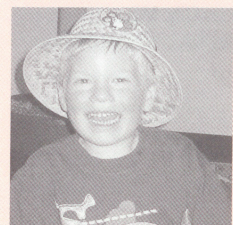




# The Gathered View

September-October 2004  
Volume 29, Number 5

National Newsletter of the Prader-Willi Syndrome Association (USA)



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## Living With 'The Sunshine Kids'

By Marge Wett

Through my years of experience working with infants, children, adults, parents and professionals, I used the phrase "the sunshine kids." I don't know if that phrase is original, but I do frequently use it when referring to the people with PWS who do not exhibit some of the more bizarre behaviors that you have all read about.

I believe we need to read about successes too, to balance our perspective. Many parents reading about PWS for the first time state that all they read is all "doom and gloom" or use such adjectives as "devastating."

Unfortunately it is reality. New parents find reality hard, a situation we wish we could change. The fact is, PWS is a rather complex set of symptoms that will affect this infant. You have lost the "normal" child you expected, and that is not easy to face.

Even with your child's handicaps, you do have a child who can achieve the goals you want for all your children. My list of goals includes happiness, reasonably good health and achievements.

My daughter Lisa, who has PWS, has accomplished everything on my list. Some people may describe Lisa as having a "mild" case of PWS. To me that was as if saying I had a mild case of pregnancy. Having been through seven pregnancies, I can say they are not all alike. PWS is not the same in every case either.

Lisa's diagnosis came in her teens, but we worked with the symptoms from the day she was born. The hard reality for us was leaving her in the hospital for the first three weeks of her life and then taking home the very flaccid baby who required gavage (tube) feeding. It was a hard reality for my late husband Dick (who was a physician) when Lisa returned to the hospital at five weeks for some tests and

he was the one who responded to the emergency call when she reacted to the testing and we almost lost her. The hardest part of the testing was that it gave us no answers,



Marge Wett, PWSA(USA)'s  
first executive director,  
with her daughter Lisa, who has PWS

and later I would find myself telling parents that they were fortunate to receive an earlier diagnosis, although they couldn't always see that.

Lisa's needs were different from our other children, and frequently more difficult, but I never had the doom-and-gloom attitude. It's true Lisa is one of "the sunshine kids," but we had our share of facing PWS problems, too. We survived Lisa's problems just as we survived our other children's problems, as well as their joys.

Like our other children, time came for Lisa to move away from home. Moving from our home into a group home situation was a matter of meeting her needs, and it has continued to meet them over the years.

*Sunshine Kids continued on page 14*

# Prader-Willi Syndrome Association (USA)

5700 Midnight Pass Road, Suite 6 • Sarasota, Florida 34242  
**1-800-926-4797** 9 a.m. to 7 p.m. Eastern Time  
Local telephone: 941-312-0400 • Fax 941-312-0142  
e-mail: [national@pwsausa.org](mailto:national@pwsausa.org) • web site: [www.pwsausa.org](http://www.pwsausa.org)

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Limited to Members: [www.pwsausa.org](http://www.pwsausa.org)  
User Name: members  
Password this issue is LEARN  
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## Board of Director's View

# Help! We NEED You!

### Carol Hearn

As your new board chair, I have an important message for you: PWSA(USA) urgently needs your financial support.

**Why?** The math is fairly simple. To continue providing our programs, products and services, promoting research, and finding new and better ways to improve the lives of people affected by Prader-Willi syndrome, PWSA(USA) must take in enough money to support those programs, products, services and new initiatives.

In recent years, our programs and volunteer efforts have been growing by leaps and bounds. Moreover, as state chapters have become more reluctant to host the large annual conference, PWSA(USA) has filled the void by hosting the conference every other year — and continues to be financially responsible during the state-hosted years.

Unfortunately, the funding necessary to keep us operating has not kept pace.

Thus, in 2003, while we provided services and information to more newly diagnosed families, more families in crisis, and more educators and medical professionals than ever before, we experienced an operating deficit of more than \$30,000. We must accelerate our fund raising to close the gap! We appear to be on a course to run an even bigger deficit in 2004, so we haven't a moment to lose.

**What has PWSA(USA) accomplished with the money you and I have already given?** Here are some highlights of our fast growing program and volunteer efforts. In addition, PWSA(USA) provided information and support to chapters and affiliates, developed a number of new and updated publications, and sponsored a bereavement program to support our families who have lost a loved one.

### Information, Education and Awareness

Every day, PWSA(USA) provides a vast quantity of *free information* to hospitals, physicians and parents worldwide. Our efforts have been rewarded by a noticeable increase in the rate at which cases of PWS are being diagnosed and referred to PWSA(USA).

In 2003, PWSA(USA) responded to educational and crisis calls at an average of 1,250 calls and more than 3,000 e-mails per month. PWSA(USA)'s Clinical Advisory Board members donated many hours of professional services to help make this possible.

We mailed 16,000 packets of educational material and 18,000 copies of *The Gathered View* in 2003.

Visitors to our web site increased 600% from 2001 to 2003. The web site is accessed more than 30,000 times daily.

PWS (USA) sponsored and staffed awareness booths at four major medical conferences in 2003, distributing educational CDs, videos and brochures to hundreds of national and international physicians at each conference. Because securing booth space costs money and finances are so tight, we have canceled plans to sponsor booths at some of this year's medical conferences.

### Research

PWSA(USA), with the help of its Scientific Advisory Board, plays a strong role in facilitating networking among researchers and advocating on a national level for appropriate research. We don't have resources to fund the numerous research projects we'd like to, so we try to focus efforts on funding projects that are within our means, and advocate for NIH and other government and institutional programs to undertake appropriate projects beyond our means.

In 2003, we became a part of an NIH rare disorders collaborative grant and collaborated with other organizations on research efforts; distributed a request for proposals regarding research on psychotropic medication; and advocated legislatively to have PWS included in the wording of funding proposals.

**Having never had to struggle to establish this organization, many of us have not fully realized that the future of PWSA(USA) is in our hands. But it is!**

### Mentoring

PWSA(USA)'s New Parent Mentoring Program served 184 families of infants and toddlers in 2003, for a total of 545 families served in the last three years. This early intervention program, which provides our free publications and volunteer mentors to parents is crucial to the future of our young children with PWS.

### Crisis Services

Our Alterman Crisis Counselor worked on 496 crisis cases in 2003, compared to 352 cases in 2002. And, thanks to the Willet Crisis Grant, PWSA (USA) provided desperately needed advice, intervention and assistance with families' struggles to secure appropriate educational and assisted living arrangements for their children.

*We Need You continued on page 5*



## A Renewed Sense of Hope

**Carolyn Loker**

Upon accepting the new position of President of PWSA(USA), I decided to donate the expense of travel and time away from my family to spend a week with the staff and volunteers at the national office. It was my hope to gain a better understanding of the workings there.

The night before I left, knowing I wouldn't see my little Anna the next morning and would be gone for seven days, I tightly tucked her in bed, listening to her prayers. As she was saying bless Daddy, bless Mommy, she stopped and looked at me sadly. "But who will say Good Morning, my little Princess?"

