The Inherited Metabolic Disorders News

Children's Hospital
London Health Sciences Centre

Winter 2016 Volume 13 Issue 1

In this Issue

From the Editor

Welcome to the latest issue of The *Inherited Metabolic Disorders News*! So far, the winter weather hasn't been too bad (at least for those close to London!) and spring will soon be here. We will be hosting the 12th annual **Low Protein Cooking Demonstration** on **Saturday May 14th, 2016**. Details are in the newsletter. Our next **Metabolic Family Workshop** will be held on **Friday, May 12, 2017.** Thank you to all who have provided us with your email address for electronic communication (including the newsletter). Those who have access to a computer who have not provided their email address, please send it to **janice.little@lhsc.on.ca**

For those without computer access, we will continue to send you a printed copy.
As always, your comments, stories and suggestions are welcome!

Janice Little

From Dr Chitra Prasad

Dear Friends
Greetings from all of us at the metabolic team!
As some of you might recall I had talked about issues related to short stature in our previous issue of metabolic newsletter. What happens when you are excessively tall as a result of a metabolic condition? You can read about "Tall Paul" and his story in this newsletter. Paul has a condition called Homocystinuria. I have summarized this condition in the next page.
Paul however is much more than his condition. He is an extremely enterprising individual with a smiling face and a very helpful nature. He is working full time and he has made a great impact on his community. I remember the first time I met him. It was hard for me to even measure his height. Since he is more than 6feet 8 inches and most of you know I am only 5ft 2 inches. Paul was very obliging. He helped me with measuring of his own height!
We all come in this world with our unique qualities and strength. It is important to remember that in times of difficulties and challenges.
My travels in India this time took me to Mumbai and my medical school Christian Medical College in Ludhiana. I did lectures for the residents and met some of my friends and colleagues
I am very thankful to all of you who have been providing emails. Soon we will be able to send every one the email version of the newsletter. For those who are still getting the print version, please let Janice know your emails. Please note we will be doing the metabolic family workshop once every two years and low protein cooking demonstration every year. Hope to see you all on the **12th May 2017** for our next metabolic family workshop!

With best wishes to all
Your friend
Chitra Prasad

Dr. Seuss

Today you are You, that is truer than true. There is no one alive who is Youer than You.

Personal Stories

Homocystinura

Paul was born Oct. 31, 1983. It was in Junior Kindergarten when Paul and his parents were told he had Homocystinura. It was a doctor in Owen Sound . Some of the features of Homocystinura are long thin body, eye lens may disconnect at anytime, and slow learner. (Paul is 6 feet 8 inches tall now at 31. Everyone calls him Tall Paul.
Had all the rest of the children tested in Toronto. Paul has 1 brother and 2 sisters. They were all fine. Paul has been taking medication, formula and vitamins ever since . He is on a low protein diet also. Paul has a check up in London once a year, eyes checked every 3 years and bones checked every 2 years.
Never any problems taking medication etc.
Always had an aide through grade school and high school . Answered questions on one to one bases. Did get a Grade 12 diploma and also did co-op through high school. Worked for landscapers, in garden centers and grocery store.
Paul now has full time work in a grocery store since grade 12 (stocks shelves, coolers and packs groceries). Does deliveries of groceries to the senior home and the hall for the meals to be catered. He also works for Humpty Dumpty potato chips. He stocks shelves in some of the local stores in the area.
Paul has been able to drive a car since he turned 16, there again they read the test to him and he answered the questions.
Paul joined the Lions Club 12 years now. Helps with volunteering in many of the Lions projects and does the decorating for there special events.
Started Tall Paul Decorating—does Weddings, Christmas Parties, Anniversaries, Fall Fairs, dinner table center pieces and graduations for the local grade and high school.
On Jan. 7, 2015 received Outstanding Citizen in the ridings of Huron and Bruce. Presented by local M.P. Lisa Thompson. The words that describes him for the award -- Paul , better known as Tall Paul, is a tireless volunteer who inspires everyone around him. Paul brings enthusiasm , energy and inspiration to all the numerous community projects that he is involved with. His passion is decorating and with that he brightens our world. Paul's volunteerism does not only touches his home town , but the surrounding towns have him helping them out to. One only has to be in a parade with him and hear the endless greetings, to realize how loved this young man is. Tall Paul's positive attitude , his constant smile and his funny laugh can motive an entire committee to do more and make it better. Paul has been involved with almost every committee and every project that happens in and around town, He took charge of cleaning the Rec. Center, he was a driving force

