November-December 2004
Volume 29, Number 6

The Gathered View

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

In this issue

PWS 'Pioneers' Can Offer Insight to Parents

By Dottie Cooper

Many years have passed since I first heard the words Prader-Willi syndrome. "What is that?" I asked, and all too soon, I knew.

Although my daughter was already 11 and I had known we were dealing with a difficult situation from her birth, it was still such a mixed blessing to get the diagnosis. Back then, there was not as much hope and promise for the future as we have now, and, while I felt a sense of relief to finally know it had a name, I also felt a sense of dread for what was to come in all our lives as we struggled together to forge into the future.

Shawn Cooper, 31, is a member of the PWSA (USA) Adults With PWS Advisory Board

What I remember most, however, about those days of receiving the diagnosis was that there was some other family in the whole world who was experiencing the same thing — who would understand the behaviors and challenges we had and most importantly, would be able to look beyond the "weirdness" to find the truly loving and caring person she is. For us, nothing in the world was more important at that time than to meet that family, or families, who would understand.

I was fortunate to live in Birmingham, close enough to go to Atlanta to meet the Witts, the Maurers and Dr. Cassidy at their semi-annual board meeting in January, and then many more families at the annual PWS conference the following summer.

From them I learned tips on how to deal with the challenges, and I found myself drawn to families whose children were older than mine so that I could draw from their wisdom and lessons learned. It was invaluable, and I would talk and ponder what I could do to improve my daughter's life and those of other families, about when she was out of school, considering a group home, or a job?

I have reached many of those goals now, and it would have been infinitely harder without the coaching and mentoring I received throughout the many years. To this day I still look up to and receive help from those who have been where I am now in the progression of life with PWS.

I've noticed that we are losing the interest and participation of so many parents who were once very involved with PWS. It seems that once things begin to stabilize and our adult with PWS has successfully moved into a (somewhat) routine period, it is all too easy to move away from the PWS association support system that sustained us in more trying times.

It is not good enough for us to sit back and think, "We had the need to actively participate in the association when our children were young, we have had our turn," and walk away. Our PWS association suffers when the collective wisdom of so many parents who have so many "lessons learned" is lost.

Pioneers continued on page 11

Prader-Willi Syndrome Association (USA)

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Our Mission: Through the teamwork of families and professionals, PWSA (USA) will improve and enhance the lives of everyone impacted with Prader-Willi syndrome (PWS) and related conditions.

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Deadline to submit items for upcoming issues of *The Gathered View*
Jan/Feb: Dec 1; Mar/Apr: Feb 1; May/Jun: Apr 1;
Jul/Aug: Jun 1; Sep/Oct: Aug 1; Nov/Dec: Oct 1

Check our PWSA (USA) web site *Members Only* section for Special Opportunities Limited to Members: www.pwsausa.org
User Name: members; Password this issue is HOLLY
If asked for Domain Name it is CIMCO

National & Chapter View

We boarded up the PWSA(USA) office four times this hurricane season! Fortunately, Sarasota was spared major damage from the storms, just the usual clean-up of leaves, tree limbs and water in the streets plus temporary power outages. We're happy to report that the sun is now shining and the beach is still beautiful!

While Floridians might "exhibit a slight twitch when introduced to anyone named Charley, Frances, Ivan or Jeanne," according to their newsletter, the **Florida** Chapter came through the hurricanes still planning for their October Fall weekend Conference in Gainesville. Go Florida!

The Florida newsletter also reported that you "might be a Floridian if….your street has more than three "No Wake" signs posted!" Many communities in Pennsylvania could say the same. At least one family from the **Pennsylvania** chapter was flooded out of their home by Ivan. We wish them better days ahead.

Congratulations, **John and Debbie Stallings,** new co-presidents of the Prader-Willi **Florida** Association! Their son Josh with PWS, age 18, was diagnosed at age 3 and, they say, "has grown into a handsome young man." Members of the Florida chapter for 12 years, John and Debbie had the huge job for several years of being responsible for the YAAP program at national conferences.

— Lota Mitchell, Associate Editor

Happy Holidays to all our Angels

We wish Tori Browning,3, of The Woodlands, Texas, who has PWS, and our PWS families a happy holiday season and a New Year of peace and prosperity.

New E-mail Group Forming for Those With PWS

A new e-mail group for people who have Prader-Willi syndrome (PWS) is being sponsored by Prader-Willi Syndrome Association (USA). It provides a method to communicate with each other. Participants in this eGroup provide support to each other, share information and discuss issues related with having PWS. No one need feel alone.

This PWS eGroup list is ONLY for people who have Prader-Willi syndrome, have their own email address and can type.

To subscribe to this list, go to http://groups.yahoo.com/group/PWSA-USA-PWS/.

Nominees wanted for PWSA(USA) Board of Directors

Who can be a Board member?

Any member of PWSA(USA), which includes parents, caregivers, adult relatives, grandparents and professionals.

Who is qualified to be on the Board of Directors?

Anyone willing to volunteer time, talents and expertise to support the mission of PWSA(USA).

How many seats are open and what is the term of office?

There are 12 board members, and typically 4 are elected each year. This year 4 seats are open. Each Board member serves a 3-year term and can run for 3 terms consecutively.

What are the expectations of a Board Member?

Board Members are required to attend two board meetings a year: at the annual PWSA (USA) conference and in January at the PWSA (USA) national office in Sarasota, Florida, plus offer their expertise by serving on at least one committee.

What are the responsibilities?

Conference calls, e-mail and mail votes are required throughout the year. Each Board Member must serve on at least one of the following committees: Leadership Development, Publications, National Conference, Crisis/ Intervention and Training, Finance, Funding and Grant Development, Board Advisory and Executive.

How to Apply

Names of members interested in or recommended for a seat on the Board of Directors should be submitted to the committee for consideration no later than April 6, 2005.

Recommendations should include a brief description of the member's qualifications to serve on the Board.

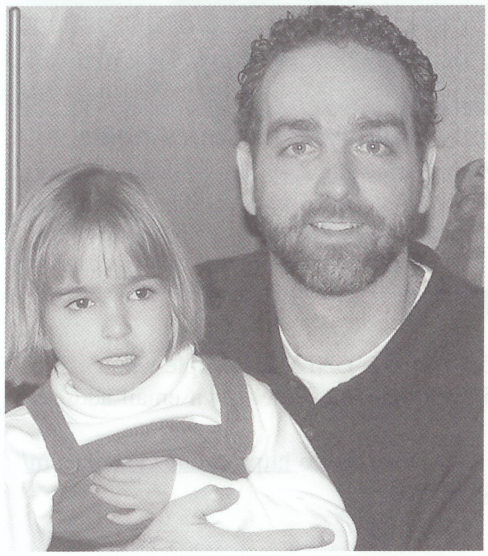
Mail, fax or e-mail recommendations to:
PWSA(USA)
Attention: Carol Hearn, Chair, Board of Directors
5700 Midnight Pass Rd., Suite 6, Sarasota, FL 34242
Fax: (941) 312-0142 or e-mail: *national@pwsausa.org*

President's View

How We're Fostering Research About PWS

Carolyn Loker

The PWSA(USA) Research Committee includes from our Board of Directors Janice Agarwal, Don Armento M.D., Dan Driscoll M.D., John Heybach, Barbara McManus, Carolyn Loker and Rob Lutz, plus David Agarwal M.D., Janalee Heinemann, Jim Loker M.D. and Ken Smith. I've asked Rob Lutz, who chairs this Research Committee, to discuss the efforts of PWSA(USA) to foster research about PWS at the highest scientific levels. Here is his very comprehensive report.



