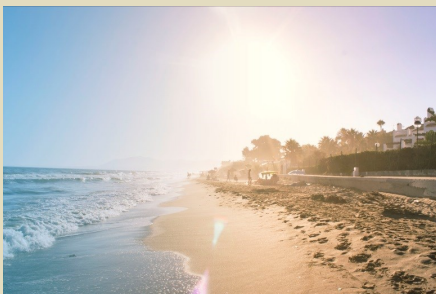


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From the Editors

Our 12th Metabolic Family Workshop and 13th Low Protein Cooking demonstration in May was a huge success! Please see the write-up under *What's New* for the full report and pictures of the events.



A big thank you to Jessica Tao, Summer Research Student, for jumping in and organizing this edition of the newsletter!

Janice Little / Jessica Tao

From Dr. Chitra Prasad

Dear Friends,

Greetings!

It was wonderful to see many of you at the 12th Metabolic Family Workshop in May. We are now holding the workshop every 2 years. I have summarized some of the highlights from this workshop in this newsletter on pages 7-8.



One of the major themes of this workshop was "Resilience" (the capacity to recover quickly from difficulties). This applies to each and every one of us. Mindfulness and "staying in the moment" are some of the powerful tools for building resilience.

Patient and family stories were amazing as always. Thanks to all the speakers, leaders and volunteers who participated in the workshop and breakout sessions.

This is a special issue of the Metabolic Newsletter as we have an assistant editor, Jessica Tao, who is also my research student.

In this newsletter, you will learn about Clara and her parents' journey. Clara had very challenging seizures in the newborn period. Appropriate diagnosis and management have been very helpful for Clara as she is doing remarkably well at 2 years of age. Dr. Narayan Prasad (her neurologist), Dr. Tony Rupar (lab director), Suzanne Ratko (dietitian) and I have had the privilege and honour to be Clara's medical team along with her parents who have tirelessly followed all the treatment plans and done a great job. A strong team and collaboration is most helpful when we are faced with a challenge.

Hope you enjoy the remaining summer and beautiful fall!

With best wishes,
Chitra Prasad

Personal Stories

Clara's Journey Living with Pyridoxine-Dependent Epilepsy (PDE)



When unexpected sicknesses happen to anyone, it's scary and the unknown about the sickness is always the worst. When an unexpected illness happened to my newborn daughter, I had never been so scared about anything in my life. Clara was born on July 23, 2015, seemingly happy and healthy. Another gorgeous baby to complete our family. My husband and I were so excited to have a baby girl who would be a sister to our now 5-year-old son. Everything seemed okay for the first 10 days of her life at home. Then, on her eleventh day of life, just after her afternoon nap, the fear all parents have about their children happened. Clara was sick. She had two seizures at home that day, both in my arms, as I sat there helpless to fix her. We called the ambulance and were taken to the hospital in Samia where we live, and were admitted overnight. It wasn't until the early hours of the next morning when she had another seizure, and then another, and then they would continue on throughout the day. We were sent to the Children's Hospital in London where we would spend the next month of her life.

As my husband and I followed the ambulance to London, the fear that something severe was wrong with our perfect little baby ran through our minds. Clara proceeded to have many more seizures daily over the next week. These seizures would last longer than five minutes and need rescue medications that thankfully would stop the seizure. Between the seizures and rescue medications though, Clara's little body would be extremely tired and she would sleep for hours. We would have to wake her up for feedings, which in her state did not go well and she lost a lot of weight. I remember crying at night when they had to give her a feeding tube, feeling so terrible that I couldn't get her to eat.

Clara's Neurology and Metabolic team ran many tests on her over the first few weeks we were there to try to find out why the anti-seizure medications were not working. MRIs, EEGs, urine samples and blood work. My heart broke every time she had to get a needle for blood work or when they had to put an IV in. You never feel as helpless as you do when your baby is sick and there is nothing you can do to make her better. Clara was beginning to experience delays with her development and muscle tone, and we were told that because of all the seizures, she may be delayed in life as she grew up.