Personal Stories - continued

that made free roller skating happen for the youth, he is an active and busy member of the Lions club and the St. Joseph Aid Society and the Nativity Scene committee. Paul is seldom the person in charge of a committee but he can sure make the grunt work happen in an up lifting and fun manner. He is a team player, a team motivator and does more than his share. Paul's unbeatable community spirit was inspired from youth through his involvement with the scout movement and through years of practice with his parents.

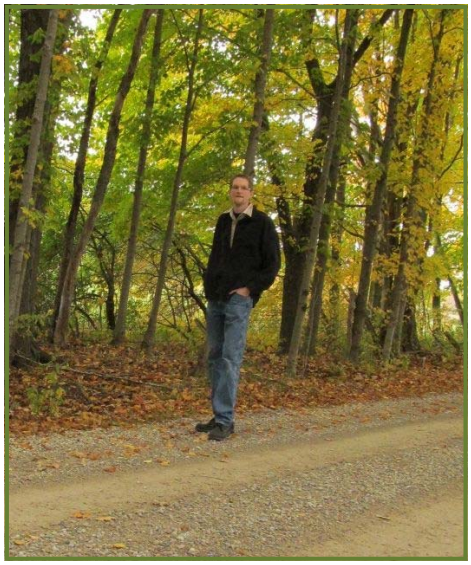
When he turned 30 he had a big birthday party for himself which was pretty well organized by himself with a tent, music, outdoor lights, supper for all 200 guests. He pulled this off with the help of his brother and sisters and Mom and Dad.

Paul is on a low protein diet-- still quite hard to follow , likes his meat. He know when he goes off the diet he gets very tired. Has to go to the local hospital every month for bloodwork.

Through all of this Paul is always happy, smiling , friendly and is always willing to help anyone who asks or if he sees something to be done, he will do it.

Paul putting up a ceiling for a wedding

"Music in the Field" in Lucknow with Chad Brownlee (Canadian Country Singer)