Research Committee Chair Rob Lutz with his daughter Isabel, who has PWS

My daughter Isabel is now close to 5 years old. She was diagnosed with Prader Willi syndrome when she was 5 months old. When my wife Debra and I first learned of her diagnosis and all the potential issues she would be facing, we vowed to do everything we could on her behalf to give her the best possible chance to be as happy and satisfied as any "typical" child. So far she is doing very well and we are cautiously optimistic about the future.

One of the efforts I made to help Isabel was to join the Board of PWSA(USA), and my current role on the Board is as chair of the Research Committee. I am optimistic that by promoting the scientific understanding of PWS and all its implications, we will improve the lives of our children and adults with PWS. It is a slow process, but looking back at the progress already made over time, I believe advances will be achieved that will help Isabel and all those with PWS.

The Research Committee is currently focused on four missions:

- Influencing government funding
- Funding valuable research projects
- Bringing scientists together to promote research
- Leveraging PWSA (USA) information

Influencing government funding

PWSA(USA) believes that the bulk of funding for research on PWS will come from the government. The National Institutes of Health (NIH) and the Centers for Disease Control (CDC), the primary government agencies involved in funding health research grants, are generally supportive of PWS research, especially as childhood and adult obesity have grown into significant national health priorities.

However, we would like for more to be spent. In that regard, Jim Kane (father of Kate, 23) and my father, Dick Lutz, led the effort of PWSA(USA) to join the Coalition for Children's Health which you may have read about in a recent Gathered View. CCH is soliciting government sources to advocate that PWS research at NIH be expanded by almost 30% and new programs be instituted at NIH and the CDC to $30,250,000.

PWSA(USA) is also seeking to influence the approval process for government research funding. For example, Joyce Opp, mother of an infant with PWS, recently introduced her son to U.S. Sen. Arlen Specter of Pennsylvania through a connection to the senator. Thanks to his heightened awareness, Sen. Specter had language inserted into a draft Health and Human Services appropriations bill supporting PWS. Efforts like that help raise the profile of PWS and could lead to greater funding over time. Also, PWSA(USA), working with an outside funding source, is in the process of working out an agreement with a professional consulting firm in Washington, D.C. to increase the scope and professionalism of our Federal advocacy effort.

Jim Kane, coordinator of our Research Advocacy Team, has been leading the effort to make connections like those in Washington and push PWS research funding among those with the power to increase PWS research. If you or anyone you know has any contacts in Washington that could be of value, please tell me (lutzer@comcast.net) or Jim Kane (jgkane@msn.com).

Funding valuable research projects

Even though the largest sources of funding for PWS projects will likely come from the government, we can't rely on independent scientists and the government to pursue all the projects we as parents would want. It is especially difficult to get the government to sponsor early stage research or research very specific to therapies for those for PWS (since PWS is relatively rare).

As an example, PWSA(USA) was concerned that the most efficacious combination of psychotropic medications

Research continued on next page

Be My Valentine...

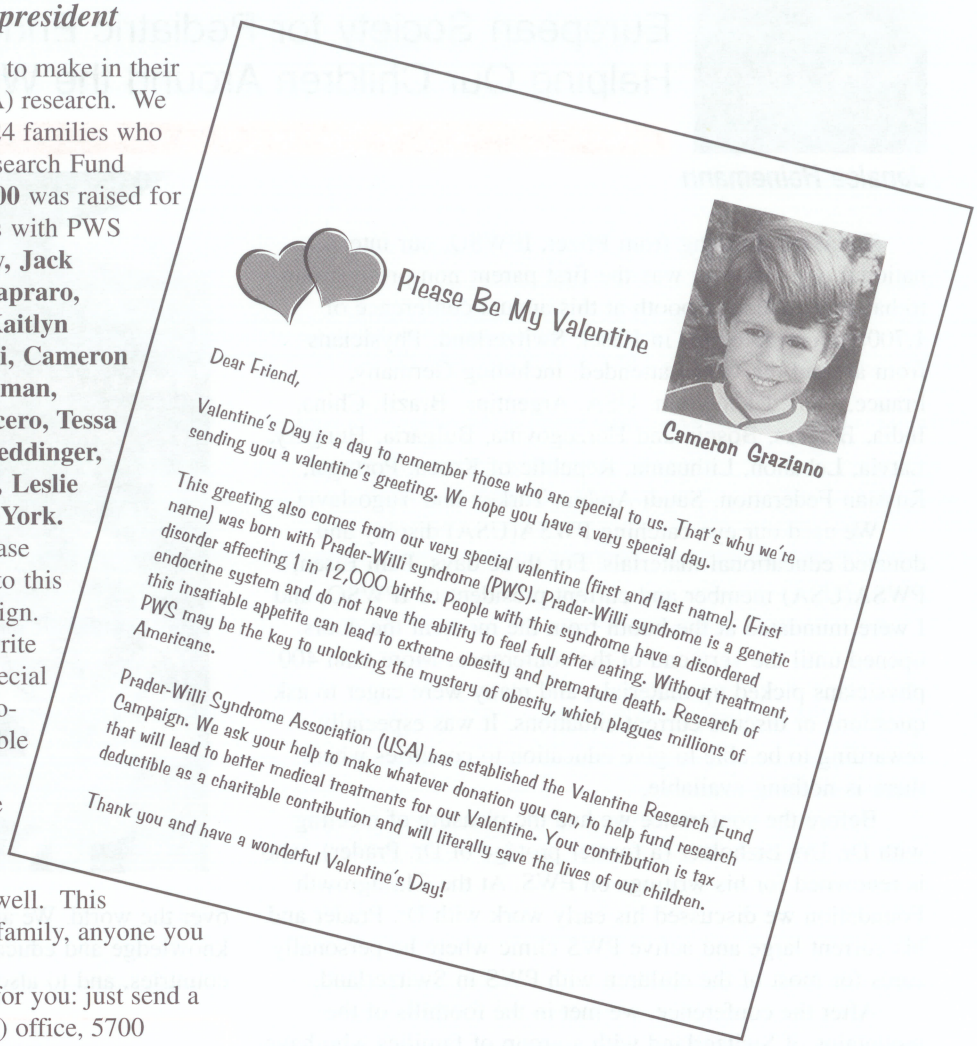
Join Our Annual Valentine Research Fund Campaign

By Carolyn Loker, PWSA(USA) president

What a difference families are able to make in their quest of raising money for PWSA (USA) research. We extend a big "THANK YOU" to these 24 families who participated in last year's Valentine Research Fund Campaign. In 2004 a total of **$33,010.00** was raised for this single event. Our special valentines with PWS were: **Rebecca Baird, Kathryn Baxley, Jack Bevacqua, Victoria Browning, Lea Capraro, Brooke Detiege, Jessika Dickinson, Kaitlyn Disney, Nekoda Erickson, Peter Funai, Cameron Graziano, J. R. Headley, Abby Heathman, Autumn Letzo, Anna Loker, R. J. Lucero, Tessa Pringle, Madison Putnam, Chelsea Reddinger, Tracy Richmond, Breanna Siegfriedt, Leslie Torbert, Clare Wilson, and Sheridan York.**

You too can make a difference! Please consider adding your special valentine to this year's Valentine Research Fund Campaign.

We make it easy for you. We will write the Valentine letter and include your special valentine's picture on it. If you're the do-it-yourself type, the letter will be available on the website beginning in November for you to download and insert a picture of your special valentine. The letter is written so that you may want to ask your family or friends to send it out as well. This letter can be sent to friends, neighbors, family, anyone you feel may want to help.



