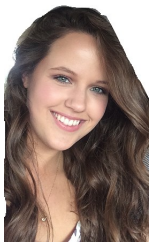
The Inherited Metabolic Disorders News

Children's Hospital
London Health Sciences Centre

Winter 2019 Volume 16 Issue 1

Help us save the trees! To receive the next issue of the newsletter by email contact meghan.zadorsky@lhsc.on.ca

In this Issue



From the Editor

I am thrilled to be taking over as editor of this exciting newsletter. I am thankful for the opportunity to get creative while sharing great information and showcasing stories of our wonderful patients! A big thanks to Dr. Prasad for the opportunity and to the whole team for supporting me as I step into this role.

Meghan Zadorsky

From Dr Chitra Prasad

Dear Friends,

Greetings!

I am very happy to share this new Metabolic Newsletter with you. Janice Little (our resource associate) whom many of you know has retired after 31years of service to LHSC. She was instrumental in the success of newsletters and the Metabolic Family Workshop. I wish her a very happy retirement. Our team is very fortunate to have Meghan Zadorsky who has done a great job as the editor of our current news-letter. Meghan is a genetics assistant who has been extremely helpful. Dorothy Michalski, our other genetics assistant, is also helping with the organization of our Metabolic Family Workshop in 2019.

Congratulations to Suzanne Ratko (dietitian) on being nominated for the President's award this year. Suzanne has worked tirelessly for many years and has impacted so many of our patients and families such as the Buchkos.

This newsletter carries the story of Jonathan Buchko, our young star with OTC deficiency (ornithine transcarbamylase deficiency- a rare urea cycle disorder). Danielle (his mother) has shared with us the challenges and opportunities in dealing with this disorder. Thanks Danielle! Jonathan is doing very well thanks to his mother and father, Suzanne Ratko, and his compliance with medications.

Preparations are underway for our next Metabolic Family Workshop on Friday **3rd May 2019**. Please note this day in your calendar. Also for patients with dietary conditions, we will be having a low protein cooking demonstration on the **4th May 2019**. Please don't forget to register for either or both events as applicable. Please let us know the names of everyone who will be attending so that we can organize well. The workshop is offered free of cost thanks to our sponsors and Children's Health Foundation.

The Metabolic Family Workshop in 2019 carries the theme "**You are not your disease**". This is the title I got from one of you during last workshop's evaluations. We all loved the title.

There is lot of new information in this newsletter. Specifically Dr. An-drea Yu has joined us as another metabolic specialist. Welcome Dr. Yu!

Please send your contributions, your achievements, and pictures for the newsletters.

Also please share your emails so we can save the trees!!

With best wishes and regards

Your Friend,

Chitra Prasad

"It always seems impossible until it's done"—Nelson Mandela

Personal Stories

Jonny's Journey

Boy or girl? What are you hoping for? Myself, I never cared to find out; happy and healthy is all I hoped for. My husband's persistence on wanting to know the gender paid off and we found out our first born would be a boy. We were absolutely ecstatic. Jonathan David Buchko, entered the world on Jan. 13, 2014. A boy, indeed, and happy and healthy. He was such an easy baby. Slept well, ate well, never fussy, he was perfect in every way.

Jonny reached 6 months and we naturally started introducing solids. He liked almost everything with the exception of fruit now and then. At about 8 months I stopped nursing him and started formula. It was then that Jonny started randomly throwing up. It was very odd, he never really spit up as a baby and now, here he is, up to 3 times a week randomly throwing up. Naturally, as a first time mom, I brought him to his family doctor several times, concerned, which was always chalked up to a "virus." There were a couple of times I was so concerned I had him at urgent care as he became lethargic. I never once left the doctor's office or urgent care feeling like they had the answer to what was going on with my son and causing him to get sick. At one point in urgent care a doctor flat out told me he was just having a 'bad day' similar to how you and I just have bad days. I knew, as a mother, something was off. It's so easy to chalk things up to a virus but I never wanted to hear the word virus again. I was not looking for something to be wrong with my son although I knew something wasn't right. At my wits end, I was in his family doctor's office in tears as she is saying "he is fine, it's just a virus" again. She knew by the look in my eyes that a mother is always right and made a referral to a gastro doctor. This ultimately was not helpful because before any appointment from a referral was even made we ended up back in the emergency room as Jonny was throwing up by now 3 times a day and was extremely lethargic. This was a 2 month journey from when he randomly started getting sick to it becoming up to 3 times a day.

We caught the attention of one amazing doctor in the emergency room who picked up on a cue of explaining that Jonny went from being days away from crawling to not even being able to get on all fours. From saying "mama" and "dada" to not saying it for weeks. Jonny was regressing in his development and this triggered this doctor to look at Jonny's ammonia level. The results from this test changed our lives forever. We sat across the table from medical professionals in a private room and were told that Jonny either had OTC or a liver disease. Of course at the time I didn't know what any of that meant. The only words I wanted to hear was "your son is going to be ok!" At this point he was admitted to critical care with an ammonia level that was at 300; a normal level is below 55.

