The Inherited Metabolic Disorders News

Children's Hospital
London Health Sciences Centre

Summer 2019 Volume 16 Issue 2

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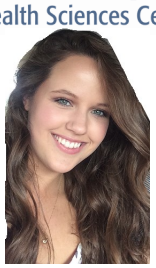
In this Issue



From the Editor

It was so wonderful seeing so many of you at our Metabolic Family Workshop in May! A big thanks to everyone who joined us and to all who were involved in making this exciting day a success!

Meghan Zadorsky

From Dr Chitra Prasad

Dear Friends,
Greetings!
We had a very successful 2019 Metabolic Family Workshop and low protein cooking demonstration. It is always very nice to see so many of the families in one place. We have included the highlights of the Metabolic Family Workshop in this issue. Thanks to Meghan Zadorsky (genetics assistant) who is instrumental in organizing this newsletter.
Our theme this year was "*You are not your disease*". It is one of the most empowering statements I have come across. It was given by one of the participants at the last workshop.
I am very grateful to the speakers, our team, volunteers, sponsors and most of all to the patients and families who made this day a grand success. I have heard many good things about our workshops. Other centers are now trying to emulate our Metabolic Family Workshop and the low protein cooking demonstration.
Our patient story is of little Anik who came to us at LHSC when he was less than a week old with high ammonia levels. His adoring parents Hardeep and Sharan are extremely committed to his care. Anik underwent a number of investigations and treatments to bring his ammonia down and I am happy to say that we did find the cause of his high ammonia levels. He has carbonic anhydrase VA deficiency which is an extremely rare condition that can cause high ammonia. He continues to do very well. You can read about their story in this newsletter.
In this issue we have also summarized some strategies for coping with medical and other psychological issues as families are dealing with various appointments, investigations and treatments.
I hope you all take some time to spend outdoors and enjoy the gorgeous summer, trees, flowers and nature's beauty.
Our journey becomes easier when we have the support of our families, friends and community. My sincere hope is that you all find this through our Metabolic Family Workshops and newsletters. Please do share your news and accomplishments.
Best wishes
Your friend,

Chitra Prasad

"When we love, we always strive to become better than we are. When we strive to become better than we are, everything around us becomes better too."

— Paulo Coelho

Personal Stories

February 25th 2019 our lives changed forever. Our sweet little boy, Anik Singh Puaar was born at 1:25pm, weighing a healthy 9.5 ounces - with ten perfect little fingers and ten perfect little toes. My husband and I were so excited to finally meet our baby boy after what felt to me, the longest labour in history – 30 hours later he was finally here.

After a long delivery process our beautiful Anik, who appeared to be a healthy and happy baby not only to us but also to his entourage of visitors who came to see him that day, was in for the fight of his life. We had a great first night at the hospital, going through all the motions new parents go through. We remember waking up the next morning, exhausted but also so proud that we survived our first night as parents.

After having a cesarean delivery, we were required to spend a couple of nights at Grand River Hospital to make sure both Anik and I were healthy and ready to go home when the day came – unfortunately that day did not come as soon as we had anticipated. That evening, February 26th we noticed a dramatic change in Anik's behavior; something wasn't right. He was crying hysterically, he was restless, his breathing was very fast and wasn't taking his feed well either. We called the nurse on duty that evening, and she felt it was necessary to check his blood sugar levels and realized that they were low. Being new parents, this scared us but the nurse assured us that this often happens with newborns and that we have nothing to worry about. She advised us to introduce formula to his diet so we can get more food into his system faster to bring his sugar levels up. This brought Anik temporary relief as he did settle down, a little. But throughout the night Anik was still being fussy and not sleeping well.

The next morning, February 27th, a new nurse was on duty and came to check in on us. We told her that we had a rough night with him and he wasn't taking his feed well again, and we distinctly remember her saying "welcome to parenthood!". The nurse, Lori then asked to hold Anik so she could try to feed him and she instantly noticed that something was seriously wrong. She left the room and immediately called the pediatrician on call that morning, Dr David Creswell. Dr. Cresswell came in to look at Anik and shared the same concerns and felt it was necessary to have Anik admitted to the Neonatal Intensive Care Unit. After a couple of hours of anxiously waiting, Dr Creswell had come back to inform us that he had run a lot of different tests and he ran one test in particular, an ammonia test – the test that saved our son's life. Anik's ammonia levels were reading at a dangerously high number for a newborn and he needed immediate treatment which was unavailable at Grand River Hospital. Dr. Cresswell told us that he had already made the arrangements for Anik to be transferred to London's Children's Hospital and the ambulance was on the way to pick him up. There are no words to describe what myself and my husband Hardeep were feeling in that moment. We were beyond terrified, feeling numb and helpless – our little boy was really sick.