Please Be My Valentine

Cameron Graziano

Dear Friend,

Valentine's Day is a day to remember those who are special to us. That's why we're sending you a valentine's greeting. We hope you have a very special day.

This greeting also comes from our very special valentine (first and last name). (First name) was born with Prader-Willi syndrome (PWS). Prader-Willi syndrome is a genetic disorder affecting 1 in 12,000 births. People with this syndrome have a disordered endocrine system and do not have the ability to feel full after eating. Without treatment, this insatiable appetite can lead to extreme obesity and premature death. Research of PWS may be the key to unlocking the mystery of obesity, which plagues millions of Americans.

Prader-Willi Syndrome Association (USA) has established the Valentine Research Fund Campaign. We ask your help to make whatever donation you can, to help fund research that will lead to better medical treatments for our Valentine. Your contribution is tax deductible as a charitable contribution and will literally save the lives of our children.

Thank you and have a wonderful Valentine's Day!

If you choose, we can do the work for you: just send a picture of your child to the PWSA(USA) office, 5700 Midnight Pass Rd. Ste 6, Sarasota, FL 34242, Attention: Diane. Tell us how many letters you want and we will scan your picture, print the letters and send them back to you. We will do everything but address them for you – what could be easier?

Deadline to have your pictures and information to PWSA(USA) is January 28, 2005.

Please call Diane at 1-800-926-4797 if you have any questions. Tell Diane if you would like a flyer to post at your workplace bulletin board.

The Valentine Research Fund Campaign is our only fund-raising appeal dedicated solely to research projects; it is separate from our annual Angel Fund Drive, which also supports essential PWSA (USA) operations and programs.

Research - *continued from page 4*

for the various types of PWS and their symptoms had never been studied. The common use of psychotropic medicines on "typical" people does not necessarily apply to individuals with PWS. Unfortunately, we did not believe that the NIH would quickly sponsor a study on that topic. Fortunately, we were able to utilize the Lea Capraro Research Funds donated to PWSA(USA) and designated specifically for psychotropic medication research. This summer we agreed to spend $30,000 to fund such a study. If successful, the project will reduce the amount of experimentation doctors must do to find the best treatment patterns — reducing the time it will take to provide relief to people with PWS and their families.

Another example is The Bear Study — Etiology of Excessive Appetite in Persons with PWS (see Janalee's article in this Gathered View). Although we did not directly fund the project, we were able to recruit the funding from our sources and help identify and find appropriate candidates for the research — as we do for many research projects.

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Janalee Heinemann

Executive Director's View

European Society for Pediatric Endocrinology Conference Helping Our Children Around the World

Thanks to funding from Pfizer, IPWSO, our international organization, was the first parent non-profit group to have an education booth at this annual conference of 1,700 endocrinologists in Basel, Switzerland. Physicians from around the world attended, including Germany, France, United Kingdom, USA, Argentina, Brazil, China, India, Belarus, Bosnia and Herzegovina, Bulgaria, Hungary, Latvia, Lebanon, Lithuania, Republic of Korea, Portugal, Russian Federation, Saudi Arabia, Turkey and Yugoslavia.

We used our eye-catching PWSA(USA) display and donated educational materials. For three days, Pam Eisen, PWSA(USA) member and current president of IPWSO, and I were inundated at the booth from the moment the doors opened until the very end of the conference. More than 400 physicians picked up materials, and many were eager to ask questions or discuss current situations. It was especially rewarding to be able to give education to countries where there is nothing available.

Before the conference we had the pleasure of meeting with Dr. Urs Eizholzer (a former protégé of Dr. Prader), who is renowned for his writings on PWS. At the Childgrowth Foundation we discussed his early work with Dr. Prader and his current large and active PWS clinic where he personally cares for most of the children with PWS in Switzerland.

After the conference, we met in the foothills of the mountains of Switzerland with a group of families who have children with PWS ranging in age from toddlers through adulthood. The enthusiastic group is an obviously well bonded "family" and the children all looked wonderful. All the young children were slim and active and most were on growth hormone.

Andreas Bächli, president of the Switzerland PWS Organization, his wife Doris, IPWSO parent delegate, and their three delightful children graciously opened their hearts and home to give us of the experience of living with PWS in a small Swiss village.

Pictured is their 11-year-old son, Pascal, who has PWS, with his 6-year-old sister Carla. As is obvious, Pascal has been on growth hormone and his parents were educated early on. Doris told us a very poignant story, reminding us of the feelings of siblings. Carla said to her, "Mom, how am I special? Pascal is special because he has PWS, but I do not know what makes me special. Perhaps if I break my arm, I will be special."

We were pleased to be joined by Giorgio Fornasier, IPWSO past president, now its first director of program development. Giorgio debuted the new IPWSO song he composed, "Ich Auch" (Me Too), with lyrics written by our own Pam Eisen. The song was inspired by Annika, daughter of Thomas Gross, who will be coordinating the 2007 IPWSO conference in Germany. The only two words Annika can say are "ich auch". She is reminding people not to leave her out — that she also counts.

Pascal Bächli, age 11, who has PWS, with his sister Carla, age 6

Yes, all of our children count, all over the world. We are grateful that we are able to share our knowledge and educational materials with so many countries, and to also learn from them.

Ich auch

Ich auch, me too, anch'io, yo tambie'n,
dear mama, dear papa help me be me.
Help me to grow, help me to be
part of this world I deserve, you will see.

When I was born You were in despair,
but I was looking for someone to care:
I am your child, I am the same
as all the children so don't be ashamed.

Ich auch, me too, anch'io, moi aussi
dear mama, dear papa what do you say?
"With us you'll grow, be as you are
part of this world that you love with your heart.

When you were born we gave you our love,
you were our angel, a star from above:
I am your mother, I am your dad,
we are so proud of the child that we had.

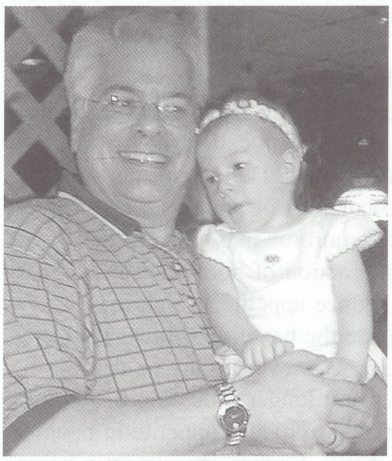
Wir auch...We too!

Fund-Raising

Here's A Win-Win Way To Give

By Stephen Leightman

Last month our new Board of Directors Chair Carol Hearn wrote a compelling article outlining PWSA(USA)'s continuing need for funds. Like many other charitable organizations, our very existence is dependent on the goodwill of our donors. Everyone is grateful for the gifts the Association has received in the past, and the generosity of all who have supported our programs, projects, conferences and counseling is greatly appreciated. Without this help we could not possibly have accomplished as much as we have so far. Despite all that has been done, there is so much more to do, and again everyone's help is needed as PWSA(USA) grows and expands its effort.



Steve Leightman with his granddaughter Josilyn Levine, who has PWS.

The checks received either for Angel Fund, our Valentine drive, in honor or in memory of those directly affected by the syndrome or as part of a fund-raising event are important parts of the budgeting and planning process. There may be other ways to efficiently make tax-deductible donations that will aid our common cause.

Consider the gift of appreciated stock

If you purchased or received a stock worth $1,000 several years ago, it may have grown in value. Assuming it is now valued at $5,000, you *will* incur a capital gains tax when you decide to sell it. If, however, you donate the same shares, worth $5,000 in this example, to PWSA/USA instead, *the Association* can sell it for $,5000 and you receive the following benefits:

- You avoid paying the Capital Gains Tax
- Because you donated to the asset to a *bona fide* charity, PWSA(USA), you can take the full amount as a current tax deduction, subject to certain limits.

