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Butchart Gardens

From the Editor

The warm weather has finally arrived!

Our 5th annual Metabolic Family Workshop on May 30th and the 4th annual Low Protein Cooking demonstration on May 31st was another huge success. Read more about these events under the "What's New" section.

As always, your suggestions and stories are welcome. Please contact me by email (Janice.little@lhsc.on.ca) or telephone: 519-685-8453 if you wish to contribute to the newsletter.

Have a wonderful summer!

Janice Little

From Dr Chitra Prasad

Dear all,

The school year has finally coming to an end. I am sure many of you are busy making plans for the summer. Our metabolic family day and the low protein food demonstration workshop were both a grand success with everyone's help.

Dr Barbara Marriage, our main speaker is also a very experienced dietitian who brought wisdom and a sense of humor to her talk on diet for metabolic disorders. Dr Stephen Lafay's inspirational talk gave us all some "food for thought". Dr Lafay displayed a lot of enthusiasm and shared with us some nice messages for life.

Shawn's story in this newsletter is extremely inspirational. His mother has done a tremendous job.

Last week I was invited to speak at the Child Neurology conference in Victoria on metabolic disorders. The weather was perfect and I could also visit the famous "Butchart Gardens". Flowers of all kinds bloomed and it was the most gorgeous day.

I leave you with the beautiful picture of the Butchart Gardens and this quote.

With best wishes for a great summer!

Your friend
Chitra Prasad

Adopt the pace of nature: her secret is patience.
Ralph Waldo Emerson

Personal Stories



Shawn Johnson's Story

Shawn Alan Johnson was born on July 13, 2006, two weeks premature. My family and I were excited to have a healthy baby boy, as he was my first child and the first grandchild for my parents. I have always wanted to be a mother and was glad that the time had finally come. Shawn was a thriving little guy and quickly grew attached to everyone in my family. However, at 2 months, it became evident that something was not right with Shawn. He began twitching one evening while I was visiting a friend. We took him to the hospital and he was admitted right away. We live in Kitchener, but Shawn was transferred

to the London Health Sciences Centre as they had more resources to assess his condition and diagnose the problem. He was diagnosed with Cryptogenic Epilepsy and was sent home with seizure medications. At 4 months, Shawn began presenting signs of spasms and he was admitted to the hospital again. He was unable to hold his head up, and was “floppy” in appearance. He was again admitted to the hospital in London, and the doctors began to investigate the cause of his spasms. The doctors noticed the unusual texture of Shawn’s hair, which is twisted and coarse. I was told these characteristics are prevalent in children with Menkes Syndrome, and at 4 months old, it was confirmed that this was the cause of Shawn’s medical condition. As a result of the disorder, Shawn has severe developmental delay, which means he cannot speak, walk and is not able to hold up his head. However, last summer I enrolled him in a swimming class for babies, which has improved his strength quite a bit. He is injected with copper chloride twice daily (which I administer) and he has a g-tube for feeding. He is now 22 months old and weighs 25 pounds, but he looks like a happy and healthy child. He is referred to by his doctors as a “miracle baby” and he loves to play with his stuffed animals, loves the colour green, and points his index fingers when he is content. He also loves Elmo and loves listening to his CDs in his Car’s boom box. He eats pureed baby food, which I blend for him. His favourite is steamed fish and mashed potatoes. He loves chocolate as well, which I give him as a treat. He occasionally makes baby sounds when someone is speaking to him, and is always looking around at his surroundings, taking in everything there is to see. My family and I are hopeful that someday Shawn can say a few words or take a few steps.

Featured This Issue

Menkes Disease

By Dr. Chitra Prasad

About one in 200,000 males is born with a genetic disorder called Menkes disease. In 1962 John Menkes, MD, and his colleagues at Columbia University in New York published a scientific article about 5 male infants with a distinctive genetic syndrome. This syndrome, now known as Menkes kinky hair disease, has been identified as a disorder of copper metabolism in the body. Some parts of the body don't have enough copper (blood plasma, liver, and brain), while other parts of the body (kidney, spleen, skeletal muscle) accumulate too much. Its symptoms include significant developmental delay, and connective tissue (collagen) abnormalities that result in soft bones and cartilage and weakened artery walls.

Menkes disease is caused by a defective X-linked gene that is involved in the metabolism of copper in the body. The disease primarily affects male infants. Affected infants may be born prematurely, but appear healthy at birth and develop normally for 6 to 8 weeks. Then symptoms begin, including floppy muscle tone, seizures, and failure to thrive. Menkes disease is also characterized by low body temperature and strikingly peculiar hair, which is kinky, colorless or steel-colored, and breaks easily. There is often extensive involvement of the gray matter of the brain. Arteries in the brain may also be twisted. This can lead to rupture or blockage of the arteries. Weakened bones (osteoporosis) may result in fractures. Bladder diverticulae can also be seen in some patients with Menkes Disease.