Ultimately, after genetic testing, we finally got an answer about the cause of Clara's seizures. When we got the results that Clara had pyridoxine dependent epilepsy (PDE), things began to change for the positive for Clara. We started her on B6 pyridoxine, arginine and left her on an anti-seizure medication to be safe. Finally, the seizures stopped! Her type of epilepsy requires taking B6 and arginine as well as a lysine (protein) restricted diet. We were able to go home and start a life as normal as possible with Clara. We started going for physiotherapy to help with Clara's development and muscle control.

Personal Stories (continued)



Over the next few months, she began to gain weight and develop as well. The therapy was a struggle at first; she wasn't where she needed to be for her age. We did lots of activities at home that the therapist suggested and worked continuously with her to get her caught up. Slowly, she started to catch up and excel. By the time Clara turned one, she was on track with her progress.

We started Clara on her lysine-restricted diet which included Amino Acid modified formula that we have to order and get sent to our home. We had a strict feeding schedule. As Clara got older and when we could start her on solids, new challenges emerged. Now, not only did we have to measure all her formula and ensure that the amount we set was consumed each day, we added in measuring food too. With the protein that she gets from her formula, we could

only allow so much protein or lysine per day too. It was a lot of trial and error, and just trying new things all the time. Together with our dietitian, we made it through the last two years.

It's unbelievable to my family the support and constant help we have received from the London Health Sciences Centre Children's Hospital. There were times when I was calling Clara's dietitian every few days for advice and she was always receptive and ready to help.

Now at two years old, I cannot believe that Clara is the same child that I just told you about. She is so determined, strong and independent. She honestly amazes me every day. She is vibrant and happy and learning all the time. She is completely developed for her age (something that I, at one time, didn't know would be possible). Clara has come so far from a baby who could barely hold her head up to a toddler who can do everything she should be doing for her age. I mean everything: she likes to get into everything—climb, jump and run around—but we wouldn't have her any other way. We go for blood work monthly to check her levels and ensure her diet is working. We continue to try new things with her diet and we are having great success with introducing new foods to her.



The first year of Clara's life was hard, but now looking back, it makes me realize that my family can get through anything. PDE isn't just something Clara has to live with for the rest of her life; it's a part of us, all of us. We embrace it, and her diet and medications are just a normal part of life for us now. I hope that any other family going through something like this can take our story and know that there is hope and that all the medications, restricted diets, tests, blood work and everything else that comes with it will someday become your normal, and it's okay to be scared at first because if you stick together, you can do it.

Thanks,
Martha Hiscocks
The proud mom of Clara Hiscocks

Featured This Issue

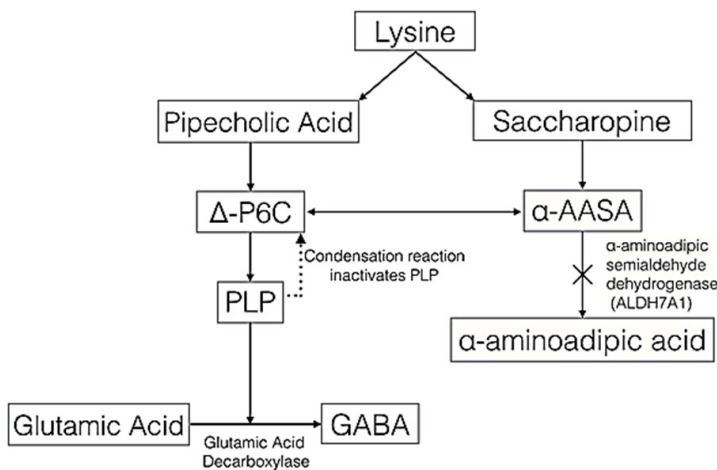
Pyridoxine-Dependent Epilepsy Compiled by Dr. Chitra Prasad

Pyridoxine-dependent epilepsy is a condition that involves seizures beginning in infancy or, in some cases, before birth. Those affected typically experience prolonged seizures lasting several minutes (status epilepticus). These seizures involve muscle rigidity, convulsions, and loss of consciousness (tonic-clonic seizures). Additional features of pyridoxine-dependent epilepsy include low body temperature (hypothermia), poor muscle tone (dystonia) soon after birth, and irritability before a seizure episode. In rare instances, children with this condition do not have seizures until they are 1 to 3 years old.

