for a few minutes
- When the maple syrup's colour becomes much lighter switch to using a wooden spoon and stir until the mixture is slightly thicker
- Pour into 8 x 8 inch pan lined with parchment paper. Let cool in fridge for a few hours. Cut into squares

ENJOY!
Suzanne Ratko, Registered Dietitian

Dietitians' Corner
Recipes continued

DID YOU KNOW?

Gnocchi (pronounced nyoh·key) is a dumpling that is popular in Italian cuisine. There are different types of gnocchi. The most common variety originated in Northern Italy and is made with potato and wheat flour. Gnocchi can be made with other vegetables (squash or spinach for example), breadcrumbs, other cereal grains (such as corn flour) or nut flours. The recipe below uses cauliflower and cassava flour making it suitable for low protein diets. Cassava is a root plant similar to sweet potato and is naturally gluten-free and nut-free. Cassava flour and cauliflower are high in dietary fiber, low in fat, rich in Vitamin C, and help regulate blood sugar levels by increasing insulin sensitivity.

Cauliflower Gnocchi

Recipe provided by Avery, PKU, Damien, PKU and their parents. A favourite in the Dent house and hopefully yours, too!

Makes 50 Gnocchi

Image source: ottosnaturals.com

Ingredients

500g cauliflower (fresh or frozen)
110g cassava flour (1 cup)

Preparation

- Steam the cauliflower until soft
- Shake off excess water or dry florets in a clean tea towel
- Blend the cauliflower until smooth, get all the lumps out
- Put the cauliflower mash in a big mixing bowl and add the cassava flour
- Knead the flour into the cauliflower
- Scoop a small ball of dough out and roll it into a long log, about a half inch in diameter
- Cut log into pieces about one inch in length
- Repeat for the rest of the dough
- Drop dough pieces into a big pot of salted boiling water and boil for 3 minutes
- When they float to the top they are done, remove with a slotted spoon

	Per entire recipe	Per Gnocchi (when 50 made)
Calories (kcal)	509	10
Protein (g)	12.9	0.3
Carbohydrate (g)	116.2	2.3
Fat (g)	2.07	0.04
Phenylalanine (mg)	391	8
Leucine (mg)	629	13

The gnocchi can be served at this point. However, for best results, after boiling give them a light fry in oil or a little butter until brown and a little crisp. If following a restricted fat diet, do not light fry.

Serve with your favourite marinara or pasta sauce and a little sprinkle of Earth Island parmesan cheese.

Recipe makes about 50 gnocchi. If you make less or more, then calculate amount of protein, carbohydrate, fat, phenylalanine or leucine by dividing the amount made into the entire recipe nutrient information. If you need help, please contact me.

Buon appetito!
Suzanne

Serving suggestion: Sprinkle cauliflower gnocchi with Earth Island parmesan cheese and garnish with a few crispy fried sage leaves (optional)

New Staff Members

Montaha Almudhry

My name is Montaha Almudhry, a mother of two lovely kids and I am a physician. I came from the eastern province of Saudi Arabia, and I landed here in Canada in 2021. I joined the metabolic team here at LHSC for a neurometabolic fellowship which is, unfortunately, passing so fast. I am fond of reading biographies outside duty time, and I like cooking Italian food. My husband thinks that I prepare the world's best pizza.

Sunita Venkateswaran

I am an associate professor in the division of pediatric neurology. I trained at Western for medical school and my pediatric residency followed by a neurology residency at McGill. I then did a 2 year fellowship between the Montreal Neurological Institute and SickKids in pediatric demyelinating diseases.
My interest is in rare disease, specifically in neurodegenerative conditions such as NBIAs and leukodystrophies. I am the PI for both the CPSP surveillance on pediatric onset leukodystrophies as well as for the Canadian site of TIRCON (treat iron related childhood onset neurodegeneration). I have a specific interest in fatty acid hydroxylase associated neurodegeneration (FAHN/SPG35) in which I have an international consortium working on bench to bedside research.

Majdina Isovic

After many years in basic research, focusing on genetics and epigenetics in development and cancer, it was time for a change...a change that would allow me to make a difference for patients whose only hope for diagnosis currently is through research studies. I am also interested in helping improve overall patient care and experience. I am excited to be part of the genetics research team!

Suzanne Nurse

I recently joined the research team in the genetics program as a Research Assistant. My husband, Mark, and I have raised our three children here in London. I am originally from Newfoundland and love to get back to Atlantic Canada when I can. My background is in neuroscience research and more recently patient advocacy. I am looking forward to combining those interests in my new role to advance knowledge, ensure the well-being of participants in our clinical research studies and, ultimately, improve patient outcomes.

Staff Acknowledgements

Samantha Colaiacovo

A big thanks to Samantha Colaiacovo, Genetic Counsellor, for doing a great job with newborn screening and genetic counselling as a valuable member of our metabolic team.

Photo Album

From the Archives

Carver, MMA with Dr. Chitra Prasad in 2010 (left) and with Suzanne Ratko, RD (right)

Karson, PKU

Margie, PKU

Kennady, MMA

Photo and Art Page

Our Stars!