Featured This Issue

Homocystinuria

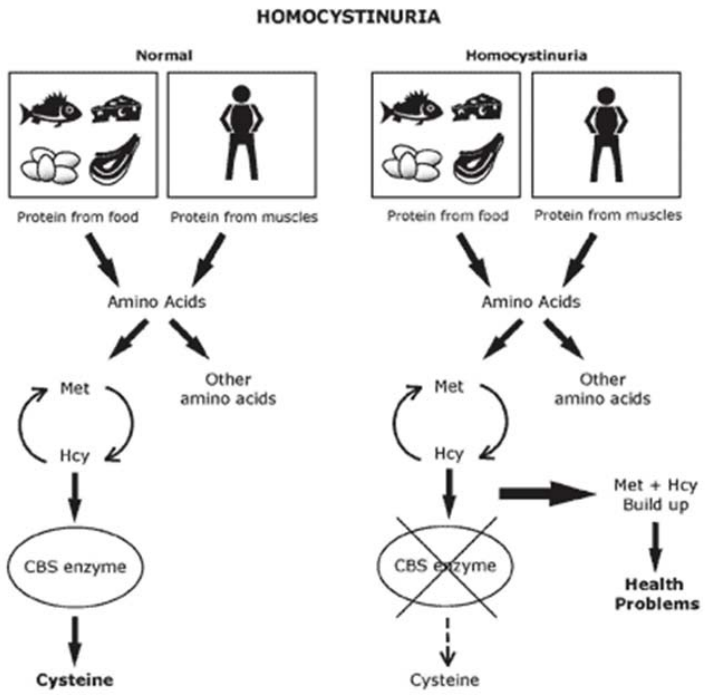
By Dr. Chitra Prasad

Homocystinuria is an inherited disorder in which the body is unable to process certain building blocks of proteins (amino acids) properly. Homocystinuria is characterized by nearsightedness (myopia), dislocation of the eye lens, an increased risk of abnormal blood clotting, and brittle bones that are prone to fracture (osteoporosis) or other skeletal abnormalities. Some affected individuals also have developmental delay and learning problems. The signs and symptoms of homocystinuria typically develop within the first year of life, although some people with a mild form of the disease may not develop features until later in childhood or adulthood. Mutations in the *CBS*, *MTHFR*, *MTR*, *MTRR*, and *MMADHC* genes cause homocystinuria. Mutations in the *CBS* gene cause the most common form of homocystinuria. The *CBS* gene provides instructions for producing an enzyme called cystathionine beta-synthase. This enzyme acts in a chemical pathway and is responsible for converting the amino acid homocysteine to a molecule called cystathionine. As a result of this pathway, other amino acids, including methionine, are produced.

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. Most often, the parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but do not show signs and symptoms of the condition.

Treatment involves taking vitamins such as pyridoxine (as some of the patients may be responsive to Pyridoxine) other medications include Betaine, Folic acid, low protein diet and special formula, Aspirin to avoid strokes etc)

Regular consultations with eye specialist, neurologist, endocrinologist for bone health, metabolic specialist and dietitian are recommended.



Reference: **http://ghr.nlm.nih.gov/condition/homocystinuria**

Metabolic Pathway Reference: **https://cornellbiochem.wikispaces.com/Homocystinuria**

Pediatric Case Study (Summerized)
Authored by Barbara Brown - 1968

Phenylketonuria

Nancy Emerson

Born on September 12, 1955, Nancy was to be the fifth child in a family of seven. She was a beautiful baby, a perfect child, or so it seemed. To her family and friends, she was no less normal than the other new arrivals in the nursery. But in the future months to come, her emotional, social, motor and mental development was to fall behind that of other children her age so much that by 18 months old she could do nothing that was normal for her level. She could not walk or run or even sit up. She spoke no words at all. She showed no love for the members of her family. She could not play with toys; she could not play! She did not laugh or show pleasure from the care given her. She had no contact with people. It was evident now that she lacked so much.

In the company of her parents she was taken to the Hospital for Sick Children for help. Through testing and evaluations she was diagnosed as a PKU child.

Nancy at 18 months of age was not placed on the diet for two reasons, one being the newness of the diet and the other, the family situation. There were at this time 5 children in the family. The father was employed as a truck driver and the mother as a housewife, putting them in a lower social economic level. They apparently did not show that much interest or enthusiasm in Nancy's situation and the Doctors felt it would be of no use to attempt the strict diet in this environment.

For the next 3 ½ years Nancy remained untreated and uncontrollable. She was allowed to do much as she pleased. No attempts at controlling her temper tantrums or disagreeable behavior were taken.

At the age of 5 in September 1960 she was admitted to the Welland District School for Retarded Children only to be taken out within the month as she was a "Behavior Problem". In July 1961 Nancy was evaluated at the Welland and District Public Health Unit by Mr. R.C. Smart, Psychologist and the following findings were sent to the principal of the Welland School.

"Given her present level of functioning, it is unlikely that she would profit from retarded children school - her habit control, incontinence, insociability and mutism would militate against her. However, the signs of improvement indicate that she should be seen every 12-18 months for the next few years in order to keep account of her development. Eventually she may be helped by Retarded Children's school but residential treatment in the O.H. would likely be essential by the time she is 9-10. She is happy and well cared for at home now and this in addition to her parents' attitude make it difficult to suggest institutionalization now."

In May of 1962, Nancy was again evaluated and the judgment was has a mental age of 2 ½ and has done better than expected, may be ready for school this fall, should have a chance

In September of 1962, the school readmitted Nancy but only to see her leave again in June as her parents found a move to Crystal Beach necessary.

In September of 1963, Nancy was admitted to the Fort Erie Area School for Retarded Children. This school had an untreated P.K.U. child who had started the diet at 6 years of age and had made remarkable progress, so they were very interested in Nancy and her condition.

