

# PKU Press

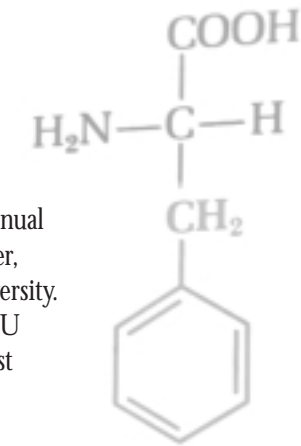
WINTER 2003

A Periodic Newsletter Published By The PKU Organization Of Illinois

NOVEMBER 30, 2002

DEAR MEMBERS,

The 33<sup>rd</sup> Annual Meeting of the PKU Organization of Illinois was a great success. We had a total of 132 attendees, including medical staff and speakers, and raised \$10,807 through our annual raffle. The centerpiece of meeting was an address by our keynote speaker, Dr. Charles Scriver, Alva Professor of Human Genetics at McGill University. Dr. Scriver covered the highlights from the June 2002 International PKU Meeting in Denmark and discussed educational topics of general interest to PKU individuals/families. His current research is focused on human genetic variations and new ways to treat genetic disease.



Phenylalanine

During 2002, the Organization sponsored a full calendar of events in addition to our annual meeting. Regular members and board members of the Organization hosted all of these events:

- *The New Parent Coffee* – hosted by Dan & Kem Baker.
- *Discovery Day* – hosted by Hazel Vespa & Rhoda Papnastassiou.
- *Cooking School* – hosted by Kimberly Gralen.
- *Cooking Days* – hosted by Dee Pratscher.
- *Southern Illinois Picnic* – hosted by Laurie Sprietsma.
- *Northern Illinois Winter Swim Party* – hosted by Patty Cosgrove.
- *Halloween Party* – hosted by Dee Pratscher.
- *PKU Family Camp* – hosted by Mary Jane and Bruce Kimbrell, Anne Kozek, & Michele Kowalski.

We hope that you were able to attend as many of these events as possible, if not this year, then perhaps next year.

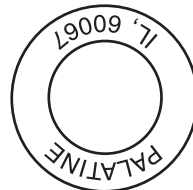
The board endeavors to keep in contact with the membership via variety of means such as our web page at [www.pkuil.org](http://www.pkuil.org) where a great deal of current information on the Organization's activities is kept along with registration forms and other related informational documents and applications. With the web page and our e-mail address at [info@pkuil.org](mailto:info@pkuil.org), the Organization is in contact with individuals and other organizations from around the world.

Sincerely,

THE OFFICERS AND DIRECTORS OF THE PKU ORGANIZATION OF ILLINOIS



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PKU ORGANIZATION OF ILLINOIS  
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The **PKU PRESS** is published three times a year as a service of the PKU Organization of Illinois. In conjunction with the three Illinois clinics, we are working together to provide information, offer support, highlight achievements, and provide services for the benefit of the PKU community. We welcome your input and ideas. Requests for additional copies or correspondence should be addressed to:

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#### 2003 PKU Organization of Illinois Officers

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Angela Koster - *Vice-President*              Gina Annunzio - *Secretary*

#### 2003 PKU Organization of Illinois Board Members

Patrice Bedenk, Larissa Bruno, Ruben Caro, Kate Chovanec,  
Bruce Kimbrell, James Leaf, Kristen Leaf, Jayne Markham

#### 2003 PKU Organization of Illinois Honorary Members

Anne Kozek, Hazel Vespa

# A TRIBUTE TO GRACE HERRON

I have been asked to pay tribute to Grace Herron, a retired Social Worker from the University of Illinois Hospital. For those of you who don't know me, I am Cindy Bouk, and I'm the mother of Karl Bouk, age 23, who has PKU. Among my first memories of PKU is the day we made that hectic trip to Chicago with a newborn, to a strange hospital, scared half out of our wits. With that memory comes the mental picture of meeting this friendly, quiet, reserved woman by the name of Grace who helped us through those first difficult hours, helping us come to terms with the diagnosis of PKU and the treatment.

Through the coming months and years, this quiet woman used her leadership skills to bring PKU parents together to help visualize and develop many PKU firsts. She was instrumental in locating and organizing the PKU camp, which has been going strong for over 20 years. Grace helped organize the first PKU picnic in Peoria, as well as promoting several Coffees and supporting the PKU Press. She made special foods and tested

new recipes. She devoted countless hours and traveled hundreds of miles to help with various PKU events, Board Meetings, and prior PKU annual meetings.

To me, and many others, Grace has been that non-judgmental person you could talk to about the challenges of PKU. She would help find the answers to your problems. Her visions have helped foster better education and communication, as well as growth for the PKU community.

With Grace's help, not only did the PKU community grow and develop, but so did many parents, myself included. Grace's quiet confidence made you think anything was possible. She had a way of getting people involved with PKU.

I can say that Grace was more than our Social Worker. She became a close friend of my family. For more years than I can remember, Grace would make the trip to Hudson for the PKU Camp or Picnic. We faced many challenges

together, each leaning on the other. Some years, we would both say that this is the LAST YEAR, just to be back the next year. Her love and concern for the PKU children and their families was evident in her tireless efforts over the past 20+ years.

In her retirement, Grace is keeping busy...she is still giving of herself. Today she is volunteering with a literacy program helping people learn to read.

