



The *Gathered View*

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

10 Years of Progress

Age of Person With PWS When First Contact Made to PWSA (USA)



Mary Patterson-Ladonne

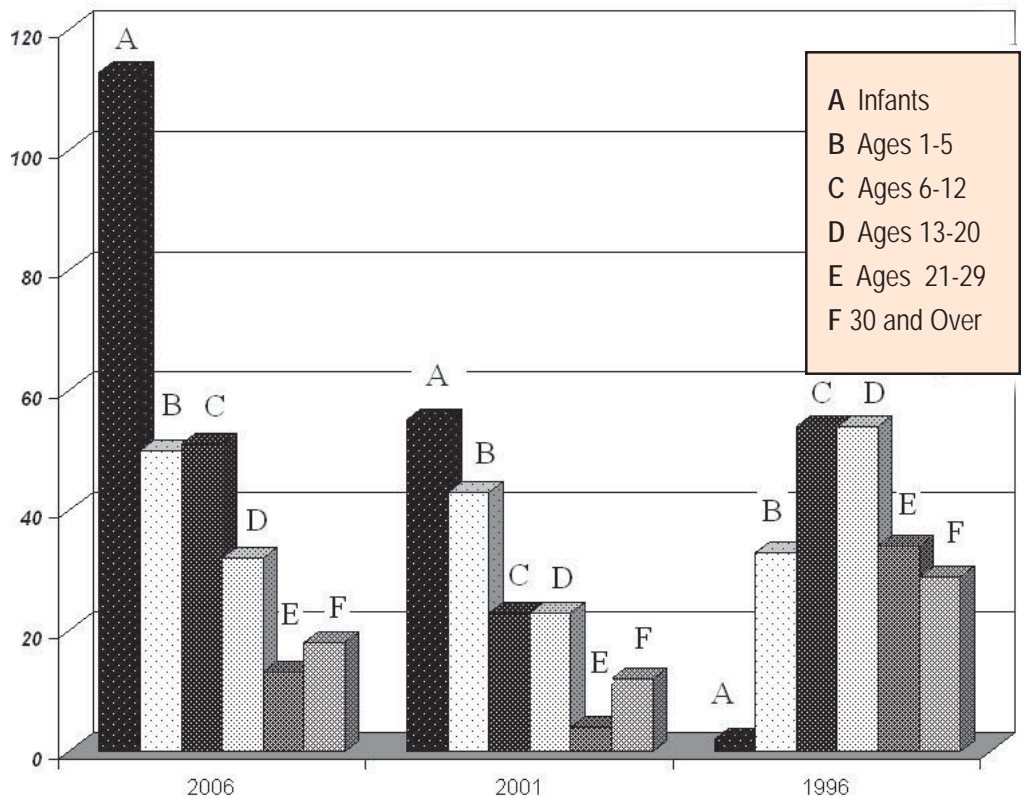
Season's Greetings

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Donations for Research

Through 9/ 30/ 2006:
\$200,000



This graph, one of the most telling statistics of how far we have come in the last 10 years, shows the number of infants being diagnosed and families joining PWSA (USA) when their child is very young compared to 10 years ago. One of the big changes from 1996 to now is the DNA methylation analysis. In 1996 Dr. Daniel Driscoll, our Clinical Advisory Board chair, in collaboration with Dr. Robert Nicholls, our Scientific Advisory Board member, had just “rolled out” the third generation DNA methylation analysis (SNRPN) which was very powerful,

sensitive and specific and which could diagnose almost 100% of all PWS. (See Dr. Nicholls’ article on page 7 for more about the test.) This, combined with better awareness (much through our New Parent Mentoring Program, extensive web site, medical awareness booths, and community events), has led physicians to do the DNA methylation analysis in infants in the NICUs and then refer them to us for education and support.

See page 3 for more on our years of progress

Prader-Willi Syndrome Association (USA)

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Deadlines to submit items to

The Gathered View are:

December 1; February 1; April 1; June 1;
August 1; October 1

Our Mission: *PWSA (USA) is an organization of families and professionals working together to promote and fund research, provide education, and offer support to enhance the quality of life of those impacted by Prader-Willi syndrome.*

Members Only: Check our website www.pwsausa.org for downloadable publications, current news, current research and much, much more limited to Members Only!

User Name: pwsamember Password this issue: bears06

Note: If you have difficulty logging in to the site, you may be using a browser that prevents you from entering authenticated websites. Try minimizing your program (e.g. AOL) and clicking on Internet Explorer or Netscape. Then type in the URL: <http://www.pwsausa.org/memberonly.htm>

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Looking Back and Moving Forward

Janalee Heinemann

Dear Members,

I am now off chemotherapy and feeling great! I have decided that it is time for me to transition into a new role with PWSA (USA) and give someone else the opportunity to move this organization forward. This was my plan even prior to being diagnosed with cancer. It has more to do with my age (61) than my medical status. As many of you know, I have been intimately involved with PWSA (USA) for more than 20 years — as a volunteer board member, president, writer, and then executive director for the last 9 years.