I remember those 9 days he was admitted in Children's Hospital like they were yesterday. We spared most of our family and friends from visiting him as the number of cords connected to my precious baby was so excessive it was hard to bare. Jonny has OTC. A metabolic disorder that limits his protein intake.

We left the hospital after 9 days and our world was turned upside down. We didn't know what the future of our son looked like. We visited Sick Kids in Toronto for a couple days to explore the option of having a liver transplant as this would be the only "cure" to OTC, however, that option comes with its own complications. We were told there's a 1% chance that a transplant is not successful which means 1 in 100 children do not survive a transplant. Jonny had a 1 in 80 000 chance of getting OTC; I didn't think the numbers were in our favour. My husband and I decided that based on our son's best interest we would not move forward with this option.

We started counting protein from 4 grams a day and gradually increased his protein intake by a gram every couple of months as he was able to manage.
High ammonia can affect behaviour and cause someone to be very irritable and aggressive. It can also cause you to refuse to eat and become sleepy. Well, for those with kids from the age of 1-3 you can imagine my demise. I felt like I was up at the hospital every 2 weeks getting a blood test done to see if Jonny's ammonia was rising. Ammonia can rise in the event of stress to the body such as an illness or broken bone. Teething, for goodness sake, could be a stress on your body. In all of these situations we were constantly getting a blood test done to ensure Jonny's ammonia was fine.

Low and behold, since we left the hospital after being diagnosed when Jonny was 10 months old we have never had to be admitted again. Jonny has been able to deal with many "actual" viruses without having an elevated ammonia. He has also had two surgeries: one for ear tubes and one for a herniated belly button. Both of these were unrelated to his condition but he didn't even get an elevated ammonia. We are so lucky. They say no one is perfect. Although Jonny has a "spelling mistake" in his genetic makeup, the kid is pretty perfect. Happy AND healthy, within his condition anyways. We are fortunate to have this under control between his medication and his diet. Jonny is a fish, loves to swim. He has played soccer, basketball, baseball, karate, skating. You name it, he has done it. He has travelled to Punta Cana twice and he is obsessed with trains, monster trucks and cars. He lives his life to the fullest. We welcomed our second son when Jonny turned 3. Charlie was tested in his first couple days of life and we discovered that he does not have OTC.

Jonny just started school this year and has been doing outstanding. He takes the bus and loves it. He has already made so many friends.

Featured This Issue

Ornithine Transcarbamylase (OTC) Deficiency (Urea Cycle Disorder)

Compiled by Dr. Chitra Prasad

Ornithine transcarbamylase deficiency is an inherited metabolic disorder that causes ammonia to accumulate in the blood. Ammonia, which is formed when proteins are broken down in the body, is toxic if the levels become too high. The nervous system is especially sensitive to the effects of excess ammonia.

OTC deficiency can become evident at any age. The most severe form occurs in the first few days of life. This neonatal-onset form of the disorder usually affects males. An infant with the neonatal-onset form of OTC deficiency may be lacking in energy (lethargic) or unwilling to eat, and have a poorly-controlled breathing rate or body temperature. Infants with this disorder may be described as "floppy" and can experience seizures or coma. Complications from OTC deficiency if untreated may include developmental delay and intellectual disability. Progressive liver damage may also occur.

In some affected individuals, signs and symptoms of OTC deficiency may be less severe, and may not appear until later in life. The late-onset form of the disorder occurs in both males and females. People with late-onset OTC deficiency may experience episodes of altered mental status, such as delirium, erratic behavior, or a reduced level of consciousness. Headaches, vomiting, aversion to protein foods, and seizures can also occur in this form of the disorder. For all individuals with OTC deficiency, typical neuropsychological complications may include developmental delay, learning disabilities, intellectual disability, attention deficit hyperactivity disorder (ADHD), and executive function deficits.

