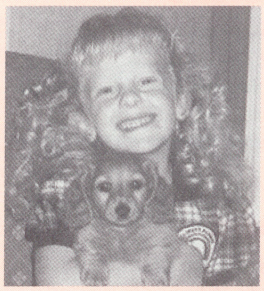
November-December 2003
Volume 28, Number 6

The Gathered View

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



In this issue

Always in Love With Aimee

By Tim Atwood

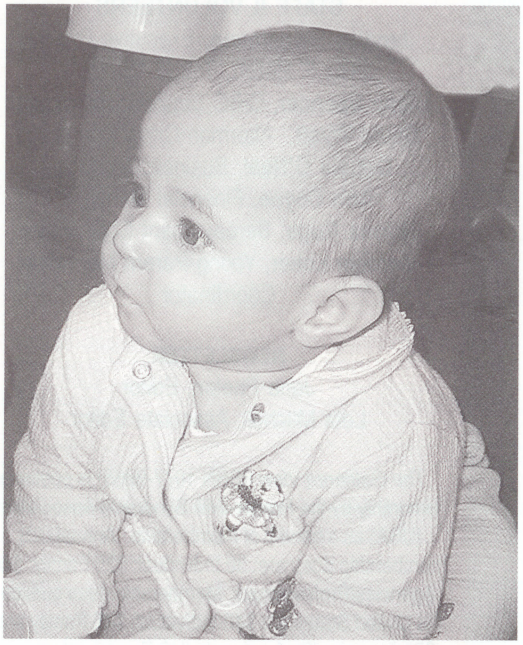
I felt compelled to write this for my wife, maybe because I don't talk about it much. There have been a few inquiries from new parents on the [0-5 e-mail] list in the past month, so hopefully this might help.

Aimee was 6 months old this past Friday. She put her own exclamation point on the occasion by rolling over for the first time. She pushed and grabbed and kicked furiously last night when I tried to coax her to finish the last ounce of her bedtime bottle. Her facial expressions now range from a crooked-lipped pout of protest, to little half smiles that would make La Gioconda proud, to gum-baring grins.

She recently started to add coos and other noises to the usual squeaks and grunts. She smacks her lips when eating her cereal. She stares at my face when she's feeding. She sticks her hands in her mouth, but hasn't figured out how to suck her thumb yet. She's got chubby little arms and legs, but not much of a belly. She has perfect skin, and a perfect little head of dark hair. She stays very clean and never spits up. She recognizes her daddy and especially her mommy.

At the many visits to many doctors, she's stoicism personified until she decides she's had enough poking and prodding, then she squeaks mightily in wide-eyed protest. She gets stirred up when her twin sister is on the floor next to her, especially when Jolie rolls onto her. She arches her back and twists around to look whenever the TV is on.

In short, she has become a little person. It was hard to imagine this when she was less than 4 lbs, with large ridges in her softball-sized skull, legs pushed out to the sides like a chicken, tube in her nose and IV in her forehead, laying very still and small in a large warming unit, a living symbol of the vagaries of fate and randomness of nature's design. For the 11 long weeks in two hospitals, she slept and slept, not seeming blissful but rather desperate to me as I watched her tearfully.



Aimee, who has PWS, is perfect in her Daddy's eyes.

During the past few weeks, she's been kissed with a new spark of life. I don't know if it's the increased GH dosage, just a natural progression in her growth, or a change in my own perception of her as I realized how much I've grown to love her.

At times I questioned whether I would ever be able to see past her condition and just love her as a baby, a little being full of potential and not a burden inflicted on my family. It was painful to contemplate, especially since I have loved my first child, Caitlin, more than anything, ever, since the minute she was born. Caitlin was so perfect and amazing it just came naturally. I had to try and set aside my preconceptions about fatherhood that had been reinforced by the painless addition of Caitlin's life to my

Aimee continued on page 15

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.

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 HOLIDAY
If asked for Domain Name it is CIMCO

Janalee Heinemann

Executive Director's View

PWS Research & Awareness – Where Do We Stand?

Exciting News in Research

A major first for PWSA (USA) is that we have been accepted as part of a major NIH research grant that was recently awarded. We will be part of one of the Centers in the Rare Diseases Clinical Research Network: Developing Collaborative Cyber-Infrastructure for Clinical Research.

A group headed by Dr. Art Beaudet from Baylor College of Medicine in Houston, Texas and including Dr. Dan Driscoll, Dr. Suzanne Cassidy and others closely involved with PWS and PWSA (USA), secured this 5-year, $6,000,000 grant from NIH to establish a new Rare Diseases Clinical Research Center (RDCRC).

The RDCRC will focus on Angelman syndrome, Rett syndrome and Prader-Willi syndrome, noting, "The RDCRC will focus on these three areas with the expectation that they may have near-term potential for meaningful therapy…. The Center will have an active affiliation with IRSA, ASF and PWSA-USA…. An extensive program is proposed for training new investigators in clinical research on rare diseases.[1]" Each of the three diseases — Rett, Angelman and PWS — will have its own research committee. Our Chair of the Clinical Advisory Board, Dr. Dan Driscoll, will chair the PWS committee for this Center grant.

This grant will include support from a Data Technology Coordinating Center (DTCC) based at the University of South Florida in Tampa, Florida. We will all be interacting with DTCC. Through PWSA (USA) research funding, our Board of Directors has approved matching the $25,000/year NIH funding to allow us to hire a full-time database coordinator. The database coordinator will be an employee of PWSA (USA) who will work in close cooperation and support with the NIH-funded Data and Technology Coordinating Center.

I believe this new database position and system offer the greatest impact for our research money by providing the questions that will peak research interest, as well as give researchers enough data to pursue funding. As an example, I believe there is a problem with gallstones among our PWS population. If in our collected data we find many children and adults who have had gallstones and gallbladder crises, this could (1) encourage a researcher to further pursue these findings; (2) provide a researcher with the data needed to elicit NIH funding; and (3) give us enough data to publish a medical alert/awareness statement.)

Also under this grant — and distinct from the PWSA (USA) database — would be a more formalized research database. This database would be maintained through the DTCC, and only patients who enrolled for a research study would become part of that research database. The PWSA (USA) database will be much more extensive regarding the number of children and adults with PWS we will be following.

Other Areas of PWSA (USA) Research Focus

- **Ghrelin/appetite research** — PWSA (USA) has approved the funding of a grant submitted by Jean-Pierre Chanoine, MD from British Columbia on "Effect of Somatostatin on Ghrelin Concentrations, Food Seeking Behavior, and Weight in patients with PWS." This research will involve giving a long-acting form of somatostatin, a hormone that inhibits ghrelin, to see if it is effective in decreasing ghrelin concentrations, food intake, and compulsive behavior toward food with PWS.
- **Behavior research** — We currently have a request for proposal for a research study on which psychotropic medications work best for those with the syndrome. Money has been designated for this very important research.
- **CoQ10 research** — Our pursuit of a CoQ10 grant has become more complex as we pursue our concept of a collaborative grant with the Mitocondrial Foundation. One key stumbling block is finding a population to study that is NOT on growth hormone. If a child *is* on growth hormone, it would be hard to distinguish the effects of growth hormone from the effects of CoQ10. After several conference calls and many e-mails with researchers from both organizations, we have identified two options: 1) a multi-center study with one coordinating center to evaluate observable responses to CoQ10; and 2) a center that could evaluate true biochemical changes. A Canadian researcher is interested in

PWS Research continued on page 4

PWSA Gets A+ for Awareness

This year, 14 to 17 newly diagnosed infants and toddlers were referred to us each month. Many were referred within the first few weeks of birth.

