

From the Editor

I hope you enjoy the Winter issue of "The Inherited Metabolic Disorders News." Inside you will pictures of some of our "Ghosts & Goblins." Please feel free to send pictures of your children for the newsletter. Email your stories and pictures anytime to janice.little@lhsc.on.ca

Janice Little

From Dr Chitra Prasad

Dear Friends,

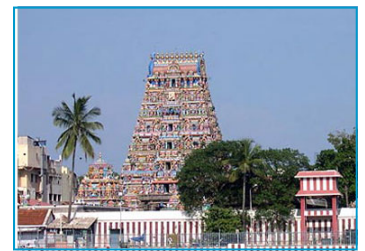
Greetings!

Our new space "The Zone B" is beautiful. We are close to the inpatient services, the cytogenetic and molecular laboratories and lab centers. It has taken a lot of planning and hard work of so many individuals to make this happen.

We are now planning for the next metabolic family workshop on the 11th May 2012 and the low protein workshop on the 12th May 2012. This year we plan to hold four simultaneous workshops (PKU, Teenage group, Lysosomal, Other metabolic disorders).

The word "inspiration" comes to mind when you read the story of "More family". The whole family and Jasper are amazing individuals. Please do read the story about young Jasper and Mucopolysaccharidosis type VI (an extremely rare disorder) in the newsletter.

I will be going to India in December. As usual I plan to attend meetings and give talks This time the neonatology meeting is in Chennai (Southern India). Chennai is a city with a lot of history (previously called Madras). Please see some pictures of Chennai temples. I also plan to visit with family and friends.



Chennai Temples

With best wishes for a wonderful Christmas, Hanukkah and a very Happy New Year.

Please send in your suggestions regarding the Metabolic Family Workshop to our Team.

Your friend
Chitra Prasad

I've learned that people will forget what you said, people will forget what you did, but people will never forget how you made them feel.

Maya Angelou, American author and poet

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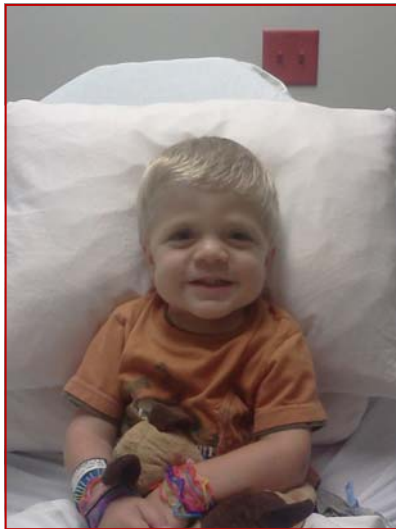
Photo by Rich Duncan - www.richduncanphotos.com

Personal Stories

Jasper's Journey

It was only April of this year when we heard the three letters that would forever change our world – MPS. These three letters were something we had never heard before our son Jasper's diagnosis. 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. When we looked at the face of our beautiful little boy, we no longer saw what we had seen even the day before but now we found ourselves searching his precious face, body and movements for signs and clues, analyzing his every move and word to see if we could pinpoint which type of the disease we were dealing with and how severe and quickly things were progressing.

Jasper was born on January 9, 2009 a healthy 10 lb 3 oz baby boy. We were so happy to bring home our beautiful boy who would complete our family. As a newborn, Jasper was a happy and content baby who slept well and was a joy to be around. About the age of three months we noticed what turned out to be bi-lateral inguinal hernia's. These were repaired by a Paediatric Surgeon and once surgery was over we were happy to have our healthy little boy back. A few months later we noticed a bump on Jasper's lower spine when he would bend over and back we went to our family



Jasper's First Treatment

Doctor to question what it could be. We were referred to an Orthopaedic Surgeon who completed a set of x-rays. After some analysis and a repeat visit 6 months later, we were then referred to the Genetics department and this where our story really begins.

Our first visit with Dr. Goobie was a series of questions and a very thorough exam of Jasper. Following our visit, both blood and urine samples were given. A couple weeks later we were called to London to discuss the results of the tests and this is when our world crashed. The test samples confirmed that Jasper has Mucopolysaccharides Disease (MPS). Never in our wildest imagination did we expect to receive such devastating news about our beautiful little boy. We continued to question how something so rare and serious could affect our family. Through much discussion and explanation she assured us it was.

Personal Stories - continued

This brings us to July in Jasper's journey and after a series of tests including more blood and urine samples, a skin biopsy and DNA mapping it is determined that Jasper in fact has MPS VI. At this point we are introduced to Dr Chitra Prasad who will head up Jasper's care. We learn that Jasper is only the 8th known case of MPS VI in Canada (there are about 70 in the United States and 1100 worldwide) but we also learn that there is a treatment available. Enzyme Replacement Therapy (ERT) using the American made drug Naglazyme. We 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.