Pathogenesis:

Copper is a trace metal in many essential enzyme systems, including cytochrome C oxidase, superoxide dismutase, lysyl oxidase, tyrosinase, ascorbic acid oxidase, ceruloplasmin, and dopamine beta hydroxylase. The deficiency or impaired function of these enzyme systems is thought to be responsible for the clinical findings of Menkes disease. The Menkes gene is located on the long arm of the X chromosome at Xq13.3, and the gene product (ATP7A) is a 1500–amino acid P-type adenosine triphosphatase (ATPase) that has 17 domains—6 copper binding, 8 transmembrane, a phosphatase, a phosphorylation, and an ATP binding.

Diagnosis is by:

- ◆ Copper and ceruloplasmin levels in blood, after the child is 6 weeks old (not diagnostic before then)
- ◆ Microscopic examination of hair will show Menkes abnormalities (Pili torti - 180° twisting of the hair).
- ◆ Skin biopsy can test copper metabolism
- ◆ DNA studies

The Inherited Metabolic Disorders News

Treatment::

With daily copper injections, the outcome may improve in Menkes disease if it is begun within days after birth. Other treatment is symptomatic and supportive and includes management of nutrition and seizures. Physiotherapy, occupational therapy and a multisystem developmental team is essential to monitor the ongoing developmental needs.

The newborn screening for this disorder is not available, and early detection is infrequent as the clinical signs of Menkes disease are subtle in the beginning.

Recent research sponsored by the NINDS can accurately diagnose the presence of Menkes disease before symptoms appear. Study results showed higher survival rates for children given the earliest copper injection treatment and improved, if not normal, neurodevelopment.

References:

Kaler SG, et al. Neonatal Diagnosis and Treatment of Menkes Disease.

N Engl J Med 2008;358:605-14.

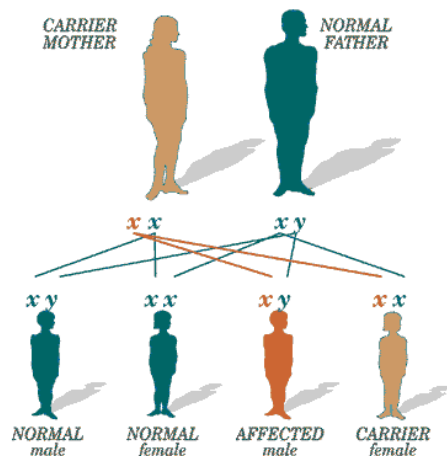
<http://www.ninds.nih.gov/disorders/menkes/menkes.htm>

<http://rarediseases.about.com/cs/menkesdisease/a/022704.htm>

<http://www.emedicine.com/neuro/topic569.htm>

http://genome.wellcome.ac.uk/doc_wtd020851.html

X-Linked Inheritance:



Examples of other X-linked recessive diseases include red-green color blindness, haemophilia and the Duchenne and Becker forms of muscular dystrophy. These diseases are much more common in males than females because two copies of the mutant gene are required for the disease to occur in females, while only one copy is required in males. The inheritance pattern of an X-linked recessive disease has the following characteristics:

The vast majority of affected individuals are male. Affected males never pass the disease to their sons because there is no male-to-male transmission of the X chromosome. Affected males pass the defective X

chromosome to all of their daughters, who are described as obligate carriers. This means they carry the disease-causing allele but generally show no disease symptoms since a functional copy of the gene is present on the other chromosome.

Female carriers pass the defective X chromosome to half their sons (who are affected by the disease) and half their daughters (who are therefore also carriers). The other children inherit the normal copy of the chromosome.