There are about 3.5 million births in the USA per year; that's about 300,000 per month, or 20 to 30 PWS births per month to be expected. It appears we have reached the 50 percent point of diagnosis and referral. That is a landmark! Only a couple of years ago we estimated less than 20 percent of infants were getting diagnosed and referred to PWSA (USA).

studying the Mitocondrial population and has the sophisticated equipment to do this type of testing properly, but he does not think he can also take on PWS. I do not see an easy resolution to making this research viable, but we will continue to pursue options.

- **Study of death grant** — PWSA (USA) presumably has the largest recording of PWS deaths in the world. Of our statistics on the 140 deaths we have followed, we have calculated both the median/average age of those studied is 27, although we know many adults with PWS live far longer. Because we feel strongly that we need to have a better understanding of the cause and prevention of deaths in PWS, I have written a grant to study PWS deaths. This would be a collaborative grant with the University of Utah, and we are waiting to hear from the potential funding source.
- **Chromosome 15** —Robert D. Nicholls, D. Phil., professor of Psychiatry and Genetics at the University of Pennsylvania School of Medicine and his colleagues discovered four new genes normally found on chromosome 15 that, when lost, may contribute to Prader-Willi and Angelman syndromes. Subsequently, the researchers determined that the mutation of one these four genes, *NIPA1*, is also responsible for a hereditary disease called spastic paraplegia. Their findings make genetic screenings and drug development for spastic paraplegia possible and can open the door for better diagnosis of chromosome 15 rearrangements, including deletions that cause PWS and Angelman syndromes, and duplications found in some cases of autism. The two syndromes are genetically-related, yet separate neurological disorders whose effects are determined based on which parent donates the faulty chromosome. Dr. Nicholls is a member of the PWSA (USA) Clinical Advisory Board. He and his colleagues announced their findings in two separate papers in the October issue of the *American Journal of Human Genetics* (available now online). Funding for this research was provided through grants from the National Institutes of Health, March of Dimes Birth Defects Foundation, and Muscular Dystrophy Association.

PWSA (USA) Web Site Usage

Compared to a national organization of a syndrome with the same population (Angelman Syndrome Foundation)

Number of hits
PWSA (USA) 933,442
ASF 261,322

Number of pages viewed
PWSA (USA) 254,890
ASF 24,726

Visits/user sessions
PWSA (USA) 23,093
ASF 11,411

Regarding PWS Research in General

Dr. Nicholls recently wrote: "It is worth pointing out that there are significant basic science (in addition to clinical interest) reasons that 'PWS-related' research may receive funding over and above the population incidence, so that the latter should not be used solely as a frame of reference. For example, PWS research has been foremost in many discoveries and understanding in human disease (and mouse models) research on genomic imprinting, mechanisms of chromosome rearrangements, of complexity of gene structure and expression, as well as other areas.

"So many insights into human genome function and genetic phenomenon have come from work directly related to PWS from many laboratories, and will be likely to come in future studies, that the funding for PWS-related research will hopefully only continue to improve in coming years. In other words, NIH research funding for PWS-related work has been good because we learn not only about PWS, but insights are gained into so much more about genetics and other human diseases in general from this work."

Advances Through Continued Support

Please remember, we can only continue to keep up the exciting advances we are making in research and awareness through your continued support. I want to extend a special thank you to those who have sponsored a fundraising event in the last year, or given personal support through donations of money or time. As I travel to Israel, Taiwan and Mexico to speak to their national organizations, I can also speak proudly of PWSA (USA) and know that the good we do in this country will shine a beacon of hope for all.

Footnote to page 3 [1] *Source- Summary Statement NIH Project Title: Rare disease CRC for new therapies and new diagnostics, 8/12/03, page 2.*

Researcher Seeks Study Participants

I am a pediatric endocrinologist at Duke University Medical Center. My research interests focus on understanding childhood obesity and specifically, the mechanism of development of obesity (including regulation of the hormone ghrelin) in healthy children, children with PWS and children with hypothalamic obesity. I invite you/your child/your patient to participate in these ground-breaking research studies looking at body weight regulation in childhood in the following subject populations:

- Infants/children with PWS (0-5 years)
- Healthy infants/children (0-5 years)
- Children with PWS (5-18 years)
- Children with organic hypothalamic obesity (5-18 yrs)
- Healthy children (5-18 years)

The procedures and time involved will vary depending on the study. No compensation or reimbursement will be offered to take part in these studies.

For more information, call Study Coordinator Donnetta Miller, at 919-660-6719 or page me at 919-970-0488. These studies are approved of by the Duke University Institutional Review Board (IRB). Andrea Haqq, MD, Principal Investigator, Office (919) 684-5091; Fax (919) 684-8613.

Highlights from Scientific Day 2003 Conference, Orlando, Fla

Chairperson: Dan J. Driscoll, Ph.D., M.D.

This day was dedicated to the memory of
Camilynn Brannon, Ph.D. 1963-2002 and Rob Wharton, M.D. 1944-2002

Editor's Note: The following is Part 2 of 2 parts, excerpted from abstracts presented at the Scientific Day. A full copy of the Scientific Conference Abstract booklet can be ordered through PWSA (USA)at a cost of $20.

Effects Of Growth Hormone Therapy In 3 Subjects With PWS

Donatella Greco, Paola Occhipinti, Letizia Ragusa, Ada Natalia, Fabio Scannella, Serafino Buono. Corrado Romano Department for Mental Retardation, OASI Institute Maria SS.(IRCCS), Troina (EN), Italy

Results and Conclusions: We have observed that the treatment of GH significantly increases lean mass, improves weight maintenance, muscle tone and physical agility. IQ remains stable, instead there are little improvements in the adaptive profile. Preliminary analysis suggests that GH-therapy improves the body composition and the behavioral phenotype.

Growth Hormone Secretion in Adult Patients with PWS

Graziano Grugni[1], Gianluca Aimaretti[3], Eros Barantani[2], Antonino Crinò[4], Alessandro Sartorio[1], Roberto Vettor[5]. [1]Division of Auxology and [2]Division of Metabolic Rehabilitation, IRCCS S. Giuseppe Hospital, Italian Auxological Institute Foundation, Verbania; [3]Division of Endocrinology, Department of Internal Medicine, University of Turin; [4]Unit of Autoimmune Endocrine Diseases, IRCCS Bambino Gesù Hospital, Rome; [5]Medical Therapy, Department of Medical and Surgical Sciences, University of Padua; Italy.

Results and Conclusions: Nine patients showed severe growth hormone deficiency (GHD, 75%), while 2 subjects had a partial GHD (16.6%). Only 1 male (8.4%) demonstrated a normal GH peak response. The majority (66%) of patients exhibited low IGF-1 levels. Moreover, no significant correlation was found between GH peak levels, age and BMI. Our results, together with those from the literature, seem to demonstrate an impaired GH secretory pattern not only in children, but also in adults with PWS. These preliminary data raise important questions about the potential beneficial effects of GH therapy in adult PWS.

