The Inherited Metabolic Disorders News

Children's Hospital
London Health Sciences Centre

Winter 2011 **Volume 8 Issue 1**

In this Issue

From the Editor

Seasons Greetings to everyone! I hope you enjoy the latest edition of "*The Inherited Metabolic Disorders News".* Please note that information for the Metabolic Family Day on April 8th, 2011 and a response form for early registration for the are inside the newsletter. Because it is so early this year, we are asking for confirmation of attendance as soon as possible. We hope that you can attend.

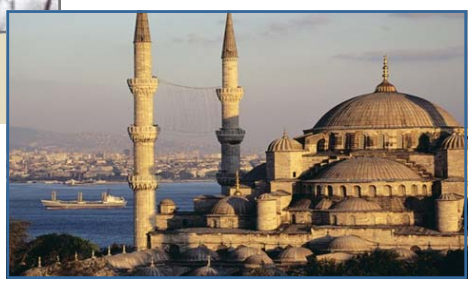
Janice Little

From Dr Chitra Prasad

Dear Friends,
This year we have had lots of snow which always reminds me about my first winter in Canada in St. John's Newfoundland. It was breathtakingly beautiful. Driving in snow of course was another matter. My travels took me to Turkey, Taiwan and India this year. Turkey is an amazing country with the old and new all in one place. Istanbul is a beautiful vibrant city with lots of old churches and mosques.
Lots of new and exciting developments are happening in our metabolic world. I would encourage you to read about "Faith's story" in this newsletter. Faith received a cord cell transplant for management of Krabbe disease in the first few weeks of life and we hope she will continue to show improvement. Her parents have shown amazing strength and resilience through their time of adversity. The Hospital of Sick Kids Toronto transplant team has been very helpful to the Jones family and many others.
You will get to read about some of our new projects and new research initiatives. Our metabolic team continues to work hard as always. I remain very grateful for their help.
We hope you will register early for the metabolic family workshop and the low protein cooking demonstration. The planning committee has spent a lot of hours to ensure that 2011 metabolic family workshop is a grand success. Your input and suggestions are always welcome.
A Merry Christmas, Happy Hanukkah and a very Happy New Year to all!

Chitra Prasad

There are only two ways to live your life. One is as though nothing is a miracle. The other is as if everything is!
Albert Einstein



Blue Mosque (Istanbul)

Aya Triada Church in Istanbul

Personal Stories

The Jones Family:

Krabbe Disease is taking precious infant lives one by one. In October, 2005, we lost our darling first born little girl, Alicia Leigh Jones to this disease. She was only 13 ½ months old when Krabbe stole her away from us. Our second daughter, Megan Marie, was born in 2008. She was perfectly normal and very healthy. On Wednesday, October 14, 2009, our third daughter was born. Faith Ann Jones was born at exactly 12:43pm. She entered this world through a C-section two weeks earlier then the due date. Faith needed to be born early so the doctors could start her battery of tests and Chemotherapy and prepare her for a Cord Blood transplant. She endured 11 days of intense Chemotherapy. Faith also had a central line put into her chest. First time it came out when it was flushed. She had a second one put in, which also came out. Poor Faith received a third line in her right bicep. Thankfully this one stays till she didn't need it anymore. It's really hard to believe just how much a baby can withstand.

Faith, 7 weeks old

On November 12, 2010, Faith received her Cord Blood which was a donor from a little boy. The doctors say it will be easier to determine how many donor cells are present in relation to Faiths cells. When the time came to do the fish test, it showed that Faith was accepting the donor cells. What a huge relief it was to hear this. However, we were not out of the woods yet.

Due to the large number of drugs required for this treatment and the doses needed, she has had some side effects. Her heart had enlarged and also had a problem with the left ventricle. Her lungs and kidneys were also affected. She was in the Critical Care Unit on a ventilator because of breathing issues as a result of the side affects and her body was unable to expel fluids. Faith became 2 liters fluid over load. At that point the doctors said there was nothing else they could do and that it was up to Faith to fight through it. It turned out that she was having a reaction to one of the medications to suppress her immune system. Once they altered that med along with her blood pressure med, she start to release all the fluid she was retaining. We finally got to hold her on Christmas morning for the first time in a month. She was in CCU for the whole month of December. She was able to go back to her transplant floor on New Years Eve. That was a nice way to bring in the New Year.