What I thought would be a difficult writing task, became very easy when I started to remember all of the wonderful (and sometimes stressful) times I have shared with Grace through the years. So, I know that I am not alone when I say

**A BIG THANK YOU TO GRACE HERRON FOR BEING A FRIEND TO PKU AND THANKS FOR ALL OF HER TIME AND EFFORTS!**

## Christmas was in the Air at the Bedenk Home... *By Dee Pratscher*

Patrice and Christopher warmly opened their home for a Cookie Exchange in December. Our hosts welcomed us along with Christopher and Gillian their two PKU children, for an afternoon of friendship and comradery. Their home was decorated for the season, children dressed in the cutest Christmas outfits, and the table was set beautifully with an array of low protein bakery items, punch, and vegetables. Only three families attended however I think the intimacy of a small group added to the comfort of the day. We had an absolute blast. The kids ate, and ate, and ate cookies galore. And played so well together on their own it really left plenty of time for the moms and dads to visit, whom I should mention also played very nicely together. It's hard to say if the six kids had more fun or the six parents, perhaps too close to call. Patrice had organized a group craft for the kids to work on, which they did with enthusiasm. We exchanged cookies, enjoyed a cocktail and watched our children to bond. A gracious thank you to the Bedenk Family to sponsor this event and to Patrice for volunteering to served on the board this year, Good Luck!



## Cooking School 2002

By Angela Koster

Well, we have had another successful cooking school at the Gralen residence. Kimberly had all sorts of wonderful recipes and tips to share with the moms and grandmothers who attended. Some of the scrumptious recipes we had the privilege to taste-test included egg rolls, tortilla chips, bagels, stir-fried beans, cinnamon and sugar coated tortilla chips, muffins, donuts and ice cream. Everyone had the chance to get their hands on the bread dough to get the "feel" for working with it. It was really wonderful to realize just how easy it can be to prepare some of the foods. It was a great experience for me personally, being a new mom myself. We left with full stomachs and some fresh ideas to try at home – just in time for the holidays! A great time was had by all. Many thanks to Kimberly for all her generous time and efforts!

### SUMMARY OF THE QUESTION ANSWER SESSION WITH THE DOCTORS

By Kristen Leaf

At the annual PKU meeting on November 16, 2002, Drs. Charles Scriver, Paul Wong, George Hoganson and Barbara Burton were open to answering questions presented to them by meeting attendees. Dr. Burton opened the discussion by responding to an inquiry regarding the status of an ongoing research project at Children's Memorial Hospital to determine what patients with PKU respond to treatment with the drug tetrahydrobiopterin (BH4). Fourteen patients with both classical and atypical PKU were involved in the research thus far. The patients were placed on a relaxed diet and taken off of their medical formulas in order to allow their blood phe levels to rise. A single dose of BH4 was then given to the subjects and their phe levels were subsequently tested 4, 8 and 24 hours later. One 8-year-old male with atypical PKU was responsive to the drug. Dr. Scriver added that he was aware of a patient with classical PKU who was also responsive to BH4 from a different study.

Dr. Hoganson reported that the University of Illinois has been conducting bone density testing of 40 subjects with PKU and will soon begin the data analysis of this research project. There are no conclusions of this study at this time.

Dr. Scriver responded to a question regarding the adult patient with PKU and symptom manifestations resulting from going off diet. He stated that adults with behavioral problems have noted an improvement in their symptoms once dietary management was reinstated.

The last question was directed at the panel, asking if there have been any advancement in home monitoring equipment of blood phe levels. The doctors were unaware of any such systems.

In conclusion, the question answer session was an interactive and informative segment of the annual PKU meeting.

## Clinic UPDATE

Each year at the PKU Annual Meeting we look forward to hearing from our local PKU doctors to learn about new developments in their clinics. We are very lucky in Illinois to have 3 experienced geneticists who treat our children – Dr. Paul Wong, Dr. George Hoganson and Dr. Barbara Burton.

Our host for the day, Dr. Wong began by sharing that his clinic at Rush has received some new patients this year and he is pleased with the results of the expanded newborn screening recently implemented in Illinois. Dr. Burton described how busy the Children's Clinic has been this year adding 16 new patients with PKU and variant disorders including 3 transferees from other clinics. She echoed Dr. Wong's statements about the benefits of expanded newborn screening. She also described the BH4 study which is now underway at Children's. Dr. Burton concluded by telling us what a pleasure and privilege it is to work with the children and families of those affected by PKU in Illinois. Dr. Hoganson brought us up to date about the University of Illinois-Chicago Clinic. They have 7 new patients and are working on the results of their bone density study. They are also participating in the BH4 study and may look into MRI studies on brain function in patients with PKU since the UIC is developing a new MRI center. Dr. Hoganson wrapped up the clinic update by reiterating what a pleasure it is to work with his PKU patients and their families in Illinois.

Thanks to each of our Drs. for taking time out of their busy schedules to spend the day with all of us. We appreciate all of your hard work on behalf of our children and loved ones with PKU.

This year's New Parent Coffee was hosted by Kem and Dan Baker. As in the past, this event has been a great source of support for new PKU parents who can come to share their concerns with other PKU parents.

Many families attended this year and were able to enjoy the afternoon making new friendships and sampling some low protein foods.

A big thanks to Kem and Dan Baker for hosting this event.

## A Special Thanks!

The Board would like to thank Patty Cosgrove and U.S. Can Corporation for the use of the phone bridge for our board meeting. The phone bridge allows board members use the phone to conference into board meeting when they cannot attend in person. This feature has proved to be an invaluable tool to the Board and it is greatly appreciated.

### Wise Foods Update

The Wise Onion Rings can be ordered directly from Wise Foods. The case of 72 bags (.5 ounce each) are \$19.53 and shipping is \$7.00. This is a little cheaper than the low protein food sources. They also sell them in different size bags. They also have a map so you can find what stores they are sold in - apparently Illinois does not sell Wise products. The website is [www.wisesnacks.com](http://www.wisesnacks.com) and the phone number is 1-800-GET-WISE.

Children between the ages 6-12 were offered a special program once again this year at the Annual Meeting on November 16, 2002. As in the past two years, the focus was based on previous feedback and input from the children, as well as suggestions from the members of the PKU Organization of Illinois. Self esteem/esteem building and a support network were the objectives of the various activities and discussions.