Our journey brings us to August 2011 when all the paperwork is filed to apply for funding. There were so many steps to be followed, the claim and supporting paperwork had to be completed and accompanied by letters written by experts in the field, sent to our insurance company for review and to the Ontario Ministry of Health. Initially we were denied funding for treatment and decided to launch a very public appeal and using the media and power of social net-working began advocating to the MOH to fund treatment. With much help and support from family, friends and The Isaac Foundation our appeal application for funding was approved. When we heard this news we were ecstatic! A huge weight had been lifted from our shoulders. Our little boy has been given the chance that every child should have – the chance to grow up.

In October Jasper had Port-a-Cath surgery in order for his treatments to be administered. Surgery went very well and mid-October, with many mixed emotions, Jasper had his first treatment. We now travel to London (a 2 hour drive from our home) once a week for treatment sessions. Treatment sessions are a full day commitment as we spend approximately 6 hours in the hospital and 4 hours travelling. On top of the weekly enzyme replacement therapy we have also had several appointments with specialists in many fields who will monitor and care for Jasper and the many issues that come with an MPS VI diagnosis.

It is now November and when we look back on the roller coaster ride we have been on since that day in April we are amazed at how far we have come. We have had numerous ups and downs in the past few months but continue to be amazed by the support we are receiving from family, friends, co-workers, community, doctors, church and our new MPS family. We feel blessed that there is a treatment option available and hope and pray it will extend the length and quality of Jasper's life. We no longer

Personal Stories - continued

look at his every movement in search of answers and analysis but see our Jasper again. We see ways we can fundraise to find a cure, we see people that we never would have met if it wasn't for this disease and we continually search for ways to make Jasper's journey positive and life changing for everyone he touches.

Please feel free to contact our family at themorefamily@wightman.ca
To keep tabs on us or read further about Jasper's journey please follow the blog we have recently created at www.thelifeofmore.blogspot.com.



Jasper



Jasper and his family



Jasper and his sister & brother

Featured This Issue

Mucopolysaccharidosis VI (MPS VI)

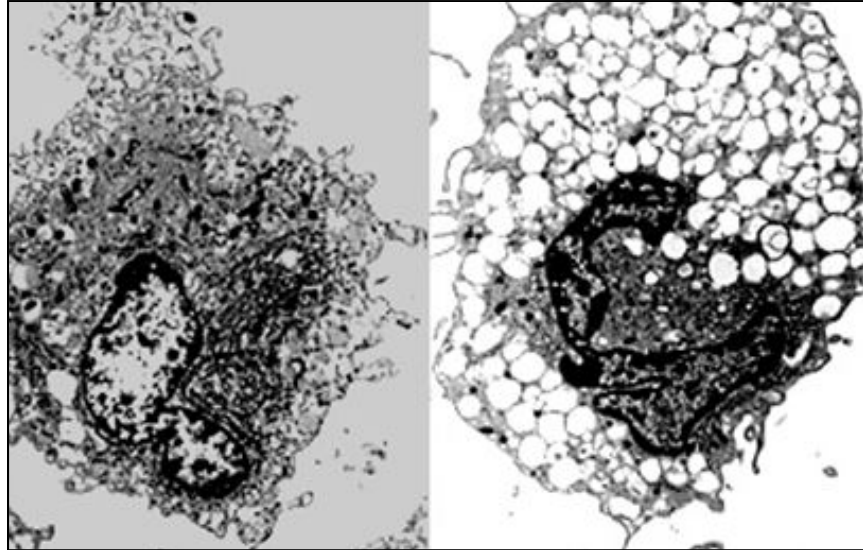
By Chitra Prasad

Mucopolysaccharidosis VI (MPS VI) is also known as Maroteaux-Lamy syndrome. MPS VI is a lysosomal storage disorder. It is a very rare disorder caused by inherited deficiency in the enzyme arylsulfatase B. This enzyme is normally required for the breakdown of certain complex carbohydrates known as glycosaminoglycans (GAGs). If the enzyme is not present in sufficient quantities, the normal breakdown of GAGs is incomplete or blocked. The cell is unable to excrete the GAG residues which then accumulate in the lysosomes of the cell. This accumulation disrupts the cell's normal functioning and gives rise to the physical manifestations of the disease. Approximately 1,100 patients in developed countries have MPS VI. In Canada there are less than ten patients with MPS VI. It is inherited in an autosomal recessive manner, affects males and females equally, and in most cases, both parents of an affected child are asymptomatic carriers of the disease.