Anticonvulsant drugs, which are usually given to control seizures, are ineffective in people with pyridoxine-dependent epilepsy. Instead, people with this type of seizure are medically treated with large daily doses of pyridoxine (a type of vitamin B6 found in food). If left untreated, people with this condition can develop severe brain dysfunction (encephalopathy). Even though seizures can be controlled with pyridoxine, neurological problems such as developmental delay and learning disorders may still occur.

Mutations in the *ALDH7A1* gene cause pyridoxine-dependent epilepsy. The *ALDH7A1* gene provides instructions for making an enzyme called α -aminoadipic semialdehyde (α -AASA) dehydrogenase, also known as antiquitin. This enzyme is involved in the breakdown of the protein building block lysine in the brain.

When antiquitin is deficient, a molecule that interferes with vitamin B6 function builds up in various tissues.



Δ -P6C: Δ -piperidine-6-carboxylate
PLP: pyridoxyl-5'-phosphate
 α -AASA: α -aminoadipic semialdehyde

Figure 1. Pathophysiology of refractory seizures in PDE. α -AASA dehydrogenase deficiency and the cerebral lysine degradation pathway. Adapted from: *Pyridoxine-Dependent Epilepsy: An Expanding Clinical Spectrum*. van Karnebeek, et al. (1)

Pyridoxine plays a role in many processes in the body, such as the breakdown of amino acids and the production of neurotransmitters, chemicals that transmit signals in the brain. It is unclear how a lack of pyridoxine causes the seizures that are characteristic of this condition.

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. Both parents of an individual with an autosomal recessive condition carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Management includes lifelong high doses of pyridoxine, lysine-restricted diet and arginine in some instances. Ongoing neurodevelopmental assessment is essential.

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2. <https://ghr.nlm.nih.gov/condition/pyridoxine-dependent-epilepsy> (Genetics Home Reference)
3. van Karnebeek CDM, Tiebout SA, Niermeijer J, Poll-The BT, Ghani A, Coughlin CR, Van Hove JLK, Richter JW, Christen HJ, Gallagher R, Hartmann H, Stockler-Ipsiroglu S. Pyridoxine-Dependent Epilepsy: An Expanding Clinical Spectrum. *Pediatric Neurology* 2016; 59: 6-12.

Featured This Issue

Pyridoxine-dependent epilepsy (PDE) (Neurologist's Perspective)

By Dr. Narayan Prasad



PDE is a rare inherited disorder that affects metabolic pathways involving the degradation of the amino acid lysine where the Vitamin B6 (Pyridoxine) is involved somewhat indirectly. The condition was first described in 1954 in an infant with treatment-resistant seizures that responded to a multivitamin cocktail that contained Vitamin B6. Some 6 decades later, a biochemical marker (pipecolic acid) was found to be consistently identified in the cerebrospinal fluid.

In 2006, the mystery was solved as a genetic basis was established following the identification of pathogenic mutations affecting a gene (*ALDH7A 1*) named *Antiquitin*. Mutations affecting the gene product lead to the accumulation of an intermediate chemical compound, alpha amino adipic semialdehyde, that inactivates the active form of Pyridoxine (pyridoxal phosphate). Consequently, several secondary deficits appear in the biosynthesis of neurotransmitters in the brain, eventually leading to the manifestations of PDE.