The day began with a "getting to know you" activity. The children individually chose from a box an unknown statement or question and shared their response with the group. This exercise generated an impressive amount of positive energy. Many ideas, thoughts, feelings and laughter were expressed amongst the children. It was a start of a dialogue which continued throughout the day.

Everyone was uniquely creative during the art activity. The children had the opportunity to design their own "wish box" and place their personal notes and comments inside. Conversations and discussions flowed while they worked toward the completion of the project.

During the breaks, the children explored the vendor fair with the goal to "seek and find". The children



brought back to the group great new discoveries and freely spoke about their likes and dislikes. Many suggestions and tips were also given to each other.

The children were the best teachers and confidantes to one another during each phase of the day. They asked thought-provoking questions, shared struggles and successes and always found a moment to have fun while exploring their world of PKU.

## ANNUAL MEETING CHILDREN'S PROGRAM

By: Susan Ruohonen, M.S.



It was extremely rewarding for me to observe the children and their participation in the events throughout the day. They discovered their own way to challenge one another and discuss their concerns and ideas. Regardless of age, bonds were formed, respect was gained and support for one another was evident in so many ways.

The challenge remains to continue to develop programs which will meet the needs of the children. It is essential to provide the avenues for them to learn, explore and develop a network of support for one another. I extend my gratitude and appreciation to everyone involved. It was my privilege to share the day with those in attendance. It is always very comforting to see familiar faces and exciting to see new ones each year. Thank you for your participation and support.

## Gene Therapy Apparently Leads to New Illness in Boy

Health Officials in U.S., France Suspend Treatments Recently Hailed as Cure for Immune Disorder

BY RICK WEISS, WASHINGTON POST STAFF WRITER – FRIDAY, OCTOBER 4, 2002: PAGE A11

An experimental genetic therapy that had seemed to cure several infants born with a life-threatening immune system disease has apparently caused a leukemia-like syndrome in one of the treated children in France, prompting health officials to suspend all such therapies for that disease in France and the United States.

Officials at the Food and Drug Administration said yesterday they had scheduled an emergency meeting in Bethesda next week, at which scientists involved in the study and other experts will analyze the latest data. One goal is to help the FDA consider whether a wider array of gene therapy studies need to be scrutinized more closely.

The ill patient, a French boy almost 3 years old, is now being treated with chemotherapy in an attempt to kill the blood cells that have begun to proliferate in his body, an ironic twist for a child whose problem at birth was the absence of such cells.

The incident is a blow to the fledgling field of gene therapy, which had only recently begun to recover its image after the widely publicized death of an Arizona teen in a 1999 experiment. The approach, which seeks to cure diseases by giving patients healthy genes, has been fraught with failure. The French treatment had recently been promoted as having delivered the first real cures in the field's 12-year history.

Despite the setback, scientists yesterday said they suspected that the complication may prove to have been caused by a molecular fluke so unlikely to reoccur that the treatment's benefits will ultimately prove greater than its risks.

The French treatment still looks "exceedingly promising," said Joseph Glorioso, director of the Pittsburgh Human Gene Therapy Center and president of the American Society of Gene Therapy. "The field of gene therapy remains vigorous and robust."

Alain Fisher, the project's lead researcher at the Necker Hospital in Paris, said he held out hope that the boy would recover, and said the family was being very understanding.

"They are extremely intelligent and courageous. They are very kind people," Fisher said. "They understood that this was experimental, and they accepted it. I admire them."

The boy was born with X-linked severe combined immunodeficiency disease, sometimes known as the "bubble boy" disease, caused by a defect in one of the body's roughly 40,000 genes. Lacking that gene, children cannot produce the white blood cells crucial to fighting off infections. Most die in the first year or so of life.

About 2 1/2 years ago, when he was 2 months old, the boy became one of a few infants enrolled in a new gene therapy study. The experiment involved the use of genetically engineered viruses to deliver copies of the missing gene to young patients' bone marrow cells. This summer, he was one of five children described in a landmark New England Journal of Medicine article as having been apparently cured.

A total of 11 children from five countries have been treated with the technique, Fisher said. All except the boy are doing well and show signs of having normal immunity.

Fisher said problems first became apparent in the boy in August, when routine tests indicated that his white blood cell count had climbed not just

to normal but to abnormally high levels. After a second test indicated even more of a rise, additional tests were done.

Using sophisticated molecular probes, the research team found that the engineered virus, which delivers its genetic payload at random locations within a cell's DNA, had in at least one cell delivered the curative gene in an awkward location: immediately adjacent to a gene called *lmo2* that controls cell division and proliferation.

The team believes that the newly arrived immunity gene disrupted that regulatory gene, predisposing the cell to divide uncontrollably. A second insult – perhaps a viral infection a year or two after the therapy – was probably needed to trigger that division process, Fisher said.

The team notified French authorities about three weeks ago, and then contacted all the families of children they had treated. It's uncertain how likely such an event is to occur again, Fisher said.

But as a precautionary move, France's public health agency and the FDA have placed all retroviral-mediated treatments for this immunodeficiency disease on "clinical hold," a status that precludes further recruitment into trials until the problem is better understood. The hold affects one study underway in London and two that were about to begin in Los Angeles and at the National Institutes of Health in Bethesda.

An FDA official said it would be premature to put a hold on all gene therapy studies involving retroviruses; many of those studies involve terminal cancer patients getting cancer-fighting genes.

Molecular glitches like the one that apparently occurred in the French boy have been considered a theoretical risk of gene therapy since the first such experiments were contemplated in the late 1980s. The first gene therapy experiment took place in September 1990 at the NIH. Since then, thousands of patients have been treated with gene therapy for a wide range of ailments — most of them with retroviruses but increasingly with other gene delivery systems. None has resulted in a case of cancer that experts believed was linked to the treatment, but FDA officials said they are now asking scientists to review their data to see if they may have overlooked such a connection. Fisher said he remains hopeful that, overall, his treatment will turn out to help more than harm. The only other therapy available for the disease is a bone marrow transplant from a close relative. Unless that transplant is from a perfectly matched sibling (available in only 20 percent of cases), the failure rate is 25 percent, more than twice as high as the complication rate shown so far in the first 11 patients, he noted.

