

## In this Issue

- ◆ From the Editor...1
- ◆ From Dr. Chitra Prasad...1
- ◆ Personal Stories...2
- ◆ Featured This Issue ...3
- ◆ Kids Korner...5
- ◆ Suzanne's Corner...6
- ◆ Tricks of the Trade...8
- ◆ What's New...9
- ◆ Resources...10
- ◆ Mind Your Heart ...11
- ◆ How to Make a Donation...11
- ◆ Contact Information ...12



## From the Editor

Hello Everyone!

The Children's Hospital of Western Ontario has changed its name to "Children's Hospital" and our newsletter has adopted the new logo and colour scheme. We hope you like it.

Plans are well underway for the 5th annual Metabolic Family Day (Friday, May 30, 2007) and Low Protein Cooking Demonstration (Saturday, May 31, 2007).

Janice Little

## From Dr Chitra Prasad

Dear Friends,

A very happy new year to you and your families! This time of the year, we all try to make new resolutions. However, today I want to share with you something that I learned at a workshop which is "Two degrees Difference". This concept comes from a book by Dr. John Trent. "Two Degrees Difference" can be implemented easily by all of us. It reminds us to take small steps, to make progress inch by inch.

On another note, please read about the story of Stefan and his family in this issue. Stefan is a very bright and intelligent boy who has Morquio syndrome. He and his family have kindly shared their story with all of us.

Our metabolic team is very proud to see Suzanne's nomination for Dr Tim Frewen Humanitarian award. Congratulations Suzanne!

Newborn Screening has taken off in a big way in a number of developed countries. In the near future I will be going to India and presenting on the newborn screening.

We all at the metabolic team would love to hear from you, your stories and experiences, triumphs and challenges. Finally, I would like to end with a quote from a favorite author of mine, Dr Wayne W. Dyer.

*"When you change the way you look at things, the things you look at change."*

Have a wonderful new year!

With best wishes  
Your friend

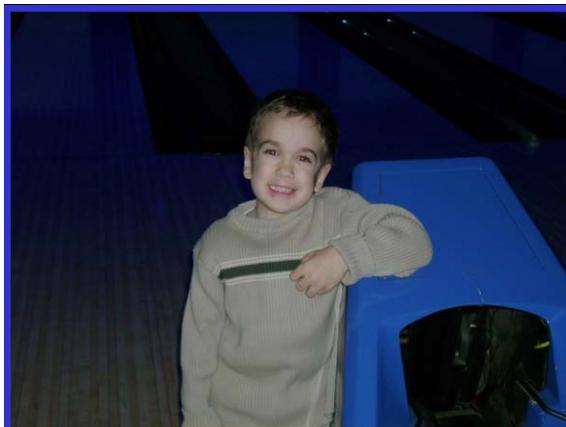
Chitra Prasad

## Personal Stories

by Verica Gacic

In January 1995, my husband, my son Boris, and myself arrived in Canada. We were fortunate to escape the civil war in the former Yugoslavia. We decided to settle in London and start a new life. We were so thrilled to be in Canada and when our second son Stefan was born in January of 2000 life seemed perfect. He was a healthy baby until the age of two. Around his second birthday, Stefan started to get sick more frequently and his growth slowed down. My husband and I were worried about our baby's declining health and embarked on an odyssey of hospital visits. In 2003, Stefan was diagnosed with Morquio Type A at the London Children's Hospital. At first we did not understand the significance of the diagnosis. We had never come into contact with Morquio before and the consequences of the diagnosis took a while to sink in. Once we understood what having Morquio meant for our baby's future, we were devastated. What kind of life would Stefan lead?

Stefan will be turning 8 in a few weeks. He is an energetic and rambunctious child in spite of the limitations that his condition imposes upon him. This summer, he had cervical fusion surgery to stabilize his neck and he was required to wear a halo for 3 months. A few weeks after the surgery and with the halo supporting his neck, he was trying to play soccer in the backyard! He is aware that he is different from his classmates at Kensal Park Public School but he does not allow that to affect his confidence or influence his quality of life. Due to his sunny disposition, Stefan has plenty of friends and is always involved in school activities. The students and staff at Kensal Park are tremendously supportive of Stefan and his needs. Like many boys his age, Stefan loves to watch baseball, soccer, and hockey. He is doing well in school and his teachers continuously praise his intelligence and wit. Although our life has changed in many ways since Stefan was diagnosed with Morquio, it has not been the negative experience we expected it to be. Thanks to the dedicated scientists who are working on enzyme replacement therapy to counteract the effects of Morquio, we believe that Stefan will have a bright and exciting future! We are grateful that we are in place where Stefan can get the care and support he needs to lead a fulfilled life.