Octreotide Potently Suppresses Circulating Ghrelin Levels In Children With PWS

Andrea M. Haqq [1], Diane D. Stadler [2], Ron G. Rosenfeld [2], Katherine L. Pratt [2], David S. Weigle[3], R. Scott Frayo [3], David E. Cummings [3] , Stephen H. LaFranchi[2], and Jonathan Q. Purnell [2]. [1]Department of Pediatrics, Duke University Medical Center, Durham, North Carolina.

Conclusions: In this pilot study, treatment with octreotide for 5-7 days markedly decreased fasting ghrelin levels in children with PWS. Children with PWS also demonstrated post-prandial suppression of ghrelin as would be expected under normal regulatory conditions. Additional controlled studies are needed to determine if long-term octreotide treatment causes sustained ghrelin suppression and weight loss in children with PWS.

PWS: Clinical Concerns for the Orthopedic Surgeon

Martin J. Herman, Department of Orthopedic Surgery, St. Christopher's Hospital for Children, Philadelphia, Pa.

Discussion: Osteopenia, poor impulse control, defiant behaviors, and diminished pain sensitivity are aspects of PWS that may complicate all facets of orthopedic non-surgical and surgical management in this patient population. The treating orthopedic surgeon must plan carefully and proceed with caution when treating children and adults with PWS.

The Relationship between Food-Related Compulsivity, Memory, Intellectual Ability, and Body Mass Index in PWS

Laura Holsen, Jennifer Zarcone, Mary Caruso, Jamie Young, David Richman, Travis Thompson, Institute for Child Development, University of Kansas Medical Center, Kansas City, Kansas.

Discussion: Performance on a short-term memory task appeared to depend on level of compulsivity when food stimuli were presented and IQ when non-food stimuli were presented. Highly compulsive people with PWS were capable of performing well on short-term memory tasks involving food-related stimuli, which are presumably highly motivating. On the other hand, on tasks involving less motivationally laden stimuli, IQ played a more important role. For participants in the food-seeking task, it appears that the degree to which their diets were restricted (resulting in lower BMI scores) may have had an effect on the amount of time spent seeking food and the amount of food covertly consumed.

Loss of Ndn results in deficiency of central respiratory drive in mice

Syann Lee, Jun Ren, Silvia Pagliardini, Matthieu Gerard, Colin L. Stewart, John J. Greer, Rachel Wevrick. Department of Medical Genetics, University of Alberta, Edmonton, Canada.

Discussion: These observations suggest that the developing respiratory center is particularly sensitive to loss of necdin activity and may reflect abnormalities of respiratory rhythm-generating neurons or conditioning neuromodulatory drive. We propose that *NDN* deficiency may contribute to the observed respiratory abnormalities in individuals with PWS through a similar suppression of central respiratory drive.

Abstracts continued on page 14

Lota Mitchell

President's View

Two Tributes — And the Lessons of Loss

Two very prominent members of our PWS family lost loved ones this fall — one suddenly, the other after long illness.

Ruth Millman, the mother of our Executive Director, Janalee Heinemann, died on Sept. 22. Ruth had been in poor health for many years, suffering from debilitating rheumatoid arthritis and other ailments. At her death at age 80, she only weighed 58 pounds.

In her eulogy of her mother, Janalee said that her parents through their example taught their children when they were young how to take care of others, how to treat them with kindness and respect and how to appreciate the basic goodness in people. Equally important, they also taught them NOT to holler at each other, NOT to swear or be cruel or criticize.

Her mother in particular taught the lesson of how to listen to others more than talk. Through the many painful years of her illnesses, Ruth demonstrated how to be strong and fiercely independent, how to love your family more than yourself, and how to go on, even when you have to work hard to find reasons to go on. Would that we all could teach our children these important lessons.

James Hanchett, M.D., husband of Jeanne Hanchett, M.D., had a massive heart attack at the Steelers football game in Pittsburgh on September 29. He died the next day, his bedside surrounded by Jeanne and their five grown children, when it became clear that further life support would be useless. He was only 68.

Until her retirement a year or so ago, Jeanne was pediatrician for Prader-Willi at the Children's Institute. She still does a PWS clinic there. She has been a member of our Scientific Advisory Board, participated in our national conferences, and has contributed so much in so many ways to the cause of PWS.

Through her, Jim, a dedicated physician with an impressive background in nephology, had become interested in the syndrome. Although he also was partially retired, he was doing some work with PWS. He probably had more knowledge on PWS care in the ICU than anyone in the world and had planned to be on the PWS Study of Death project. His death is a great loss, not only to his family, but to PWSA(USA).

An insert in Jim's memorial service program ended with the words, "The Hanchett family has been richly blessed by this good man's life." We in PWSA(USA) have been richly blessed by both these lives, directly or indirectly. We offer our love and sympathy to Janalee and Jeanne.

And we ourselves are reminded once again not to lose sight of the blessings in our lives, and the joy of the wonderful people we love and are loved by, even in the midst of grieving for that perfect child we expected to have, or struggling with the stresses that often come with PWS. Treasure those dear ones, give them a hug and tell them how much we care for them — today.

Peace, Lota

Chapter View

Here's what some of our busy chapters have been doing. For more information about a chapter in your area, call the national office for a contact name and number.

Let's all move to Florida. The **Prader-Willi Florida Association** held their Spring Conference in Gainesville, enjoyed the National Conference in Orlando in July, and then planned their Fall Conference for September 19 and 20 at Biscayne Bay in Miami. Aren't they lucky to have all those lovely places to have their meetings!

PWSA of Michigan held a September Appreciation Party to honor Jim and Carolyn Loker for their years of service as co-presidents and continued service to PWSA(USA). New Co-Presidents are Jon and Chris Hendricks.

The home of Roo and Peter Wood was the site in September for the annual Summer Picnic of **PWSA of Maryland, Virginia and D.C.**

Prader-Willi California Foundation planned an Oct. 18 daylong meeting on "Social Skills: Life's Most Important Tool", featuring distinguished experts on PWS and social skills training Scott Stiefel, M.D., and Rick Clemmons, M.A. Maybe we should live in California!

PWSA of New Jersey is sponsoring a workshop led by B.J. Goff for staff who work with adults with PWS in residential, vocational, support, or other settings on Oct. 30. B.J., a nationally known disabilities consultant specializing in PWS, is an assistant professor of education at Westfield State College in Massachusetts and coauthor of *The Student with Prader-Willi Syndrome: Information for Educators*. The fall membership meeting in November will feature a full program of presentations.

PWSA of Wisconsin presents "Educating Educators about Students with PWS" at three different locations in September, October and November. This 1½ hour training is to help educate those who work with the student with PWS. Busy Wisconsin also did a golf benefit in August.

