The Inherited Metabolic Disorders News

Winter 2006 Volume 3 Issue 1

In this Issue

From the Editor

Spring is just around the corner and so is the 3rd annual Metabolic Family Workshop. This year it will be held on Friday May 5th at the Best Western Lamplighter Inn, just south of the hospital on Wellington Road followed by a low protein cooking demonstration on Saturday for those who wish to attend.

We are very excited about the upcoming workshop as there will be group specific talks for low protein diet, neurological issues and lysosomal storage disorders. Please mark your calendars and plan to attend!

Janice Little

From Dr. Chitra Prasad

Dear Friends,

Greetings! Hope you have had a good start to year 2006!
Every New Year brings new resolutions and new beginnings. If only we could keep up the promises we made to ourselves!

Our metabolic family continues to grow. We have a number of "family members" on our planning committee for our upcoming metabolic family day in May. The newborn screening (new technology) will be in place in a few months and we all are really looking forward to the new era of metabolic medicine in Ontario!

This year we plan to hold our family day in a different venue (please check out the details in the newsletter). I am very pleased that under the leadership of Suzanne Ratko (dietitian) we have been awarded grants to support this family day by Garrod association and Children's Hospital Foundation London. The families have been with us at every step to make all our projects successful. I thank each and every one of you for being a health advocate in your own unique way.

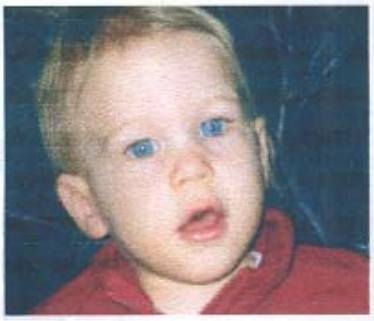
Lastly I am extremely grateful to all of our dedicated metabolic team members who work quietly behind the scenes but make all the difference!

I am closing with words of Etteine De Grelletwhich which are very inspiring!
" I expect to pass through this world but once; any good thing therefore that I can do, or any kindness that I can show to any fellow creature, let me do it now; let me not defer or neglect it, for I shall not pass this way again."

May you have a peaceful and safe year 2006!
With best wishes
Chitra Prasad

Personal Stories

NICKELS FOR NICHOLAS GOLF TOURNAMENT



On September 25, 2005 a golf tournament named “Nickels for Nicholas” was held at Maple Ridge Golf Course. It is a fund to help extra expenses for Nicholas’ needs and for Tay-Sachs research. Nicholas was diagnosed with Tay-Sachs disease in April 2005. Since then a lot of support and money raising has been done by friends, family and strangers in the community. A special thanks to Hi Tech Auto Care, Vipond Sprinkler Systems and the Valkeyrie Motorcycle Club of Southwestern Ontario for being strong contributors for the event. Thanks you also to both Dr. Prasads for attending and supporting the event. This is to be an annual event and we look forward to meeting more people next year.

Nick and Dana Walczak would like to thank everyone involved in Nicholas’s life, from all the doctors and nurses, family and friends and contributors for “Nickels for Nicholas”.

Nicholas is also involved in a trial drug study in Washington DC. It is a 6 month study using a drug called “Zavesca”. It may help slow his disease down and will give doctors and scientists more to go on for the future of finding a cure for Tay-Sachs disease. We hope the study will help other kids in the future born with this disease.

Written by Nick Walczak, father of Nicholas

Participants at the golf tournament

Nicholas and the medical staff in the Washington Hospital where Nicholas is getting treatment

Nicholas and his parents in front of the White House

Resources

- **Canadian Association for Tay-Sachs and Allied Diseases**
  www.catsad.ca

- **March of Dimes**
  www.marchofdimes.com

- **PKU Perspectives** (provides low protein food products for the dietary management of metabolic disorders and medical conditions requiring low protein diets)
  pkuperspectives.com

Kids Korner

January, February, March birthdays:

Child's Name	Metabolic Disorder	Birthday	Age
Jesse	PKU	January 5, 2000	6
Abrahem	PKU	March 31, 1993	13
Megan	PKU	March 31, 1998	8
William	PKU	February 26, 1997	9
Charis	PKU	January 26, 2005	1
Madeline	CDG	March 28, 1998	8
Kody	Gaucher's	January 3, 1991	15
Seoyoung	GSD	January 31, 1997	9
Tyler	Homocystinuria	March 26, 1992	14
Elvina	MCADD	February 5, 2001	5
Lucas	Morquios	March 7, 1997	9
Zane	Morquios	February 12, 1999	7
Stefan	Morquios	January 15, 2000	6
Megan	Rett Syndrome	February 7, 1994	12
Curtis	SLO	February 2, 1994	12
Nicholas	Tay-Sachs	February 3, 2004	2
Edward	Transcobalamin II Deficiency	March 6, 2001	5
Ethan	Metabolic Disorder	January 1, 2002	4
Gideon	Metabolic Disorder	February 28, 2003	3

CDG – Congenital Disorder of Glycosylation
GSD – Glycogen Storage Disease
MCADD – Medium Chain Acyl CoA Dehydrogenase deficiency
SLO – Smith-Lemi-Opitz

Happy Birthday Everyone!

Cole (arginase deficiency featured in the Winter 2005 issue) and his baby sister

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!

Suzanne's Corner

Suzanne Ratko
Registered Dietitian

Spiced Apple Muffins

SHS International

These muffins are full of flavor, and make a great addition for breakfast or anytime snack!

Ingredients:
1/3 cup + 1 Tbsp (100 g) soft margarine
1/2 cup white granulated sugar
1 2/3 cup (200 g) Loprofin Baking Mix
2 tsp egg replacer
2 tsp baking powder
1 tsp ground allspice

6 fluid ounces Loprofin Drink Mix
2 medium apples
1 Tbsp honey

Preparation:
1. Preheat oven to 375 deg.F
2. Place the margarine and sugar in a large bowl. Beat well until light and creamy
3. Combine the Loprofin Baking Mix, egg replacer, baking powder and allspice.
4. Add to the creamed mixture with 5 fl oz Loprofin Drink Mix.
5. Beat the mixture for 1 minute with an electric mixer (2-3 minutes with a wooden spoon) until a thick, smooth batter is formed.
6. Add the remaining Loprofin Drink Mix, if necessary, to achieve a smooth batter.
7. Halve the apples and remove the pits.
8. Cut 10 slices and set aside.
9. Chop the remainder of the apples and stir into the cake mixture.
10. Divide the mixture evenly between about 10 muffin tins.
11. Place the honey in a small bowl.
12. Dip the saved apple slices in the honey and press a slice into the top of each muffin.
13. Bake in a preheated oven for 20-25 minutes, until well risen and firm to touch.
14.Cool on a wire rack.

Makes 10 large muffins.

	Per Serving	Per Recipe
Calories	212	2124
Protein, g	0.31	3.1
Fat, g	8.5	85
Phenylalanine, mg	9.7	97
Methionine, mg	4.2	42
Tyrosine, mg	6.7	67
Isoleucine, mg	10.9	109
Leucine, mg	19.7	197
Valine, mg	14.1	141

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Announcements

3rd Annual Metabolic Family Workshop

Mark your calendars!

- We have grown! This year we will be holding the 3rd annual Metabolic Family Workshop on Friday May 5th 2006 at the Best Western Lamplighter Inn on Wellington Road in London
- There will be many interesting speakers including Dr. Joe Clarke, a metabolic specialist from the Hospital for Sick Children in Toronto and Dr. Simon Levin, a pediatric neurologist from LHSC
- This year there will be talks specific for diet management, neurological issues and lysosomal disorders
- There will be a LOW PROTEIN DIET EXCHANGE. Please send the recipes you would like to submit to janice.little@lhsc.on.ca and include serving size, number of servings, amount of protein per serving and if you have the phe content per serving
- On Saturday May 6th there will once again be a low protein cooking demonstration @ Loblaws Wonderland Market (Southdale & Wonderland location)
- Further details for the workshop & cooking demonstration will be mailed out in March