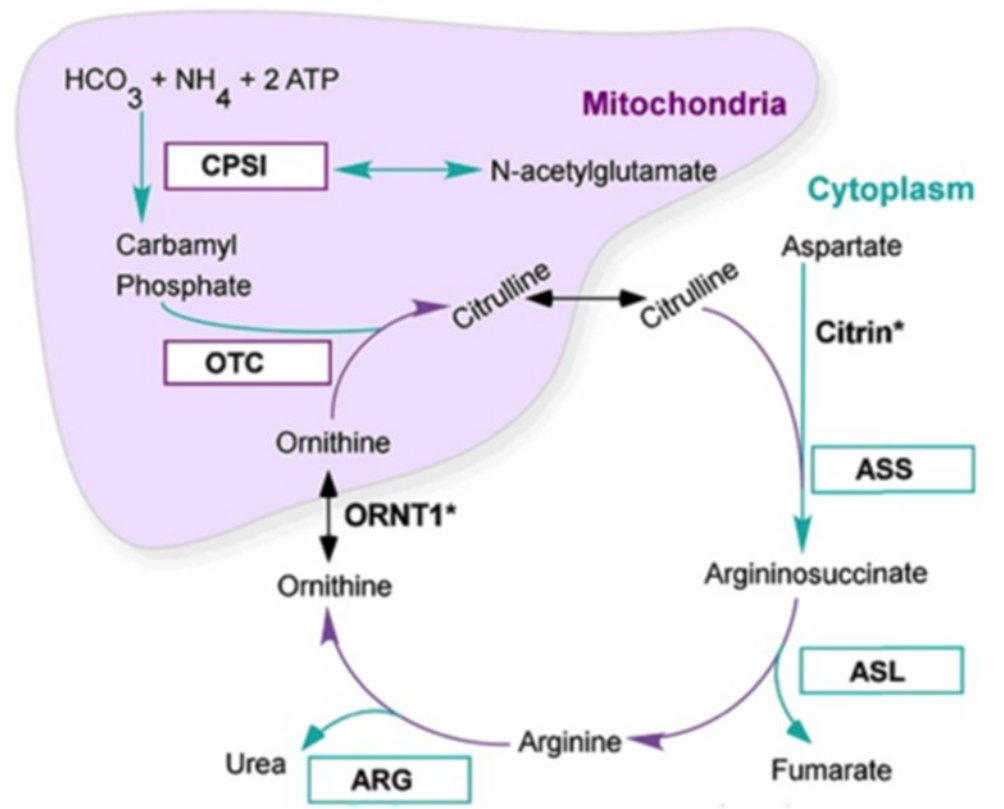
OTC deficiency is inherited in an X-linked manner. If an affected male reproduces, none of his sons will be affected and all of his daughters will inherit the pathogenic variant. Females who are carriers of the pathogenic variant have a 50% chance of transmitting the pathogenic variant with each pregnancy: males who inherit the pathogenic variant will be affected; females who inherit the pathogenic variant may or may not develop clinical findings related to the disorder.

Diagnosis is made by biochemical and molecular studies (plasma amino acids and urine for orotic acid which may be elevated). Mutation testing detects 60-80% of mutations. Previously, liver biopsies were performed for confirmatory diagnosis; however, now we use blood DNA to assess for pathogenic variants.

Management includes a specific diet with restriction of protein under the supervision of a metabolic dietitian. Nitrogen scavenger therapy is used (sodium phenylbutyrate (Pheburane/Ravicti)), as well as citrulline. Regular monitoring in the metabolic clinic is an essential part of management. Liver transplant has been done for severe forms of OTC deficiency, especially neonatal forms. This has been successful in some cases. Liver transplant requires proper assessment, selection, and post-transplant management of each case.

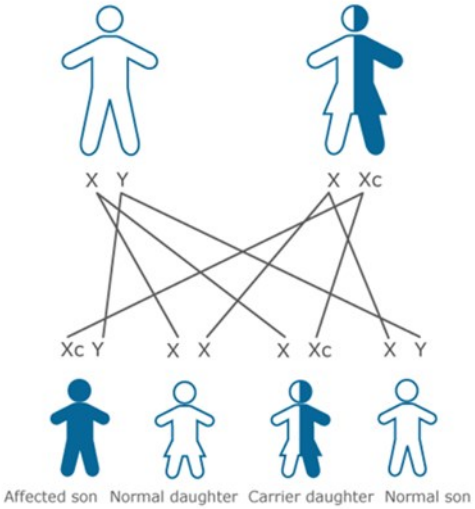
References

https://www.ncbi.nlm.nih.gov/books/NBK154378/
https://ghr.nlm.nih.gov/condition/ornithine-transcarbamylase-deficiency



Urea Cycle

https://faisal8670.wordpress.com/2016/08/23/1957/



X Linked Inheritance
https://www.rch.org.au/genmed/clinical_resources/Inheritance_Patterns/

Suzanne's Corner

Brooke`s Favorite Bread Recipe
SERVING SIZE 50 g (1 slice)
SERVINGS PER RECIPE 20

Ingredients

- 1 3/4 cups plus 2 Tbs (420g) of warm water
- 2 Tbs (23g) sugar
- 3 tsp (9g) active dry yeast
- 4 1/4 cups (packed) (518g) CBF Baking Mix
- 1/4 cup (40g) Metamucil®
- 1 1/4 tsp (9g) salt

Directions

- Add all ingredients to bread machine pan and follow the manufacturer's instructions for your bread maker

