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From the Editor

It's hard to believe that this is the 10th year of the *Inherited Metabolic News!* Time has just flown by!

As always, your stories and suggestions are welcome! Enjoy!
Janice Little

From Dr Chitra Prasad

Dear Friends,

I would like to wish each and every one of you happy holidays and a very Happy New Year.

There have been lot of new research developments in our clinic. We have finished 2 research studies (on PKU 016 and Psychosocial aspects of inborn errors of metabolism). Thanks to all those who participated. I would like to take this opportunity to also thank you for your generosity in helping our junior residents and fellows learn.

We are delighted to have our new social worker "Kim". Some of you have already met her.

May 10th 2013 is our next metabolic family workshop. This will be the 10th anniversary!

We would love to have you join us on this milestone.

The metabolic family workshop has been a labour of love where so many people come together, bring their ideas and then devote their time and energy to make it a grand success each year.

My sincere thanks to the parent representatives and our entire metabolic team for their hard work and vision for the workshop.

In this issue Shane a gentleman with Fabry disease has shared his experience of how his diagnosis was reached. There are so many people who get diagnosed with inborn errors of metabolism in adulthood. Educating families and the professionals will help in earlier diagnosis and help in management.

Finally, a quote from Margaret Mead -

Never doubt that a small group of thoughtful, committed citizens can change the world. Indeed, it is the only thing that ever has

With best wishes

Your friend
Chitra Prasad

Personal Stories

Hello, my name is Shane and I would like to tell about my experience with Fabry disease. This is a hereditary condition, so I had it all my life but only found out about it at the age of 35.

My wife Karen and I left our home in Pennsylvania in 1998 to teach at a school for missionary children in Indonesia. After three years, we had to exit the country in order to renew our visa. We went to Thailand to do the paperwork since we had friends there we wanted to visit. It was pretty hot in Thailand and I noticed my feet were quite swollen. I did not think too much about it, but during the next year in Indonesia the swelling became more common. We moved to Canada in 2002 and went through a two-year missionary training course in Durham, Ontario, after which we stayed for 3 more years as teachers/trainers. During this time the swelling in my feet became more and more persistent until it became noticeable even in the winter.

We planned to return to Indonesia in 2007 but felt that before doing so we should really determine what was causing the swelling (edema). I went to our family physician around March that year. He referred me to a tropical medicine specialist because of our time overseas. This doctor took one look at my legs and determined that the cause of the swelling was poor blood return due to faulty valves in my veins. When I went back to see our family physician, he was away and a different doctor looked at my lab results and commented that my kidney function was pathetic. He said I could expect a kidney transplant in 20 years. He sent me on to Dr. Garg, a nephrologist at LHSC (Victoria Hospital). He ordered a number of tests and sent me to a cardiologist. By this time fall had arrived and we were getting ready to leave for Indonesia.

I had an appointment with Dr. Garg sometime in October and he took a kidney biopsy. The results of that biopsy indicated a strong possibility of Fabry disease, so he set up one more appointment for a blood test specifically for this condition. He asked us to consider postponing our trip, but we had already purchased the tickets. A few days later, we flew to Indonesia not knowing the results of the test. About a month after arriving in Java, Dr. Garg called and said that he was sad to inform me that I did, in fact, have Fabry disease. He suggested that it would be better if I returned to Canada. However, we were still convinced that we were supposed to be in Indonesia. I began going for blood work and urinalysis every two weeks to keep an eye on my creatine levels (a marker of kidney function). After several months of this, it was apparent that my kidney function was not deteriorating and Dr. Garg said I could have the lab work once a month for monitoring. We stayed in Java for the next 4 years and sent monthly lab results back to Dr. Garg.

Personal Stories- continued



In November 2011, we returned to Canada and immediately got started with doctor visits and lab work with the intent of starting the Enzyme Replacement Therapy (ERT) as soon as possible. I had appointments with different doctors and had many different tests. During this time, primary responsibility for my treatment was transferred to Dr. Prasad in the Metabolic Clinic. It took quite a long time before I was able to start the

ERT, but the day finally came – May 23, 2012.