The ambulance arrived at GRH hospital around 3pm that afternoon to transport Anik to London and unfortunately neither Hardeep or myself were able to ride with him. Still in extreme pain from the cesarean surgery, Hardeep and I packed our bags – called our parents who arrived at the hospital shortly after, got into our car and followed the ambulance to hospital in a terrible snowstorm. That will always be one of the longest, most silent care rides of our lives.

Arriving at LHSC at 6:20pm, we met with the team who were looking after our son. The team escorted us into a room where our baby was in an incubator with tubes attached to every part of his body. I remember thinking in that moment, is this really happening? This has to be a nightmare. My pregnancy was so perfect, we were so happy only a couple of hours ago… how could this be happening to us. Every feeling of disbelief took over our minds and bodies.

Shortly after being there, we met Dr. Victor Han, the neonatologist on duty that evening. At this point all we knew was that Anik's ammonia level had now increased from 265 to 325 since he had left Grand River Hospital. After a few hours of waiting Dr. Han had informed us that the issue Anik was having was a metabolic one and it was very serious. I think the only sentence Hardeep and I remember from this initial conversation was "Anik is very sick...", after that sentence it was so difficult to process anything beyond that. How does something like this happen? But Dr. Han reassured us that we were in the right place, and we had the full support of the metabolic team behind us and they were going to do whatever it took to save our baby.

That night we were put up in a hotel room by LHSC as there were no beds available at the hospital. The early morning of February 28th at 3am we received a phone call informing us that a medication they had given to Anik did not work and we were asked to come back to the hospital right away. Arriving at the hospital, we were told his ammonia level did not come down as they had hoped, and it had actually spiked since he first arrived. Due to this, Hardeep and I had to give our permission for Anik to undergo hemodialysis in the Pediatric Critical Care Unit (PCCU)- one of the scariest decisions we have ever had to make.

Shortly after, we were introduced to Dr. Chitra Prasad – Director of Metabolic Services at LHSC. Dr. Prasad informed us she was aware of the situation and was working very hard to find a diagnosis so she could prescribe the correct medicine cocktail for our son. Later that morning, our son was transferred to the PCCU as they prepped him for hemodialysis, a process which would last six hours. With the support from close loved ones, and the PCCU team we were able to get through the hardest six hours of our lives. Later that evening, in a group meeting with the doctors, staff and family we were informed that the dialysis was successful, and the ammonia levels had dropped from 365 to 96. Still not as low as they had hoped, but a great step in the right direction. The path moving forward was to monitor his ammonia levels in hope that they would continue to drop with the concoctions of medicine put together by the metabolic team.

The next two days Anik remained in the PCCU recovering from dialysis and we couldn't have been happier with his progress. Anik's levels were dropping by the hour, getting him to a place the team was happy with. Regrouping with the team, we were introduced to Dr. Tony Rupar – Director of Biochemical Genetics who had been working very closely with Dr. Prasad in helping to find a diagnosis for Anik. They were very happy with the progress Anik was making. We remember Dr. Rupar telling us how he was stumped by Anik's case as it had many mysterious elements that he had not seen before. Anik was now responding to the medications well and was introduced to formula which was being monitored by dietician Suzanne Ratko. Although we still did not have a diagnosis, we were all very hopeful that the management plan that we had in tact was working well. As days went by, each day had its own small victory as one more tube was coming off our baby. March 5th, 2019 we were discharged from LHSC without a diagnosis, but with the right tools, support and guidance to take our baby home.

Today we have diagnosis for our son. Anik has a rare condition known as carbonic anhydrase VA deficiency. There have only been 10 reported cases of this condition worldwide, and Anik is the only reported case in all of Canada. As rare as this disease is, the team of doctors are very happy with the outcome and diagnosis as this is a transient condition. They are very hopeful that our son will have a normal life as he gets older and stronger.

