The Inherited Metabolic Disorders News

Summer 2012 Volume 9 Issue 2

Children's Hospital
London Health Sciences Centre

In this Issue

From the Editor

Summer is finally here, and the weather has been beautiful!
Our 9th annual Metabolic Family Day on May 11th and the 8th Low Protein Cooking Demonstration on May 12th were a huge success. See the section "What's New" for a full report of the events.
We encourage families to send their information and news. Please contact me by email (**janice.little@lhsc.on.ca**) or by telephone: 519-685-8453 if you wish to contribute to the newsletter.
Have a safe and happy summer!

Janice Little

From Dr. Chitra Prasad

Greetings!
Hope you are having a great summer! Our current newsletter summarizes the stories and information from three wonderful families (April Willison, Michael Dremo and Jasper More). The information was presented at the 9th metabolic family workshop and was the highlight of the day. I am always impressed with our young patients and their families who are so courageous and teach us to enjoy each and every minute of our lives. Dr. Sharan Goobie's talk on role of media and metabolic disease was very informative. Karen Sappleton, Interprofessional Education Specialist from Hospital for Sick Children Toronto gave us some excellent ideas about how to introduce transition strategies for our patients. There have been a number of recent updates about research studies, newer therapies that you can read about in the newsletter. My trips this year took me to Australia, a beautiful continent. I enjoyed the meeting (international child neurology), scenery, beautiful beaches and also the other lovely sights and people. The koalas and kangaroos were a treat to watch. Here are some pictures. Enjoy! Have a wonderful summer!

With best wishes,
your friend,
Chitra Prasad

Too often we underestimate the power of a touch, a smile, a kind word, a listening ear, an honest compliment, or the smallest act of caring, all of which have the potential to turn a life around.
Leo F. Buscaglia

What's New

Metabolic Family Workshop 2012

Members of the metabolic team from the Genetics Program at Children's Hospital who organized the annual event include (back from left) Dr. T. Rupar, Dr. N. Prasad, Dr. G. Filler, Dr. Gordon, Dr. McNeill, Dr. S. Goobie, Sarah Denomme, Joanne Psiuk-Rodgers, Joanne Weir and (front from left) Jennifer DiRaimo, Dr. C. Prasad, Melanie Napier, Janice Little, Suzanne Ratko, and Bridget Reineking

"The aim of the workshop is to educate families and health care providers about various aspects of metabolic conditions," says Dr. Chitra Prasad, Clinical Geneticist and Director of Metabolic clinic. "This is an opportunity for patients and families to network and make connections with others who have a family member or are themselves affected with metabolic disease."

The workshop provided a host of sessions on specific metabolic disorders and guest speakers including family presentations which were appreciated by all. Throughout the day and lunch, workshop participants interacted with the guest speakers, learned about different metabolic formulas and foods, visited displays and discovered ways to care for themselves and their families.

For Debbie White, whose six-year-old daughter has an extremely rare metabolic disease, the workshop provides an opportunity to gain more knowledge to support her daughter and to network with other parents. This year White, who also sits on the Family Advisory Council, participated in a panel discussion.

At the workshop I get to meet others in a similar situation, so I feel I'm not alone, there are others walking the same path," says White. "I feel very strongly that parents learn from other parents. "Inherited metabolic disorders are genetic conditions that interfere with the body's metabolism of protein, fats and carbohydrates. These disorders affect many organs and can be complex.

"Inherited metabolic disorders can occasionally be fatal. With recent advances in treatments, such as

What's New - continued

enzyme replacement and dietary therapies, the quality of life for individuals affected with metabolic disorders has significantly improved. In addition, newborn screening has made an enormous impact on the prognosis of these disorders with early detection and intervention.

Save the Date:

Metabolic Family Workshop 2013 will be held on Friday, May 10th, 2013 at the Best Western Lamplighter Inn and Conference Centre.

Family Stories

At this years Metabolic Family Workshop, we were fortunate to have 3 family presentations.

Michael Dremo did a break dance, April Willison shared her story and the More family talked about their journey to get enzyme replacement therapy for their son Jasper.

Here are the summaries of their presentations:

Michael has classic PKU (phenylketonuria). He shared with us many ways in which he deals with PKU, especially through his break dancing.