Kids Korner



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

Child's Name	Metabolic Disorder	Birthday	Age
Daniel	PKU	April 25, 2003	5
Anabella	PKU	September 29, 2007	11
Zacharia	PKU	June 6, 2004	4
Hailey	PKU	August 19, 2003	5
Julia	PKU	July 29, 2001	7
Abilene	PKU	June 12, 2005	3
Stathie	PKU	May 7, 1992	16
Dusan	PKU	July 21, 2003	5
Carter	PKU	September 5, 2001	7
Elijah	PKU	June 18, 2003	5
Samantha	PKU	July 31, 2000	8
Pramothan	6PTPS deficiency	August 7, 2007	1
Cameron	Alpha-1 Antitrypsin Deficiency	July 10, 2004	4
Alexis	Biotinidase Deficiency	June 21, 2007	1
Olivia	Biotinidase Deficiency	May 11, 2007	1
Rebecca	Biotinidase Deficiency	August 23, 2007	1
Susanna	Carnitine Uptake Disorder	August 20, 2007	1
Cameron	Ethylmalonic Aciduria	April 26, 1995	13
Ema	Galactosemia	August 9, 2005	3
Alexander	Galactosemia	May 8, 2001	7
Joan	Glycogen Storage Disease	June 20, 2003	5
Michelle	HMG CoA Lyase Deficiency	September 1, 1999	9
Emma Lynn	Hurler	September 9, 2004	4
Austin	MCADD	July 17, 1996	12
Myah	MCADD	April 4, 2007	1
Stanley	MCADD	August 26, 2003	5
Stephanie	MCADD	June 16, 2006	2
Shaylin	MCADD	July 13, 2006	2
Kelly	MELAS	July 10, 1995	13
Shawn	Menkes	July 13, 2006	2
Thomas	3MCC	July 26, 2007	1
Natasha	3-Methylglutaric aciduria	August 19, 2004	4
Chelsey	Mitochondrial Complex 1 Deficiency	August 4, 1994	14
Mackenzie	3-Methylcrotonyl-CoA Carboxylase Deficiency	June 10, 2007	1

The Inherited Metabolic Disorders News

April, May, June, July, August, September Birthdays Continued...

Aaron	ML4	Aug 30, 1993	15
Jordynn	5-MTHFR	May 30, 2000	8
Jared	OTC	September 22, 1993	15
Jack	OTC	August 8, 2005	3
Marlo	Pyruvate Dehydrogenase Deficiency	August 15, 2005	3
Hamza	Sanfilippo	April 3, 2005	3
Ethan	VLCADD	June 13, 2007	1
Paisley	Metabolic Disorder	September 24, 1999	9
Zachary	Metabolic Disorder	September 4, 2005	3
Wyatt	Metabolic Disorder	July 18, 2002	6
Alexa	Metabolic Disorder	September 29, 1993	15
Jeff	Metabolic Disorder	June 24, 2001	7
Madison	Metabolic Disorder	June 23, 2000	8
Nicholas	Metabolic Disorder	September 1, 2002	6
Ali	Metabolic Disorder	September 13, 2000	8
Mariam	Metabolic Disorder	July 10, 2003	5

PKU Phenylketonuria

6PTPS 6-Pyruvoyltetrahydropterin Synthase Deficiency

MCADD Medium Chain Acyl-CoA Dehydrogenase Deficiency

3MCC 3-Methylcrotonyl-CoA Carboxylase Deficiency

MELAS Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, Stroke

5-MTHFR Methylenetetrahydrofolate Reductase Deficiency

OTC Ornithine Transcarbamylase Deficiency

ML4 Mucopolipidosis Type IV

VLCADD Very Long Chain Acyl-CoA Dehydrogenase Deficiency

Happy Birthday Everyone!

Tricks of the Trade

Tyler Allen has another suggestion -

Mix until smooth:

3 Tablespoons DS Peanut Butter Flavoured Spread + 1 Tablespoon Regular Honey + 1 whole banana
This tastes great on bread and crackers. Enjoy.

Jenn Culp (Mom of Daniel) says -

Keep trying new foods on a regular basis. The first time your child tries something they may not like it but after a few times they may enjoy it. We always have to try new things and if they don't like it that's okay but they have to try it. This goes for all children whether they have PKU or not. (Example: Daniel now loves the new Brooklyn Dogs from Cambrooke but the first 2 times he tried them he didn't like them)

Get your child involved in their diet early on - it will make it easier for everyone. Daniel (age 5) understands the basic diet, how to turn on his scale and weigh his 'milk'. He also knows how to mix it in the pitcher and with help can pour it into his sippy cups.

If you don't like baking try to get your children to help out. It makes for lots of fun in the kitchen and time spent together. It's also great for counting and math skills. Daniel loves making his bread for his friends - he takes the dough and shapes it into pretzels and other shapes. He's made this for his class several times this year and really enjoys it. His sister gets to help out and we all get a snack when they are done.

Suzanne's Corner



Ravioli with Toasted Bread Crumbs *****REVISION*****

The following recipe from last year's low protein cooking demonstration contained incorrect nutrition facts. The correct information is below.