PWSA(USA) receives every cent of the amount you intended to donate in cash, and this method benefits you also. The Prader-Willi Syndrome Association(USA), for its part, will not incur a capital gains tax because it is a non-profit organization as defined by Section 501(c)3 of the tax code.

For more information on how to implement this strategy, please call the national office at 800-926-4797 and speak with either Financial Manager Ann Coyne or Business Manager Steve Dudrow.

Note that PWSA(USA) does not give tax advice, and any action you decide to take should be after consultation with your own tax advisor.

Stephen Leightman serves on the PWSA(USA) Board of Directors. He and his wife Michele live in Cherry Hill, N.J.

Grassroots Fund-raisers Net Support for PWSA(USA)

The Donovans Decide to 'Walk-A-Mile' for Riley

Joe and Carmen Donovan are literally going the extra mile for their 3-year-old son Riley, who has PWS.

Beginning on September 4, Riley's birthday, they pledged to walk at least 5 miles a day through December 31, 2004 to raise awareness and support for Prader-Willi syndrome research. The Donovans committed to walk 1 mile for every $5.00 contributed to PWSA(USA) in Riley's name, and set a goal to complete 1,000 miles.

So far, they have raised more than $6,000!

The Donovans distributed a one-page handout about Riley and PWS that included a donation coupon at the bottom. The Donovans' Walk-A-Mile campaign caught the attention of their local newspaper in Montezuma, Iowa, which helped them garner even more support. Carmen told the newspaper that she and Joe were Riley's "best advocates."

Like Carmen and Joe Donovan, you can be the best advocate for your child who has PWS. If you'd like to organize your own grassroots fund-raising effort, PWSA(USA) will help you. Call Steve Dudrow at the PWSA(USA) national office, 1-800-926-4797.

And please let us know about your efforts to raise funds for PWS. Tell us what you did and how it turned out, and include flyers, letters, and newspaper clippings. We want to thank you for your accomplishment, and your ideas might help others who want to try their own grassroots fund-raising. ***— Jane Phelan, Editor***

Carmen Donovan with her son Riley, 3, who has PWS.

Effect of Ghrelin Levels On Appetite Needs Further Study

By Tony Goldstone, M.D., Ph.D.

Reducing high levels of the appetite-enhancing hormone ghrelin with the hormone somatostatin did not reduce the appetite of the four adults with Prader-Willi syndrome participating in this study.

Dr. Tony Goldstone

Ghrelin is a hormone produced principally by the stomach that increases appetite. Levels of ghrelin in the blood are high when fasting and before meals, but fall after eating food, suggesting that changes in ghrelin may normally contribute to changes in appetite before and after eating. People who are obese usually have low levels of ghrelin in the blood.

People with Prader-Willi syndrome develop an uncontrollable appetite from early childhood which persists through the rest of their lives, and can lead to extreme obesity at an early age. Prevention and management of their obesity is vital to avoid complications of obesity, such as diabetes, heart and lung problems.

There are no effective treatments currently available for the insatiable hunger. It was recently found that, despite being obese, patients with Prader-Willi syndrome have very high levels of ghrelin in their blood. The cause for this is unclear. It has been suggested that high ghrelin levels may cause or contribute to their increased appetite, raising the possibility of developing a drug treatment to reduce their hunger. This possibility has been investigated in our research study led by Dr. Tricia Tan, Royal Free Hospital, London, and me at St. Bartholomew's Hospital, London (both of us now working at the University of Florida, Gainesville), and colleagues, using a different hormone that lowers the levels of ghrelin in the blood.

We studied four men with Prader-Willi syndrome. After fasting overnight, they were allowed to eat as many sandwiches as they wanted over one hour, halfway through receiving a slow injection — over 5 hours — of a hormone called somatostatin, which reduces ghrelin secretion. The results were compared to those obtained when the men were given an inactive infusion of water on a separate day under the same conditions. The patients and investigators were not aware on which day the somatostatin or water was being used.

Despite somatostatin lowering the levels of ghrelin by 60 percent to a normal level, there was no reduction of appetite. A similar amount of food was eaten when receiving somatostatin as when receiving water, averaging 14 slices of bread over 1 hour. These results suggest that the high levels of ghrelin in the blood may not be the only, nor even be a significant, factor causing the increased appetite in these patients.

Ghrelin and other hormones produced by the intestine that control appetite are thought to work through a part of the brain called the hypothalamus. Defects in the hypothalamus of patients with Prader-Willi syndrome may prevent the normal action of hormones and signals that reduce appetite, which may override the benefits of lowering ghrelin. However it may be that a larger and longer reduction of ghrelin is needed to reduce appetite in people with PWS. Studies are currently being planned in North America using a

Ghrelin continued on page 13

The Bear Study and Appetite in PWS

If you recall, we were collaborating with, and helped find the funding for, an exciting research project with Ralph A. Nelson, M.D., Ph.D, FACP, former head of the Department of Internal Medicine for the University of Illinois College of Medicine at Urbana-Champaign and current director of Research at Carle Foundation Hospital in Urbana, Illinois. Dr. Nelson is a world-renowned expert on the subject of metabolism in bears and in the area of clinical nutrition.

Dr. Nelson has been studying the denning habits of bears and their physiology over 30 years and has focused on the control of appetite in bears. Bears are remarkable since they are able to go many months without eating or drinking. They prepare for this denning period with several weeks of ravenous foraging and feeding.

Dr. Nelson's research team has been comparing substances that potentially control this process in the blood of both the child/adult with PWS and the bears. Our hope is that this study of bears and people with PWS will be helpful in finding a way to control appetite in those with PWS.

When this comparison was complete, only one protein that appeared in the blood of the anorexic bear did not appear in the blood from subjects with PWS. This means that a particular protein present in black bears allows them to "turn off" their appetite, while subjects with PWS cannot. Because the weight of this protein is close to an established appetite-control protein called Cholecystokinin-8 (CCK-8), Dr. Nelson believes that this protein can be considered an appetite suppression protein.

Because I am under a non-disclosure agreement, I cannot tell you much more at this time, but suffice to say that the sequencing of this protein promises to be of great interest to pharmaceutical companies for future clinical studies.

— Janalee Heinemann
Executive Director

Abstracts from 2004 Scientific Conferences

Huron, Ohio • Chairs: Suzanne B. Cassidy, M.D. and Shawn McCandless, M.D.

Part 2 of two parts, excerpted from abstracts presented at Scientific Day. A full copy of the Ohio Scientific Conference Abstract booklet is available through PWSA(USA) at a cost of $10.

The Therapeutic Role of Growth Hormone Replacement Therapy for Persons with PWS Across the Life-Span

Barbara Y. Whitman[1#],, Myers SE[1*#], Bekx, T[*], Moerchen V[*], Carrel A[*], Allen D[*], Mogul H[#], Frey M[4#], Lee P[#], Pinyard B[#], Zipf W[#]; [1]Dept. of Pediatrics, St. Louis Univ., St. Louis, MO; *Infant study; #Adult study*

Although not curative, recombinant human growth hormone replacement therapy (rhGHRT) can alleviate many of the somatic morbidities associated with PWS. The effects of rhGHRT in children with PWS include acceleration of linear height velocity, optimization of final adult height, increased lean mass, increased bone mineralization, improvements in strength and agility, and possible augmentation of dietary effects on weight control and body fat. Following the initial studies of children ages 4 to 16 years of age, at least two questions regarding opposite ends of the age spectrum emerged: (1) would earlier initiation of rhGHRT in infants and toddlers with PWS lessen body fat accumulation and hasten accrual of lean body mass, perhaps modifying the natural history of the disorder; and (2) could rhGHRT positively and safely alter the body composition abnormalities of adults with PWS previously naïve to rhGH or previously treated adults whose rhGH therapy had been discontinued for greater than one year.