Moreover, Fisher said, the quality of life for bone marrow recipients is not as good as it has been for the children who got new genes. Yesterday's announcement of the French complication coincided with the release of a new report on human medical experimentation produced by the Institute of Medicine — a report prompted in large part in response to the death of Jesse Gelsinger, the Arizona teen who died in 1999.

The report calls for increased federal oversight of research involving human subjects, including unprecedented federal oversight of privately funded studies; a revamping of the nation's ethics review boards that judge proposed experiments; compensation for people harmed in experiments; and the creation of a new federal advisory body to consider issues relating to experimentation on humans.

# Cooking Days Continue to Grow...

By Dee Pratscher



Last we had 6, this date 10 have responded thus far with two more weeks to go. Last menu we created donuts, sausage, quiche, spinach ravioli, waffles, sausage ravioli, and perogies. February's date has requests for nuggets, quiche, hotdogs, sausage ravioli, English muffins, soup and TV dinners. These items will be made as well as cinnamon rolls, breadsticks/rolls and a side dish or two. For those of you that have never attended one of these Cooking Day; basically we create a menu of what we will make by taking the wants and needs of those in attendance. Then on that Saturday we cook and visit all afternoon, pack up our fresh baked treats to take home to our little taste testers and there is always some exchanging of recipes, phone numbers and various tricks of the trade before leaving. It's informal but informative. If you'd like to attend call me (Dee Pratscher 708 877-2761) or check the website ([www.pkuil.org](http://www.pkuil.org)) for upcoming dates. I generally hold these every 3 months. If you'd like to host one in your area, I'd be happy to help you get started. Some quick tips for using the Tortilla Maker from Cambrooke Foods: preheat maker for 20 minutes or until light turns off, do not lock maker while cooking, once heated and batter/dough is distributed onto maker, slowly and gently lay the top down and watch batter spread depressing top gingerly. Appropriate heating time and delicate closure is essential in using the tortilla maker.



## PKU ORGANIZATION OF ILLINOIS

### 33<sup>rd</sup> ANNUAL MEETING

NOVEMBER 16, 2002

The PKU Organization of Illinois gratefully acknowledges the following persons and companies who contributed raffle prizes or made donations which helped us bring this year's Annual Meeting to you:

#### RAFFLE PRIZES:

**One Week Trip to Puerto Rico – The Caro Family: Hotel, PKU IL: Airfare**

Winner – Pat Fitzpatrick

Seller – Beth Mullen

**Medinah Golf Outing – The Critchfield Family**

Winner – C. Janka

Seller – Joshua Bennett

**Charter Air Service to champagne breakfast at lake Lawn Resort in Lake Geneva – 4 people - Tim Koster from Midwest Avtech**

Winner – Deb Skopes

Seller – Angela Koster

**Color Television – The LaGioia Family**

Winner – Kathy Anderson

Seller – Annette Egan

**Mill Creek Hotel – Lake Geneva – 2 nights**

Winner – Tom Grabowski

Seller – Tom Grabowski

**\$100 American Express Gift Certificate – Jodi and Ruben Caro**

Winner – Melinda Murphy

Seller – Scott Stevens

**Chicago Cubs Tickets – The Critchfield Family**

Winner – Heidi Ramos

Seller – Heidi Ramos

**Six Box Seats – Arlington Park Race Track**

Winner – John Hentrich

Seller – Joshua Bennett

**Chicago White Sox autographed pictures**

Winner – Nick Humphrey

Seller – Juan Santillan

**Tickets to Second City**

Winner – Jim Fruzyna

Seller – Joshua Bennett

**Family Pass to the Children's Museum**

Winner – Casey Talbot

Seller – Tom Keegan

**Chicago Architecture Loop walking Passes – 4 tickets**

Winner – Shawn Murphy

Seller – Tom Keegan

**Family Membership to the Museum of Broadcast Communications**

Winner – Doug Rardin

Seller – Herschberger Family

**Terra Museum of American Art – 6 month membership**

Winner – Kevin Kauffman

Seller – Herschberger Family

**Kohl's Children Museum – Family Pass**

Winner – Joe O'Connor

Seller – Beth Mullen

**"Thank You"**

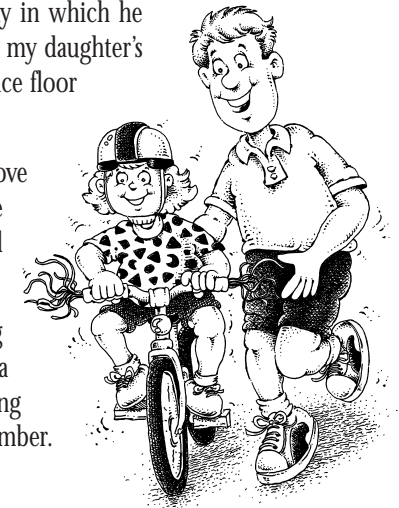
## Papi's Perspective *By Ruben Caro*

It is hard to believe how time goes by! Yes, after a brief absence, Papi's Perspective is back! This is an informal column with a perspective of a daddy ("papi" in Spanish) of a daughter with PKU. I hope to continue to bring my perspective about issues, ideas and feelings that are relevant to us dads, as we deal with a child that is growing so fast in front of our eyes. In this column, I want to share a recent experience that reminded me of the importance of enjoying and cherishing every moment of my daughter's life.

I was with my wife at a wedding of one of my colleagues. There is part of the wedding where the bride dances with her father. No problem, right? Wrong! As the music began, I started to listen to Bob Carlisle's 1996 hit song "Butterfly Kisses." This is an emotional song about a father's tribute to his daughter on her wedding day in which he recounts several important events of her life along the way. I was crying like a baby, as my daughter's life started to flash in front of me. She is only 9 now, however, I saw myself on that dance floor in front of my beautiful daughter telling her how proud I am of her.

