The Inherited Metabolic Disorders News

Summer 2011 Volume 8 Issue 2

In this Issue

From the Editor

I hope everyone is enjoying their summer thus far!
Our 8th annual Metabolic Family Day and 7th annual Low Protein Cooking Demonstration were once again a huge success. See the section "What's New" on page 9 for a full report of the events, as well as pictures.

As always, your suggestions and stories are welcome. Please contact me by email: janice.little@lhsc.on.ca or telephone 519-685-8453 if you wish to contribute to the newsletter.

I hope everyone has a safe and happy summer!

Janice Little

From Dr Chitra Prasad

Dear Friends,

Hope you all are having a wonderful summer. We had a great metabolic family workshop with around 198 registrants this year. This was truly phenomenal. Thanks to all the team members and the families in the planning committee for doing such a fantastic job. I was really touched when one of our young metabolic patients told her parents that she would like to attend the metabolic family workshop! Please see some of the highlights of the metabolic workshop in the newsletter for those who could not make it. The speeches by our youth (Sadiq, Leanna and Laura) were appreciated by everyone. Big thanks to Jill Tosswill (our previous social worker) who coordinated their talks. We now have a new social worker Heather Small whom some of you have already met. Karen Reid (mom) has written her personal experiences about dealing with X-linked Adrenoleukodystrophy (ALD). Thank you Karen for sharing your story with us.
Russ Candler has also shared with us his challenges with a disease that is in the same category as ALD but of a different form "adrenomyeloneuropathy". Despite his struggles, Russ remains very gracious and optimistic.

Our genetics and metabolic section has finally moved to the 5th floor B building of the new Women's and Children's hospital. Those of you who have already had a chance to visit us in the clinic found it very spacious and bright.

I end with a quote from Rabindranath Tagore (Indian poet, philosopher, educator, thinker and Nobel laureate). This is his 150th birth anniversary year.

"It is very simple to be happy, but it is very difficult to be simple."
Rabindranath Tagore
Indian educator & Bengali poet (1861 - 1941)

Personal Stories

DAVID A. STAMPER (1989-1999)

David

by Karen Reid

On September 20th, 1989, I was blessed with my first son, David. David easily reached the milestones indicative of childhood. David loved school and loved to learn. Reading, math and spelling were his favorites. Before entering grade one, David would go to the barn on his own. There his grandpa and he would go through the addition tables. During David's grade 3 year , well established skills began to become challenging. In November of 1997, David's teacher called me and expressed concerns with David academically, as well that he seemed depressed. His teacher also mentioned that David seemed to have trouble hearing, if you stood behind him. In December of 1997, David was diagnosed with a severe auditory processing challenge. A week after this, David had a grand mal seizure lasting around 3 minutes. I recall going into David's Doctor demanding " What is going on!?" It really felt like a piece of David was being taken away piece by piece. My heart was breaking for David. With assistance of the tests ; CT scan, MRI, EEG and bloodwork, a diagnosis was reached. Bloodwork indicated that David's long chain fatty acid was elevated. MRI indicated that one third of David's white matter in his brain was destroyed . One never forgets the moment in time that life stood still.

David was diagnosed was a rare genetic disorder called Adrenoleukodystrophy ALD (Adrenal- affects the adrenal glands , Leuko – affects the white matter of the brain – Dystrophy – deterioration) The most heart breaking news, there is no cure. In this disease, the very long fatty acid destroy the myelin sheath around the nerves in the white matter of the brain , thusly , destroying the abilities presented by that area. Other testing indicated that David had Addison's Disease (adrenal gland under functions, Cortisone treatment was issued). Since the disease was genetically related , David's 2 brothers were tested . Brendan gratefully was clear, however , Brady was found to have the gene for the same disease that inevitably claimed his brother. Brady also had Addison's Disease.

