The Inherited Metabolic Disorders News

Children's Hospital
London Health Sciences Centre

Winter 2014 Volume 11 Issue 1

In this Issue

From the Editor

Welcome to the latest issue of The *Inherited Metabolic Disorders News*! Spring is just around the corner (we hope!) and our Low Protein Cooking Demonstration plans are now underway. This year it will be held on **Saturday May 10th**.

As always, your comments, stories and suggestions are welcome!

Janice Little

From Dr Chitra Prasad

Dear Friends,

Greetings!

As always it is a pleasure to write to you all. Here in London and this part of Canada (like many others), we continue to experience a long winter. Of course it also means sitting and watching some of the winter Olympics. The athletes show their dedication and hard work to represent their country. I know that each and every one of you shows the same dedication and resilience when dealing with the challenges of everyday life. In spite of the health related issues, there are so many of you that bring smiles to others. Abdul, our 16 year old young patient with Pompe disease who has so many health related problems, still manages to play chess and give tough competition to many others.

In this issue you will also find the story of Laframboise family and how demanding their journey has been first to find the right diagnosis for their son Connor and then how to provide the best care for him. Even when the whole process has not been easy, both Michelle and Vince remain cheerful and loving.

This brings me to how can we remain resilient in life. There are many a time when we want to give up but then we don't. As I look around, I realize it's the love of our families and friends that keep us going when we are ready to stop and also our own resilience.

This year in 2014, our metabolic team decided to not have the metabolic family workshop. We are going ahead with the low protein workshop on May 10th 2014. We appreciate your suggestions about how we can further improve the metabolic family workshop. We will be having the metabolic family workshop in 2015. So keep tuned to our newsletters. 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.

From Dr Chitra Prasad - continued

Recently I watched a most inspiring film about Nelson Mandela “Invictus”. The title is based on a poem by William Ernest Henley. “I am the master of my fate: I am the captain of my soul”.

I am ending with a quote from Nelson Mandela himself, a man, a peaceful warrior who has taught the entire world the message of forgiveness and resilience.

“The greatest glory in living lies not in never falling, but in rising every time we fall.”
“Nelson Mandela”

With best wishes
Your friend
Chitra Prasad

Personal Stories

THE WINDSOR STAR

Michelle, left, and Vince Laframboise with their children Rebecca, 6, and Connor, 3, at their Amherstburg home on Tuesday, April 30, 2013. Connor suffers from a rare disease and the family is holding a fundraiser to help buy a specialized van.
(DAN JANISSE/The Windsor Star)

Amherstburg child lives with rare genetic disorder

Tues, April 30, 2013
Julie Kotsis

It took numerous doctors almost three years to figure out what was causing Connor Laframboise’s symptoms - visual impairment, severe respiratory infections, aspirating food and liquids.

At almost four years old, Connor is unable to walk or talk and now must be fed through a feeding tube.

A London team – Dr. Chitra Prasad, medical geneticist and director of the Metabolic Clinic at Children’s Hospital, and her husband, Dr. Narayan Prasad, a pediatric neurologist - finally diagnosed Connor with a rare genetic disorder called multiple sulfatase deficiency or Austin’s disease.

Only about 30 cases of MSD have been diagnosed worldwide over the last 60 years. It is a death sentence. “It doesn’t get better,” said Michelle Laframboise, Connor’s mom. “They can only guess (how long he’ll live) because every case is different.

Personal Stories - continued

It was last October when Michelle and her husband, Vince, were given the news. The doctors' rough estimate is that he'll live until 10 years old.

Michelle said that since MSD affects both the body and the brain, "it depends which one breaks down first" and his brain could degenerate or he may get a respiratory infection that he doesn't recover from.

"This is difficult," Vince said. "It's been a really tough process.

"I don't think it gets worse. It really takes the hope away." "It's like the worst thing you can have happen," Michelle said. "We're pretty used to crying all the time."

But Connor is like any happy, carefree three-year-old boy. He loves walks outside in his wheelchair, listening to music, his dog Toby and playing with his six-year-old sister Rebecca, who likes to read nursery rhymes to him.

He spends three days a week at the John McGivney Children's Centre where he participates in physiotheraphy, occupational and speech therapy, as well as crafts, outings and interactive play.

"He's incredibly happy. He smiles and laughs all the time," Michelle said. "He almost never cries … except when he's getting sick or something's changing.

