



Illinois PKU Press
Fall 2014



Family FunDay Group Photo: May 10, 2014

Recap: Family FunDay

Saturday, May 10th, 2014 we gathered together at Cosley Zoo in Wheaton, Illinois. The day started at 10 am with families registering. After registration, families were able to meet different vendors that serve the low protein metabolic community. While the parents were busy with vendors, children were able to take part in various games, face-painting, temporary tattoos, arts & crafts, and other activities.



Children had fun creating their own sticker farms.



Families enjoying getting to pet and learn about the various pets that live at the zoo.

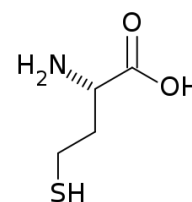
About an hour in, our walk started and we toured the different parts of the zoo. When the walk concluded we all enjoyed a catered lunch from Caliendo's Restaurant and Cambrooke Foods. After lunch, one of the zoo keepers brought out a few different animals that we were able to enjoy petting and learning about.

IN THIS ISSUE



Annual Meeting Highlighted Speaker:
Dr. Kristen Skvorak

Come see the advances Dr. Kristen Skvorak is making in stem cell research in relationship to PKU and the Allied Disorders community.



Who's Who in PKU and Allied Disorders:
Spotlight on:
Homocystinuria



Recipes from the Chef: Eric Sartain



Roasted Peaches with Vanilla Spice and Honey



Ingredients:

Peaches 4-5 ripe
Cinnamon Sticks 2
Star Anise 2
Vanilla Bean 1
Honey 3-4 tbsp.
Peach Schnapps or Brandy (optional) 1oz

Method:

- 1) Preheat oven to 375 degrees.
- 2) Cut peaches in half and pull out the pits and then slice into wedges.
- 3) Then add spices, honey, vanilla bean and liqueur in a 9x9 pan.
- 4) Bake for 10-20 minutes until tender in the middle.
- 5) Serve with So Delicious Coconut Yogurt, or your favorite sorbet.

Serves 4- 100g serving
20 PHE per serving

Persian Style Onion Soup:

Ingredients:

Olive Oil 3 Tbsp. divided
Spanish Onions 5 large sliced
Dried Mint ½ Tsp
Baking Mix 2 Tbsp
Vegetable Stock 3 Cups
Cinnamon Stick 1
Turmeric ½ Tsp
Juice of Lemon 1
Sugar 1 Tsp
Salt and Pepper to taste



Method:

- 1) Heat 2 Tbsp. Olive Oil and onions, and cook for 12- 15 minutes covered in a 4 QT. sauce pan.
- 2) Next add dried mint, 1 Tbsp. olive oil and stir in the baking mix. Cook, stirring frequently for 3-4 minutes.
- 3) Gradually stir in stock. Whisk, making sure to prevent lumps. Add spices and cinnamon stick.
- 4) Simmer over a low heat partially covered for 30-40 minutes. Add lemon juice and salt and pepper to taste.

Serves 4: about 1-1 ¼ Cup.
PHE: 72 per serving.



Back to School With: Jacob King



Marinated Vegetables:

Ingredients:

- Carrots Sticks
- Celery
- Radish
- Cauliflower
- Green Peppers
- Mushrooms



Method:

- 1) Choose any raw vegetables from list.
- 2) Cut decoratively or plan match sticks size.
- 3) Place into individual zip lock bags and toss with Italian Dressing (no cheese variety).
- 4) Refrigerate overnight for best flavor.
- 5) Great for LOW PRO lunch Snack!

5 Sandwich Ideas

- Diced avocado, Miracle Whip and tomato
- Brown sugar and margarine
- Sliced cucumbers, onions and tomatoes
- Lettuce, onion, tomatoes and Miracle Whip
- Grilled mushroom, onions and tomatoes

Soup in a Minute

- 1/2 c dry low pro pasta noodle of your choice
- 1/3 c thinly slice or diced carrots
- 1 package Seasoning and broth mix or 1 tsp. of Wyler's bouillon granules
- 2 C. water
- 1tsp. of salt
- Dash of dried herbs of your choice

Directions:

1. Mix all the ingredients together including uncooked pasta, in a medium saucepan.
2. Cover and bring to a boil over high heat
3. Turn heat down the to low, and cover and let simmer 12 minutes until fork tender

Yummy PKU Trail Mix

Ingredients:

- Low protein pretzel circles (suggestion Wyldes brand)
- Raisins or Craisins (or other dried fruit)
- Chocolate chips or butterscotch chips
- Veggie straws –broken apart

In a sandwich bag combine 18-20 pretzel rounds, 1/8 cup of dried fruit, small handful of chocolate or butterscotch chips, and 5-6 veggie straws.

