The Inherited Metabolic Disorders News

Children's Hospital
London Health Sciences Centre

Winter 2009 Volume 6 Issue 1

In this Issue

From the Editor

Greetings Everyone!

I hope everyone had a wonderful holiday season.

Plans are now underway for the 6th annual **Metabolic Family Workshop** which will be held on Friday May 29th, as well as the 5th annual **Low Protein Cooking Demonstration** on Saturday May 30th, so please mark your calendars. More information to follow in the next few weeks.

Janice Little

From Dr Chitra Prasad

Dear all,

Happy New Year!

I wish you and your families the very best in the year 2009. A couple of months ago, I was able to travel to Mumbai India for a pediatric gastroenterology conference. It was wonderful to see so much interest in genetic and metabolic disorders. In this issue please read the very nice write up by Karen regarding her daughter Kennady. This reminds all of us to remain vigilant as some of the severe metabolic disorders may present even before we get results from the newborn screening.

The metabolic family group is growing. We as a metabolic team are very grateful for the support and input that we receive from the families.

Plans are already underway for 2009 Metabolic Family Day. I would encourage your participation. I am delighted as my mentor Dr Mark Korson (Tuft University Boston) has agreed to be our main speaker.

In the last year's resolution I had mentioned about the "two degrees change". This year my focus will be on being grateful for everything. I even found a website that is devoted to being grateful.

http://www.gratefulness.org/brotherdavid/a-good-day.htm

With best wishes

Your friend
Chitra Prasad

Personal Stories

Kennady Jorja Stone

My husband and I were like two kids counting down the days until Christmas…..for 9 months, in anticipation of our first child. We were really lucky. Pregnancy was very kind to me. No significant morning sickness, no swollen feet or hands, my fatigue level was tolerable and I was fortunate enough to gain only as much weight as necessary. Then came the labour. Again, we were lucky. Induction allowed a little more predictability and preparation, and the epidural was timed perfectly. A half hour of pushing and our perfect little peanut arrived. No cone head to speak of, 10 fingers and toes, a rosy pink skin colour, a great APGAR score, and she breastfed immediately like a little pro. Sounds almost too good to be true, you say? It took only 3 days for this fairytale to turn into a living nightmare. As new parents, we had prepared as best we could prior to the arrival of our first daughter. The baby's room was ready, the car seat was in, prenatal classes had been attended, and lots of reading was done. When we brought Kennady home 24 hours after delivery, life was good. Marking down her feedings, her poops, and the number of wet diapers on our hospital sheet indicated she was right on track. Then she didn't seem to want to eat very much. Her little nose was always cold no matter how much we wrapped her up. Her limbs stopped fighting us when we tried to get her to eat and her healthy cry became soft and weak. Most of her symptoms had logical explanations when I called for assistance three times that first day home. We were told of a breastfeeding clinic that would be open Monday morning that might help us out, but I needed someone to see my daughter immediately and reassure me that I was imagining the worst. 36 hours after bringing her home, our perfect little peanut was back at the hospital and barely responsive. What did we do wrong? Immediately the emergency staff recognized that Kennady was a very sick baby and they began testing and doing everything they could to ensure she survived. The initial suspicion was an infection and by starting antibiotics, we were told that she should be back home with us in a few days. At the time, we did not understand that the emergency doctors were saving her life by drawing blood to test her ammonia level. It was through the roof and fortunately we were too naïve to understand the severity of these numbers. All that we knew was that she had a metabolic disorder and my husband and I were the ones who had passed on the faulty genes. (We were later told that Methylmalonic Acidemia (MMA) was the name of her particular disorder.) As Kennady was brought up to the Critical Care Unit, I'll never forget the nurse telling us to go home and get our camera. She wanted to make sure we had a picture of our little girl in case she didn't make it. We eventually did go home to pick up a few

Personal Stories - continued

things, but the camera was not one of them. There was no way we were going to bring any negative energy near our little peanut as she was fighting for her life. The next week was a blur of numbers, tests, tears, medical jargon, and specialists, but since that first week, we have returned to working on our fairytale one step at a time.

Kennady spent her first month in the Children's Hospital receiving very special attention as we quickly learned that she was a pretty unique little girl. (As far as we know, she is the only one in Southwestern Ontario with MMA.) Never-ending testing continued and her diet became the main focus of her treatment. Most of the specialists told us that it was too early to tell how much damage had been done with such a high ammonia level but she should be followed closely. It seemed like we were booking appointments with anyone whose title ended in 'ology'. Nephrology, neurology, ophthalmology….. and we will be forever thankful for the exceptional care Kennady received and continues to receive. She has become a well-known frequent visitor to the PMDU but we are happy to report that the number of visits has reduced significantly. The only unfortunate thing about that is Kennady misses out on seeing her 'adopted' aunt as often, her dietician, Suzanne.