"He gets so much from little things that he's always happy. It's easier for us … at least he's finding joy in things." It took almost 14 months of testing and perseverance by the Prasads to finally diagnose Connor with the degenerative disease.

"They really wanted to figure out what it was," Vince said. "All the diagnoses … the next one was worse than the last. "(MSD)'s so rare – there's no funding for stuff like this."

Michelle and Vince's mothers, Maureen Byrne and Marilyn Laframboise, came up with an idea a year ago to hold a fundraiser for the family. But Michelle said she and Vince were hesitant and wanted to wait until there was a diagnosis and a prognosis for Connor.

On Saturday, the Verdi Club in Amherstburg will be overflowing with supporters attending Connor's CareVan, a pasta dinner and silent auction. Connor's CareVan is a group of family members and friends who have taken on the challenge of raising funds to help provide for Connor's specialized needs.

Michelle said Countryside Chrysler Dodge Ltd. helped them buy a 2013 Grand Caravan.

"They've been just amazing to get us a brand new Caravan that we can send off to get converted," Michelle said.

The van will have a fold-out side entry platform to allow Connor's wheelchair to roll inside. Anchors will be installed to secure the chair.

Money raised at the sold-out fundraiser will be used for the conversion. The family also plans to build ramps at the front and back of their Amherstburg home as soon as the weather co-operates.

Anyone wishing to make a donation can send a cheque, made out to Connor's CareVan, to Marilyn Laframboise, 2690 Front Rd., LaSalle, or email Marilyn at connorscare-van@bell.net for further information.

Featured This Issue

Multiple Sulfatase Deficiency (MSD)

Multiple sulfatase deficiency is a disorder with many different presentations. There is a combined deficiency of various lysosomal enzymes causing features suggestive of metachromatic leukodystrophy, mucopolysaccharidoses, chondrodysplasia punctata amongst others thus making diagnosis challenging. (MSD) is an extremely rare lysosomal storage disease (only about 50 cases have been reported).

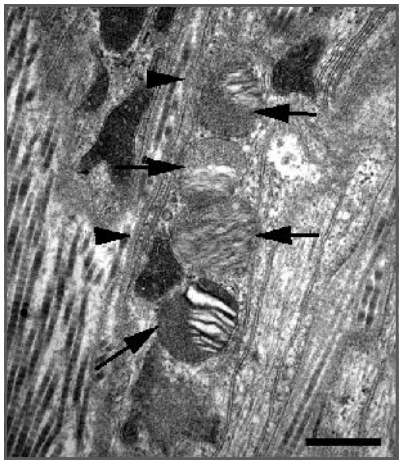
The most classically encountered form begins around the age of 1 or 2 years, but infantile cases appearing at birth have been described. Urine may typically show presence of mucopolysaccharides and sulfatides. Skin biopsy may show lipid inclusions. (see figure below) Results are confirmed by measuring the activity of several sulfatases. The gene SUMF1 is located on chromosome 3p26 and many mutations have been identified.

The disorder is inherited in an autosomal recessive manner. As with other autosomal recessive conditions there is a 1 in 4 risk of having another affected child.

Treatment remains supportive at present.

Reference: http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=585

Skin biopsy ultrastructural findings



The lysosomes (arrows) in the Schwann cell contain lipid with pleomorphic morphology.

Suzanne's Corner

A simple and fun recipe to make.
From **myspecialdiet.com.** Check out this site for other tasty treats.

Low Protein Crispy Marshmallow Treats

- 1 box LP Flakes
- 4 cups miniature marshmallows
  (or one 7 oz jar of marshmallow cream)
- 3 tbsp margarine (stick)

1) In large saucepan melt butter over low heat. Add marshmallows and stir until completely melted and smooth. Remove from heat.
2) Add the LP flakes and blend well.
3) Grease a 13 x 9 pan with oil or cooking spray. Use wax paper to press the mixture into the pan. Allow to cool, and then use a knife to cut into 2-inch squares.

If you prefer to make with a microwave:
In microwave-safe dish, heat butter and marshmallows on HIGH 1 minute intervals and stir. Continue until mixture is smooth. Usually, product will be smooth after about three 1 minute intervals but microwave cooking times will vary. Once your mixture is melted and smooth, follow steps 2 and 3 above.