"With all that I have done wrong, I must have done something right to deserve her love every morning. And butterfly kisses, I couldn't ask God for more, man, this is what love is. I know I've gotta let her go, but I'll always remember every hug in the morning and butterfly kisses..."

Truth is I am glad she is only 9 and I don't have to let her go - yet! I will have a long time to cherish every moment, watch her grow, and see her transformation from a little girl into a woman. There are going to be many challenges along the way, including those associated with PKU, but there will also be plenty of moments to always remember. I can still get my hugs every morning and butterfly kisses!!



PERSPECTIVE

### **Dr. Charles Scriver - Overview of Phenylketonuria**

By Jodi Caro

Dr. Charles Scriver was the keynote speaker at the PKU Organization of Illinois 33rd Annual Meeting held on November 16, 2002. Dr. Scriver is a Professor of Human Genetics at McGill University in Montreal Canada. He traveled to Illinois to bring us his unique perspectives on the history and treatment of PKU.

Dr. Scriver began his talk with a story about 2 sisters he treated with PKU. One sister was late diagnosed and although she was treated she suffered permanent impairment. The other sister was diagnosed at birth, treated and led a normal life. The vastly different life experiences of these two sisters taught Dr. Scriver a lot about PKU. Dr. Scriver educated us about some early pioneers in the field of PKU. Ashborne Furline was a Swedish scientist who in 1934 recognized a genetic disease which fit the rules of inheritance and disrupts biochemistry. In 1946 another scientist, Lionel Penrose gave this genetic disease the name Phenylketonuria. He published a paper which put forth the idea that a way to treat PKU is to change the metabolic environment of the patient and as a result he anticipated the treatment of PKU. PKU became one of the first treatable genetic diseases. Of course as we all know Dr. Robert Guthrie developed the test for PKU which has become the standard in newborn screening.

Dr. Scriver reviewed the genetics of PKU and the identification of the many gene mutations within PKU. The mutations of PKU vary by family and geographic region. Mutation analysis is often done to provide patients and their families with additional information about their own specific genetic makeup and may prove useful as future treatments are explored for PKU.

Dr. Scriver briefly reviewed the geography of PKU and how it the flow of the PKU has followed the flow of the general population. He then discussed some new thoughts on PKU treatment including BH4. He described how a subset of PKU patients might respond to BH4 (tetrahydrobiopterin). BH4 acts as a chemical chaperone and can work to lower phe levels in certain PKU patients. Unfortunately BH4 treatment is extremely expensive but Dr. Scriver and others are looking into a way to make a similar drug in a less expensive manner. He believes about 40 PKU mutations will respond to PKU treatment.

Dr. Scriver emphasized the importance of treating PKU as early as possible and maintaining treatment for life. He highlighted the growing interest in improving the quality of the PKU diet. Dr. Scriver also reviewed other possible treatments such as gene therapy and PAL (Phenylalanine Ammonia Lyase) enzyme replacement but noted that there is a lot of work to be done in both areas.

At the conclusion of his talk Dr. Scriver took questions from the audience and asked us to consider the challenges associated with treatment of adults with PKU. Since pediatric geneticists have the primary experience in treating PKU and most internists lack the experience necessary to provide proper treatment, adults with PKU must struggle to find adequate care. Their options are to seek out pediatric doctors with experience treating PKU or push for the proper training of internists.

Thank you Dr. Scriver for sharing your insights with all of us.

# PUMPKINS... PONIES... POTIONS...

By Dee Pratscher

This year's Halloween event was held at Siegel Cottonwood Farms. More than fifty participated in the festivities. Partaking train rides, hay mazes, feeding farm animals and plenty more. The weather cooperated, the sun was hot and there wasn't too much wind, a great Saturday for October in Chicagoland. Lunch included low pro: chili, mac-n-cheese, cupcakes, sweet breads, pasta, rice krispie treats, chips, pop, and popcorn balls. Hotdogs and pizzas were served for our non-pku friends. The kids were given treat bags with prizes and plenty of candy they immediately began nibbling. Our favorites were Taco and Chocolate. Believe it or not they weren't on the menu but a couple of the ponies we rode. The fort playground was the other big hit with our pumpkins. Everyone was able to take home a pumpkin to continue the celebration. Admittedly I missed seeing the kids in costume and awarding prizes. Next year we will have to include a craft and costume contest but the pumpkin patch was a great family centered experience. Thanks to all who came and to those who brought treats.



## PKU Organization of Illinois Scholarship Winners

In 2002, the PKU Organization of Illinois offered two \$1,000 scholarships to individuals who are continuing their education. This year's winners are Brent Woody and Robert Wollenzien. Brent is currently attending Missouri Baptist University and majoring in Kinesiology. Robert is attending North Central College and earning a degree in Broadcast Communications. Congratulations to Brent and Robert. Best of luck to you both and all members who are pursuing continuing education.

### A Gratitude of Thanks... To the PKU Organization,

I want to sincerely thank you for generously awarding me a PKU scholarship. With the many expenses of getting a college education, I greatly appreciate your assistance and generosity in my pursuit to succeed academically and prepare for my future. Currently, I am attending North Central College in Naperville, Ill., and I plan to major in broadcast communications (most likely sports broadcasting). This career field is very exciting and I am enjoying my broadcast experiences at North Central thus far. Although I had planned to attend the November PKU Meeting in Chicago, unexpected health problems arose and prevented me from publicly accepting the scholarship. Once again, I cannot express my gratitude for the PKU Organization's kindness and generosity; I can only try to put into words how great of a feeling it was when I learned that I had won the scholarship. If you would like, feel free to publish this in the PKU Press so that I may spread my thanks to as many people as possible. Happy holidays to all!