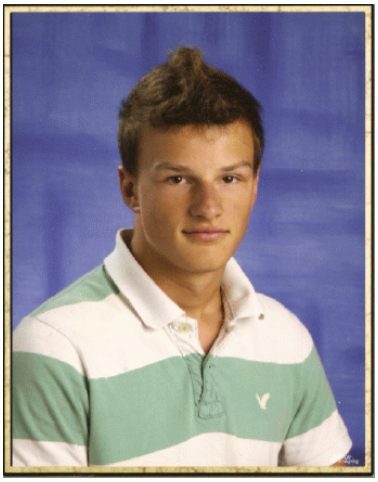
David and Brady were started on a research treatment "Lorenzo's Oil" (for ALD). As shock and devastation settled in, reality of our new life began. After diagnosing (January '98)

Personal Stories - continued

David's abilities declined very quickly. By September of that year , David required the use of a wheelchair , pureed food and assistance with feeding. Speech was with great difficulty. It was like an escalator going down hill- FAST.

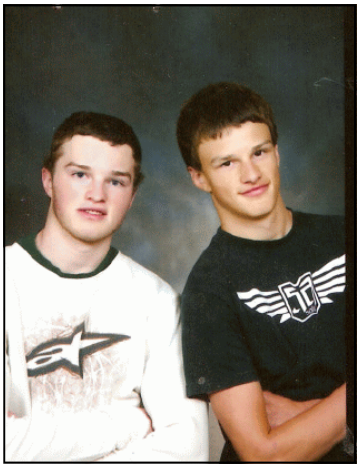
David would have mystery infections requiring antibiotics - each bout of these would affect his eating. In January 1999, David was admitted to the hospital; he was having troubles eating plus a fever. A feeding tube was inserted. David was in the hospital from January 1999 to his passing December 16th 1999. During this time David's life consisted of being turned every 2 hours, painful contractures and the feeding tube. David was unable to see at this time.

Currently, Brady is doing wonderfully. Brady continues to take the medication Cortef for Addison's Disease. For the hope of ALD prevention, Brady now takes a new research medication, Lovastatin. At the age of 16, Brady is involved with activities all teenagers enjoy. Brady was captain of his soccer team this past summer. In high school, Brady plays soccer and basketball on the school teams. At 6'2 Brady has grown into a fine young man. Brady keeps busy working on the family farm. This past summer , he worked at the CAN AM Motocross Championship in Walton. I have been blessed with 3 fantastic sons. While the road took a tragic turn with David, I can't imagine missing time I had with him while he was here with me. As one reflects, one is humbled by the support of friends and family through challenging times. Appreciation goes out to the medical team, past and present in London who walked with us through the tears and successes experienced.



Brady

Brady



Brendan & Brady

Featured This Issue

A Little About Russ

I am a 68 year old retired school teacher with AMN (Adrenomyeloneuropathy). I have weakness in my legs and very poor balance. I have a neurogenic bladder; kidneys function at 50%. I also have Addison's. I'm in the hospital once a year with Addison's Crisis or Shock. I am under the care of CCAC; a man comes twice a week to shower me, and a nurse comes every 8 weeks to change my catheter. I am interested in computers and learning about the latest information on ALD. I also love going to restaurants.