Quentin, VLCAD deficiency

Drawing by Michael Karp, "Flower man and friends"

Katilynn, PKU and sister Adalynn

Katilynn, PKU

Emily, PKU and Kaitlynn, PKU
– friends forever

Kasper, hyperphenylalaninemia

Celebrating Achievements

Karson, PKU, won his Kids Triathlon in July. He swam 150 m, biked 10 km and ran 2 km. Great job Karson!

METABOLIC FAMILY WORKSHOP:

Save the date! April 26, 2024

STAY TUNED.... MORE DETAILS TO BE ANNOUNCED IN THE NEXT ISSUE OF THE INHERITED METABOLIC DISORDERS NEWS

WE ARE EXCITED TO BE PLANNING THIS IMPORTANT EDUCATION EVENT FOR NEXT YEAR. IT'S ALSO A GREAT OPPORTUNITY TO MEET OTHER FAMILIES.

Research and Presentation Updates

Coughlin CR 2nd, Tseng LA, Bok LA, Hartmann H, Footitt E, Striano P, Tabarki BM, Lunsing RJ, Stockler-Ipsiroglu S, Gordon S, Van Hove JLK, Abdenur JE, Boyer M, Longo N, Andrews A, Janssen MC, van Wegberg A, **Prasad C, Prasad AN**, Lamb MM, Wijburg FA, Gospe SM, van Karnebeek C, International PDE Consortium. Association Between Lysine Reduction Therapies and Cognitive Outcomes in Patients with Pyridoxine-Dependent Epilepsy. *Neurology*, 2022 Aug 25; 99(23): e2627–36.

Yang JH, Friederich MW, Ellsworth KA, Frederick A, Foreman E, Malicki D, Dimmock D, Lenberg J, **Prasad C, Yu AC, Anthony Rupar C**, Hegele RA, Manickam K, Koboldt DC, Crist E, Choi SS, Farhan SMK, Harvey H, Sattar S, Karp N, Wong T, Haas R, Van Hove JLK, Wigby K. Expanding the phenotypic and molecular spectrum of NFS1-related disorders that cause functional deficiencies in mitochondrial and cytosolic iron-sulfur cluster containing enzymes. *Hum Mutat*, 2022 Mar 1; 43(3): 305-15.

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. Family Experiences with Care for Children with Inherited Metabolic Diseases in Canada: A Cross-Sectional Survey. *Patient*, 2022 Mar 1; 15(2): 171-85.

Judy Ibrahim, Suzanne Ratko, Melanie Napier, Natalya Karp, C Anthony Rupar and Chitra Prasad. Carbonic Anhydrase (CA-VA) deficiency: an under recognized cause of neonatal hyperammonemia with excellent outcome on proactive management. *Arch Dis Child*, 2022 Aug 17; 107(Suppl 2): A200-1.

Presenters at the Metabolic National Case Series. **Natalya Karp**, **Suzanne Ratko** and Luis Altamirano-Dias (Cardiology) presented at the Metabolic National Case Series, 2022 Dec 12. Title: “Infant with severe VLCAD deficiency on Dojolvi in the past 4 months: our experience”.

Chitra Prasad. Carbonic anhydrase deficiency and underrecognized because of hyperammonemia, 2022 Nov 1, Delhi, India, Continuing Medical Education

Dr. Natalya Karp

Chitra Prasad. Lectures on carbonic anhydrase deficiency and Exome sequencing for developmental delay syndromes, 2022 Oct 30, Rishikesh, Uttaranchal, India, Continuing Medical Education

Chitra Prasad Lecture on organic acidemias for the North American Metabolic Academy, 2022 Oct 9, Georgia, United States, Continuing Medical Education

Presenter at Paediatric Grand rounds. **Dr Chitra Prasad** January 10th 2023 Inborn Errors of Metabolism and Million-dollar therapies

Ataxia update (Workshop for Movement disorders October 2022) **Chitra Prasad** and Mary Jenkins

Dr. Jean-Marie M. Saudubray (centre) and colleagues at the 2022 meeting of the North American Metabolic Academy (NAMA)

Meeting of the Metabolic Minds

Rare Disease Day

Children's Hospital
London Health Sciences Centre

RARE DISEASE DAY®

We observed Rare Disease Day on February 28th!

On Rare Disease Day, Children's Hospital at London Health Sciences Centre (LHSC) officially became a member of the Canadian Organization for Rare Disorders (CORD), the second children's hospital in Canada to join this national network representing all those with rare disorders.