Personal Stories - continued

Because of the ventilator, Faith was being fed by tubes going down her nose. Eventually she had a feeding tube inserted. To this day she still has the feeding tube. We are slowly introducing her to bottle and spoon feeds. She will have to have a Barium Swallow test done to see if she is clearing her throat properly. She has aspirated in the past and had reflux issues when she was at Sick Kids in Toronto. Once we got home Faith had a nurse visit daily to care for the Pic line in her arm. In May, she got an infection in the Pic line which put her back in the Hospital for another 10 days. The doctors determined they no longer required it and to have it removed and left out. I guess you could say it was a blessing in disguise. Now, Faith no longer has nursing care and is on track to being a very healthy little girl. Now at 9 months of age she is a few months behind in motor skills, but everyday she is doing 1 thing more then she did the previous day.

Faith is the first in Ontario and second in Canada to have this procedure done. From birth to discharge, the whole process took 4 months. Needless to say, the entire experience was long and very emotional. However, we can say today that Faith is a living miracle as she continues to fight and beat this terrible disease. We continue to hold together as a family and hope the worst is far behind us and nothing but clear skies ahead.

Faith (4 months old) and Dr. Gasses at Sick Kids

Faith, 1st Birthday: October 14, 2010

Faith, 9 months old

Featured This Issue

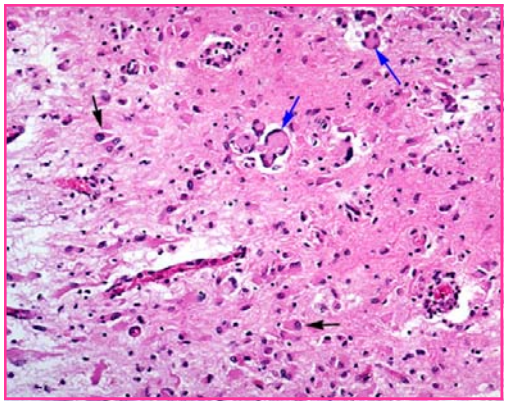
Krabbe Disease:

Chitra Prasad MD FRCPC (Clinical and Metabolic Specialist)
(Originally published in Volume 4 Issue 1 Winter 2007)

Krabbe disease is a rare neurodegenerative disorder often beginning in infancy where affected infants typically start to regress and lose developmental skills that have been acquired. It affects the nervous system at the levels of the brain and the peripheral nerves selectively, with sparing of other organ systems such as the heart, liver and kidneys. The hallmark of this disease is the presence of "globoid cells" which have more than one nucleus with neurons swollen with lipid storage material. There is a breakdown of the protective myelin coating and destruction of brain cells (features common with other leukodystrophies). Myelin is an important constituent of the white matter regions of the brain; consequently disorders affecting the white matter are termed "leukodystrophies". Krabbe disease is caused by the deficiency of enzyme "galactocerebrosidase" a key enzyme involved in the biochemical pathways of the body that handles myelin, a nervous system compound made of protein and fat (lipid).

Clinical symptoms of this condition include; irritability, unexplained fever, limb stiffness, seizures, feeding difficulties, vomiting and slowing of mental and motor skills. Eventually muscle weakness, spasticity, deafness and blindness develop. The Infantile form of the disease is fatal. Krabbe disease is a genetic disorder in which both parents are carriers of one altered gene copy for Krabbe disease. Parents themselves are completely healthy, but do carry a 25% risk of having an affected child in each pregnancy. Genetic counselling is recommended in families with this diagnosis.

Although there is no cure for Krabbe disease, the progressive course can sometimes be modified when children receive early bone marrow transplantation or cord cell transplantation prior to development of symptoms.



http://missinglink.ucsf.edu/lm/ids_104_Demyelination/Didactic/Leukodystrophies.htm

This section of a brain with Krabbe's disease shows severe astrogliosis (black arrows) and globoid cells around blood vessels (blue arrows).