PDE presents in early life with a wide variety of seizures that are often treatment-resistant. At times, the diagnosis may be delayed due to presentation mimicking more common conditions such as sepsis. Long term outcomes include; intractable and treatment-resistant epileptic seizures, and developmental delay affecting all aspects of development, motor, cognitive, language and communication. Outcome is dependent on many factors including; age at diagnosis, the age at which specific replacement therapy was introduced, presence of associated brain abnormalities, etc. Treatment of this condition involves administration of pyridoxine or pyridoxal phosphate, and folinic acid in appropriate doses, as well as dietary modifications designed to reduce the accumulation of toxic intermediary compounds through lysine restriction. Early diagnosis through detection of AASA in the urine and cerebrospinal fluid, and identification of pathogenic mutations in the *Antiquitin* gene using molecular genetic testing has been a major breakthrough. Treatment-resistant seizures presenting in the neonatal period and early childhood should be given a trial with pyridoxine and folinic acid while diagnostic testing is pursued to rule out this rare disorder. With proper diagnosis of PDE, a number of other anticonvulsants can be reduced or stopped altogether.

Dr. Richa Agnihotri, Senior Pediatric Resident



I had the pleasure of being involved in Clara's care during her admission to hospital as a neonate, and following her story in the Genetics and Metabolics follow-up clinic with Dr. Chitra Prasad when she was 12 months of age. She was a bright and delightful, appropriately growing and developing child!

Her story is both inspirational and educational, as she has done so well following an early diagnosis and start of appropriate therapy, which was practiced diligently by her family and healthcare team.

I have worked along with Dr. Chitra Prasad and Dr. Narayan Prasad to write up her case to raise awareness among health professionals, of the condition and the appropriate early management. It raises a key differential diagnosis for us to consider as pediatricians dealing with seizures in the neonate, and shows the positive impact a timely diagnosis and therapy can have. This was presented to the Genetics and Metabolics Department, Paediatrics Department and as a Poster Presentation at Paediatrics Research Day in 2017. I am also in the process of submitting the case for publication in a journal to reach a wider audience.

The Inherited Metabolic Disorders News

What's New

Our Staff



Thanks, Sam!

Thank you to our Genetic Counsellor, Samantha Colaiacovo, for all your help with Metabolics & Newborn Screening! Don't worry, Sam will be staying with us at Medical Genetics, LHSC!

Welcome Back Melanie!

Melanie Napier, Genetic Counsellor, is returning this fall from her maternity leave. The Metabolics Team has missed her greatly!



Welcome Dr. Maha Saleh, Geneticist (in place of Dr. Sharan Goobie)!



I have recently completed my residency training in Medical Genetics at the University of Toronto. Prior to residency, I was a research student at SickKids. I had the privilege of working with Dr. Joe Clarke and Julian Raiman on Lysosomal Storage Diseases. I have developed an interest in Education for which I received the Joe Clarke teaching awards; I have helped revamp the undergraduate and postgraduate genetics teaching curriculum, and completed a Stepping Stones Teacher Development Program. Last year, upon completion of my training, I obtained my Fellowship in Pediatric Simulation. I will be covering Genetics and Newborn Screening, and look forward to working at LHSC.

Welcome Back Dr. Karp!

Dr. Natalya Karp, Geneticist, is also returning this fall from her maternity leave. We are glad to have her back!



What's New (continued)

Metabolic Family Workshop 2017



Some of the organizing committee and special guests of the Metabolic Family Workshop:
(L-R) Janice Little, Rana El-Shourafa, Jo-Anne Psiuk-Rodgers, Karen Groeneweg, Erika Clements, Dr. Bhooma Bhayana, Dr. Tony Rupar, Dr. Chitra Prasad, Dr. Rana Chakrabarti, Samantha Colaiacovo, Dr. Paoyun Tang, Sue MacLean