Adrenoleukodystrophy and Adrenomyeloneuropathy

Summarized by Dr. Chitra Prasad

Adrenoleukodystrophy (ALD) is one of a group of genetic disorders called the *leukodystrophies* that cause damage to the myelin sheath, an insulating membrane that surrounds nerve cells in the brain. People with ALD accumulate high levels of saturated, very long chain fatty acids (VLCFA) in the brain and adrenal cortex because they do not produce the enzyme that breaks down these fatty acids in the normal manner. The loss of myelin and the progressive dysfunction of the adrenal gland are the primary features of ALD. ALD has two subtypes. The most common is the **X-linked form (X-ALD),** which involves an abnormal gene located on the X-chromosome. Women have two X-chromosomes and are the carriers of the disease, but since men only have one X-chromosome and lack the protective effect of the extra X-chromosome, they are more severely affected. Onset of X-ALD can occur in childhood or in adulthood. The childhood form is the most severe, with onset between ages 4 and 10. The most common symptoms are usually behavioral changes such as abnormal withdrawal or aggression, poor memory, and poor school performance. Other symptoms include visual loss, learning disabilities, seizures, poorly articulated speech, difficulty swallowing, deafness, disturbances of gait and coordination, fatigue, intermittent vomiting, increased skin pigmentation, and progressive dementia. The milder **adult-onset form (adrenomyeloneuropathy),** typically begins between ages 21 and 35. Symptoms may include progressive stiffness, weakness

Featured This Issue - continued

or paralysis of the lower limbs, and ataxia. Although adult-onset ALD progresses more slowly than the classic childhood form, it can also result in deterioration of brain function. A **mild form of ALD** is occasionally seen in women who are carriers of the disorder.

Symptoms include progressive stiffness, weakness or paralysis of the lower limbs, ataxia, excessive muscle tone, mild peripheral neuropathy, and urinary problems.

X-ALD is diagnosed by a simple blood test that analyzes the amount of very long chain fatty acids; the levels of these molecules are elevated in X-ALD. While the test is accurate in males, in about 20% of women who are proven carriers, the test shows normal results and thus gives a "false negative" result. A DNA-based blood test is available. This test permits accurate identification of carriers by genetic testing, and if it is normal can assure a woman that she is not a carrier. Diagnostic testing, carrier screening and prenatal diagnosis are available.

Adrenal function must be tested periodically in all patients with ALD. Treatment with adrenal hormones can be lifesaving. Symptomatic and supportive treatments for ALD include physical therapy, psychological support, and special education. Recent evidence suggests that a mixture of oleic acid and erucic acid, known as "Lorenzo's Oil," administered to boys with X-ALD can reduce or delay the appearance of symptoms. More research on Lorenzo's oil and lovostatin is required. Bone marrow transplants can provide long-term benefit to boys who have early evidence of X-ALD, but the procedure carries risk of mortality and morbidity and is not recommended for those whose symptoms are already severe or who have the adult-onset or neonatal forms.

A movie "Lorenzo's oil" was made in 1992 and depicts the story of a family where a couple's young boy was affected with Adrenoleukodystrophy.

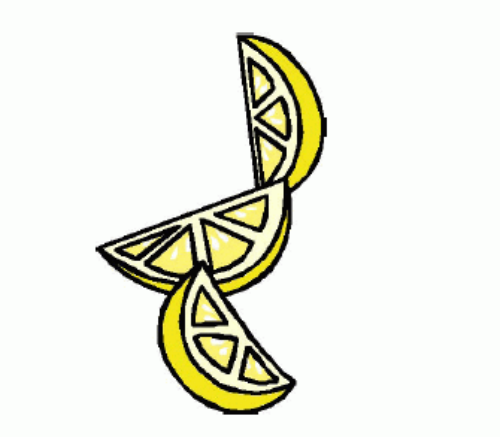
References: http://www.ninds.nih.gov/disorders/adrenoleukodystrophy/adrenoleukodystrophy.htm

http://www.ulf.org/

Suzanne's Corner

Lemon Sugar Cookies

1/2 cup butter flavored Crisco
3/4 cup powdered sugar
1 1/2 tsp egg replacer
1 tsp lemon extract
1/2 tsp lemon zest
1/2 cup potato starch
1/2 rice starch
1/2 cup Wel-Plan baking mix
1/2 tsp baking soda
1/2 tsp cream of tartar
1 tsp lecithin
1/4 tsp salt



In a large mixing bowl, cream together the Crisco, powdered sugar, egg replacer (mixed with 2 tbsp water), lemon extract and lemon zest. Add the potato starch, rice starch, Wel-Plan baking mix, baking soda, cream of tartar, lecithin, and salt. Mix well. Chill the dough. Roll out and cut into cookies. Bake at 375º for 10 minutes. Makes 40 cookies.