Michael Dremo

Family Stories - continued

April's Story

Hi, my name is April Willison. I am 23 years old. I graduated from the Hotel Management program at Fanshawe College in 2010. I am now completing my food service worker certificate through online courses. I currently work at Windermere on the Mount Retirement Home as a concierge and People Care nursing home as a dietary aide. I also work for my landlord as a building supervisor. I work about 60 hours a week. I live on my own with no roommates. I have a car, a boyfriend and two cats named Simon and Buddy; they are like my kids. I am also a runner. I run about 50 kilometers a week. On April 29th I ran in the forest city road race, it was my first half marathon. My goal time was 2 hours and I ran it in 1 hour 52 minutes. It felt amazing. Based on everything I have told you I may seem like a normal young woman, but I am different. When I was one year old, I was diagnosed with a metabolic disorder called MCADD. My parents were lead to my diagnoses because my brother Adam died when he was 2 from the same disorder. Babies get screened for MCADD as part of the newborn screening, but it was not always part of this testing. So when Adam was born we thought he was a normal baby. He was sick with the flu and since we didn't know, the proper precautions were not taken and he passed. Because of my brothers death I was tested and we know that I have it too. My older brother saved my life.

The easiest way for me to explain MCADD would be that: say you have the flu and you cannot keep anything down. Your body will go to your stored fat and break it down to get energy to keep your body working. My body cannot do that. I cannot break down certain kinds of fat. When my body needs extra energy it can't use fat, so unless I eat something my body shuts down. If I do get sick and cannot keep anything down I have to go to the hospital for a sugar IV to keep a constant energy source.

Having MCADD certainly isn't the worst thing to have but it is frustrating. I have to take medicine called carnitine 3 times a day, which I have trouble doing sometimes. So I have learned that I have to keep it everywhere so that I remember.

I make my own doctor appointments now and have been coming to these appointments on my own since I was 17. I have learned to make an appointment to see Dr. Prasad and Megan, the dietician, when I want to try something new like preparing for my half marathon. I want to make sure I am doing the right things. I also have to make sure I always have medicine on hand. When I was young my parents would always order it when I was getting low and go and pick it up. Now it is my job. I am now responsible for knowing my disorder, how to deal with it and how to handle it day to day and especially when I am sick. I am on my own now and I have learned how to take care of myself.

Family Stories - continued

I have had a pretty normal life. I did well in school. I was active as a child; I was on every sports team. And when you have a disorder, it doesn't make you different from other kids. But because I have MCADD there are certain things that I had to learn about the hard way because when you're a kid or teenager, you think that nothing bad will happen to you. I didn't fully understand this until I got a bit older. But we do have to make our own mistakes and learn on our own.

Looking back on my experiences, some advice that I would give to parents is that "kids will be kids". Don't bubble wrap them. Let them be as normal as possible, they still need to grow up and experience life even though they have a disorder. They will make mistakes and they will push your buttons and push their limits. But they will learn and they will achieve great things. If anything, having a disorder makes you want to achieve more so you can say that I CAN do it and I will. I can ride my bike from Grand Bend to London and back, I can run a half marathon and I can speak about my life in front of 150 people. Because you have a metabolic disorder, doesn't mean you can't achieve your goals. If you want to do something, do it. Just make sure you are doing it right. We all can and we will. Thank You.

Media and Metabolic Disease

By Pam More

When I sat down to put together a few words for today, I can't help but think of how much has happened to our family in just one short year. We have learned medical terms we didn't know existed, we have met countless incredible professionals, we have fought battles we could have never imagined, we have made connections that will last a lifetime and we have learned how strong and supportive our family really is. I will never forget that day in April of 2011 when Dr Goobie had the unfortunate task of meeting with Darren and I to discuss the results of earlier testing she had done on our son Jasper. It was then that we heard the three letters that would forever change our world – MPS. We struggled to understand their meaning and to figure out how we were going to deal with all of the information and emotions that were suddenly and quite unexpectedly thrown our way. We were plagued with feelings of helplessness and confusion, guilt and despair.

Fast forward to July in Jasper's journey it is determined that Jasper in fact has MPS VI, otherwise known as Marateaux-Lamy Syndrome. We learn that Jasper is only the 8th known case of MPS VI in Canada but we also learn that there is a treatment available- Enzyme Replacement Therapy (ERT) using the American made drug Naglazyme. We

Family Stories - continued

learn the drug is not recognized in Canada at this point in time and it is very expensive to the cost of $300,000 to $1,000,000 per patient per year. We become aware at how difficult it may become to convince the Ontario Ministry of Health to fund treatment for Jasper.

