The Inherited Metabolic Disorders News

Winter 2005 Volume 2 Issue 1

In this Issue

From the Editor

Spring is just around the corner (we hope!) and our Metabolic Family Workshop Day (formerly PKU and Related Disorders Day) plans are now underway. This year it will be held on **Friday May 13th**. Along with this issue of the newsletter I have included a tentative schedule for the day with a form that I ask you return to me so we can get an idea of how many people will be attending.

This year there will be activities planned for the children during the sessions that will be supervised by volunteers.

I am pleased to present our first personal story on page 2. I hope to receive more for the next issue!

Janice Little

From Dr. Chitra Prasad

Dear all,

Greetings! Lots of wonderful things have been happening at the clinic. The Lab Test Centre for children will finally move closer to the clinic, thus making it so much easier for families to get their blood work drawn.

The Metabolic Family Workshop Day is on Friday May 13th and I would like to extend invitation on behalf of our metabolic team to all of you to come and be a part of this day.

This year we have tried to hold a concurrent session to involve our families who have disorders other than PKU. There will also be one concurrent session on PKU. We have great speakers who have agreed to come and participate. Children will also have activities organized for them with the help of volunteers. I'm sure it will be a great fun-filled day!

My sincere thanks to our planning committee- the metabolic team and Jennifer Culp (Daniel's mom), Darlene Elliott (Olivia's mom), Trudy Ridout (Laura's mom) and Sam Smith (Jordynn's mom).

Please send in your suggestions and contributions for our newsletter.

With best wishes

Dr. Chitra Prasad

"Dare to dream great dreams"

Personal Stories

Ewa's Story *as told by her mother, Teresa Kowalczyk, to Laura and Trudy Ridout*
January 2, 2005

Ewa was born to the Kowalczyks on Saturday, September 27, 1975 in Olawa, Poland. Olawa is a small town, about 600 km east of Warsaw. Ewa was a healthy, beautiful baby at birth, with a weight of 3.5 kg and a length of 53 cm. The birth was uneventful, except that the umbilical cord was wrapped around her neck, and it had to be removed in the usual manner. Teresa and Ewa returned home from the hospital the following Monday, and the family resumed their new life together with their daughter. As was the usual practice at the time in Poland, Ewa did not receive testing for PKU.

When Ewa was 7 months old, her parents were uneasy about her development, and noticed that Ewa's muscles seemed unusually tight, and she suffered from muscular tremors. They took Ewa to the doctor, and the doctor reported that everything was fine. He felt that her difficulties were due to some trauma at birth. The Kowalczyks were not satisfied with the doctor's report, so in time, they took her to see many different doctors, none of whom gave her parents any more information than did the first doctor.

The muscular problems with Ewa persisted, as did the conviction of her parents to find out the root of the problem. At this time, Ewa was about 8 months old, and she was being fed a normal diet that was appropriate to feed a baby without PKU. Finally the parents heard about a special children's clinic in Wroclaw and they were able to get their daughter admitted there. Ewa had to stay at the clinic for 3 months, where she was tested for a variety of conditions. Teresa made the 60 km round trip to be with her daughter every day. At the end of her 2-month clinical stay, her parents were advised by the doctors to massage her muscles daily.

Although the Kowalczyks were not satisfied by the answers given by the doctors, they were not in a position to question the doctor's authority. At the time, Poland was under Communist rule, and the rights of the average citizen were very restrictive. The health system was a two-tiered system for example. There was one system of medical care for the average citizen, and another system with special doctors and hospitals for the "elite". Night curfews were also enforced. Everyone had to be in their homes by 8 pm, and the streets were patrolled by the police.

Finally, when Ewa was four and a half years old, the Kowalczyks were able to have their daughter assessed by a child psychologist at the Institute of the Mother and Child in Warsaw. Brain scans of Ewa were done, and it was discovered that the patterns present were not normal. The child psychologist suspected that Ewa had PKU, and upon administering the tests, it was concluded that this was the case.

