The Inherited Metabolic Disorders News

Children's Hospital
London Health Sciences Centre

Summer 2014 Volume 11 Issue 2

In this Issue

From the Editor

Greetings everyone!

After such a long and cold winter, the warm weather has finally arrived!

We have been busy planning the Metabolic Family Workshop and Low Protein Cooking Demonstration for 2015.

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.

Janice Little

From Dr Chitra Prasad

Dear Friends,

Greetings!

Summer is finally here after a very long winter. I hope you are enjoying the greenery and the beauty of the flowers. In this current issue please find another story of courage and resilience. I know over the years I have introduced remarkable families to you.

The Alsemgeests are also one such family. David and Kazumi have two children affected with a very severe neurodegenerative disorder called 'Batten disease'. On June 9th 2014, Kazumi received one of only four Home Healthcare Heart of Home Care awards for the care she provides to two of her three children suffering with Batten Disease. Both children require around the clock care and supervision for all their daily activities in addition to managing their feeding and medication for seizures. As always I remain in awe of all the patients and their families who radiate joy and happiness in spite of such difficult circumstances.

I am pleased to inform you that we will be going ahead with the 2015 metabolic family workshop on Friday May 8th. Please don't forget to check the details in this issue of the newsletter. Please also make sure you sign up your names and names of people attending with you and send to Janice little our resource associate. Hope to see you all there!

Since we are trying to go digital with our metabolic newsletters, kindly provide your current email address to Janice Little (Resource Associate) Janice.Little@lhsc.on.ca.

This will be very helpful. *continued .....*

Our metabolic clinic is growing. We now have another new geneticist Dr. Natalya Karp who will be helping out in the metabolic clinic apart from her other clinical duties. Nicholas Watkins is the new metabolic counsellor who is covering Melanie Napier's maternity leave. Kara Bigelow is the new metabolic secretary. We thank Joanne Psiuk-Rodgers who has been the metabolic secretary for many years for her support to the metabolic clinic. Some of you might recall her as Jo-Jo the clown in the earlier metabolic family workshops.

This year the Garrod meeting (national meeting of inborn errors of metabolism) and newborn screening meeting were held together in Ottawa. It was a very successful meeting with over 200 members participating. There were discussions about newer therapies and management of metabolic conditions.

I would like to leave you with this beautiful quote from Maya Angelou, poet, activist and writer.

"My mission in life is not merely to survive, but to thrive; and to do so with some passion, some compassion, some humor, and some style"

With best wishes

Your friend
Chitra Prasad

Personal Stories

Kazumi and David Alsemgeest and their son, Rey. Two of the couple's three children suffer from Batten Disease.

London mother given Home Care award

By Gerard Creces

Reprinted from thelondoner.ca with permission
Original print date: June 13, 2014

For Kazumi Alsemgeest, providing 'round the clock care to her children is just a part of parenting. However, on June 9, Kazumi received one of only four VHA Home Healthcare Heart of Home Care awards for the care she provides two of her three children suffering with Batten Disease.

Kazumi and her husband, David, have three children Rey, 8 Mei, 5 and Ken, 13. Their two youngest were both diagnosed with late infantile Batten Disease - a degenerative neurological disorder that is the most common form of Neuronal Ceroid Lipofuscinoses (NCL).

Children with Batten Disease eventually suffer from seizures, mental and physical impairments and loss of sight and motor skills. It is fatal.

On one side of the family's living room is a large dining room table dominated by a computer, a care schedule and binders of nursing reports. The other side is occupied by a hospital bed.

Personal Stories - continued

Rey is home from school today, so Kazumi is kept busy with different tasks as the interview is conducted.

David sits in front of the computer, bringing up photos of the family and the award ceremony. He was the one initially nominated for the award by a nurse at the Kids Country Club in London.

However, he suggested the nomination be transferred to Kazumi, who he noted is always 100 per cent focused on her children's care.

"If you could see how dedicated she is to the children," he said. "How at any given point in time she is thinking not only about the kids' immediate needs but calculating their future needs for the next hour or the next week." The entire family is involved in caring for Rey and Mei. The couple's oldest son Ken, is also very much a part of caring for his siblings. While Rey is immobile, Mei still has the ability to crawl and use a walker, and is able to be fed.

"People think of quality of life differently," Kazumi said. For her, it means taking Mei outside and seeing her smile, or seeing basic expression from Rey. Not only is the interaction good for the two children, but for Kazumi, David and Ken as well.