To address the first question, 29 children with genetically confirmed PWS ages 4 to 37 months were enrolled in a 24 month long, two-site, controlled study of the effect of early administration of rhGHRT. Treated subjects caught up to the population mean for length, had decreased percent body fat, and accrued more lean body mass. Further, after 1 year, the treated group demonstrated statistically significant gains in language, cognition and motor development when compared to untreated controls.

To address the second question, a four-site study seeks to enroll 40 genetically confirmed adults with PWS meeting inclusion criteria for growth hormone deficiency. Change in body composition serves as the primary outcome (efficacy) measure. Preliminary 6-month analyses of baseline changes in total body weight, BMI, waist and hip circumference, lipids, and behavioral assessments administered monthly to parents and assistants of the study subjects are in progress. *Major support for both studies was provided by Pharmacia/Pfizer.*

Assessment of sleep disorders with special attention to PWS

Carol L. Rosen, M.D., Pulmonology Division, Case Western Reserve University/ University Hospitals of Cleveland and Sleep Center, University Hospitals of Cleveland. Summary by Shawn E. McCandless, M.D.

Sleep is a mandatory part of life, and good sleep is important to maintaining good health. The rapid eye movement (REM) stage is the sleep that is maximally restorative, but only accounts for 25% of the sleep cycle. REM becomes longer and more frequent toward the morning. Stages I–IV are progressively deeper stages of sleep. A variety of parts of the brain are involved in the regulation of our sleep-wake cycle and our "internal clock" (circadian rhythm).

A sleep study involves a variety of non-invasive but not necessarily comfortable monitors, including EEG, muscle activity, blood oxygen, airflow and nasal pressure, heart function, and expired CO_2. The process, polysomnography, is best performed by child-friendly, experienced technicians.

A variety of risk factors for sleep disordered breathing may be present in children and adults with PWS, including decreased muscle tone, brain developmental abnormalities, differences in arousal to low oxygen and high CO_2, and obesity. Other factors not related directly to PWS include enlargement of tonsils and adenoids and acute respiratory infections. The effects of sleep apnea in childhood can include excessive daytime sleepiness, bed-wetting, growth impairment, behavioral problems, high blood pressure and heart failure. In some ways, people with PWS may appear to have a pattern of findings similar to people with narcolepsy, including sleep onset REM periods. Individuals with snoring, restless sleep, episodes of not breathing during sleep, bed-wetting that starts after they have been previously dry, morning headaches or excessive daytime sleepiness should have an evaluation of sleep. Likewise, it is recommended that children with PWS have a sleep study before, and 6 to 8 weeks after, starting recombinant human growth hormone therapy.

Treatment of sleep disordered breathing involves reducing airway obstruction by removing redundant tonsillar and adenoidal tissue, or by using an external device to keep the airway open, such a continuous positive airway pressure machine. This machine is often not tolerated well by children. Weight loss is also an important treatment. If nothing else works, tracheotomy (placement of a permanent breathing tube into the trachea through the neck) may be needed. More work is needed to understand the full spectrum of sleep problems associated with PWS, and to evaluate treatment strategies.

Clinical features, psychological and academic profiles of PWS: comparison of genetic subtypes

D. Greco[1], S. Buono[1], P. Occhipinti[1], A. Costanzo[1], L. Ragusa[1], G. Maiorana[1], F. Scannella[1], P. Bosco, and C. Romano[1] Dept. for Mental Retardation and IRCCS, Troina, Italy

Advances in genetics have led to an increased understanding of the role of the genotype on psychological functioning in PWS, in particular regarding cognitive and behavioral phenotype. Recent papers have reported phenotypic differences associated to the main genetic subtypes (deletion and maternal uniparental disomy), including lower birth weight in the deletion group, shorter birth length in males with UPD and shorter course of gavage feeding and later onset of hyperphagia in females with UPD. Other features previously reported with UPD were: less typical facial appearance, mild hypotonia, minor genital hypoplasia, skill with puzzles, high threshold of pain, and delay in diagnosis.

Cassidy, et al. (1997) reported relatively less impairment in articulation and skin picking among PWS individuals with UPD in comparison to those with deletion. Roof, et al. (2000) and Thompson (2002) examined differences in intellectual functioning related to the two genetic subtypes, and reported that subjects with UPD had significantly higher verbal IQ scores than those with deletion, while performance IQ scores did not differ between the two PWS genetic subtypes. It isn't clear if these intellectual differences might influence school performance; however, Butler, et al. (2004) reported difficulties in reading and math skills as well as visual-motor integration in deletion group.

Dykens, et al. (1999) have found that the deleted cases showed significantly higher maladaptive ratings on Child Behavior Checklist as well as more symptom-related distress on the Yale-Brown Obsessive-Compulsive Scale. Recently Butler et al. (2004) identified more behavioral and psychological problems in individuals with type I deletions.'

The purpose of this study is to examine the presence of such differences related to these two genetic subtypes in our sample.

Abstracts continued on page 14

Happy Birthday, Mary Lou Watson!

On Dec. 2, Mary Lou Watson, of Framingham, Massachusetts, who has PWS, celebrates her 51st birthday. She was born in 1953, three years before Drs. Prader and Willi described the syndrome. Her parents had no idea what was wrong with her. As her weight ballooned, her doctors scolded her mother for feeding her too much — a familiar story.

When she was 15 and up to 360 pounds, Mary Lou's brother read an article by Dr. Hans Zellweger, one of the giants in Prader-Willi history, who was at the Iowa University Hospital. Her mother Mary first called Dr. Zellweger, then took Mary Lou to Iowa, where he confirmed the diagnosis. After a bypass operation there, she lost 100 pounds the first year.

Mary Lou weighs 168 pounds now, is healthy and happy. The group home which she entered when she was 18 is close enough for her to go home every weekend. The only sad note in her life right now is that she lost her beloved father, Bob Watson, in October. We think he'd be very proud of Mary Lou on her birthday.

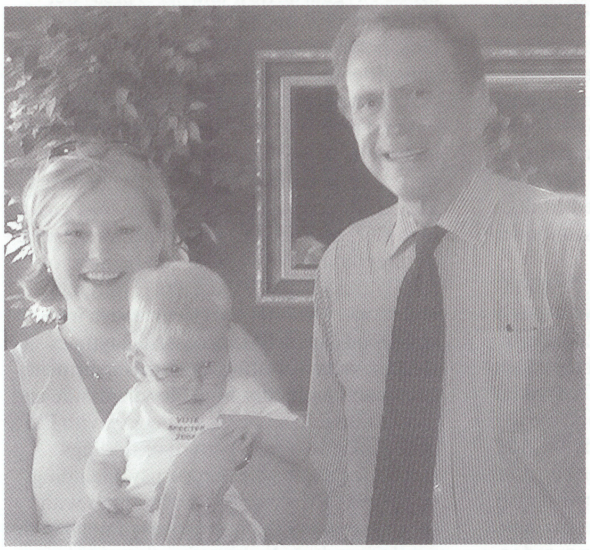
— Lota Mitchell, Associate Editor

Research - *continued from page 4*

Bringing Scientists Together

PWSA(USA) believes that another role it can play in spurring research is to foster communication and networking among researchers. Through the Scientific Conference each year and through communication with doctors and researchers, PWSA(USA) is in a unique position to be a focal point for the network of researchers working on PWS.

