# PKU & Related Disorders Newsletter

Fall 2004 Volume 1 Issue 1

*In this Issue*



## From the Editor

Welcome to the first issue of our newsletter! We encourage your help and suggestions. If you have a question to ask, medical or practical, please let us know so we can find and publish answers to them under the "Q & A" section. We would love to hear from you!

Back in July a survey was sent to 40 metabolic families. Ten completed surveys were returned. If you did not receive a copy of the survey and would like to participate I would be happy to send you one. Please contact me at 519.685.8453 or toll-free at 1.800.243.8416.

So far families that have expressed an interest in forming a support group have PKU (6 families), Arginase deficiency (1 family), and MTHFR (1 family).

*From Dr. Prasad, Director of the Metabolic Clinic:*

Dear All,
It gives me great pleasure to see our first PKU and related disorders newsletter up and running.
The idea of having a strong support group is to form a strong circle of friends. There are a number of issues that can only be addressed when families, patients and the professionals work together.

We will try and keep you informed on all the latest developments regarding our program through this newsletter and our website. A project like this doesn't come about without dedication and committment of a number of individuals who work behind the scenes. I would like to express my sincere thanks to Janice Little (Resource Associate Provincial IODE Genetics) for her help with this newsletter and the website. I am also very grateful to all the members of our metabolic team.

I would love to hear from you and my little (and big) friends!

Best wishes for the Christmas/Hannukah/ Id/Diwali/New year!

I would like to end my note with this quote from Mahatma Gandhi (A great soul)

***"The simplest changes are often the hardest to make; we spend too much energy believing in things and not enough in ourselves. Believe in yourself! Live your dreams! Be the change you want to see in the world."***

Chitra Prasad

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**The Metabolic Team**

Front row from left - Dr. Tony Rupar (Biochemical Geneticist), Dr. Jack Jung (Geneticist, Director of Genetics), Dr Chitra Prasad (Director of the Metabolic Team). Middle row -Dr. Beverly Antle (Guest Speaker), Kathy Baker, Jo-Ann Kane, Robin Dyson (Administrative Associate), Susan Conacher, Sue Taber, Angela Kerr. Back row- Joanne Weir (Administrative Associate), Suzanne Ratko (Registered Dietitian), Kathy Corley (Professional Practice Leader, Metabolic Coordinator), Carla Campagnolo, Janice Little (Genetics Resource Associate) Dr. Victoria Siu, (Geneticist), Brian Seeley (Social Worker)

**Resources**

- OMIM (Online Mendelian Inheritance in Man)
http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM

- NORD (National Organization for Rare Disorders)
http://www.rarediseases.org/

- CLIMB (Children Living with Inherited Metabolic Diseases)
http://www.climb.org.uk/

- National Coalition for PKU & Allied Disorders
http://www.pku-allieddisorders.org/

- PAHdb (Phenylalanine Hydroxylase Locus Knowledgebase)
http://www.pahdb.mcgill.ca/

- Organic Acidemia Association
http://www.oaanews.org/

- FOD (Fatty Oxidation Disorders Family Support Group)
http://www.fodsupport.org/

- National PKU News
http://www.pkunews.org/

**Kids Korner**

October, November and December birthdays:

| | | | |
|---|---|---|---|
| MIchael | PKU | October 12, 2001 | 3 years old |
| Mitchell | PKU | November 19, 1990 | 14 years old |
| Emma | PKU | November 2, 2000 | 4 years old |
| Adam | PKU | November 2, 1998 | 6 years old |
| Cole | Arginase deficiency | November 1, 1997 | 7 years old |
| Raphael | PKU | October 23, 1999 | 5 years old |
| Laura | PKU | December 29, 1993 | 11 years old |
| Vanessa | PKU | October 6, 2003 | 1 year old |

**How to Make a Donation**

Donated funds are used for future PKU (and related disorders) 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

Your donation is tax deductible, and an income tax receipt will be mailed to you.

***Thank you!***

**Suzanne's Corner**

Suzanne Ratko
Registered Dietitian

# SPANISH RICE

Peter Henkel, Culinary Instructor
Johnson & Wales – College of Culinary Arts

Serves 6 – 8
Total Recipe provides: 241 mg Phe & 7.2 g Protein

INGREDIENTS

| | |
|---|---|
| 1 tsp | Oil |
| 40 grams (1/4 cup) | Low Protein Rice (Cambrooke Original Rice) |
| ½ cup | Water |
| 30 grams | Fresh Tomato, diced |
| 1/8 tsp | Ground Cumin |
| ½ packet | GW Washington Broth Powder |

1. Heat oil in small sauce pan, add imitation rice
2. Cook over medium heat until rice browns
3. Add rest of ingredients & bring to a boil
4. Reduce heat & cover pan
5. Let simmer over low heat until all liquid has been absorbed

Enjoy this wonderfully delicious rice dish!

**What's New**

Trudy Ridout and her daughter Laura, one of our PKU children, recently spoke to Laura's grade 5 class about PKU. The children were very receptive about the information they were given and asked thoughtful and relevant questions. We at the metabolic clinic are very proud of Laura!

Every year her school raises money for a charitable cause by holding a walk-a-thon. The principal of Laura's school is giving consideration for the Metabolic Clinic at LHSC as next years recipient for the funds raised in their walk-a-thon. How exciting!

***Christmas Party***: Jennifer Culp, our parent support contact, is organizing a Christmas party for November 27th. Here are the details:

Everyone will meet at the Amber Lounge (Conference Room at Labatt's) at 3:30 PM. A Christmas movie will be put on the big screen from 4:00 to 5:00 for the kids giving time for parents to visit.