Anik is doing very well, and that is all because of the incredible team at LHSC. He takes biotin vitamins daily and been getting his ammonia levels checked weekly. After going through such a traumatic and devastating experience, we are now enjoying our son and looking forward to the adventures ahead of us.

We will forever be grateful and indebted to the teams at London and Grand River Hospitals. Thank you from the bottom of our hearts for saving our son's life.

Featured This Issue

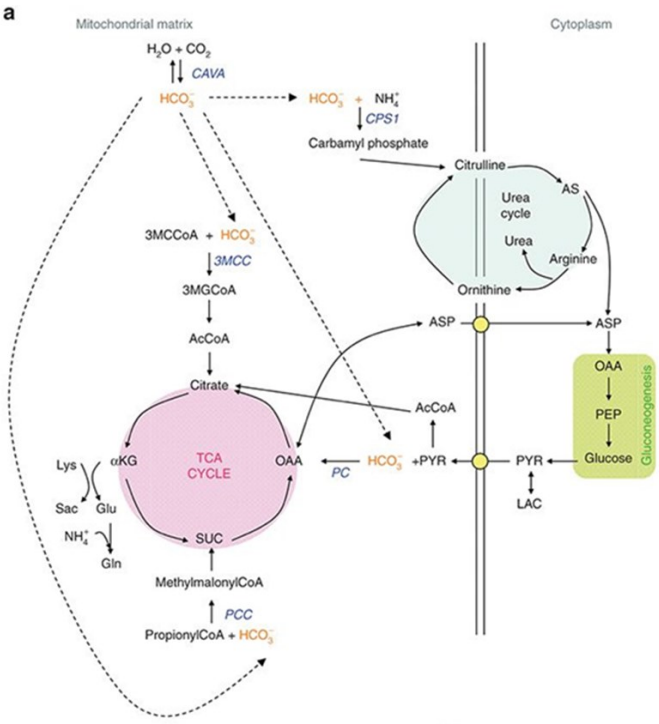
Carbonic Anhydrase VA Deficiency

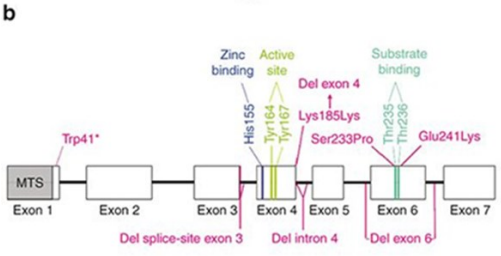
Compiled by Dr. Chitra Prasad

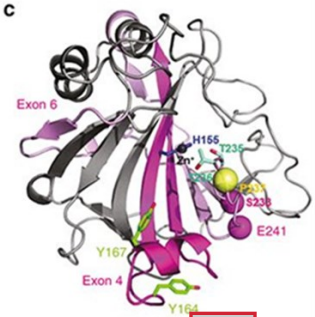
Carbonic anhydrase VA (CA-VA) deficiency can present between day two of life and age 20 months with hyperammonemic encephalopathy (i.e. lethargy, feeding intolerance, weight loss, rapid breathing, seizures, and coma). It is an extremely rare metabolic disorder with less than 10-15 patients described. Diagnosis is suspected based on neonatal, infantile, or early-childhood metabolic hyperammonemic encephalopathy combined with high lactate and metabolites suggestive of multiple carboxylase deficiency, and molecular testing with biallelic pathogenic variants in *CA5A*. Most individuals only have one episode of hyperammonemic encephalopathy. A suggested explanation for this phenomenon is that the CAVB isoform, the only other carbonic anhydrase expressed in liver mitochondria undergoes a maturation effect. Acetazolamide should be avoided as it inhibits carbonic anhydrase activity. During acute decompensation, the laboratory findings are consistent with dysfunction of all four enzymes to which CA-VA provides bicarbonate as substrate in mitochondria (i.e., carbamoyl phosphate synthetase 1 (CPS1) , propionyl CoA carboxylase, pyruvate carboxylase, and 3-methylcrotonyl CoA carboxylase), thereby differentiating CA-VA deficiency from other urea cycle disorders (see pathway below).

