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From the Editor

Hello everyone,

The air is crisp, the days are shorter and once again thoughts turn to planning the next Metabolic Family Workshop (Friday May 11, 2007) and Low Protein Cooking Demonstration (Saturday May 12, 2007)

There will be more details will follow as soon as they are available.

Janice Little

From Dr. Chitra Prasad

Dear Friends,

A new school year has started, there is lots of activity going on not to forget the amount of construction going on with the London roads recently. This is true for our lives, too...an endless process of starting and finishing our jobs.

Our most recent meeting for the Metabolic Family Day was full of enthusiasm and new ideas. We would like to focus on the families and relationships in the upcoming Metabolic Family Day 2007.

In our metabolic team we had some changes, too! Robin Dyson has left us for a job closer to her community. We'll miss you Robin!

Joanne Psiuk-Rodgers has joined us and we are very happy to have her in our team.

Do send your ideas and stories for the newsletter and workshop to us.

My sincerest best wishes to each and everyone.

Your friend,

Chitra Prasad

"It takes a whole village to raise a child"
African Proverb

Personal Stories

Living with Gaucher's Disease and Accepting It

My name is Patricia McDowell and I have Gaucher's disease. I was diagnosed in nineteen ninety two, when I was just five years old. I was brought to the doctor's office with a low grade fever and a cough, which the doctor prescribed antibiotics. When I did not seem to get better, he followed up with some blood-work. The results concluded that there was something else wrong with me besides having a common virus. The call came midmorning on Easter Sunday and everything was packed up for a trip to London Hospital for Sick Kids. I was in the hospital for five days and during that time underwent many tests to rule out different diseases. The test that I remember the most is the spinal tap that was performed to rule out leukemia. The biopsy of my liver was the test that diagnosed my Gaucher's disease. This occurred fourteen years ago, and I am now nineteen years old and attending the University of Windsor in second year of the nursing program. I believe that having the disease had a lot to do with my decision to go into nursing. My life has always revolved around the medical field, seeing many

doctors and nurses every time I had to go to the hospital. I just hope that I will be able to make a difference in someone's life the way that many have done for me.

The way that I take care of my health now is a lot different from what I had to do when I was younger. In elementary school, I was not allowed to participate in the contact sports that were taught in physical education because of my enlarged spleen and liver. Most of the time I had to sit out and watch everyone else. This was hard for me because all that I wanted was to be a normal kid like everyone else. I was discouraged from having sleepovers at friend's houses because it was hard for my parents to have to explain to another child's parent about the disease, when the only illness that they had to deal with was colds and the flu. A few of my close friend's parents knew but most of the time, I had my friends over to my house for a sleep over.

Now that I have been performing the enzyme replacement therapy for eleven or twelve years, the way that I take care of my health has changed. I consider myself to be a quite normal nineteen-year-old university student. I go to the gym when I can find time and take my vitamins. I am still somewhat cautious about the activities that I do, but I am not limited from any tasks or sports. I am quite active because of school, work and a social life. I am not as tired as I used to be when I was younger and I find that I have a lot more energy to do the things that I want to do. Overall I would say that I am a lot healthier than I used to be.

Living with Gaucher's disease has been challenging at times. When I was younger and the teacher's were learning the students' names at the beginning of the year, they always knew my name first because of talking to my parents. Every year before the start of the school year, my parents would call the school to talk to my teacher and brief them about their daughter and the genetic disease. I used to get uncontrollable nosebleeds because of my low blood counts, sometimes as many as a two or three a day. Getting them at school was the worst because no one really seemed to know how to deal with it. I would have to go and sit in the office with an ice pack until my parents were called. The nosebleeds had to be timed, and if they exceeded more than twenty minutes, I had to go to the hospital and possibly get my nose packed or cauterized. Since starting the enzyme replacement therapy, I do not get the nosebleeds as often. They are not completely gone, but they have lessened in severity and I have not had to go to the hospital because of uncontrollable bleeding in about seven years. My low blood counts caused me to feel tired all of the time. I did not notice exactly how tired I felt until I started the replacement therapy because I was so young and I did not really know how it felt to not feel tired.