That's how I greet Anna each morning. So I gave her seven hugs and seven kisses for each day I would be away saying, "Good morning, my little Princess" each time. "Will that do?" I asked.

Anna smiled, closed her eyes and fell fast asleep.

Monday morning, as I listened intently to each person at the national office explain his/her job responsibilities, there was this feeling I had that, it wasn't just about their jobs, but it was about their love and dedication to helping in their own way to make a difference in the many lives of the children and families affected with this syndrome.

How heartbreaking it was listening to the countless numbers of calls, and hearing staff say with despair, "That makes the fourth crying mother today, I feel so bad for them." Then another person saying, "We had three families with new babies call in today. That is so many! We are

happy, though, that they found out so early and can get wonderful interventions that will change their future." And another person saying, "A doctor called reporting a death of a young child and wondered how he could have stopped it." One staff person was dismayed that a grant to study the causes of death had been denied.

Looking around the crowded offices of desks, computers, file cabinets, copiers and people squished in their own little corners, I saw hundreds of pictures of our children taped to practically every space on the walls, and a handwritten sign with an arrow pointing down to some pictures of our beautiful children. It said "Why I'm here!"

Not only did I leave with a unique understanding of the workings at the office, I left with a renewed sense of hope that our staff and volunteers are working because they want to make a difference for our children and families.

In future messages, I hope to bring you an understanding of each position at the office, the board of directors and committees, outlining who they are and what they are accomplishing, so you can have that same sense of hope I feel. The hope I know I have to hold onto as I walk into the airport... meeting Anna... arms reaching out for me, waiting for me to say, "Good morning, my little Princess!"

**I left with a renewed sense of hope that our staff and volunteers are working because they want to make a difference for our children and families.**

## Give Equal Time for PWS Activities As for Kids' Sports

One of the biggest challenges is just getting enough time from anyone, to do anything. In our chapter, it is time for the younger families to take over a lot of responsibility, but they all have families with multiple children going in five different directions, both working spouses, etc. I think this is a systemic issue throughout today's society where there are more school and sporting options than ever before, a much greater percentage of both spouses working, and also many single parents, which is another level of complexity.

The real challenge I keep bumping up against is not a lack of time, but how to get volunteer activities as a higher priority against the other choices that we have available daily. I have gone on some near rants in a couple of board meetings when board members say that they do not have time for our activities and tasks/responsibilities to run the

organization because of a soccer tournament, hockey tournament, etc. with their other kids. I try to nicely lay out an argument that as parents we need to make decisions that distribute our time across all of our children. Because most kids with PWS are not in organized sports, the PWS activities need to be considered the sport of our kids with PWS!

Put in that light, the time that we as parents would allot to board meetings, activities, etc. would grow 10 times. PWS parents think nothing of running a child to a sport 2-3 times a week for at least 1 hour. How much time do we dedicate to enhancing the sporting ability of our children with PWS, which is the ability to survive life with greater educational opportunities, greater vocational opportunities, better medical care and better social services?

— Jay Behnken, President, PWSA of Minnesota



## We Need You - continued from page 3

### Conference

More than 1,000 people attended the 2-½-day conference in Florida in 2003, featuring a 1-day scientific symposium, 1-day care-providers' conference and 1-day conference for presidents of our state chapters and affiliates.

If we increased registration fees to cover conference costs, few families could afford to attend. Despite lower fees, many families can't attend without financial aid. In 2003, PWSA(USA) and our state organizations awarded grants of more than \$30,000 to help families attend the conference.

### *What has PWSA(USA) done to try to raise funds?*

Until two years ago, PWSA(USA) fund-raising was done by our executive director and a few volunteers. Anticipating that the need for its services would soon outstrip our resources, we decided to hire a fund-raising professional.

Unfortunately, two recent trials with professional fund-raisers have not been successful. On reflection, we realize that fund raising for a rare disorder like PWS must be done from the heart by people who truly understand the condition and the significant impact it has on the lives of the families affected by it.

*So instead of retaining another professional, we will help the national office do a more effective job of facilitating fund raising at the grassroots level by people like you.* Future articles in *The Gathered View* will keep you updated.

***What will happen if we fail?*** We don't want to think about cutting any of the valuable programs PWSA(USA) offers. In fact, we are bursting with new ideas. So, together, we must determine *not* to fail. Who could we possibly turn away? The grieving parents and grandparents of a newly-diagnosed infant who are desperate for information? The frustrated parents of a boy who is being denied access to an appropriate education? The distraught parents of a young

## Chapter View

The **Pennsylvania** Chapter has exciting plans to host a Mini-Conference October 16. It will feature Linda Gourash, M.D. and Jan Forster, M.D., internationally known experts speaking on behavioral, psychiatric, and medical management of children and adults with PWS. This will be an expanded version of their presentations at the 2003 national conference which rated raves by those who attended. Neighboring states are welcome. Please check the Web for more information as it becomes available, or contact Maria Silva, 412-369-4433, [KahlanRule@aol.com](mailto:KahlanRule@aol.com).

The Annual Swim Party for people with PWS and their staff, family and friends, was planned for Sunday, August 8, at the home of Judy O'Leary, **Missouri** Chapter president.

The Prader-Willi Alliance of **New York** had a very successful Annual Conference this past spring with nearly 200 attending. New York's Chapter President, Judge Daniel Angiolillo, comments that this validates the Alliance's belief that "a regional PWS conference is extremely valuable."

adult who is being prosecuted for a crime he/she can't begin to understand? The family of a child or adult whose weight and health are dangerously out of control? The list goes on.

***What can you do to help?*** First, make as generous a donation as possible to PWSA(USA)'s general operating fund. Second, you can encourage your friends and relatives to do the same. Last but certainly not least, consider doing a fund-raiser (either yourself or through your state chapter) to benefit PWSA(USA). Contact our national office (800-926-4797) about how to stage a fun and successful fund-raiser.

***Why does PWSA(USA) need you?*** Let's face it. No one advocates for your child or adult with PWS better than you do. At the doctor's office, you provide the most recent, up-to-date information on PWS. At school, you are the advocate who goes to the meeting prepared with the publications necessary to educate teachers and therapists who will be working with your loved one. Similarly, when funds are needed to hold a national conference or continue or improve programs and services your person with PWS requires, you will be the most effective advocate for raising those funds.

***The future of PWSA(USA).*** It is difficult for some of us — especially those of us who have relatively young children with PWS — to realize that PWSA(USA) is vulnerable to financial stress. From our first introduction to the syndrome, PWSA(USA) has been there with information and services that have proven to be timely and reliable. *Having never had to struggle to establish this organization, many of us have not fully realized that the future of PWSA(USA) is in our hands. But it is!*

Time and again, all of us have seen how the actions of one person can make a positive difference in the lives of others. You can be that person. With your help, we can continue our mission.

