The Inherited Metabolic Disorders News

Children's Hospital
London Health Sciences Centre

Winter 2010 Volume 7 Issue 1

In this Issue

From the Editor

Greetings Everyone!

It's hard to believe the *Inherited Metabolic Disorders News* is in it's 7th year of publication! We hope you find the information helpful. As always, we welcome your input and stories.

Janice Little

From Dr Chitra Prasad

Dear all,

Wishing you a very Happy New Year!

We bring you another newsletter with stories of triumphs and challenges.
In this issue you will read about a very courageous family dealing with the uncertainties of a diagnosis of Metachromatic Leukodystrophy in their son.
Dr. Rupar has summarized some of the information on Metachromatic Leukodystrophy. His laboratory has been at the forefront for research on this condition.
Planning is underway for our 7th metabolic family workshop. We have a great line up of speakers. Please read about the details of the workshop on the 14th May 2010 and mark the day in your calendars. A number of families are now receiving the newsletters. We would love to receive your feedback, suggestions and contributions.

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From Dr Chitra Prasad - continued

A few months back I was honoured to give a talk and workshop on pediatric metabolic disorders in the Post Gradate Institute of Medical Education and Research Chandigarh India. This is the institute where I trained in Pediatrics before coming to Canada. Chandigarh is a very beautiful city. It was built by a French architect Le Corbusier. Chandigarh has a unique rock garden, with no similar edifice found in any other place in the world. It consists of the artwork that has been made from industrial and urban waste. Working in secret for over 18 years, Nek Chand the untutored genius built a paradise and created a gigantic 12-acre series of palaces, pavilions, and courtyards, all filled with his unique rock sculptures.
Here is a picture from this unique garden.

With best wishes

Your friend
Chitra Prasad

To accomplish great things, we must dream as well as act.

Anatole France
French novelist (1844 - 1924)

"Swans" in the Rock Garden

Personal Stories

Hi there,

I am writing this letter to tell you our story and journey as a family. Our youngest son, Christopher was diagnosed last Sept 25th, 2008 with Metachromatic Leukodystrophy. Christopher was born a healthy baby boy, August 18th, 2000. He weighed 8lbs 5oz. I breastfed him until he was almost 2 years old, then he went straight to a sippy cup. He was an active toddler, liked to play with his older brother and the neighbour children….he learned to ride his two wheeler with no training wheels at age 4, he would zoom up and down the sidewalk.

We moved to a new neighborhood when he was 5. He started grade 1, and that's when we noticed something was a little different. Christopher had a little tremor in his arm; we thought it was just nerves with starting a new school….then we started to do strength exercises with him for his fine motor skills. As the year passed with not much change, grade 2 approached and his teacher had noticed his walk was a little different. At that point we had noticed a little bit that Christopher's attention wasn't all that great…so we had him tested for Central Auditory Processing, that test came back normal. He started to let out a little squeal and banging a lot on things…..We saw a psychologist and he told us that Christopher had signs of Tourette syndrome.

We and our family doctor thought we would try ADD medicine but it put Christopher into a drugged out state…no personality and not the fun little boy he usually is…..so we took him off the medication after 1 week. Our family doctor told us that it was just behavioral. I felt there was no way, as he is a well behaved child, a mother seemed to know her child. I would come home and tell my husband, maybe it's this, maybe it's that? My husband was also unsure and we were both unclear as to what was happening to our son. Sad to say, we were getting frustrated with Christopher and thought that he was just being lazy as he wouldn't tie his shoes anymore or dress himself.

The breaking point was that Christopher couldn't get down on the floor or ride his bike or swim anymore.

I went to a Pediatrician and told him my thoughts and that I wanted a CT scan, as something just wasn't right or connecting properly. He approved and that's when the devastating news came to us…Christopher then had a MRI and skin biopsy to confirm it all.

Personal Stories - continued

The hardest part of all of this was telling his older brother as they are so close! He took it very well, 3 weeks later we took the kids to Disney World as we had no clue how long Christopher had to live as we had never really heard much about the disease.

Since the diagnosis, 14 months ago, Christopher is now incontinent and spends 90% of time in a wheel chair. His vocabulary is very limited as well. He needs leg braces now, as his left foot has started to tighten in and causes him some pain and doesn't help with his balance and standing. We have noticed a little problem with his swallowing too and we have found that he has become quite disinhibited….He also puts everything in his mouth, just like a baby again! We have been told that all these are due to progression in his disease. He is still in school with 2 full time EA's and is overall a happy 9 yr old boy. He loves music and Hannah Montana! It has been a growing experience and a lot of stress on the family, but we all work together and have had some great help and friends. We are looking forward to going to Disney again in Jan 2010 through Make A Wish.