Because my current role as executive director is so multi-faceted, in the next year we will be segmenting what I do into two separate positions. I will become the Medical Affairs Liaison working with medical crises, research, publications and conference. This plays to my strengths, and allows the next executive to be able to focus on the true role of an executive director.

I reflect here on the growth of our organization over the last 10 years and thank our past and current board members and officers who have all been a part of that growth.

Services offered when I started in 1997:

- Newsletter
- National conference
- Chapter support
- Research – in 1996, we gave \$5,800 in grants
- A few publications
- 800 phone number with me answering all questions
- Scientific Advisory Board

Services added over 9 years to bring us to date in 2006:

- **New Parent Mentoring Program** — 865 families served
- **Crisis Intervention and Prevention Program** — 483 families served in 2005. This number has already been exceeded in the first nine months of 2006, and September had a record number of 91 crises served.
- **Bereavement Program**
- **Extensive 2,851-page web site** — from 1 page in 1997!
- **Publications** now include 34 booklets and books, 14 brochures, and DVD's and videos — the largest number of publications on PWS in the world
- **Clinical Advisory Board**
- **Medical Awareness booths & national medical mailings**

- **Research expanded** — more than \$200,000 a year in grant commitments and impact research in many other ways
- **Extensive e-mail support and eight specialized e-mail support groups**
- **Grandparent Mentoring Program** — more than 700 in the database and 400+ as members
- **Medical database** — 1,471 children & adults with PWS – the largest in the world
- **Study of deaths** — 178 deaths – the largest study on PWS deaths in the world
- **Adults with PWS Advisory Board** — Support specifically for those with PWS was non-existent in the past. The board was founded, a book, exercise DVD, and rap CD were all created for the person with PWS.
- **International support** has gone from a few e-mails and letters to daily e-mails to and from other countries. Our extensive educational materials have been translated into many languages, and communication with key physicians worldwide has increased our visibility. Through our outreach we have assisted more families and professionals around the world than any other country.
- **Community fundraisers** — In 2005, PWSA (USA) had 29 families and friends rally to our cause with community projects. In 2006, at least 45 events are planned or have been completed! This does not include the chapter events. Imagine how this spreads the word to thousands through media coverage, flyers, word of mouth, etc.
- **PWSA (USA) is now approved by give.org, the Combined Federal Campaign, and is registered to accept donations in every state.**

The succeeding executive director needs to take PWSA (USA) to the next level while also acknowledging the people who have done so much — who have helped turned PWSA (USA) into one of the most respected organizations for rare disorders. The fact that as PWSA (USA)'s executive director I have been asked to present in France at a symposium of professionals on the topic of "What We Can Learn From Parents And Associations" demonstrates our high standing in the worldwide PWS community.

I am proud of my role with this association and hope to remain a viable team member for a long time, but the time is near for me to pass the torch of leadership.



President's View

Join Our 5th Annual Valentine Research Fund Campaign

Carolyn Loker

Boy, do we have some exciting news to share with you! Our fabulous Valentine families raised \$87,500 in our 4th Annual Valentine Research Fund Campaign.

This amount more than doubled our 3rd Valentine campaign which raised \$38,000. Since the campaign began 5 years ago, our grand total is \$193,657.

Many thanks to our 44 families and their sweet little Valentines who made 2006 so successful.

It is exciting to see the research that is being funded by PWSA (USA). Check our website at www.pwsausa.org and take a peek at the studies being funded and also the results of those studies.

Please join us and let's see if we can reach a goal of \$100,000 or even greater for the 2007 Valentine Campaign.

Here's the scoop: You can mail in your special Valentine picture with the number of ready-made letters you want us to print to PWSA (USA), 5700 Midnight Pass Road, Suite 6, Sarasota, FL 34242 Attn. Diane. Or e-mail Diane Spencer at national@pwsausa.org with a scanned picture and number of letters you wish to send, along with your mailing contact information.

The deadline to send in your special Valentine picture is January 25, 2007. Please call Diane at 800-926-4797 with any questions. We'll send the letters back to you ready to sign and address. The letter will be available online in January at www.pwsausa.org for you to

Working For A Cure Tomorrow – Helping Our Families Today.

Please Be My Valentine



Jacob Fiske

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 girls. 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.

On Valentine's Day we often give a loved one a heart-shaped box of delicious candies. Today, the heart-shaped box we give (First Name) contains not chocolates, but all our love and hope. With your help, our dream of a better future for (Name) and for all people who have Prader-Willi syndrome can become a reality. Instead of a heart-shaped box filled with chocolates, please send your love in the form of a contribution to the PWSA (USA) Valentine Research Fund.