Per recipe: 18 mg ohe, 0.6 g protein
Per cookie: trace phe, trace protein
Created by Evelyn Gower

Source: ***http://depts.washington.edu/pku/recipes/lemoncookie.html***

Suzanne's Corner - continued

Low Protein Food List for PKU

Third Edition

by Virginia E. Schuett

The Low Protein Food List for PKU has been an indispensible resource for everyone involved in the treatment of phenylketonuria (PKU) since 1995. This third edition (2010) contains over 6,000 entries, the most extensive listing yet of foods that are potentially suitable for the diet, nearly double the number found in the previous edition. It includes many new foods found at health-oriented grocery stores, expanding choices for the diet.

The book provides information on phenylalanine (phe), protein, and calorie content of foods based on serving portions, both in common measures and gram weights, in an easily searchable format. The handy "mg phe/gm food" column allows users to compare phenylalanine density of foods and make appropriate food choices for individual diet needs. People using an "exchange" system for counting phe will find exchanges are calculated for each food portion as well. The front section of the book also contains valuable information and tips for managing the PKU diet.

Virginia E. Schuett is founder and director of National PKU News, a non-profit organization devoted to providing news and information to people living with or treating PKU since 1989 via a newsletter that is produced three times yearly. A nutritionist, she has been working with PKU-affected families since 1972. She is the author of Low Protein Cookery for PKU, Apples to Zucchini: A Collection of Favorite Low Protein Recipes (with co-author Dorothy Corry), You and PKU, and numerous articles and educational materials related to PKU

At last, the newest edition of the **Low Protein Food List for PKU** is available. This resource may be used by individuals and families with PKU as well as anyone who follows a low protein diet.

Please see the website for ordering information: **www.pkunews.org**

What's New

Justin Massicotte (MPS1-H) has been attending Kindergarten full-time since September 2010. On March 4, 2011, his school presented him with an award for showing respect towards his peers and his teachers. We are so proud of you Justin, Love Maman et Papa!

Great job Justin! from all of us on the Metabolic Team at LHSC

Kareem's Arctic Quest *www.corddonate.ca/KareemsArcticQuest*

Please read Kareem's story about having the metabolic disorder *Niemann Pick B*, and his "Arctic Quest " to raise money for the Canadian Organization for Rare Disorders (CORD).

Good luck from your friends on the Metabolic Team at LHSC!

New Metabolic Team Member

Hi Everyone. My name is Heather Small and I am the new social worker on the Metabolics and Neurology teams. I might look familiar to some of you as I have worked at LHSC for the past eleven years in a couple of programs including child and adolescent mental health and paediatric oncology. I have enjoyed meeting many amazing families in my new role within the Metabolics program and look forward to meeting many more of you during your clinic appointments. Please feel free to contact me if something comes up outside of a clinic appointment at 519-685-8500 ext. 56149. Bye for now.

What's New - continued

"*The nice thing about teamwork is that you always have others on your side*"

By Margaret Carty

8th Annual Metabolic Family Workshop a Grand Success!

The 8th annual Metabolic Family Workshop held on April 8, 2011 provided an excellent opportunity to share information on metabolic disorders as well as a venue for families and health care providers to share their experiences and make a few friends along the way. This year 198 individuals attended the event with their families, our highest number to date.

Families were invited to interact with speakers, learn about various metabolic formulas and foods, get updates about treatments and research, visit displays of metabolic food products and learn about the resources available in their community.

The day began with a warm welcome from Dr. Chitra Prasad, Director of the Metabolic Clinic at Children's hospital LHSC. Jill Tosswill (previous social worker with the metabolic program) introduced Sadiq Chevelwalla, Laura Ridout and Leanna Glasbergen (three of our youth/young adults living with various metabolic conditions) who presented an insight into what it is like living with a metabolic condition. They discussed challenges they have faced and accomplishments they have made along the way. Their talks were appreciated by all.