One way that PWSA(USA) is bringing scientists together is slated to occur in November. PWSA(USA) is supporting a "Scientific Roundtable" discussion on hunger in PWS. Created, organized and led by Jim Kane, the Scientific Roundtable will bring six to eight major PWS and hunger/obesity researchers together with representatives from the NIH for a weekend to discuss the top 10 research projects that would help discover the causes and treatment of hunger in PWS. Then we'll work to get those projects funded and under way.



U.S. Sen. Arlen Spector of Pennsylvania meets Joyce Opp and her son Daniel, who has PWS.

Leveraging PWSA(USA) Information

PWSA(USA) is the largest organization in the U.S. related to PWS and therefore has access to information about PWS that is unavailable to anyone else. Barb McManus (grandmother to Jessika, 12) has begun circulating a questionnaire on the web (and in paper form) about people with PWS. The goal is to get a response for each PWSA(USA) member who has PWS. Questions concern the age, height, weight and type of PWS, as well as a checklist of other PWS health issues (hypothyroidism, sleep apnea, use of growth hormone, etc.) When complete, PWSA(USA) will have ready access to information about hundreds of people with PWS that would help all families and could provide insight for further research topics.

As a parent and as chair of the Research Committee, I am committed to seeing that PWSA(USA) does what it can to fund and spur research that will improve the life of my daughter and all those with PWS. If you have any ideas, thoughts, or questions, please feel free to contact me or the PWSA (USA) office.

Rob and his wife Deb and daughters Isabel and Natalie live in Bryn Mawr, Pa.

The Chuckle Corner

Try, Try Again

I was in the kitchen about a month ago and Mikey, age 5, who has PWS, came over and asked me for a hot chocolate. Being the middle of June I told him it was too hot and I wasn't making it.

He came back shivering a minute later saying, "I TOLD (cold)."

He ended up with chocolate skim milk, but you have to give him points for originality!

Nicole Henshaw, Pittsburgh, Pa.

Please send your joke or funny story to the PWSA (USA) office. Be sure to include your name, phone number and address in case we have any questions.

2005 CONFERENCE HOLD THE DATE!

2005 National PWSA (USA) Conference will be held at the Sheraton World Resort in Orlando, Fla. Scientific, Providers and Chapter Presidents programs are on **July 27**, the General conference and YAP/YIP programs are **July 28 and 29**.

An outstanding youth & adult program is planned. Watch our web site and upcoming issues of *The Gathered View* for details.

Pioneers - *continued from page 1*

Perhaps there is a sense of loss felt by the parents of older adults, as the PWSA National Conference currently includes their person with PWS only every other year. I think there have been many benefits to the smaller conference that every even year is centered around the young children — focusing on their needs and research into opportunities to impact their future at a very young age. I have been to one of those conferences and it was wonderful!

However, we can fill that loss by taking on projects, communicating by e-mail, and using our collective efforts to do a greater good. Let us not forget that there is a collective good that can come from the efforts of parents of older adults; and as parents of older adults, let us not become disenfranchised. There is still so much more to learn from and about the aging adult with PWS. Perhaps we can even build a "mini-conference" for adults with PWS.

I think it is time for us to consider establishing an affiliate association of these parents who are entering retirement years — sort of a "Pioneer" group. We could take on special project(s) at the behest of our beloved national association. Perhaps it could be a work effort in support of the national conference, in support of research, or an accumulation of our collective experiences in group homes and other alternative living experiences, physical exercise, or work alternatives for the adult with PWS. Wouldn't we all benefit from that?

Interestingly enough, I have found that I am still drawn to the national conferences — certainly for new pieces of information I can pick up, but as much or more so for the benefit of the many young families who are where I was so many years ago.

These young families gaze lovingly and curiously at my daughter, perhaps trying to get a glimpse of what the future might hold for them, just as I did. They draw Dale (my husband) and I aside to ask about how we handled certain situations they are faced with, or to ask what is was like so many years ago when so much less was known about PWS. I'm sure it gives them a sense of comfort to reflect on how much more support they have now — something positive about the situation they find themselves in.

Also, they want to talk about us, our lives, how we have coped, how we manage the demands of work and everyday challenges, and how our other children have coped with the stress of a family dealing with PWS. This is so important to these young families, and we can give them hope for their ability to persevere and, yes, even prosper with their own families.

I think parents of older children/adults with PWS have a unique opportunity to contribute to the lives of many families in the midst of extreme challenges and even crisis as they try to go through their everyday lives. Our contribution may be different for each of us, but contribute we can. I am sure each of us can remember the times of extreme stress in our families — when a helping hand or just an understanding smile meant so much. Let us not forget how important that association was in our lives and that many more families are still in the throes of those challenges.

It is difficult for many of them to get through each day, much less volunteer their time to the association. But we can. We know what the needs are and we can offer support by making phone calls, writing articles, hosting a fund-raiser, talking with families who need support, or a thousand other "beacons of light" that can make such a difference for all families impacted by PWS.

Dottie and Dale Cooper live in Alpharetta, Ga. She is the mother of Shawn, 31, who has PWS.

View From The Home Front

Living With PWS Means Challenges as well as Triumphs

Robin MacGillivray writes:

Robin MacGillivray

I wrote an article that appeared on page 1 of the July-August 2004 newsletter about my accomplishment last year. My intention was to spread hope and encouragement that it is possible for individuals with PWS to reach their own goals and dreams.

I'm overjoyed and honored to hear from several parents of young children with PWS that my accomplishment has been encouraging in ways that I could have never have imagined. However, I'm heartbroken that my graduation story may have been painful for some parents whose children have not done as well, and and remorseful that I ignored the importance of sharing my personal challenges as well as accomplishments.

I hope that the following article written by my mother will serve as a supplement to the July-August 2004 article. It acknowledges some of the challenges associated with PWS that I've had to deal with. I wish you all the best of luck on accomplishing any success possible with the difficult challenges of PWS. In addition, I need to make it clear that although I was fortunate enough to get as far as I have, the success that you may have with your child who has PWS may not be the same as mine. However, I don't want to be discouraging because there is ALWAYS hope that there can be some success in dealing with PWS.

Robin's mother, Lorrie MacGillivray, continues:

After the publication of her article in *The Gathered View*, Robin received a handful of very positive e-mails, all from parents of young children. It has been pointed out to us that some parents of older children found her article to be very disheartening, and even wondered if she really has PWS. I will give you a little back-ground which may help.

Robin had all the classic signs of PWS as an infant and child. She was evaluated for PWS at age 3 and again at age 5, first by an endocrine specialist who ran an inconclusive genetic test, and then by the Twin Cities specialist in PWS at Gillette-Children's hospital. They both felt that they could not make a diagnosis of PWS.

As time went on and the Internet became available, I did more research through organizations like PWSA(USA) and became convinced that Robin indeed had PWS. But my husband and I hesitated to have another diagnostic consultation — everything we read about PWS was so lacking in hope that rightly or wrongly we avoided dealing with the situation.

Meanwhile, of course, Robin gained weight and had various problems including speech, physical (surgery for scoliosis), and social. But on the positive side she was determined to do well in school, plus she had an interest in trying a variety of activities. She took music lessons, tried out for plays, and joined a softball team. Plus, she was lucky enough to have some loyal friends.