Jon and Chris Hendricks, co-presidents of the **Michigan** chapter, report that their PWS Awareness Walk raised more than \$5,500, which went to help with the 2004 National Conference. A Mother's Day Flower Sale raised another \$900, which sent three Michigan families to the conference.

After their July fund-raiser golf outing, PWSA of **New Jersey** now looks toward October 23 for the Fall Meeting, which will be a Resource Fair. There'll be something for everyone, with representatives from a variety of agencies speaking briefly about their services, a special speaker for young adults with PWS, and childcare for the younger set.

Prader-Willi **California** Foundation plans their General Education Meeting on November 6 in Sacramento, providing an in-depth look at behavioral issues of PWS and featuring internationally known speakers Suzanne Cassidy, M.D. and B.J. Goff, Ed.D. The following day there will be a training program for care providers in group homes, supported living, and vocational workplace settings.

— Lota Mitchell, Associate Editor

# We Are All About Saving Lives

By Janalee Heinemann, Executive Director

I recently received a call from a physician who told me that one of our mothers brought our Medical Alert articles with her to the emergency room. He said, "If she had not brought the articles and insisted I go to your web site, this child would have died. This information saved her life."

His patient, a slim 15-year-old, had an episode of binge eating. She came in with vomiting and belly pain. The physician said typically he would have treated it like the flu for a couple of days. Due to our alerts, the doctor pursued this further, and found the girl with PWS had such a bad hernia that her spleen, stomach, and duodenum were in her chest. She is now recovering from surgery.

Unfortunately, not all parents carry the Medical Alert articles with them, and not all physicians heed our warnings. In another recent situation, a slim young man had an episode of binge eating and the ER and hospital did not take his symptoms seriously enough, soon enough. Even though we had one of our physicians called as a consultant and emphasized the urgent need for exploratory surgery, there was a 14- to 16-hour delay in surgery before the local hospital physician believed how life-threatening his condition was.

This young man had been doing very well prior to this incident, and a few hours after the eating episode, initially only exhibited signs of stomach pain and vomiting.

See the late Dr. Robert Wharton's article below, which was initially printed in *The Gathered View* in 1999. What Dr. Wharton described was "acute idiopathic gastric

dilation," a situation where part of the stomach tissue dies, which is similar to a heart attack where part of the heart tissue dies.

It comes on suddenly, is very life-threatening and needs immediate surgery. I have been speaking to several medical professionals, including our GI specialist, Dr. Ann Scheimann, and the pathologist who did this report with Dr. Wharton about the cause.

Our conjecture is that if a person with PWS greatly distends his/her stomach with food (slimmer people may be more at risk) and does not get the normal message of fullness or pain, the person may distend it to the point that it cuts off the blood supply, thus causing necrosis (meaning the stomach becomes blackened and dead).

Another risk of binge eating that can create a serious medical emergency is GI perforation. In addition, when there is severe stomach pain, a physician should consider an ultrasound due to the possibility of gallstones and pancreatitis. The pancreatitis can be differentiated by chemistry analysis of the blood and a CT of the abdomen.

We are working on a *Medical Alert Resource Guide* that will contain all of the appropriate articles needed for an emergency situation. It will be published in a format that is professional looking, will not get tattered, and is easy to keep in the glove compartment of your vehicle. We hope to have it available by the time you get the next *Gathered View*.

At PWSA(USA) we are about research for tomorrow — and saving lives today.

## Stomach Problems Can Signal Serious Illness

By Robert Wharton, M.D.

Previously published in *The Gathered View*, March-April 1998

We have recently recognized and reported\* an important medical condition in individuals with Prader-Willi syndrome which families and other care providers should know more about. Although the condition is not common in individuals with PWS, it is much more common in these individuals than in anyone else. It is important to recognize the condition because it can cause severe medical problems when diagnosis and treatment are delayed. The condition can be successfully managed, however, when recognized in a timely fashion.

We have called the condition **acute idiopathic gastric dilatation**. The condition typically begins suddenly in individuals in their 20s or 30s. There is generally no known cause. The first symptoms of illness are vague central abdominal discomfort or pain and vomiting. Bloating of the abdomen, caused by swelling or distention of the stomach, may also appear at this time. The person's temperature may also begin to become elevated at this point. In addition, the individual often begins to look and feel quite ill.

Individuals in whom these symptoms appear should receive **immediate medical attention**:

- abdominal pain
- bloating or distention, and/or
- vomiting

A simple X-ray or CT scan of the abdomen should be taken to look for abdominal distention. If abdominal distention is present and the individual has pain but is relatively well appearing, a test called an endoscopy should next be performed to test the person's stomach lining for signs of inflammation. If the individual has distention on X-ray and is quite ill, emergency surgery might be necessary to more closely examine the person's stomach for signs of inflammation and necrosis [death or decay] of the tissue lining the stomach wall. When severe distention and necrosis is present, treatment consists of surgical removal of a significant portion of the stomach.

Anyone who has knowledge of other individuals who have had severe stomach problems or would like more information on the syndrome, please call PWSA(USA) at 1-800-926-4797.

\* Wharton RH et al. (1997) Acute idiopathic gastric dilation with gastric necrosis in individuals with Prader-Willi syndrome. *American Journal of Medical Genetics*, Dec. 31; Vol. 73(4): page 437-441.

## Aid PWS Research Through Brain & Tissue Bank Donations

A donation of brain tissue may be the key to unlocking discoveries about Prader-Willi syndrome. Many people think about brain tissue donation but find it a difficult issue, especially when death of a loved one with PWS is imminent. Yet this important gift may help researchers find new therapies, treatments or even a cure for PWS.

Currently, PWSA(USA) is in contact with two organizations: the Evelyn F. & William L. McKnight Brain Institute at the University of Florida, and the Brain and Tissue Bank for Developmental Disorders. Both have pre-registration programs and will send you written information.

Here are facts that may help with decision making.

### **How many hours after death is brain tissue viable?**

After death, human brain tissue is viable for 24 hours, or even up to 96 hours, provided the body is cooled, writes Lillian M. Rodriguez of the Brain & Tissue Bank for Developmental Disorders (BTBDD). But tissue is best recovered for research no later than 6 hours after a donor's death, writes Barbara Fankhouser, coordinator at the Human Brain Tissue Bank (HBTB) of the Evelyn F. & William L. McKnight Brain Institute at the University of Florida.

### **Who absorbs the cost?**

Both organizations will absorb most costs associated with tissue procurement. "If the donor dies at a hospital, the procedure could take place there (with hospital permission). If a donor dies in his/her home or some other place, the body would need to be transported to a local facility that would allow the procurement to take place. We are able to procure tissue nationally using these methods. The HBTB can not cover the cost of transportation of the body," Fankhouser noted. PWSA(USA) can help with transportation costs in some cases.

### **What is more or different than a regular autopsy?**

Brain tissue harvest is similar to a regular autopsy, except that half of the tissue is rapidly frozen and used for subsequent RNA and DNA studies, according to Rodriguez.

The HBTB uses the same procedure to recover brain tissue as for a regular brain autopsy. If the body is going to be embalmed, the funeral home should have no difficulty performing this procedure after the brain tissue is removed. You could have an open casket funeral after this procedure.