Thank you and have a wonderful Valentine's Day!

download and insert a picture if you wish to do this yourself. For those wishing to participate, we will make a special donation web page with your special Valentine's picture on it. Call Diane if you would like a flyer including your special Valentine's picture for the workplace bulletin board.

If you'd like to make a single donation specifically for research, you can donate online at our website or detach the form on page 5 naming your special Valentine and mail to the PWSA (USA) office.

Each year we have exceeded expectations.

I can hardly wait to see what 2007 brings!

In 2006 our special Valentine sweethearts were: Aimee Atwood * Rebecca Baird * Sophia Bolander * Isabella Burnham * Joshua David Carter * Laura Cassady * Clara Ciuriuc * Grace Culley * Tanner Dean * Brooke Detiege * Kaitlyn Disney * Erin Favret * Tristan Ferdig * Jacob Fiske * Peter Funai * Brandon Greco * Anna Guthrie * Claudia Haverfield * Morgan Heffner * Hannah Herne * Jayden Hippert * Jakayla Howard * Casey Howell * Kate Kane * Jake Klauber * David, Ben, & Caroline Knopf * Dylan Krambeer * Autumn Leigh Letzo * Anna Loker * Jake Pawulak * Callasandra May O'Connor * Dallas Rincon * Ryland Sanders * Reagan Seely * Abby Heathman * Phillip Fusco * Jacob Thorne Summerlin * Leslie Torbert * Jacob Ziliox * Faith Morse * Ben Hopkins * Pat Rupe * Lindsay & Erin Anderson * Cameron Graziano



Holiday Alert



Last holiday season, in spite of all of our warnings and our massive mailing to all providers, we had two more unexpected deaths due to food-binging episodes that led to necrosis of the stomach wall and perforation (tear) in the stomach.

We also had a recent death of a young woman whose overeating binge occurred while she was hospitalized! **(If your child is hospitalized, take a copy of the Medical Alert for Inpatient Providers from the Sept.-Oct. 2006 Gathered View and now on www.pwsausa.org).**

In these cases, the person with PWS was slim, so there was no great concern about weight gain. Except for the one woman with PWS in the hospital, the others were typically in festive, family group situations, where everyone was watching, which meant no one was watching.

Keep in mind that a person with PWS who is slim does not necessarily have total food control. Add too many temptations at hand, the lack of feeling full and the high pain threshold, and you have the potential of filling the stomach dangerously full.

Because we have many food-binging episodes of our children and adults with PWS — most without such disastrous results — we think there are probably other factors that play into this life-threatening scenario. But until we have completed our research on the study of deaths, we feel obliged to again forewarn parents, grandparents and caregivers of this potential threat.

We and our death study committee members, spearheaded by David Stevenson, M.D., are working to get this information out by reporting at major medical meetings. We have also submitted for publication in an emergency room journal for medical professionals.

Another outcome of this research sponsored by PWSA (USA) is an article on “Deaths Due to Choking in Prader-Willi syndrome.” It will be published in the *American Journal of Medical Genetics*.

PWS Gifts for the Season

Wonderful Wrist-A-Bouts

Brooke Slabaugh, 9, of Alanson, Michigan, who has PWS, models one of our orange **PWSA (USA) wrist-a-bouts** in the photo below. What a great way to promote PWS awareness! Why not give them as holiday gifts for your friends and family? They're only \$2 each, free shipping for orders of 5 or more.



Online PWS Store

Our **PWS Store** of logo items is open for business 24/7 — shop for unique and useful items and avoid holiday crowds at the mall. Choose shirts, mugs, tote bags, beach towels, and more! All items over \$10 are 20% off!. See www.pwsausa.org for more details and for other great deals, too.

PWSA (USA) Angel Fund

And, for the gift that will keep on giving, why not contribute to this year's **PWSA (USA) Angel Fund** campaign? Angel Fund donation cards are in the mail, and your support is essential. Remember, PWSA (USA) is the only national membership organization that supports and educates all about PWS, including research, awareness and education, family support and expanding knowledge of PWS-related medical issues. We can't do it without YOU!

Valentine Research Fund Donor Form

Mail to: PWSA (USA), 5700 Midnight Pass Rd., Suite 6, Sarasota, FL 34242

The Valentine Research Fund Campaign is our only fundraising appeal dedicated solely to research projects. It is separate from our annual Angel Fund Drive, which also supports essential PWSA (USA) operations and programs. Please help us find treatments to help our children. Many thanks!