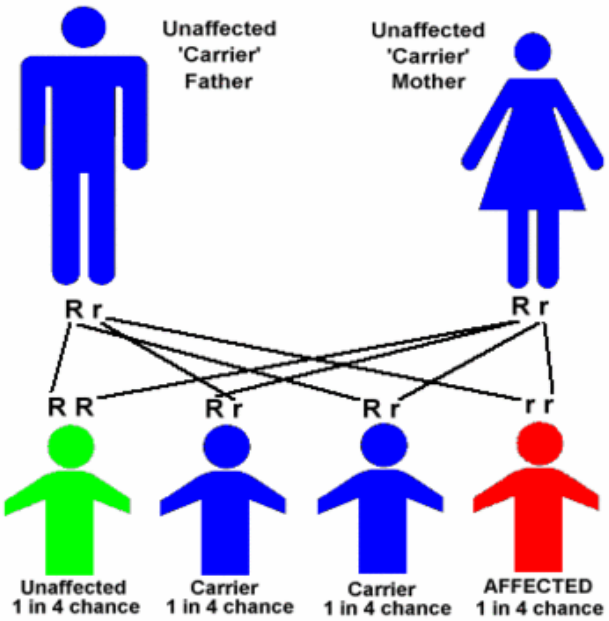
Featured This Issue

What is Tay-Sachs Disease?

Compiled by Chitra Prasad

Tay-Sachs disease in its infantile form 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 condition is caused by insufficient activity of an enzyme called *beta-hexosaminidase A* that catalyzes the breakup of acidic fatty materials known as *gangliosides*. Gangliosides are made and broken down rapidly in early life as the brain develops.

Infants 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. The child unfortunately becomes blind, deaf, and unable to swallow. Muscles begin to atrophy and paralysis sets in. Other neurological symptoms include dementia, seizures, and an increased startle reflex to noise. A much rarer form of the disorder occurs in patients in their twenties and early thirties and is characterized by an unsteady gait and progressive neurological deterioration. Persons with Tay-Sachs also have "cherry-red" spots in their eyes. In fact the disease was first recognized by Warren Tay (1843 - 1927) a British ophthalmologist by the presence of cherry red spot at the back of eye. This sign is very useful and helped us diagnose patients with Tay-Sachs disease and GM1 Gangliosidosis among others. The incidence of Tay-Sachs is particularly high among people of Eastern European and Askhenazi Jewish descent. With carrier screening programs people of Ashkenazi Jewish ancestry are very well aware of the condition however another group where this disorder is seen frequently is French Canadian. Patients and carriers of Tay-Sachs disease can be identified by a simple blood test that measures beta-hexosaminidase A activity.



Genetics: Both parents must carry the mutated gene in order to have an affected child. In these instances, there is a 25 percent chance (1 in 4) with each pregnancy that the child will be affected with Tay-Sachs disease. Prenatal diagnosis is available if desired.

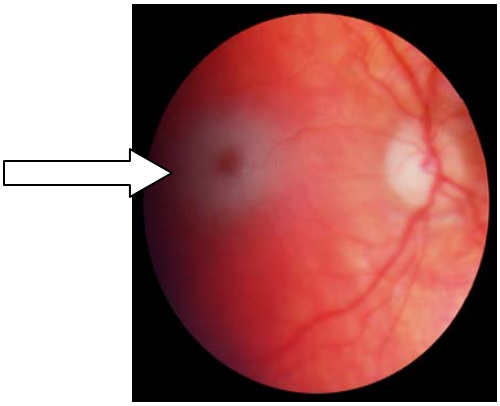
Source: www.answers.com

Tay Sachs disease remains one of the lysosomal disorders where presently there is no curative treatment. A number of barriers to treatment have faced the scientists over last few decades. Anticonvulsant medicine may initially control seizures. Other supportive treatment includes proper nutrition and hydration and techniques to keep the airway open. Children may eventually need a feeding tube. Even with the best of care, children with Tay-Sachs disease usually die by age 4, from recurring infection. The families will require lot of support to deal with such a challenging disorder.