Featured This Issue - continued

New Developments:

Dr. Joanne Kurtzberg's group at Duke University has pioneered the use of cord blood cells for transplantation which appear to provide a better and faster correction of enzyme deficiencies than does adult bone marrow. The Hospital for Sick Children (HSC) in Toronto helped the Jones family with this new therapy. Cord or bone marrow transplantation is a very challenging treatment and requires various subspecialists and a very knowledgeable team. Dr. Adam Gassas MBChB MSc FRCP DCH (Associate Professor of Pediatrics, Division of Haematology/Oncology/ Blood and Marrow Transplantation) was involved in the cord cell transplantation for Faith. Dr. Julian Raiman Metabolic specialist in HSC Toronto has also provided his expertise with ongoing care. Here at LHSC we are very grateful to the pediatric hematologists and oncologists and the entire metabolic team for helping with the ongoing management of children who have received transplantation. The HSC bone marrow transplantation team has performed almost 50 transplants for different types of metabolic diseases. It is the biggest pediatric program in Canada and worldwide. Over last couple of years newborn screening for Krabbe disease has been initiated in New York State. Long term follow up of children detected and managed presymptomatically will help us with understanding this devastating condition.

While there are still many hurdles to cross with the therapies of lysosomal storage disorders such as Krabbe disease, the newer advances are encouraging for patient families and professionals alike.

References:

- Global Organization for Lysosomal Diseases
- www.goldinfo.org
- United Leukodystrophy Foundation
- www.ulf.org
- The Myelin Project of Canada www.myelinprojectcanada.ca
- Newborn screening for Krabbe disease: the New York State model.
- Pediatr Neurol. 2009 Apr;40(4):245-52; discussion 253-5.
- Boelens JJ. Trends in haematopoietic cell transplantation for inborn errors of metabolism. J Inherit Metab Dis. 2006 Apr-Jun;29(2-3):413-20
- Duffner PK, Caviness VS Jr, Erbe RW, Patterson MC, Schultz KR, Wenger DA, Whitley C.The long-term outcomes of presymptomatic infants transplanted for Krabbe disease: report of the workshop held on July 11 and 12, 2008, Holiday Valley, New York. Genet Med. 2009 Jun;11(6):450-4
- http://www.dukehealth.org/physicians/joanne_kurtzberg

Suzanne's Corner

Savory Dressing

Recipe from Low Protein Cookery for PKU *by Virginia E. Schuett*

Ingredients:

1-1/3 cups (85 gm)
3/8-inch low protein bread cubes
1/3 cup (35 gm) finely chopped fresh mushrooms
1 teaspoon Nucoa margarine
1 tablespoon (9gm) chopped onion or green onions
2 tablespoon (13 gm) diced or thinly sliced celery
1/8 to 1/4 teaspoon sage or poultry seasoning
1 tablespoon melted Nucoa margarine
1/4 cup hot water
salt and pepper to taste

Cut off the crusts of 2 to 3 slices of low protein bread.
Cut into cubes for a total of 1-1/3 cups. Dry in a single layer in a 300-degree oven for 30 to 40 minutes until very dry.

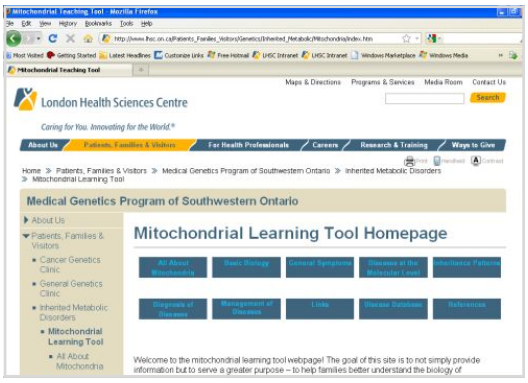
Meanwhile, prepare vegetables. Sauté mushrooms in 1 tsp. of margarine in a small skillet or saucepan for 2 minutes. Combine mushrooms, celery, and onions with dry bread cubes and seasoning in a small greased baking pan. Dribble melted margarine over all. Dribble hot water over to moisten (use slightly more or less depending on whether you like drier or moister dressing, realizing it will become some-what moister in baking). Add salt and pepper to taste. Bake at 350 degrees for 40 to 45 minutes.

Yield: 1-1/3 cups

Per recipe: 73 mg phe
Per 1/3 cup serving: 18 mg phe

Resources

Mitochondrial Website



The *Mitochondrial Learning Tool* is now available on our home page.
It is a teaching tool developed to give information to parents, caregivers and professionals about mitochondrial disorders.
Web page manager- Janice little
We would appreciate any comments and feedback.
http://www.lhsc.on.ca/Patients_Families_Visitors/Genetics/Inherited_Metabolic/Mitochondria/index.htm.