The Kowalczyks were told that Ewa had to be on a special diet that excluded meat, and that it was basically "too late" for Ewa. They were advised to put Ewa in an

institution and forget about her, and proceed to have another child; maybe that child would not have PKU. This advice did not agree with the Kowalczyks.

By word of mouth, they came into contact with another mother of a child with PKU, and through her they connected with a PKU support group that the parents had formed on their own. The group consisted of about 40 parents and 20 children from all over Poland. Through this support group, Teresa found out more information about the diet that Ewa would need, and that she needed to be in touch with a specialist and a dietician. They also learned that they would need a special type of formula for Ewa. The support group also wrote letters to the newspapers in Poland, to educate prospective parents about PKU. Eventually they were connected with a private doctor who had done his studies in the United States, and his advice to them was "If you want to help your daughter, you must leave the country." Finally they had some advice from a doctor that they felt comfortable taking.

They applied to the Polish government for a passport enabling them to leave, but they were refused. Mr. Kowalczyk was allowed to come to visit Canada, and he stayed with his godmother who lived in London. While here, he visited Victoria Hospital, where he was able to purchase some PKU food and he sent it home. In the mean time, Ewa's regular diet consisted of potatoes, some vegetables, and at Christmas the family was allowed to purchase 1 kg of oranges. Vegetables were difficult to obtain during the winter, so Teresa canned them in the summer to make sure that Ewa would have enough to eat in the winter.

The Kowalczyks persisted in trying to obtain passports to leave, and they were finally advised to get a letter from the doctor to explain Ewa's condition. They went to their doctor where they begged and pleaded for a letter. The doctor was in a difficult position, because if he did this, he would go to jail. He said that he could get them another 25 extra milligrams of normal white rice. At the same time, a PKU formula was introduced into Poland. It was brown like coffee, and the smell and taste were very bad.

At this point, Ewa was in terrible shape, and she was having seizures on a regular basis. She was not admitted into school, because the schools did not feel that they could meet her needs, although school attendance was mandatory for all children. She was finally accepted at a boarding school adjoining the original clinic that she went to when she was very young. This was run by nuns, and there she started her formal education at the age of nine years.

Finally they met a police officer who said that he would help them to get passports to leave Poland. This was done for a considerable sum of money of course. Mr. Kowalczyk was given an International passport, and Teresa and Ewa were given European passports only. Only their immediate families were aware of their intentions, and they told friends and neighbors that they were leaving to go on vacation. It was August, so it made perfect sense.

The Kowalczyks arrived in Germany with only their personal suitcases, and the clothes on their backs. In Germany, they found that the people were very helpful and hospitable, but they were faced with the task of learning German, and finding

employment. For the first two weeks, they stayed at an old age home, and then they were given an apartment by the German government. A priest from the Polish parish quickly put them in touch with a couple of whom the lady was Polish, and the man was a doctor. Help was on the way for Ewa! Although they enjoyed their time in Germany, their dream was to come to Canada.

In 1987, they applied to the Canadian Embassy in Bonn, and on January 26, 1988, they arrived in Canada when Ewa was 13 years old. The family moved to London, and Ewa was put under the care of Dr. Clarson and dietician, Nancy Strange. Recently, like all of the PKU patients in the London area, she was transferred to the care of Dr. Prasad and dietician Suzanne Ratko.

Presently, Ewa is doing well on her diet. She is happy and active. She graduated from St. Pius school, and she currently attends Hutton House three times per week, she volunteers in the Sherwood Forest Branch Library, and she attends "Night Life" on Thursdays to socialize with her friends.

In Teresa's opinion, Canada is like paradise compared to the world that they left behind in Poland. She is very grateful for the medical system in Canada, and to the doctors, the dieticians and the personnel that have helped to care for Ewa.