Every two weeks I traveled to the IV clinic at LHSC for an ERT infusion, each time being given Benadryl and Tylenol to minimize any allergic reaction before the infusion is started. I have now had eleven of these treatments; the first eight were administered at the IV clinic and the last three at my home. A nurse comes to the house for each infusion. Thus far I have not experienced any negative reactions. Most of my symptoms have changed little: the swelling, lack of sweat, intolerance of temperature extremes, and angiokeratomes (the little red dots on the skin south of my bellybutton). My intermittent IBS has been noticeably more rare and it's been a while since I noticed any of the little "phantom" stinging in my hands. I am very thankful for this treatment and for the excellent care I have received. I appreciate Dr. Garg's continued interest and concern while we were overseas and how he immediately scheduled an appointment upon my return. I appreciate the way in which Dr. Prasad and Melanie Napier of the Metabolic Clinic have taken a personal interest and have been so helpful every step of the way. When I consider all the different specialists I have seen and all the tests that have been done, I realize how very fortunate we are to live in Canada and to have so very much in the way of medical care. Truly we have much for which to be thankful.

Lastly, I am thankful to my Creator whose care for me has never faltered. I happened to be memorizing Psalms 139 when Dr. Garg first called us in Java to say that I did have Fabry disease. The whole psalm is wonderful, but verses 13 and 14 were especially meaningful: "For you created my inmost being; you knit me together in my mother's womb. I praise you because I am fearfully and wonderfully made: your works are wonderful; I know that full well." The phrase "inmost being" in verse 13 comes from the Hebrew word for kidneys. That stood out to me because it was my kidneys that seemed most at risk at that time. I did not know about it until I was 35; He knew before I was even born.

Featured This Issue

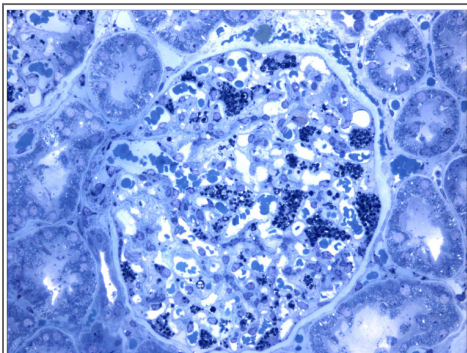
Fabry Disease

Compiled by Dr. Chitra Prasad MD FRCPC FCCMG

Fabry Disease is one of several dozen Lysosomal Storage Disorders that interfere with the body's ability to break down specific fatty substances. Fabry patients are missing or lack sufficient a-gal A enzyme which results in sugars and fatty acids (Gb3) accumulating in the cells throughout the body and impairs the function of several major organs including the kidney and heart. This can become a major problem in parts of the body that depend on small blood vessels, since the built-up Gb3 can clog these vessels. The areas that are most affected by the closing of small blood vessels are the kidneys, heart, nervous system, skin and inner ear. It is a very rare disease and because the rate of occurrence is less than 1 in 200,000 is considered one of the many "orphan" diseases. The diagnosis of Fabry Disease in one family member may lead to the evaluation and diagnosis in other relatives, as Fabry disease is caused by a faulty gene in the X-chromosome. Men have an X and a Y-chromosome whereas women have two X-chromosomes. This is why men generally suffer more than women although there are cases where women also have all the symptoms. Blood tests confirm the level of a-gal A enzyme in the blood and DNA studies are used to confirm the diagnosis. Care should be exercised in diagnosing females as the level of the a-gal enzyme is not a true indicator of Fabry Disease and they should be checked for the specific gene mutation. Enzyme Replacement Therapy (ERT) was approved in Canada in January 2004.

ERT requires an intravenous infusion once every two weeks and may be done in a hospital although some patients eventually receive infusion at home. There are two enzyme replacement therapies Replagal and Fabrazyme. Research is ongoing for new therapies (oral, gene therapy). Canadian Fabry Disease Initiative led by government and the enzyme therapy companies enables many Canadian Fabry patients to receive therapy as well have their data monitored.

Ref: <http://www.fabrycanada.com/Fabry-Disease-an-Orphan-Disease.html>



Lysosomal inclusions in kidney in Fabry Disease

Suzanne's Corner



Polish Cabbage and Noodles

Category > Main Dishes

1 medium head of cabbage (shredded)
(about 4 cups)

1 c. sweet red onion (cut into strips)

1/2 c. butter or margarine

1 lb. aproten tagliatelle pasta noodles

Sea salt and fresh ground black pepper



Cook pasta (al dente) in a large pot of boiling salted water. Heat butter in a skillet over medium heat. Sauté cabbage and onions until tender. Drain pasta and return it to pot. Add cabbage and onion mixture and toss gently. Add salt and pepper to taste. Serve immediately.

Enjoy!!!

Notes: Total Phe (phenylalanine) per recipe = 277 mg Phe, 9.1 grams protein and 2748 calories.

