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***The most astonishing thing about miracles
is that they happen."***

***–G.K. Chesterton English author
(1874-1936)***

From the Editor

Greetings everyone!

I hope you enjoy the newest section entitled "Food For Thought" This will be a regular section in the newsletters, and provides some excellent suggestions for keeping your heart healthy.

Have a safe and happy summer, everyone!

Janice Little

From Dr. Chitra Prasad

Dear Friends,

I am sure you are all quite busy making plans for the summer.

We had the most wonderful Metabolic Family Day, celebrated on the 11th of May. You can read all about it in our newsletter. As our main speaker, Dr. Martin Collis emphasized "Our wellness in all spheres is the best gift we can give to ourselves and others."

I am always amazed at the strength and commitment that you all possess when placed in extremely challenging situations. You will read about one such inspiring story of BJ Lawrence, who brings joy and happiness to others even in the midst of his own suffering.

Janice, our editor, has put the beautiful butterfly picture on this newsletter, which reminds me of this poem:

A Butterfly

A butterfly lights beside us
Like a sunbeam.
And for a brief moment
Its glory and beauty
Belong to our world.

And then it flies on again
And though we wish
It could have stayed
We feel blessed to have seen it.

-- Anonymous

Have a wonderful and glorious summer with your friends and families.

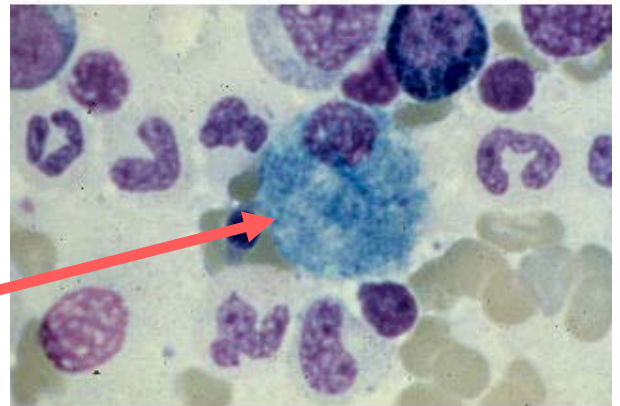
Your friend,

Chitra Prasad

Featured This Issue

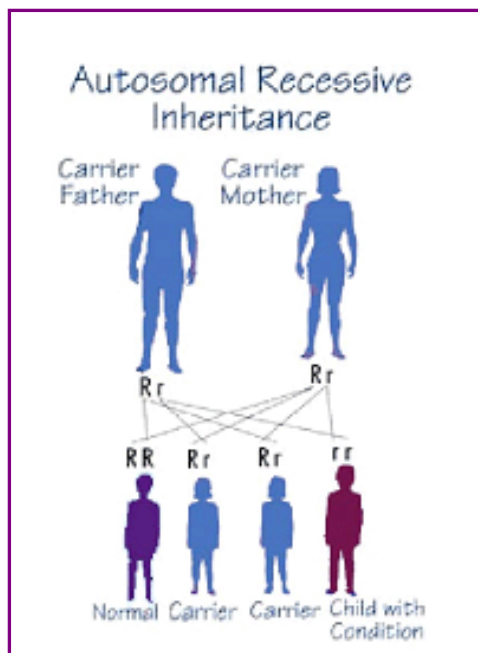
Niemann Pick C compiled by Dr. Chitra Prasad

Niemann Pick C is a storage disorder of the lipids where harmful quantities of a fatty substance (lipids) accumulate in a number of body organs such as the spleen, liver, lungs, bone marrow and the brain. The worst problems happen when the brain is involved. Affected people can have problems walking, talking, and remembering things. Swallowing difficulties occur, leading to choking episodes and pneumonia. Diagnosis for this disorder can be difficult, as there are numerous presentations.



Niemann Pick C cell (blue cell)

Source: www.icampus.ucl.ac.be/PEDIHEPA/document/pedihepa/seabluehist.jpg



Niemann Pick C is an autosomal recessive genetic disorder. This means that one defective gene for Niemann Pick C was inherited from each parent as shown in the diagram.

