The Inherited Metabolic Disorders News

Fall 2007 Volume 4 Issue 3

In this Issue

From the Editor

I hope everyone has enjoyed the beautiful early fall weather we have been blessed with.

The tentative date for the 2008 Metabolic Family Workshop is Friday May 30th, and the Low Protein Cooking Demonstration is Saturday May 31st. More details will be included in the next newsletter

We have added a new feature entitled "Tricks of the Trade" which allows families to share practical tips and information with each other.

As always, we would love to share your stories, suggestions and questions.

Janice Little

From Dr. Chitra Prasad

Dear Friends,

The fall is always the best time of the year for me. I love my walks and am always amazed at the spectacular changes in the color of the leaves. The beautiful picture of the pinecones on this page reminds me that pinecones are a symbol of the "evergreen fruit". Pinecones are also revered in many different faiths as bringing various people together as members of one big family.

Our newsletter continues to evolve with your input, stories and suggestions. Jennie, and her story reveals the true meaning of courage. I have now known Jennie for a couple of years and am very impressed by her strength and her spirit to overcome all obstacles.

A number of new people have joined the metabolic team. Our sincere thanks go to Brian Seeley (Social Worker) and to Susan Alexander (Social Worker who provided coverage for the summer). We now have Jill Toswill our new social worker that is taking Brian Seeley's place. Jennifer MacWhirter is our newborn screening coordinator and Andrea Barton is the new newborn screening secretary. Do read about them in the newsletter.

With best wishes to all of you.

Your friend
Chitra Prasad

"Happiness is an inside job" By John S. Powell

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Personal Stories

Jennie's Story on Maple Syrup Urine Disease

Hi my name is Jennie Verbeek I am 33 years old. When I was 7 days old I was diagnosed with Maple Syrup Urine Disease at the Hospital for Sick Children. This made me the first diagnosed case with this disease in Ontario and even though the prognosis was poor at the time, I was able to beat the odds. They started treating me at 8 days old, but a day later I had 5 Cardiac arrest one right after the other, which left me with mild Cerebral Palsy.

I am very good with my diet, because I don't cheat. Even when I go out with my friends I still watch what I eat and I am very good at calculating my equivalents. My disease has not kept me from going places.

Four years ago I went with my parents to Aruba and had a great time. Last year I went with them to Holland for a second time. I had no problems with my diet traveling even though it would have been nice to have had the M.S.U.D. Express. This would have been a lot easier than dragging 2 cases of Keaton-next around, which is my daily diet. Because I can not run my blender in some places, I usually travel with a battery operated one or in a pinch I'll use a spoon to mix my diet. What ever works, but one thing I never travel without is my doctor's 'To whom it may concern-Letter' in case of an emergency.

I hope everyone enjoyed reading my story and I hope I helped other people.

By Jennie Verbeek

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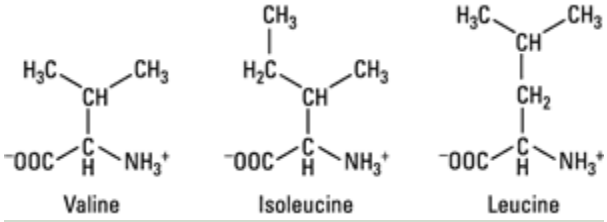
Featured This Issue

Maple Syrup Urine Disease (MSUD)

Compiled by Dr Chitra Prasad

John Menkes, the pediatrician who later described Menkes disease, first characterized this disease in 1954. Maple syrup urine disease (MSUD) is due to a disruption in the metabolism of the branched-chain amino acids (BCAAs)—leucine, isoleucine, and valine—and the resulting buildup of α-ketoacids. The isoleucyl ketoacid gives the urine its distinctive odor and the disease its name.

The disease has an incidence in the United States of ~1/200,000. There is increased incidence in Old Order Mennonite population. MSUD is inherited as an autosomal recessive disorder. MSUD can arise from mutations in several genes. These genes encode for some of the proteins involved in the mitochondrial multienzyme complex branched-chain α-ketoacid dehydrogenase (BCKD).



Branched-chain amino acids.
A buildup of α-ketoacids during amino acid metabolism can be dangerous to a newborn infant.

The early symptoms of the disease include lethargy and a loss of appetite within the first few days after birth as the infants go into ketoacidosis. Unrecognized and untreated, the disease leads to vomiting, seizures, coma, and sometimes death. Thus, it is critical that the disease be caught early by a simple blood test to determine the serum levels of the BCAAs and their respective α-keto acids. Several efforts have been undertaken to add MSUD to the list of diseases for which newborn infants are regularly checked. In Ontario the newborn screening for this disorder is ongoing as part of the expanded newborn screening program. One way to limit α-ketoacid buildup is by eliminating the problem amino acids from the diet, but they are three of the "essential" amino acids. Therefore, MSUD patients must ingest special formulas that limit their dietary intake of BCAAs, and because the severity of symptoms varies greatly from person to person, diets must be carefully tailored. The role of dietitian is absolutely essential in the management of this condition.