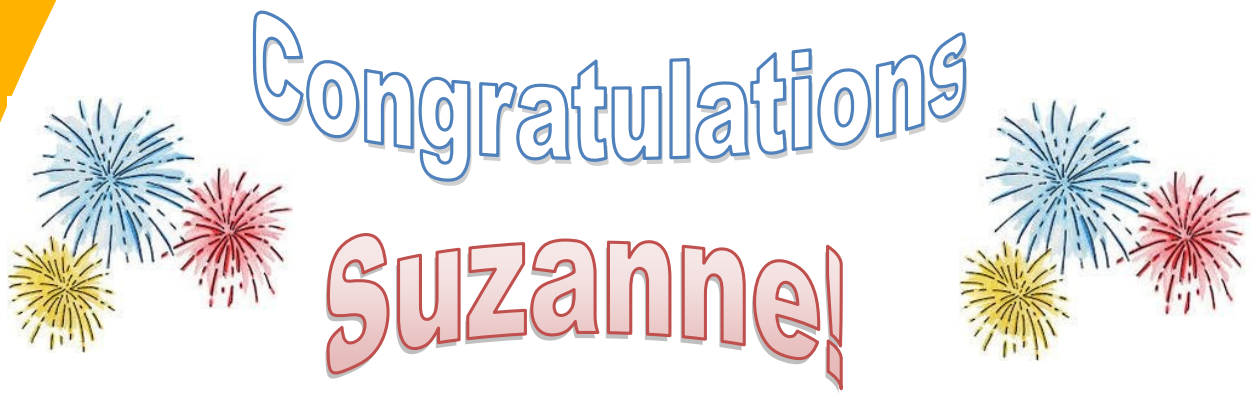
- FOR BEST RESULTS: Pour water into bread machine pan; add sugar, then yeast. Stir briefly, and let sit for 10 minutes (mixture should develop a creamy foam on top). Add the remaining ingredients and follow directions for bread maker. Yields a 2 lb. loaf

- TIP: Add 1/2 teaspoon vitamin C to dough! The literature suggests adding ascorbic acid (powdered Vitamin C) to the dry ingredients in bread recipes. The `C` may interfere with the yeast and sugar during the beginning stages. ASCORBIC ACID (Vitamin C) has the ability to strengthen weak flours and is routinely used by commercial bakers. It creates an acidic environment for yeast which helps the yeast work better. It allows the yeast to react quickly by strengthening the protein structure and enabling the dough to trap the carbon dioxide which is also produced more effectively. This causes the dough to rise faster. If you can`t find pure ascorbic acid crystals, you can crush a vitamin C tablet, but measure accordingly

INSTRUCTIONS FOR MAKING BREAD WITHOUT A BREAD MACHINE: Mix the warm water, sugar and yeast together with a wooden spoon and let stand for 5 or 10 minutes. Add and stir in remaining ingredients until the dough stiffens. Add a Tbsp of baking mix at a time if dough is too sticky to handle. Dump the dough out onto a floured surface and knead until the smooth and slightly resilient. Grease a big bowl (about 2-3 qt.) Lubricate dough by rolling around in the greased bowl. Cover with a cloth or buttered Saran wrap and put someplace warm to rise until nearly doubled in size, about 45 -60 minutes. When almost doubled, form a loaf by shaping then put in a greased loaf pan or on a greased baking sheet. Let rise a second time until almost doubled in size and place into a pre-heated 350 degrees F oven and bake until golden brown, about 35-45 minutes. Turn bread out onto a rack to cool.

Nutrition

	Per Recipe	Per Serving
Protein, g	6.23	0.31
Calories	2300	110



Congratulations Suzanne on being nominated for the 2018 President's Award! We are so proud of you and all of the incredible work you do!

Low Protein Foods Update

Products that have been discontinued by the manufacturer are listed below:

Applied Nutrition Homestyle Blueberry Muffin Mix
Applied Nutrition Cinnamon Chip flavour Muffin Mix
Applied Nutrition Homestyle Sugar Cookie Mix
Applied Nutrition Homestyle Yellow Cake Mix
Apoten Low protein bread
Cambrooke Foods – Tomato Tortilla Wraps
Country Sunrise Soft Tortillas
DS Tri-coloured shells
DS Herb & Garlic Ziti
Homestyle Fudge Brownie Mix
Juvela Low Protein Mix (Nutricia)
Kingsmill Vacuum Packed Unimix Bread 550 g
Kingsmill Unimix All Purpose Baking Mix
Loprofin Pasta: vermicelli 250 g
Milupa lp Bar
PKU Perspectives – Sandwichmate Imitation Cheese Slices
PKU Perspectives – Shredmate Imitation Shredded Mozzarella Cheese
Wel-Plan Baking Mix 400 g