<http://www.hawaii.gov/health/family-child-health/genetics/newimg/autosomalrecessive.gif>

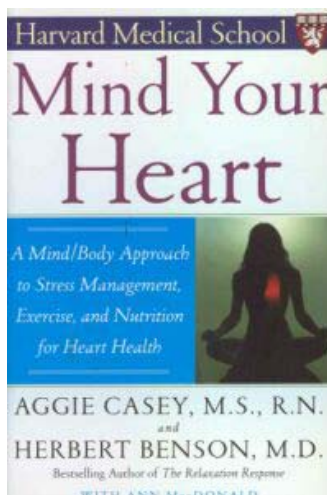
The diagnosis of NPC is confirmed by biochemical testing that demonstrates impaired cholesterol esterification and positive filipin staining in cultured fibroblasts. Biochemical testing for carrier status is unreliable. Most individuals with NPC have NPC1, caused by a mutation the *NPC1* gene; fewer than twenty individuals have been diagnosed with NPC2, caused by mutations in the *NPC2* gene . Molecular genetic testing of the *NPC1* and *NPC2* genes detects disease causing mutations in about 94% of individuals with NPC.

Management of NPC is supportive. Symptomatic therapy may be helpful in managing seizures and abnormal movements. A night time sedative may be indicated for sleep. Chest physical therapy with antibiotic therapy for infection appear beneficial in preventing secondary complications. A regular bowel program for mobility-impaired individuals is important. Physical therapy is indicated to maintain movement as long as possible. Swallowing is monitored to allow placement of a gastrostomy tube when aspiration or inability to eat is a concern.

References Researchers, such as Dr. Marc Patterson, Director of Pediatric Neurology, The Neurological Institute of New York New York are working very hard to find a cure for Niemann Pick C.

www.geneclinics.org

Food For Thought



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

- ◆ Find some time during the day to meditate or listen to a relaxation tape.
- ◆ Organize your work; set priorities. Make a list daily, then evaluate your various tasks and divide them into three columns:

A (must do), B (can wait), and C (let it go)

- ◆ Don't try to be perfect. Don't feel like you have to do everything. It's okay to make a mistake. You're only human.
- ◆ Don't try to do two or three or more things at a time
- ◆ Reduce the noise level in your work or home environment, if possible. Instead, listen to soothing music.

Kids Korner

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



Child's Name	Metabolic Disorder	Birthday	Age
Daniel	PKU	April 25, 2003	4
Zacharia	PKU	June 6, 2004	3
Olivia	PKU	September 6, 1991	16
Hailey	PKU	August 19, 2003	4
Patrick	PKU	July 2, 1991	16
Julia	PKU	July 29, 2001	6
Abilene	PKU	June 12, 2005	2
Stathie	PKU	May 7, 1992	15
Dusan	PKU	July 21, 2003	4
Carter	PKU	September 5, 2001	6
Elijah	PKU	June 18, 2003	4
Samantha	PKU	July 31, 2000	7
Brock	ALD	April 6, 1991	16
Royce	ALD	April 6, 1991	16
Cameron	Alpha-1 Antitrypsin Deficiency	July 10, 2004	3
Cameron	Ethylmalonic Aciduria	April 26, 1995	12
Ema	Galactosemia	August 9, 2005	2
Alexander	Galactosemia	May 8, 2001	6
Joan	Glycogen Storage Disease	June 20, 2003	4
Michelle	HMG CoA Lyase Deficiency	September 1, 1999	8
Emma Lynn	Hurler	September 9, 2004	3
Austin	MCADD	July 17, 1996	11
Stanley	MCADD	August 26, 2003	4
Stephanie	MCADD	June 16, 2006	1
Shaylin	MCADD	July 13, 2006	1
Kelly	MELAS	July 10, 1995	12
Shawn	Menkes	July 13, 2006	1
Natasha	3-Methylglutaric Aciduria	August 19, 2004	3
Chelsey	Mitochondrial Complex 1 Deficiency	August 4, 1994	13
Aaron	ML4	Aug 30, 1993	14
Jordynn	5-MTHFR	May 30, 2000	7
Jared	OTC	September 22, 1993	14
Jack	OTC	August 8, 2005	2
Samantha	OTC	July 19, 1991	16
Marlo	Pyruvate Dehydrogenase Deficiency	August 15, 2005	2
Hanza	Sanfilippo	April 3, 2005	2

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

Paisley	Metabolic Disorder	September 24, 1999	8
Zachary	Metabolic Disorder	September 4, 2005	2
Wyatt	Metabolic Disorder	July 18, 2002	5
Alexa	Metabolic Disorder	September 29, 1993	14
Jeff	Metabolic Disorder	June 24, 2001	6
Madison	Metabolic Disorder	June 23, 2000	7
Nicholas	Metabolic Disorder	September 1, 2002	5

PKU Phenylketonuria
MCADD Medium chain acyl-coA dehydrogenase deficiency
MELAS Mitochondrial myopathy, encephalopathy, lacticidosis, stroke
5-MTHFR Methylene tetrahydrofolate reductase deficiency
OTC Ornithine transcarbamylase deficiency
ML4 Mucopolipidosis type IV (4)

Happy Birthday Everyone!