Serves: One

Approx.: PHE is 45mg per serving

Applesauce Muffins

Ingredients:

- 1c Wel- Plan baking mix
- 3 tablespoons granulated sugar
- 2 teaspoons baking powder
- 1/2 teaspoon baking soda
- 1/2 teaspoon cinnamon
- 1/4 teaspoon salt
- 3 tablespoons Vegetables oil
- 1/3 cup water
- 1/3 cup applesauce



Method:

- 1) Preheat oven to 400*.
- 2) In a medium mixing bowl, stir together Baking Mix, sugar, Baking powder, baking soda, cinnamon, and salt.
- 3) Mix together oil, water, and applesauce in a liquid measuring cup: add all at once to dry ingredients and mix by hand, 20 to 30 seconds until smooth (do not use an electric mixer).
- 4) Spoon batter into six greased 2 1/2 – inch muffin cups (they will tend to stick to cupcake liners). Bake 15 to 18 minutes.

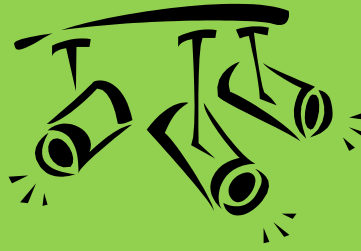
Yield: 6 muffins.

Per muffin PHE (mg) 1

Protein (gm) 0.1

Who's Who in PKU and Allied Disorders:

Spotlight on:



Homocystinuria

By: Danae' Austin & Matthew Bartke

Did you know the PKU Organization of Illinois serves more than just those with Phenylketonuria (PKU)? The PKU Organization of Illinois serves approximately 20 other allied disorders. PKU, along with the other allied disorders falls into one of three metabolic disorder categories: Amino Acid Disorders, Organic Acid Disorders, or Urea Cycle Disorders. All the disorders require life-long treatment, care and a special diet. Anyone who is affected by PKU or an allied disorder is more than welcome to come join the organization. We are always glad to see new faces. Our spotlight this time around is on Homocystinuria.

What is Homocystinuria?

Homocystinuria or HCU is a recessive genetic disorder, meaning both parents must be carriers of the gene to have a chance (25%) to pass it down to their children, which affects the metabolic system. The genetic defect most commonly causes a deficiency in the Cystathionine beta synthase (CBS) Gene, but can also be caused by mutations in the MTR, MTRR, MTHFR and MMADHC genes. The effect of this mutation is a buildup of Homocysteine in the blood stream which can cause numerous defects in the body. HCU Affects around 1 in 200,000 people worldwide.

Complications

The issue lies with the consumption of the essential amino acid, Methionine and protein intake must be watched. In many cases doctors tell one to limit their total protein as opposed to methionine itself, but those affected should take into consideration the methionine level of their foods. Mustard, for example, is labeled as 0g protein per 1tsp, but actually is high in methionine at 3.9mcg per tsp. If you increase the serving size to 1 cup, the protein is now 11g and methionine is at 194mg. So one should be aware, because it is often the case that a package may be labeled 0g of protein, yet still have some, but not enough where companies required to count it or

may be high in methionine. <http://nutritiondata.self.com> is a good resource for finding protein and methionine levels (including other amino acids) in foods. Remember to check multiple sources if there is a question because no one source is 100% accurate.

The amount with HCU varies, but for the sake of this article, 15-20g of protein is the upper limit for those that I know that have HCU as prescribed by their doctor while taking the prescribed medications, which I'll go over briefly later. If one's homocysteine level exceed the normal range (10 and lower), there is potential for numerous complications including, but not limited to: Blood clots, mental retardation, retinal detachment,

cataracts, seizures and vascular disease and osteoporosis to name a few. Those with HCU tend to have: flush cheeks, tall/thin builds, have knocked-knees and high-arched feet. It is highly-recommended that those with HCU stay on diets at all times because other than the accumulating effect of some symptoms like retinal detachment, there are quick-setting dangers like blood clots. Blood clots occur when homocysteine levels are high which causes "sticky platelets." This can result in the loss of a limb, a stroke (if in the brain), a heart attack and may even cause death.

Stay on Diet and Don't Forget To Ask

The first line of defense against HCU is your diet which consists of a primarily vegetarian diet with little to no dairy. There are low-protein foods available like low-protein breads and pastas. Also, look into the Gluten Free sections of stores. Gluten free foods tend to be lower in protein because wheat is often swapped with sugars, starches and fats. You can make your own low protein pastas and breads using Wheat Starch and Xanthan Gum is a good egg replacer, although you can also use a normal boxed egg-replacer, but Xanthan Gum is lower in protein. Wheat starch may be difficult to find in the stores, so if you can't find them there, it is available inexpensively at amazon.com.