MPS VI is a clinically heterogeneous disease with a wide variation in the rate of disease progression, the severity of symptoms, and the organ systems affected. Unlike MPS I, MPS VI does not typically affect intelligence level. Over time the disease progresses, and depending on the degree of enzyme deficiency, patients experience severe disabilities and possibly early death. Some of the features of the disease include short stature, large head, progressively coarse facial features, corneal clouding, recurrent ear infections, impaired hearing, sleep apnea, reduced pulmonary function, cardiac abnormalities and valvular disease, hepatosplenomegaly, umbilical and inguinal hernias, reduced joint range of motion and dysostosis multiplex (bone deformities).

Historically, treatment of MPS VI has been limited primarily to palliative care that addresses the multi-systemic symptoms of the disease. Today, however, there is an approved therapeutic option specifically for the treatment of MPS VI - enzyme replacement therapy with Naglazyme[®] (galsulfase). Clearly making the diagnosis earlier is very important. The enzyme therapy is given once every week over a period of 4 hours. We hope that continued research in this condition will help improve the health status of Jasper and many other individuals affected with MPS VI.

Featured This Issue - continued



MPS VI is a lysosomal storage disorder. Excess GAG accumulate in lysosomes, where they would normally be degraded. The photo on the right shows lysosomes engorged with GAG.

Information on Resources

B:OMARIN[®]

Matching proven science with proven needs

<http://www.bmrn.com/patients-physicians/mps-vi.php>

<http://www.maroteaux-lamy.com/English/HCP/Resources.aspx>

<http://www.maroteaux-lamy.com/English/HCP/BiochemBasis.aspx>

Suzanne's Corner



Anthony's Favorite Meat*Less*Balls



(serve with any pasta dish)

- | | |
|---------------------------------|---|
| 1 c. chopped zucchini | 1 tsp. garlic powder |
| 1/4 c. chopped button mushrooms | 1 tsp. garlic salt |
| 2 T. minced onion | 1/2 c. Success instant brown rice
(cooked) |
| 1 large egg yolk | 1 T. fresh parsley (minced) |
| 1 c. lo pro bread crumbs | |

Notes: Total Phe (phenylalanine) per recipe = 284 mg Phe, 6.4 grams protein and 258 calories.

In a medium sized sauce pan boil 4 cups water and cook rice. In a medium size mixing bowl add all ingredients. Mix well (using hands). Shape small balls. Place balls in a lightly coated fry pan (using canola oil or grape seed oil) over medium heat, cook until golden brown. In a preheated oven 350 degrees, place meat*less*balls on a cookie sheet or a pizza pan and bake for an additional 15 minutes. Top on your favorite pasta dish and enjoy!!!

The Inherited Metabolic Disorders News

What's New



GHOSTS & GOBLINS !!!



Carver

Kennady



Jasper



Research / Talks/ Presentations/ Meetings

1. Mhanni AA, Prasad C, Rockman-Greenberg C. Ornithine transcarbamylase deficiency presenting as recurrent abdominal pain in childhood. *Pediatr Emerg Care*. 2011 Sep;27(9):850-3.
2. Prasad AN, Rupar CA, Prasad C. Methylenetetrahydrofolate reductase (MTHFR) deficiency and infantile epilepsy. *Brain Dev*. 2011 Oct;33(9):758-69. Epub 2011 Jul 22.
3. Prasad AN, Levin S, Rupar CA, Prasad C. Menkes disease and infantile epilepsy. *Brain Dev*. 2011 Sep 15. [Epub ahead of print]
4. Prasad C, Rupar T, Prasad AN. Pyruvate dehydrogenase deficiency and epilepsy. *Brain Dev*. 2011 Sep 9. [Epub ahead of print]
5. Khan A, Mackrell M, Maranda B, Melancon S, Patterson MA, Poulos V, Prasad C, Raiman J, Sirrs SM, Stockler S, Canadian NP-C Guidelines. Calgary, Canada. (June) 2011
6. Death and Dying in Childhood. Experiences from the neurogenetic/neurometabolic clinic and Palliative care at Children's Hospital LHSC. Narayan Prasad, Lisa Pearlman and Chitra Prasad
7. Iyer H Sen M, Prasad C, Rupar CA, Lindsay RM. Coma, hyperammonemia, metabolic acidosis, and mutation: Lessons learned in the acute management of late onset urea cycle disorders. *Hemodial Int*. 2011 Sep 19. doi: 10.1111/j.1542-4758.2011.00591.x.
8. Ratko,S. Dietetians in Metabolics, Toronto, November 2011.