The 12th Metabolic Family Workshop took place on May 12th, 2017 at the Lamplighter Inn & Conference Centre. The Low Protein Cooking demonstration was held on May 13th, 2017. There were close to 175 registrants (including patients, families, friends, relatives, professionals, sponsors, service groups and medical teams). Dr. Chitra Prasad - Director of Clinical Metabolic Services, Children's Hospital at London Health Sciences Centre (LHSC), introduced Jackie Schleifer Taylor - Vice President Children's Hospital and Women's Care, who gave very inspiring welcoming remarks. Jackie had the opportunity to meet with many families and sponsors. She brought the message of hope to our families amidst all their challenges. Dr. Bhooma Bhayana, who is a part of the Family Medicine Group in London, was our main scientific speaker. Through interesting anecdotes, stories and real-life situations, Dr. Bhayana spoke about "Navigating the course; can your family doctor be your first mate on this journey?". Many of our patients have complex metabolic disorders—having a family physician who can understand their ongoing issues and also ensure that the rest of their health care is looked after would be truly ideal.

Geneviève Lafrance - Metabolic Dietitian, Fleurimont Hospital, l'Université de Sherbrooke, led the discussions about phenylketonuria (PKU) and diet (PKU group). Families have always appreciated advice from experts. She was ably helped by Suzanne Ratko - Registered Dietician, Children's Hospital at LHSC, and Amanda McManaman - mother of a child with PKU. This year, we had a new breakout session on MCADD (Medium-chain acyl-CoA dehydrogenase deficiency), a metabolic disorder in the pathway of fat metabolism. Jill Sangha - Patient & Family Centred Care Specialist, Children's Hospital at LHSC, April Willison - adult with MCADD, and Samantha Colaiacovo - Genetic Counsellor, Metabolics, Children's Hospital at LHSC, were leading this workshop. April has also spoken about her experiences with MCADD in previous workshops—her input was most valuable.

We had a workshop on coping strategies for parents who have children with inborn errors of metabolism (IEM). Dr. Chitra Prasad, Darren Connolly - Family Resource Centre Advisor, Children's Hospital at LHSC, and Sue MacLean - Social Worker, Children's Hospital at LHSC, shared some useful strategies.

Lysosomal Storage Disorders were another breakout session. Dr. Tony Rupar - Biochemical Genetics Laboratory Director, LHSC, has led this group for many years. His expertise and knowledge about this group of conditions and many other metabolic conditions is always much appreciated. Dr. Rana Chakrabarti - Research Assistant, LHSC, also participated in this workshop. He also helped with the stretch break.

The Inherited Metabolic Disorders News

What's New (continued)



Dr. Jonathan Mahn

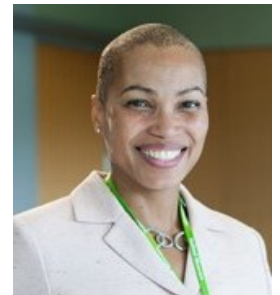
The family presentations included Dr. Jonathan Mahn - adult patient with PKU, Heather Aitken - adult patient with Mitochondrial neurogastrointestinal encephalopathy syndrome (MNGIE), and Lisa Gearey - mother of a child with lysosomal acid lipase deficiency (LAL-D). The three wonderful speakers shared their insights, day-to-day challenges and how to overcome these.

Dr. Akshya Vasudev - Psychiatrist, LHSC, led the adult metabolic group and the entire group in a discussion about strategies for resilience. Dr. Vasudev brings the knowledge of psychiatry and his interest in ancient philosophies, meditation as his expertise.

We were very privileged to have Child Life Therapy and Art Therapy for our younger patients and their family members. Through imaginative and innovative techniques, our specialists kept the children and adolescents engaged while providing some interesting education. Mind Over Matter was the theme for the preteens. Erika Clements and Karen Groeneweg are the Child Life Specialists, and Todd Wharton and Gill Yealland are the Art Therapists at Children's Hospital at LHSC.



Dr. Bhooma Bhayana



Jackie Schleifer Taylor

Tracy Loosemore - Chief Operating Officer, Children's Health Foundation, LHSC, addressed the Metabolic Family Workshop after lunch and brought greetings from the Foundation. The Children's Health Foundation has given us strong support from the time we started this journey.