Makes about 12 marshmallow treats.

Nutritional Information

	Calories	Carbohydrates	Sugar	Fat	Protein	Phenylalanine
Per treat	208	43.6 g	18.5 g	3.0 g	.4 g	10 mg

Resources

Many people are using the internet to connect with other individuals with the same condition or to learn more about the condition. Below is a list of known support groups that provide information and support for individuals and their families, as well as raise awareness and advocate for individuals with these conditions. Many of these groups host annual or bi-annual family conferences, so it is encouraged that you to take a look at the following websites and learn if there is a conference that you may wish to attend.

It is very difficult to keep track of all of the various support and advocacy groups, so please let us know if we have overlooked a support group!

Adrenoleukodystrophy (ALD) - ***ALD Support***: **adrenoleukodystrophysupport.org**
Biotinidase deficiency - ***Biotinidase Deficiency Family Support***: **biotinidasedeficiency.20m.com**
Fabry Disease - ***Canadian Fabry Association***: **www.fabrycanada.com**
Fatty Acid Oxidation Defects (MCAD, VLCAD, CPT2, etc) - ***FOD Family Support Group:*** **www.fodsupport.org**
Gaucher Disease - ***The National Gaucher Foundation of Canada***: **www.gauchercanada.ca**
Glycogen Storage Diseases (GSD) - ***Association for Glycogen Storage Disease:*** **www.agsdus.org**
Krabbe Disease - ***Hunter's Hope Foundation:*** **www.huntershope.org**
Leukodystrophies - ***United Leukodystrophy Foundation***: **www.ulf.org**
Maple Syrup Urine Disease (MSUD) - ***MSUD Family Support Group:*** **www.msud-support.org**
Metachromatic Leukodystrophy (MLD) - ***MLD Foundation:*** **mldfoundation.org** and ***Bethany's Hope:*** **www.bethanyshope.org**
Mitochondrial diseases (MELAS, MNGIE, etc) - ***Mito Canada***: **www.mitocanada.org**
Mucopolysaccharidoses (MPS) - ***MPS Society:*** **www.mpssociety.ca**
Niemann-Pick Disease - ***Canadian Chapter of the National Niemann-Pick Disease Foundation:*** **www.nnpdf.ca**
Organic Acidemias (MMA, Isovaleric Acidemia, etc) - ***Organic Acidemia Association:*** **www.oaanews.org**
Phenylketonuria (PKU) and Hyperphe - ***CanPKU:*** **www.canpku.org**
Pompe Disease - ***International Pompe Association***: **www.worldpompe.org**
Porphyria - ***American Porphyria Foundation:*** **ww.porphyriafoundation.com**
Urea Cycle Disorders (OTC Deficiency, ASA Lyase Deficiency, etc) - ***National Urea Cycle Disorders Foundation:*** **www.nucdf.org**

What's New

IMPORTANT NEWS ABOUT THE METABOLIC FAMILY WORKSHOP AND LOW PROTEIN COOKING DEMONSTRATION!!

As you may be aware, we will not host a Metabolic Family Workshop in 2014. We are taking time to reorganize this day to better meet the needs of our patients and families. We plan to host our new and improved Workshop in 2015.

By now, you may have received a survey in the mail requesting feedback on our Metabolic Family Workshop. If you have not filled it out yet, please consider doing so. Your feedback will provide us with useful information and help us to create a Workshop that you will hopefully find educational and enjoyable. You do not have to have attended a past Workshop to fill out this survey.

If you have filled out your survey and you/your child has a clinic appointment prior to April 30, 2014, you can bring your survey to clinic. Otherwise, please mail the survey back to us at the following address:

Medical Genetics Program
ATTN: Melanie Napier
800 Commissioners Road East
P.O. Box 5010
London, ON
N6A 5W9

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.

Thank you!

What's New - continued

Hello, my name is Kevin Dube and this is my sweet little niece Avery. She was born on August 24, 2013 and was diagnosed with PKU just a few days after she was born.

In June, I will embark on a journey across this great country. My niece is my inspiration for this ride, however, this is also a national issue and I hope my ride across Canada will:

Raise awareness regarding PKU;

Advocate so that better coverage for medical food, formula and other treatments may be available to all of those who have PKU, including Avery;

Raise money for Canadian PKU & Allied Disorders (www.canpku.org) to help them accomplish their goals; and

Promote a sense of community for Canadians living with this rare disorder.