With early detection, there will be improvement in the overall health care of these individuals. We hope that newer definitive treatments will become available with further research on the genes and proteins.

References

http://pubs.acs.org/subscribe/journals/mdd/v05/i03/html/03disease.html
http://nordrdb.com/search/rdbdetail_abstract.html?disname=Maple%20Syrup%2Urine%20Disease
http://www.msud-support.org/

From Dr. Victoria Siu, Medical Geneticist:

MSUD affects about 1 in 400 babies in the Old Order Mennonites of Pennsylvania, where the carrier frequency for a specific DNA change (mutation) is about 1 :10. We have identified the same DNA change in the Old Order Mennonite community in southwestern Ontario. Married couples in the community are now being offered carrier testing. If both husband and wife are found to be carriers, each of their children will have a 25% chance of having MSUD. When the babies are born, a blood sample will be taken at day 1 of life and sent directly to the Biochemical Genetics Laboratory at CPRI for testing of branched chain amino acids. Identification of infants with MSUD before they develop symptoms will allow early institution of diet so that they can remain healthy.

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Welcome

We are pleased to welcome new members to the metabolic team:

Jill Toswill, Social Worker, replaces **Brian Seeley**.

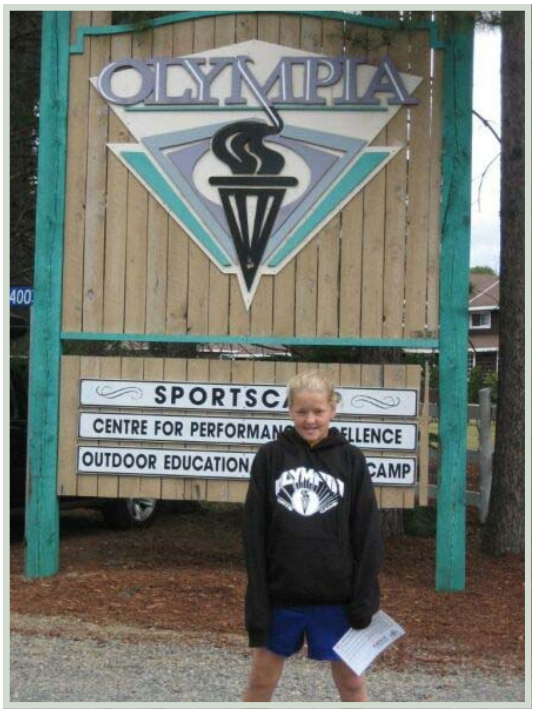
Megan Edwards, Dietitian, joins the team to help support the growing metabolic program.

Jennifer MacWhirter, Newborn Screening Program Coordinator.
Read more about Jennifer on page 5 under *"What's New".*

Andrea Barton, Newborn Screening Administrative Associate.

We would also like to thank **Brian Seeley**, Social Worker, for all his hard work and dedication with the Metabolic Team over the last few years, as well as **Susan Alexander** who filled for the summer during his absence.

Success Stories



Laura was selected by her teachers to be the recipient of the Leadership Development award at her school. It is awarded by the Rotary Club of London to a student who shows good leadership ability among their peers, as well as good grades and athletic ability. The award was a week at Camp Olympia. She had a fabulous time and had many new experiences and met some great new friends.

Congratulations Laura!

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What's New

Newborn Screening Update

Hello everyone,

My name is Jennifer MacWhirter. I am a Genetic Counsellor and Coordinator of the Newborn Screening Program in Southwestern Ontario. I have taken a leadership role within the Newborn Screening Program since May of this year and have been enjoying my time working with amazing families and their newborns.

Newborn screening has been around for many years, however last year the government expanded the original screening panel and by the end of 2007 it will include 27 conditions for which treatment and early intervention exists. This will ultimately reduce the number of new-borns who will fall sick with life threatening complications related to their diagnosis.

On 19 October 2007 the Children's Hospital of Eastern Ontario will be holding a special workshop for physicians, coordinators, and medical experts to discuss how Newborn Screening Programs across Ontario are working at individual centres. I will be attending this workshop and will be happy to give a summary of the meeting in the next edition of this newsletter.

Warmest,
Jennifer MacWhirter, MS

Correction: In the Spring/Summer issue it was reported that Newborn Screening for Cystic Fibrosis had begun in Ontario. This is not the case. However, it should begin sometime before the end of 2007.

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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Suzanne's Corner

Update from Nutricia

Effective September 24, 2007, **Loprofin Drink Mix** will no longer be available. It is replaced by **Milupa Low Protein lp-drink** (product code at Specialty Food Shop 12653).