Sincerely,

Rob Wollenzien

## ILLINOIS DEPARTMENT OF PUBLIC HEALTH Newborn Screening Program Highlights

### ● WHAT'S NEW IN NEWBORN SCREENING?

Universal expanded newborn screening of all babies born in Illinois became a reality on 7/1/02. On that date IDPH Chicago laboratory began utilizing tandem mass spectrometry (MS/MS) to test all newborn screening specimens. Due to the efforts and support of our medical specialists, parent organizations, like the PKU Organization of Illinois, and our Newborn Screening Advisory Committee, Illinois is now among the few states in the nation to provide newborn screening for additional metabolic disorders. The Illinois newborn screening panel now includes testing for amino acid disorders, including tyrosinemia type I, maple syrup urine disease (MSUD), homocystinuria and several urea cycle disorders, fatty acid oxidation disorders, including MCAD (medium chain acyl CoA dehydrogenase deficiency), and several types of organic acid disorders.

Between the dates of 7/1/02 and 9/30/02 over 49,500 dried blood spot newborn screening specimens have been tested at the IDPH Chicago lab. As a result 9 babies were identified and diagnosed with PKU, 1 baby was diagnosed with hyperphe, 1 baby was diagnosed with MSUD, 2 babies were diagnosed with MCAD, 1 baby was diagnosed with SCAD (short chain CoA dehydrogenase deficiency), and 2 babies were diagnosed with an organic acid disorder.

Please visit the IDPH website, [www.idph.state.il.us](http://www.idph.state.il.us). A search under the topics of "genetics" or "newborn screening" accesses information about the Genetics/Newborn Screening Program. Our recently updated fact sheets about newborn screening disorders and parent brochure, "Babies First Steps", are also available.

### ● STAFF CHANGES

Unfortunately for IDPH, Barb Toepfer, the staff member who processes formula orders, will be retiring 12/31/02. Barb is busy training a new IDPH staff member, Lauren Whalen. Lauren will soon begin taking over some of Barb's duties. While Barb looks forward to her much deserved retirement, she will miss the parents, dieticians and counselors with whom she has worked over the past few years.

### ● FORMULA ORDERS

Illinois is proud to be one of the few states in the nation that provides medically necessary formulas for individuals diagnosed with PKU who are under the care of IDPH designated medical specialists. IDPH will also provide medically necessary formula for infants diagnosed with several of the new metabolic disorders identified through expanded newborn screening.

IDPH appreciates your understanding when the Department is sometimes forced to send less of the product than is requested at a given time. Funding for formula is not unlimited. At times, out of necessity and our commitment to provide formula to all Illinois individuals with PKU, an adjustment of sending less of the product than is requested may be necessary. The Department is constantly attempting to improve our service to individuals diagnosed with PKU.

As a reminder, the Genetics/Newborn Screening Program is now located at a new address, 500 E. Monroe St., Ridgely Building, First Floor, Springfield, Illinois 62701. Please make certain that you are using current order forms as the older forms may still have our old address and fax numbers. Using outdated forms may result in delayed processing and shipment. New order forms may be requested by calling 217-782-6557 or 217-557-5379, or requests may be faxed to 217-557-5211. Please keep track of your formula usage and place orders early, allowing at least 10 days for IDPH to receive your orders.

### ● INSURANCE REIMBURSEMENT

Currently IDPH is attempting to obtain insurance reimbursement for PKU formula provided to families during calendar year 2001. The cost of formula is just under 1 million dollars per year; and insurance reimbursement helps the Department recoup some of the Newborn Screening Program expenses, and helps keep our Program costs down. Program costs are always an important issue during State budget crunch years.

### ● NEWBORN SCREENING FEE INCREASE

Although expanded newborn screening was implemented without an increase in the screening fee, the Department has gotten approval to increase the fee from \$37 to \$42 per test effective for any baby born after January 1, 2003.

# National PKU Teen and Adult Conference

**DATE:** 7/25/2003

National Coalition of  
PKU and Allied Disorders

The National Coalition  
for PKU and Allied  
Disorders will be  
hosting a conference  
for PKU Teens, Adults,  
Parents, Spouses,  
Siblings, etc. of PKU  
Teens and Adults  
and the professionals  
involved in the  
treatment of PKU.

It will be held Friday,  
July 25 and Saturday,  
July 26, 2003 at the  
Crowne Plaza Hotel  
and Resort at the  
Crossings,  
801 Greenwich  
Avenue, Warwick, RI  
02886

**For more information  
contact:**

Trish Mullaley  
President  
National Coalition  
for PKU and Allied  
Disorders  
coalition4pkud@  
aol.com  
877-996-2723  
www.pku-  
allieddisorders.org

## The New Board Members are Here!

After a delicious lunch and an extremely successful Raffle, it was time to take care of business – Board business that is. Each year the Board is required to elect members to fill vacant Board seats. The Board of Directors of the PKU Organization of Illinois consists of 11 voting members in addition to ex officio and honorary members. The Board meets periodically throughout the year to plan all of the events our families enjoy. Once the general membership elects the Board, the Board will then elect four officers (President, Vice President, Treasurer and Secretary) at the first Board meeting. Board members are elected to a two-year term, with a limit of three consecutive terms.

*The following Board members have 1 year left on their term and will be returning to service next year:*

- Bruce Kimbrell
- Ruben Caro
- Angela Koster
- Jayne Markham

*The general membership also elected the following Board members:*

- Gina Annunzio
- Joe Annunzio
- Kate Chovanec
- Jim Critchfield
- Patrice Bedenk
- Kristen Leaf
- James Leaf
- Larissa Bruno

*The Honorary Board Members for 2002-2003 will be:*

- Anne Kozek – UIC
- Hazel Vespa – CMH

Congratulations to the 2002-2003 Board. We appreciate your dedication and hard work on behalf of all of our children and families!

