The Inherited Metabolic Disorders News

Children's Hospital
London Health Sciences Centre

Summer 2013 Volume 10 Issue 2

In this Issue

From the Editor

I hope everyone is having a wonderful summer!

Our 10th Metabolic Family Day and 9th Low Protein Cooking Demonstration were once again a huge success. See the section "What's New" for a full report of the events, as well as pictures.

As always, your suggestions and stories are welcome. Please contact me by email or telephone if you wish to contribute to the newsletter.

I hope everyone is having safe and happy summer!

Janice Little

From Dr Chitra Prasad

Dear Friends,

Greetings! This year was the 10th anniversary of our metabolic family workshop. The event was very successful thanks to each and every one of you, our metabolic team, our sponsors, Children's Health Foundation and many supporters. It was also a very special occasion for me since both my mother Dr. Tara Saxena (from India) and my mother-in law Mrs. Mani Bhashyam attended this incredible event. For those of you who could not make it, please read about the event in our newsletter. As always your suggestions and feed back helps us immensely.

There are a couple of stories of courage and resilience we would like to share with you in this newsletter. (The Hummell and Pearson families). As I read their stories I was reminded of the quote:

"Anyone can give up, it's the easiest thing in the world to do. But to hold it together when everyone else would understand if you fell apart, that's true strength".

-- Christopher Reeves

My best wishes to all

Your friend

Chitra Prasad

Personal Stories

Carver David Hummell

Our journey began with the birth of our son; a planned c-section for a breech (feet first) baby made planning the delivery easier and more exciting!! On February 10/2010, following an uncomplicated section was the birth of our beautiful and healthy baby boy; Carver David Hummell weighing in at 7 lbs, 2 oz. and 21" long. The perfect day shared with family, friends and even big brother Caleb. The joy and excitement continued into the next day only I had an unsettling feeling, Carver was sleeping more and feeding less. With a slight decrease in temperature, I was to wrap him tightly in blankets, and keep pushing baby to feed. By the next morning, Carver's weight had become a concern, barely hovering over 5 lbs I was to push feeds and top him up with infant formula. By that night, Carver became weak and sluggish and his breathing pattern had changed. Dave was on his way out to prepare for our arrival home in the morning, but he was pacing by the door uncertain if he should go. I assured him everything would be fine that the nurses were keeping their eye on Carver. Attempting to feed again, I held his limp and lifeless body against mine and cried, filled with fear and panic, what was wrong???? The nurse came in to check on us, without hesitation, she rushed him out of my arms and down to the nursery. I sat there waiting alone, the minutes felt like hours, then I slowly made my way to the nursery where I watched the nurses through the glass busy with a tiny, fragile newborn; it took me a moment to realize that it was Carver. I cried and couldn't help but wonder what had I done wrong???

Carver required constant monitoring throughout the night along with being intubated, IV's, antibiotics, X-rays, blood work, and even a lumbar puncture (a large needle to withdraw spinal fluid to check for infection). With no results, Carver was then transported to Children's Hospital in London. I watched a group of strangers rush my critical newborn baby away.

Once we arrived at Children's Hospital we were directed to the PCCU (pediatric critical care unit) where we were flooded with questions (about the pregnancy, family history) I could barely answer the questions coming at me as I stood there staring at Carver attached to so many beeping machines, monitors and IV's. We were then told that Carver had hyperammonemia (toxic blood ammonia levels) his levels were over 800 (normal range 15-55) which can lead to brain damage, coma and death without immediate treatment. The doctors were inserting a dialysis catheter through his skull and then he would undergo hemodialysis to 'clean his blood'. What were supposed to be the happiest days of our lives had become a living nightmare. Carver was 3 days old and there was no guarantee he would even survive the required treatment.

As Carver began to stabilize, with ammonia levels under 100, he no longer required dialysis. He continued with many more treatments and tests in the PCCU. At this time, we

Personal Stories - continued

were introduced to the metabolic team explaining that Carver had an inherited metabolic disorder called Methylmalonic Acidemia (MMA). Carver spent nearly 3 months in the ill infant nursery at Children's Hospital. During this time he continued with extensive blood tests, constant monitoring, medication, and special formula via NG-tube (nasogastric tube). We had frequent visits from Suzanne Ratko, Carver's dietician, reviewing weight gain, poops, diet, tube feeds, and blood values, while I would ask her a dozen questions a day. I can remember Suzanne rhyming off medical jargon and numeric values that we could not even pronounce let alone spell or understand, but (just as she assured us) we did learn them very quickly. Dr. Prasad would pop in and out to 'brief' us on Carver's specific metabolic disorder, complications, treatment, diet restrictions, medications, emergency protocols, sick day guidelines, SO MUCH TO LEARN!!! "

Carver's blood become more and more difficult to get from his veins, which meant uncertainty with lab values, and a lot of needle pokes for the poor little guy!! A pediatric doctor who recommended the insertion of a portacath then approached us, and after much consideration, we agreed. After the port insertion, his blood work was actually coming back in the normal range. This meant it was time to prepare for Carver's discharge home. That word, discharge, filled me with both relief and panic!!! How would we care for a child with such a severe and complex disorder at home, were we ready????