Newly available at the Specialty Food Shop
www.sickkids.on.ca/specialtyfoodshop

NEW Low Protein Products covered:
1327 Maddy's Homestyle Yellow Cake Mix
1328 Maddy's Homestyle Sugar Cookie Mix
1329 Maddy's Homestyle Blueberry Muffin Mix
47067 Phlexy Add Ins

Cheddar Broccoli Soup

Ingredients

1/4 cup onion chopped
1/4 cup butter or margarine
1/4 cup CBF All Purpose Baking Mix
3 Tablespoons (27g) CBF Shake 'N' Cheeze
1 teaspoon CBF Chicken-Flavored Consomme and Seasoning
1 cup water
1 1/2 cups non-dairy liquid creamer
1 cup broccoli, chopped

Directions

1. In a saucepan, sauté the onion in butter until tender. Stir in CBF All Purpose Baking
2. Mix and CBF Shake 'N' Cheeze. Cook and stir until smooth and bubbly.
3. Add the CBF Chicken-Flavored Consommé and Seasoning, water and non-dairy creamer all at once.
4. Cook and stir until the mixture boils and thickens.
5. Add the broccoli. Simmer, stirring constantly, until heated through.

Makes 3 servings: **1 serving = ~ 1 cup (225g)**

Total Protein: 1.7 g per serving

Phe per serving: 40 mg per serving

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Tricks of the Trade

This new section gives the opportunity for families to share special tips about foods, and formula preparation. Please email janice.little@lhsc.on.ca if you have anything to share.

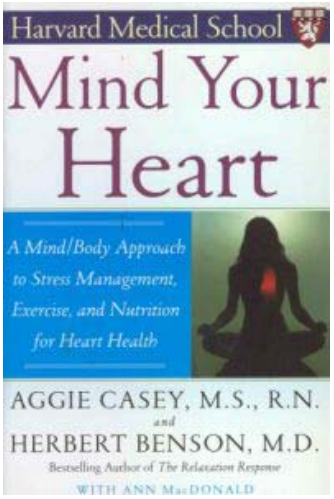
From Suzanne Ratko: Update on two popular Cambrooke products:

Portabella Spinach Ravioli & Tortilla Wraps are unavailable from Cambrooke at this time. Both of these products are being redeveloped and will be back soon. No date is available.

Tyler Allen highly recommends:

Dietary Specialties Peanut Butter Flavoured Spread (from the Specialty Food Shop—SHS) mixed with Dandelion or Wildflower Honey. In a separate container, gradually mix together some of the Peanut Butter and Honey to a spreadable consistency and sweetness. Tyler likes it really sweet.

Mind Your Heart



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

- Don't take your job home with you or on breaks. If you can't avoid it, at least limit the time you spend worrying.
- Remember to get enough sleep at night..
- Develop with friends and coworkers your own brand of happy hour, parties, birthday celebrations and other events that break up routine.
- Look at unavoidable stress as an avenue for growth and change.
- Avoid people who are stress carriers or negaholics.

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Kids Korner

Birthdays: October, November, December

Child's Name	Metabolic Disorder	Birthday	Age
Nolan	PKU	October 18, 2005	2
Michael	PKU	October 12, 2001	6
Emma Lynn	PKU	November 2, 2000	7
Nathan	PKU	October 6, 2006	1
Adam	PKU	November 2, 1998	9
Raphael	PKU	October 23, 1999	8
Laura	PKU	December 29, 1993	14
Vanessa	PKU	October 6, 2003	4
John	PKU	October 25, 2004	3
Brady	ALD	October 2, 1993	14
Joseph	Amish Microcephaly	December 7, 2002	5
Cole	Arginase Deficiency	November 1, 1997	10
Michael	CPS Deficiency	December 16, 1996	11
Riley	MCADD	December 17, 2006	1
Colleen	MELAS	October 24, 1992	15
Warda	Metachromatic Leukodystrophy	November 14, 2000	7
Ryan	MSUD	October 25, 1992	15
Kareem	Niemann Pick-B	November 23, 2002	5
Joseph	PDC	October 17, 2000	7
Sarah	Sanfillipo	December 4, 1997	10
MacKenzie	Not Diagnosed	November 16, 1999	8
Marrisa	Not Diagnosed	December 10, 1998	9
Gavin	Not Diagnosed	November 22, 2003	4

PKU - Phenylketonuria
ALD - Adrenoleukodystrophy
CPS Deficiency - Carbamyl Phosphate Synthetase Deficiency
HMG CoA Lyase Deficiency 3-Hydroxy-3-Methylglutaryl-Coenzyme A (CoA) Lyase Deficiency
MCADD - Medium Chain Acyl-coA Dehydrogenase Deficiency
MELAS - Mitochondrial Myopathy, Encephalopathy, Lactacidosis, Stroke
MSUD - Maple Syrup Urine Disease
PDC - Pyruvate Dehydrogenase Complex

Happy Birthday Everyone!

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