Other speakers included Ashraf Ghadban (parent) from the Canadian Organization for Rare Disorders (CORD) who informed us about Orphanet (European internet site for rare conditions). Dr. Michael Geraghty, medical advisor to the newborn screening at CHEO, brought us up to date with the Newborn Screening process in Ontario. Our motivational speaker was Michael Moore, (***www. motivationalplus.com***) whose message stressed the importance of caring for the caregivers using humor, music among other things.

The surprise of the day came from Nikki Hummel, mother of Carver Hummel (young boy) with Methylmalonic Acidemia, who donated close to $4500 to the Inherited Metabolics Disorders Program. Dr. Chitra Prasad and Debbie Comuzzi, President and CEO of the Children's Health Foundation, accepted the generous donation.

What's New - continued

We also had four group sessions: a diet group led by Dr. Chitra Prasad and Suzanne Ratko who discussed diet-related independence strategies; a neurological group led by Dr. Sharon Goobie as well as the team from the Thames Valley Children's Centre which focused on navigating the school system; a lysosomal storage disorders group led by Dr. Tony Rupar, Veronica Kokavec and Karen Sappleton (Sickkids Toronto) discussed updates for enzyme replacement therapies; and a teen and young adult group led by Heather Small and Dr. Erica Gold focused on motivational strategies for taking responsibility of their own health to lead a more independent lifestyle with their metabolic conditions.

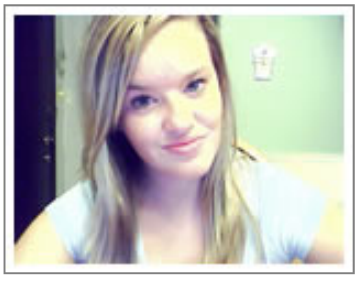
Suzanne Ratko, Registered Dietitian and her team once again ran the annual "low protein" cooking demonstration on the 9th April, which featured food and recipe ideas by Karen Gough, Nutricia. Families got practical demonstration for the low protein recipes.

Submitted by:
Oana Morar MSc
Genetic Counsellor and Newborn Screening Coordinator
(On behalf of the metabolic team)

Congratulations to the 3 youth speakers who shared their stories about living with a metabolic disorder. They all did an excellent job.

Sadiq Chevelwalla

Leanna Glasbergen



Laura Ridout

Metablic Family Workshop

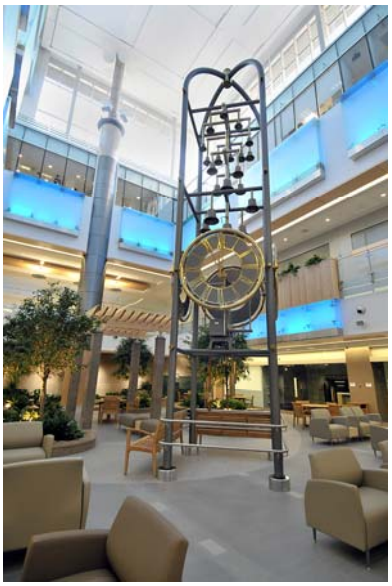
LHSC Child and Family Resource Centre

Lisa Hawthornthwaite and Jennifer Banting would like to welcome you to the new Child and Family Resource Centre. We are located on the first floor of Children's Hospital directly beside the atrium, room B1-006. Once located on the 3rd floor our new space offers an open, bright, quiet and comfortable setting where patients, family members and staff can find current and reliable information about health and medical care. Lisa and Jennifer are available to help you find information about specific conditions, hospitalizations parenting, child development, nutrition and much more.

We look forward to meeting you!

"Knowledge is a Powerful Resource!"