Thank you for sharing our life!
Janice, Mike, Michael and Christopher Thompson

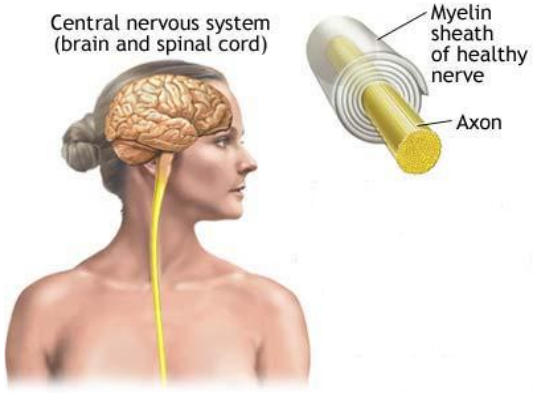
Featured This Issue

Metachromatic Leukodystrophy

Dr. Tony Rupar, PhD, FCCMG, Director, Biochemical Genetics Laboratory and Bethanys Hope Leukodystrophy Research Laboratory, London Health Sciences Centre and the University of Western Ontario

Metachromatic leukodystrophy (MLD) is a progressive neurodegenerative disorder. As you will read in Christopher's story, a baby born with MLD is well at birth but later in childhood, usually about the second year of life but often later, the child stops gaining new skills and starts to lose existing ones. Abilities like walking and swallowing are lost. Behavior and learning abilities deteriorate and the child becomes totally dependent on others for care. These are all symptoms of the deterioration of both the central and peripheral nervous systems

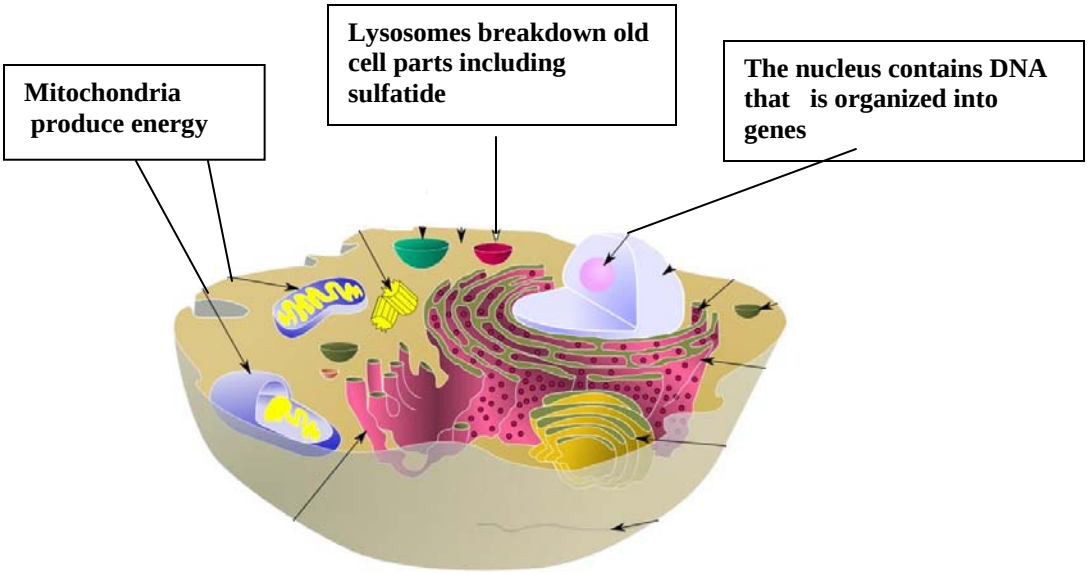
MLD is caused by the loss of the ability to breakdown a fatty substance, sulfatide that is present in nervous tissue. The figure below illustrates the role of sulfatide in forming the myelin or white matter around nerves.



Sulfatide is a component of the myelin sheath that wraps around nerves in "onion –like "layers providing insulation to the nerves that transmit electrical impulses required for thought and movement and also some nutrition to nerves.

In all of us components of cells are constantly being renewed. Old structures that may be damaged are replaced with new structures. In this renewal process, old sulfatide is transported to lysosomes for degradation. This is accomplished by the lysosomal enzyme arylsulfatase A (ARSA). Individuals with MLD lack functional ARSA and are unable to break down sulfatide. Sulfatide accumulates in lysosomes and lysosome-derived vesicles and ultimately results in cell death, tissue damage and the disease MLD.