From www.pku.com under [Main Dishes](#)



What's New

Ottawa Marathon



In May of this year, we traveled to Ottawa to take part in the 5k event of the Ottawa Marathon. We did this as fundraising venture combined with the Isaac Foundation. Our team of family and friends were a small percentage of the more than 42,000 participants in the weekend long event but were able to raise just over \$9,000 in donations and combined, the Isaac Foundation raised just over \$18,000, 100% of which will be spent on research grants, searching for a cure for MPS VI and MPS in general.

We have joined forces with the Isaac foundation in the effort to find a cure for our boys, and everyone else affected by this disease. If anyone would like more info or to donate or get

involved, please feel free to contact me by email darren@theisaacfoundation.com anytime, for any reason.

Thank you. The More Family.



The Isaac Foundation

Researching a cure for MPS VI
www.theisaacfoundation.com

What's New- continued



10th Anniversary



Metabolic Family Workshop



Friday, May 10, 2013
Best Western Lamplighter
591 Wellington Road London, ON



Low Protein Cooking Demonstration

Saturday, May 11, 2013
Loblaw on Wonderland
3040 Wonderland Road South London, ON

For further information, contact Janice Little at 519-685-8453 or
Joanne Psiuk-Rodgers at 519-685-8500 Ext 56131



Metabolic Family Workshop

Friday, May 10, 2013

Best Western Lamplighter

Number Attending: _____

Contact Name: _____

Phone Number: _____

Workshop Attending: (indicate number of persons)

Diet Teen Other Metabolic Disorders Lysosomal

Low Protein Cooking Demonstration

Saturday, May 11, 2013

Number Attending: _____

Please return above information to:

Janice Little janice.little@lhsc.on.ca Tel : 519-685-8453
LHSC Victoria Hospital Fax: 519.685.8214
800 Commissioners Rd E London, ON N6A 5W9



*Please send this page back to pre-register for the Metabolic Family Workshop
and/ or the Low Protein Cooking Demonstration*

What's New- continued

Bethany's Hope Foundation's 1st Annual Project Hope - 5K Walk, Run and Roll was held in Harris Park on Saturday, October 13th, with close to 250 participants joining in to help 'move to the CURE!' With the support of numerous generous sponsors, remarkable volunteers and a special \$10,000.00 donation from our friends - Lori and Renee - with Athena's Hope, \$40,000.00 dollars was raised to help find the cure for Metachromatic Leukodystrophy (MLD). Money raised will support world-class research being conducted under the direction of Dr. Tony Rupar at the Bethanys Hope Leukodystrophy Research Laboratory in conjunction with Western University.

For more information, visit www.bethanyshope.org



Running Factory Charity Challenge 8km road race SUNDAY SEPT. 16, 2012

Matt, Blaire and Karson were given the opportunity to enter their own charity (the Children's Health Foundation).

Through this charity, Matt and Blaire were able to donate directly to the PKU and Metabolic workshop fund. This fund helps Karson and the other kids with PKU and other metabolic disorders at London Health Sciences Centre



What's New- continued

New Metabolic Team Member



Hello! My name is Kim Tiemens and I am the new social worker on the Metabolics and Neurology teams. I have a Masters of Social Work degree from the University of Calgary. I am new to LHSC, however I have been working in paediatric medical social work for approximately 10 years. I work part-time and can be reached at 519-685-8500 x56149. I look forward to the opportunity to meet many of you during

your clinic appointments. Please feel free to call if you feel there is a way social worker can support you on your journey.

Research accomplishments

1. PKU 016 study completed (Dr. Chitra Prasad, Melanie Napier, Suzanne Ratko, Dr. Erica Gold, Pharmacy and Research lab).
2. Chitra Prasad, Marina I. Salvadori and C. A. Rugar. Severe Phenotypic Spectrum of Mevalonate Kinase Deficiency with Minimal Mevalonic Aciduria. *Molecular Genetics and Metabolism*
3. Cartagena A, Prasad AN, Rugar CA, Strong M, Tuchman M, Ah Mew N and Prasad C. Recurrent Encephalopathy: NAGS (N-acetylglutamate synthase) Deficiency in Adults. *Canadian Journal of neurological Sciences*.
4. Potter B, Khangura S, Chakraborty P, Feigenbaum A, Hernandez M, Little J, Mitchell J, Prasad C, Rockman-Greenberg C, Wilson B, on behalf of the Canadian Inherited Metabolic Diseases Research Network (CIMDRN) The Canadian Inherited Metabolic Diseases Research Network (CIMDRN). Joint ICBDSR and CCASN Annual Scientific Meeting, Ottawa November 12
5. Canadian Association of Genetic Counsellors Annual Education Conference - October 2012 - Saskatoon (Presentation: Metabolic Genetic Counselling by Melanie Napier)
6. Exploring the current and future state of ERT for Gaucher Disease - October 2012 - Toronto (Melanie Napier)

