The Inherited Metabolic Disorders News

Winter 2007 Volume 4 Issue 1

In this Issue

From the Editor

Greetings to everyone!

Once again the parent planning committee and the metabolic team are actively organizing the 2007 **Metabolic Family Workshop and Low Protein Cooking Demonstration**. In the "Announcements" section of the newsletter you will find a lot of information about the talks that are being planned as well as plans for a "Silent Auction".

This year's workshop is unfolding to be even bigger and more exciting than ever. A full brochure with all the events details will be mailed out in the next few weeks. We hope to see you there!

Please continue to send your contributions, ideas and stories for the newsletter, as well as topic ideas for the Metabolic Family Workshop.

Janice Little

From Dr. Chitra Prasad

Dear friends,

Next month I will be attending my 25th year medical school reunion in India. Time flies so quickly. Each passing moment has taught me about the courage and resilience of the human spirit.

I thank all my team members and parents in our committee.

Please send your comments and suggestions to us. Hope to see you all there.

Best wishes,

Your friend Chitra Prasad

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

Albert Einstein

Winter 2007 **Volume 4 Issue 1**

Personal Stories

BREANNA'S STORY as told by Corrine Arthurs

Breanna Corinne Arthurs was born on September 19, 2002, weighing 5 lbs 4 ounces…such a beautiful little girl. Even through my pregnancy I knew something was not right and that feeling continued after she was born. She was such a quiet patient baby. We took her home and introduced her to her big sisters. Breanna, better known as Bree had completed our family. I first noticed a seizure when she was only 4 weeks old, on October 10, 2003. It lasted only seconds. Then I was not even sure it was a seizure but now I know.

Sweet Bree never picked up many milestones. She was never able to hold her head up. I just kept thinking "OK all children develop at different rates." That did help a bit. But there was still this feeling that I always had that something was not right. Bree was 2 1/2 months when the crying started. Oh boy, unless you have heard a Krabbe baby cry, you have heard nothing. In December she would cry for maybe 6-8 hours a day. Yes that sounds like a lot but nothing compared to what was to come. She seemed to be in so much pain. We tried everything. And what I found amazing is that not once did anyone ever get upset about the crying. We all would just cry along with her.

Breanna started to smile and laugh in January. It was a real task to get her to smile and laugh but when she did it, it was like music to our ears; so pure, sweet and innocent. She used to love it when her daddy would put her in her car seat and run her around the kitchen. Oh, she just laughed. I can still hear it. It is now like a dream.

January was also when things got so bad that we searched and searched for a doctor, however, none would take her on. We took her to the hospital weekly. She was twitching a lot (now we know they were myoclonic jerks), her chest was always so loud, and vomiting like crazy. And crying, crying, crying. The hospital would always do x-rays and then send us home saying they found the problem. It was an ear infection. I was getting so sick of hearing that nothing else was wrong, because something was terribly wrong.

Winter 2007 **Volume 4 Issue 1**

In the beginning of June I notice again another seizure and panicked I called a doctor from the phone book. I have called this doctor before but this time I was not taking no for an answer. I said to him, "something is very, very wrong with my little girl" and he said bring her right in. I packed her diaper bag and we left right away. When we first got there he called us right back. She was crying but he had me lay her on the bed. He checked her hips and she was so tight and boy did she scream. She tried to check to see what her neck control was and she did not have any. I told him she has never been able to hold her head. For me this has been part of our life now for 9 months so it was normal for us but it shocked the heck out of the doctor that the hospital had not said something. I also told him that she had never been able to do that. He examined her for only a few minutes. That was all it took to know something was really wrong. I heard him talk to his secretary. He told her to book her in to see a pediatrician and try to get her in ASAP. He told her Bree was very sick. When we got to the pediatrician a week later he told us to get her to the Emergency ASAP. He said she sounded like she was in respiratory distress. I tried to explain that is how she had been for months now and the hospital kept sending us home. Well we took her to a different hospital; London's Children's Hospital of Western Ontario. Because of Bree's non-stop crying her heart rate was up to almost 200 beats per minute. They took x-rays right away and they were clear, so they called the neurologist. Well that got everything rolling. Finally we were not just paranoid parents. The neurologist witnessed a seizure and admitted her that day.

I had told the neurologist our trouble with trying to get medical help he was so upset. He said it is so obvious that something is very wrong. They could not get blood from her arm so they had to use her head. Now Breanna was not able to suck anymore so we had to use "thick it" in all of her formula. I would put it on a spoon and put it in her mouth. Then I would have to rub her neck to get her to swallow it. NOW HOW NORMAL IS THAT? My heart was just breaking for her.

The neurologist came in every morning saying the same thing, "How is Breanna the irritable doing this morning?" How true. I wanted her to be happy again so bad. He came into our room the day before we went home and said he wanted to talk to us and he took us to another room to talk to us. Oh that is never a good sign. He said Breanna had a terminal illness however he didn't have a name for it yet. He said she would most likely not make it to 2 years old. Her MRI showed her white matter in her brain was abnormal and showed demyelination in parts of her brain. The padding around the wires of her brain was disappearing. Without it her brain cannot send out messages to the body. Eventually the brain will be unable to tell the body to breathe, for her heart to beat. He said it looked like a Leukodystrophy. All of them are terminal.