Next thing we knew a year had flown by and we had succeeded, only returning to the hospital for his required appointments. Most frequently, we would 'visit' the PMDU where Carver had captured so many of the nurses' hearts with his charm and smile. In Carver's first year we were able to watch him grow and develop and reach so many milestones; holding his head up, rolling over, first tooth, first words, sitting up, crawling, first steps, and his 1st Birthday!!!

Our next big decision was to have a G-tube inserted. Carver had been feeding pretty well orally and gaining weight, but with a lack of appetite and hypotonia (low muscle tone) he tired quickly resulting in more frequent feedings. We were feeding anywhere from 2-4 X's during the night along with his daytime feeds. We were tired!!! Carver's weight began to plateau and then slowly drop. Our decision was clear at this time, but another surgery meant possible complications or metabolic crisis. However, his surgery was a success and Carver's weight began climbing rapidly, he was thriving again and we were all able to sleep at night.

Another year has flown by and Carver is now in his 'Terrible Two's'. Typical busy boy, always running, jumping, and climbing; his favorite word is NO; he loves tractors, trucks, and trains; books and painting; swimming and playing in the sand; playing with big brother Caleb; and his friends at daycare. Terrible two's have also been terrifying for us! As Carver started daycare in Feb/12 with a low immunity, and inability to properly fight infection he picked up (I swear) every single common cold virus. Runny nose, cough, and congestion not a big deal for the typical 2 yr old, but for Carver a simple cold rushes us

Personal Stories - continued

into the ER, resulting in an admission, to Carver's favorite place, the paeds floor at Children's Hospital. With nasal drops and suctioning, IV medication and hydration, continuous g-tube feeds, daily weights, daily blood work, and weighing diapers Carver slowly begins to 'bounce' back. Try keeping a busy two year old confined in a small room, the days feel endless!!! He would be discharged and it would seem like we were right back in hospital only a few weeks later. The winter months passed and Carver has been hospital free for 5 months, but with winter soon upon us again, we can only hope that it will be a better season for us this year. Keeping our house clean, frequent hand washing, flu shots for the entire family and screening all visitors for illness and infection is our plan for this upcoming cold/flu season.

Raising a child with a complex disorder has changed our lives completely. We have learned to be more organized, we manage to juggle appointments, activities, diet restrictions, preparing formula, weighing foods and formulas, g-tube feeds, medications, sick day guidelines, poop and puke logs. We do so in a single binder with so many calendars, charts and checklists. We've also learned to be more prepared, with a bag at the door for ER trips, packing extra clothes, a 'puke bucket', g-tube supplies, and an emergency protocol to name a few.

There are so many uncertainties with MMA, therefore we are very uncertain of what the future holds for Carver. Long-term complications can include feeding problems, developmental delays, kidney failure, pancreatitis (inflammation of pancreas). We manage Carver at home by following his strict diet and medication, monitoring his blood work very closely as well as weight gain. We continue with a ton of appointments with several specialists such as nephrology, neurology, cardiology, ophthalmology, PT, OT, speech, metabolic clinic and our pediatrician.

Most importantly, we have learned to ENJOY Carver everyday!!! His charming personality always keeps us laughing, his big, bright smile and wickedly handsome looks melts our hearts. He never ceases to amaze us and always keeps us on our toes!!! We've learned to live day by day or more importantly, we have learned to live in the moment.

This pretty much sums up our story for now, or as I like to call it Carver Hummell's journey….

Featured This Issue

Methylmalonic acidemia (MMA)

By Chitra Prasad MD FRCPC FCCMG

Originally printed in the Winter 2009 newsletter

Methylmalonic acidemia is an inherited disorder in which the body is unable to process certain proteins and fats (lipids) properly. The effects of methylmalonic acidemia, which usually appear in early infancy, vary from mild to life-threatening. Newborns can present with lethargy, metabolic acidosis, hyperammonemia and coma. Affected infants experience vomiting, dehydration, weak muscle tone (hypotonia), excessive tiredness (lethargy), and failure to gain weight and grow at the expected rate (failure to thrive). Long-term complications can include feeding problems, developmental delays, kidney disease, and inflammation of the pancreas (pancreatitis). Without treatment, this disorder can lead to coma and death in some cases.

