The Inherited Metabolic Disorders News

Spring/ Summer 2005 Volume 2 Issue 2

In this Issue

From the Editor

Welcome to the latest issue of The *Inherited Metabolic Disorders News*! This issue is a combined spring and summer issue. We are still undergoing some growing pains, so you might notice some new items in the index, as well as name changes.

Our goal is to share information on various metabolic disorders. We now have expanded our newsletter circulation to over 80 families in the Southwestern Ontario region.

Please send your stories, comments and suggestions!

From Dr. Chitra Prasad

Dear All,

Hope everyone is enjoying the summer! As always, it's so wonderful to see the trees full of leaves, lots of beautiful flowers and the whole world seems to come alive.

Our Metabolic Team has worked very hard, and all members deserve special recognition for making a difference in so many people's lives. Our Metabolic Family Day on May 13th was a great success. Please read more about it in the newsletter. We hope more families will continue to join our metabolic support group.

Our patients and their families continue to show us the path of courage, how to live even with the most difficult situations. In the newsletter you will find such stories of courage, determination and a clear evidence of what good teamwork is capable of.

Please send your suggestions and contributions to our newsletter.

Finally, a quote I like very much. *"Happiness is not a state to arrive at, but a method of travelling!"*

Dr. Chitra Prasad

Personal Stories

From A Young Lady with MCADD:

Adam is my older brother who passed away from MCAD he was only 2 and a half years old. He had big blue eyes, blonde hair, and the cutest smile. He was my older brother and I looked up to him and know he is looking over me. My name is April I was not tested for MCAD when I was born, I was only tested for it when Adam died, in a way he saved my life and I thank him for that. As a kid it was a bit of trouble having MCAD when I got the flu, which was many times, I would have to make a trip to the hospital. And I remember whenever we would go to McDonalds I was only allowed to have six french fries because I have to watch my weight. As you get older it gets easy. Now since I am able to make my own choices I choose healthy foods and take my medicine three times a day. I'm my opinion I don't mind having MCAD because I just have to take my medicine and watch what I eat. I am now sixteen years old and going into grade eleven, receiving good marks. And I am happier then ever with my family and friends. To all the families with children who have MCAD, be strong and I wish you all the best in the bright future to come.

April Willison

Tammy Clark is a mother of a child who died from undetected MCADD. She shares her family's story on this website: www3.sympatico.ca/tammy_roger/Jennastory.htm She is also an advocate to expand newborn screening in Canada. *Saving Babies Through Newborn Screening Inc*. www.savebabies.org/canada_plea.php is an organization whose main goal is to raise public awareness of the need for the Canadian government to take an initiative to guarantee access to expanded newborn screening for all Canadian babies.

Resources

- **National Tay Sachs and Allied Disorders**
  www.ntsad.org
- **National Niemann-Pick Disease Foundation, Inc.**
  www.nnpdf.org

Kids Korner

April, May, June, July August, September birthdays:

Child's Name	Metabolic Disorder	Birthday	Age
Mohammed	MMA	July 26, 2004	1
Austin	MCADD	July 17, 1996	9
Aaron	ML4	August 30, 1993	12
Rozlin	PKU	August 7, 1990	15
Daniel	PKU	April 25, 2003	2
Zackaria	PKU	June 6, 2004	1
Olivia	PKU	September 6, 1991	14
Wyatt	Metabolic disorder	July 18, 2002	3
Lama	PKU	September 1, 1990	15
Patrick	PKU	July 2, 1991	14
Alexa	Metabolic disorder	September 29, 1993	12
Julia	PKU	July 29, 2001	4
Hailey	PKU	August 19, 2003	2
Jeff	Metabolic disorder	June 24, 2001	4
Joan	Glycogen Storage Disease	June 20, 2003	2
Stathie	PKU	May 7, 1992	13
Dusan	PKU	July 21, 2003	2
Carter	PKU	September 5, 2001	4
Elijah	PKU	June 18, 2003	2
Samantha	PKU	July 31, 2000	5
Samantha	PKU	May 9, 2002	3
Michelle	HMG CoA Lyase Deficiency	September 1, 1999	6
Jordynn	5-MTHFR	May 30, 2000	5
Madison	Metabolic disorder	June 23, 2000	5
Nicholas	Metabolic disorder	September 1, 2002	3

Happy Birthday Everyone!

MMA- Methylmalonic Acidemia
MCADD- Medium chain acyl-coenzyme A dehydrogenase deficiency
ML4- Mucolipidosis type 4
5-MTHFR- Methylenetetrahydrofolate reductase deficiency

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!

Suzanne's Corner

Suzanne Ratko
Registered Dietitian

Here is a low fat summer drink from *MEALS FOR YOU* that should cool you on a hot summer day!

Creamy Orange Fizz

For 4 servings:

- ⇨ 2/3 cup low fat vanilla frozen yogurt
- ⇨ 1 1/3 cup orange juice
- ⇨ 1 Tbsp + 1 tsp confectioner's sugar
- ⇨ 1 Tbsp + 1 tsp lemon juice
- ⇨ 1/3 cup ice cubes
- ⇨ 1 ¼ cup orange-flavoured or unflavoured sparkling water, chilled

Mix all ingredients, except sparkling water, in a blender.
Process until smooth.
Combine juice mixture with sparkling water in a pitcher.

Stir until mixed thoroughly and serve.