It was at this point we decide to contact the Isaac Foundation. Started a few years earlier by Andrew and Ellen McFadyen of Campbellford Ontario, parents of Isaac who has also been diagnosed with MPS VI as well. We immediately began the process for applying for funding to cover the Enzyme Replacement Therapy that is available for MPS type VI. On one hand our doctors were telling us there was treatment available but on the other hand it looked like obtaining the necessary funding was going to be very difficult. We knew in our hearts we wouldn`t stop fighting until we had the answer we wanted and funding for Jasper was approved but at the same time we didn`t know where to start. We felt helpless. Our medical team began the process of completing the necessary paperwork to allow the drug into Canada and secondly applying for funding. They took the time and care necessary to research requirements and procedures, gather information and expert referrals.

Up until this point we hadn't talked a lot about Jasper's diagnosis to many people outside our immediate family. We had shared some information with a hand full of close friends and co-workers but we still hadn't really spread the word to everyone we knew. We decided we had no choice but to become prepared and we jumped in, with both feet.

Our first step was to start emailing our MPP. We figured making him aware that this was likely to become an issue was the best place to start. We quickly learned that to get a response all you had to do was add a simple carbon copy to a member of the opposition. We were finally contacted by our MPP who requested he come to our house for a visit. Once he arrived he went over the process of application and the various stages of appeal we could expect before us. He sounded to us like he was smoothing over the "No" he knew we were about to receive on our request for funding. We had a bad feeling that things were not going to go the way we wanted them to.

We spoke again with the Isaac Foundation who encouraged us whole heartedly to launch a very public campaign. When we started to survey the political landscape we realized we had the timing of a provincial election on our side. We had about 3 weeks before the election was to be called. If we could make things happen in those three weeks by threatening to be in the faces of politicians during election campaigns it should definitely help our case.

Family Stories - continued

Sure enough, 2 days after the visit from our MPP, our application for funding was denied. The press release that had been previously prepared was immediately dispatched out and the fight was on! The release was picked up by various larger newspapers across the province as well other parts of Canada. We began sending out the press release to all of the local papers in our area and spreading the word wherever we could. A boy in Ontario has been denied life sustaining treatment! We suddenly had reporters contacting us, wanting to write an article, have a photo of Jasper to print alongside their article and requesting interviews with us. The first one was the hardest and most stressful and the more we did the easier they became. Or so he says, I let Darren handle most of these.

At the same time this was going on, we engaged the help of social media as well. Often thought to be brushed off as fluff, Facebook and Twitter became our next tool. The response we got was nothing short of amazing. Friends and family went to town and within hours, the Ministry was flooded with calls. We had people calling from across the province and country and even from outside Canada. We had people contacting us to report they couldn't get through so we requested they start calling their own MPP's. This continued tirelessly for three days. We began sending tweets to Deb Matthews and Dalton McGuinty. We wanted them to know we weren't going away. We had a television crew travel to our home from Hamilton to do a piece and a radio station from Owen Sound and Goderich requesting to share our story. We had a friend of a friend hand deliver a copy of the press release to Deb Matthews. Finally Dr. Goobie called to tell us that funding had been approved. Jasper was going to be able to receive funding for his treatment that we had so desperately been lobbying for! It took us awhile to digest this but we most definitely could not keep this news to ourselves for long. Once again we turned to social networking to spread the joyous news and updated reporters that were working on stories for us.

As I said earlier, looking back over the past year and all we have learned and all we have accomplished is amazing. We travel to London each week for Jasper to receive his enzyme replacement therapy. All our reports continue to be positive and the health care professionals we look to for guidance in his care have been wonderful. Even our shy Jasper is warming up to them …. Melanie often gets a colourful picture for her wall, Dr Goobie has a special nickname and gets a smile and Dr Prasad even gets a hug. However, ladies, I must tell you that when it comes to our weekly Wednesday visits, Terry is tops!

Family Stories - continued

We know Jaspers journey is going to be unique and we aim to be the best parents we can be to him and support his every endeavour. We have since joined forces with the Isaac Foundation and strive to fundraise to support research efforts. It is certain all forms of media will again come into play as we continue to raise awareness of MPS and set our sights on new achievements. We have proven to ourselves that we can accomplish what we set out to do. Our next goal is improved treatment options and eventually a cure. It is out there somewhere and we are going to find it.