When I was nine years old, I had my first surgery to have a titanium port-a-cath inserted into my chest, so that I could do the infusions out of my house instead of having to go to the hospital. A V.O.N. nurse would come to my house every other Thursday morning to access the port and set up the infusion of the enzyme replacement therapy. During that time, we only had one complication, which was a bubble of fluid that started to build up in my neck. London hospital, where I had the port put in was called and an emergency trip was taken to ensure that the port had not become dislodged from the jugular vein and the fluid bubble was not blood. It turned out that the needle was not inserted all of the way to the back when it was accessed and when the saline was flushed through the line, it built up causing a bubble.

Eventually my mother was trained on how to access the port and how to prepare and set up the infusion. After the V.O.N. nurse trained us, none of the medical supplies were covered under insurance so my parents had to pay for all of the needles and supplies out of their own pocket. My mother found it emotionally difficult to access the port after the V.O.N. nurse left, because she knew that it hurt me, but she overcame it because she knew that it had to be done. She used to tell me that it probably hurt her more than it hurt me.

I had that port-a-cath in for almost seven years, when it started to wear out. I had my second surgery at the age of fifteen to have a new one inserted. The surgeon was not able to put it back in on the same side of my chest, so it was inserted on the other side. The V.O.N. came back and retrained my mother on how to access the new port. Today, I am performing my own injections and setting up the enzyme replacement therapy.

The largest issue that I deal with now is the keloid scarring on my chest from the surgeries. I have gone to the hospital numerous times to receive cortisone shots to reduce the swelling and redness of the scarring. The shots did not take them away but they reduced their visibility. They are still noticeable but I do not get questioned about them as much as I did before. It is also difficult to find time to do the enzyme replacement therapy because it takes about three hours to do. It is hard to try and juggle work, school and a social life when I have to dedicate about six hours a month to doing the infusions. I am receiving four thousand units a month of Cerezyme, which is broken up into one therapy every other week.

Overall I believe that having Gaucher's disease has made me who I am today. I think that it has helped me in my school work because of all of the predisposition that I have had to the medical field. I also think that it will help me in becoming a better nurse because I will have been on both ends of the spectrum, the nurse side and patient side. I can understand the fears and concerns that families have as well as the fears and concerns that the patients have. I would like to thank Dr. Clarke and his staff for all of his work as well as Genzyme and the National Gaucher Foundation for their support. Without their persistence I would not be where I am today.

To accomplish great things we must dream as well as act

French Novelist Anatode France

Kids Korner



October, November and December Birthdays:

Child's Name	Metabolic Disorder	Birthday	Age
Nolan	PKU	October 18, 2005	1
Michael	PKU	October 12, 2001	5
Mitchell	PKU	November 19, 1990	16
Emma Lynn	PKU	November 2, 2000	6
Adam	PKU	November 2, 1998	8
Raphael	PKU	October 23, 1999	7
Laura	PKU	December 29, 1993	13
Vanessa	PKU	October 6, 2003	3
John	PKU	October 25, 2004	2
Brady	ALD	October 2, 1993	13
Joseph	Amish Microcephaly	December 7, 2002	4
Cole	Arginase Deficiency	November 1, 1997	9
Michael	CPS Deficiency	December 16, 1996	10
Colleen	MELAS	October 24, 1992	14
Warda	Metachromatic Leukodystrophy	November 14, 2000	6
Ryan	MSUD	October 25, 1992	14
Kareem	Niemann Pick-B	November 23, 2002	4
Joseph	PDC	October 17, 2000	6
Sarah	Sanfillipo	December 4, 1997	9
MacKenzie	Not Diagnosed	November 16, 1999	7
Marrisa	Not Diagnosed	December 10, 1998	8
Gavin	Not Diagnosed	November 22, 2003	3

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

Happy Birthday Everyone!

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!