<u>Other Supplements</u>
Flavour Pacs – Orange
Flavour Pacs - Lemon
Flavour Pacs - Raspberry
Flavour Pacs – Black Currant
Flavour Pacs –Tropical flavour

Jeffrey McManaman at our 14th Annual Low Protein Cooking Demo – May 2018

Research/Publications

- Aref-Eshghi E, Bend EG, Hood RL, Schenkel LC, Carere DA, Chakrabarti R, Nagamani SCS, Cheung SW, Campeau PM, **Prasad C**, Siu VM, Brady L, Tarnopolsky MA, Callen DJ, Innes AM, White SM, Meschino WS, Shuen AY, Paré G, Bulman DE, Ainsworth PJ, Lin H, Rodenhiser DI, Hennekam RC, Boycott KM, Schwartz CE, Sadikovic B.BAFopathies' DNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffin-Siris and Nicolaides-Baraitser syndromes. Nat Commun. 2018 Nov 20;9(1):4885.

- Remtulla S, Emilie Nguyen CT, **Prasad C**, Campbell C.Pediatr Neurol. 2018 Aug 9. 018.08.007. [Epub ahead of print].Twinkle-Associated Mitochondrial DNA Depletion.

- Wang PZT, **Prasad C**, Rodriguez Cuellar CI, Filler G. Nephrological and urological complications of homozygous c.974G>A (p.Arg325Gln) OSGEP mutations. Pediatr Nephrol. 2018 Nov;33(11):2201-2204. doi: 10.1007/s00467-018-4060-x. Epub 2018 Aug 23.

- Zapata-Aldana E, Kim DD, Remtulla S, **Prasad C**, Nguyen CT, Campbell C. Further delineation of TBCK - Infantile hypotonia with psychomotor retardation and characteristic facies type 3. Eur J Med Genet. 2018 Aug 11. pii: S1769-7212(18)30002-8. doi: 10.1016/j.ejmg.2018.08.004. [Epub ahead of print]

- Datar R, Prasad AN, Tay KY, **Rupar CA**, Ohorodnyk P, Miller M, **Prasad C**. Magnetic resonance imaging in the diagnosis of white matter signal abnormalities. Neuroradiol J. 2018 Aug;31(4):362-371. doi: 10.1177/1971400918764016. Epub 2018 Mar 8.

- O'Connor K, Jukes T, Goobie S, **DiRaimo J,** Moran G[1], Potter BK, Chakraborty P, **Rupar CA**, **Gannavarapu S**, **Prasad C**. Psychosocial impact on mothers receiving expanded newborn screening results. Eur J Hum Genet. 2018 Apr;26(4):477-484. doi: 10.1038/s41431-017-0069-z. Epub 2018 Jan 29.

- Brady L, Sadikovic B, **Rupar CA,** Tarnopolsky MA. Complete elimination of a pathogenic homoplasmic mtDNA mutation in one generation.Mitochondrion. 2018 Feb 1. pii: S1567-7249(17)30328-8. doi: 10.1016/j.mito.2018.01.010. [Epub ahead of print]

- Inborn Errors of Metabolism in Newcomer and Refugee Populations in Ontario, CA. **Srinitya Gannavarapu*, Madhavi Prasad* and Chitra Prasad** **contributing equally (Submitted to University of Western Ontario Medical Journal)*

Presentations

- Role of Clinicians in the Multigene panel/Exomic/Genomic era. **By Chitra Prasad.** Presented to Department of Paediatrics in New Delhi India March 2018.
- Failure to Thrive, Short Stature and Abnormal Bones: Recent Advances in Diagnosis and Management of Hypophosphatasia Presented at Hospital of Sick Children Toronto Paediatric Update. April 2018
- **Chitra Prasad**. Kidney disease and Organ Transplantation in Methylmalonic Acidaemia Damien Noone, Magdalena Riedl, **Suzanne Ratko,** Yaron Avitzur, Ajay P Sharma, Guido Filler, Komudi Siriwardena Children's Hospital
  Oral presentation at Garrod Association meeting May 2018 in Edmonton
- Homocysteine Metabolism and Clinical Disorders: the long and short of it By **Dr. Chitra Prasad** (Presentation in St. Johns and Halifax Nova Scotia) June 2018
- Acid Lipase Deficiency: from Diagnosis to Therapy in Canada
  Prasad C, Dhandapani A, **Colaiacovo S** and **Rupar CA**
  Presented at the 1st Canadian Lysosomal meeting in Sherbrook Quebec, September 2018

Highlights from Melanie: Canadian Association of Genetic Counsellors Annual Education Conference

Even though I am currently on secondment, I had the ability to attend a Genetic Counselling conference this year. I attended the Canadian Association of Genetic Counsellors Annual Education Conference in St. John's, Newfoundland. I was able to speak with colleagues from across the country and learn from their experiences. Some of the highlights include:

-Newborn Screening Workshop
-Strategies for Rare Disease Diagnosis
-Ethical Considerations in Genetic Counselling
-Overview of Treatment for Genetic Diseases

I am looking forward to coming back to the clinic in March. I am hoping I can implement some of this new information into my practice. I am also looking forward to seeing and working with all of the wonderful individuals and families that receive care through our clinic.