## Featured This Issue

### Morquio Syndrome (MPS IV) By Chitra Prasad MD FRCPC

The mucopolysaccharidoses (MPS) are a group of inherited lysosomal storage disorders. Lysosomes function as the primary digestive units within cells. Enzymes within lysosomes break down or digest particular nutrients, such as certain carbohydrates and fats. In individuals with MPS disorders, deficiency or malfunction of specific lysosomal enzymes leads to an abnormal accumulation of certain complex carbohydrates (mucopolysaccharides or glycosaminoglycans) in the arteries, skeleton, eyes, joints, ears, skin, and/or teeth. These accumulations may also be found in the respiratory system, liver, spleen, central nervous system, blood, and bone marrow. This accumulation eventually causes progressive damage to cells, tissues, and various organ systems of the body. There are several different types and subtypes of mucopolysaccharidosis.

**MPS IV** is one of the subtypes of mucopolysaccharide disease which is also known as Morquio syndrome. Mucopolysaccharides are long chains of sugar molecules used in the building of connective tissues in the body.

"Saccharide" is a general term for a sugar molecule (think of saccharin). "Poly" means many". Muco" refers to the thick jelly-like consistency of the molecules. There is a continuous process in the body of replacing used materials and breaking them down for disposal. Children with Morquio syndrome are missing an enzyme which is essential in cutting up the mucopolysaccharides called keratan sulfate. The incompletely broken down mucopolysaccharides remain stored in cells in the body causing progressive damage. Babies may show little sign of the disease, but as more and more cells become damaged, symptoms start to appear.

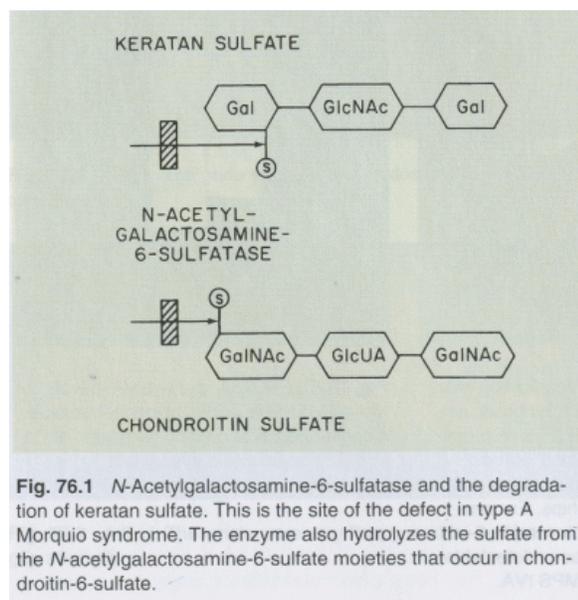
This disorder is inherited as an autosomal recessive trait thus there is a risk of 1 in 4 of having another affected child. MPS IV takes its name from Dr. Morquio, a pediatrician in Montevideo, Uruguay, who in 1929 described a family of four children affected by this condition. As the same condition was also described in the same year by Dr. Brailsford from Birmingham, England, it is sometimes known as Morquio-Brailsford syndrome. Morquio syndrome (mucopolysaccharidosis type IV; MPS IV) is a mucopolysaccharide storage disease that exists in two forms (Morquio syndromes A and B) and occurs because of a deficiency of the enzymes N-acetyl-galactosamine-6-sulfatase and beta-galactosidase, respectively. A deficiency of either enzyme leads to the accumulation of mucopolysaccharides in the body, abnormal skeletal development, and additional symptoms. In most cases, individuals with Morquio syndrome have normal intelligence.

## Featured This Issue

Morquio Syndrome (MPSIV) continued...

The clinical features of MPS IV-B are usually fewer and milder than those associated with MPS IV-A. Symptoms may include growth retardation, a prominent lower face, an abnormally short neck, knees that are abnormally close together (knock knees or genu valgum), flat feet, abnormal sideways and front-to-back or side-to-side curvature of the spine (kyphoscoliosis), abnormal development of the growing ends of the long

bones (epiphyses), and/or a prominent breast bone (pectus carinatum). Hearing loss, weakness of the legs, and/or additional abnormalities may also occur.