Nancy had difficulty fitting in to the school program, she was noisy and disruptive, ate anything and everything including floor tile and cried "copious" tears for no apparent reason. She did not respond to gestures of friendship and never looked directly at anyone. She had violent tremors and at times shook so badly that she was unable to lift a glass to her mouth. To quote her teacher "at times she seemed tormented". This possible could be due to violent headaches which accompany her condition. Her balance was greatly disturbed making her general mobility poor, she could not run, jump or skip. She exhibited a marked left sided weakness. Her hands were flaccid and she shook them frantically. The only parts of the school program that she readily enjoyed were the musical activities and during these sessions she was so nervous and excited that it was impossible to conduct the class. They found that if they seated her next to the teacher at the piano (Mrs. Foden) that she was less noisy.

She made only unintelligible sounds. She did not use her hands for crayons or pencils but made some attempts at finger painting. Her attention span was short. She carried her dishes to the sink and put her placemat away and responded to a few simple commands such as some, sit, no. She was very strong willed and exhibited strong outbursts of temper. If denied something, these outbursts were immediate. She always seemed ravenously hungry and ate vast quantities of cookies which never seemed to satisfy her. Fortunately, she absolutely refused to drink milk. Certainly she did not present the picture of a model student and was a challenge to her teachers.

Parent-teacher interviews were set up after she had been to school a few months and the possibility of the mother placing her on the diet for P.K.U. was discussed and encouraged by the staff of the school. The mother said she'd think it over and later informed the principal that she and her husband had seen enough Doctors for Nancy and they were not going to put out any more money to be told that Nancy was retarded.

Nancy remained in school until October 1964, when it was decided by the Admissions Committee that unless some measure was taken to help Nancy to become more comfortable and to modify her behavior, she would be excluded from the school.

The mother made an appointment with the Children's Rehabilitation Centre and Dr. R. Warner in Buffalo to have Nancy evaluated and to seek his opinions as to the advisability of placing Nancy on the diet. The mother was most encouraged after this initial visit with Dr. Warner and reported to the school that Dr. Warner was most optimistic that Nancy's behavior and development would be greatly improved by the diet.

Nancy's mother discussed her visit to the Dr. with her husband and as a result of this discussion sent a letter to the school informing them that they considered it physically impossible to even attempt the diet. They said they had tried to remove a few things from her over the weekend and that this had triggered a chaotic situation in the already tension strained home.

The principal (being a nurse) wanted Nancy to have her chance and with Dr. Warner's encouragement, asked the parents' permission to rake her into her home and institute the diet. The idea was that once Nancy was on the diet and no longer a behavior problem, the family would be able to manage.

Much to everyone's surprise and delight, the parents agreed to this move.

A battery of tests were done on Nancy at the clinic in Buffalo and the included visits to Dr. Weinstein, a neurologist, a speech therapist, an occupational therapist, an education counselor, x-rays, lab tests and visits with Dr. Fischer, a psychologist and a nutritionist. After the tests were done a professional conference was held to which the principal and the school supervisor were invited to attend, where reports of the various disciplines were read, discussed and recommendations made.

Following the professional conference a parent interview was held and last and very important a conference with the principal and her husband (Mr. and Mrs. Emerson) and Dr. Warner to discuss the problems which they were likely to meet in the first few months of Nancy's new regime. Mrs. E says that "backed by so much support and knowing that professional help and moral support were as near as the telephone made the undertaking less formidable." A social worker was assigned to Nancy's case and a nutritionist was always available to assist her with any dietary problems.

On January 1, 1965 Nancy became a temporary member of the Emerson household which is comprised of Cathy 15, Ted 16, a dog, a cat, and of course a father and a mother. The Emerson's live 7 miles from Nancy's family in a large house on the shore of Lake Erie. It seemed like an ideal environment for Nancy (except for the water of which she was extremely fearful and would not go near) who came from a crowded rundown house with no yard in a poor neighborhood in the village of Crystal Beach.

She was started immediate on her diet which she refused to take. The first 10 days were very trying. Nancy went through a withdrawal stage. She vomited and lost a great deal of weight, her eyes were sunken and her appearance

seemed to indicate that she was gravely ill. If it had not been for the encouragement of the Dr. and his assurance that she was alright, they may have given up. After 10 days she started to eat a potato salad and drink a bit of formula. She refused the low protein bread which is obtained from the Faculty of Food Services of the University of Toronto at a $1.25 a loaf. Each loaf contains 30 mg of phenylalanine and will cut into 15 slices making each slice containing 2 mg. This is an excellent filler and can be used to satisfy the child. As of today, Nancy still will not eat it.