Be My Valentine…

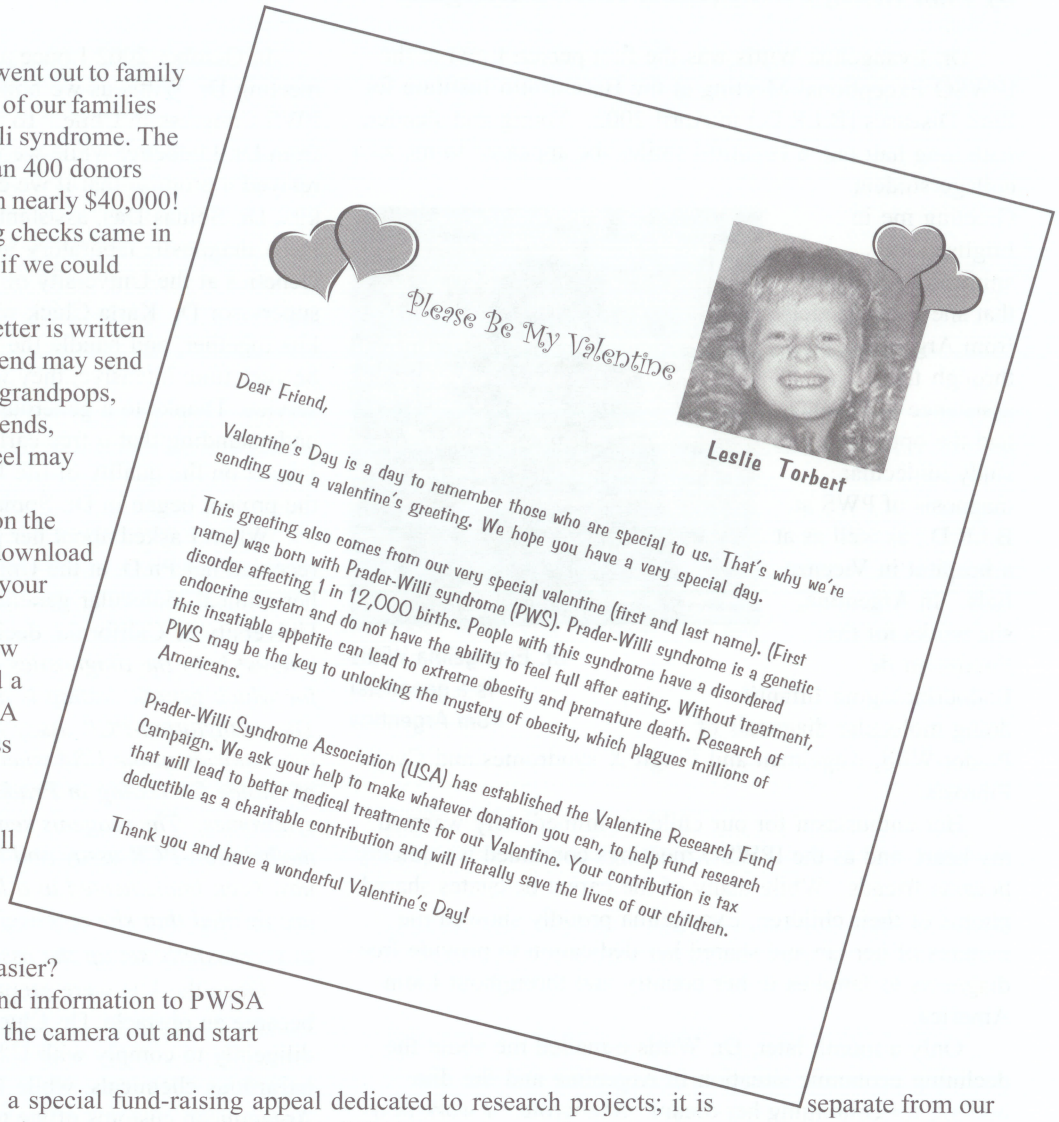
Our 2nd Annual Valentine Research Fund Campaign

Last year, a warm greeting went out to family and friends from a small number of our families whose children have Prader-Willi syndrome. The response was amazing, more than 400 donors provided the Research Fund with nearly $40,000! Small donations of $2.00 and big checks came in daily. Wouldn't it be wonderful if we could double that number this year?

Here is how it works! The letter is written so that any family member or friend may send it… aunts, uncles, grandmoms, grandpops, etc. The letter can be sent to friends, neighbors, family, anyone you feel may want to help.

The letter will be available on the web site next month for you to download and put your favorite picture of your child in it.

Oops, you say, "I don't know how to do that!" Then 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 the 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 9, 2004, so get the camera out and start snapping pictures.



Please Be My Valentine

Leslie Torbert

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!

The Valentine Research Fund is a special fund-raising appeal dedicated to research projects; it is separate from our annual Angel Fund Drive, which also supports essential PWSA (USA) operations and programs. The Valentine Fund has helped fund some of the research grants listed in this month's *Gathered View.*

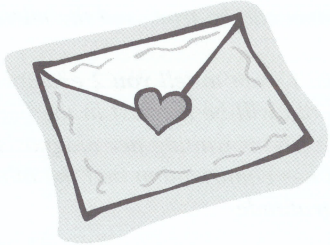
Hugs,

Carolyn Loker, VP PWSA (USA),
Parent Mentor Program Coordinator

Holding good thoughts for you and waiting to hear from you…

Diane Spencer, Support Coordinator

PWSA (USA) Valentine Research Fund Campaign



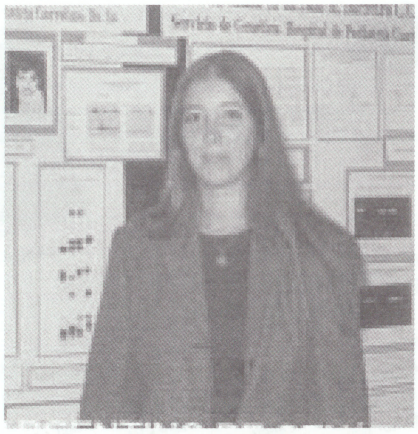
+ $$ = PWS Research

International View

'We Can Do It!' — Dream Realized in Argentina

By Pam Eisen, PWSA (USA) IPWSO Delegate

Dr. Evangelina Wittis was the first person I met at the IPWSO Exceptional Meeting at the Baschirotto Institute for Rare Diseases (B.I.R.D.) in April 2002. Young and slender, with long hair and a beautiful smile, she appeared to me as a college student.



Dr. Evangelina Wittis is a geneticist from Argentina

Greeting me in English, she animatedly told me that she is a geneticist from Argentina, and through the assistance of IPWSO had the opportunity to study molecular diagnosis of PWS at B.I.R.D., as well as at a hospital in Vicenza, Italy. In Argentina, she works for the Fundacisn de Endocrinologma Infantil doing molecular diagnosis of Prader-Willi, Angelman and Fragil X syndromes and Cystic Fibrosis.

Her enthusiasm for our children immediately warmed my heart, and as the IPWSO meetings continued we quickly became friends. While many of the parent delegates shared photos of their children, Evangelina proudly showed me pictures of her lab and shared her dedication to provide free diagnosis to families in her country and throughout Latin America.

Only a month later, Dr. Wittis e-mailed me about the declining economic situation in Argentina and the dim outlook for continuing her dream. She wrote, *"I work in a Foundation that receives donations and then collaborates with a very important Children's Hospital in Buenos Aires. This hospital assists children from all over the country and our neighboring countries and their families can't pay the studies to confirm the diagnosis because now these studies are very expensive for them. They need the study to be free. Since the kit I need is made in USA, I thought that maybe you could find the way that a laboratory manufacturing the kit for diagnosis could donate only one so that we could use it for 50 or 100 patients."*

To assist Evangelina, I contacted Dr. David Ledbetter (University of Chicago), who has long been interested in PWS and who developed the meththylation-PCR test for PWS in his lab. These two doctors were introduced over the Internet and then met personally in Buenos Aires, when Dr. Ledbetter attended a conference and gave a presentation on prenatal diagnosis. He kindly agreed to do everything possible to help Dr. Wittis and the countries of Latin America in obtaining the kits necessary for genetic testing.