I pretty much knew that before he told us but that was really hard to take. We took her home a got a camcorder and a camera and went crazy with them. It was really hard to know Bree was going to die and there was nothing we could do but watch her as her body shut down.

July 2, 2003 when she was 10 months old and something strange happened. She was sitting with her grandma and it was quiet. I ran over to her I thought something was wrong. Well she was sleeping and well, that was it for the crying. Just like it came it ended. Now instead of crying all the time she was now sleeping all the time. I think this is the point where Breanna went from Stage I to Stage II. She was now only awake for approximately 2-4 hours a day. We continued to fight for a diagnoses and our neurologist sent us to a metabolic doctor.

December 23 was her first major pneumonia and she was admitted in the PCCU. She was so sick. I stayed with her again like I always did. Oh, that was so hard. Dec 23 to Dec 31 we spent in the PCCU. Christmas was very difficult. I just prayed God would not take her on Christmas. I could not even hold her because she was hooked up to so many machines. She was released January 1, 2004.

Winter 2007 **Volume 4 Issue 1**

January 16, 2004 was the day we got the horrible news. Our little Breanna had Krabbe disease. I had done so much research. I knew what Krabbe disease was. It was one of the diseases I was praying she did not have, with a life expectancy of 16 months. She was 16 months old the day we got her diagnosis. Oh, how hard to have a doctor tell you that your 16 month old little princess was going to die and soon. And that there was nothing you or anyone can do.

January 18, 2005 - Bree is 2 years 3 months and 30 days

Our little angel Bree flew up to heaven the morning of January 18, 2005. Our little Bree never woke up. She had been sleeping since Sunday at 10 am. We ran her up to the emergency early Monday morning because her seizures got so bad.

Her oxygen level was in the 60's heart rate 189. Her blood pressure kept dropping. She was not responsive and our angel had reached the 3rd stage of this devastating disease. So we made the choice to bring her home. Breanna loved being home more than anywhere else. We told the doctors to do what ever they could do for her. They gave her 15 liters of oxygen at 100%. Her colour was still very poor and her oxygen levels would not improve. The doctors did not think she would make it much longer.

The hospital called us an ambulance and we took her home. Our little Bree was fighting so very hard. Her breathing was still very shallow and fast. At 4:00 am on Tuesday Bree was making some noises I have never heard. I gave her some Motrin. After I gave her the Motrin she made noises, beautiful little sighs. I keep thinking she must have seen Heaven. At 4::20 she made a large gasp. I was holding her and praying. I just kept telling her please Bree please Bree don't go. I just wanted her to keep fighting. This happened so quickly and I was begging her to breathe, but she had already seen Heaven. I put the stethoscope to her chest and I could no longer get a heart beat. I hugged her and put her down. I went upstairs to get Brian. I kept her oxygen on just in case. He came down and I again picked her up and checked her heart rate and still no heart rate. Oh, no, not now. Not ever. My beautiful little Bree has left us; however, she had gone to a far better place.

Winter 2007 **Volume 4 Issue 1**

Kids Korner

Birthdays: January, February and March

Jesse	PKU	January 5, 2000	7
Abrahem	PKU	March 31, 1993	14
Megan	PKU	March 31, 1998	9
William	PKU	February 26, 1997	10
Cody	PKU	March 26, 2006	1
Charis	PKU	January 26, 2005	2
Madeline	CDG	March 28, 1998	9
Kody	Gaucher's	January 3, 1991	16
Hannah	GSD	February 21, 2006	1
Christine	GSD	January 31, 1997	10
Tyler	Homocystinuria	March 26, 1992	15
Elvina	MCADD	February 5, 2001	6
Shawn	MCADD	January 2, 2002	5
Lucas	Morquios	March 7, 1997	10
Zane	Morquios	February 12, 1999	8
Stefan	Morquios	January 15, 2000	7
Megan	Retts	February 7, 1994	13
Curtis	SLO	February 2, 1994	13
Edward	Transcobalamin II Deficiency	March 6, 2001	6
Ethan	Metabolic Disorder	January 1, 2002	5
Gideon	Metabolic Disorder	February 28, 2003	4

CDG - Congenital Disorder of Glycosylation
GSD - Glycogen Storage Disease
MCADD - Medium Chain Acyl CoA Dehydrogenase deficiency
PKU - Phenylketonuria
SLO - Smith-Lemi-Opitz

Happy Birthday Everyone!

How to Make a Donation

Donated funds are used for future Metabolic Family Workshops 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!

Winter 2007 **Volume 4 Issue 1**

Suzanne's Corner

Brownies

Ingredients
1/2 cup butter (1-stick) softened
1 1/2 cups packed brown sugar
2 tsp vanilla extract
1/4 cup corn syrup
3 Tbsp unsweetened powdered cocoa (14 g)
1 tsp salt
2 tsp baking powder
2 cups packed CBF All Purpose Baking Mix (250 g)
1/2 cup water

Directions

1. Cream together butter and brown sugar until fluffy. Add vanilla.
2. Add the remaining ingredients and stir until all ingredients are moistened.
3. Spread brownie mixture into a greased 8-inch square baking pan.
4. Bake at 300 degrees F for 1 hour. When brownies are cooled, cut into 16 brownies

Enjoy!