MMA can also occur due to maternal dietary B_{12} deficiency, which is found in vegetarians. Complementation studies have revealed the presence of at least 8 different complementation groups (mut0, mut-, cblA, cblB, cblC, cblD, cblF, cblH) that cause MMA. The incidence of MMA is 1 case per 48,000 infants.

Management includes very close monitoring of the diet to remove the offending amino acids and to ensure proper nutrition. Both liver and kidney transplant has been tried in some severe patients of MMA. A team of specialists and other health care professionals are required for ongoing follow up and monitoring such as dietitians, nephrologists, neurologists, metabolic physicians, pediatricians, occupational and physiotherapists and social workers.

This condition is inherited in an autosomal recessive pattern, which means both copies of the MUT, MMAA, or MMAB gene in each cell have mutations. The parents of an individual with an autosomal recessive condition are carriers of one copy of the mutated gene but do not show signs and symptoms of the condition.

Special dietary formula for MMA

Suzanne's Corner

Medical Formula/Food Update

The Inherited Metabolic Diseases (IMD) Program which covers many foods, formulas and drugs has released an updated list effective July 25, 2013:

Food
Applied Nutrition Cinnamon Chip Flavour Muffin Mix
Cambrooke Bagels plain
Cambrooke Pita Pockets
Cambrooke Veggie Meatballs
Country Sunrise Chicken Flavoured Pattie/Nugget Mix
Country Sunrise Soft Tortillas
Loprofin Chocolate Cake Mix
Loprofin Rice
PKU Perspectives – Sandwichmate Imitation Cheese Slices
PKU Perspectives – Shredmate Imitation Shredded Mozzarella Cheese

Formula

If you are interested in trying a new product please let me know. You will not be able to order it until I have contacted the Shop or Centre.

The new formulas will be listed according the disorder which they are approved to be used with.

<u>Homocysinturia</u>
HCU Cooler 10 red
HCU Cooler 20 red
HCU Express 20
HCU Lophlex LQ Mixed Berry Blast

<u>MSUD</u>
BCAD1
MSUD Cooler 10 red
MSUD Cooler 20 red
MSUD Express 20
MSUD Lophlex LQ Mixed Berry Blast

<u>PKU</u>
PKU express 20 unflavored
PKU Lophlex LQ Juicy Orange
PKU Lophlex LQ Mixed Berry Blast

<u>Urea Cycle Disorders</u>
UCD Anamix Junior
UCD Anamix Junior, vanilla

Suzanne's Corner

Strawberry and Kiwi Shake

Ingredients (for 2 servings):
200 g (7 oz) strawberries, chopped (about 8 medium strawberries, or about 1 and 1/3 cup chopped strawberries)
1 fresh medium kiwi, peeled and chopped
25 g (1 oz) superfine granulated sugar (or baker's sugar) (1 and 1/2 tbsp)
200 mL (about 7 fl oz) prepared LP Drink

Method:
1. Place the strawberries and kiwi in a blender or food processor and blend for one minute.
2. Add the sugar and LP Drink and blend for another 1-2 minutes.
3. Pour into a glass and serve.

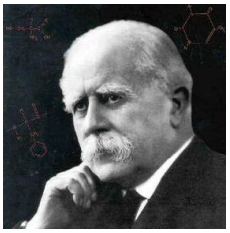
Nutritional Information	Calories	Protein	Phenylalanine	Fat	Carbohydrate
Per serving	156	1.6 g	10 mg	3.2 g	31 g

What's New

Garrod Association:

Dr. Chitra Prasad has become president of the Garrod Association of Canada. (inborn errors of metabolism)

Please see the following website for more details.
www.garrod.ca



Sir Archibald Garrod

IMPORTANT UPDATES TO METABOLIC FAMILY WORKSHOP AND LOW PROTEIN COOKING DEMONSTRATION

We would like to alert our families that in 2014 we will be hosting the Low Protein Cooking Demonstration in May. Information regarding the date will be communicated shortly. This event is for patients/families that require a low protein diet as part of their medical management.

Please note, we will NOT be hosting the Metabolic Family Workshop in 2014 as we are taking time to reorganize our Workshop to better meet the needs of our patients and families. We will be hosting the Workshop again in 2015 and hope to see you there!