Featured This Issue



A diagram of a typical cell. Imagine this cell as a hard boiled egg that has been cracked across the middle to show the inside parts. There are many parts to a cell but only three are identified in this illustration. Lysosomes in cells in individuals with MLD and other lysosomal storage diseases accumulate undegraded substances and eventually cause cell death.

Diagram modified from the A.D.A.M. Medical Encyclopedia (http://www.nlm.nih.gov/medlineplus/encyclopedia.html)

MLD is one of a group of more than forty lysosomal storage diseases. The diseases are individually rare but taken together affect about 1 in 5000 newborns. All lysosomal storage diseases have similarities to MLD but differ in the enzyme that is deficient and the material that is stored in lysosomes. Different lysosomal storage diseases affect different tissues. Muscle, liver, spleen, bones, kidneys, heart, as well as brain are all the targets of lysosomal storage diseases.

Supportive treatment to maximize the quality of life helps patients with all types of lysosomal storage diseases. For some diseases including Gaucher disease, Hurler syndrome, Hunter syndrome, Fabry disease, Pompe disease, and Maroteaux Lamy syndrome enzyme replacement therapy is available. In enzyme replacement therapy enzyme is infused intravenously every week or two to replace the enzyme that is deficient in the particular disease.

Featured This Issue

There is no enzyme replacement therapy available for metachromatic leukodystrophy and most lysosomal storage diseases. The brain is a target organ in many lysosomal storage diseases and enzyme replacement therapy has not been successful in treating the brain. Bone marrow and hematopoietic stem cell transplants have been used in many diseases with only modest success. Bethanys Hope Foundation (http://www.bethanyshope.org/), a London based charitable organization is dedicated to helping families with metachromatic leukodystrophy and supporting research to develop a treatment for MLD. More information about research into a treatment for MLD is available at http://www.biochemgenetics.ca/asa/.

On May 14, 2010, the annual metabolic clinic family day will have a session devoted to lysosomal storage diseases. Dr. Julian Raiman from the Hospital for Sick Children in Toronto will be our guest at this session. There are a number of children and adults with a variety of lysosomal storage diseases who attend the metabolic clinic and we encourage you to attend this session.

CPRI Laboratory

Suzanne's Corner

Quick Pasta Sauce Ideas

From the Canadian PKU Society Newsletter, Fall 2004

Herb and Garlic Pasta

Add some butter or margarine to freshly cooked pasta, and then add some dry "herb and garlic" spice. It can be purchased in the spice section of the grocery store. Stir pasta and serve immediately.

Mushroom and Caramelized Onion Pasta Sauce

Dice the mushrooms and onion. Place them in a frying pan with some oil and sauté until mushrooms turn golden brown and onion is soft and golden in colour. Add salt and black pepper to taste and serve on top of pasta.

Tomato and Parsley and Garlic Sauce

Dice 2 ripe tomatoes and place them in a heated frying pan with some oil. Chop the parsley and garlic. Stir tomatoes carefully to avoid burning. Once they soften up and become mushy, add the garlic and stir vigorously to create sauce consistency. Take the pan off the heat and add parsley just before serving.

Pasta With Vegetables

Pull out of the freezer store bought veggie mix and microwave about 1-2 cups (depending on the amount of pasta you're cooking). Then add the veggies to the freshly cooked pasta; add some butter, salt and pepper to taste. Mix well and serve immediately.

What's New

Metabolic Family Workshop and Low Protein Cooking Demonstration 2010

Please watch your mail for the brochures with all the details to be sent out in March

Donation to Neurometabolic Clinic by Chris & Donna Stickles

Dean Stickles was an active healthy baby boy up until he reached 18 months when he was brought to London in a static seizure. After 7 weeks in the hospital with a newly diagnosed terminal mitochondrial disorder, Dean's parents Chris and Donna decided to bring Dean home where he could spend his last days. He was home for 3 months when he succumbed to his illness and passed away peacefully in his parents arms on September 11, 2009. A deep heart felt thank you to Dr. Chitra Prasad, Dr.Nayran Prasad, Lisa Pearlman, Dr. Rupar, and the rest of the Neuro-Metobolic team in their efforts to an early diagnosis which allowed the Stickles family to take Dean home.

Dean Stickles

On October 19, 2009 on what would have been Dean's 2nd birthday, a donation of $18,854 was presented to the Neuro-Metabolic Clinic to aid in providing the same early diagnosis to other families that they provided the Stickles family.