5 Tips for Mental Health

1. Build Confidence

Identify your abilities and weaknesses together, accept them build on them and do the best with what you have.

2. Eat right, Keep fit

A balanced diet, exercise and rest can help you to reduce stress and enjoy life.

3. Make Time for Family and Friends

These relationships need to be nurtured; if taken for granted they will not be there to share life's joys and sorrows.

4. Give and Accept Support

Friends and family relationships thrive when they are "put to the test".

5. Create a Meaningful Budget

Financial problems cause stress. Over-spending on our "wants" instead of our "needs" is often the culprit.

Accomplishments

Tammy Beadle, age 34

Diagnosis—Ornithine Transcarbamylase Deficiency (OTC)

26 YEARS OF RIDING AT SARI!

Tammy Beadle was only 8 years old when she mounted her first pony named Gretel at SARI. Tammy's Mother Sandy remembers those first lessons when Tammy was so tiny that her legs stuck straight out across Gretel's chubby little frame! Tammy is now 34 years old and is *still* riding at SARI!

As a child, Tammy was very weak and had poor muscle tone. Her doctor recommended therapeutic riding for her as a form of physiotherapy. As a result of her regular riding Tammy's legs are stronger. She is involved in other activities but none for as long as she has stayed with SARI. Riding is part of a regime to keep Tammy's mind and body active; it keeps her healthy.

Tammy keeps coming back because she loves the horses and she loves the people. Over the years she has come to know many different volunteers and Instructors. She likes to socialize, she likes learning and she likes the environment.

Her Mother has asked her on many occasions if she'd like to give up riding and Tammy's response is "No way"!



A highlight of Tammy's riding at SARI was performing in 1990 at the Canadian National Exhibition pictured in Toronto when she came home with a ribbon.

From the November 2011 SARI newsletter:

Kareem Ghadban, age 9

Diagnosis - Niemann Pick Type-B

Kareem's Arctic Quest

On Aug 10, 2011, my self my dad and 8 others made our way up north to a small town called Pangnirtung. This place is in Nunavut and is so remote that you can only get there by plane or boat. It took 8 hrs to get there from London and 3 plane trips. Once we got there we stayed at a lodge for a few days and had got a chance to see some things in the town. I also went on a boat trip to see some whales and ice bergs, it was amazing. The land up north is all mountains and dirt, there are no trees and grass like we have here in London. On Aug 15th we packed all our bags in the early morning

Accomplishments - continued

(5am) and took a 1 hr boat ride to the edge of Auyittug national park. When we got there we had to climb big rocks to get to the land and then we started to trek through the spongy land, sand , rocks, mountain side cliffs, It was amazing. I started to get tired near the end of the day after we went 6 km. Day 1 we set up camp and had a great dinner and I went right to bed. Day 2 in the morning we headed out to our goal of the arctic circle and almost made it there but we fell short going 8 km and setting up camp. That day was long and hard as the land was very rough and we had to cross a lot of glacier water streams that were to high for me so my dad carried me across. On Day 3 we were determined to make it to the arctic circle and we did trekking 7.5 km there .. it was amazing I was the first to cross the line in the group and was so happy I made it. We had lunch and then headed back. That day we trekked 15 km, for a small guy like me that was a lot , but I didn't feel that tired...

Day 4 we started to head back and we took the same route we did in coming. We got back to the drop spot and the boat was there to pick us up. Some things I made note of:

1. The weather was not as cold it was 5 to 15 deg and sunny every day except when a cold arctic breeze came by.
2. The terrain was not what I expected it was rough and a lot of different things we had to go through that 42 km is like walking 50 km on a paved street.
3. It was always sunny, because we are so north the days are long in the summer sun set was 12:15 am and sun rise was 2 am, but I slept well.
4. No Trees or grass, it was all dirt and distance is a problem because we don't know how far things are, everything is so big.
5. No animals, the only thing I seen was an arctic bunny in the park.
6. Water tide drops and rises 10 feet in 3 hrs.

On the last day we had a traditional Inuit dinner it was great we had Carabobo stew and I also tried raw seal... yuck

So why go to all this trouble, I wanted to raise awareness of my disorder called Neiman pick Type B and show that anyone with a disorder can do anything ..