Keep warm this winter! See you in 2019!
Melanie Napier
Metabolic Genetic Counsellor

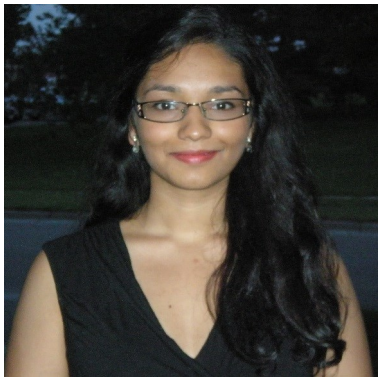
Student Projects

Project: Inborn Errors of Metabolism in Newcomer and Refugee Populations in Ontario, CA
Researcher(s): Madhavi Prasad, Srinitya Gannavarapu and Chitra Prasad
Summary: In Canada, newborn screening programs allow early detection of many inborn errors of metabolism, drastically improving the overall outcomes of affected children. However, many countries in the world do not have the resources to support such programs. With Canada's expanding population of refugees, the detection and diagnosis of these conditions becomes more challenging given that children start developing symptoms. In our work, we aimed to discuss treatment and management needs for these vulnerable populations, to better understand the gaps in our understanding of the cultural and psychological impacts related to migrant populations.

Project: Psychological Aspects and Stressors for Parents of Children with Inborn Errors of Metabolism
Researcher(s): Prashanth Rajasekar, Srinitya Gannavarapu, Melanie Napier, Asuri N. Prasad, Akshay Vasudev, Andrew Mantulak, Beth K. Potter and **Chitra Prasad**
Summary: Diagnosis of an inborn error of metabolism (IEM), the associated frequent hospitalizations and medical intervention impact psychosocial functioning in parents. To better understand the psychosocial stressors and strategies, we interviewed parents of children with IEMs over the course of four years. We aimed to better understand specific resources and support systems they sought and gain perspective on how their tumultuous journey impacted their daily lives and overall life choices.

Project: Gene Discovery in a Complex Neurological Disease
Researcher(s): Srinitya Gannavarapu, Tony Rupar, Asuri N. Prasad, **Tugce B. Balci and Chitra Prasad**
Summary: Receiving a clear diagnosis for a disease is often very important for the affected individual and family, as it provides a sense of context for the psychological and emotional experiences they go through. The Care For Rare Research program has attempted to give a definitive diagnosis to numerous complex, rare diseases through the use of whole exome sequencing. As part of the same initiative, we are working on identifying the genetic basis of a unique, progressive, degenerating neurological disease that was seen here at LHSC over the past decade.

Prashanth Rajasekar



Srinitya Gannavarapu

What's New

Staff Updates

Dr. Tugce Balci

Dr. Tugce Balci joined the Genetics team in September 2018. She completed her residency training in Ottawa where she participated in research, specifically within the Care4Rare initiative, a pan-Canada research program aimed at providing care to families with rare diseases. She is currently building her research program here in London. She aims to explore new ways of diagnosing patients with unsolved genetic disorders using new genomic technologies and characterizing novel genes using the zebrafish model.

Andrea Djolovic

I have been working as a Genetic Counsellor since 2016 and I have been lucky to call the Windsor Genetics Outreach Clinic of LHSC my home since July 2017. Our Windsor Genetics Clinic has a new location based at the John McGivney Children's Centre where we run monthly full day clinics with geneticists. We are very fortunate to have physicians from London Health Sciences seeing patients from the Windsor-Essex area locally. The clinic's main focus has been in general, pediatric and prenatal genetics. Recently we started seeing metabolic patients alongside Dr. Chitra Prasad and Dr. Natalya Karp. In the future we are really looking forward to having Dr. Andrea Yu seeing Windsor metabolic patients too!

Dr. Andrea Yu

I joined LHSC in November as a new Medical Geneticist who will be involved in General Genetics, Metabolics and Newborn Screening. I completed my residency training in Medical Genetics and Genomics at the University of Ottawa. I recently finished a Clinical Biochemical Genetics Fellowship at the Children's Hospital of Eastern Ontario. I look forward to being a part of the LHSC team!