What's New—continued

Metabolic Camp

The 16th annual metabolic camp will be held in Atlanta, Georgia from June 21 - 26, 2010.

This camp offers an awesome chance for young women aged 12 years and up with PKU or MSUD to "live and learn together in a supportive environment".
Check out the website. Deposit deadline is March 8, 2010.

www.metcamp.org

New I Phone Application

For those who have an I Phone, there is a new PKU tracker application. Visit the link **http://www.appstorehq.com/pkutracker-phenylketonurialookupandlog-iphone-55781/app**

Let us know what you think.

Conferences and Research Publications

Chitra Prasad and Narayan Prasad Neurocon Meeting Chandigarh India- Organized workshop on genetic metabolic basis of developmental delay, October, 2009.

Chitra Prasad. Lecture on "Approach to a child with developmental delay: when should we think of genetic and metabolic etiology?" in Neurocon Meeting, Chandigarh, India. October 2009.

Narayan B, Prasad AN, Prasad, C, Kronick J, Ramsay, D, Tay, K. and Rupar, CA. MELAS: Molecular, Pathological and Radiological Correlates. United Mitochondrial Disease Foundation June 2009.

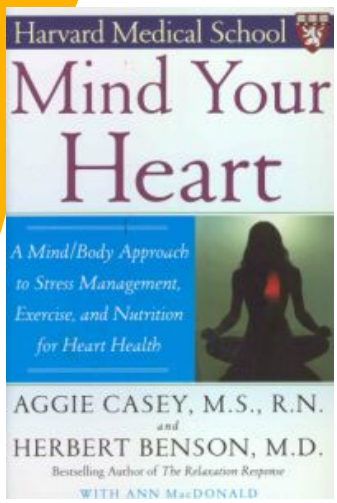
Faghfoury H, Feigenbaum A, Blaser S, Prasad C, Donner E, Hahn A, Rupar CA, Crow Y.AICARDI-GOUTIERES syndrome may masquerade as a metabolic disease. SSIEM. September 2009.

C. Prasad, J. DiRaimo, P. Chakraborty, S. Goobie and C.A Rupar. Positive Newborn Screening for Carnitine Transporter Deficiency: A Marker for Maternal Genetic/Metabolic Diseases? American Society of Human Genetics, October 2009.

PKU & Acute Lymphoblastic Leukemia (ALL): Challenges in Management!. Ratko S, Prasad C, Rupar CA. Conference on PKU, Munich, Germany, January 2010 (accepted).

Workshop on Newborn screening was held at LHSC by Children's Hospital of Eastern Ontario in November 2009.

Mind Your Heart



Here are a few helpful tips to keep your heart healthy from the book "Mind Your Heart"

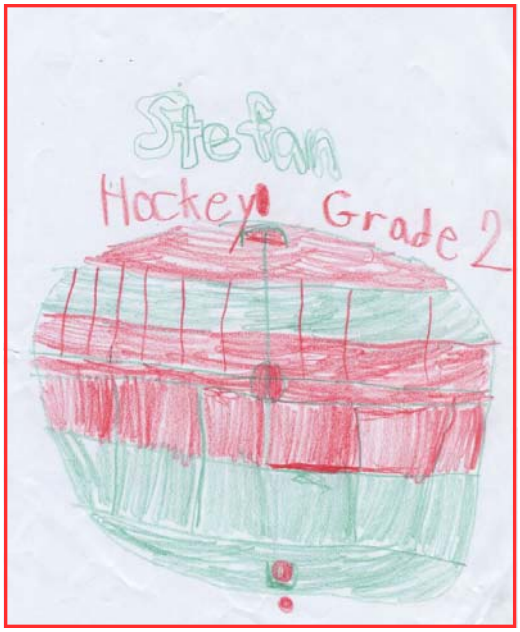
- **Breathe before speaking; then you won't interrupt or finish other people's sentences.**
- **Be flexible with change; things don't always go as planned.**
- **Say a short prayer or observe a moment of silence before meals.**
- **Practice an affirmation each day**
  - **I am relaxed.**
  - **One day at a time.**
  - **I am doing the best I can.**
  - **I am healthy and strong.**

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, you may send a cheque to:

The Children's Health Foundation C/O Medical Genetics Program of Southwestern Ontario
Attention: Janice Little
800 Commissioners Road East
London, Ontario, N6A 4G5
Charity # 118852482 RR0001
Your donation is tax deductible, and an income tax receipt will be mailed to you.

Thank you!



Stefan, Age 7
Morquios
Stefan's story was featured in the Winter 2007 Issue

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