It is no less than remarkable the improvement that Nancy has shown. She loves and is loved by all members of her new family who legally adopted her in 1967. From schizoid introvert she has blossomed into a loving and happy extrovert.

Singing and Music: Nancy responds to music and has developed a sense of rhythm. She plays the drum in the rhythm band. She is showing steady improvement in the dance classes especially tapping, toe heel, brush step, and sliding side step. Nancy was in the Christmas concert and playing her Moroccans on cue and sang “la, la, la” in a song. She was quiet, exhibiting model behavior for 1 hour before an audience that filled the room. Unbelievable to those present.

Nancy is able to wash and dry dishes and set a table in sequential steps. She gets out toothbrushes and combs, sets out the necessary equipment for making her formula, as her teacher says “her efficiency is in direct ratio to her fondness for the task”. Nancy is given ample opportunity to try her hand at all household tasks. She loves to help in anyway.

It will be interesting to see what the future holds for Nancy. Dr. Warner says that she has improved “one hundred fold” and the parents say “you can’t ask for more”. Nancy is happy and content and she has a full day of activities planned for her. She is living at school and at home in a loving but non permissive atmosphere. Her activities are balanced between the active and passive and she is being provided with meaningful experiences which will help her become a useful member of her family and an acceptable member of the community.

“As each star differs in brightness so do the children of God yet each is entitled to develop his full potential physically, mentally, socially and spiritually”. Pearl S. Buck

Suzanne's Corner

Hummus

Nutritional Facts

Serving	Recipe
Energy	104 kcal
Protein	0.5 g
PHE	21 mg
TYR	12 mg
LEU	26 mg
VAL	21 mg
ILE	15 mg
MET	5 mg
LYS	22 mg

Yield

Number of servings: 8
Serving size: 1/4 cup (60 ml)

Ingredients

- ½ cup (125 ml) eggplant (50 g)
- ½ cup (125 ml) zucchini squash (80 g)
- ½ cup (125 ml) onion (70 g)
- ½ cup (125 ml) red bell pepper (70 g)
- 1 Tbsp (15 ml) olive oil
- 2 cups (500 ml) cooked Aproten orzo, which is 2/3 cup dry or 140 g (0218)
- 3 grilled garlic cloves
- 1 Tbsp (15 ml) olive oil
- 2 Tbsp (30 ml) lemon juice

2-3 Tbps (30-45 ml) hot water

Preparation

- The day before, or whenever you use the oven, prepare the grilled garlic. Cut the top of the garlic head, wrap it in foil and bake it for 45-60 minutes. Keep it in fridge and use it everywhere! Try it on grilled bread or pizza crust!
- Preheat the BBQ or oven to broil.
- Cut the vegetables into. Brush with olive oil, salt and pepper. Grill the vegetables. Keep aside.
- Meanwhile, cook the orzo in plenty of boiling water for 9 minutes. Rinse for a few minutes after cooking.
- In food processor, place the orzo, grilled vegetables and grilled garlic. Pulse to chop coarsely. Add the olive oil and lemon juice. Pulse again to obtain a purée. Add water, enough to obtain a silky texture. Spread on crackers or vegetables. Perfect in a sandwich or wrap with vegetables!

From Genevieve Lafrance

www.lowprorecipes.com

12th Annual Low Protein Cooking Demonstration

Saturday, May 14, 2016

Time: 10:00 am to 12:00 pm (registration at 9:30 am)