## Young Adults with PKU find Support and Encouragement from Peers

A new addition to this year's annual meeting was the development of a Young-Adult Round Table Discussion with Dr. Scriver. The group of teenagers and young adults with PKU joined Dr. Scriver to discuss the issues and challenges they face as they go through the teenage and adult years. The discussion was informal and gave the teens and young adults an opportunity to hear words of encouragement from each other and to know that they were not alone. Some of the teens and adults were doing well on their PKU diets while others were struggling and considering going back on the diet. Each person had a different perspective and insight to offer especially those who were living alone and were away at college. Dr. Scriver provided the group with useful insight and understanding regarding new research in the treatment of PKU.

Sarah Foster participated and shared information about a national Adult PKU Conference which is being planned for next July on the east coast. Email Sarah Foster at pkuadult@aol.com for an invitation to the conference or for further information about the PKU Teens and Adult Listserv [a discussion list for PKU teens and adults from around the world to provide support and raise questions]. When contacting Sarah please include your name, age, and city/state/country.

Overall, participants were supported and encouraged as they talked with others who face similar challenges during the transition from the teenage years to adulthood. They also provided suggestions for on going programs ideas, which have been forwarded to the Board.

Melinda Erickson, B.A.  
Social Work Intern  
Children's Memorial Hospital



## Please Help Us Make The Annual Meeting Even Better.

Please send us your comments, thoughts, and criticisms of the 2002 Annual Meeting. Please tell us what you liked and did not like about the 2002 Annual Meeting. What would you like to see different at the 2003 Annual Meeting?

Please provide us with your ideas about the theme, presentations, speakers, break out sessions, special programs etc.

Please feel free to send us your comments at:

### E-Mail:

info@pkuil.org

### Voice Mail:

(630) 415-2219

### Regular Mail:

PKU Organization of Illinois  
P.O. Box 102  
Palatine, IL 60078-0102

Thanks and best regards,  
The PKU Organization of Illinois Board of Directors.



## Workshop:

### PARENTING THROUGH TRANSITIONS - SELF MANAGEMENT OF PKU

Guidelines and an overall approach to manage PKU and how parents handle some typical challenges at various development stages from childhood through young adulthood were considered. Participants received reprints of materials authored by Christine M. Trahms, MS, RD., Seattle, WA entitled: **Achieving Healthy Outcomes for Children with PKU (Part 1 and Part 2.) National PKU News, Vol. 13, No. 2, Fall, 2001 and Vol. 13, No. 3, Winter 2002.** and the PKU Self-Management Timeline (University of Washington, 1999 revised edition). Really Living with PKU, a video produced by Christine Trahms and the PKU Action Group, 2001, Seattle, WA, was viewed and used to illustrate how a number of parents react to the initial diagnosis and how they and their children cope and adapt over time. These educational resources serve to highlight what we hope can become on going conversations with your families, with friends in PKU support groups - formal and informal, and with PKU clinic staff in on-going contacts.

For a detailed and comprehensive approach to look at steps in health supervision from infancy through adolescence, explore: **Bright Futures Guidelines for Health Supervision of Infants, Children and Adolescents (www.brightfutures.org)** – a model that is well known and accepted by health care professionals. This model provides a structure that parents can consider as they shape expectations related to health as well as quality of life issues. The *Bright Futures* model is the foundation from which Christine Trahms developed the above articles, specifically addressing parenting leadership and supervision issues pertaining to PKU.

Effective management of PKU requires a thoughtful parenting process, which includes understanding your child's psychosocial needs and skills at varying ages; being aware of your own parenting style and capacities; and identifying and using the supports you have within your family, friends and larger community. Participation in the programs and activities of the PKU Organization of Illinois such as this Annual Meeting confirms how effective parent to parent support can be. Remember to turn to parents you have met at Annual Meetings, other programs, or as you have visited PKU Clinics. Keep the 2002 PKU Directory close at hand when you need support. Call on any of your PKU Clinic Team members who can also help guide you to resources.

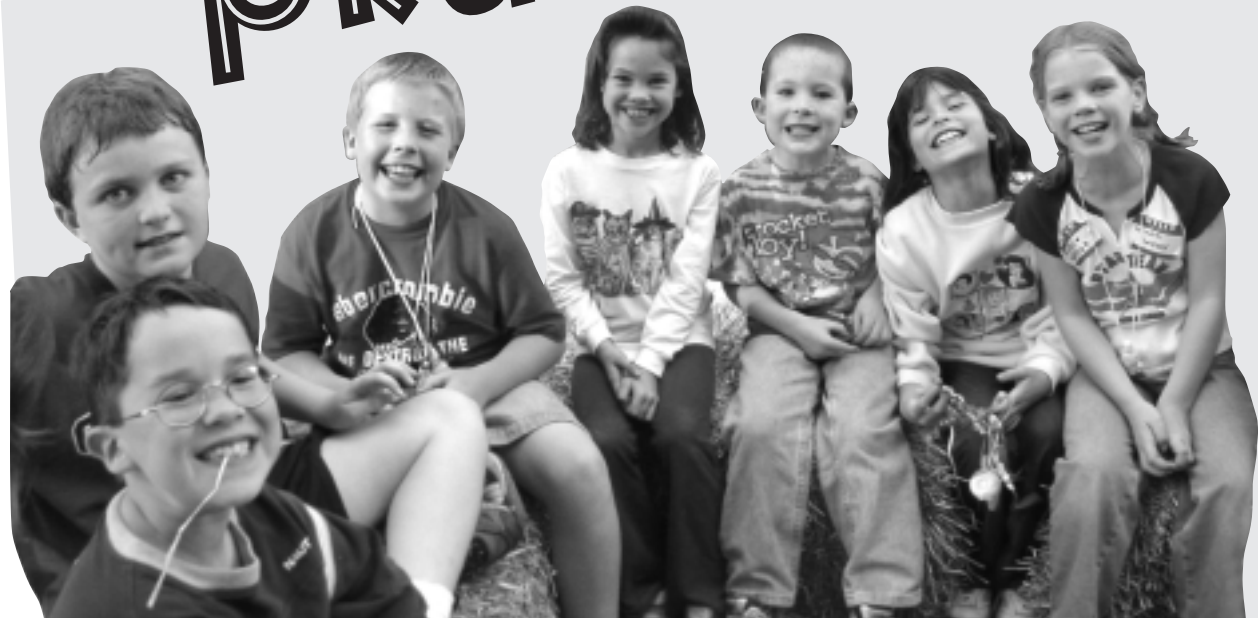
If you are interested in obtaining any handouts used in this workshop, contact Rhoda Papanastassiou, dietician or Hazel Vespa, Social Worker, workshop facilitators.