### **What happens if I want a traditional embalming?**

"Funeral homes are equipped to handle this embalming procedure. It will take them a little longer to do it, but I am told it does not take much longer," Fankhouser stated.

If the body has been embalmed, it still can be used, but it is not as helpful, Rodriguez responded.

### **Why is it important to let the PWSA(USA) national office know?**

The PWSA(USA) national office can often help families make arrangements for brain tissue donation. National also needs to know that tissue banks have available PWS tissue for researchers who may contact us. Both Rodriguez and Fankhouser welcome discussion with family members to answer concerns, and are willing to help however they can.

## Keep These Articles Handy and Make Copies for Your Care Providers

At present, we encourage all families to keep in their car and give a copy for all care providers the following articles:

- 1) Medical Alert for the ER;
- 2) Anesthesiology article;
- 3) Respiratory article;
- 4) Clinical Advisory Board consensus statement on *Recommendations for Evaluation of Breathing Abnormalities Associated with Sleep in PWS*;
- 5) Wharton article on acute idiopathic gastric dilation (for older children and adults with the syndrome).

All these articles are on our web site under the Medical Alert button. If you cannot access the web, call 1-800-926-4797 and request a copy. A new *Medical Alert Resource Guide* containing articles needed in emergency situations will soon be available.

"At no time will we ever pressure a family to make a tissue donation. This is a very personal decision and the family needs to talk it over," Fankhouser stated, adding, "we exist as a resource for families if or when they choose to become a tissue donor. It is always better if donors are signed up prospectively, but if we are called quickly at the time of death, there is still time to try and get the procurement team in place to recover tissue in an optimal fashion.

Fankhouser noted that "we will do everything in our power to fulfill our commitment... but we need the support of the PWS community with regard to becoming prospective as well as post-mortem brain tissue donors." She plans to attend the 2005 PWS Conference in Orlando, Florida.

Rodriguez said BTBDD is available 24 hours a day, seven days a week. "It is our goal to be a bridge between the families who wish to donate and the investigators who are studying these diseases affecting our children."

### **How can I contact these organizations?**

• Barbara C. "Bobby" Fankhouser, M.S.W.,  
Coordinator, Human Brain Tissue Bank, Evelyn F. & William L. McKnight Brain Institute, University of Florida  
P.O. Box 100015, Gainesville, FL 32610-0015  
Office: 352-294-0537; Toll Free Pager: 888-836-0919  
[bfankhouser@mbi.ufl.edu](mailto:bfankhouser@mbi.ufl.edu)  
<http://www.mbi.ufl.edu/facilities/brainbank/index4.php>

• Sally Wisniewski, B.A., Project Coordinator  
Brain & Tissue Bank for Developmental Disorders  
The University of Maryland at Baltimore  
655 West Baltimore Street, Baltimore, MD 21201  
1-800-847-1539  
e-mail: [tbumab@umabnet.ab.umd.edu](mailto:tbumab@umabnet.ab.umd.edu)  
<http://www.som1.ab.umd.edu/BrainTissueBank/main.html>

If you need this information immediately, remember that the phone numbers for both these tissue banks are available on our answering system at 1-800-926-4797.

— Jane Phelan, Editor

# Seen and Heard at PWSA(USA) Ohio Confe

Great educational opportunity. Great networking opportunity. Great opportunity as a provider to meet and talk to parents and others and openly share with others. Thank you!

\* The two sessions for 6-16 I attended on Friday morning were great. Educational yet fun. I love the resources you provide for us \* Fabulous sharing with other families. Excellent educational opportunity



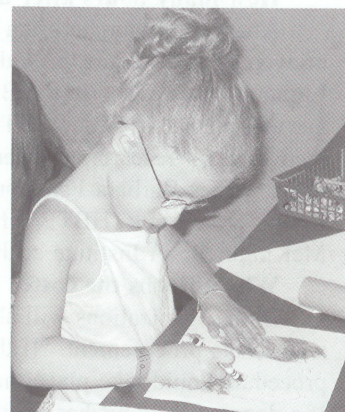
Excellent scientific overview \*  
Presentation was great, MD's very knowledgeable \* Really like the new alternating small/large conference. Everything at this one was great. Keep up the good work!



New PWSA(USA) Board Chair Carol Hearn with outgoing Chair Ken Smith (above left) \*  
Outgoing President Lota Mitchell hands the gavel to new President Carolyn Loker (above right) \*  
Executive Director Janalee Heinemann (center, below left) presents Bereavement Coordinator Norma Rupe and Office Volunteer Annie Durell with awards from PWSA(USA) for their dedication as volunteers at the national office .



\*Very helpful that scientific overview was reviewed \*  
Dr. McCandless (above with Anna Loker) was the best!



# ence 2004



Great presentation by Dr. Linda Gourash and Dr. Jan Forster (shown top right). Lots of valuable information \* As a provider this was the best and most knowledgeable presentation of the conference \* Best ever adult session \* Good combo team --clear, great ideas and expertise \* Linda Gourash was so interesting we didn't want her to quit talking — quite a learning experience \* These classes will definitely help our 22-year-old daughter who lives at home \* Hope someday this can be published \* Top notch!



Dr. Daniel Driscoll and Robert Lutz were re-elected to the PWSA(USA) Board of Directors. Elected to serve with them for three-year terms are John Heybach and Stephen Leightman. Donald Armento was elected to fill the one year remaining of Pam Tolbert's term. Pam resigned because of the increased time constraints of her growing young family.



It's so helpful to bring the children into the program. It's why we are here \* Very overwhelming to see the support, love, dedication and passion of our folks with PWS



Hard-working Conference Team (shown below) earned thanks and praise from all who attended!



# Highlights from Scientific Day 2004 Ohio Conference

**Chairs: Suzanne B. Cassidy M.D. and Shawn McCandless M.D.**

*Editor's Note: The following is Part 1 of two parts, excerpted from abstracts presented at Scientific Day. A full copy of the Scientific Conference Abstracts booklet is available through PWSA(USA) at a cost of \$10.*

## **Gene expression studies in Prader-Willi and Angelman syndromes**

Merlin G. Butler<sup>1</sup>, Douglas C. Bittel<sup>1</sup>, Nataliya Kibiryeva<sup>1</sup>, Zohreh Talebizadeh<sup>1</sup>, and Daniel J. Driscoll<sup>2</sup>: <sup>1</sup>Section of Medical Genetics and Molecular Medicine, Children's Mercy Hospitals and University of Missouri-Kansas City School of Medicine, Kansas City, MO; <sup>2</sup>Department of Pediatrics, University of Florida College of Medicine, Gainesville, FL

The genetic subtypes of PWS and Angelman syndrome S are known to have specific clinical phenotypes. It is unclear how subtle changes in gene expression due to the loss of both imprinted genes and reduced expression of nonimprinted genes lead to the clinical features associated with PWS and AS. In our continuing efforts to understand the molecular genetics of these disorders, we generated a custom cDNA microarray representing 77 genes/transcripts from the 15q11-q13 region. We used our custom microarray to compare gene expression from actively growing lymphoblastoid cell lines established from male subjects.