What's New

Conferences and Research Updates

1. Prasad AN, Rupar CA and Prasad C. Methylene Tetrahydrofolate Reductase deficiency (MTHFR) and infantile epilepsy. International Symposium on Epilepsy in Neurometabolic Diseases (ISENMD). 13th Annual Meeting of the Infantile Seizure Society and14th Annual Meeting of Taiwan Child Neurology Society. Abstract Book. A10. Oral Platform Presentation. March 2010
2. Prasad C, Rupar CA, and Prasad AN. (ISENMD). 13th Annual Meeting of the Infantile Seizure Society and14th Annual Meeting of Taiwan Child Neurology Society. Congenital Lactic acidosis (Pyruvate Dehydrogenase Deficiency) and Epilepsy. International Symposium on Epilepsy in Neurometabolic Diseases. Abstract Book. A26. Oral Platform Presentation. March 2010
3. Prasad AN, Rupar CA and Prasad C, and Levin S. (ISENMD). 13th Annual Meeting of the Infantile Seizure Society and14th Annual Meeting of Taiwan Child Neurology Society. Menkes Disease and Infantile Epilepsy. International Symposium on Epilepsy in Neurometabolic Diseases. Abstract Book. A32. Oral Platform Presentation. March 2010
4. C. Prasad, S. Venance, A. N Prasad, S. Levin, C. Campbell, H Rosenberg, M Geraghty and C. A Rupar. Spectrum of Pompe Disease (Metabolic Myopathy) in South Western Ontario: Role of Enzyme Replacement Therapy. Oral presentation at the Garrod association meeting in June 2010.
5. Prevention of Disability-Role of Early Detection and Newborn Screening for Some of the Genetic-Metabolic Disorders Causing Disability". Oral presentation in Maulana Azad Medical College in New Delhi India November 2010. Chitra Prasad, Charles A Rupar and Asuri Narayan Prasad.
6. Prasad AN, Prasad C. Genetic evaluation of the floppy infant. Semin Fetal Neonatal Med. 2010 Dec 3. [Epub ahead of print] PubMed PMID: 21131247.
7. Epilepsia 0 Suppl. 0 (Abst. 3.305), 2010 Methylenetetrahydrofolate reductase (MTHFR) deficiency and infantile epilepsy Authors: Asuri Prasad, C. Rupar and C. Prasad.

What's New - continued

Welcome to Our New Newborn Screening Counsellor!

Hello everyone,

My name is Oana Morar and I recently joined the Medical Genetics team as the Newborn Screening Genetic Counsellor. In my role, I meet with families to discuss the results of the Newborn Screen, a test that all newborns in Ontario have which tests for 28 conditions (mainly Metabolic). My educational background includes an HBSc. in Human Biology from the University of Toronto in 2007. This year I also graduated with a Master's of Science in Genetic Counselling from the University of Toronto. I am very excited to be part of the team and look forward to meeting you at our Metabolic Family Workshop on April 8, 2010.

Congratulations to Jennifer DiRaimo, Genetic Counsellor, Newborn Screening, on the birth of her second child, Matteo Luigi Diraimo born 22 Nov 2010,weighing 8lbs and 1oz

Tentative Vendors/ Booths That Will Be in Attendance

- Abbott
- Applied Nutrition
- Cambrooke Foods
- Mead Johnson
- Easter Seals
- Epilepsy Support Centre
- Kids Country Club
- National Food Distribution
- Nutricia
- Specialty Food Shop
- Vitaflow
- Pratten One - CPRI
- Mitrochondrial Support Group

Metabolic Family Workshop

Friday, April 8, 2011

Bestwestern Lamplighter

Number Attending: _____

Contact Name: _____

Phone Number: _____

Workshop Attending: (Indicate number)

Diet	Teen Group	Neurological Issues Navigating School System	Lysosomal
_____	_____	_____	_____

Low Protein Cooking Demonstration

Saturday, April 9, 2011

Loblaw on Wonderland

Number Attending: _____

Please return above information to:

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800 Commissioners Rd E London, ON N6A 5W9

Children's Hospital
London Health Sciences Centre

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!

Luke, age 11

Contact Information

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Parent Support Contact: Jennifer Culp

Tel: 1.519.632.9924

Email: donjen2000@hotmail.com

On healing---

All healing is first a healing of the heart.
-- Carl Townsend