Over the years, the Lamplighter Inn has provided a beautiful venue and lunch (including low-protein options).

My heartfelt thanks to our volunteers who take care of the young children, and Jennifer Culp (PKU mother) who is always there to set up the daycare room for families, sponsors, families and service groups.

The Metabolic Family Workshop has become a unique experience for the families and professionals alike.

Hope to see you all in 2019!

Respectfully submitted,

Dr. Chitra Prasad

(On behalf of the Metabolic Team: Suzanne Ratko - Registered Dietitian, Janice Little - Resource Associate, Rana El-Shourafa - Administrative Assistant, Jo-Anne Psiuk-Rodgers - Administrative Assistant, Samantha Colaiacovo - Genetic Counsellor, Dr. Rana Chakrabarti - Research Assistant, Dr. Tony Rupar - Biochemical Genetics Laboratory Director, Dr. Narayan Prasad (Neurometabolic), Sarah Denomme - Registered Dietician, Sue MacLean - Social Worker)



"There are only two ways to live your life. One is as though nothing is a miracle. The other is as though everything is a miracle." - Albert Einstein

The Inherited Metabolic Disorders News

Metabolic Family Workshop 2017



**We hope to see
you all in 2019!**

The Inherited Metabolic Disorders News

Suzanne's Corner



Low Protein Cooking Demonstration 2017



The Low Protein Cooking Demonstration was held on May 13th, 2017. Thank you to Suzanne Ratko, Sarah Denomme, Leslie Harden, Nicole Marleau, Bethany Staubitz, Emily Gagnier, Chef Patrick, Chef Neal (U.K.), and Janna Boloten (from our sponsor Vitaflo) for a fantastic day!

TSA Cares



For our families with special medical needs and disabilities who travel by air to and within the USA, check out the **TSA Cares** link. **TSA Cares'** toll free helpline assists travelers with disabilities and medical needs.

<https://www.tsa.gov/news/releases/2011/12/22/tsa-announces-launch-tsa-cares-toll-free-helpline-travelers-disabilities>

Suzanne's Corner (continued)

"Pigs" in a Blanket

Number of servings: 24



This fun, low-protein version of an all-time favourite snack is great for the entire family, and all ingredients can be easily found at your local grocery store!

Ingredients

1 recipe of car"hot"-dog

- 8 peeled carrots

Marinade

- 3 Tbsp (45 ml) vinegar
- ¾ cup (175 ml) water
- ¼ tsp (1 ml) de salt
- 2 tsp (30 ml) sesame oil
- 1/3 cup (75 ml) soy sauce
- 1 clove of garlic, minced
- ¾ tsp (4 ml) liquid smoke

1 recipe of pizza dough

(½ will be used in this recipe)

- 2 Tbsp sugar
- 1 ¾ cup (425 ml) warm water
- 6 g (1 ½ tsp) traditional yeast
- 2 Tbsp (30 ml) olive oil
- 350 g (3 ½ cups) Wheat starch (0454 or 2203)
- 60 g (1/3 cup) Métamucil
- ½ tsp (2.5 ml) salt

Preparation

Prepare the car"hot"-dogs

- In a pot, place the whole carrots with water. Once the water is boiling, cook for about 12 minutes. Carrots should be al-dente.
- To stop the cooking, put the carrots in cold water.
- During cooking, mix together the ingredients for the marinade and place it in an airtight container.
- Once cooked, place the cooled carrots in the marinade. Refrigerate for at least 24 hours.

Prepare the pizza dough

- In a bowl, mix warm water, sugar and yeast. Let stand for 5-10 minutes to activate the yeast.
- Meanwhile, mix together the wheat starch, Métamucil, salt and baking powder.
- When the yeast is activated, add oil to the mix. Make a well in the center of the dry ingredients. Pour the liquid ingredients. Mix well. Knead the dough for a few minutes.
- Place in an oiled bowl and let rise for 25-30 minutes with humidity (Heat a cup of water in the microwave oven-it creates the perfect humid place to let the dough rise).