Kennady is now 9 ½ months old and has become quite a character. Her 3-tooth grin is infectious and so far she has achieved many milestones. She still surprises any medical professional that we meet for the first time who has looked up her history and has envisioned what this little girl might 'look' like. Most don't expect to meet our bright, blue-eyed little peanut chewing on her colourful toy and excited to be laying on the 'crinkly' paper in the examining room. As new parents, every day we learn something new. Unfortunately, we have had to learn things about feeding tubes, special diets, emergency protocols, sick day recipes and other important issues as with any metabolic disorder. Luckily, we are also enjoying learning about first smiles, reading books together, hearing 'mama' for the first time, and trying all kinds of crazy things to hear that priceless giggle. Unsure of what the future holds and what is in store for our little family, we can only look ahead one day at a time, continue to enjoy life's small pleasures, and do the best we can as we keep working on our very own fairytale.

Featured This Issue

Methylmalonic acidemia (MMA)

By Chitra Prasad MD FRCPC FCCMG

Methylmalonic acidemia is an inherited disorder in which the body is unable to process certain proteins and fats (lipids) properly. The effects of methylmalonic acidemia, which usually appear in early infancy, vary from mild to life-threatening. Newborns can present with lethargy, metabolic acidosis, hyperammonemia and coma. Affected infants experience vomiting, dehydration, weak muscle tone (hypotonia), excessive tiredness (lethargy), and failure to gain weight and grow at the expected rate (failure to thrive). Long-term complications can include feeding problems, developmental delays, kidney disease, and inflammation of the pancreas (pancreatitis). Without treatment, this disorder can lead to coma and death in some cases.

MMA can also occur due to maternal dietary B_{12} deficiency, which is found in vegetarians. Complementation studies have revealed the presence of at least 8 different complementation groups (mut0, mut-, cblA, cblB, cblC, cblD, cblF, cblH) that cause MMA. The incidence of MMA is 1 case per 48,000 infants.

Management includes very close monitoring of the diet to remove the offending amino acids and to ensure proper nutrition. Both liver and kidney transplant has been tried in some severe patients of MMA. A team of specialists and other health care professionals are required for ongoing follow up and monitoring such as dietitians, nephrologists, neurologists, metabolic physicians, pediatricians, occupational and physiotherapists and social workers.

This condition is inherited in an autosomal recessive pattern, which means both copies of the MUT, MMAA, or MMAB gene in each cell have mutations. The parents of an individual with an autosomal recessive condition are carriers of one copy of the mutated gene but do not show signs and symptoms of the condition.

Special dietary formula for MMA

Featured This Issue - continued

References
http://ghr.nlm.nih.gov/condition=methylmalonicacidemia
http://emedicine.medscape.com/article/947154-overview

Olaf A Bodamer, MD, PhD, FACMG, Professor, Department of Pediatrics, Biochemical Genetics and Neonatal Screening Laboratories, University of Vienna Children's Hospital, Austria
Brendan Lee, MD, PhD, Associate Professor, Department of Molecular and Human Genetics, Baylor College of Medicine

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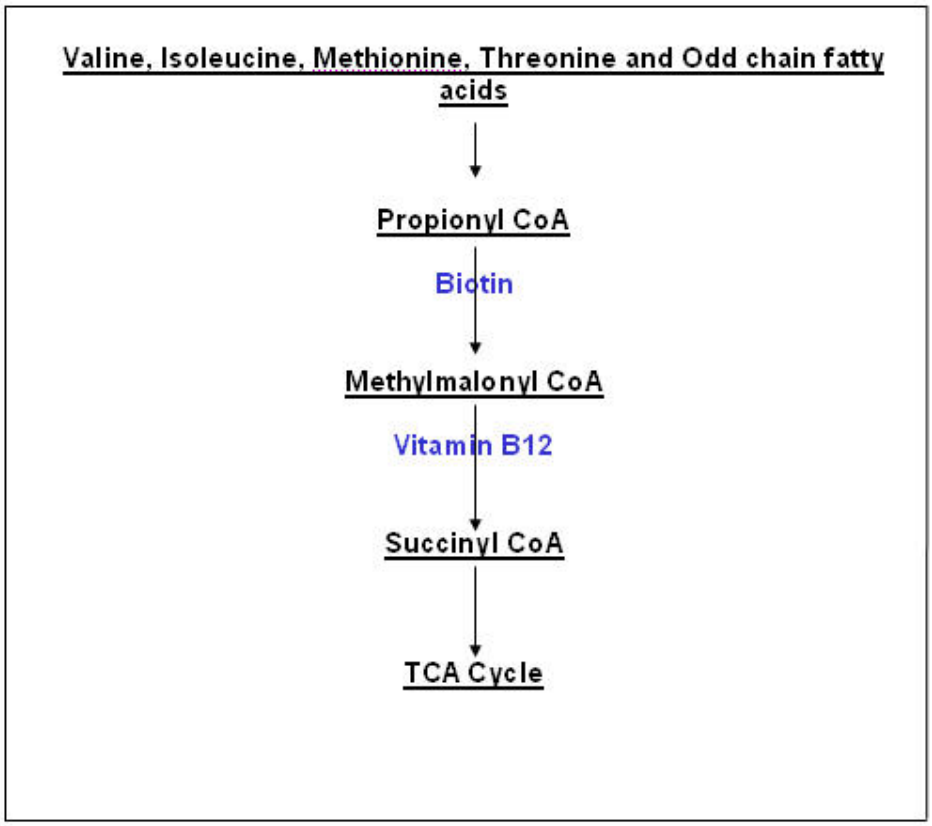
Methylmalonyl CoA And Its Metabolism