It may be frustrating going out to eat with friends and family or on vacations, but there are things you can do. Don't be afraid to special order your food. Tell them that you can't have cheese in your dish or maybe they can take the beef patty off of that burger and replace it with something else, like extra tomatoes, a portobello mushroom or whatever they might have for other dishes. You can also bring your own item for them to put on the burger for you or your own low protein pasta for them to use for your dish. Most places will try to cater to you. If you're going somewhere like Disneyworld, call ahead. They already have people dedicated to accommodating those with special diets.

Common Medication, Supplements and Tips

The second line of defense is your medication. If your diet is bad, your medication won't help you much. With a diet of 15-20g of protein, you'll run into the issue of a protein deficiency, so you'll be required to supplement. VitaFlo's HCU Express is available to meet this need. It has all of the essential amino acids, plus assorted other nutrients that people on a low-protein diet tend to miss out on in one package. There are both liquid and powdered forms of this product. You will need to take more if you are active and/or workout than if you are sedentary. Contact your doctor if you are unsure of the minimum protein you need to consume per day. The powdered version of this product is quite bitter, so it is wise to mix it with something if you can't drink it mixed with water. One person I know mixes it with instant

if you can't drink it mixed with water. One person I know mixes it with instant pudding and eats it that way. Another one mixes it with a low-calorie powdered instant-beverage mix, in a shaker bottle with a blending ball to make it as smooth as possible. She'll combine her other powdered medications in it as well. A side-effect is that it tends to create a lot of gas. I have found that if you mix a quarter teaspoon of baking soda with a quarter cup of water and mix it up and drink it, this will sometimes immediately settle your stomach.

A lucky few with HCU are called B6 responders. These people can just take some Vitamin B6 and their homocysteine levels will be under control and they can eat standard diet at the same time.

The primary medication for the others is called Betaine Anhydrous (BA) AKA Trimethylglycine (TMG). You'll often find over-the-counter versions of this under the TMG name. BA is extracted from sugar beets and has been shown to significantly decrease plasma homocysteine levels. Doses vary, but a common dose is around 3g taken 3 times a day. You want to take this throughout the day like that because homocysteine levels vary throughout the day and this helps keep it low. BA will increase the remethylation of homocysteine and thus make up for low or no CBS Enzyme levels pretty well. It can be taken with water or mixed with your protein supplement, but is heat sensitive and should not be mixed in advance with any liquids. The fresher the mixing, the more affective it is. Also, Betaine Anhydrous has a 5 year shelf life when stored in the fridge. Adding

beats to your diet is a good idea since every 100g of beats has 256mg of Betaine Anhydrous.

Other commonly prescribed medications include Vitamin B12, B6 (as stated), Folic Acid (B9), Calcium/Magnesium/Zinc and Vitamin C. These are to fill in the gaps of the Remethylation Pathway because if there is a deficiency in any of these, homocysteine levels can and will dramatically rise. Calcium/Magnesium/Zinc is a combined supplement with three different products that work together to increase bone health. Contact a doctor for dosages and advice before taking any medication I mentioned. I am not a doctor or medical expert. Everyone is different and where a series of medications might be right for one person, it may have a detrimental effect on another.

Stay Active

There is also a third line of defense and that is exercise. Weight training and cardio are thought to bring plasma homocysteine levels down. If you exercise, make sure you are taking enough of your HCU Protein Supplement because your body's needs will be greater. Your muscles, especially when strength training, will need more protein to aid in the rebuilding of your muscles. Losing weight is good, but make sure you're mostly losing fat because if you lose weight too fast, you'll be burning off muscle, which is detrimental to your

health whether or not you have HCU or not, but is worse if you do because your muscles are made of protein and if your body catabolizes them too much it can actually raise your plasma homocysteine levels. The most you ever want to lose is 2lbs per week. If you lose more than that, you're losing a great amount of muscle which is not good for your homocysteine levels and it isn't good for your health. Another advantage of lifting heavy weights (around 5-8 reps per set) is that it will build up your joints and ligaments and strengthen your bones and since a person with HCU has an increased risk of osteoporosis, it is to their benefit to do what they can to preserve and increase bone density. Don't rely on medication to do this for you.

Remember to be proactive and take an active stance to minimize the effects that having Homocystinuria has on your life. If you stay on diet, take your HCU Protein supplements, take your medication and work out, you can live a happy and full life. HCU is a manageable genetic disorder and you should do all you can to stay in front of it.