The National Institute of Neurological Disorders and Stroke (NINDS), a part of the National Institutes of Health (NIH), conducts research about Tay-Sachs disease in laboratories at the NIH and also supports additional research through grants to major medical institutions across the US.

We hope that in near future there will be a breakthrough so that these children will be able to survive and lead productive lives.

Cherry red spot in eye
Source: www.snof.org



Sources: http://www.ninds.nih.gov/disorders/taysachs/taysachs.htm

Organizations

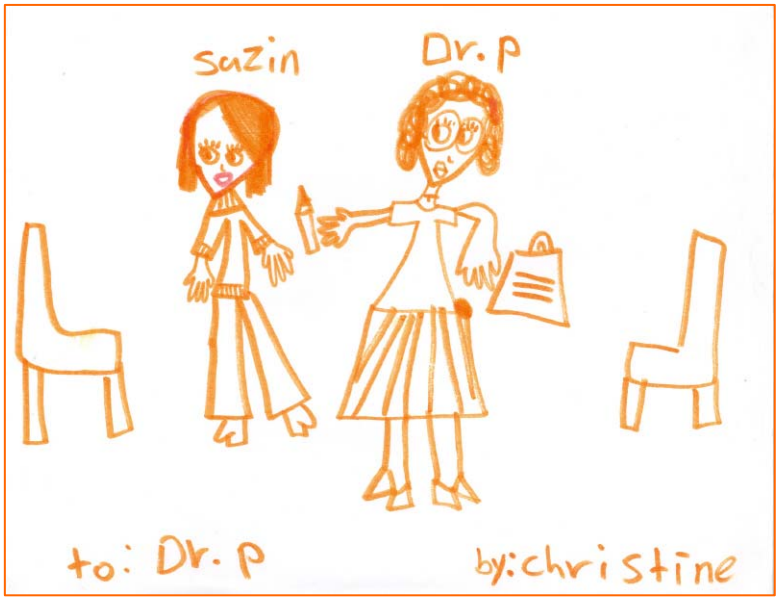
National Tay-Sachs and Allied Diseases Association
2001 Beacon Street
Suite 204
Brighton, MA 02135
info@ntsad.org
http://www.ntsad.org
Tel: 617-277-4463 800-90-NTSAD (906-8723)
Fax: 617-277-0134

March of Dimes Birth Defects Foundation
1275 Mamaroneck Avenue
White Plains, NY 10605
askus@marchofdimes.com
http://www.marchofdimes.com
Tel: 914-428-7100 888-MODIMES (663-4637)
Fax: 914-428-8203

National Organization for Rare Disorders
PO Box 1968 55 Kenosia Avenue
Danbury, CT 06813-1968
orphan@rarediseases.org
http://www.rarediseases.org
Tel: 203-744-0100 Voice Mail 800-999-NORD (6673)
Fax: 203-798-2291

Genetic Alliance
4301 Connecticut Avenue, N.W.
Suite 404
Washington, DC 20008-2369
info@geneticalliance.org
http://www.geneticalliance.org
Tel: 202-966-5557 800 336-GENE (4363)
Fax: 202-966-8553

Canadian Assoication for Tay-Sachs and Allied Diseases (CATSAD)
569 Laural Drive
Burlington, Ontario, Canada, L7L 5E1
info@catsad.ca
http://www.catsad.ca
Tel: 905.634.4101



Joan and Christine (artist of picture)

Sisters with Glycogen Storage Disease

Contact Information

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1.519.685.8453

Email: janice.little@lhsc.on.ca
Website: www.lhsc.on.ca/ programs/rmgc/met/metaboli.htm Please note there has been a slight address change and you will need to update your bookmarks.

Yahoo Online Chat: health.groups.yahoo.com/group/metabolic_disorders
Post Message: metabolic_disorders@yahoogroups.com

Parent Support Contact: Jennifer Culp

Tel: 1.519.632.9924