Though the award focuses on Kazumi's dedicated support, she is quick to point out the family doesn't do it alone. "There are a lot of people involved in Rey and Mei's care," Kazumi said. "We shouldn't forget everyone fighting with us."

Home caregivers are the heart and soul of the healthcare system in Ontario, though their work is seldom recognized outside their homes. Stoikopolous said from VHA's perspective, the Alsemgeest family all deserve recognition.

Still, as David puts it: "This story is as much a story about a family fighting against the odds to remain as much of a family as possible - thanks to Kazumi and her dedication to all."

Rei, Kamuzi, Mei

Kamuzi, Mei, David, Rey

Courage doesn't always roar. Sometimes courage is the quiet voice at the end of the day saying, "I will try again tomorrow".

Mary Anne Radmacher

Featured This Issue

Batten Disease

Prepared by Dr. Chitra Prasad

Batten disease is a fatal, inherited disorder of the nervous system that typically begins in childhood. Early symptoms of this disorder usually appear between the ages of 3 to 10 years, when parents or physicians may notice a previously normal child has begun to develop vision problems or seizures. In some cases the early signs are subtle, taking the form of personality and behavior changes, slow learning, clumsiness, or stumbling. Over time, affected children suffer mental impairment, worsening seizures, and progressive loss of sight and motor skills. Eventually, children with Batten disease become blind, bedridden, and lose awareness of their surroundings. Batten disease is often fatal by the late teens or twenties. It is the most common form of a group of disorders called the neuronal ceroid lipofuscinoses, or NCLs. There are many other forms of NCLs. Batten disease and other forms of NCL are relatively rare, occurring in an estimated 2 to 4 of every 100,000 live births. These disorders appear to be more common in Finland, Sweden, other parts of northern Europe, and Newfoundland, Canada.

Childhood NCLs are autosomal recessive disorders; that is, they occur only when a child inherits two copies of the defective gene, one from each parent. When both parents carry one copy of the changed gene, each of their children has a one in four chance of developing NCL. Symptoms of Batten disease and other NCLs are linked to a buildup of substances called lipofuscins (lipopigments) in the body's tissues. These lipopigments are made up of fats and proteins. The lipopigments build up in cells of the brain and the eye as well as in skin, muscle, and many other tissues. The substances are found inside a part of cells called lysosomes.

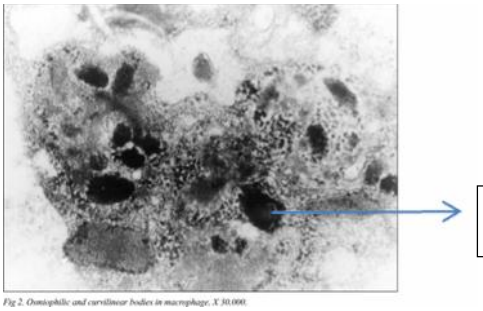
Because vision loss is often an early sign, Batten disease may be first suspected during an eye exam. An eye doctor can detect a loss of cells within the eye that occurs in the childhood forms of NCL. In order to diagnose NCL, the neurologist needs the individual's medical and family history and information from various laboratory tests. Diagnostic tests used for NCLs include: *Skin or tissue sampling*. The doctor can examine a small piece of tissue under an electron microscope. The powerful magnification of the microscope helps the doctor spot typical NCL deposits. These deposits are common in skin cells, especially those from sweat glands. *Measurement of enzyme activity*: Measurement of the activity of palmitoyl-protein thioesterase involved in CLN1, the acid protease involved in CLN2 in white blood cells or cultured skin fibroblasts (cells that strengthen skin and give it elasticity) can be used to confirm or rule out these diagnoses.

Featured This Issue - continued

DNA analysis: DNA analysis can be used to confirm the diagnosis or for the prenatal diagnosis of this form of Batten disease. As yet, no specific treatment is known that can halt or reverse the symptoms of Batten disease or other NCLs. However, seizures can sometimes be reduced or controlled with anticonvulsant drugs, and other medical problems can be treated appropriately as they arise. At the same time, physical and occupational therapy may help patients retain function as long as possible.

Support and encouragement can help patients and families cope with the profound disability and dementia caused by NCLs. Often, support groups enable affected children, adults, and families to share common concerns and experiences. Research is ongoing for this disorder in National Institute of Neurological Disorders and Stroke, a part of the National Institutes of Health USA. Meanwhile, scientists pursue medical research that could someday yield an effective treatment.