“Our Kids”



Dean's brother Lucas and Jasper More

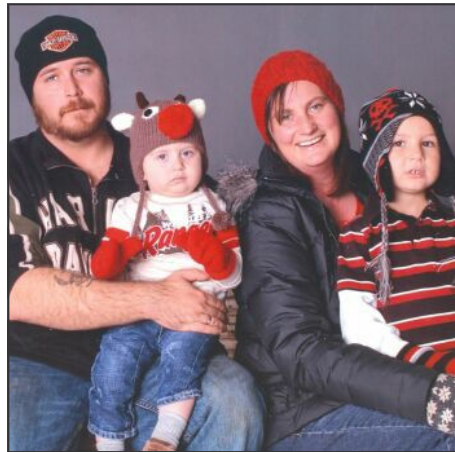
"A treasured gift" in
memory of Dean Stickle



Dean 2009



Felix Deschênes



Carver Hummell's
Family



Cole Lucas



Charlotte Morris and her
parents



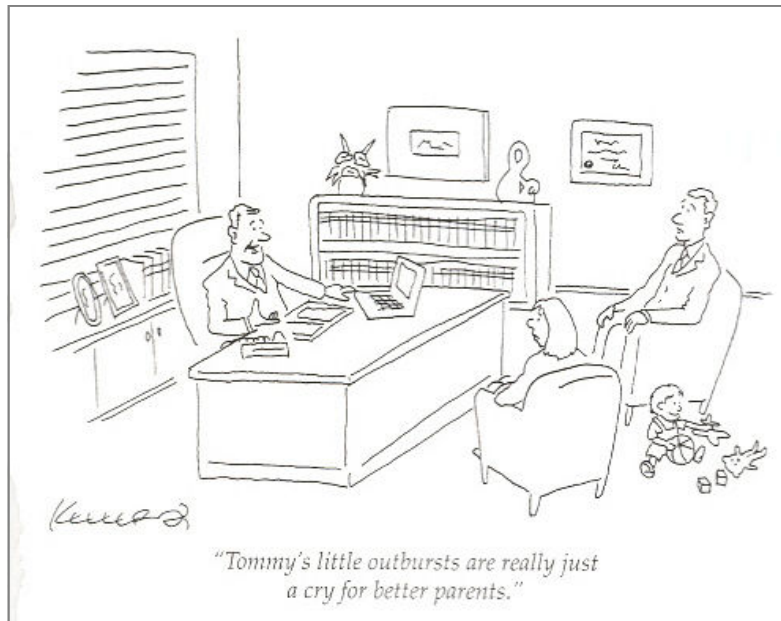
Michael McManaman



Elvis Meets Dr. Prasad !!!
Jasper More

Art of Extreme Care

- ◆ Do one brave thing everyday
 - ◆ Play
- ◆ Treat yourself to something
- ◆ Wear something that makes you feel beautiful or handsome and confident
 - ◆ Have an “All Day PJs Day”



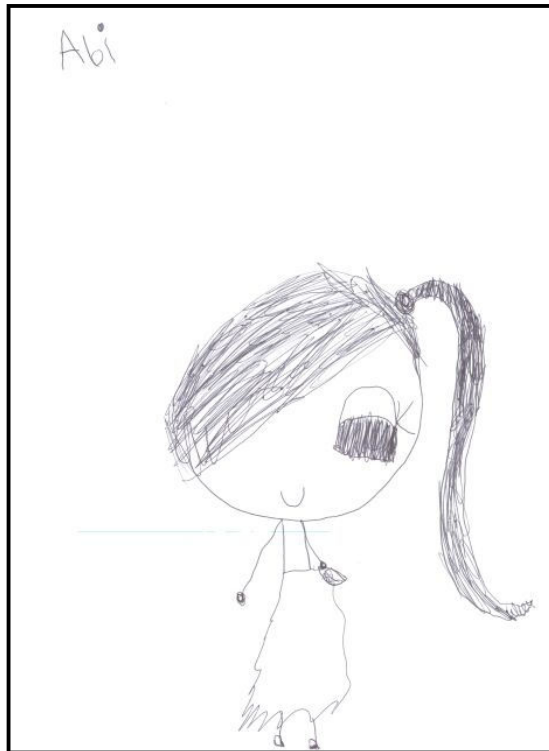
How to Make a Donation

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!



**Abi, age 7
PKU**

Contact Information

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Parent Support Contact: Jennifer Culp

Tel: 1.519.632.9924

Email: jennc2011@hotmail.ca