Sarah Muto

Sarah's health care career at LHSC spans over 12 years, and her educational background includes a combined honours Bachelor of Arts degree in English and Political Science from the University of Western Ontario, and a post-graduate certification in Public Relations from Toronto's Humber College. Sarah has been Coordinator of Medical Genetics and The Fertility Clinic since October 2017. Prior to this role, Sarah was a project consultant with the Clinical Redesign team at LHSC where she led a number of large -scale clinical projects, including; the pay for results program, medicine discharge project, quality based procedures within surgery, medicine, women's and children's, and most recently the mental health/ED patient access projects. Prior to her work as project consultant, Sarah was a Communications Consultant with Corporate Communications & Public Relations.

Meghan Zadorsky

After completing my Bachelor's degree in Genetics at Western University, I began volunteering in the Genetics Clinic in May 2018 working mostly with cancer genetics. During this time I learned so much and began to get to know the incredible people that make up the genetics team. In September 2018 I had the opportunity to join the team full time as a Genetics Assistant. I love my new role as I have the opportunity to learn something new each day, and to be surrounded by all of these brilliant minds! Outside of work I enjoy volleyball and volunteering for various organizations including most recently, becoming a youth mentor with the Huntington's Society.

Dorothy Michalski

I obtained both my Master's degree in Cell & Molecular Biology and Bachelor's degree in Genetics and Biochemistry from the University of Western Ontario. While completing my Master's degree I began volunteering in the Genetics Clinic, and upon graduating was hired on as a Genetics Assistant. I have been thoroughly enjoying the first few months in my new role and continue to learn so much from both patients and the dedicated staff in the clinic! Outside of work I volunteer as a peer-supporter with the Canadian Cancer Society and enjoy swimming, playing tennis, watching horror movies and going for long walks with my dog.

Melanie Napier & Samantha Colaiacovo

We are looking forward to welcoming back Melanie Napier (left) to the genetic counselling team in March 2019. Melanie has been working hard to help ensure the hospital was in tip-top shape for accreditation. A very special thanks to Samantha Colaiacovo (right), who has been providing spectacular genetic counselling for our Metabolic Genetics Clinic's patients and families, especially in Melanie's absence!

Welcome back Rana!

We are very excited to welcome back Rana who returned from her maternity leave in September!

A Word from Scott Fortnum, President and CEO of Children's Health Foundation

The Metabolic Family Workshop is free for families, year-after-year, because of the caring generosity of donors to Children's Health Foundation. Our community is proud to support you and your loved ones as you face the challenges presented by a metabolic disorder. You too can become a donor! Please consider making a gift at childhealth.ca *or text CHILD to 41010.* Children's Health Foundation is the fundraising for Children's Hospital at London Health Sciences Centre, Thames Valley Children's Centre and Children's Health Research Institute.

Children's Hospital
London Health Sciences Centre

Metabolic Family Workshop

Friday, May 3, 2019

Best Western Lamplighter

591 Wellington Road London, ON

"You are not your disease"

Low Protein Cooking Demonstration

Saturday, May 4, 2019

Real Canadian Superstore

825 Oxford St E London, ON

Metabolic Family Workshop
Friday May 3, 2019

Number Attending: _____

Names: _____

Contact Name: _____

Phone Number: _____

Email: _____

Morning Workshop Attending: (indicate names)

PKU/Diet	MCADD	UREA CYCLE	Other Metabolic Disorders	Lysosomal
_____	_____		_____	_____

Afternoon Workshop Attending: (indicate names)

Preteen (8-13)	Teens (14-20)	Adults (with metabolic disorder)	Parents
_____	_____	_____	_____

Low Protein Cooking Demonstration
Saturday, May 4, 2019

Number Attending: _____ Names: _____

Please return above information to:

rana.elshourafa@lhsc.on.ca **Tel : 519-685-8500 ext 56131**
LHSC Victoria Hospital **Fax: 519.685.8214**
800 Commissioners Rd E London, ON N6A 5W9

Children's Hospital
London Health Sciences Centre

Our Stars

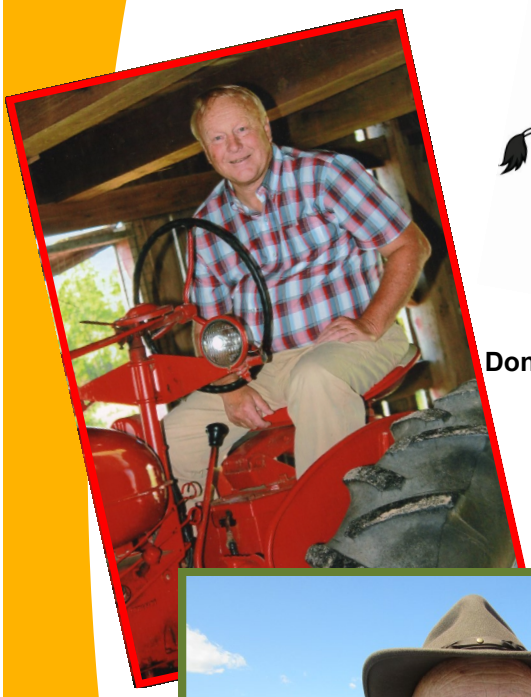
Quach Family as The Wiggles! Clockwise from top right: Vu, Ryder (8), Logan (8), Quorra(1) and Jenny. (A-beta–lipoproteinemia)