Resources

- ◆ **The Canadian Society for Mucopolysaccharide & Related Diseases Inc.**
www.mpssociety.ca
- ◆ **Chippery**
www.pkuchips.com (Low Protein Chips)



Check out these favourite snacks –

Tracey suggests: Munchy Bites, Sweet BBQ flavoured from Cambrooke Foods. A 16 gram serving has only 0.03 g Protein and 1 mg Phe. These are really low protein and great tasting!

Daniel recommends Cassava Chips from your local Bulk Barn Store. A 28 gram serving of chips 20 mg Phe & 0.4 g Protein (see page 123 *Low Protein Food list for PKU*, Virginia Schuett, 2nd ed.).

Apple Crisp

Ingredients

4 apples, peeled, cored and sliced
1 1/2 packed cups (187g) Cambrooke Foods (CBF) All Purpose Baking Mix
1/4 cup sugar
1/2 cup brown sugar
1 tsp. cinnamon
1/2 cup butter or margarine, cut into pieces
dash salt, optional

Optional: For an awesome variation, add 3/4 cup cut up strawberries to the apples and sugar portion of the recipe.

Directions

1. Heat oven to 350 degrees F.
2. Mix apples and sugar together and spread evenly in the bottom of a greased baking dish.
3. Blend CBF All Purpose Baking Mix, brown sugar, cinnamon, salt and butter pieces together until mixture is crumbly using a fork or food processor.
4. Spread evenly over apples and press down firmly covering all of the apples.
5. Bake for 30-35 minutes or until the top is golden brown.
6. Makes six servings.

Serve with a dollop of frozen non dairy creamer such as cool whip.

	Per Recipe	Per Serving
PHE:	84 mg	14 mg
LEU:	79 mg	13 mg
Pro:	2.3 g	0.4 g
Calories:	2410	400

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Framingham, MA 01701
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Announcements

Tiny Hands Provide Big Pay-cheque to LHSC's Medical Genetics Program



Article from **the Page**

LHSC staff newsletter Vol: 11, Iss: 13 - Aug 15, 06

Parents Dana and Nick Walczak Sr. present a cheque of \$5,000 to the Medical Genetics program to Dr. Chitra Prasad, Medical Geneticist, Paediatrics LHSC. Proud son Nicholas Jr. approves as he sleeps in his wagon.



July 5, 2006 was a very special day for the Medical Genetics program at LHSC. They received a gift of \$5,000 for further research, from one of their smallest patients, Nicholas Walczak.

Nicholas was diagnosed with Tay-Sachs disease in April 2005 and receives care through the Medical Genetics program at LHSC. Tay-Sachs disease is a fatal genetic lipid storage disorder in which harmful quantities of a fatty substance called ganglioside GM2 build up in tissues and nerve cells in the brain.

The Walczak's raised the money for the Medical Genetics program through a fundraising golf tournament, Nickels for Nicholas.

"We hope the money raised can help find solutions for Tay Sachs Disease and other fatal genetically inherited diseases," says Nicholas' dad, Nick Walczak Sr.

"Dealing with metabolic disorders especially the fatal neurodegenerative disorders such as Tay-Sachs Diseases can be extremely challenging for any family," says Nicholas' doctor, Chitra Prasad, Medical Geneticist, Pediatrics LHSC. "As Nicholas' physician I have always admired the Walczak family for their love, dedication not only for their son, but also for all children affected with similar disorders. The Walczaks have truly made a difference by their positive attitude and thinking."

Those with Tay-Sachs disease appear to develop normally for the first few months of life. Then, as nerve cells become filled with fatty material, a relentless deterioration of mental and physical abilities occurs. Those affected may become blind, deaf and unable to swallow. Their muscles begin to atrophy and paralysis sets in. Other neurological symptoms include dementia, seizures and an increased startle reflex to noise.

Both parents must carry the mutated gene in order to have an affected child. In these instances, there is a 25 percent chance with each pregnancy that the child will be affected with Tay-Sachs disease. A simple blood test can determine if the parents are carriers.

The second annual golf tournament was held at Maple Ridge Golf Centre and sponsored by many community businesses including Hi Tech Auto Care, the Valkyrie Riders of Southwestern Ontario and VIPond Sprinkler Systems. Community members made donations to help further the important research of the Medical Genetics program at LHSC. Tay Sachs disease remains one of the lysosomal disorders where presently there is no curative treatment.