	Per Recipe	Per Serving
PHE:	190 mg	12 mg
LEU:	187 mg	12 mg
Pro:	4 g	0.2 g
Calories:	3370	210

Servings: 1 brownie
Total Protein: 0.2 g per brownie
Phe per serving: 12 mg

From the Cambrooke Foods website : www.cambrookefoods.com

Winter 2007 **Volume 4 Issue 1**

Announcements

Metabolic Family Workshop
Bestwestern Lamplighter Inn
London, Ontario
Friday May 11, 2007

Low Protein Cooking Demonstration
Loblaws on Wonderland
London, Ontario
Saturday May 12, 2007

Tentative Schedule for the 4th Annual Metabolic Family Day May 11, 2007

Time			
1030 – 1100 h	Registration		
1100 - 1130	Welcoming Remarks, Metabolic Disorders Simplified - *Dr. Tony Rupar, Laboratory Director, CPRI*		
1130 - 1200	Obtaining Accurate Internet Searches - *Lance Le Roux*		
1200 – 1300	Lunch		
1300 – 1400	Key note speaker Maintaining a Balanced Lifestyle - *Dr. Martin Collis, Psychologist, Motivational Speaker*		
1400 – 1430	*Lindsey Moir - School Advocate*		
1430 – 1440	Groups splits		
	Group 1 The Child with Disabilities	**Group 2** For Teenagers/ Young Adults Only	**Group 3** Coping Strategies for a Lifelong Disease or Diet Management?
1440 – 1600 (20 min break included)	*Paul Strickland Siskinds Law Firm* Henson Trust Break	*Beth Mitchell Psychologist and Brian Seeley Social Worker* Break	*Rénald Richer & Martin Collis* Break
1600 – 1610	Groups reconvenes		
1610 - 1630	Newborn Screening Update, Closing Remarks *Dr. Chitra Prasad, Metabolic Director London Health Sciences Centre*		

Winter 2007 **Volume 4 Issue 1**

Announcements

Host Families

Both Sam Smith and Stacey Taylor have graciously offered to host out-of-town families requiring overnight accommodation for those attending the workshop and cooking demonstration. Please call Janice Little at 519.685.8453 for further information.

Accommodations at the Best Western Lamplighter for the Metabolic Family Workshop

15 rooms have been set aside until April 11, 2007 at a cost of $139.00 per night. Call the hotel to book a room at 519.681.7151 as soon as possible to ensure your room is reserved. Be sure to tell them you are with the Metabolic Family Workshop to get the special rate.

Silent Auction

This year to help offset the cost of the Metabolic Family Workshop we are planning a "Silent Auction" on the event day. Donations of items and services are welcome. Please contact Brian Seeley at 519.685.8300 extension 52456 if you wish to make a donation.

Past and Future Meetings Update

Newborn Sreening Meeting

There will be a provincial meeting on February 23rd, 2007 in Ottawa to discuss issues emerging from the expanded newborn screening program, as well as resource planning.

Garrod (National Metabolic Experts) Meeting

To be held on May 5th and 6th, 2007 in Vancouver, BC.

Winter 2007 **Volume 4 Issue 1**

Featured This Issue

Krabbe Disease

Chitra Prasad MD FRCPC (Clinical and Metabolic Specialist)

Krabbe disease is a rare degenerative 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".

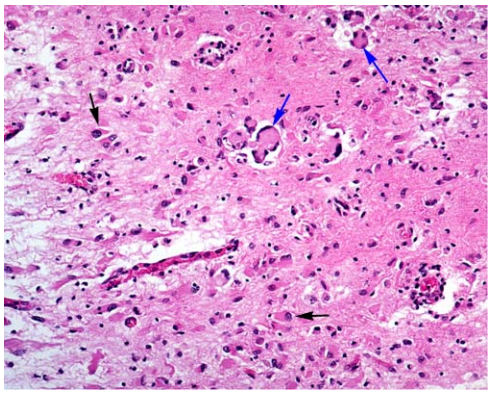
Krabbe disease is caused by the deficiency of enzyme "galactocerebrosidase" a key enzyme involved in the biochemical pathways of the body that handles myelin, a nervous system compound made of protein and fat (lipid).

Clinical symptoms of this condition include; irritability, unexplained fever, limb stiffness, seizures, and 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. 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.

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. I hope as our understanding of the functioning of genes and proteins continue to advance, there will one day be a cure for this devastating disorder.

Resources

- Global Organsation for Lysosomal Diseases
  www.goldinfo.org
- United Leukodystrophy Foundation
  www.ulf.org
- The Myelin Project of Canada
  www.myelinprojectcanada.ca



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).

Winter 2007 **Volume 4 Issue 1**

Carter, age 4
PKU

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