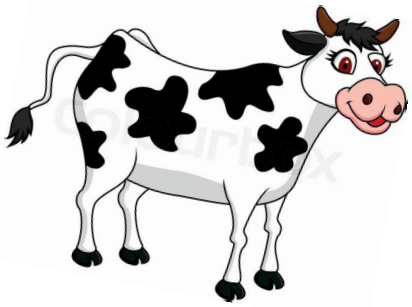
Karson Gervais (8), PKU

Kaitlynn Broad (1), PKU

Michael (9), PKU and Jeffrey McManaman

Karson Gervais (8), PKU





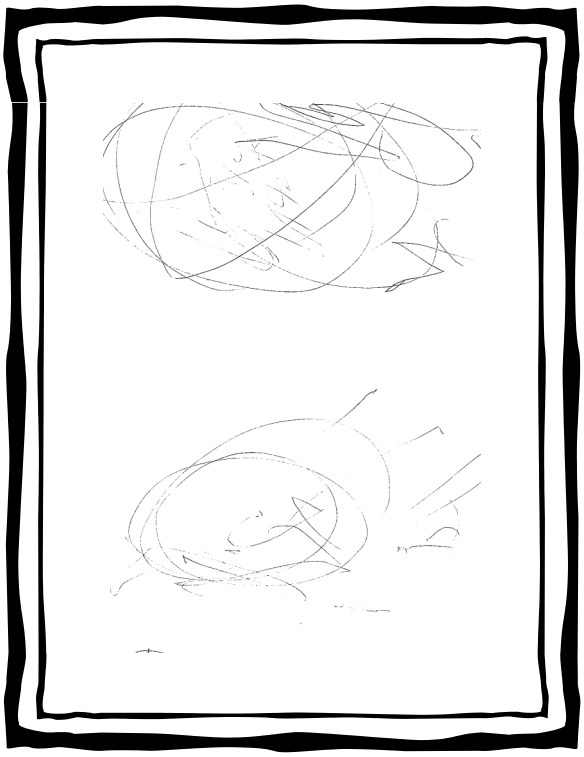
Donald Doan (70), OTC deficiency

"Master Breeder", Donald Doan!
We are so happy for you Donald!

Congratulations!

Inspirations

Evelyn Fehr (4), hypophosphatasia



Abbas Dirani (3), PKU

"Invent your world. Surround yourself with people, color, sounds, and work that nourish you."
– Susan Ariel Rainbow Kennedy

These Feet

These feet have walked a thousand miles, visited across the country of Canada.
Raced with fellow athletes, and bravely conquered mountains.

These feet have lived through pain, sweat and tears. Hiked up a thousand stairs, stepped on sharp rocks, hot sand and waded through cool shallow waters.

These feet have traveled across the world! They are my feet, my memories, I cherish to keep. I have set goals to work hard and achieve them all, for an athlete is a believer and is not a quitter!

Written by: Jacqueline Gravel (34), PKU

STORY Bout Margie
I like to Doing LOTOF WALKING
I like Doing Lot OF Swimming
I like to going the YMCA
I like to PLay BASK BALL
and I like to work in the Bake shop
I am a good Bowling
Iosht to Be in the STeiPL OLYMTICS
I like Da Lot OF singeing in the Chir
I want to Tornto For the steiAL OLymtics
I like Going on Hall day
I like to warch tV
I like to Go no DAY TIPS
Some Time I like to go the CN Towner
in tornto and i like go to the FISh
In tornto
I like to Learn More Riping For PKU
in the Sumer Time I like to go
swimming and sit out in the sun wan it is
warm out sid
I like to Be in
I like Do Lot OF Swimming IN the Summer time
I like Going to Tim Honton some Get some tea
some POP to have
Tray to cut Down on POP

FromYourFrined MARGIE Cartier

for swimming and Bowing
when I was little
New Food

Margie Cartier (60), PKU

We would love to hear from you! To send pictures and stories for future editions of the newsletter or to be added to our email list, please contact meghan.zadorsky@lhsc.on.ca

"An empty lantern provides no light. Self-care is the fuel that allows your light to shine brightly."
– Unknown

"Self-compassion is simply giving the same kindness to ourselves that we would give to others."
– Christopher Germer

Contact Information

Editor: Meghan Zadorsky
LHSC - Medical Genetics Program of Southwestern Ontario
Tel: 1.800.243.8416
1.519.685.8453

Email: meghan.zadorsky@lhsc.on.ca

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!