When she was 17, her scoliosis physician referred to "her genetic disorder," and Robin actually asked us to be re-evaluated for PWS. She said she was ready now to handle the results (as if anyone could ever be ready for this diagnosis!).

This time we had the FISH test, from which I quote: "...a microdeletion of the Prader-Willi critical region of one chromosome 15 is identified by flourescent in situ hybridization using a research molecular probe for the SNRPN locus located in G-band region 15q11-15q13. In the FISH analysis, ten metaphases were studied and, in each, a flourescent SNRPN signal was seen on only one chromosome 15 thus suggesting a microdeletion of the Prader-Willi critical region (within the limits of FISH technology)." This test is dated April 14, 1999, and was run by Allina Cytogenetics laboratory.

This diagnosis offered Robin about 2 hours of joy ("This proves that it is not my fault!") followed by at least 2 years of emotional crisis, anger and turmoil. We were lucky enough to come in contact with Bonny Bates, an advocate for people with PWS, who really helped us understand the ramifications of PWS and the requirements for successful lives for those with PWS.

Before the PWS diagnosis, we had always raised Robin to assume that she would go to college. After the diagnosis, the PWS specialist in our area told her that "PWS kids don't go to college." This, along with other very negative information we received, angered Robin a great deal — and anger can be a great motivator. I really think that her desire to show this physician that he was wrong was a great contributor to her desire to succeed.

Why has Robin done as well as she has? I don't have an answer to this. Maybe your medical contacts can read something into that FISH test that I can't. Are there levels of severity of PWS?

Robin struggled to get into this college, but she had her heart set on it. She took the ACT 3 times (finally achieving an adequate score when she was allowed to do extended time testing). She went to the college rep visit when they came to her high school, took notes, and went up and explained her situation to the rep. When her application was being reviewed at the college and was going automatically into the "No" pile, this rep remembered Robin and took her part — getting her into the "maybe" pile until the final ACT score was in.

We have tried several housing and food situations since she entered St. Thomas. We actually interviewed and hired a

Robin continued on next page

Julie Donates Locks for Love

Julie Mitchell (shown here with her mother, PWSA(USA) past-president Lota Mitchell), let her thick dark hair grow and grow... and grow. Last summer when she decided to get it cut, she donated her shorn tresses to Locks for Love, an organization that uses human hair to make wigs for children who have lost theirs due to chemo and cancer.

Julie, who has PWS, is very proud that she was able to help someone else.

What a good deed, Julie, and your new short 'do looks great!

Ghrelin - *continued from page 8*

longer acting, injectable version of somatostatin to investigate this further.

Since we showed that somatostatin also lowered the levels of another hormone in the blood called peptide YY, which normally reduces appetite, this action could have cancelled out the beneficial effect of lowering ghrelin on appetite. The future development of drugs that block the action of ghrelin more specifically will hopefully enable this possibility to be examined. While it is obviously disappointing that we were not able to demonstrate any reduction of appetite with our treatment, the results have been extremely informative and have laid the foundation for future studies to investigate the cause and possible treatment of the excess appetite in Prader-Willi syndrome.

The results of this study have been published in the Journal of Clinical Endocrinology and Metabolism (Tan T.M., Vanderpump M., Khoo B., Patterson M., Ghatei M.A., Goldstone A.P. Somatostatin infusion lowers plasma ghrelin without reducing appetite in adults with PWS. J Clin Endocrinol Metab. 2004 Aug;89(8):4162-5).

We thank all the individuals, caregivers and their families for taking part in our study, and also the UK Prader-Willi Syndrome Association for financial support.

Robin - *continued from page 12*

roommate freshman year — that turned into a disaster and a complete blowup by the end of the semester. The housing director (bless her) found a new roommate, a senior majoring in social work, who was perfect. She was the dorm RA and got Robin involved in a bible study group that has remained her main social outlet in college.

Robin is still in school, having finished her B.A. in sociology, but needing to complete her field experience and student teaching to finish the B.A. in elementary education. This semester she will also be taking a couple of graduate-level courses on special education. She has an on-campus apartment by herself (she is difficult to room with because of the food situation).

For the past 3 years I have packed every meal and every snack in baggies and brown paper bags. She has absolutely no money — she has to bring her laundry home because the quarters were going for candy instead of laundry. She exercises three times a week with a fitness consultant, and once a week with her Dad or me. I know she is lonely living alone and we would all like to remedy this with the right roommate.

Last summer Robin lived in a PWS group home with several other women. She did this against her will, but to her credit she accepted the situation. She did lose weight, but was very discouraged by the distance she felt between her abilities and those of the other women. Sometimes she feels very isolated — she does not feel a true kinship with other people with PWS, but she sees that she is different and doesn't quite fit with "normal" individuals, either.

Why has Robin done as well as she has? I don't have an answer to this. Maybe your medical contacts can read something into that FISH test that I can't. Are there levels of severity of PWS?

While we have been attentive parents and have always tried to be strong advocates for our daughter, so have the parents of people with PWS with less positive outcomes. I don't think that we have done anything differently, or more aggressively, than most parents.

I do know that Robin has the ability to reflect on her disorder in a way that seems extraordinary to me. Just tonight she presented me with a new food plan that she wants to try, based on the PWS food pyramid and other research she has done via the medical resource indexes on her college library internet (articles printed out and highlighted with the info she wanted me to know about).

I hope my letter puts to rest some of the skepticism about the validity of Robin's PWS. (What a ridiculous thing to say — as a parent, I don't want to defend the validity of her PWS, I just don't want her to have it.) I realize that Robin is blessed to be as highly-functioning as she is, and that we are certainly blessed to have her as our daughter.

My heart goes out to those who might be saddened by Robin's story — that reaction is certainly the opposite of what Robin wanted. I think these families are correct that they have done everything possible — and I hope that this follow-up article might be helpful, perhaps on the range of what's "normal" for those with PWS and outcomes that might be expected with PWS.

Prevalence of poisoning in people with PWS

Ulrika S. Lange[1], Karen Potter[2], and Shawn E. McCandless[3] [1]Univ. of North Carolina at Greensboro, Genetic Counseling Program; [2]Dept. of Pediatrics, Div. of Genetics and Metabolism, Univ. of North Carolina, Chapel Hill; [3]Departs. of Genetics and Pediatrics, Case Western Reserve Univ., Cleveland, OH

This pilot study assessed the prevalence of poisoning in people with PWS. The goal was to examine whether this unusual food-related behavior would lead to an increased prevalence of ingesting a poisonous substance. Subjects in this study included those with PWS ages 4 and older. Participants who completed the anonymous online survey were either the parents or the primary caretaker of individuals with PWS. The participants also provided information about non-PWS siblings, who served as controls. One hundred forty-one responses were submitted using the online survey.

This study found a six times higher prevalence of poisoning in people with PWS compared to their non-PWS siblings. Several features of PWS, including the food-related behaviors (specifically "looking and searching for food"), decreased cognitive ability, eating unusual objects, and history of extreme behaviors appear to correlate with this increased prevalence. Awareness should be raised by alerting parents and caregivers of the increased prevalence of poisoning in people with PWS. Primary care providers and poison control centers also need to be aware of this association, as well as the implications of PWS for diagnosing and treating ingestions.