Prepare the pigs in a blanket

- Preheat oven to 350 F.
- Dry the carrots with a paper towel.
- Roll dough to obtain a rectangle of 35 x 25 cm (17 x 10 inches).
- Place marinated carrot on the dough. Roll to cover the dough (dough should overlap slightly below the carrot).
- Cut into 3 sections and place on a baking sheet.
- Repeat the steps to use all the carrots and half the dough recipe.
- Bake for 17-20 minutes.

Serve with ketchup, mustard or your favorite sauce.

Serving	1 "pigs" in a blanket
---------	-----------------------

Energy	47 kcal
Protein	0.2 g
PHE	15 mg
TYR	10 mg
LEU	25 mg
VAL	17 mg
ILE	18 mg
MET	5 mg
LYS	25 mg

The Inherited Metabolic Disorders News

Our Stars



Doris (left), the incredible adoptive mother of Nancy Emerson (right) - patient with PKU, celebrated her 92nd birthday this year!

Happy Birthday Doris! Thank you for providing us with so much joy!



Congratulations Laith Sharma - patient with Maple syrup urine disease (MSUD) for being recognized for his amazing service as a volunteer at the Hindu Temple and Cultural Center of Windsor!



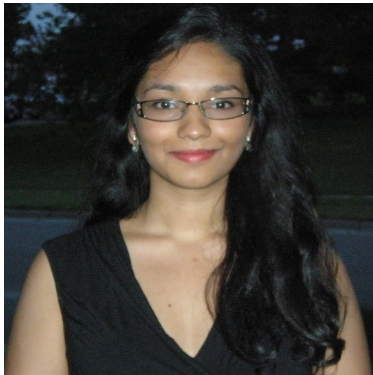
Research / Publications

Jessica Tao

My name is Jessica Tao, and I have the pleasure of working as Dr. Chitra Prasad's Summer Research Student. I started as a volunteer at Medical Genetics 2 years ago, and now, Dr. Prasad and I are conducting a pilot study with the Metabolics Team.

Our pilot study is investigating the **use of complementary and alternative medicine (CAM) in patients with inborn errors of metabolism (IEM)** through patient questionnaires. Currently, there is a lack of research in this field, and we aim to use this research to educate health care professionals about CAMs and help us understand how to further improve the care of patients with IEM.

I will be leaving LHSC at the end of August to attend medical school at the University of Ottawa. However, I am beyond excited to continue this amazing project in our nation's capital in collaboration with LHSC. Thank you Dr. Prasad, Janice Little and the entire Medical Genetics team for making my time at LHSC so rewarding and memorable.



Srinitya Gannavarapu, BSc.

Over the past three years, I've had the wonderful opportunity of working on multiple projects under the supervision of Dr. Chitra Prasad and Dr. Tony Rupar.

Most recently, I've been working on identifying the disease-causing gene variant in an unclassified white matter disease. With exome sequencing and *in silico* analyses, I've compiled a candidate gene list. Functional tests with patient and unaffected control fibroblasts are being carried out to assess the possible pathogenicity of candidate gene variants and to investigate molecular implications of identified mutations.

I've also contributed in the completion and writing of two other studies centered around the psychological and emotional impacts of newborn screening (NBS) processes on mothers and family members. The crux of this research is to highlight and discuss the possible psychological stressors invoked by either false positive results and/or living and accommodating to a child with an inborn error of metabolism. Our understanding of psychosocial aspects of NBS and subsequent treatments is limited. Exploring the intricacies of these peripheral effects, often clinically unidentifiable, is an integral part of addressing many of the challenges and limitations



Prashanth Rajasekar

Prashanth has been working with Dr. Chitra Prasad and Team on a study entitled "Psychosocial Aspects of Inborn Errors of Metabolism".