4 CBF Raviolo or Pierogi
1 Tbsp butter
1/4 cup low protein bread crumbs (recipe follows using CBF HomeStyle Slice White Bread or CBF Artisan Bread– see below)

1. Cook Ravioli or Perogi in boiling water for no more than 3 minutes and drain.
2. Transfer to pan and fry in butter for about 2 minutes
3. Add bread crumbs and toss well. Fry until golden brown and crispy, about 3 minutes. Season with salt and pepper.

Serve with warm marinara sauce to dip in!

Bread Crumbs

1 slice of CBF HomeStyle Sliced White Bread or 1/3 of an Artisan Bread roll.

Directions:

1. Break the bread into pieces, drop into the food processor, and grind until fine. Add dried herbs and/or salt and pepper if desired.
2. In a baking dish, place the bread crumbs in a 200 degree F oven for about 15-20 minutes, tossing occasionally until lightly browned and completely dry.

Recipe makes 1 serving.

Nutrition facts per serving (4 pieces)

PHE: 31 mg
LEY: 47 mg
Pro: 0.63 g
Calories: 317



Resources

New Canadian Galactsemia Support Group

Canadian Families with Galactosemia: www.canadianfamilieswithgalactosemia.com

Low Protein Cooking Websites

Taste Connections: www.tasteconnections.com

LowProtein.com: www.lowprotein.com

What's New

International PKU Teen and Adult Conference August 15 and 16, 2008

Embassy Suites Chicago O'Hare/Rosemont
5500 North River Road
Rosemont, IL 60018 USA

For further information, visit www.pkuworldlink.org

Metabolic Family Workshop May 30, 2008

The 5th annual Metabolic Family Workshop at the Lamplighter Inn was another great success. Approximately 154 family members and professionals from South-western Ontario & the Toronto area participated. The first speaker was Barbara Marriage, Dietitian, who explained nutrition for inherited metabolic diseases. The attendees then split into 4 groups: *Ask the Expert* (a panel of medical professionals), *Lysosomal Storage Disorders*, *Financial Security for the Child with Disabilities* and *For Teenagers/ Young Adults Only*. When the groups rejoined, they listened to the keynote speaker, Dr Stephen Lafay, motivational speaker, give a talk entitled "Transforming Stressings into Blessings" .

The overall feedback has been very positive with families providing ideas for future events. The professionals and vendors that attended were thoroughly impressed by the organization and family attendance.

Low Protein Cooking Demonstration May 31, 2008

The 4th annual Low Protein Cooking Demonstration was once again very well attended and enjoyed by all. Malathy Ramanujam demonstrated some interesting recipes, and shared her first-hand knowledge of low protein cooking.



LHSC Metabolic Team

Newborn screening updates:

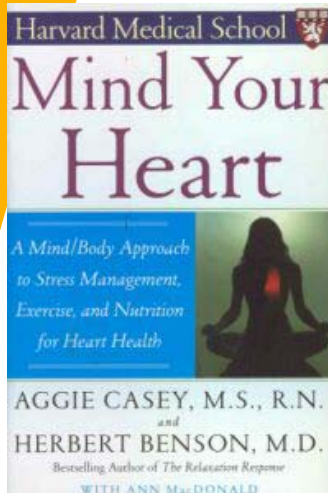
In October 2007 the Children's Hospital of Eastern Ontario (CHEO) held an educational meeting for all health care professionals involved with newborn screening. Currently there are 5 centres within Ontario involved with the Ontario Newborn Screening Program. The objective of the meeting was to learn how positive screens are handled at various centers across the province to learn from each other and identify common challenges. It is hoped that within the next year we will learn better ways to work with new parents/families dealing with positive newborn screens to enhance patient satisfaction and patient care.

Sincerely,
Jennifer A. MacWhirter, MS
Genetic Counsellor

Welcome to Stephanie Ho, Genetic Counsellor, who is covering Jennifer's position and Wendy Curren, who has taken over administrative support for newborn screening.

Addition of Tests: On April 6, 2008, Cystic Fibrosis screening began in Ontario.

Mind Your Heart



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

- ◆ Don't overlook emotional resources who are close at hand: spouse, friends, family and co-workers.
- ◆ Don't be afraid to ask questions or to ask for help.
- ◆ Allow an extra fifteen minutes to get to work or appointments.
- ◆ Check your breathing throughout the day. Practice diaphragmatic breathing when you find yourself shifting into upper chest breathing.

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!

Picture drawn by Shawn
(Menkes), age 21 months



Contact Information

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Post Message: metabolic_disorders@yahoogroups.com

Parent Support Contact: Jennifer Culp

Tel: 1.519.632.9924

Email: donjen2000@hotmail.com