We encourage you to contribute your thoughts on ways that we can improve this day for you and your family. Please contact Melanie Napier (519-685-8500 x 53510) to share your ideas!

What's New - continued

10th Annual Metabolic Family Workshop

The 10th annual Metabolic Family Day was held on Friday, May 10 at the Best Western Lamplighter Conference Centre, offering participants the opportunity to share information on the unique challenges that accompany metabolic disorders.

Metabolic disorders are multi-systemic in nature and children living with them can have problems with growth, developmental delays and issues with organs such as the heart and kidneys. These disorders can require special restrictive diets, as well as enzyme replacement therapies. Children's Hospital at LHSC currently treats close to 500 patients with metabolic disorders.

Families in attendance were invited to interact with speakers, learn about various metabolic formulas and foods, see what is new with treatment and research, visit displays of metabolic food products and learn about the resources available in their community. The day also provided an excellent opportunity for families to talk with each other, share their experiences and make a few friends along the way.

The day began with a warm welcome from Dr. Chitra Prasad, director of the metabolic clinic and medical geneticist at LHSC and opening remarks from Bonnie Adamson, president and CEO of London Health Sciences Centre and Dr. Victoria Siu, Director of the Genetics Program.

This year's keynote presentation was by Dr. Tony Rupar who spoke on "Getting Into Your Genes", addressing new developments in diagnosis and treatment of metabolic disorders.

What's New - continued

Presentations held throughout the day offered families a chance to share their experiences of living with metabolic disorders and finding coping strategies to deal with challenges. The afternoon sessions began with a welcome by Susan Crowley, president and CEO of Children's Health Foundation, the conference's main sponsor. Melanie Napier and Kim Tiemens talked about "sharing your story" . Nichole Meyers led everyone through a "stretch break" that all seemed to enjoy.

Learning continued into Saturday with a demonstration in low-protein cooking at the Loblaw Wonderland Market by Children's Hospital Dietitian Suzanne Ratko, and Nutrition Specialist Karen Gough of sponsor Nutricia.

Jasper in pensive mood

Dr. Rupar

Listening to an information talk

Dr. Beary Goode and friend

Kenney boys

Stretch Break

Dr. Chitra Prasad with Mrs Mani Bhashyam and Dr. Tara Saxena

Art of Extreme Care

- Order dinner in (or have someone else make it)
- Feel the sun (or rain) on your face
- Listen to your favourite music
- Turn off the computer, cell phone and TV for 24 hours
- Take a walk with your camera - take pictures of all you see that delights you

Research Accomplishments

Poster Submissions

Submitted to the Garrod Association for the Symposium on May 30/31st ,2013 in Sherbrooke, Quebec.
Biochemical and Hematologic Manifestations of Gastric Intrinsic Factor (GIF) Deficiency: Three cases in the Mennonite Population of Southwestern Ontario. A. Ferrand, M.P. Napier, C.A. Rupar, O. Y. Al-Dirbashi,

Krabbe Leukodystrophy across the Life Span; Experience at the London Health Sciences Centre South-Western Ontario Neurometabolic Service Prasad C, Rupar CA, Florendo-Cumbermack A, Dayal A, Napier MP, Raiman J, Levin S and Prasad AN

Submitted to the WORLD Symposium in Orlando in February 2013
Multiple Sulfatase Deficiency: Disease in Search of a Treatment! Chitra Prasad, C. Anthony Rupar, Craig Campbell, Melanie Napier, David Ramsay K.Y Tay, Sapna Sharan and Asuri N. Prasad

Assessing therapeutic and clinical outcomes in patients with Lysosomal Storage “Disorders (LSDs) in Southwestern Ontario over 20 years. Ladha, A, Rupar, C.A., Prasad, A.N, Seabrook, J.A and Prasad, C.

(Dr. Alysha Ladha also received first prize for paediatric resident symposium in May 2013 for this project.)

Submitted to ACMG in March 2013
Psychosocial Aspects of Inborn Errors of Metabolism. Prashanth Rajasekar, Melanie Napier, Andrew Mantulak, Narayan Prasad, Akshya Vasudev, Beth Potter and Chitra Prasad.

PKU 016 study has just been finished at our centre. Thanks to the entire team and our participants.

Submitted to the Newborn Screening Meeting Ottawa April 2013
The Emotional Impact of Expanded Newborn Screening: Exploring Mothers’ Experiences and Perceptions of Parenting K. O’Connor, T. Morley, J. DiRaimo, B. Potter, S. Goobie, G. Moran, P. Chakraborty, C.A. Rupar, & C. Prasad.