The study aims at identifying the psychosocial stressors that families with inborn errors of metabolism face, and proposing possible support systems to help families cope with these stressors.

Research / Publications (continued)



Ravi Datar

Ravi has been working on a project entitled “Magnetic Resonance Imaging in the Diagnosis of White Matter Signal Abnormalities” with Dr. Chitra Prasad and Team.

This project has been presented at the Garrod Symposium, May 2017, and is now being written up as a publication.

Dr. Chitra Prasad is now a full professor at Western University, Schulich School of Medicine & Dentistry, Pediatrics, Genetics/Metabolism.

Congratulations Dr. Prasad!

Hunter’s Hope Medical Symposium

In July, Dr. Tony Rupar attended the Hunter’s Hope Medical symposium in New York State. This symposium focused on the diagnosis, treatment and care for patients with leukodystrophies especially Krabbe disease. There was an extensive discussion of newborn screening for Krabbe disease detailing potentially significant improvements to existing methods. As in other diseases, there is evidence that diagnosing children with this rare disease at as young an age as possible is likely to improve the success of treatments. Research on improving treatments especially by using gene therapy was presented at the meeting.

Dr. Tony Rupar is also a member of the Canadian gene therapy trial for Fabry disease team.

Publications

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Publications (continued)

Acid Lipase Deficiency from Diagnosis to Therapy in Canada (Poster Presentation at Garrod Symposium, May 2017) by Chitra Prasad, Dhandapani Ashok, Samantha Colaiacovo and Tony Rupar.

Beth K. Potter, Michael Pugliese, Alana K. Fairfax, Laure Tessier, Monica Lamoureux, Pranesh Chakraborty, Doug Coyle, Jonathan B. Kronick, Kumanan Wilson, Alette Giezen, Robin Hayeems, Shailly Jain, Anne-Marie Laberge, Julian Little, John J. Mitchell, Chitra Prasad, Komudi Siriwardena, Rebecca Sparkes, Kathy N. Speechley, Sylvia Stockler, Kylie Tingley, Yannis Trakadis, Jagdeep Walia, Brenda J. Wilson. Parent-reported experiences with health care for children with inherited metabolic diseases in Canada: results from a multi-centre survey, CIMDRN SMDM conference.

Inspirations

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Life is a journey... not a destination. Enjoy the trip!

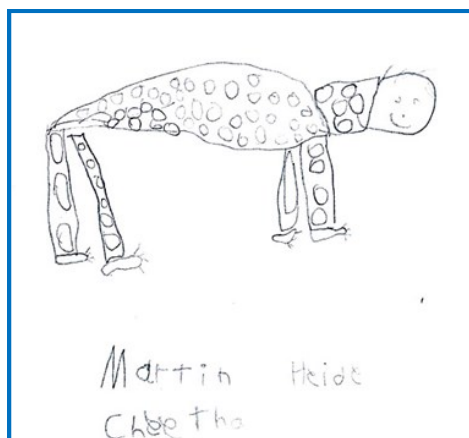
He who laughs... lasts.

Life's precious moments don't have value, unless they are shared.

Success stops when you do.



Evelyn, 3



**"Cheetha"
Martin, 7**

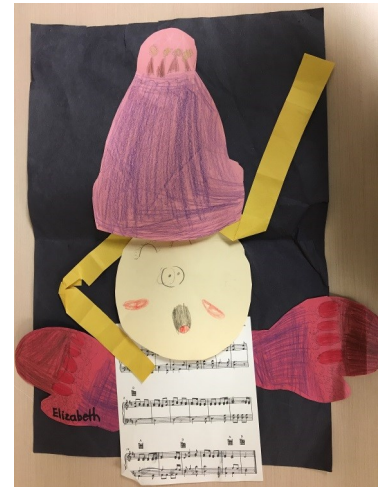
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Inherited_Metabolic/index.htm](http://www.lhsc.on.ca/Patients_Families_Visitors/Genetics/Inherited_Metabolic/index.htm)

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