Suzanne's Corner

Taste connections(TC) Chocolate Chip Cookies by Malathy

2 ¼ cups (260 gm) TC Multi-baking mix
½ -3/4 cup tightly packed brown sugar
½ Cup butter or margarine
1 teaspoon vanilla
1 pkt (3.5 oz) vanilla instant pudding mix
¼ c (38 gm) Semi sweet chocolate chips
1 ½ teaspoons baking powder
Water to make stiff dough

- In a bowl measure the baking mix, and baking powder, and add one instant pudding mix packet. Using a wire whisk mix the dry mixture well. Preheat oven to 375 degrees F (190 degrees C).
- Spoon brown sugar into a mixing bowl, add margarine or butter, vanilla, and beat well until fluffy.
- Add the dry ingredients and mix thoroughly. Add water slowly until you get a stiff dough*.
- Mix in the chocolate chips to the mixture. Roll into walnut size balls, place on ungreased cookie sheet 2 inches apart. Bake for 12 - 15 min. Makes 2 dozen cookies (24)

Hint: * Add water slowly until you get really stiff dough. If it is too stiff to form the dough add extra water, ½ teaspoon at a time until you get the right consistency. Using an electric mixer speeds up the beating process.

Nutrition Information

	kcal	Protein	Phe	Met	Leu
Per Recipe	3213	4.86	185	65	318
Per Cookie	134	0.20	7.7	2.7	13.3

Tricks of the Trade

Jenn Culp (Mom of Daniel) says -

At an early age let your child have a shelf (or area) in your fridge and in your cupboard. Daniel and Celeste both have their own shelf in the cupboard for their snacks. Whenever they are hungry for a snack they ask and go and get one. Daniel shows me what he wants to eat and then I can mark it down in his book. Daniel also has a shelf in the fridge. His bread, cheese, milk etc is on it. When he wants to have something he goes into the fridge and grabs it. He knows he can eat anything on his shelf. I think this gives him the responsibility of learning about food/beverages that he can have.

What's New

URGENT: FOOD RECALL NOTICE from Cambrooke Foods

Dear Valued Cambrooke Customer,
Cambrooke Foods® is announcing a voluntary recall on its Imitation Cream Cheese and a market withdrawal of its Peanut Butter(TM) product. During routine testing, *Listeria* was detected in a new batch of our Imitation Cream Cheese before it was sent to customers. This batch of Imitation Cream Cheese products was destroyed while in our facility. While no reports of adverse events have been reported with any product shipped to our customers, we are taking the precautionary step to recall all products made on this machinery. Please read the Official Recall Notice below. If you have purchased any of these products since May 2008, please take the following actions immediately:

1. discard all units of these products remaining in your possession;
2. complete the online Recall Response Form click here to open form

http://survey.constantcontact.com/survey/a07e2f473t6fom1oqeq/a01hbfpqz5cd4/questions

We apologize for the inconvenience and concern that this recall has caused for you and your family and want you to know that your good health and the health of your family is our paramount concern. For more information, visit:

www.cambrookefoods.com/information/product_recall.php

Sincerely,
The Cambrooke Foods Family

What's New continued

A New Artificial Sweetener

Please be advised there is a new artificial sweetener on the market called "Neotame" that replaces "Aspartame" . Since it is a new product, warning labels stating it contains phenylalanine are not required .