Jasper's first 'official' T-Ball practice - age 3

Art of Extreme Care NEW

- Keep a gratitude journal
- Take a walk with your camera - take pictures of all you see that delights you
- Find and notice something beautiful every day
- Admire beautiful artwork
- Do absolutely nothing on some days

Suzanne's Corner

This recipe was a smashing hit at our Low Protein Cooking Demonstration on Saturday, May 12th. Thanks to Chef Bill and Nutricia for providing the recipe:

EGGPLANT DIP

Yield	Serving Size	Per serving
16 servings	28 g or 1 oz	Protein 0.2 g Phenylalanine 8 mg

Ingredients

2 cup (1 medium)	Eggplant
6 cloves	Garlic
½ tsp	Salt
½ cup	Miracle Whip
½ cup	Onion
1 each	Lemon

Preparation

- Preheat oven to 400 degrees
- Cut the eggplant in half & place on a baking sheet
- Rub oil on each half & bake for 30 minutes or until tender
- Allow 15 minutes for the eggplant to cool slightly & then peel off the skin
- Discard the skin
- Place the eggplant and the remaining ingredients in a blender or food processor
- Whip until fully blended
- Season as necessary
- ENJOY

The Inherited Metabolic Disease (IMD) Program finally launched a new list of formulas and foods coverage by the starting APRIL 2012

FOODS

Walden Farms Peanut Spread
La Tiara Taco Shells
Loprofin Pasta Lasagna
Cambrooke Brooklyn Dog Buns
Country Sunrise Vegetable Hot Dog Mix
Homestyle Fudge Brownie Mix

FORMULAS

If you are interested in trying a new formula, please contact Suzanne

TYR Cooler 15 Red
HCU Cooler 15 Red
MSUD Cooler 15 Red
PhenylAde 60 Unflavoured Drink Mix Pouches and Cans
PhenylAde 60 Vanilla Drink Mix Pouches
PKU Cooler 10 Red
PKU Cooler 15 Red
PKU Cooler 20 Red

Achievements

Lama Kadri

Hello my name is Lama Kadri and I am 21 years old. I have always envisioned nursing as the profession I wanted to be a part of.

In following this vision, I enrolled in the practical Nursing program at St. Clair College in Windsor, Ontario. I knew that I would need to work harder than my classmates to achieve my diploma and with a few bumps along the way I finally made it with just one course to finish up in nursing and the completion of my board exam.

I have decided to continue on in my education as I have always wanted to be a Registered Nurse. I will be attending to the University of Windsor in the fall of 2012 to start my journey to becoming a Registered Nurse. With the background I have gained through the
Practical Nursing Program, I hope to upgrade my marks and realize my dream.

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, do so on the **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 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.
Thank you!

Research News

Psychosocial Aspects of Inborn Errors of Metabolism

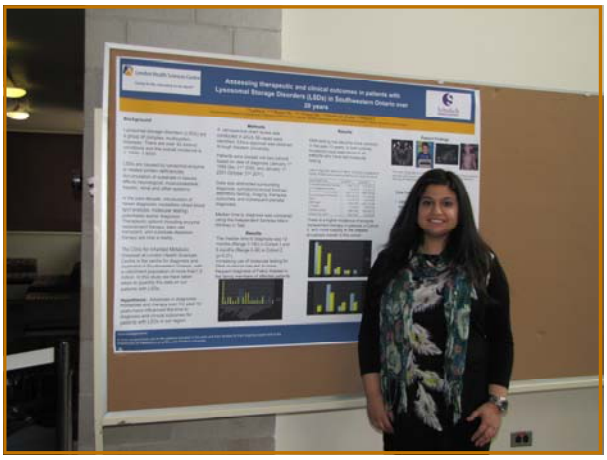
The primary goal in the healthcare industry has always been to tend to the mental, social and physical well-being of our patients. In that respect, we feel it is crucial to think past the clinic to truly understand what families experience in their day-to-day lives. In our new study entitled *Psychosocial Aspects of Inborn Errors of Metabolism*, led by Dr. Chitra Prasad, we aim to speak to families with children diagnosed with an Inborn Error of Metabolism (IEM), to identify the difficulties they face while dealing with such a disorder. Inborn Errors of Metabolism are genetic disorders which often create challenges in the lives of children and their families. Dietary restrictions, genetic testing, repeated blood work, enzyme replacement therapy and even organ transplants are some of the challenges families must face.

Our study aims at identifying the psychosocial stressors these families face, and proposing possible support systems to help families cope with these stressors. We hope to ultimately better our understanding of the difficulties families dealing with an IEM face in order to improve the level of care provided to them.