In October 2002 I once again had the pleasure of meeting Dr. Wittis as we both attended the Latin American PWS Congress in Chile. To our delight, an e-mail arrived from Dr. Ledbetter while we were together. His message relayed a promise that if we could find $1,000 to cover the kits, Dr. Somas Das, assistant professor and director of the DNA diagnostic laboratory in the Department of Human Genetics at the University of Chicago, and her laboratory supervisor Dr. Karla Cluck would order the supplies, put the kits together, and handle the shipping. Although this would be very time intensive, they would not charge for their service. Thanks to a generous USA Angel, with an understanding that a free early diagnosis will have a huge impact on the quality of life for many individuals with PWS, the project began in Dr. Soma Das's laboratory.

When I asked about her work, Dr. Somas Das, who received her Ph.D. at the University College in London and her clinical molecular genetics training fellowship at the University of California, declared, *"My primary focus and interest is in the diagnostics of rare orphan genetic diseases for which genetic testing is not readily available elsewhere. The methylation-PCR assay has been adapted by several laboratories in the USA where this assay is now the method of choice for testing in Prader Willi and Angelman syndromes. The reagents sent to Vangie were for the methylation-PCR assay and I am glad that this assay has now been implemented in a laboratory in Argentina... .we are thrilled that she received them safely and has been able to successfully set up the assay in her laboratory."*

Once the kits were prepared, difficulties with shipping became an obstacle. Dr. Cluck and Dr. Soma Das worked diligently to comply with U.S. regulations for shipment of hazardous chemicals, while Dr. Wittis worked with the Argentinean customs office to help reduce the heavy taxes. I contacted our Angel for more help and was assured that the additional expensive shipping costs would be covered.

Finally, the reagents arrived in Argentina in August 2003 and Dr. Wittis wrote: *"When I read your last letter, I was very emotional. I haven't words to thanks to the man (our Angel Sponsor) to pay everything. He knows that it is for improving the diagnosis in Latinoamerica." I want to tell you that I enjoy doing all I do. I put dedication and love in my work. My motive is solidarity issues. When I can help some one, I feel completed. Sometimes I feel that nothing is impossible, that my dreams can be true, and the miracle happened.*

"About the patients, I want to tell you 2 examples: The first, a woman 17 years old will be the first in the list of patients that I will study. She is waiting her diagnosis 17 years. The important of her confirmation and the other similar cases, it is my gratitude.

Argentina continued on page 9

International View

Chile, U.S. and the World — Connected by a Song

By Carlos Molinet Sepulveda

When we look to our children, still small, we do not know what they will look like in the future, but when we have the chance to share with so many persons affected by Prader-Willi syndrome, of different ages, we can say, that they are so tender, kind, and loving the same way, they are now.

That was our experience attending to the Prader-Willi syndrome congress in United States, a wonderful congress, with many children and families, from many states and countries.

At this beautiful congress, where so many people and professionals of the highest level attended, is now a tradition to feel the warm of the family, to feel that we are together and the main objective is the well being and the love of our children. To feel also, that no matter the social condition, language or race, we hold the hand of the person who is close to us and then raise it with emotion and pride, when Giorgio Fornasier sings the song which represents all PWS organizations around the world.

That was the emotion we felt at the congress, to see the unity, dedication and love of so many persons that worked very hard to make the congress a success.

To feel the love of the children of Janalee and her husband, when they entertain all those persons affected by the PWS at the different halls of the hotel, organized games, singing songs, or when they hugged, looking to each other eyes, when dancing those romantic songs. I had never enjoyed that much looking to all those little persons fulled of love, purity and joy, that moved me, as if my daughter was there.

As Chileans, we also felt in our hearts, to pay homage to the United States organization on its independence day, dancing our national dance, "La Cueca Chilena." It was very nice to see all the persons, along with the children,

Janalee Heinemann with Carlos and Jan Sepulveda and Pam Eisen

clapping their hands to the rhythm of the music, to see Janalee and Giorgio dancing, it was really a pride for us as Chileans.

The final day, at the hall of the hotel with many people, then suddenly appeared all the little persons affected by PWS, dancing and singing the song which since now will identify them. "My Name is Not Willi." This song describes the characteristics of the persons affected by this rare sickness, they were all happy, they felt loved, by their proud parents and no matter how hard are the things, the sun is always shining on our lives and to the love of our hearts.

Children of the world affected by the PWS, thanks many thanks for allowing use to overcome so many things and to know, thanks to all of you, the solidarity and the love.

Carlos Molinet Sepulveda is president of the Prader Willi Sindrome Association – Chile

Argentina - *continued from page 8*

"The second, the last years people of different countries wrote to me told me their difficulties about obtaining a confirmation. Now I have a good answer to them. We can do it."

In recognition of PWSA (USA)'s role in making this dream a reality, the Management Board of the Foundation de Endocrinologia Infantil wrote to thank us for the *"privilege of having reached the first stage of such an important project oriented to offer molecular studies to confirm the diagnosis of PWS in patients who lack the economic means to have access to them on account of the difficulties all Latin American countries are going thorough. We are thankful for all the good will, time and effort you devoted to this cause and also for understanding the importance of such a generous contribution."*

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Fund-raising for the love of Patty

Brothers Meet Prader-Willi English Channel Challenge

They made it!

The five Roberts brothers — Andy (43), Matt (42), Courtney (41), Michael (39), and Jim (38) — swam 22 miles across the English Channel in August as a tribute to their sister Patty, who has PWS. In the process, they created world awareness about Prader-Willi syndrome.

"We want to experience the challenge of swimming this ultimate open-water swimming event... and, most importantly, to recognize and honor Patty, who has struggled and achieved so much and continues to do so in a way we will never fully understand," Andy Roberts said prior to the swim. "Our lives have been made better, our experiences richer, our memories sweeter because of Patty."

Patty (center) with her brothers and the Prader-Willi English Channel Challenge team

The brothers accomplished their Prader-Willi English Channel Challenge in 11 hours, 23 minutes and 14 seconds. "We swam the English Channel," they shouted, "Can you believe it!"

"We were proud of our accomplishment, happy for Patty, and glad that God gave us the talents to honor her in such an extraordinary way," said Mike.

When her brothers arrived at the dock, Patty was waiting. She proudly displayed a large neon colored poster with a picture of the team and her. On it Patty had written: "You Did it! Thanks Bros. Love, Patty."

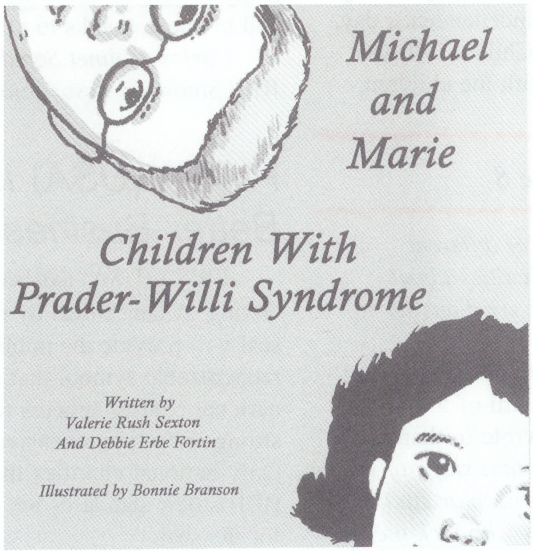
A special thank you to all who donated to this cause.