CORD Canadian Organization for Rare Disorders

Dr. Craig Campbell
Chair, Department of Paediatrics and Neurologist

Nash Syed
President, Children's Hospital at London Health Sciences Centre (LHSC)

Durhane Wong-Rieger
President & CEO, Canadian Organization for Rare Disorders (CORD)

There were inspiring remarks from the above speakers as well as representatives from patient organizations and families living with rare disorders including:

- ◊ Perry Esler, Chief Executive Officer, Defeat Duchenne Canada dedicated to curing Duchenne muscular dystrophy
- ◊ Dave McIntyre, Bethanys Hope Foundation, supporting the goal of putting an end to Metachromatic Leukodystrophy
- ◊ Hailey Renneboog and Family — the importance of a diagnosis
- ◊ Nine-year old Daniel Kinchela, Author, and his mother Chris celebrating the official release of his children's book *The First Fire Dragon: An Autoinflammatory Adventure*

Centre: Dr. Tugce Balci (Medical Geneticist) and Dr. Sunita Venkateswaran (Pediatric Neurologist) organizers of Rare Disease Day at Children's Hospital LHSC along with (L-R) Maria Bata, Jack Reilly, Majdina Isovic and Suzanne Nurse

INTERVIEW with Casey Renneboog, Dr. Balci and Dr. Venkateswaran on Rare Disease Day

CBC LONDON

www.cbc.ca/listen/live-radio/1-158/clip/15969118

Hope!

Hope is an important factor in health and well-being. Hope involves the belief that one has the power and ability to influence one's circumstances and create a positive change. There are many benefits to having hope. Research has shown that having a hopeful mindset can result in better health outcomes for children and adults living with chronic conditions. By encouraging and fostering hope, we can better equip ourselves to manage difficult life circumstances.

"In studying hope, so too have I observed the spectrum of human strength. This reminds me of the rainbow that frequently is used as a symbol of hope. A rainbow is a prism that sends shards of multicolored light in various directions. It lifts our spirits and makes us think of what is possible. Hope is the same — a personal rainbow of the mind."

- C.R. Synder, psychologist known for Hope Theory

The blogpost, "How to Face Adversity and Become More Hopeful", by Jennifer Guttman Psy.D., describes ways you can learn to incorporate hope and hopeful thinking into your daily life. There are explanations and concrete examples provided. This is a skill, and with practice each of us can learn to have a more hopeful outlook.

Six techniques to help you be more hopeful in your day-to-day life:

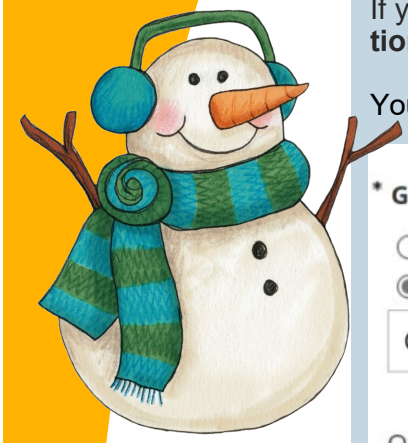
1. Manage negative, hopeless thoughts.
2. Learn to cope with challenges.
3. Contribute positively to someone else's life.
4. Try to surround yourself with other positive people.
5. Learn to focus on solutions to problems you can control.
6. Concentrate on what is working in your life.

From: How to Face Adversity and Become More Hopeful, *Psychology Today* (2020, July 15).

"Hope: How to Face Adversity and Become More Hopeful" is part one of a four-part series from *Psychology Today* on skills for better mental health. Links to each blogpost in the series are below. The series was published in 2020 during the first year of the COVID-19 pandemic and was written with this particular challenge in mind. However, the techniques can be applied to many different circumstances.

Introduction: H.E.A.R. A New Series Using 4 Skills For Better Mental Health
Part I. **H**ope: How to Face Adversity and Become More Hopeful
Part II. **E**mpowerment: Utilizing Empowerment to Manifest More Self-Confidence
Part III. **A**daptation: Learn How Adaptation Is a Key to Embracing Change and Growth
Part IV. **R**esilience: How It Can Help Improve Your Mental Health

"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



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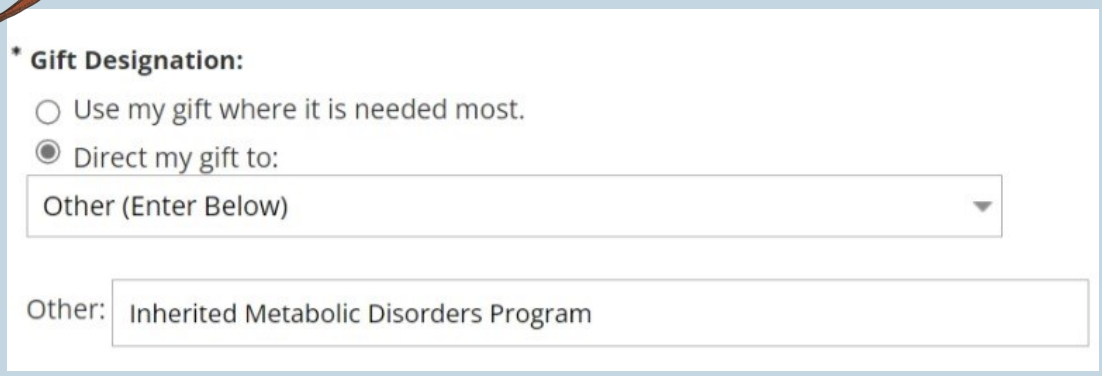

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