Potluck Supper (of finger food) around 5:00 PM

Around 5:30 to 5:45 people could walk three blocks to downtown London to watch the Santa Claus Parade that begins at 6:00 PM. People may want to walk downtown earlier to get a good spot but the earlier you go the longer it is on the kids to sit and wait, especially if it is a cold night.

Please RSVP to Jennifer if you are planning to attend by calling her at 519.632.9924 or by emailing her at- donjen2000@hotmail.com.

**In the Next Issue**

Hopefully for the winter issue we will have some family stories, questions for the Q & A area, new resources to share, pictures (including the up and coming Christmas party), and Arginase deficiency in the “Featured This Month” area. Future research studies as well as the planning process will also be discussed in the next issue.

**Featured this month**

*What is PKU* by Virginia E. Schuett, MS, RD, Director, National PKU News

PKU (phenylketonuria), in its "classic" form, is a rare, inherited metabolic disease that results in mental retardation and other neurological problems when treatment is not started within the first few weeks of life. When a very strict diet is begun early and well-maintained, affected children can expect normal development and a normal life span. (Not all elevations of blood phenylalanine require treatment; any child with a level less than 360 umol/L (6 mg/dL) does not need to be on a special diet and is not risk for mental retardation.)

*The Enzyme Deficiency*
The disease arises from the absence of a single enzyme (phenylalanine hydroxylase). This enzyme normally converts the essential amino acid, phenylalanine, to another amino acid, tyrosine. Failure of the conversion to take place results in a buildup of phenylalanine. Through a mechanism that is not well understood, the excess phenylalanine is toxic to the central nervous system and causes the severe problems normally associated with PKU. Not every child has the same degree of enzyme deficiency, however; some have enough enzyme activity that the diet can be quite liberal, while others must have the very strict diet. The nature of the diet for an individual child must be determined by an experienced PKU treatment program.

*Genetics*
PKU is carried through an "autosomal recessive" gene. This means that two people who conceive a child must both be "silent carriers" of the gene in order for there to be a chance that the baby will have PKU. When two carriers conceive a child, there is a one in four (or 25%) chance for each pregnancy that the baby will have PKU. The incidence of carriers in the general population is approximately one in fifty people, but the chance that two carriers will mate is only one in 2500. Carrier tests are available only through PKU treatment programs.

*Screening*
Because of the very positive outcome when children are treated early and well, newborn screening for PKU is carried out in every state in the US and in many other countries.

Such screening started in the mid-1960's. Although PKU affects only one out of every approximately 10,000 babies born in the US, there are several hundred babies diagnosed and put on the diet each year. Children with the severe form of PKU once were destined to become mentally retarded and spend their lives in institutions. Children with the disease are now growing up normally. They are attending college and becoming productive adults as doctors, lawyers, teachers and engineers because of early diagnosis and strict adherence to the diet.

*Diet Composition*

The diet for the most severe form of PKU eliminates all of the very high protein foods since all protein contains phenylalanine. This means that all concentrated sources of protein must be eliminated from the diet in order to limit the amount of phenylalanine.

Except in rare circumstances, the diet does not allow consumption of meat, fish, poultry, milk, eggs, cheese, ice cream, legumes, nuts, or many products containing regular flour. A synthetic formula is used as a nutritional substitute for the eliminated foods. This formula is very expensive. Fortunately, a number of states have mandated insurance coverage for the cost, or in some instances the state health department may provide the formula to families of affected children. The diet is supplemented with special low protein foods and weighed or measured amounts of fruits, vegetables and some grain products. These foods are allowed in quantities that suit the individual child's tolerance or phenylalanine; some can have fairly liberal diets and still maintain good control of blood phe, while others must have a very strict diet. Some states now have laws mandating insurance coverage of the expensive special low protein foods.

*Diet Maintenance*

In the early days of treating PKU, clinicians believed that the diet could be stopped at an early age (6-10 years). However, it is now known that stopping the diet can result in a variety of serious problems. These include drops in IQ, learning disabilities, behavior problems such as hyperactivity and irritability, neurological problems such as tremors, eczema (a skin disorder) and personality disorders (including schizophrenia, panic attacks and agoraphobia). s a result of the problems that have developed in young people who have discontinued the diet, it is believed that the diet should be maintained for a lifetime. It is also now generally believed that keeping blood phenylalanine levels in the range of 120-360 umol/L (2-6 mg/dL) is the safest, especially in infancy and early childhood. Frequent blood monitoring to achieve this goal is critical. All individuals, male or female, who have stopped the diet would be wise to return to the diet under the supervision of a PKU treatment program.

*Maternal PKU*

Returning to the diet is especially important for young women with PKU who want to become pregnant. Women with PKU who have high levels of phenylalanine in their blood have a very large probability of harming their unborn baby. "Maternal PKU" has become a significant concern as young women who were once taken off the diet are

now reaching childbearing ages in ever-increasing numbers. A large collaborative study has shown that returning the woman to diet before conception and keeping blood phenylalanine levels below 360 umol/L (6 mg/dL) results in the best outcome for the baby. Many children born to mothers with PKU are developing normally because of early and strict treatment.

*PKU Treatment Programs*

There is at least one specialized PKU treatment program in most US states. In some less-populated states, families may need to travel to another nearby state, or the treatment may be coordinated through a state health department. All children with PKU ideally should be followed by a specialized PKU program with adequate monitoring facilities. Information about the nearest PKU treatment program can be obtained through the state newborn screening program.

*"Please go buy batteries for the remote so Timmy can do his homework."*

*Contact Information*

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Post Message: metabolic_disorders@yahoogroups.com

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