— Jane Phelan, Editor

New Storybook Helps Children Learn About PWS

Michael & Marie: Children with Prader Willi Syndrome, was written by two teachers, Valerie Rush Sexton and Debbie Fortin.

It addresses a void in the educational system, as there are no storybooks about PWS. The story was designed to be read to elementary school age children. Classmates of special needs children need to be educated to understand and even help create a friendly and safe environment for all children.



The story and illustrations reflect the authors' vision of how to explain the difficulties and achievements that two children with Prader-Willi syndrome encounter daily in a school system.

At the end of the story is an accompanying lesson plan that provides a simulated activity of how a child with PWS feels while waiting for snack time. The lesson plan has been successfully implemented with a group of fourth grade students.

Valerie Rush Sexton is a speech language pathologist, and Debbie Erbe Fortin, a special education paraprofessional. Bonnie Branson is a graduate from the School of Visual Arts with a BFA in Illustration.

This book would make a great birthday gift to your school library, inscribed inside as donated by your child. It is also just in time for holiday gift giving. We are happy to add this book to our "Educator's Awareness" inventory.

The book can be ordered by phone at 1-800-926-4797, on the Internet at www.pwsausa.org or just by mailing in your order to the PWSA (USA) national office. The price is $8.00 plus $1.50 shipping (if only ordering this one item).

— Diane Spencer, CIBC Support Coordinator

View From The Home Front

A Journey of Hope...

By Debbie Robinson

Above my son's crib there was a wall hanging, a gift from his Godfather. Its message was obscured among the medical supplies at the never-ending hum of the tube feeding pump. It read miracles still happen. My eyes were too bleary to see. I could not wrap my brain around the irony; I could scarcely focus on day-to-day activities. Perhaps the miracle is surviving.

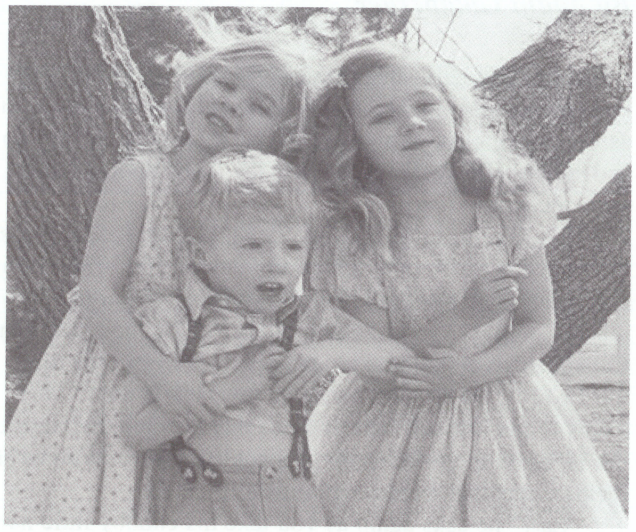
Hope was given to me and promises made by strangers who called. They said it's not that bad, things will get better. Slowly with each phone call I began to believe and then I began to pray.

At five months of age my son's feeding tube was removed. Slowly he began to make progress. I knew that by nine months he'd be sitting, but I was wrong. On his first birthday he could sit momentarily and would then fall back.

A theme kept running through my mind. God helps those who help themselves. If I do my best then God will do the rest.

I learned all I could about low muscle tone, sensory integration and sign language. I learned to teach my son things that came naturally to other kids. I balanced my time among my son and my other two small children. My husband worked long hours to support us because I could no longer work. He somehow found the time to study about medicine and to look for answers.

As for now, my son can communicate well enough to make his needs known. Not only can my son walk but he is starting to run. He can chase chickens and sometimes even catches one. After it rains my 3-year-old goes outside and looks for toads and usually finds one or two, and I believe that one day he will catch a lightning bug.



Andrew Robinson, age 3, who has PWS, with his sisters Andrea, age 5, and Ashley, age 6½

It may not be manna from heaven, nor is it the parting of the red sea. But the energetic, fun-loving little boy my son has become as well as the joy he gives is nothing less than a miracle.

The road ahead is long. But we have come so far. I know that there will be miracles not only for my son but for all our children.

...And A Journey of Gratitude

I felt isolated and afraid of being deserted. I told my neighbor I couldn't hold him. I just want to go in another room and pretend he's not here. She came and she held him and I was grateful.

I am grateful for all who come to visit, for all who care and for all those who pray. I am grateful for my case manager who supports me and my family and for all the professionals who come to do the things that I cannot.

I am grateful to the doctors who care and know what to do. I am grateful for the hope of modern technology.

I am grateful for my husband and family. I am grateful that the grass is green and that there is a sun in the sky. Best of all I am grateful for feeling grateful.

Debbie and Tom Robinson live with their three children, Ashley, Andrea and Andrew, who has PWS, in Westerville, Ohio.

The Chuckle Corner

A Very Special Club

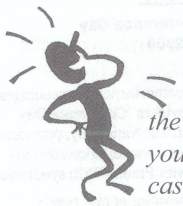
The youngest child of Shanin and Gage Haverfield of Uniontown, Ohio, is their daughter Claudia, age 4.

A couple years ago whenever her mother mentioned anything about going to do something for Prader-Willi syndrome, her big brothers Clay and Camden, then 5 and 6, would say, "Oh, that's the club that Claudia belongs to."

They thought she belonged to a special club called "The Prader-Willi Club." (Shanin suspects they may still think this.)

They didn't quite understand the meaning of it all, but they knew when kids get together it is fun — just like their Faith Weavers (a church group) and Tiger Scouts.

-- Lota Mitchell, Associate Editor

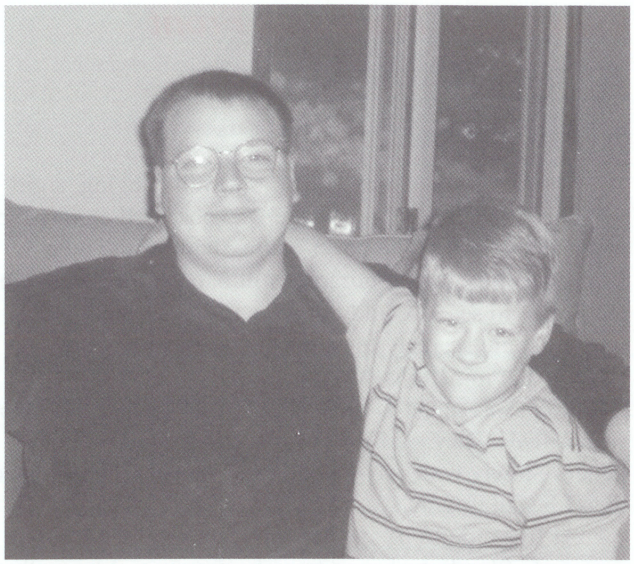


Please send your joke or funny story to the PWSA (USA) office. Be sure to include your name, phone number and address in case we have any questions.