Real Canadian Superstore

825 Oxford Street East at Gammage Street
London, ON

CAMBROOKE THERAPEUTICS

Children's Hospital
London Health Sciences Centre

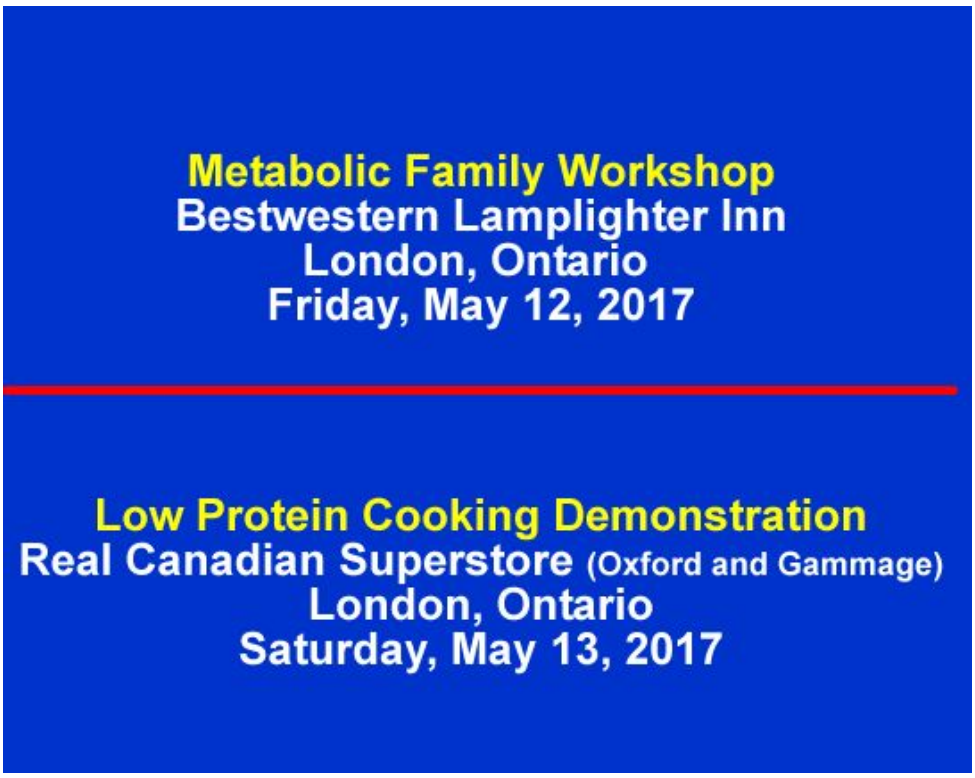
For further information, contact Suzanne Ratko
519-685-8500 Extension 52469

What's New

11th Annual Cooking Demonstration
Real Canadian Superstore
825 Oxford Street East
London, Ontario
Date: Saturday, May 14, 2016
Time: 10:00 am – 12:00 pm (Registration starting at 9:30 am)
Sponsored by Cambrooke Therapeutics
Special Guest: Geneviève Lafrance, Dietetiste (Dietitian)
Creator of www.lowprorecipes.com
Geneviève Lafrance, Dietetiste

In 2007, Geneviève began working with clinical inherited metabolic diseases in Sherbrooke, Quebec at the Centre Hospitalier Universitaire de Sherbrooke. Along with her passion about food in all forms and a very curious nature, Geneviève looks for new low protein products in the stores. In 2013, she began to explore different ways to help her families follow a healthy and restricted protein diet, which also would be tasty and appetizing. The idea of creating a recipe website came to her. Since the official launch of www.lowprorecipes.com which took place in 2014, Geneviève has been adding an average of one recipe per month on this site.
Geneviève has been acknowledged on numerous occasions for her work. In 2014, she received the "Innovation Award'' from her peers and College, Order of Dietitians. In connection with her low protein recipe website, Genevieve has led several cooking demonstrations mainly in Quebec and New Brunswick.
Her objective is simple: prove that cooking a reduced protein meal can be simple, attractive and most importantly delicious!
We are thrilled to have Geneviève join us on May 14, to leading us in some fun and delicious cooking.
Thank you to Cambrooke Therapeutics for sponsoring our event!
Space is limited.
Please contact Suzanne Ratko to secure your spot
519 685 8500, ext 52469

Save the Date in 2017 !!!



Behind the Scenes

Melanie Napier

As many of you are aware, I am the metabolic genetic counsellor working with the individuals/families that are followed by Dr. Chitra Prasad and Dr. Natalya Karp. Many of those seen in our metabolic clinic were diagnosed through newborn screening - early detection and treatment is important for so many of these conditions! My role has now expanded to include working with families whose children screen positive on newborn screening. If you have a child with a condition that was diagnosed on newborn screening and are currently pregnant, please let me know! I can follow-up on the newborn screening results for the newest addition to your family.