## Thank you Gina !!

By Dee Pratscher

Gina Annunzio has been handling the PKU Press for a couple of years now. She worked hard collecting, organizing, writing articles, and proofing the final product. It's a huge responsibility and I think I speak for everyone when I say she has done a wonderful job so on behalf of the group I want to say thank you to her for delivering us concise and pertinent information. The PKU Press is our link to each other, it is vital to keeping us informed and connected. Remembering that, the Press has always had an open door policy if you have information or recipes you would like to share, contact a board member or e-mail the article to info@pkuil.org. Ruben Caro will take over this phenomenal task and to him I commend his efforts and self sacrificing time he will be dedicating to this project. I wish you the best of luck and offer any support I can be.

pkU



# family weekend!

By Michele Kowalski

September 27, 28 and 29<sup>th</sup> proved again to be super weekend at East Bay Camp in Hudson, IL. The PKU Family Weekend was hosted by the PKU Organization of Illinois for the 21<sup>st</sup> year. There were 15 families in attendance. Some started the weekend Friday night at camp arriving to a roaring campfire. This is what Ian, who's 10, waits all year for. He rates this the best PKU event he attends all year and he attends quite a few events during the year.

Early Saturday there was a fishing competition. Several fish were caught and everyone felt like a winner. We all meet for lunch at the main dining hall. This was the chance for people to become reacquainted with each other and to meet new families. This is the official kick off for the weekend. After lunch the action was non-stop. We had a moon bounce for the afternoon, swimming, baseball and games in the fields. There were also some great crafts for kids and adults. We had hay rides where everyone got to go out on a 30 minute ride. On the second ride several deer were spotted. It was then time for dinner; we had lo-pro make your own pizzas and much, much more lo-pro and regular dishes. After dinner the campfire was



started and at the same time the cotton candy machine was going strong. So after many marshmallows and cotton candy was consumed we needed an outlet for the energy still left in all the kids. It was time for dancing and games at the Recreation Center. Everyone played either basketball or danced, some just were content to sit. There were some very tired folks by this time of the day. It was then back to our cabins for a good nights rest.

Sunday we had a brunch, with lo-pro pancakes and lo-pro crepes made fresh. There was much to eat as everyone brings their favorite lo-pro and regular dishes to share. It was then time to sadly say goodbye until next year's camp.

Thanks to the PKU Organization of Illinois for sponsoring the event each year and many thanks to all that helped organize the weekend. To all the families that come to camp many thanks, you help makes it so successful every year.

Hope to see you there next year. I know Ian can't wait for next year's camp.

food





games



fun

crafts



sports



...and sleep!

# "2002 ANNUAL MEETING RAFFLE – THE BEST YET"

2002 will go down in our history as one of the best years ever for our largest fundraising event, "THE RAFFLE".

*The 2002 raffle raised \$10,807!*

What made this year's raffle so successful? It's simple. Member participation and involved parents generated these sales. No matter how much any of our members

sold, in 2002 everyone seemed to contribute greatly towards this important annual fundraising event.

Certain members in our organization "went the extra mile" in raffle ticket sales. This year's top producers were:

- Steve & Lisa Bennett (\$2,142 in ticket sales – AWESOME!)
- Nelson & Anna Herschberger (\$1,310 in ticket sales – GREAT JOB!)
- Tom Keegan (\$600 in ticket sales – IN ONLY TWO DAYS!)

For their efforts, the Bennett's, the Herschberger's and Mr. Keegan received a \$100, \$50, and \$25 American Express gift certificate respectively. Just think, almost 40% of our raffle ticket sales came from these three families – THANK YOU!

This year's raffle was also very successful because of raffle prize contributions from our member families. Specifically:

- 1 week trip to Puerto Rico – The Caro Family (this was our grand prize)
- Medinah Golf Outing on Course #3 – The Critchfield Family
- Charter Air Service to champagne breakfast at Lake Lawn Resort in Lake Geneva – 4 people - Tim Koster from Midwest Avtech
- A \$100 American Express Gift Certificate – The Caro Family
- Color Television – The LaGioia Family, Parents of Lisa Hughes
- Chicago Cub Tickets – The Critchfield Family

Many of our vendors contributed product samples for everyone to try. A special "thanks" goes to our good friends at Ross Products (represented by Ann Levin, Herman Miller, and Mario Pierre-Jerome), Applied Nutrition (represented by Sara Foster), and SHS North America (represented by Sally Ritz) for being there to personally answer questions and to allow our families to sample their new products.

The Organization would like to express our gratitude to the following vendors for the monetary donation given for the Annual Meeting:

SHS, Inc. ....	\$350.00
Applied Nutrition Corporation .....	\$500.00
Ross Products Division .....	\$500.00
Cambrooke Foods .....	\$100.00

Next year's raffle is right around the corner – if you have prizes that you can contribute or know of companies or organizations that will contribute a prize, or would be willing to help write the raffle prize solicitations, LET US KNOW.

The individual prize winners of the 2002 Annual Meeting Raffle are listed separately in this newsletter.

THANK YOU TO EVERYONE WHO PARTICIPATED BY SELLING TICKETS, BUYING TICKETS, AND CONTRIBUTING PRIZES – YOU MADE THIS YEAR'S RAFFLE ONE OF THE BEST EVER.