Lisa & Jennifer



Lobby on the 1st floor outside of the Child and Family Resource Centre

Please Mark Your Calendars for Next Years Date

Children's Hospital
London Health Sciences Centre

Metabolic Family Workshop

Friday, May 11, 2012

Best Western Lamplighter

Low Protein Cooking Demonstration

Saturday, May 12, 2012

Loblaw on Wonderland

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

Research & Presentations

1. The Expanding Phenotype of MELAS Caused by the m.T3291C tRNA Mutation. Chitra Prasad, C. Anthony Rupar and Asuri N. Prasad. Presented at American College of Medical Genetics Meeting March 2011.

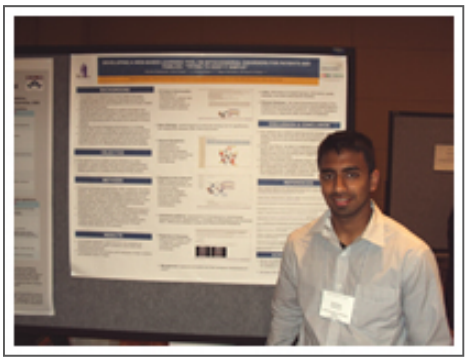
2. Menkes disease, Mitochondrial dysfunction and Emerging Therapies Prasad C, Rupar CA, Goobie S, Levin S, Kokavec V and Prasad AN. Presented at Garrod Meeting June 2011.

3. Rupar CA-Presentations at World Lysosomal meeting Las Vegas February 2011, Lysosomal storage diseases club June 2011, and Metachromatic Leukodystrophy association meeting in Baltimore on June 2011. Recent updates on metachromatic leukodystrophy.

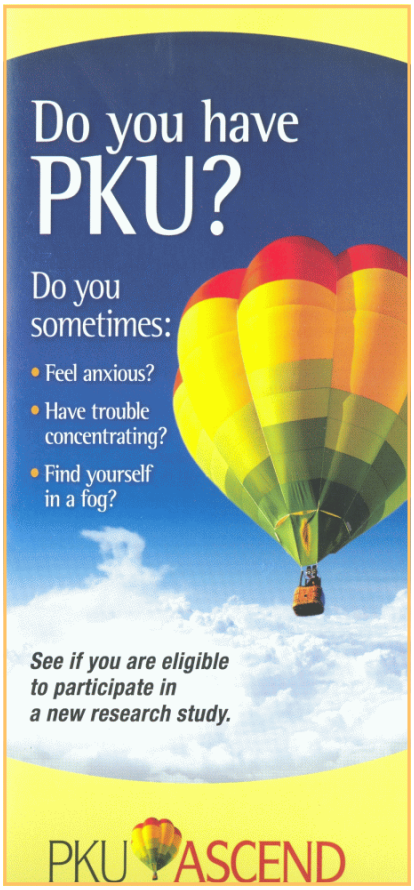
4. Nachiket Deshpande, Chitra Prasad, C. Anthony Rupar, Vijaya Chevendra, and Asuri N. Prasad. Developing a web-based learning tool on mitochondrial disorders for patients and families: “trying to keep it simple!” Presented at United Mitochondrial Foundation June 2011 in Chicago.

5. Bock DE, Rupar CA, Prasad C. Acta Paediatr. 2011 Feb 25. doi: 10.1111/j.1651-2227.2011.02208.x. Asymptomatic critical hypoglycaemia: a dangerous presentation of glycogen storage disease type 1b in infancy.

6. Veronica Kokavec: Health Care Advocates Meeting in Calgary 2011 and World Lysosomal meeting Las Vegas February 2011.



Nachiket Deshpande is a 2nd year BSc biomedical sciences student at the University of Western Ontario. Here he is presenting a poster at the United Mitochondrial Foundation in Chicago June 2011. (with thanks to the Stickles family)



A new Study for those with PKU is starting at LHSC soon. Please contact Veronica Kokavec, Suzanne Ratko or Dr. Prasad at 519-685-8500 Extension 56131 if you would like more information.

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!

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