**Do you have
an Allied
Disorder?**

**We'd like to
hear from
you!**

Please Contact:

pkullinois@gmail.com

“Great Pops of Fun!”



SEPTEMBER 13, 2014

LOW PROTEIN COOKING WITH TASTE CONNECTIONS

Cooking Workshop

The PKU Organization of Illinois, along with Nutricia North America would like to invite you for an exciting day of low protein fun. Malathy Ramanujam of Taste Connections will be demonstrating and leading us in a hands on workshop on how to make “Great Pops of Taste”. We will be using cake pop machines along with other equipment to make “meat” balls, pancake balls, cake balls and so many other delicious eats!



- **New tasty recipes**
- **Free raffle**
- **Free PKU and Non PKU Lunch**
- **Must register in advance.**
- **Only 35 spaces available!**
- **Ages 6 to Adult**
- **To Register-
Please contact:
Danae' Austin
(630) 546-6452
or
Danaeaustin@gmail.com**

95TH COMMUNITY CENTER

Culinary Arts Room 303

2244 West 95th St.
Naperville, Illinois

Registration 9:30AM
Workshop: 10A.M-2PM



**NEW
LOCATION**
2601 N.
MILWAUKEE
AVENUE
(LOGAN SQUARE)



WEBSTER'S WINE BAR

**WINE
TASTING**

**SATURDAY OCT 4,
2014**
5 TO 8:30 P.M.

**WINE TASTING AND
SILENT AUCTION
FUNDRAISER**

**WE ARE STILL IN NEED OF
AUCTION DONATIONS!**



**SAVE
THE
DATE**



Working Together for a Cure

2014 Annual Meeting

November 8, 2014

Skokie Holiday Inn

5300 W. Touhy Ave. Skokie. Illinois 60077

Meet Our Highlighted Speaker:



Kristen J. Skvorak, PhD

I received my BS in Biology from Lycoming College in 2002. While there, experiences with a Muscular Dystrophy support group first piqued my interest in inherited disease. I attended the University of Pittsburgh, focusing primarily on cell transplantation to treat inborn errors of metabolism (IEM), receiving my Ph.D. in 2008. My research now focuses exclusively on cell transplant for PKU. There is currently a clinical trial at University of Pittsburgh investigating cell transplantation for IEM.

Rather than transplant whole liver (expensive, invasive, organ shortage, lifelong immunosuppression), healthy cells could instead be infused into a patient's diseased liver providing the missing enzyme to improve symptoms. Initial studies with liver cells transplanted into a MSUD and PKU mouse model were very promising, and so investigation with human placental stem cells (plentiful, low immune response by host so may not require immunosuppression) began. Placental cell transplant in MSUD and PKU mice decreased amino acid toxicity in blood and brain and restored balance, and improved neurotransmitter levels. Our major project goal is to bring this therapy to reality clinically.

Breakout Sessions:

Our breakout sessions are led by RDs, Physicians, social work and PKU representatives. This year we are organizing our breakout sessions by age group.

Age Groups:

Early Childhood

Elementary

Middle School

High School

Adulthood

Allied Health Disorder

KUVAN[®]
(sapropterin dihydrochloride)
Powder for Oral Solution

KUVAN Powder—The convenience is

CLEAR.



KUVAN[®] (sapropterin dihydrochloride) Powder for Oral Solution provides a convenient option.

- Dissolves completely within 15 seconds
- Sweet taste with lower acidity
- Easy-to-carry packets



Every patient should be offered a trial of KUVAN except those with two null mutations in *trans*¹



Visit KUVAN.com to learn more about the benefits of KUVAN Powder.

Reference: 1. Vockley J, Andersson HC, Antshel KM, et al; for the American College of Medical Genetics and Genomics Therapeutic Committee. Phenylalanine hydroxylase deficiency: diagnosis and management guideline [published online ahead of print January 2, 2014]. *Genet Med*. doi:10.1038/gim.2013.157.

Indication

KUVAN® (sapropterin dihydrochloride) Tablets for Oral Use and Powder for Oral Solution are approved to reduce blood Phe levels in people with a certain type of Phenylketonuria (PKU). KUVAN is to be used with a Phe-restricted diet.

Important Safety Information

It is not possible to know if KUVAN will work for you without a trial of the medicine.

Your doctor will check your blood Phe levels when you start taking KUVAN to see if the medicine is working.

Starting KUVAN does not eliminate the need for ongoing dietary management. Any change to your diet may impact your blood Phe level. Follow your doctor's instructions carefully. Your doctor and dietitian will continue to monitor your diet and blood Phe levels throughout your treatment with KUVAN **to make sure your blood Phe levels are not too high or too low**. If you have a fever, or if you are sick, your Phe level may go up. Tell your doctor and dietitian as soon as possible so they can make any necessary changes to your treatment.