Modified from http://www.ninds.nih.gov/disorders/batten/detail_batten.htm



www.scielo.br/scielo.php?pid=S0004-282X2000000400001&script=sci_arttext

Going Paperless !!!

In an effort to streamline newsletter production and help the environment, we are going to email future newsletters to as many people as possible. Please help us by sending an email to **janice.little@lhsc.on.ca** using the title “Newsletter” indicating that you would like to receive the newsletter by email.

Suzanne's Corner

Lo-Pro Chicken Nuggets

Makes 4 servings (4 nuggets per serving)

2 cups	Carrots, cooked and pureed
2 Tablespoons	Potato flakes
1 Tablespoons	Egg replacer
1 Tablespoons	Onion flakes, dry
½ cup	Wheat starch (can use Loprofin Mix)
½ teaspoon	Salt
½ teaspoon	Seasoned salt
¼ teaspoon	Pepper
1 Tablespoon	Worcestershire sauce
2 Tablespoons	Water
½ cup	Low protein bread crumbs
2 Tablespoons	Canola oil

In large mixing bowl, add carrots, potatoes flakes, egg replacer, onion flakes, wheat starch and dry seasonings. Stir together.
Add Worcestershire sauce and water 1 tablespoon at a time as needed, until mixture sticks together.
Refrigerate for several hours or overnight until mixture is stiff.
Form mixture into 16 nuggets and coat with bread crumbs.
Fry in canola oil and serve warm.

Convenience tip: Nuggets can be stored frozen in airtight container up to 4 months. To cook nuggets remove from freezer and bake in preheated oven 5 minutes at 300°F, microwave 30 seconds, or pan-fry in skillet with 1 teaspoon oil.

Nutrition Information	Calories	Protein	Phenylalanine
Per serving (4 nuggets)	237	1.1 grams	61 mg

What's New

For more information or to donate, please visit :

www.canpku.org/pku-news/209-averys-ride-4-pku-new

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Welcome to Dr. Natalya Karp !

I am thrilled to join the genetics-metabolics team at LHSC! Several words about myself. I obtained my Medical Doctor degree from Kazakh State Medical Academy in the city of Astana, Kazakhstan (in the former Soviet Union). I then immigrated to Canada and completed my Master of Science degree in Human Genetics at McGill University in 2004. After successfully passing the Medical Council of Canada Licensing Examinations, I was accepted to a residency program in Medical Genetics at the University of Toronto. I successfully competed the residency program and obtained my Royal College Certification in Medical Genetics in June of 2014.

What's New - continued

Congratulations to Melanie & Mike

As many of you know, I am currently on maternity leave. My husband and I are pleased to introduce you to our son, Jack, who was born in May. I am learning that being a mom brings a lot of joy as well as sleepless nights! It feels like time is flying by - he is growing so fast ! Before I know it, I'll be back at work. Looking forward to seeing you again in 2015 and hope you are all well

Welcome to Nicholas Watkins

Nicholas is a graduate from the University of Toronto MSc program in Genetic Counselling, class of 2014. Prior to his enrollment in the genetic counselling program Nicholas completed a MSc in Molecular Genetics at the University of Toronto and a BSc in Genetics at Western University. In a ddition to genetics Nicholas enjoys cycling, soccer and superheroes, not necessarily in that order.

Welcome Back Kara

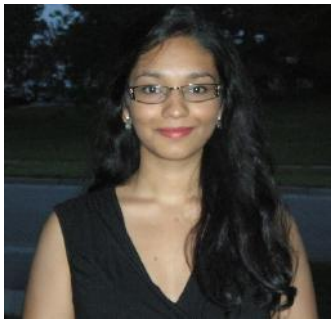
Some of you might already know Kara, who worked in pediatric neurology for the past 3 years. Prior to that, she worked in prenatal genetics. The team is very excited to have Kara back as the new administrative support member for Dr. Prasad and metabolics.

In her spare time, Kara enjoys spending time with her horse Ebony.

University Student Researcher

Hello!

I am a 4th year undergraduate student at Western University, studying genetics and computer science. This summer I've had a wonderful opportunity to work with Dr. Prasad and numerous members of the Medical Genetics department. Our study looked at the clinical, enzymatic, and molecular aspects of biotinidase deficiency amongst newborns in Ontario.



What's New - continued

Biotinidase deficiency is an autosomal recessive disorder, which disrupts several essential metabolic pathways resulting in a wide range of clinical presentations. Early detection and life-long treatment of biotin can allow children to remain asymptomatic. Because of this, it has being screened through newborn screening.