Meetings and Symposiums

1. INCIDENCE OF MEDIUM CHAIN ACYL-COENZYME A DEHYDROGENASE DEFICIENCY (MCADD) IN CANADA USING THE CANADIAN PAEDIATRIC SURVEILLANCE PROGRAM (CPSP) (SEPTEMBER 2005-AUGUST 2008)

Presented by C Prasad, KN Speechley, S Dyack, CA Rupar, P Chakraborty and JB Kronick at the Canadian College of Medical Genetic meeting September 2008

2. ENZYME REPLACEMENT THERAPY AS ADJUNCT TO CORD CELL TRANSPLANT IN HURLER SYNDROME

K. Corley, C. A Rupar and C. Prasad

Lysosomal Disease Network WORLD Symposium 2009
(Co-organized by Lysosomal Disease Network and the National Institutes of Health)

Will be presented on February 18-20, 2009

Newborn Screening Update

The Culp Family's experience with Newborn Screening:

November 2003

Our story begins on April 25th, 2003 when our wonderful baby boy, Daniel, was born. We were thrilled to bring him home and proudly showed him off to family and friends. His sister, at 22 months of age, was delighted to hold him and lovingly welcomed him into her world. Our midwife came to our home for his routine newborn screening in his first few days of life, which included obtaining blood from the baby's heel to test for phenylketonuria (PKU). It came as a total shock when she phoned to tell us that Daniel's PKU screening test was positive and that we must travel to London's Children's Hospital the next day for further assessment.

Daniel was only twelve days old, and although he was somewhat fussy, he appeared perfectly healthy to us. We were terrified by the news because we really had no idea what his diagnosis entailed. With great uncertainty and apprehension, my husband and I traveled to London to meet with a doctor and dietitian who specialized in this condition.

The news was overwhelming…Daniel was lacking an enzyme that prevented him from breaking down phenylalanine (PHE), an essential amino acid found mostly in foods that contain protein such as meat, fish, eggs, cheese, milk products and even bread. His blood test revealed that his levels of PHE were very high and consequently, he was immediately prescribed a special formula (Lofenalac). Left unchecked, the excess PHE would accumulate in the blood and lead to retarded physical and intellectual development. It was devastating news and, though the professionals had provided us with some information, our minds were spinning with unanswered questions and concern for the well being of our child.

Meanwhile, at, home, our families were investigating the topic of PKU on the internet, looking for some hope and reassurance to ease our fears. We spent the following week reading and digesting some of the information, making note of questions which we brought back to the hospital a week later at our next appointment. We continued to travel to London twice a week for the next six weeks until Daniel's levels had fallen to within the normal range. After that, we visited the hospital almost weekly until Daniel was almost six months old. At each visit, he would have blood drawn and then we would return home and eagerly await the results. With the support of the dietician, I was also to continue breastfeeding for the first few months, supplementing with the special formula as instructed. Later, we learned to balance the Lofenalac with infant formula off the shelf.

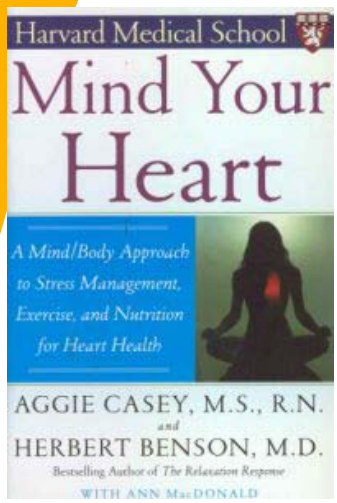
His special formula with be a staple in his diet throughout his lifetime, and he will always need to adhere to a special diet. We are still working with the dietitian as we introduce new foods and record everything he eats and drinks to assure that he gets only the allotted amount of intake of PHE (which he requires for normal growth) and his inability to break down this amino acid. The intake of PHE required varies with each individual child and may change depending on what period of growth they are in.

The condition that Daniel has is hereditary – both my husband and I are carriers and, as such, there was a twenty-five percent chance that our child would be born with PKU. Our daughter does not have this condition, though she may be a carrier, and she will likely undergo testing at a future time to determine her status.

The necessity of the heel prick-screening test done on newborns is sometimes questioned because a positive result is so rare (less than one in 10, 000 children). Daniel is the reason this test in so worthwhile. Thanks to early detection and swift intervention, Daniel will be able to enjoy a 'normal' childhood and a promising future.

Jennifer and Don Culp

Mind Your Heart



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

- **Humour is a great coping strategy. Try to find something funny in a situation.**
- **Take a "mental health" day.**
- **Practice being patient. Create patience practice periods; for example, when waiting in line. Watch your thoughts; focus on your breathing.**
- **Understand that we do not all see or do things in the same way.**
- **Practice mindfulness. Learn to live in the moment.**

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!

Faith G
age 7

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