The care of the children and older patients affected with Morquio syndrome involves multidisciplinary approach with a number of sub-specialists (metabolic specialist, eye specialist, Orthopedic and cardiac specialists among others). In the past few years new progress has been made in the treatment of a number of lysosomal storage disorders such as for Gaucher disease, Fabry

disease, Pompe disease etc. Enzyme replacement therapy is also being tried in conjunction with other therapies for MPS1 (Hurler syndrome), MPSII, MPSVI.

These are truly exciting times as a number of scientists, physicians and other professionals are working together to find treatment for these complex disorders. Enzyme replacement therapy for Morquio syndrome will hopefully be available in the coming years.

References:

<http://www.bchealthguide.org/kbase/nord/nord299.htm>

[http://www.mpsociety.org/content/4040/MPS\\_IV/](http://www.mpsociety.org/content/4040/MPS_IV/)

Morquio syndrome chapter from Atlas of Metabolic Disease by William Nyhan and Pinar. T. Ozand

## Kids Korner



### Birthdays: January, February, March

Jesse	PKU	January 5, 2000	8
Abraham	PKU	March 31, 1993	15
Megan	PKU	March 31, 1998	10
William	PKU	February 26, 1997	11
Cody	PKU	March 26, 2006	2
Charis	PKU	January 26, 2005	3
Madeline	CDG	March 28, 1998	10
Hannah	GSD	February 21, 2006	2
Christine	GSD	January 31, 1997	11
Tyler	Homocystinuria	March 26, 1992	16
Elvina	MCADD	February 5, 2001	7
Andres	MCADD	February 26, 2007	1
Shawn	MCADD	January 2, 2002	6
Jenine	Mitochondrial Disorder	March 23, 1993	15
Lucas	Morquios	March 7, 1997	11
Zane	Morquios	February 12, 1999	9
Stefan	Morquios	January 15, 2000	8
Megan	Retts	February 7, 1994	14
Curtis	SLO	February 2, 1994	14
Edward	Transcobalamin II Deficiency	March 6, 2001	7
Ethan	Metabolic Disorder	January 1, 2002	6
Gideon	Metabolic Disorder	February 28, 2003	5

CDG Congenital Disorder of Glycosylation

PKU Phenylketonuria

MCADD Medium chain acyl-coA dehydrogenase deficiency

MELAS Mitochondrial myopathy, encephalopathy, lacticidosis, stroke

MTHFD Methylene tetrahydrofolate reductase deficiency

OTC Ornithine transcarbamylase deficiency

ML4 Mucopolipidosis type IV (4)

SLO Smith Lemli Opitz

**Happy Birthday Everyone!**

## Suzanne's Corner



For this newsletter, we are fortunate to have a recipe from Malathy Ramanujam from Taste Connections.



*TASTE CONNECTIONS MULTI-BAKING MIX*  
[www.tasteconnections.com](http://www.tasteconnections.com)

### *Taste Connections Tortillas*

#### Ingredients

<b>TC Multi-baking mix</b>	<b>2 ½ cups</b>
<b>Taco seasoning (optional)</b>	<b>½ tsp</b>
<b>Water</b>	<b>½ - ¾ cup (enough to make dough)</b>

#### Directions

1. Combine the baking mix and the seasoning in a bowl; add enough water to make a stiff dough.
  2. Knead for 3 - 5 minutes to be soft enough to roll into circles.
  3. Divide the dough into 12 equal parts.
  4. Using a rolling pin roll each ball into a thin tortilla (dusting baking mix so that the dough doesn't stick), approximately measuring 8 inches in diameter.
  5. Heat a griddle or a frying pan and cook one tortilla at a time. Place one rolled tortilla on the hot pan and cook for 30 seconds on one side.
  6. Turn it to the other side and cook on the other side until small bubbles appear on the surface.
  7. Repeat the cooking and turning processes until both the sides of the tortilla are covered with small bubbles that indicate the tortilla is cooked.
  8. Remove from the pan and spread the cooked tortilla with melted margarine or vegetable oil. Keep it covered in a cloth towel or a tortilla warmer.
- Cook all the tortillas similarly and keep them wrapped in a cloth towel. Makes 12 tortillas.