As a student, most of my learning is through classes and books. This is why I truly cherish this summer's experience with this project. My involvement has really educated me with a wealth of knowledge, work experience with professionals, and an appreciation for everything families and patients undergo.

Thank you,
Srinitya Gannavarapu

Conferences, Invited Lectures & Publications

February 2014 Melanie Napier and Dr. Rupar attended the World Lysosomal Conference San Diego California.

April 24 - 26, 2014 Suzanne Ratko attended Genetic Metabolic Dietitian's International - Broadening Horizons in Clinical Practice, Grapevine TX. New practice guidelines for management of various metabolic disorders and the latest research were presented during lectures and round tables.

April 2014. Dr. Rupar attended the Canadian Advisory Board on Niemann-Pick Disease Type C

May 2014. Dr. Rupar, Dr. Chitra Prasad and Jennifer DiRaimo (genetic counsellor) attended the Garrod and newborn screening meeting in Ottawa Canada.

May 2014 Dr. Chitra Prasad and Dr. Rupar attended the lysosomal diseases group meeting in Quebec City Canada.

May 2014. Suzanne Ratko and Dr. Chitra Prasad gave a lecture to the genetics journal club on The Role of Dietary Therapy and Dietitians in IEM (Inborn Errors of Metabolism).

May 2014. Trakadis YJ, Alfares A, Bodamer OA, Buyukavci M, Christodoulou J, Connor P, Glamuzina E, Gonzalez-Fernandez F, Bibi H, Echenne B, Manoli I, Mitchell J, Nordwall M, Prasad C, Scaglia F, Schiff M, Schrewe B, Touati G, Tchan MC, Varet B, Venditti CP, Zafeiriou D, Rupar CA, Rosenblatt DS, Watkins D, Braverman N. J Inherit Metab Dis. 2014 May;37(3):461-73.Update on transcobalamin deficiency: clinical presentation, treatment and outcome.

June 2014. Nicholas Watkins (genetic counsellor) attended the Candian Fabry Disease Initiative meeting in Vancouver Canada.J

June 6-7, 2014 Suzanne Ratko attended Annual Multidisciplinary European Phenylketonuria Symposium "PKU: a lifetime of challenges" This symposium reviewed the most important research achievements in the field of PKU and provided a unique opportunity to share knowledge and best practice in the clinical management of PKU.

July 2014. Dr. Rupar has been invited as a participant and speaker in meetings on research in metachromatic leukodystrophy in Pittsburg.

Prasad M, Narayan B, Prasad AN, Rupar CA, Levin S, Kronick J, Ramsay D, Tay KY and Prasad C. 2014. MELAS: A multigenerational impact of the MTTL1 A3243G MELAS mutation. Can J Neurol Sc 41: 210-219

What's New - continued

Presentations

June 2014. Ferrand A, Napier MP, Rupar CA, Al-Dirbashi OY, Chakraborty P, Siu VM, Prasad C. Biochemical and Hematologic Manifestations of Gastric Intrinsic Factor (GIF) Deficiency: Three cases in the Mennonite Population of Southwestern Ontario. Paediatric Research day London ,Canada; 2014.

May 2014. Beth K Potter, Pranesh Chakraborty, Doug Coyle, Jonathan B Kronick, Kumanan Wilson, Marni Brownell, Alicia Chan, Linda Dodds, Sarah Dyack, Annette Feigenbaum, Deshayne Fell, Michael Geraghty, Jane Gillis, Cheryl Rockman-Greenberg,Astrid Guttmann, Monica Hernandez, Maria Karaceper, Aneal Khan, Sara D Khangura, Anne-Marie Laberge, Julian Little, Jennifer MacKenzie, Bruno Maranda, Aizeddin Mhanni, Fiona A Miller, John J Mitchell, Grant Mitchell, Meranda Nakhla,Murray Potter, Chitra Prasad, Komudi Siriwardena, Rebecca Sparkes, Kathy N Speechley, Sylvia Stockler, Kylie Tingley, Yannis Trakadis, Lesley Turner, Hilary Vallance, Clara Van Karnebeek, Brenda J Wilson, Nataliya Yuskiv,Building a pan-Canadian practice-based research network for inherited metabolic diseases:the first two years of the Canadian Inherited Metabolic Diseases Research Network (CIMDRN) on behalf of the Canadian Inherited Metabolic Diseases Research Network presented at Garrod meeting.