During acute episodes patient should be admitted to the hospital. Always provide IV fluids (with glucose at maintenance doses) and extra calories via IV lipids; restrict protein intake if plasma ammonia is elevated. Monitor plasma ammonia, serum lactate, serum glucose, blood gases, electrolytes, and liver parameters. Consider administration of carglumic acid (which – though not approved yet for this indication – enhances CPS1 activity and thus partially compensates for reduced HCO_3^- resulting from CA-VA deficiency). Other ammonia-lowering medications such as sodium benzoate would also be reasonable.

CA-VA deficiency is inherited in an autosomal recessive manner. At conception, each sibling of an affected individual has a 25% chance of inheriting two *CA5A* pathogenic variants.







Reference: Carbonic Anhydrase VA Deficiency

Gene reviewshttps://www.ncbi.nlm.nih.gov/books/NBK284774/

By Clara van Karnebeek, MD, PhD, FCCMG and Johannes Häberle, MD.

Pathway
From: Defective hepatic bicarbonate production due to carbonic anhydrase VA deficiency leads to early-onset life-threatening metabolic crisis

https://www.nature.com/articles/gim2015201?proof=true

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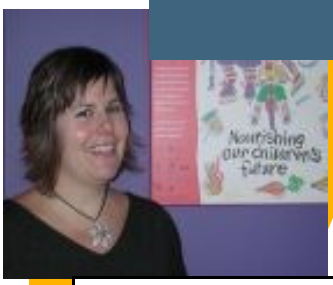

Suzanne's Corner

From Jon's Kitchen

Pulled Jackfriut

"Our kids like it plain on a low protein bun while we like it 'grown up' with pickles and coleslaw on top"

	Amount in grams
Jackfruit in brine	400 g (2 cans)
Onion, red, chopped	200 g
Garlic, fresh, minced	6 g
Paprika	2 g
Ginger, ground	1 g
Bullseye Guinness BBQ sauce	350 g
Salt, table	to taste
Oil	15 mL (1 tablespoon)

Instructions:

- Drain and thoroughly rinse jackfruit in colander. Remove the tough core and pull the jackfruit apart. Then rinse again in colander
- In a skillet, heat oil, sauté onion and garlic until translucent for 3-5 minutes
- Add rinsed jackfruit to onions. Stir
- Add seasonings and sauté for another 2-3 minutes until slightly brown
- Add BBQ sauce to coat the jackfruit
- Salt to taste

Entire recipe contains**

Protein (g)	10.9
Phenylalanine (mg)	297

**based on howmuchphe.org

Carnitas

"Serve in tacos, quesadillas or as a salad topper"

	Amount in grams
Jackfruit in brine	200 g (1 can)
Onion, white, chopped	100 g
Cumin	2 g
Chili powder	4 g
Paprika	1 g
Garlic powder	1 g
George Washington golden broth mix	2 packets
Salt, table	to taste
Oil	15 mL (1 tablespoon)

Instructions:

- Drain and thoroughly rinse jackfruit in colander. Remove the tough core and pull the jackfruit apart. Then rinse again in colander
- In a skillet, heat oil, sauté onion and garlic until translucent for 3-5 minutes
- Add jackfruit and rest of seasonings
- Stir to coat and sauté for 5 minutes
- Add Coca-Cola and stir in. Continue to cook at medium to high temperature. This will caramelize and become slightly crisp. This takes about 10 minutes

Entire recipe contains**

Protein (g)	4.1
Phenylalanine (mg)	139

**based on howmuchphe.org

Jon is the proud father of Avery (5) and Damien Dent (3)– both with PKU seen here making their own low protein pizza

What's New

Metabolic Family Workshop 2019

Medical Genetics Program hosts 13th Metabolic Family Workshop

The 13th Metabolic Family Workshop was held on May 3 at the Best Western Lamplighter Inn. In following with this year's theme, *You are not your disease*, the full-day workshop aimed to build and enhance support and connections amongst patients, families and care providers, and to educate on metabolic disorder management. With more than 200 patients, families and physicians in attendance, it was the most successful workshop to date.

Metabolic disorders are multi-systemic in nature, and children and adults living with them can have problems with growth, developmental delays, and issues with organs such as the heart and kidneys, brain and others. These disorders can require special restrictive diets, as well as enzyme replacement therapies, medications and organ transplant.