7,382 km
Across Canada
Saturday, June 7 to Sunday, August 17, 2014
To learn more, please visit:
www.rideforpku.ca

Announcements

10th Annual Low Protein Cooking Demonstration

Saturday, May 10, 2014

Time: To Be Announced

***** New Venue *** Real Canadian Superstore**

825 Oxford Street East at Gammage Street
London, ON

Sponsored by:

NUTRICIA
Advanced Medical Nutrition

Children's Hospital
London Health Sciences Centre

For further information, contact Suzanne Ratko
519-685-8500 Extention 52469

Conferences, Invited Lectures &Publications

Conferences

Melanie Napier and Dr. Rupar are attending World Lysosomal Conference San Diego California February 2014

Dr. Chitra Prasad. Presented “Recent advances in newborn screening of Cystic fibrosis” at regional meeting of international society of newborn screening meeting in New Delhi September 2013

April 24 - 26, 2014 Genetic Metabolic Dietitian’s International – Broadening Horizons in Clinical Practice. (Suzanne Ratko)

June 6, 7, 2014 2014 Annual Multidisciplinary European Phenylketonuria Symposium Teaching (Suzanne Ratko)

Teaching

Suzanne Ratko: January, 2014 - Bresica University College Masters in Nutrition Program: Nutritional Management of Metabolic Disorders during Infancy and Childhood

Publications

1. P**rasad C**, Melançon SB, **Rupar CA**, **Prasad AN**, Nunez LD, Rosenblatt DS, Majewski J. Exome sequencing reveals a homozygous mutation in TWINKLE as the cause of multisystemic failure including renal tubulopathy in three siblings. Molecular genetics and metabolism, 2013 Mar 1; 108 (3): 190-4, DOI: 10.1016/j.ymgme.2012.12.007.

2. Kevelam SH, Bugiani M, Salomons GS, Feigenbaum A, Blaser S, **Prasad C**, Häberle J, Baric I, Bakker IM, Postma NL, Kanhai WA, Wolf NI, Abbink TE, Waisfisz Q, Heutink P, van der Knaap MS. Exome sequencing reveals mutated SLC19A3 in patients with an early-infantile, lethal encephalopathy. Brain: a journal of neurology, 2013 May 1; 136 (Pt 5): 1534-43, DOI: 10.1093/brain/awt054.

3. Cameron F, Xu J, Jung J, **Prasad C**. Array CGH Analysis and Developmental Delay: A Diagnostic Tool for Neurologists. The Canadian journal of neurological sciences. Le journal canadien des sciences neurologiques, 2013 Nov 1; 40 (6): 777-782.

4. Farhan SMK, Wang J, Robinson JF, Lahiry P, Siu VM, **Prasad C**, Kronick JB, Ramsay DA, Rupar CA, Hegele RA. Exome sequencing identifies NFS1 deficiency in a novel Fe-S cluster disease, infantile mitochondrial complex II/III deficiency. Mol Genet Genomic Med. 2014 Jan;2(1):73-80. doi: 10.1002/mgg3.46. Epub 2013 Nov 18.

"Our Kids"

Justin - Age 7
MPS 1

Jasper - Age 4
MPS VI

Accomplishments

Abdoul - Age 16
Pompe

Dear Dr. Prasad

This is Abdul. Everything is going well so far, after I have gotten out of the hospital. It was really rough in there. All my teachers and friends were really happy when they saw me back at school. I am in grade 10. My courses this year are anthropology, science, English, Canadian history, business, American history, math, civics, and careers. I have passed all my courses in grade 9. There is a chess club at school and I'm in it. I've been playing chess ever since I was 8 years old. I have went to 5 chess tournaments, 1 to the playoffs 2 times, 1 second place both times, and won 2 silver medals. I love school. When I graduate from high school, I plan on going to college and doing something with business or English.

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!***

Art of Extreme Care

- Buy yourself some flowers
- Play in the snow
- Sip a mug of hot, soothing tea
- Keep your daily to-do list to 3 items or less
- Take a long walk with your dog

Abeline
Age 8
PKU

Shaon
Age 12
MELAS

Contact Information

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Parent Support Contact: Jennifer Culp

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Email: jennc2011@hotmail.ca