Children younger than 7 years old treated with KUVAN doses of 20 mg/kg per day are at an increased risk for low levels of blood Phe compared with children 7 years and older. Frequent blood monitoring is recommended in this population to ensure that blood Phe levels do not fall too low.

Tell your doctor if you have ever had liver or kidney problems, have poor nutrition or have a loss of appetite, are pregnant or plan to become pregnant, or are breastfeeding or plan to breastfeed.

KUVAN is a prescription medicine and should not be taken by people who are allergic to any of its ingredients. KUVAN and other medicines may interact with each other. Tell your doctor about **all the medicines you take**, including prescription and over-the-counter medicines, vitamins, herbal and dietary supplements.

If you forget to take your dose of KUVAN, take it as soon as you remember that day. Do not take 2 doses in a day. If you take too much KUVAN, call your doctor for advice.

The most common side effects reported when using KUVAN are headache, runny nose and nasal congestion, sore throat, diarrhea, vomiting, and cough. Additional adverse reactions reported in connection with worldwide marketing include sore throat, heartburn or pain in the esophagus, inflammation of the lining of the stomach, indigestion, stomach pain, and nausea. These are not all the possible side effects seen with KUVAN. Call your doctor for medical advice about side effects. You may report side effects to FDA at 1-800-FDA-1088.

KUVAN can cause serious side effects, including:

- **Severe allergic reactions.** Stop taking KUVAN and **get medical help right away** if you develop any of these symptoms of a severe allergic reaction:
 - Wheezing or trouble breathing
 - Nausea
 - Flushing
 - Lightheadedness or fainting
 - Coughing
 - Rash
- **Inflammation of the lining of the stomach (gastritis).** Gastritis can happen with KUVAN and may be severe. **Call your doctor right away if you have any:**
 - Blood in your vomit or stool
 - Severe upper stomach-area discomfort or pain
 - Nausea and vomiting
 - Black, tarry stools
- **Too much or constant activity (hyperactivity) can happen with KUVAN.** Tell your doctor if you have any signs of hyperactivity, including fidgeting, moving around or talking too much.

For more information, call BioMarin Patient and Physician Support (BPPS) at 1-866-906-6100.

Please read the attached full Patient Information.

Reference: 1. Vockley J, Andersson HC, Antshel KM, et al; for the American College of Medical Genetics and Genomics Therapeutic Committee. Phenylalanine hydroxylase deficiency: diagnosis and management guideline [published online ahead of print January 2, 2014]. *Genet Med*. doi: 10.1038/gim.2013.157.

BiOMARIN

BioMarin Pharmaceutical Inc.
Copyright 2014. All rights reserved.
USPKU0475ENJUN2014

KUVAN[®]
(sapropterin dihydrochloride)
Powder for Oral Solution

Dear Phebea,

When I go to clinic my dietitian is always asking for a diet record. I have a really hard time remembering to keep track of my diet. My parents used to keep diet records for me, but I am busy with school, sports and an after school job. My parents want me to learn how to do this on my own. Do you have any ideas or tips to help me keep track of my diet?

Thanks,

Daisy Dietrecordless

Hi Daisy,

That's a great question! It's something I hear from a lot of people that have PKU. Keeping a diet record is a very important part of your PKU treatment. I understand that this can be really hard to do, especially if you're busy. If this is something you have never done, start by keeping track of your diet before your blood draws; it's best if you do not change what you eat. Make sure to write down everything you eat, even if it is just a small snack, and measure the foods you eat with a scale or measuring cups. This helps your PKU team get a better idea of what you are actually eating. It also helps the team understand what you like to eat and can give you suggestions for new foods to try! Try to provide as much detail as you can, including the brand, how you prepared the food, and the PHE content if you know it. There are a lot of ways to write down your diet. You can keep track on a notepad. Some patients keep track on their computer. Other patients use applications on their phones. There are a few applications that are specific to PKU including AccuGo for PKU (\$0.99) and Dietwell for PKU (\$4.99). There is also a new website called howmuchphe.org. You can buy a membership to this website for \$45 a year. You can look up the phe content of a number of foods. It also has an application on your Iphone too. If you really struggle with writing down your diet record and remembering what you eat, try taking a picture of your plate before all your meals. You can look at the pictures to help you remember what you ate, or you can send the pictures to your dietitian. I know it's hard to keep track of your diet. But it is a very helpful skill to maintain good PHE levels, so that you can feel and think your best!

Phebea