Dr. Chitra Prasad, Medical Geneticist and Director of LHSC's Metabolic Clinic, says that awareness about metabolic disorders in the general public is quite limited, underscoring the value of the workshop for patients and families. "Families generally feel lost as they receive a diagnosis of a metabolic disorder," she says. "Our workshop aims to support the families through their journey by providing them with educational resources, resiliency strategies and opportunities for networking."

The workshop, held every two years, features a combination of scientific lecture covering broad topics of newer treatments and other self-management skills, and breakout groups that educate about different metabolic disorders. The workshop is offered free of cost for patients and families, and child care is available to allow parents the opportunity to fully benefit from the day's activities.

The day opened with greetings from LHSC's President and CEO Dr. Paul Woods. President and CEO of Children's Health Foundation Scott Fortnum welcomed the families during the second half of the workshop. The foundation has been a strong supporter of the workshop and helped fund the day's activities and provided volunteers.

Guest speakers throughout the day covered topics such as therapies and their impact on metabolic disorders, coping strategies and adult resiliency. Dr. John Mitchell, Associate Professor from McGill University, delivered the keynote lecture, *New therapeutic advances in rare disease: How do you know if it is right for you?*

Breakout groups, run by medical professionals and families, were divided into morning and afternoon sessions. Morning sessions were condition-specific to ensure relevant information was presented to participants, and afternoon sessions were organized by age groups.

A lunch (including low protein items) was served and enjoyed by participants. In the afternoon, three families spoke to the entire group regarding their journeys with metabolic conditions through the years and coping strategies. This is an important part of the workshop that allows families to share how to remain resilient when facing uncertainty.

The following day a low protein cooking workshop was conducted by Dietician Suzanne Ratko and Chef Patrick Wiese. This event allows families to connect with each other while also learning new low protein recipes to help make meals more exciting and tasty.

The event is organized every two years by the Metabolic Family Workshop Committee, consisting of physicians, parents/patients, administrative staff, genetic counsellors, social workers, dieticians, art therapists, child life specialists, and representatives from Children's Health Foundation. The committee distributes surveys to allow families to evaluate the workshop, and will utilize this feedback while planning their next workshop, scheduled for 2021.

"We are proud and grateful for the hard work of the Metabolic Family Workshop Committee, sponsors and service groups which make this unique event possible," says Dr. Chitra Prasad. "We are all driven by the belief that families benefit greatly when they belong to a supportive community, and this event goes a long way in helping them to cope with challenges. We are thankful to the Lamplighter inn for providing a beautiful venue for our workshop. Many metabolic centres are getting inspired from our metabolic workshop and are considering similar programs."

Article written by Cassie Dowse and the Metabolic Team

Lessons From Families

A focus group came together at the Metabolic Family Workshop to discuss how families and individuals can preserve energy on their medical journey. These are some of the words of wisdom that **families** shared. Special thanks to the participants of the "Other Metabolic Disorders" workshop group for their contributions and to Darren Connolly for compiling.

Self care

Add something into your routine that you enjoy

Educate people and advocate

Acceptance

Respite

Self-compassion

Recognize when you are tired

Be organized and prepared

Follow a routine

Contact community organizations

Pace yourself

Take some quiet time

10 Ways to Build Resilience
The American Psychological Association

Special thanks to Dr. Julie Richard for providing these powerful suggestions in the Adult Resiliency workshop group.

Make connections

Avoid seeing crises as insurmountable problems

"Happiness is a journey not a destination"

Accept that change is a part of living

Move towards your goals

Take decisive actions

Look for opportunities for self-discovery

Nurture a positive view of yourself

Keep things in perspective

Maintain a hopeful outlook

"The flower that blooms in adversity is the rarest and most beautiful of all."

— Walt Disney Company, Mulan

Take care of yourself

Workshop Summaries

MCADD

The Freeman family spoke to the group and shared their story starting with the diagnosis of MCADD in their daughter, including the newborn screening process and what it has been like having a child with this condition. Families in this session asked questions of each other and shared their thoughts, feelings, concerns and experiences.

Lysosomal Storage Disorders

Drs. John Mitchell and Tony Rupar led an informal discussion on the basics of lysosomal storage diseases and the principles, successes and limitations of various therapeutic strategies. Families participated with questions about specific diseases and future therapies currently being developed.