The tortilla can be cooked in an electric tortilla press or flatbread maker.

## NUTRIENT INFORMATION:

	KCAL	PROTgm	PHEmg	METmg	LEUmg
Per Recipe	1050	0.9	27.5	5.8	50
Per Tortilla	87.5	0.08	2.3	0.5	4.2



On Saturday, May 31, 2008 **VitaFlo** will be sponsoring our 4<sup>th</sup> Annual Low Protein Cooking Demonstration featuring Malathy at the Loblaws Wonderland.

## Malathy Ramanujam

Malathy Ramanujam graduated from University of Madras, India, in 1980, with a Bachelor of Science in Chemistry. She joined her husband in London, U.K., in 1983. In 1985, her son was diagnosed at the age of two with a rare metabolic condition called Homocystinuria. She moved to the United States in 1989.

The treatment of Homocystinuria also requires a special diet similar to PKU. Since there were not many options available for his diet she started experimenting with the limited resources to formulate various baking mixes and use them to prepare special meals for her son. Since 1994, she has been teaching families with similar metabolic condition how to prepare special meals. She graduated Magna Cum Laude from Bachelor of Arts (Liberal Studies major) from Cal State, Dominguez Hills in spring 1999. She created the website [www.lowprotein.com](http://www.lowprotein.com) in 2000 to share her recipes with everyone. Encouraged by Kathryn Moseley, dietitian at CHLA, and Dr. Toma (Professor, Food Science in CSULB) she pursued Master's degree in Nutritional Science in Fall 2000. She graduated (Fall, 2004) in the top 1 % of the graduating class (included in the Dean's List). She started her company, Taste Connections in 2003.

She has been presenting workshops on low protein cooking around the United States and Canada and still continues to do so, inspiring metabolic patients to stay on such life-saving diet.

## Tricks of the Trade

### Jennifer Culp says:

"Check out **President's Choice**® mini Chef's apple smooshies" This real fruit snack comes in a 60 mL tube format. Easy to carry and Daniel (cPKU) loves them!

Another great product to try may be found at your local **BULK BARN** store:

**Cassava Chips** - 28 g weight has 0.4 g protein & 20 mg phe. This is based on the information from *Low Protein Food List* by Virginia E Schuett (2<sup>nd</sup> ed, 2002), page 123 under Snacks (Chiffles).

### Please note regarding Cambrooke products:

**It is always important to re-check the labels of all purchased food items.**

Cambrooke Foods have recently reduced the phe and protein in some of their bread products as a result of changes to some of their recipes.

### **Good news - Product Update:**

Cambrooke Wraps will be available again from the Specialty Food Shop in the next few weeks.

### Unavailability of Aproten products in Canada:

From Kathryn Cormier RD @ Specialty Food Shop (SFS):

"The SFS Shop On-line will have up-to-date messages as Aproten item becomes out of stock. The message will read "**Temporarily unavailable due to labeling issues**". There will also be a message explaining the situation on the "What's New" -"Metabolic Information" section and on the front page of the Metabolic Shop On-line section in the next few days. This message will be updated as we have more information. Those who use the 1-800 line will also be informed with a brief message and the store staff will have information pertaining to this situation as well. Unfortunately, some of your patients are going to need to switch to a different pasta."

### From Virginia Schuett, Nutritionist and Editor, National PKU News:

I wanted to let you know that recently several parents have pointed out to me that data for a few items in the food list didn't look right to them. I have looked at those items and agreed. So I have now made corrections that are available on my website at [www.pkunews.org](http://www.pkunews.org) in the Diet Related Information section (Food List corrections) for hearts of palm, as well as pinenuts and several other of the nuts listed on page 184-185 of the food list. These are items that I assume most of you are not using, but I wanted to let you know just in case.

I really appreciate parent input on the food list. If you notice anything that looks odd or are aware that an ingredient has changed in an item to potentially change the protein/phe content please let me know! You are the ones using the list everyday and will be much more likely to notice discrepancies or odd numbers than I will be. There are so many numbers in the book that there are bound to be a few errors.

## What's New



**SUZANNE RATKO**  
Registered Dietitian, Children's Hospital

"I must have driven her crazy with all my questions, e-mails and concerns!"