Family-Friendly PKU Recipes: Deliciously Simple Recipe Ideas for People with PKU and their Families

The Mid-Altantic Connection for PKU and Allied Disorders Inc.
www.macpad.org
email: info@macpad.org
tel: 717 872 7546

Creative Family Cooking: Recipes & Menu Planning Ideas for PKU

The Mid-Altantic Connection for PKU and Allied Disorders Inc.
www.macpad.org
email: info@macpad.org
tel: 717 872 7546

Kids Korner

January, February, March birthdays:

Edward	Transcobalamin Deficiency	March 6, 2001	4 years old
Devin	PKU	March 7, 1987	18 years old
Jesse	PKU	January 5, 2000	5 years old
Abrahem	PKU	March 31, 1993	12 years old
Megan	PKU	March 31, 1998	7 years old
Seoyoung	Glycogen Storage Disease	January 31, 1997	8 years old
Kody	Gaucher's	January 3, 1991	14 years old
William	PKU	February 26, 1997	8 years old

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

APPLE PORRIDGE

Chef David F Dawson
Johnson & Wales University – Denver, Co

Amount	*Ingredient*
8 oz	Water
¼ cup	Low Protein Porridge
¼ tsp	Salt
1 tsp	White sugar
3 tbsp	Apple butter
1 tsp	Butter
2 tbsp	Non-dairy creamer
dash	Cinnamon

Preparation

1. Bring water to a boil, remove from heat
2. Slowly add porridge, whisking to avoid lumps
3. Simmer for 30 seconds and add remaining ingredients
4. Serve warm

May add dried fruits like cranberries and substitute brown sugar for white sugar.

Servings	Total Yield/weight (g)	Total recipe		
		Phe (mg)	Protein (g)	Phe g/weight
4	475	9	0.8	0.02

What's New

Metabolic Family Workshop
Friday May 13th, 2005
1130 am- 400 pm
Place: London Health Sciences Centre
7th Floor Amphitheatre
800 Commissioners Road East
London, Ontario

Lunch will be provided

Please RSVP by returning the bottom of the tentative schedule (pink sheet enclosed with envelope) ASAP

Cooking Demonstration

I had the opportunity to participate in a great hands-on cooking demonstration in Ottawa. Malathy from Taste Connections showed everyone how to use her product and SHS sponsored the event. It was a one-day event that was well worth the trip. We made pizza crusts, buns, bread as well as several different cookies, vegetable nuggets and a squash filled with lots of wonderful vegetables. What I enjoyed as much as the cooking was meeting other families that were going through the same things as we were. Talking about how they manage food and formula along with siblings, school and family events was a great relief. You realize that you are not alone and that others manage this 'diet' the same as we do. It was also great to meet some adults with PKU that have been through this entire process and how they manage day to day. I got some great tips on things to do with Daniel now so that when he gets older he will take ownership of his PKU rather than me doing for him. I look forward to helping organize a demonstration for our area in the spring.

Jennifer Culp
Mom to Daniel and Celeste

Christmas Party

On a cool afternoon in November we had our 1st Annual PKU and Related Disorders Christmas Party. With a little help from our friends at Labatt's we had a great room and place for the event. We had 24 people attending (including adults, kids and extended family), which was a great turnout for our first time. We had a potluck meal with lots of low protein foods and snacks for everyone to enjoy. The parents got a chance to mingle and meet a few other families as the kids watched the movie and meet a few new friends. We shared recipes and just had an opportunity to get to know each other a little better. After eating we headed down to watch the Santa Claus Parade which all the children enjoyed. I look forward to other events to give parents and kids a chance to meet again.

Jennifer Culp
Mom to Daniel and Celeste

The Foran Family

The Davidson Family

Jennifer and Celeste Culp, Trudy Ridout, Ewa and Teresa Kowalczyk

AJ Foran, Mom and Dad Foran, Joe Davidson, Gerald Foran

Pictures of the Christmas party at Labatt's

Genzyme Presentation

Drs. Prasad and Jung accepting a cheque from Pavel Krysiak (Genzyme Canada)

The Metabolics program here at Children's received the distinction of being named a "Centre of Excellence" by Genzyme Canada. Genzyme identifies metabolic centres across Canada and provides funding for education and other initiatives. We thank Genzyme for its continued support.