Child Life

In a group session facilitated by Child Life Specialists Erika Clements and Karen Groeneweg school-aged attendees participated in Coping Bingo and Mindful Slime. Each child took away a tool kit of items and strategies to help them relieve stress, increase attention and manage their emotions.

PKU

Families learned what 's past, present and new in the monitoring and treatment of PKU. Dr Ellen Vriezen explained neuropsychological testing and why we need it. Lama Kadri, a nurse with PKU, showed the group how to do blood dot card monitoring at home. Justin Denbok, PKU, and his wife Katie shared the struggles and successes of living with PKU. Dr Andrea Yu discussed the treatment of PKU.

Parents and Caregivers– Coping Strategies

Social Worker Sue MacLean and Darren Connolly provided a presentation on the topic of Resilience and Coping – using the tools of Self Care and Compassion. The audience was invited to participate in the session, which generated excellent discussion and reflection on the strategies that help us to reframe our negative thinking and experiences. Parents shared powerful content that facilitated a mutually supportive dialogue!

Urea Cycle Disorders

Dr. Natalya Karp led a discussion with individuals with a diagnosis of a urea cycle disorder and their family members to review aspects of management, diagnosis, and some of the challenges that this patient population faces. Participants were able to share stories from their own journeys and provide supportive suggestions to others in attendance.

Art Therapy

In keeping with the theme "You Are Not Your Disease", the Art Therapy workshop encouraged young teens to share aspects of themselves beyond their medical condition. This created an opportunity for discovering their individual uniqueness, in addition to their similarities. The art activity involved transforming a blank mask into a representation of their interests, hobbies, desires and strengths. This led to positive sharing and promoted a sense of group cohesiveness.

Low Protein Cooking Demonstration

On Saturday, May 4th, we held the 15th annual low protein cooking demonstration. We had the largest crowd thus far and we had the best volunteers ever!!

I would like to acknowledge those that helped make the event so successful:
Vitaflo for sponsoring the event
Chef Patrick for delicious recipes and ideas
Volunteers
Leslie Harden
Alissa Morrison
Rachel Miller
Kaitlyn Studenny
Nicole Clouthier
and Sarah Denomme. Sarah has helped out for the past 10 years! I do not know what I would do without her!

Suzanne

Breanne Freeman– mother of child with MCADD

Erin Little– mother of child with cystinosis

Paul Kuntz– homocystinuria

A very special thanks to all of the patients and family members who shared their experiences at the Metabolic Family Workshop. We appreciate the courage to share your stories and your willingness to help build a supportive community.

Donald Doan– OTC deficiency

Chantal Massicotte– mother of child with MPS I

Lama Kadri– PKU

We look forward to seeing everyone at the next Metabolic Family Workshop in 2021!

Publications

Remtulla S, Emilie Nguyen CT, **Prasad C,** Campbell C.
Twinkle-Associated Mitochondrial DNA Depletion.
Pediatr Neurol. 2019 Jan;90:61-65.

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Health services use among children diagnosed with medium-chain acyl-CoA dehydrogenase deficiency through newborn screening: a cohort study in Ontario, Canada.
Orphanet J Rare Dis. 2019 Mar 22;14(1):70.

Aref-Eshghi E, Bend EG, **Colaiacovo S, Caudle M**, Chakrabarti R, **Napier M**, Brick L, Brady L, Carere DA, Levy MA, Kerkhof J, Stuart A, **Saleh M**, Beaudet AL, Li C, Kozenko M, **Karp N, Prasad C, Siu VM,** Tarnopolsky MA, Ainsworth PJ, Lin H, Rodenhiser DI, Krantz ID, Deardorff MA, Schwartz CE, Sadikovic B.
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Am J Hum Genet. 2019 Apr 4;104(4):685-700.

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Infantile-Onset Multisystem Neurologic, Endocrine, and Pancreatic Disease: Case and Review.
Can J Neurol Sci. 2019 May 6:1-5.

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Parental Psychosocial Aspects of Inborn Errors of Metabolism. *(manuscript currently under revision)*

Gannavarapu S, Prasad M, Prasad C. Inborn Errors of Metabolism in Newcomer and Refugee Populations in Ontario, CA.
UWO Medical Journal. 2019 *(accepted)*

Madhavi Prasad, Srinitya Gannavarapu and Chitra Prasad
A case of mistaken identity: succinic semialdehyde dehydrogenase deficiency in a refugee child in Ontario

Srinitya Gannavarapu, Chitra Prasad, Asuri N. Prasad, Tugce B. Balci and Tony Rupar
Hypomyelinating leukodystrophy: do heterozygous variants in HSPD1 deserve a closer look?