Jennifer DiRaimo

As of 1 October 2015 I will no longer be the primary newborn screening genetic counsellor at LHSC. The torch will be passed to Ms. Melanie Napier, Genetic Counsellor, within the metabolic program. Melanie will handle all the metabolic/ SCID newborn screening referrals received from Newborn Screening Ontario (NSO).

It's been almost ten years since I started working in the area of newborn screening. I've enjoyed working with such an incredible team. I am proud to have worked in the area of NBS. I will be transitioning into a full time Cancer Genetics Genetic Counselling position here at LHSC, beginning of October, however I will be providing coverage for Melanie whenever she is away. So, I will remain connected to the metabolic team. I wanted to thank the metabolic team at LHSC. They have been wonderful to work with and I will cherish the wonderful memories and families I've had the chance to meet through newborn screening.

Warmest Regards,
Jennifer DiRaimo, MS, CCGC
Genetic Counsellor

From Dr. Chitra Prasad -

Janice Little

Many of you know her as the lady who sends you the metabolic newsletter, who handles registration and many other aspects of the metabolic family workshop. Janice, to me is a very creative person who has embraced the concepts of newsletters and the metabolic family workshop over the last 13 years. She has great skills at the computer and can make the Newsletters look pretty and also full of information. She is an integral part of our metabolic team and I am very thankful to her for all her support.

Angela Kerr

I am sure you all are familiar with Angela, our clerk who greets you as you first come to the “window”. Angela has a delightful British accent and an infectious smile. She also calls many of you to remind you of the appointments. All of us in the genetics and metabolic team would not know what to do without her! She knows

What's New - continued

all about the appointments. Again I am very thankful for all her help and support to the metabolic team and the metabolic clinics.

Janice & Angela

New member of the team:

Cara Gordon

I began my social work role with the Metabolic team in October 2015. My role on the Metabolic team Iincludes many aspects such as providing emotional support and linking individuals to appropriate community resources. Before beginning this role I worked with individuals 16 and over with mental health challenges and individuals experiencing crisis in the community. I also have clinical experience working with children with multiple neurological disorders as well as mental health challenges. Throughout my undergraduate and graduate studies I gained experience in social and neuroscience research. I look forward to continuing to work with the Metabolic team and continuing to meet the individuals and families that our team works with**!**

Research, Presentations, Lectures

Hello Everyone,
I attended the annual symposium for the SSIEM (Society for the Study of Inborn Errors of Metabolism) in September 2015. This is a large international conference and I was able to learn about new and ongoing research in all areas of metabolic disease from top experts from around the world! Here are a few highlights from the meeting that I hope are of interest to you.

PKU

When someone is diagnosed with PKU or hyperphe, we often order genetic testing to learn what genetic mutations that person has in their PAH gene. One of the reasons we do this is to find out if the mutations are predicted to be "responsive" to Kuvan (meaning, will the medication work for the person with PKU). Kuvan ssentially helps to lower a person's phenylalanine levels and allows them to eat more protein in their diet. However, our predictions about who will respond are not always correct based on our current information. Dr. N. Shen and colleagues in Germany are currently studying the interaction of into which combination of genetic mutations will respond to Kuvan treatment, which will allow us to identify which patients will receive the most benefit from this medication.

Research, Presentations, Lectures - continued

different genetic mutations in the PAH gene and determining the level of responsiveness to Kuvan. It is hoped that they will be able to provide some more insight

Acute Intermittent Porphyria (AIP)

At this point in time, there is only one treatment for individuals with severe AIP called Normosang, which is an IV therapy given every 2 weeks. An individual is only offered this therapy if they have frequent "attacks", which can happen in this condition. An "attack" may include severe abdominal and/or back pain, abnormally fast heartbeat, muscle weakness/ cramping/twitching, dark coloured urine, constipation, nausea/vomiting, insomnia and/or anxiety. Dr. L. Gouya and colleagues in France are studying the use of another therapy that was shown to prevent and treat acute attacks in mice. They are currently starting pre-clinical trials in humans with early results showing that it may be more effective than Normosang! Hopefully we will hear more about this new therapy soon!