May 2014. Rupar CA, Khan A, Au BC, Tailor C, Rothe M, Bischof D, Sirrs S, Auray-Blais C, Prokopishyn N, O'Hoski P, Huang J, Paul G, Benabid R, Viswanathan S, Morel C, Raiman J, Schambach A, West M, Keating A, Cornetta K, Foley R, and Medin JA . Towards a Clinical Trial of Lentivirus-Mediated Gene Therapy for Fabry Disease: In Vitro Patient Cell Mobilization and Transduction Outcomes. 2014. Garrod Association meeting

May 2014. Sara D Khangura, Pranesh Chakraborty, Doug Coyle, Jonathan Kronick, Anne-Marie Laberge, Julian Little, John J Mitchelle, Chitra Prasad, Komudi Siriwardena, Rebecca Sparkes, Kathy Speechley, Sylvia Stockler Kylie Tingley, Yannis Trakadis, Brenda Wilson, Kumanan Wilson, Beth K Potter on behalf of the Canadian Inherited Metabolic Diseases Research Network (CIMDRN) Patient and Family Experiences with Inborn Errors of Metabolism: perspectives of support & advocacy groups presented at Garrod meeting

June 2014. Sharan Goobie, Paul Atkison, Paul Gibson and Chitra Prasad. Severe thrombocytopenia in CDG type 1a. Presented at European Human society of Genetics in Milan.

May 2014. McAllister R, Liu J, Barr SD, CA Rupar. The analysis of therapeutic lentiviral vector integrations sites in CNS ependymal cells after delivery to the lateral ventricular in the murine model of metachromatic leukodystrophy. American Society of Gene and Cell Therapy meeting

May 2014 Khan A, Au BC, Tailor C, Sirrs S, Auray-Blais C, Rupar T, Prokopishyn N, O'Hoski P, Huang J, Benabid R, Viswanathan S, Morel C, Raiman J, West M, Keating A, Cornetta K, Foley R, and Medin JA. Pre-Clinical Patient Cell Mobilizations and Transduction Outcomes in Preparation for a Clinical Trial of Lentivirus-Mediated Gene Therapy for Fabry Disease. American Society of Gene and Cell Therapy meeting.

Student project

May 2014. Srinitya Gannavarapu, Chitra Prasad, Jennifer DiRaimo, Melanie Napier, Sharan Goobie, Murray Potter, Pranesh Chakraborty, Michael Geraghty, Maria Karaceper, Andreas Schulze, Tatiana Munoz, Jennifer MacKenzie, Lihua Li and Charles. A Rupar· Biotinidase deficiency: Spectrum of Molecular, Enzymatic and Clinical Information from Newborn Screening Ontario, Canada.

Children's Hospital
London Health Sciences Centre

Metabolic Family Workshop

Friday, May 8, 2015

Best Western Lamplighter

591 Wellington Road London, ON

Low Protein Cooking Demonstration

Saturday, May 9, 2015

Real Canadian Superstore

825 Oxford Street E London, ON

For further information, contact Janice Little at 519-685-8453

Metabolic Family Workshop
Friday, May 8, 2015

Number Attending: _____ Names: _____

_____

Contact Name: _____

Phone Number: _____

Morning Workshop Attending: (indicate names)

Diet _____ Other Metabolic Disorders _____ Lysosomal _____

Afternoon Workshop Attending: (indicate names)

Youth (8-13) _____ Adolescents (14-20) _____ Adults (with metabolic disorder) _____ Parents _____

Low Protein Cooking Demonstration
Saturday, May 9, 2015
Loblaw on Wonderland

_____ _____

Please return above information to:

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Children's Hospital
London Health Sciences Centre

Things to Remember

- Find and notice something beautiful every day
- Keep a gratitude journal
- Lay in the grass and watch the clouds
- Call friends you've been meaning to call
- Schedule a day of "no schedule"

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, do so on the **The Children's Health Foundation website** www.childhealth.ca

Ways to Give/ Under Giving Options: donate now/ Select: Make a Donation or Join Caring Heart Monthly Giving /Follow the prompts and it will give an online form with a comment box that you can type in and instruct the funds go to the *Inherited Metabolic Disorders program.*

If you would like to donate by phone with your credit card, please call 519.432.8564 or toll-free at 1.888.834-2496, Monday to Friday, 9 am to 5 pm.

Your donation is tax deductible, and an income tax receipt will be mailed to you.
Thank you!

Shaylin
Age 8
MCADD

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