Abnormal Food Motivation in PWS: Relationship between Neural Dysfunction and Obesity using fMRI

Laura Holsen[1], Jennifer Zarcone[1], Mary Anderson[1], Jamie Young[1], Merlin Butler[2], Travis Thompson[3], and Cary Savage[1] [1]Univ. of Kansas Medical Center, [2]Children's Mercy Hospitals & Clinics; Univ. of Missouri-Kansas City; [3]Minnesota Autism Center and Univ. of Minnesota

The behavioral phenotype of PWS includes hyperphagia and obsessive-compulsive disorder (OCD). It is proposed that the interaction of these behaviors may be related to regional brain dysfunction. Based on a model of OCD and abnormal food motivation, it was predicted that specific areas of the brain that respond to food-related images (i.e., orbitofrontal cortex, insula, limbic cortex and anterior cingulate gyrus) may differ from areas of activation during viewing of non-food images. Further, it is predicted that different states of motivation (pre-meal vs. post-meal) may predict specific sites of activation (LeBar et al., 2001 *Behavioral Neuroscience, 115,* 493-500). This study examined the relationship between hyperphagic and food-related compulsive behaviors and localized brain activity in PWS.

Data are reported for eight individuals with PWS (7 females, 1 male; mean age = 15.4 years, SD = 5.2) In contrast to normal-weight individuals, for whom pictures of food activate specific areas of the brain to a greater degree when a person is hungry (versus during a state of satiation; LeBar et al., 2001), our recent findings using fMRI demonstrate that individuals with PWS display greater activation post-meal than pre-meal. These findings may indicate a specific neural contribution to the gene-brain-behavior interaction in hyperphagic behaviors in PWS, as well as possible delineation of neural mechanisms of obesity.

Facial phenotype of PWS becomes more normal with early growth hormone treatment

Suzanne B. Cassidy[1], Ellen Simpson[1], Shauna Heeger[2] [1]Div. of Human Genetics, Dept. of Pediatrics, Univ. of California, Irvine; [2]Case Western Reserve Univ.

In recent years, short stature in PWS, as well as altered body composition, has been attributed to growth hormone deficiency, since they improve with replacement therapy. Many newly diagnosed people are being treated from the time of diagnosis, which is often infancy. Observation of treated patients has shown significant impact on phenotype.

Published reports of double-blind cross-over studies of recombinant human growth hormone (rhGH) treatment in PWS have demonstrated dramatic increase in growth rate (especially in the first year of treatment) and a variety of other effects, including: 1) improved body composition (higher muscle mass, lower fat mass); 2) improved weight management; 3) increased energy and physical activity; 4) improved strength, agility and endurance; and 5) increased respiratory muscle forces. No study has specifically addressed facial appearance.

We have been conducting an ongoing multi-system detailed standardized phenotypic evaluation of individuals with PWS over the past few years in a study designed to make genotype-phenotype comparison. However, given the recent shift to rhGH treatment, recently enrolled patients with rhGH replacement can be compared phenotypically to earlier enrolled patients, who were not rhGH treated. Such comparisons, as well as review of patients followed in a multi-disciplinary PWS management clinic and of studies by others, indicate a number of effects on phenotype. These comparisons primarily document the normalization of facial appearance, as well as improvement towards normal body habitus. Although comparisons are difficult to make quantitative or objective, this presentation will show, using multiple examples, the differences in facial appearance between those who have, and those who have not, been treated with rhGH. Body habitus will also be compared. Such photos show impressive differences, with those treated with over a period of years lacking the typical facial gestalt of PWS.

This study will demonstrate that normalization of facial and body appearance can be added to the benefits of growth hormone therapy in PWS.

New Zealand Scientific Conference – Part 2 of 2

Skin-Picking and Other Self-injurious Behaviors in PWS

Chris Oliver, BSc, MPhil, PhD., AFBPsS; University of Birmingham, UK

Evaluation of a high prevalence of skin-picking and other self-injurious behaviors in PWS is analyzed in three studies, which conclude the usual risk markers for these do not seem to be evident and alternative causes need to be considered.

Poor Evidence for Contraindication of Growth Hormone for PWS Patients With Respiratory Problems

T. Nagai, K. Obata, J. Murakami, A. Yoshino, E. Takahashi, R. Sakuta; Dokkyo Univ. School of Medicine, Koshigaya Saitama, Japan

A study of growth hormone treatment following the death of two PWS patients in Zurich, Switzerland with several respiratory problems since birth, since the association between GH treatment and death was not clear. A survey questionnaire of 318 responses showed 1) Respiratory problems were noted in about one-third of all PWS patients before GH treatment; 2) Among these respiratory problems, a similar number of central and obstructive apnea were the most common; 3) GH treatment was not harmful to PWS patients with any respiratory problems. Rather, it seemed to have ameliorated central apnea in those patients.

Impact of Long-term Growth Hormone Treatment on Body Composition in PWS

Ann Scheimann, M.D., W. Klish, B. Hayslett, N. LaFuente, D. Orellana, B. Gordon, R. Thompson, Phillip D.K.Lee, M.D.; Johns Hopkins University School of Medicine, Baltimore, MD; UCLA School of Medicine, Los Angeles, CA; Baylor College of Medicine, Houston, TX.

An IRB-approved retrospective of 84 patients with PWS was reviewed at Texas Children's Hospital between 1998 and 2002. Patients were divided into two GH-treated groups (between 0-5 years treatment and greater than 5 years GH treatment) and an untreated group.

GH treatment appears to have long-term beneficial effects on body composition and linear growth. GH replacement therapy does not appear to increase the likelihood of development of scoliosis.

Abstracts continued on page 15

Contributions In Memory Of

Mrs. Sarah Avin
Michael Burns & Sybil Cohen
Julie Burhans
Mary Burkhardt
Jerry & Kathy Buyze
Patricia & Roger Cooper
Sharon Manning
Jack & Lucille Pearson
Eva Frazin
Marshall & Rene Gratz
Anne Matsoff
James Golden
Bill & Kathy Noffsinger
Elinor S. Hearn
Target
Harold & Helen Crowe
Rodney & Doris Danielson
Bill & Marolyn Halverson
Hilda Mary Schoon
Willis & Rebecca Wong
Gene Higginbotham
Overland Park Automotive
Midwest Cinema Group
Patricia & Ernest Cruwell, Jr.
June Davis
Joann & Lawrence Hanus
Dennis & Karen Higginbotham
Kathy Hassler,Jacky Jenkins & Janet Miller
Tony & Carole Katsantioness
Kenneth & Paula Manley
Paul & Ruth Meyn
Mike Schmable
Marcella Shanavarger
Robert & Jeanne Shaw
Frank & Karolyn Sotolar
Adam Romagnoli
Hymen & Ruth Chausow
Charles & Geraldine DiCosimo
Wm. Rhett Eleazer
Dennis & Sharon Saacks
Hank Lee
Bill & Marolyn Halverson
Julie Manning
Jack & June Link & Family
Clyde Mays
Elissa Gruenhut
Tim & Carol Hearn
Stewart & Bronnie Maurer
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Perspectives on an Association Between GH Therapy and Mortality in Children With PWS

David B. Allen, M.D.; Univ. of Wisconsin Children's Hospital, Madison

Discussion: In children with PWS, unexpected deaths due to respiratory illnesses and/or respiratory failure are disproportionately high. Breathing abnormalities include restrictive airways, reduced oxygen or elevated carbon dioxide levels, sleep apnea and arousal disorders. Growth Hormone (GH) treatment has been shown to increase respiratory muscle strength and measures of pulmonary function and reduce the number of hypoapnea and apnea events. Recent unex-pected deaths reported in very obese children with PWS treated with GH have created concern about a possible association. Until more information is known to guide a risk/benefit analysis, it is recommended that children with PWS have a sleep study and ENT exam prior to GH therapy. Excessive obesity (e.g.> 200% expected weight for height) and evidence of nocturnal airway obstruction are considered contraindications to GH therapy until corrected.

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