The Sibling View

Caring for Callaghan

By Adam Goldberger



Adam Goldberger helps care for his nephew Callaghan, 6, who has PWS.

A chair quietly slides across the linoleum floor of the kitchen over to a high cabinet. You walk into the room just as Callaghan climbs onto the chair. After picking him up and setting him back on the floor, he looks up to you and asks one word: "Eat?"

You have to smile and slowly shake your head because he has eaten lunch only a half an hour earlier. Callaghan, a 6-year-old boy, has Prader-Willi syndrome. Among many other symptoms, an insatiable hunger is the most prevalent. Callaghan's verbal apraxia prevents him from speaking most words, but he can manage a few simple ones. He wants to get into the cabinet since that is where the keys to the locks on the refrigerator and pantry reside. Without the locks, Callaghan would eat himself to death.

"Eat?" he asks again as you take his chubby little hand and lead him back to the couch and the book he was looking at 2 minutes before. You notice another ripped-out page lying on the carpet. You pick it up and throw it away as you've done twice before that day.

He smiles brightly and asks once more, "Eat?" You tell him that he can have an apple, and he smiles and laughs excitedly.

Glinting off Callaghan's hair, the light makes his blonde hair look golden. He looks at you with his twinkling blue eyes and flashes a heart-warming smile. Returning his attention to his book, he sits there for the next few hours, occasionally looking at you and asking, "Eat?"

Dinnertime rolls around, and when Callaghan hears you getting out the pots and pans for cooking, he runs into the kitchen and asks, "Eat?" The glow that radiates from his face as you tell him you are cooking dinner shines extraordinarily. "Eat!" he exclaims before he gets out plates and cups for himself and his younger brother.

Dinner finishes cooking and the rest of the table is set. After saying prayers, Callaghan hands you his plate. You dish out a small serving for him and his two brothers. You finish dishing out your own serving right before he asks for more. Taking his plate, you dish out a little more food and tell him that that is all. You have accustomed yourself to eating fast to prevent Callaghan from asking for more food, since he's always the first to finish. However, it doesn't help.

After dinner, the boys get ready for their bath. You tell the oldest to make sure his younger brothers wash their hair well. You take some time to clean the house while the boys are in the shower.

Callaghan is the first one out of the shower. He walks into the living room with a towel draped over him, water dripping off his shivering body. He smiles brightly as you dry him off and asks once more, "Eat?" You tell him that he can have an apple, and he smiles and laughs excitedly.

While you get the apple for him, Callaghan puts an animated movie in the VCR. He walks over to the silverware drawer and gets out his purple plastic knife. He takes the apple from you and sets it on the cutting board and proceeds to cut the apple in half horizontally. Walking back to the couch, he watches the movie, occasionally rewinding it so he can watch some parts a second or third time.

Callaghan continued on page 13

About Adam

Adam Goldberger, 23, is a Microsoft certified systems engineer. He is studying at the University of Illinois at Springfield to earn an advanced degree in Computer Science.

He lives with his sister Regina Hartnett and her three boys, Callaghan, 6, and Reilly, 5, who both have PWS, and Devereaux, 11.

Adam and his 25-year-old brother Aaron were sharing an apartment, but when Aaron got a job in another town, Regina offered Adam the opportunity to come live with them. Adam helps with the care of the boys, including assisting Dev with his homework and giving Callaghan and Reilly their growth hormone shots.

Adam had planned on living with Regina no more than a year, but after her husband Bob died, Adam decided to stay with her to help with the boys. Regina has returned to school, intending to earn a Ph.D. in family counseling.

Conference 2004

Plan Ahead: Sawmill Creek Resort will be a WOW!

By Lota Mitchell, Associate Editor

On a crisp late September day just dripping sunshine, I left the hills of Pittsburgh for the flatlands of Ohio. Destination: Sawmill Creek Resort and Conference Center in Huron, site of next year's national PWSA(USA) mini-conference. Purpose: meeting of conference Co-Chairs Carolyn Loker, Johanna Costello, and Janice Agarwal and committee members to review space and start planning (well, more accurately, continue planning; lots of work had already gone into Conference 2004).

I had been at the Lodge at Sawmill Creek almost two years earlier — and fell in love with it — when I attended an Ohio Chapter family social which took place there. At that time this beautiful resort was in the process of "growing up" to be a conference center as well. The finished conference center is just as wonderful (I am so enthusiastic about it I feel I can gush a bit) as the Lodge.

There are all sorts of resort and area activities, like tennis, swimming in the big indoor pool or the big outdoor pool, Nature Reserve walking trail, a marina (close to Lake Erie), golf, and the well-known Cedar Point Amusement Park just minutes away. In other words, Wow! It's a great vacation spot!

Our committee met in the Bobcat Room, in between Wilderness Hall, the big conference area, and an indoor waterfall graced by a… bobcat?… mountain lion?… lynx?… well, some variety of the cat family that you don't pet. Other meeting rooms have nature-related names like Sycamore, Brook Trout, Wolf and Coyote. Indian motif and inviting spots with fireplaces and comfortable chairs to just "hang out" abound. Sleeping rooms are in buildings with names like Shawnee and Mohawk. Everything from the Lodge to the Conference Center except the Sawmill shops are connected, so if it rains, you don't get wet.

Mini-conference 2004 will take place on Thursday, June 30 and Friday, July 1 with Scientific Day, Providers Day, and Chapter Presidents meeting on Wednesday, June 29. Mark those dates!

Rooms are at a discounted rate of $99 per night for up to four people, and, good news, parking is free. Make your room reservations through Globetrotter Travel by calling 800-322-7032 or email pwsa-use@globetrottermgmt.com. The committee has booked several rooms to remain discounted for Saturday night, so stay over and vacation. Rates for conference and YIP registration aren't available yet; they are still in the process of being worked out. Soon!

Child care will be provided for infants and children up through age 9. The Youth & Infant Program (YIP) is for those with PWS and their siblings in this age range. It is **very important to sign up early for the YIP.** The numbers in Orlando for kids 9 and under was high (110), and space at Sawmill Creek is limited.

In fact, it is important for everyone who plans to go to sign up early as possible in order to be assured of having a room at the Lodge.

It is also essential to understand that **programming for the general conference will be for parents and caregivers of individuals with PWS of all ages.** Although child care is limited to certain ages, the conference is for everyone. The goal is to have speakers of high quality for all, speakers who make their listeners say Wow!

Driving back to Pittsburgh the next day, I felt a sense of exhilaration and anticipation — and the certainty that Conference 2004 is going to be another great one, a real WOW! Be part of it. Meet other parents. Share your experiences. Learn more about the syndrome and current research. Join us!

Callaghan - *continued from page 12*

When the movie ends, you get the keys to the medicine cabinet and take out Callaghan's evening medication. Hearing the jingle of the keys, he walks over to you and asks, "Eat?" You tell him that it's just time for his pills and shot. You give them to him and have him get ready for bed.

After tucking Callaghan and his brothers in for the night, you take the next couple hours for yourself before going to bed.

"Eat!" You open your eyes and look at the clock. The red block numbers read 5:23 a.m.

"Eat!" Callaghan yells again. You get out of the bed and walk out into the living room. Callaghan looks up at you and quietly asks, "Eat?" Nodding sleepily, you get the keys from the cabinet.