Gaucher Disease

Gaucher disease is a lysosomal storage disease (LSD). It was the first LSD to have therapy , called enzyme replacement therapy, which is given through an IV. Early treatment in children with Gaucher has been shown to improve bone pain/crises, decrease the size of their liver and spleen and improve their platelet deficiency (which can cause increased bruising and bleeding). There is now a new form of treatment, called substrate reduction therapy, which has been shown to be effective in clinical trials. This treatment is currently only available to people with Gaucher disease enrolled in clinical trials, and there are certain criteria to be met before someone can be given this new therapy. What is the best news about this treatment? It is an oral therapy! No more IVs/infusions! We look forward to learning more about this therapy and if it will be available to more patients in the coming years.

Sincerely,
Melanie Napier, MSc, CGC, CCGC
Genetic Counsellor

1. Karaceper MD[1], Chakraborty P[2], Coyle D[3], Wilson K[4,5], Kronick JB[6], Hawken S[7], Davies C[8], Brownell M[9], Dodds L[10], Feigenbaum A[11], Fell DB[12], Grosse SD[13], Guttmann A[14,15,16], Laberge AM[17], Mhanni A[18], Miller FA[19], Mitchell JJ[20], Nakhla M[21], **Prasad C**[22], Rockman-Greenberg C[23], Sparkes R[24], Wilson BJ[25], Potter BK[26]; Canadian Inherited Metabolic Diseases Research Network. The health system impact of false positive newborn screening results for medium-chain acyl-CoA dehydrogenase deficiency: a cohort study.Orphanet J Rare Dis. 2016 Feb 3;11(1):12. doi: 10.1186/s13023-016-0391-5.
2. Gannavarapu S[1], **Prasad C**[2], DiRaimo J[1], Napier M[1], Goobie S[1], Potter M[3], Chakraborty P[4], Karaceper M[4], Munoz T[5], Schulze A[6], MacKenzie J[7], Li L[8],Geraghty MT[9], Al-Dirbashi OY[9], Rupar CA[10]. Biotinidase deficiency: Spectrum of molecular, enzymatic and clinical information from newborn screening Ontario, Canada (2007-2014). Mol Genet Metab. 2015 Nov;116 (3):146-51. doi: 10.1016/j.ymgme.2015.08.010. Epub 2015 Aug 31.
3. Farhan SM[1], Wang J[2], Robinson JF[2], Prasad AN[3], **Rupar CA**[4], Siu VM[4]; FORGE Canada Consortium, Hegele RA[1]. Old gene, new phenotype: mutations in heparan sulfate synthesis enzyme, EXT2 leads to seizure and developmental disorder, no exostoses. J Med Genet. 2015 Oct;52(10):666-75. doi: 10.1136/jmedgenet-2015-103279. Epub 2015 Aug 5.

Presentations: Invited faculty Dr. Chitra Prasad
The Department of Pediatrics, Seth GS Medical College & KEM Hospital, Mumbai, workshop, *'Cracking the code of genetics: a case-based workshop on genetics & metabolic diseases*' on 16th October, 2015.

Lectures:
1. No sweat! Hypoglycemia simplified
2. Neonate with cholestasis: Clearing the grey zones
3. Learning through cases (panel discussion)

Dr. Prasad recently attended the advisory board on hypophosphatasia (organized by Alexion), a rare metabolic condition involving the bones. Enzyme replacement therapy has been made available for a couple of years on research basis and now will be available on clinical basis.

Our Young Stars

"Hello, My name is Eryka Barnes. I'm six years old and have PKU. Last spring during our track and field races I came in first place! I usually tell people that it's my shoes that make me run super fast!"

Jasper is celebrating his 25th "at home" enzyme replacement infusion

Art of Extreme Care

- Browse your favourite bookstore (or music store)
- Write thank you notes to those who have touched your life and inspired you
- Practice random acts of kindness
- Admire beautiful artwork
- Let go of belongings you no longer love or use

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.

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Thank you!

Abby W
Age 7

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