The team working on this study includes Dr. Chitra Prasad (Medical Geneticist), Dr. Akshya Vasudev (Psychiatrist), Dr. Beth Potter (Epidemiologist), Dr. Narayan Prasad (Paediatric Neurologist), Melanie Napier (Genetic Counsellor), Andrew Mantulak (Social Worker) and myself, Prashanth Rajasekar (Student Researcher). If you wish to participate in this study, please contact me at 519-685-8500 ext 56795 or **prajase@uwo.ca.**

Cheers,

Prashanth



Dr. Alysha Ladha (Paediatric resident) presented the paper “Assessing therapeutic and clinical outcomes in patients with Lysosomal Storage Disorders (LSDs) in Southwestern Ontario over 20 years” at the Garrod meeting in Winnipeg in May of 2012

Research Meetings & Presentations

1. Emotional aspect of newborn screening: by C. Prasad, T. Morley, K.O'Connor, J.Diraimo, S. Goobie, B .Potter, G. Moran, P. Chakraborty and C.A. Rupar. Preliminary data presented at Children's Hospital of Eastern Ontario in Ottawa
2. Erythropoietic Protoporphyria: Clinical, diagnostic and therapeutic experiences from Southwestern Ontario by Gonzalez S, Napier, MP, Prasad C and Rupar T - Presented at Garrod meeting May 2012 Winnipeg
3. Assessing therapeutic and clinical outcomes in patients with Lysosomal Storage Disorders (LSDs) in Southwestern Ontario over 20 years by Ladha, A, Prasad, C, Prasad, N, Rupar, C.A. Presented at Garrod meeting May 2012 Winnipeg
4. World Lysosomal meeting Feb 2012 SanDiego (Dr. Tony Rupar)
5. American Society Gene and Cell Therapy May 2012 Philadelphia (Dr. Tony Rupar)
6. Palliative care issues in the setting of neurometabolic clinic by Dr. Chitra Prasad, Lisa Pearlman and Dr. Narayan Prasad. Presented at International Child Neurology Association May 2012 Brisbane Australia
7. 6 PTPS deficiency by Dr. Narayan Prasad, Dr. Tony Rupar and Dr. Chitra Prasad. Presented at International Child Neurology Association May 2012 Brisbane Australia
8. Canadian Paediatric Society May 2012 Emerging therapies for metabolic disorders for paediatricians (Dr. Chitra Prasad and Dr. Julian Raiman)
9. Canadian Paediatric Society May 2012. Workshop on hypotonia (Dr. Narayan Prasad, Dr. Kym Boycott, Dr. Craig Campbell and Dr. Chitra Prasad)
10. International conference for genetic / metabolic dietitians and conferences on Kuvan therapy in PKU New Orleans (Suzanne Ratko)
11. Health Care Advocate Meeting - March 2012 Halifax (Melanie Napier)
12. Canadian Fabry Disease Initiative Annual Scientific Meeting - June 2012 Halifax (Melanie Napier)
13. Canadian Fabry Association National Patient Conference - June 2012 Halifax (Melanie Napier)

Newer Therapies and Research Trials that our patients are participating in:

1. Kuvan therapy: A number of patients are now receiving BH4 (Kuvan) for PKU through Kuvan assistance program.
2. A double-blind, placebo-controlled, randomized study to evaluate the safety and therapeutic effects of sapropterin dihydrochloride on neuropsychiatric symptoms in subjects with phenylketonuria (PKU).
3. Oral drug therapy for Gaucher disease- This is an advance as previously only intravenous enzyme replacement therapy was available.
4. Use of Hyperion product for urea cycle instead of sodium phenylbutyrate.
5. New enzyme therapy for Morquio disease
6. Fabry disease- various trials (through Canadian Fabry Disease Initiative and replegal studies)

What's New in the Clinic

Welcome to Melanie !

My name is Melanie Napier and I'm the new metabolic genetic counsellor working with the Metabolic Team; I started this position January 2012. My educational background includes a BSc in Genetics from the University of Western Ontario in 2006, an MSc in Biochemistry and Molecular Biology from the University of Calgary in 2009 and an MSc in Genetic Counselling from the University of Toronto in 2011. After graduation I held two positions before I came to London Health Sciences Centre: interim-coordinator of the DNA Resource Centre at the Hospital for Sick Children in Toronto and genetic counsellor at McMaster Children's Hospital in Hamilton. I am pleased to be part of the Metabolic team here in London. For those of you I haven't had an opportunity to meet through clinic or the Metabolic Family Workshop, I look forward to getting to know you in the future.

"She thinks that if she calls it 'e-broccoli' we'll eat it."

Olivia, Age 6

Contact Information

Editor: Janice Little
LHSC - Medical Genetics Program of Southwestern Ontario
Tel: 1.800.243.8416
1.519.685.8453

Email: janice.little@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