In the Next Issue

An explanation of metabolic disorders

Featured This Month

Dr. Tony Rupar PhD and Dr. Chitra Prasad MD

Cole's Story

Cole Lucas is now a 7 ½ year old boy with a lot of spirit and determination. He has **arginase deficiency**. Most people have not heard of this disorder. Cole was born at the right time with no problems during pregnancy or after birth. During the first year itself parents had noticed motor delays. He was provided with different forms of services from the CPRI. Unfortunately he also developed seizures through this period. He was diagnosed as having diplegic cerebral palsy (involving the lower legs). Developmental delays were noted. At about 4 years and 8 months he was seen by the metabolic service when analysis of plasma amino acids was carried out. This showed clearly that he had this rare metabolic condition called the arginase deficiency. Further studies have also identified the exact mutation in the DNA. Cole remains on a restricted protein diet with the help of Suzanne Ratko (Dietitian). He is learning new tricks all the time and is also beginning to walk with the help of walker! His family and all of us at the metabolic clinic are extremely proud of his achievements!

Arginase Deficiency

- Arginase deficiency is one of the least common of the urea cycle disorders. This entity also presents somewhat differently from the other urea cycle disorders. The disease is caused by deficiency of enzyme arginase type I in the liver.

- Arginase is one of six enzymes that play a role in the breakdown and removal of nitrogen from the body by a process known as the urea cycle, which is the major route for waste nitrogen disposal. The lack of the arginase enzyme results in excessive accumulation of nitrogen, in the form of ammonia (hyperammonemia), in the blood and arginine (hyperarginemia) in the blood and cerebrospinal fluid. The reaction normally mediated by arginase is the final step in the urea cycle, which liberates urea with regeneration of ornithine (Fig 1). The severe hyperammonemia observed in other urea cycle defects rarely is observed in arginase deficiency. As an autosomal recessive trait, the disease affects each gender equally. Parents have 1/4 risk of having an affected child with each conception.

- Affected infants and children may exhibit mental retardation, seizures, and spasticity (difficulty in walking). As an inherited disorder, the age of onset is typically during the neonatal period. Because of its atypical form of presentation, most cases may be easily missed in the neonatal period and only recognized in later infancy or early childhood. Some cases likely go undiagnosed with clinical symptoms attributed to “cerebral palsy”.

- Diagnosis is made by special blood tests called plasma amino acids. Enzyme level can be measured in the blood however specialized laboratory help is required for this. In some situations DNA mutation analysis is also available. Prenatal diagnosis is possible based on DNA mutation analysis.