We identified and confirmed several paternally biased genes/transcripts which had significantly greater expression from the paternal allele. This group of genes/transcripts included the GABA receptor genes, GABRA5 and GABRB3. GABA is an inhibitory neurotransmitter that is widely distributed throughout the central nervous system. GABAergic mechanisms have been implicated in a number of symptoms associated with PWS. Presumably, even a modest decrease in the synthesis of these receptor proteins could have a significant effect on brain development, with permanent consequences on central nervous system function.

Our findings suggest multiple layers of complex control of gene expression on chromosome 15. Interconnected mechanisms can produce subtle and unexpected changes in gene expression when perturbed by the different mechanisms causing the genetic subtypes found in PWS and AS. The resulting alterations in gene expression may help to explain, at least in part, the phenotypic differences observed among the genetic subtypes.

## **Basis for neonatal failure to thrive in a PWS mouse model**

Mihaela Stefan<sup>1</sup>, Hong Ji<sup>5</sup>, Kentaro Yamasaki<sup>1</sup>, David E. Cummings<sup>6</sup>, Klaus H. Kaestner<sup>2</sup>, Rexford S. Ahima<sup>3</sup>, Mark I. Friedman<sup>5</sup>, Irena Suponitsky-Kroyter<sup>4</sup>, Rebecca A. Simmons<sup>4</sup>, and Robert D. Nicholls<sup>1,2</sup>: <sup>1</sup>Departments of <sup>1</sup>Psychiatry, <sup>2</sup>Genetics, <sup>3</sup>Medicine and <sup>4</sup>Pediatrics, University of Pennsylvania, Philadelphia, PA; <sup>5</sup>Monell Chemical Senses Center, Philadelphia, PA; <sup>6</sup>Department of Medicine, University of Washington, Seattle, WA.

PWS results from loss of function of several clustered, paternally expressed genes in a 1.5-Mb region of chromosome 15q11-q13. Most of the primary PWS-region genes appear to have nuclear RNA regulatory functions, suggesting that multiple genetic pathways could be secondarily affected in PWS. To identify the basis of PWS, we characterized a PWS mouse model with an equivalent 4-Mb deletion that has severe failure to thrive and shares similarities with the first stage of the human syndrome.

Our data suggest a widespread abnormality in energy metabolism with severe shortage in available fuel supplies that may underlie the neonatal failure to thrive phenotype of the deletion PWS mouse model.

To identify downstream genetic pathways controlled by the PWS genes, we performed microarray analysis of brain and liver mRNA from P1 PWS vs. WT mice. Surprisingly, the same genes were upregulated in an equivalent manner in brains of mice with Angelman syndrome (AS), which results from the same deletion but of maternal origin. These findings suggest a model in which the *trans*-effect is due to decreased expression of a non-imprinted gene within the PWS/AS mouse deletion.

In conclusion, despite the severe neonatal phenotype of the PWS mouse model, our data suggest that the imprinted PWS loci do not regulate transcript levels but may regulate mRNA modification or structure.

## **Zebrafish: a model for the neurodevelopmental causes of PWS?**

Jennifer L. Unger and Eric Glasgow, Department of Neurobiology, Northeastern Ohio Universities College of Medicine, Rootstown, OH and the Graduate Program in Cellular and Molecular Biology, School of Biomedical Sciences, Kent State University, Kent, OH

Zebrafish are an ideal model for many aspects of brain development because of the many experimental advantages of this animal. These advantages include the ability to

perform large-scale genetic screens, targeted loss and gain of function assays and embryological manipulations. We have characterized the development of isotocin producing cells in zebrafish as a paradigm for the molecular control of hypothalamic development. Zebrafish isotocin is orthologous to mammalian oxytocin. The results of these experiments demonstrate that the genes controlling isotocin cell development are structurally and functionally conserved between zebrafish and mammals.

In PWS there is altered development of the hypothalamus through a reduction in both the total number of cells and oxytocin-containing cells in the paraventricular nucleus. The transcriptional regulatory gene *sim1* is required for oxytocin cell development in mice. Likewise, we have shown that *sim1* is required for isotocin cell development in zebrafish. It is important to note that haploinsufficiency of *sim1* leads to obesity in humans, further linking the molecular control of oxytocin cell development to PWS. Further re-search is needed in order to understand the underlying molecular genetic basis of oxytocin/isotocin cell development. Understanding the molecular genetic control of isotocin cell development will identify candidate genes and pathways involved in PWS, and perhaps lead to novel therapeutic approaches to this disorder.

## **Developmental neuronal abnormalities in mouse models of PWS**

Rachel Wevrick<sup>1</sup>, Syann Lee<sup>1</sup>, Christine L. Walker<sup>1</sup>, Barbara Karten<sup>2</sup>, Alysa A. P. Tennese<sup>1</sup>, Sharee L. Kuny<sup>1</sup>, Silvia Pagliardini<sup>3</sup> and John J. Greer<sup>3,1</sup>: <sup>1</sup>Department of Medical Genetics, University of Alberta, Edmonton, Alberta, Canada and <sup>2</sup>Centre for Neuroscience, Department of Physiology, University of Alberta, Edmonton, Alberta, Canada.

Only a few genes are functionally inactivated in individuals with PWS, although the typical 15q11-q13 deletion extends over several Mb. Most PWS candidate genes are conserved in mice, and all are located in an orthologous region on mouse chromosome 7C. Simultaneous inactivation of PWS-equivalent genes in the mouse is usually lethal in the first postnatal week, because of failure to thrive. Several mouse models in which one or more of the PWS candidate genes are silenced partially recapitulate various aspects of the PWS phenotype. The analysis of expression patterns of murine orthologues of human disease genes is valuable for identifying sites of gene

*Highlights continued on next page*

expression that correlate with disease phenotype.

We now concentrate our studies on two related PWS candidate genes, *Magel2* and *Ndn*. *Necdin* has been proposed to act in the survival of post-mitotic neurons, perhaps supporting the maintenance of the postmitotic state and/or in preventing apoptosis. New-born pups from several strains of mice with a paternally inherited *necdin* deletion succumb to respiratory insufficiency. Observations suggest that the developing respiratory center is particularly sensitive to loss of *necdin* activity.

To further define the nervous system defect in the *necdin*-null mice, we analyzed embryonic brain development in the *necdin*-null mice using a combination of microarray-based expression analysis, immunohistochemistry with neuronal markers, and RNA in situ hybridization. Together these changes point to a possible disruption of cytoskeletal elements in neurons and glia, which could together lead to a defect in axonogenesis.

As *necdin* is expressed in neurons, we hypothesized that changes in gene expression may be due to changes in neuronal identity, development or differentiation. We identified molecular interactions between *necdin* and proteins important in axonal outgrowth and integrity. Data point to a novel *necdin*-mediated intracellular process essential for neurite outgrowth in the central nervous system. We now propose a model whereby defects in axonal elongation and fasciculation may also contribute to the phenotype in PWS.