**T**hroughout many letters of nomination, families and colleagues expressed their appreciation for Suzanne's guidance through a world of medical turmoil. Metabolic patients are extremely medically complex. The details of the daily nutrition and care plan can become overwhelming for families, but absolutely necessary for the growth and health of the child. More than a dietitian she has become a friend, supporter and confidant as families struggle to deal with the complexities

of special diets, continual monitoring and food sourcing. Suzanne is always there for those families. As a member of a multi-disciplinary team, she is well respected and in at least one instance, her knowledge of a new product led to a breakthrough in a child's care. From early morning phone calls, late night emails to 'just checking in' on her days off, she shows care and compassion each and every day.

*Nominated by  
Dr. Chitra Prasad and  
Dr. Tony Rupar*

**2007**

*Dr. Timothy C Frewen  
Humanitarian Award*

Congratulations to Suzanne Ratko who was nominated for the Dr. Timothy C Frewen Humanitarian Award.

The Dr. Timothy C. Frewen Humanitarian Award recognizes and honours medical health professionals, support staff and residents at London Health Sciences Centre, Children's Hospital and St. Joseph's Health Care London Neonatal Intensive Care Unit, who exemplify the qualities of outstanding compassion and humanitarianism in the provision of patient and family care.

## Meetings Update

### PEDICON 2008

Dr. Prasad will making a presentation on Newborn Screening at the PEDICON 2008 International/ National pediatric meeting in India late January.

Society for Inherited Metabolic Disorders (SIMD)    [www.simd.org](http://www.simd.org)

Suzanne Ratko will be attending this conference from March 2-5. It is being held in Pacific Grove, California.

**Metabolic Family Workshop**  
Bestwestern Lamplighter Inn  
London, Ontario  
Friday May 30, 2008

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**Low Protein Cooking Demonstration**  
Lowbaws on Wonderland  
London, Ontario  
Saturday May 31, 2008

## **Metabolic Family Workshop**

This year, the Metabolic Family Workshop begin a bit earlier in the morning. This will allow more time for family interaction. Registration will begin at 10 am and the event will wrap up at 430 pm.

We are looking at several exciting speakers. A final timetable will be included in the brochure, which will be mailed out sometime in March. We hope as many families as possible can attend.

There are hotel rooms set aside at the Lamplighter for a special rate. If you require a room Friday and /or Saturday night, please call the hotel to book your room at 519.681.7151 as soon as possible. Be sure to tell them you are with the Metabolic Family Workshop to get the special rate.

## **Resources**

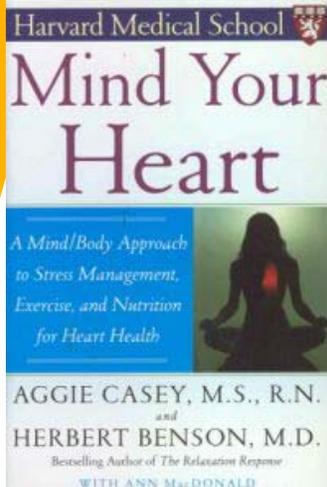
**The Canadian Society for Mucopolysaccharide & Related Diseases Inc.**  
[www.mpssociety.ca](http://www.mpssociety.ca)

**National MPS Society**  
[www.mpssociety.org](http://www.mpssociety.org)

**Taste Connections (Low Protein Foods)**  
[www.tasteconnections.com](http://www.tasteconnections.com)

**Low Protein.com (Low Protein Recipes)**  
[www.lowprotein.com](http://www.lowprotein.com)

## Mind Your Heart



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

- ◆ Adopt a pet.
- ◆ Don't watch the 11 PM news.
- ◆ Praise yourself regularly; give yourself a pat on the back for something you're proud of.
- ◆ Develop a wide variety of resources for gratification in your life, whether it's family, friends, hobbies, interests, special weekends or vacations. Treat yourself to "new and good things."
- ◆ Be assertive. Learn to express your needs and differences, to make requests, and to say "no" constructively.

## 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!**

*Action may not always bring happiness; but there is no happiness without action.*

**Benjamin Disraeli**

## Contact Information

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Website: [www.lhsc.on.ca/ programs/rmgc/met/metaboli.htm](http://www.lhsc.on.ca/programs/rmgc/met/metaboli.htm)

Yahoo Online Chat: [health.groups.yahoo.com/group/metabolic\\_disorders](http://health.groups.yahoo.com/group/metabolic_disorders)  
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**Jesse, 8 on January 5th, PKU**