Per Serving (1/4 of recipe)	
Calories	80
Fat	0.3 grams
Carbohydrates	18.9 grams
Protein	1.7 grams

Courtesy of www.mealsforyou.com

Metabolic Family Day

The Metabolic Family Day on May 13th was another grand success with 84 family members and over 40 staff registrants. Dr. Tim Frewen, (Chair of Pediatrics at LHSC) gave the introductory speech. After the combined session given by Dr Tony Rupar regarding the current status of newborn screening in Ontario, we had two concurrent sessions:

1- PKU
2- Neurological aspects of metabolic disorders.

Dr Blair Seifert (Clinical Pharmacist from Manitoba) gave two talks. One was on Biopterin (a PKU drug) and one was about herbal therapies for seizure disorders. CCAC (Community Care Access Centre) was represented in session 2. Dr. Narayan Prasad (Pediatric Neurologist) spoke on neurological aspects of metabolic disorders. We were fortunate to have a mother of a child with PKU (Dr. Darlene Elliott-Faust, psychologist) and an adult with PKU (Sarah Foster) speak about the challenges and triumphs of living with PKU.

Suzanne Ratko, dietitian, organized a low protein food cooking demonstration for next day. More than 20 people participated. This was held at Loblaws and very much appreciated by the families.

Our grateful acknowledgments to the Children's Hospital of Western Ontario, Children's Health Foundation, Child Life for their donation of gifts for the children, Genzyme, metabolic food companies, metabolic team, administrative staff, genetics support staff, speakers, members of planning committee (which also included four mothers of children with inborn errors of metabolism), patients, families and volunteers.

Families and guests enjoy lunch and social time at the Metabolic Family Workshop

Families attending the herbal therapy for seizure disorders session

Cooking Demonstration

After an informative day learning about PKU many people were excited to have an opportunity to learn about cooking for the PKU diet. With the help of Dianne Sullivan from Cambrooke we were able to taste and see just how exciting cooking low protein can be. She made an amazing dish "Haluski" that tasted awesome and I'm sure several people went home to try it (I know I did with great success). We also had a chance to see new products that have been developed by Cambrooke (Camburgers and wraps) as well as see items that may already be in our kitchens and different ways to use them. Dianne also gave hints about cooking once and eating twice - a helpful hint no matter whom you are cooking for. It was fun to see the kids tasting the new foods and enjoying them. It was a great day with 20 people attending the event, held at Loblaws Wonderland Market. Thanks to all who attended and if anyone has ideas for next year please let me know.

Jennifer Culp

Low protein cooking demonstration at Loblaws May 14th 2005

Announcements

MCADD (Medium chain acyl-coenzyme A dehydrogenase deficiency) Study

We are very pleased to announce that soon a clinical study on estimating the incidence of MCADD (fatty oxidation disorder) will begin. This is a Canada wide study and will be timely,

as newborn screening for this disorder is not occurring in all provinces. Our metabolic group has taken the lead for this study. Dr. Chitra Prasad along with participants Dr. Tony Rupar and Dr. Kathy Nixon-Speechley (London), Dr Pranesh Chakraborty (Ottawa) Dr Jon Kronick (some of you will know him as the previous metabolic director at LHSC) and Dr Sarah Dyack (Halifax).

Invited Speakers

Congenital Disorders of Glycosylation (CDG)
Dr. J Jaeken, an extremely prominent researcher (the person who discovered this group of disorders) will be coming in November to LHSC to speak about CDG. All families with this disorder are welcome to attend.

Conferences

The Association for Glycogen Storage Disease 27th annual conference for families will be held in Calgary Alberta September 16th and 17th 2005. For more information, please visit www.agsdus.org

The 42nd annual symposium for the SSIEM (Society for the Study of Inborn Errors of Metabolism) will take place in Paris, France. World-renowned experts in the field of metabolics are meeting and sharing information on metabolic issues.

Featured This Issue

Dr. Tony Rupar PhD and Dr. Chitra Prasad MD

MCADD (Medium chain acyl-coenzyme A dehydrogenase deficiency)

Medium chain acyl-coenzyme A dehydrogenase deficiency (MCADD) is a rare hereditary disease that is caused by the lack of medium chain acyl-coenzyme A dehydrogenase enzyme required to convert fat to energy. Individuals with MCADD cannot fast for very long. Lack of prompt treatment can lead to low sugar and other changes in various chemicals in the blood resulting in seizures, coma, and development delay and, even death. The good news is that all these effects are preventable if MCADD is detected before symptoms arise.

MCADD is inherited in an autosomal recessive fashion. In this type of inheritance, both parents must carry a copy of the non-working MCAD gene, and both must pass it on to their child for the disease to occur. Parents who are both carriers have a 1 in 4 chance of having an affected child with each pregnancy. Children affected by MCADD could go for weeks, months or even years before it is noted that something is wrong. A simple flu could trigger the devastating affects of MCADD such as irreversible brain damage or death. Unfortunately even today, as many as 25% of individuals die in their first crisis, so it is imperative to know if a person is affected before they develop symptoms, to prevent a crisis.

Newborn screening for this preventable disorder using tandem mass spectrometry continues to pose a major challenge. The same blood spot that is used for PKU screening can be utilized to detect MCADD. The Ontario Ministry of Health is looking at this new development in newborn screening.

Lexi Age 11

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