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!

Resources

The Canadian Society for Mucopolysaccharide & Related Diseases Inc.

www.mpssociety.ca

National Niemann-Pick Disease Foundation, Inc.

www.nnpdf.org

Suzanne's Corner



Pizza

A popular dish with the whole family and it's also very versatile. Pizzas can be used as main meals, snacks and cold in packed lunches too.

Serves: 4

Oven Temperature 190°C, 375°F

Base

225g (8 oz) Loprofin Low Protein Mix
A pinch of salt
2 tsp baking powder
50g (2oz) margarine
150ml (5 fl oz) water (approximately)

Topping

25g (1 oz) margarine
1 onion, chopped
50g (2oz) mushrooms, sliced
Pinch of mixed herbs
2-3 tomatoes, skinned and sliced
1 green pepper, deseeded and sliced
Black pepper

Sieve together the Mix, salt and baking powder; rub in margarine. Add enough water to give a soft dough. Turn out on to a smoother surface lightly dusted with mix and knead until smooth. Roll or press out to a 20cm (8 inch) round and place on a greased baking tray.

For the topping, melt the margarine and gently fry onion and mushrooms until soft. Place on the pizza base. Sprinkle on the mixed herbs and then place the tomato and green pepper slices on top. Sprinkle with black pepper. Bake in a pre-heated oven for 30-35 minutes until the base is cooked and the topping browned.



Nutrition Facts

Amount per serving: ¼ of pizza

Calories	388 kcal
Protein	1 g
Fat	20 g
Carbohydrate	51 g
Sodium	455 mg
Potassium	205 mg
Phosphorus	275 mg

Source: SHS International, found at: www.shsweb.co.uk

What's New

Metabolic Family Workshop & Low Protein Cooking Demonstration

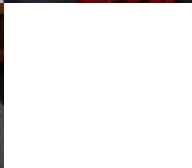
The 4th Annual Metabolic Family Workshop & 3rd Annual Low Protein Cooking Demonstration were held on Friday, May 11, 2006 at the Lamplighter Conference Centre and Saturday, May 12, 2006 at the Loblaw's Wonderland, respectively.

On Friday, 180 family members and professionals from Southwestern Ontario & the Toronto area listened to the keynote speaker, Dr Martin Collis, motivational speaker, discuss achieving well being & balance in your life with humour, exercise, and laughter. Lindsay Moir, school advocate spoke to the captive audience on "Current Issues in Ontario Special Education". After which the attendees split into 3 groups for the concurrent sessions on the following topics: "Financial Security for the Child with Disabilities – The Henson Trust" – Paul Strickland; "Living with someone who has a metabolic disorder" – Rénald Richer & Dr Martin Collis; and "For Teenagers/Young Adults Only" – Dr Beth Mitchell and Brian Seeley.

Both Dr Beary Goode & Ollie Pale visited the children in the Play Room. The A-Channel, Roger's Television and Fanshawe College Radio provided media coverage of this fantastic event. The formula and food vendors offered an excellent opportunity for families to try new products. Most importantly, the families were able to network with other families dealing with similar issues and staff in relaxed, interactive environment.

On Saturday, 47 family members and professionals from Southwestern Ontario, Hamilton, & Toronto watched Erica Lesperance RD from Cambrooke Foods (Boston) prepare many delicious low protein products using medical foods and fresh fruit & vegetables.

The overall feedback was absolutely positive. Families provided ideas for future events. The professionals and vendors that attended were thoroughly impressed by the organization and family attendance.



Spring/ Summer 2007

Volume 4 Issue 2

Silent Auction

The silent auction was very successful, raising over \$450.00 which was used to help cover costs for the Metabolic Family Workshop and Low Protein Cooking Demonstration. We are hopeful this fundraising effort will grow every year.

A big thank you goes to Richard Rodgers, who helped immensely organizing the auction and collecting the money.



Newborn Screening Update

Currently in Ontario, 23 endocrine and metabolic disorders are being screened for. Recently added disorders include biotinidase deficiency and galactosemia. In the near future, screening will begin for congenital adrenal hyperplasia and cystic fibrosis .

For more information, visit:

The Ontario Newborn Screening Program : www.health.gov.on.ca/newbornscreening



Ewa, adult with PKU

Contact Information

Editor: Janice Little
LHSC - Medical Genetics Program of Southwestern Ontario
Tel: 1.800.243.8416
1.519.685.8453

Email: janice.little@lhsc.on.ca
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
Post Message: metabolic_disorders@yahoogroups.com

Parent Support Contact: Jennifer Culp

Tel: 1.519.632.9924

Email: donjen2000@hotmail.com