Invited lectures

Department of Pediatrics, at Sackler School of Medicine, Tel Aviv University Israel Invited lectures (Neurometabolic)

Considering metabolic diagnosis for neurological disorders: Approaches based on clinical experience at a neurometabolic clinic. Dr. Narayan Prasad and Dr. Chitra Prasad

Psychosocial aspects of inborn errors of metabolism and newborn screening By Dr. Chitra Prasad

Exome sequencing, expansion of phenotype of twinkle mutations By Dr. Chitra Prasad

Dr. Narayan Prasad- Plenary lecture in honor of Dr. Pinchas Lerman (Pioneer Paediatric Neurologist in Israel) "Algorithm for approach to epilepsy associated with in born errors of metabolism"

Dr. Narayan Prasad- 6PTPS deficiency

Events

Ben Pearson Memorial Golf Tournament

We lost our beloved Ben, after a very short illness from an OTC deficiency on the 2nd of October 2010. We were approached by the girlfriend of one of Ben's close friends, wanting to create a golf tournament in memory of Ben. You can only imagine how very much we were moved and how honoured we were for our son to have touched people so deeply, that they would want to do this. Working with his friends the first" Ben Pearson Memorial Golf Tournament was born. In just under three months. A tourney with 99 golfers, prizes and raffle items made a profit of $5,100.00

We decided to award two post secondary scholarships to members of Ben's hockey team who displayed his leadership skills and personality strengths and attributes on and off the ice. It was very healing for all involved.

This year on the 16th of June, with a year to plan, we had our second annual tournament. We went to a bigger course because we knew that there were many more who wanted to come but were unable the year before.

I had been attending our local bereavement center trying to make sense of his death and trying to find a way to move on without him. It was like a sign for my husband and I. We took the girls to a meeting there so they could see what a wonderfully supportive, welcoming and caring place the Coping Center and to hear from the creators of "Coping" exactly what and why they do what they do. They were convinced, as were we that the center is where we wanted the funds raised in the tournament to go. They help people of all ages deal with their grief at no cost to the participants. All of the staff there know intimately what its like to lose a loved one as they all have suffered a loss. This they do without any government funding and fully dependent on donations. Ross & Glenn are the most caring and compassionate people you could ever meet and the service they supply in our community is invaluable. For the first two weeks of July they run an adventure camp for children who have had a significant loss. In most cases it's a parent or a sibling. We knew immediately that this is where the funds had to go.

Our Ben loved to go to camp every year and we knew he would be smiling down on us for our decision to partner with the Coping center especially for the camp program. I'm very proud to say this year we had 180+ golfers, plus an extra 60 for dinner. After we sold out foursomes for 18 holes the course had to open the back nine as well. We had prizes for the golfers, raffle prizes again and this year we also added a silent auction, all was donated in Ben's memory, including the luncheon for the golfers. We raised over &14,000.00 dollars this year!! But more than that, our son was honoured and remembered in a very special and meaningful way. We have turned the tragedy of his death around and have found something positive to do in his memory.

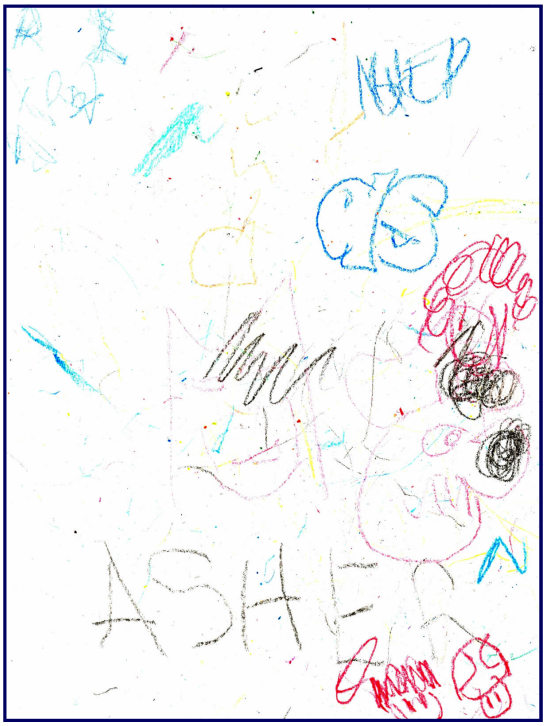
"Would you folks mind if I told my friends I was adopted?"

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!



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