Supported by Canadian Institutes of Health Research and March of Dimes(USA).

#### Ghrelin levels in children with PWS less than six years of age

Christine R. Erdie-Lalena M.D.<sup>1</sup>, Vanja A. Holm M.D.<sup>3</sup>, Pat Kelly D.O.<sup>1</sup>, David E. Cummings M.D.<sup>4</sup>, R. Scott Frayo<sup>4</sup>, and Troy H. Patience<sup>2</sup>; <sup>1</sup>Department of Pediatrics, Division of Genetics and Development and <sup>2</sup>Department of Clinical Investigation, Madigan Army Medical Center, Fort Lewis, WA; <sup>3</sup>Center of Human Development and Disability, University of Washington, Seattle WA; <sup>4</sup>Division of Metabolism, Endocrinology and Nutrition, Department of Medicine, University of Washington, Seattle WA

PWS is the most common genetic obesity syndrome. An intriguing progression from failure-to-thrive to morbid obesity occurs between 2 and 6 years of age in these patients. Ghrelin is a hormone mainly produced in the stomach; which, when given to humans, stimulates appetite and increases food intake. Levels of ghrelin are significantly increased in adults with PWS. Fasting ghrelin levels have been found to be 3- to 4-fold higher in children 5 to 15 years of age with PWS when compared to obese controls. Information on ghrelin levels in younger children is not available in the literature.

**Results:** There were 14 patients - 7 with PWS and 7 healthy controls. In the

PWS group, all patients had positive methylation studies. In PWS, fasting ghrelin levels were not significantly different compared with age/BMI-matched controls; however, statistical significance is approached ( $P=0.07$ ) when comparing only the UPD PWS. There was no correlation between age and ghrelin levels in either group.

**Conclusions:** Unlike adults and older children with PWS, elevation in total ghrelin level was not found in seven non-obese children less than 6 years of age with PWS when compared to age mates. The reason for this discrepancy is unclear. The sample size is small and we are presently planning to increase the size of the study. Elevated levels of ghrelin in obese persons with PWS is an important finding that, if treatable, promises a better outlook for the quality of life for individuals with this condition. Additional studies in younger children with PWS are needed to better understand ghrelin's role in appetite and eating behaviors in PWS.

#### Somatostatin infusion corrects hyperghrelinemia without reducing appetite in adults with PWS

Anthony P. Goldstone<sup>1,2</sup>, Mike Patterson<sup>3</sup>, Mark Vanderpump<sup>5</sup>, Nila Kalingag<sup>2</sup>, Bernard Khoo<sup>3</sup>, Audrey E. Brynes<sup>4</sup>, Mohammad A. Ghatei<sup>3</sup>, Stephen R. Bloom<sup>3</sup>, Ashley B. Grossman<sup>2</sup>, Márta Korbonits<sup>2</sup> & Tricia M-M. Tan<sup>5</sup>; Division of Pediatric Genetics, University of Florida College of Medicine, Gainesville, USA; <sup>2</sup>Dept. of Endocrinology, St. Bartholomew's Hospital, London, UK; <sup>3</sup>Depts. of Metabolic Medicine and <sup>4</sup>Dietetics, Imperial College School of Medicine, Hammersmith Hospital, London, UK; <sup>5</sup>Dept. of Endocrinology, Royal Free and University College Hospital Medical School, London, UK.

PWS is characterized by severe childhood-onset hyperphagia, obesity and, uniquely, high plasma levels of ghrelin, the orexigenic gastric hormone, which has been postulated as contributing to their hyperphagia. We have found that fasting ghrelin levels are 2.0- to 2.2-fold higher, and post-prandial ghrelin levels 1.8-fold higher, in PWS ( $n=10-27$ ) than non-PWS adults ( $n=17-32$ ), adjusting for % body fat. At least part of the hyperghrelinemia in PWS may be explicable by their relative hypoinsulinemia, since insulin lowers plasma ghrelin. Fasting ghrelin levels were 1.3- to 1.8-fold higher, and post-prandial ghrelin levels 1.2- to 1.5-fold higher in PWS than non-PWS adults, adjusting for insulin levels or insulin sensitivity. Hyperghrelinemia is not seen in patients with hypothalamic obesity from craniopharyngioma.

Somatostatin suppresses ghrelin secretion in normal subjects. We therefore examined the effect of somatostatin on plasma ghrelin and appetite in four male PWS adults fasted overnight in a double-

blind, placebo-controlled, randomized crossover study. Despite somatostatin lowering fasting plasma ghrelin by  $60 \pm 2\%$  ( $p=0.04$ ) to levels seen in non-PWS men, there was no associated reduction in food intake. Somatostatin also lowered plasma PYY levels by  $45 \pm 16\%$  ( $p=0.04$ ), and produced post-prandial hyperglycemia ( $p=0.04$ ). In a separate study, there was no reduction in fasting or post-prandial plasma levels of somatostatin or PYY in PWS adults, but pancreatic polypeptide secretion was impaired.

We conclude that either (i) hyperghrelinemia may not contribute to hyperphagia in PWS adults; (ii) the persistent orexigenic effect of underlying hypothalamic or other hormonal abnormalities in PWS, which impair satiety, override changes in ghrelin; or (iii) concomitant reductions in anorexigenic gastrointestinal hormones, such as PYY, by somatostatin counteracted any anorexigenic effect of lowering orexigenic ghrelin. Somatostatin analogues may therefore not be an effective therapy for hyperphagia in PWS. However larger chronic studies with long-acting somatostatin analogues will be needed to determine their benefits and risks in treating PWS obesity.

#### Effects of growth hormone on sleep patterns in PWS

Jennifer Miller<sup>1</sup>, Janet Silverstein<sup>1</sup>, Abby Wagner<sup>2</sup>, and Daniel J Driscoll<sup>3</sup> Department of Pediatrics, Divisions of <sup>1</sup>Endocrinology, <sup>2</sup>Pulmonology, and <sup>3</sup>Genetics, University of Florida College of Medicine, Gainesville, FL.

Children with PWS have significant sleep-related respiratory problems, including obstructive sleep apnea (OSA), central sleep apnea (CSA), abnormal arousal, and abnormal cardiorespiratory response to hypercapnia. Polysomnographic studies (PSS) of PWS patients show that these respiratory problems are unrelated to weight, tonsillar/adenoid size, or evidence of airway obstruction.

Treatment of PWS patients with recombinant human growth hormone (rhGH) has been shown to improve CSA and increase the respiratory response to hypercarbia. However, recent reports document that nine PWS patients have died suddenly during sleep soon after starting rhGH treatment. Although it is uncertain whether the deaths were related to rhGH therapy, a postulated mechanism is increased OSA due to IGF-1 mediated hypertrophy of the tonsillar/adenoid tissue. While increases in OSA have been seen in non-PWS rhGH-treated patients, no studies have documented the incidence of OSA in PWS patients before and after rhGH treatment. This study was undertaken to evaluate the effect of rhGH on OSA and ventilatory drive in patients with PWS.