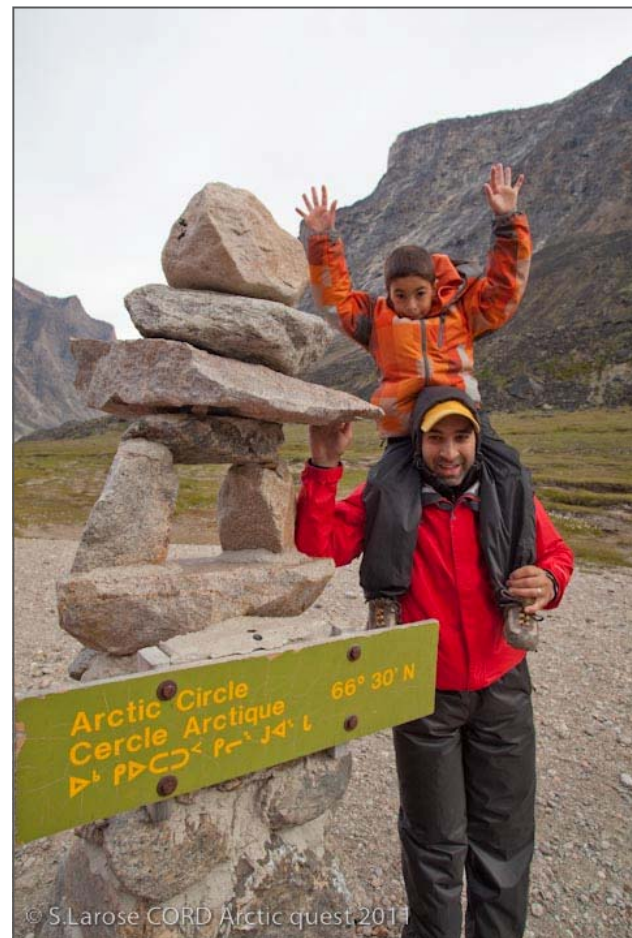
Thanks to all the people who helped me get to my goal and journey

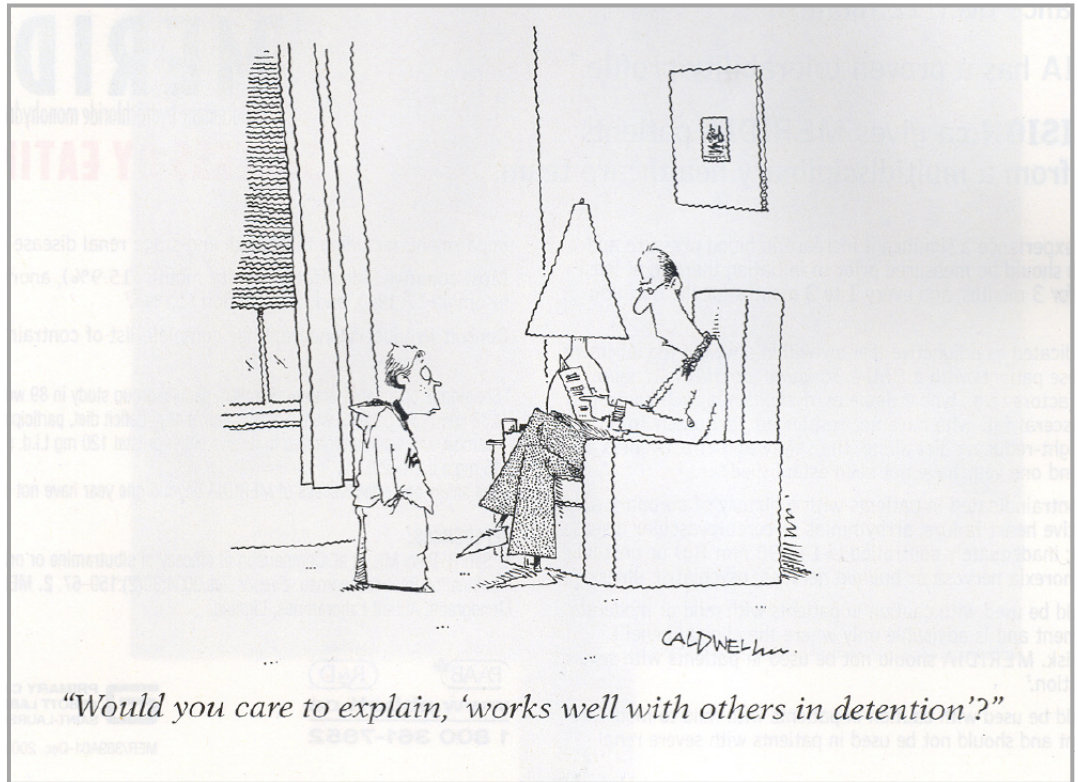
One more note, as per parks Canada , I'm the youngest person to trek to the Arctic Circle at 8 years 8 months

Kareem Ghadban

The Inherited Metabolic Disorders News

Accomplishments - continued





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!



**Elvina,
Age 10**

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