Oral Presentations

Chitra Prasad, Dhandapani Ashok, **Samantha Colaiacovo**, Joanna C. Walsh, Robert A. Hegele and **Tony Rupar**
Acid Lipase Deficiency from Diagnosis to Therapy in London Ontario Canada.
Sherbrook Canadian Lysosomal meeting

Chitra Prasad
Lectures presented at All India Institute of Medical Sciences in New Delhi, India (February 2019)

- Hypophosphatasia: 3 years follow up on Enzyme replacement therapy (Strensiq)
- Acid Lipase-Disease to therapy (Kanuma)
- Homocysteine and Neurological disorders
- Laboratory and Clinical metabolic approach (SUCLA, SUCLA2-Related Mitochondrial DNA Depletion Syndrome, Encephalomyopathic Form with Methylmalonic Aciduria and Molybdenum cofactor deficiency) Unusual presentations

Suzanne Ratko
Case study presentation: Cow's milk protein allergy and VLCADD
2019 Abbott Nutrition Metabolic Conference in Memphis, Tennessee (May 2019)

Chitra Prasad
International guidelines for Propionic and methylmalonic and isovaleric acidemia
Garrod meeting (May 2019)

Jessica Tao, Dr. Tony Rupar, Suzanne Ratko, Dr. Michael R. Miller, **Dr. Chitra Prasad**
Use of complementary and alternative medicine in patients with inborn errors of metabolism: A single -centre study.
Garrod (May 2019)

Poster Presentations

Chitra Prasad, C. Anthony Rupar, A. Narayan Prasad
Clinical, biochemical and molecular characterization of rare inborn errors of metabolism (IEM) presenting as movement disorders.
Garrod (May 2019)

London Health Research Day 2019, Child Health Research Day 2019 & Garrod International Symposium 2019

Prasad M, Gannavarapu S, Mailo J, **Prasad C**.
A case of mistaken identity: succinic semialdehyde dehydrogenase deficiency in a refugee child in Ontario.

Gannavarapu S, Prasad C, Balci TB, Prasad AN, Rupar CA.
Hypomyelinating leukodystrophy: do heterozygous variants in *HSPD1 deserve a closer look?*

Prasad C, Prasad AN, Rupar CA.
Clinical, biochemical and molecular characterization of rare inborn errors of metabolism (IEM) presenting as movement disorders.

Our Stars and Inspirations

Emily Mclennan (1), PKU

Kaitlynn Broad (1), PKU with dad, Chad

> **"Perfection is no small thing, but it is made up of small things."**
>
> **– Michelangelo**

Evelyn Fehr (5), hypophosphatasia

Jasper More (10), MPS VI

Aaliyah Solomon-Brown (8), Gaucher

Achievements

Jesse Davidson

We would like to send a BIG congratulations to Jesse Davidson, 19 (PKU) for finishing his first year at University of Windsor in the mechanical engineering with aerospace program with a 96% average! Jesse had the highest marks in several of his classes and also one of the highest averages in his year!

Jesse said that he went into this year prepared to spend the majority of his time studying and to avoid procrastination which helped reduce stress, get excellent grades, and made the transition into university seamless. Jesse says that "university is all about the work you put into studying. Put the time and work in and it will pay off". The PKU diet has been a part of Jesse's life for quite some time now, but he does mention that he finds it difficult to think straight when he misses a day of Kuvan, so he makes sure he doesn't forget it, especially before any major exams.

In the future, Jesse hopes to work for Space X or Blue Origin (the big space companies) as an engineer for one of the rockets that will go to Mars. He would also like to complete his Masters and PhD at Massachusetts Institute of Technology (MIT).

Congratulations Jesse all of your accomplishments this year, we are so proud of you and we can't wait to see what your future holds!

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Website: http://www.lhsc.on.ca/Patients_Families_Visitors/Genetics/Inherited_Metabolic/index.htm

Parent Support Contact:
Jennifer Culp

Tel: 1.519.632.9924

Email: jennc2011@hotmail.ca

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!