Highlights continued on page 15

## *Balancing the Needs of Siblings... Our Family's Experience*

*By Mary Hill*

Our oldest son, Abraham, was only two when Oscar was born and diagnosed with PWS. I distinctly remember grieving for him — he was not getting the brother he imagined any more than I was getting the child I had imagined. But Abraham did not know that yet — he did know that Oscar's muscles were weak and that he would need extra help to get stronger. We told him it would take longer for Oscar to learn to walk and talk but that someday he'd be able to do all the same things Abraham could do. This simple explanation was perfect for a 2 year old, and Abraham went about loving Oscar and not thinking too much about PWS.

As Abraham has gotten older — he's now almost 6 — we have had to slowly adjust how we talk about PWS and how we balance his needs. I think of it as another aspect of the syndrome that needs managing. Our goal, and sometimes we are better at achieving this than other times, is to attend to Abraham's needs just as we attend to Oscar's need for PT, OT and speech. Here are some of the things we've noticed with Abraham over the past 3+ years and how we've approached them.

As a toddler, Abraham would often accompany me to Oscar's therapies and would, quite honestly, have a great time. At times I could tell he was sad that the therapists weren't as excited to play with him as with Oscar. But most therapists would also find ways to include Abraham or bring him a special toy to play with. And if they didn't, I'd try to schedule that therapy at a time when Abraham was in preschool.

As both boys got a little older, I could see Abraham making more comparisons between the younger siblings of his peers and Oscar. Abraham would often delight in playing with these other babies — they were much more active and responsive than Oscar was early on. We tried to acknowledge the differences when Abraham would talk about those children. Often we would say things like, "You're right, Teddy can crawl already. His muscles are strong. Oscar will crawl too. He just needs more time to get strong."

If Abraham was really frustrated, I would often also say, "I'm sad/angry/frustrated about that too. I wish Oscar were stronger. It's ok to be sad/angry/frustrated. It doesn't mean we don't love Oscar." We were trying hard to teach Abraham that his feelings, whatever they were, were ok to talk about... and that we shared some of the same feelings of anger and frustration too.

About a year ago, when Abraham was 4 ½, he started to withdraw a little and often complained about stomach aches. When playing with Oscar, he would sometimes show minor signs of aggression. Though he is very articulate I think he was holding in a lot of feelings and thoughts that he did not understand and was afraid to talk about. Oscar was definitely getting a lot of attention and starting to get into Abraham's toys. He was walking, beginning to talk, and



Abraham, 5½, with his brother Oscar, 3½, who has PWS

turning into a very charming 2 year old. But his communication was lagging, making it hard for Abraham to play with him.

We did lots of things to try to help Abraham through this frustrating period. Abraham and I had a weekly date which we called "Mommy and Abraham Special Time." As stressed as I often was about Oscar and his care, I tried to make that time all about Abraham. We would be silly, talk about anything he was interested in, browse the local bookstore. Because Oscar needed to be watched pretty carefully, and because I always felt that I needed to be stimulating Oscar when with him, both Abraham and I felt "free" when it was just the two of us.

Our café time was also an opportunity to let have Abraham have a treat. When Oscar was born our whole family switched to healthier eating habits, and as a result we rarely have non-fruit desserts. Abraham, by age 4½, was noticing that many of his peers had more cookies and other treats and I think this was adding a little to his feelings of resentment. I try never to say things like "Oscar's not here so we can have a cookie," but even Abe notices that those rare treats are usually without Oscar. When he asks, I say that it's ok for our bodies to have sweets once in a while, but Oscar's body can't handle as many treats. It's a fine balance between being honest with Abraham, not depriving him, but also trying not to add to any resentment he may have toward Oscar.

Abraham always had a lot of trouble with separation, but his anxiety worsened, and dropping him off at preschool

*Siblings continued on next page*

became very difficult when he turned 5. At times he seemed withdrawn and depressed. We tried always to acknowledge his feelings, often having to guess, but gave him lots of opportunities to talk, in an attempt to lay down the groundwork for a future of open communication.

About a year ago, we met another family in our neighborhood who has a child with a disability. Abraham hit it off immediately with their typically developing son Oliver, and slowly realized, even if he couldn't articulate it, that Oliver's older brother Eli, age 6, who he also considered his friend, had a disability too. In some ways, I think this normalized our family, and even if Abraham couldn't express it, I think he felt less "alone." (I know I did!) Abraham and Eli would often play together, especially if Oliver wasn't home. But when Oliver, Eli, and Abraham played together, Abraham's aggression would often surface toward Eli. Jennifer (Eli and Oliver's mom) and I often speculated that Abraham's aggression toward Eli stemmed in part from his anger about Oscar having PWS. We talked a lot with Abraham about treating people with respect and rewarded him with praise when he was respectful of other

**I knew recently that we were on the right track with Abraham when he felt comfortable enough to voice some big fears about the baby we are expecting this fall.**

kids, especially Oscar and Eli. This tactic helped a little, but the underlying feelings of anger still surfaced in inappropriate ways from time to time.

One day, as Eli arrived for a play date with just Abraham, Abraham came running down the hall yelling, "Mom, there's something WRONG with Eli! He can't run fast. And I'm bigger and he's almost 7!" Abraham was clearly upset, almost panicked with the realization that he was right about Eli being different. And he had finally found the words to describe *how* Eli was different.

We were both happy for the clear opportunity to talk to Abraham. Jennifer immediately took Abe aside and explained that he was right, Eli has Down syndrome. She told Abe she wanted to spend some time talking about Down syndrome with him. But for that afternoon, she asked Abe to try to just be Eli's friend.

Abe's anxiety subsided as soon as his observations and feelings were validated, and Abraham and Eli had a great afternoon together. Once Abraham's fears were acknowledged, he was able to relax and really enjoy playing with Eli. If I didn't realize it already, it became clear in this moment that the best path forward with Abraham would be to make sure he had opportunities and felt safe voicing his feelings and concerns.

Now, at almost 6, we still have "Mommy and Abraham Special Time" which is now an entire morning. Paul finds days when he can take extra time and ride bikes with Abraham to preschool, something they both love to do. And we also try to make sure Abraham gets lots of opportunities to play with typical peers.

And, finally, we try not to put Oscar's therapy needs over Abraham's typical kid needs. We've also decided to send him to private school for kindergarten in the fall. His new school specifically teaches that each person has gifts to offer, each person learns differently and that we all have our strengths as well as our weaknesses, and that is ok. This philosophy is an extension of our family philosophy and I think Abraham will thrive there.

I knew recently that we were on the right track with Abraham when he felt comfortable enough one night to voice some big fears about the baby we are expecting this fall.

Both Abraham and Oscar had been showing a lot of excitement about the baby, but one night Abraham seemed angry about having a new baby. He told me he didn't want the baby to come, and then he asked through his tears if the baby was going to have Prader-Willi syndrome.