"We hope and pray everyday for a miracle or a cure for my son's disease, and maybe this money can help. But if not him, it may help a baby girl or boy in the future to have a better life," says proud Dad, Nick Sr.

What's New

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

Metabolic Family Workshop and Low Protein Cooking Demonstration

There are a couple of local families who have graciously volunteered to host families that require overnight accommodation.

More details will follow in the next newsletter.

New Metabolic Team Member

Welcome to Jo-anne Psiuk-Rodgers, Administrative Associate for the Metabolic Team. Jo-anne previously worked in Hematology at LHSC



Congratulations to Metabolic Team member **Brian Seeley** on the birth of his daughter!

Marin Nicole Seeley
Born Sept 20, 2006
8 lb 8 oz

Featured This Issue

Compiled by Kathy Corley
Professional Practice Leader, Genetics/Metabolics LHSC

Gaucher Disease

The term "lysosomal storage disorders" applies to over 40 different rare inherited metabolic diseases

Gaucher disease is the most common lysosomal storage disorder. This disorder is caused by a deficiency of the enzyme glucocerebrosidase, which is required for the breakdown of a specialized lipid called glucocerebroside. This breakdown occurs throughout the body, particularly in the liver, spleen and bone marrow. Accumulation of glucocerebroside in people with Gaucher disease leads to organ damage (specifically the liver, spleen and bone marrow). These accumulated cells are called Gaucher cells. Gaucher disease is inherited in an autosomal recessive manner, meaning that an individual must inherit two "altered" genes in order to be affected. Carriers of this disorder are healthy.

Individuals with Gaucher disease are often chronically anemic as a result of replacement of normal bone marrow by "storage cells". Anemia can sometimes become severe enough to require periodic transfusions. Accumulation of glucocerebroside in the bone marrow causes weakening of the bone, causing fractures. In a few, disturbances of the circulation to the bone causes periodic attacks of excruciating pain in the hips, knees, and shoulders. In these individuals, interruption of the circulation to the bone often causes destruction of joints resulting in a requirement for major surgical treatment. People with Gaucher disease are also more apt than others to develop bone infections. Accumulation of glucocerebroside in the liver may cause cirrhosis, resulting in bleeding into the stomach and gut, jaundice, swelling of the ankles and eventually death. Many individuals with this disorder suffer only mild signs and symptoms and remain undiagnosed. Even those with major complications often face a long journey, visiting many diagnosticians before their diagnosis is confirmed.

In the past, treatment of Gaucher disease focused on symptomatic treatment of pain, surgical treatment of fractures, infections, and avascular necrosis of bone, surgical removal of the spleen to relieve the thrombocytopenia caused by hypersplenism and blood transfusions to correct the anemia of the disease. Patients with very severe disease have also been treated by bone marrow transplantations. However, this requires the availability of a suitable donor and the procedure is associated with prolonged hospitalization and morbidity as well as a high mortality rate.

Since the early 1990's, treatment of Gaucher disease by enzyme replacement therapy represents a major advance in the management of this disease. The missing enzyme, glucocerebrosidase was originally extracted from human placentas and chemically modified to enhance its effectiveness. The modified enzyme was given by intravenous infusion and proved to dramatically decrease the hematologic and bone complications of the disease. Over the years, this enzyme was produced en masse using new biotechnology which allowed adequate supplies of the drug Cerezyme, which is now routinely used to treat individuals with Gaucher disease.

In Ontario patients with Gauchers are routinely monitored and when necessary, enzyme replacement therapy is instituted. This allows individuals to lead normal lives, free from the signs and symptoms of disease. It is important to recognize that not all individuals require this therapy, however those that do now have hope where only a few years ago there was none.

Please watch in future newsletters for more information related to enzyme replacement therapy. While these treatments provide new hope for patients with rare disease, there are many issues related to adapting to lifelong therapy involving frequent infusions and medical tests.



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