Callaghan busies himself getting a bowl and a spoon while you unlock the refrigerator and pantry. You take out the jug of milk, Rice Crispies and Cheerios to prevent the morning fit he'll throw if he doesn't have two different types of cereal. Fetching his morning medication, you place the pills in his mouth then go about locking everything up. You return the keys to the cabinet, placing them behind the coffee can, making sure they are well hidden, and securing the child lock back in place.

Callaghan finishes his breakfast as you crawl back into bed. He settles down on the couch with his book, and just as you start drifting off back to sleep, you hear the tearing of a page.

We welcome writings from siblings for *The Sibling View:* good experiences, bad experiences, we want to hear what you think. They can be signed or anonymous, whichever you prefer. Send them to the attention of Lota Mitchell at the PWSA (USA) national office, or e-mail to her at ljecholsm@juno.com.

Effects of Growth Hormone on Respiratory Function and Sleep Patterns in PWS

Jennifer Miller, Janet Silverstein, Abby Wagner, Daniel J. Driscoll.
Divisions of Pediatric Endocrinology, Genetics & Pulmonary, Department of Pediatrics, College of Medicine University of Florida, Gainesville, Florida.

Discussion: All patients with PWS studied had some degree of either obstructive sleep apnea (OSA) or central sleep apnea. Further studies must be done to determine which sleep patterns are ameliorated with GH treatment and to determine which, if any, sleep abnormalities are worsened by GH treatment. The only normal studies in our cohort were two children on GH, one of whom had had a previously abnormal study.

Adrenarche In Prader Willi Syndrome Appears Not Related To Insulin Sensitivity And Serum Adiponectin

Nancy Unanue [1]*, Germán Iñiguez [1], Fanny Cortés [2], Alejandra Avila [1] and Verónica Mericq [1]. [1] Institute of Maternal and Child Research, University of Chile, Santiago, Chile, and [2] Institute of Nutrition and Food Technology, University of Chile, Santiago, Chile.

Discussion: Significantly lower blood glucose and higher 17 OH prog and DHEAS levels were found in PWS patients. Serum adiponectin levels were similar in PWS and obese matched controls (OC); therefore it is unlikely that differences in insulin sensitivity and adiponectin are related to the development of premature adrenarche in these patients.

Growth Hormone Deficiency in Adult PWS Patients: *Preliminary Findings from a 4-site Study of Genotropin Treatment*

Harriette Mogul[1], Michael Frey[1], ***Barbara Y. Whitman[2]***, Susan E. Myers[2], Phillip D.K. Lee[3], William Zipf[4], Belinda Pinyard.[4] [1]Medical College of New York, Valhalla, NY; [2]Department of Pediatrics, St. Louis University School of Medicine, St. Louis, MO 63104; [3]University of California at Los Angeles, Los Angeles, CA; [4]Columbus, Ohio.

Results and Discussion: Growth hormone replacement was well tolerated by all study subjects with no reported emergent adverse events. Most adults, including those in group homes, were able to administer their own injections with supervision. An illustrative case example including lab values and anthropometrics was presented. Results from this study will provide detailed information on the efficacy and safety of short-term GH treatment in GH deficient adults with PWS.

Maladaptive And Compulsive Behavior In Prader-Willi Syndrome: New Insights From Older Adults

Elisabeth M Dykens, Ph.D., UCLA Neuropsychiatric Institute, Los Angeles, Ca.

Results and Discussion: As expected, gains in certain maladaptive and compulsive behaviors were found across the child and adolescent years. Young adults in their twenties appear to be at particularly high risk for elevated symptoms, even compared to the adolescent period. For the first time ever, we show that both maladaptive and compulsive symptoms diminish to an astonishing degree in older adults with PWS. Sweeping, across-the-board declines were found in externalizing behaviors, aggression, overall problems, and the number and severity of compulsive symptoms, including skin picking. We also examined predictors of these patterns, specifically gender, IQ, BMI, and living status. Controlling for the effects of age, gender and the BMI emerged as the most consistent, significant predictors of skin picking and other symptoms in both children and adults. Possible reasons for declines in problems among older adults were discussed, including the selective survival hypothesis. Findings pave the way for longitudinal studies of aging in PWS, and possible medical, hormonal, and environmental predictors of more successful outcome in older adults with PWS.

Divergence of the Prader-Willi Syndrome Imprinting Center

Karen A. Johnstone, James L. Resnick and Camilynn I. Brannan. Department of Molecular Genetics and Microbiology, University of Florida, Gainesville, Florida.

Results and Discussion: We previously tried to exploit the similarities between the two species by using a human transgene to study the mechanism of imprinting in mouse. However, we found that the human gene was not imprinted in mouse, suggesting that the sequences and factors that mediate imprinting have diverged. To study the processes involved in more detail, we have engineered mice in which imprinting of the region is under the control of human sequences. These mice demonstrate that the divergence between the species is limited to the imprinting sequences. The processes affected involve both parental-specific gene silencing and parental-specific gene activation indicating that both are active processes. In addition, the mice are likely to provide some further insights into the phenotypes associated with PWS.

Transgenic Rescue of the PWS-IC Deletion Mouse

Stormy Jo Chamberlain, James Resnick and Camilynn I. Brannan. Department of Molecular Genetics and Microbiology, University of Florida, Gainesville, Florida.

Results and Discussion: In total, we have found that the PWS phenotype is less severe on certain strain backgrounds, suggesting the presence of modifier genes that may ameliorate the PWS phenotype. We have also determined that *Mkrn3*, *Magel2*, *Ndn*, *Snrpn*, and 5 species of snoRNAs are not solely responsible for neonatal failure to thrive. Finally, we demonstrated that the small stature phenotype of PWS-IC deletion mice could be rescued by a single transgenic line that is the only one showing expression of one particular snoRNA species, suggesting that this tandemly repeated snoRNA cluster is involved in the

Abstracts continued on page 15

Highlights - *continued from page 14*

regulation of growth of PWS-IC deletion mice, and most likely PWS individuals.

Regulation of Imprinting at the H19/Igf2 Locus
Marisa S. Bartolomei, Joanne L. Thorvaldsen, Nora I. Engel, Andrew Fedoriw. Department of Cell & Developmental Biology, University of Pennsylvania School of Medicine, Philadelphia, Pennsylvania.
Results and Discussion: These experiments suggest that the sites governing repression of the paternal *H19* allele and those that mediate insulator activity at this locus are distinct. Furthermore, these experiments show that mutation of only 9 CpG dinucleotides within a 2 kb region that contains over 50 CpG dinucleotides is sufficient to perturb the ability of the paternal allele to confer and maintain *H19* repression. Experiments are also in progress to determine whether CTCF protects the maternal DMD from DNA methylation.

Aimee - *continued from page 1*

family. I thought I had signed up for more of the same with the twins, along with the ordinary difficulties that come with multiple children, but certainly not for the anguish of dealing with a birth defect.

The weekend after Aimee's diagnosis was the worst time I've ever faced, but I somehow survived it. It took a while, and it's not something a parent probably ever gets over, but I'm happy to say that I'm growing more in love with my smallest girl every day.

Tim and Dawn Atwood are the parents of Caitlin, Jolie and Aimee, who has PWS. They live in Burlington, Mass.

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