That was the first time he ever referred to the syndrome by name. I could tell he was upset. I was honest — I told him we were worried about that too, but I was able to reassure him (since we had actually just completed the testing) that no, this baby would not have PWS. He thought about that for a moment and then said, "What about Down syndrome?" And his final question was, "So the baby will be like me then, Mom?"

I was stunned by how much he already understands about the risk of having a baby. But I was also just so glad he knew it was ok to ask these questions. And, finally, I realized what a burden Abraham already carries even though Oscar is now a very active, talkative, interactive little guy with whom Abe absolutely *loves* to play.

Abraham is clearly working through some feelings of grief, and we are doing our best to help him. I never thought I'd be relieved to hear him say, "I hate Prader-Willi syndrome. I'm *angry* that Oscar has Prader-Willi syndrome."

Me too, Abraham, me too.

I know that saying it and knowing it's okay to say it helps Abraham let go of some of that anger and frustration, leaving more room for the enormous love he feels for Oscar. And I also know that if we continue to meet Abraham's needs as a child and a sibling, as well as acknowledge some of those scarier feelings, he will always feel that same strong, brotherly bond that he so clearly has felt since Oscar was born.

PWSA (USA) gratefully acknowledges the printing and mailing of this newsletter is made possible by a grant from CIBC World Markets Corp./Miracle Day USA

# We Remember...

Every person has something special to offer this world — and we, along with their families, want to share who they were and what they meant to the people who loved them.

## Charles “Clyde” Walden Mays

Clyde Mays, 24, died July 22, 2004. He was the only child of Hope Mays, the long-time executive director of our Georgia chapter. Clyde’s dad died 12 years ago of brain cancer, and Hope dedicated her life to Clyde. A memorial service to celebrate his life was held on July 24 in Mountain Park, Georgia.



In 1999, Clyde’s accomplishments were featured in a front-page article in *The Gathered View*. In his essay application for membership into the National Honor Society, Clyde wrote: “I believe I have the qualities of service, leadership, and character that are required to be in the National Honor Society. I have learned in service work to help others. I have learned in leadership to set a good example. I have learned that having a good character means having a good attitude and self-esteem, no matter what names people call me. I want to join the National Honor Society so that I can be a good example to students at RHS.”

In spite of all of his challenges, Clyde was slim and healthy, running in the world’s largest 10K, Atlanta’s Fourth of July Peachtree Road Race, for the past 4 years. He was inducted into the National Honor Society and Beta Club, volunteered for the fire department and coordinated annual Earth Day activities in his community. He graduated from Roswell High School in 2001. Clyde was a remarkable young man and was indeed a “good example” and an inspiration to all who knew him.

“No one had a bigger heart than Clyde,” said State Sen. Tom Price of Roswell, who was team doctor of the Roswell High School football squad at the same time Clyde was its student manager. “Clyde gave his all to everything he did.”

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## The Sunshine Kids - continued from page 1

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At age 39, Lisa now lives in a group home with three other women and works in the community through the guidance of a sheltered workshop. She looks forward to a weekend a month and special trips for family celebrations and holidays, when she comes “home” just like our other children.

When your child is young, I believe in facing the reality of the syndrome. Your child can accomplish goals despite “differences,” even if he or she isn’t one of the sunshine kids.

*Marge Wett was PWSA(USA)’s first executive director. She and her late husband, Richard “Dick” Wett, M.D., devoted years to establishing the early PWSA organization.*

“Clyde was a hugely enthusiastic advocate for people with disabilities, especially their wish to live independently,” said Linda Pogue of Atlanta, adviser to People First, a self-help organization.

“At his high school commencement, when his name was called to receive his diploma, the other 400 or so graduating seniors rose and gave Clyde a standing ovation,” said his grandmother, Patricia Walden of Macon, Georgia.

Dale Cooper wrote, “He always called me ‘Mister Cooper’.... Now with Clyde gone, I feel very cold, and alone. Special spirits live in our souls and Clyde Mays lives in mine.... I suggest you fall in love with a syndrome and their victims, and there are many. I would invite you to fall in love with Prader-Willi syndrome. It will enrich your life.”

His mother said that over the last few years, Clyde had accomplished three of his life-long goals: to “have a real job” as a school custodian, to live “on his own” in a community-based group home, and to become engaged to his high-school sweetheart. He lived a very full life and had an impact on many people.

We are grateful that Hope felt it was important to have an autopsy and to have brain tissue donated. This can be difficult to acquire in a timely manner but crucial to PWS research. With this legacy, Clyde will continue to impact many.

— Janalee Heinemann

## On The Wings Of Angels

Special thanks to Dotty and Dale Cooper for donating their frequent flyer miles so I could attend Clyde Mays’ memorial service. I really wanted to be able to attend, since we have all been in the PWS trenches together for 20 years. The Coopers arranged the flight, housed and fed me. I was also able to spend time with their delightful daughter, Shawn, who is a member of our PWS Advisory Board.

Most of my trips have been funded by outside sources: Taiwan, by their Rare Disorder organization, Israel by the Israel PWS Association, Pharmacia formerly funded medical awareness booths, PWS families around the world feed and house us, and I pay some of my own expenses. In 2003, we only used \$143 from operating funds for all of my travel!

You can be our “angel’s wings” by donating your frequent flyer miles as we work to help educate people across the world. We know that setting up awareness booths at major medical conventions, distributing educational materials and answering questions at these events has greatly increased awareness and early diagnosis of PWS.

— Janalee Heinemann

## Contributions In Memory Of

### Ferdaus Albukhari

Pediatric Associates of West Essex

### Margaret Bailey-Schwartz

Patrick & Maryanne Patten

### Eileen Bringman

Mr. & Mrs. Kenneth Foster

Niel & Cindy Haseley

Casmer & Dianne Hill

Carl & Elizabeth Meyers

### Harold Carlson

Mary Ingalls

### Grandma Collier

Kenneth & Catherine Smith

### Vagn Blicher Dam

The Antioch Company

### Renee Davis

Melvin & Schelley Childress

### Angelo DeBernardis

Brenda Lidwell

### Goldie Effune

Richard Abrams

Albert & Marcia Bryant

### Jerri Evetts

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## Highlights - continued from page 11

**Methods:** PSS was performed in a standard sleep laboratory with 23 PWS patients. Studies were evaluated for evidence of sleep-disordered breathing (SDB) including CSA, central hypopnea, OSA, obstructive hypopnea, hypoxemia, and hypercarbia.

**Discussion:** All patients with PWS studied had some degree of OSA and CSA as a baseline. rhGH did not worsen obstructive events, central events, or duration of events in the majority of patients. rhGH decreased the number and duration of obstructive events in several of our patients, with no significant effect on central events. rhGH treatment improved SDB during REM in all but one of our patients when IGF-1 levels were normal. Our results suggest that elevated IGF-1 levels may worsen obstructive sleep disturbances, necessitating close follow-up of sleep studies and rhGH dose. It is unclear at this time which patients may have high IGF-1 levels on the recommended starting dose of rhGH, so is important to monitor IGF-1 levels routinely in this patient population.

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