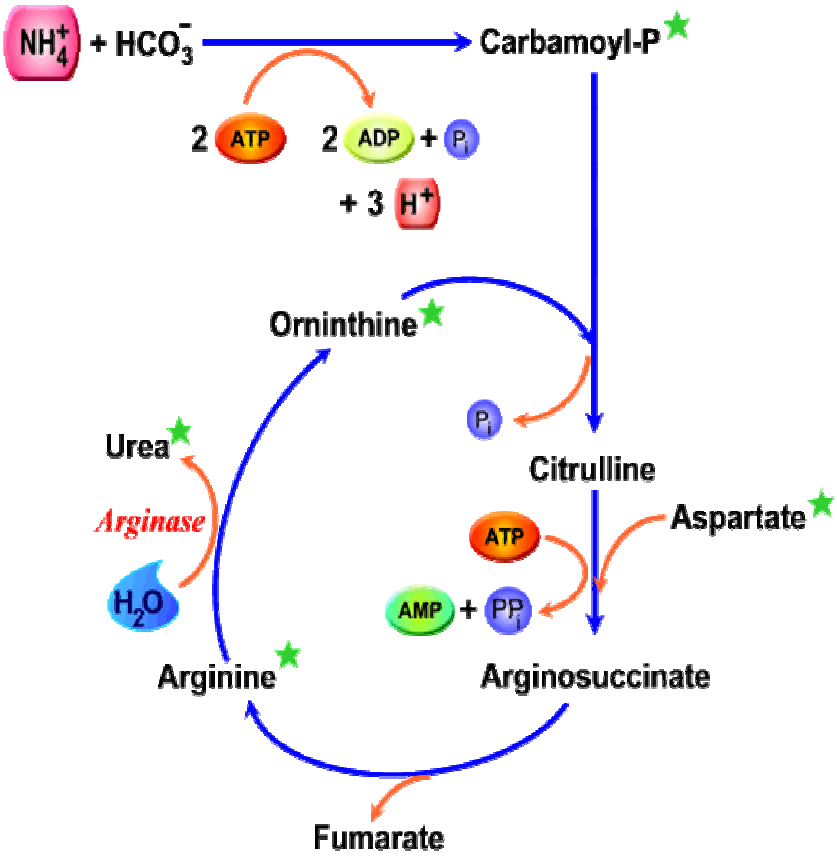
Management of Arginase Deficiency

- Reduce risk of hyperammonemia
- Normalize plasma arginine levels
- Genetic counselling
- Low protein diet
- Drugs: Sodium phenyl butyrate/ benzoate to divert nitrogen from urea cycle

Prognosis is guarded as most cases are unfortunately diagnosed late. Awareness about arginase deficiency and other such metabolic disorders is essential to help the

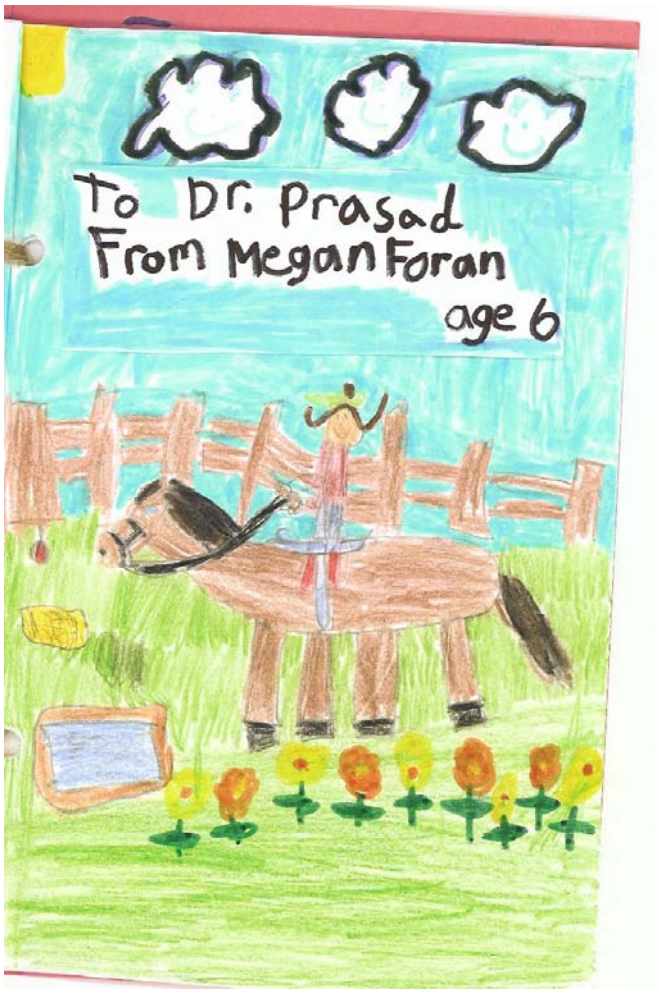
affected children and families by making earlier diagnosis and helping with the management.

References:
Emedicine article on Arginase deficiency by Karl Roth MD
OMIM (Online inheritance in Man)
http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=207800



Urea Cycle
(Figure 1)

Urea Cycle diagram obtained from:
www.lander.edu/flux/ 301_aminoacid_catabolism.htm



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/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