My Valentine is _____ My Name is _____

My Address _____ City _____ St _____ Zip _____

Telephone # _____ Donation \$ _____ By Check _____ By Credit Card _____

Credit Card # _____ Exp Date _____

Name on Card _____ Your Signature _____

Please make checks payable to PWSA (USA). Or make your donation online at www.pwsausa.org.

Click on "Valentine Research Donation." THANK YOU!

Research View

Brain Developmental Abnormalities in PWS Detected by Diffusion Tensor Imaging

Yamada K, Matsuzawa H, Uchiyama M, Kwee IL, Nakada T., *Pediatrics* 2006 Aug;118(2): 442-448

Abstracted by Jamie Bassel, D.C.

This latest study out of the Center for Integrated Human Brain Science at the University of Niigata in Japan looked at studying the developmental brain irregularities thought to be present in PWS. It is believed to be one of the first objective studies to use technology to study various aspects of the brain to determine the regions that may be affected by PWS.

Using a technique called Diffusion Tensor Imaging, 16 patients were studied (8 age- and gender- matched patients with PWS and 8 age- and gender-matched control patients). Diffusion tensor imaging (DTI) is a technique based on MRI (Magnetic Resonance Imaging). It visualizes the location and the orientation of the brain's white matter tracts. The imaging of this white matter property is an extension of diffusion magnetic resonance. Clinical applications of DTI are the tract-specific localization of white matter abnormalities.

Based on the study, the PWS group demonstrated higher measurements in Trace Values (TV) in two areas of the brain: the left frontal white matter and the left dorsomedial thalamus. Minimal orientation changes were noted in other areas, such as the splenium of the corpus colosum, right frontal white matter and posterior limb of the internal capsule (PLIC) on both sides.

There appears to be a high correlation between structural differences viewed in this study compared to established clinical features described at length in other medical literature. This study provides additional support suggesting de-

velopmental changes that can be visualized in developing brains of PWS patients. Further studies may be enhanced and focus on the structure and neurologic function of those with PWS.

Motor dysfunction, one of the hallmark features of PWS, has been shown to be a central nervous system problem rather than a muscular issue. Motor dysfunction has been related to the extent of the altered structure of the PLIC.

Clinical DTI studies have shown that changes in fractional anisotropy (FA) have been related to conditions such as interrupted connectivity, deterioration and disturbance of white matter. FA is a value used in this kind of imagery to display the directional changes observed in the brain's white matter. This study showed that there was FA in the PLIC.

The posterior portion of the corpus colosum is responsible for a connecting area between the right and left sides of the brain. This region includes spatial, cognitive and visual perceptions. The PWS group displayed diminished FA in this region. Developmental disturbances in the corpus colosum have also been observed in conditions such as autism.

Those with PWS have demonstrated strengths in organizing and spatial perception, while short-term memory and visual perception are weaker. This study suggests that decreased levels of diffusion in these regions may be from an altered mechanism with which the right and left sides of the brain communicate in PWS.

School Days, PWS Daze: PWSA (USA) Resources for Educators

By Barb Dorn, R.N., PWSA (USA) Crisis Counselor

As the school year progresses, it is not uncommon for educators and students to face questions, issues and challenges. PWSA (USA) has several resources for educators that may help. We are very excited to debut a brand new DVD that parents and those who are involved in the education of a child of school age need to know about.

PWSA (USA), in collaboration with Prader-Willi California Foundation, is producing an important new DVD for school staff. Its purpose is to provide a tool to help education personnel (school sites) better understand the specific needs, challenges and rewards of working with students with PWS. The DVD provides an overview of PWS, interviews teachers sharing their successful strategies, and shares classroom footage of upper and lower grades demonstrating some of the strategies. We hope to have this DVD available for distribution by late November, but it is possible that the release may not be until December 2006.

In addition, there are a number of other resources that teachers may use to manage behavior in the classroom and to create an optimal learning environment.

- (Educator's Resource Packet) **Pupils Wanting Success: Resources for Those Who Support Students with Prader-Willi syndrome** (2003), by Barbara Dorn and Barbara J. Goff, Ed.D. Includes **The Teacher's Handbook for the Student with PWS**, an accompanying worksheet about PWS related issues and interventions for school staff as well as related Gathered View articles, and assorted brochures.
- **Teacher's Handbook for the Student with PWS: Information for Educators** (2003) from the Educator's Resource Packet may be ordered by itself. The book is important for all who work with these students to gain knowledge about PWS and the many factors that influence their learning. The manual will provide valuable information to assist in working with students with PWS at all ability levels.
- **What Educators Should Know about PWS** is a brochure that offers guidelines and strategies for helping the student with PWS stay focused, develop skills and knowledge, and minimize problems associated with the syndrome in the school setting.
- **Medical Alert booklet.** Describes important considerations for routine or emergency treatment the school may encounter.

What The DNA Methylation Test For PWS Can And Cannot Do

By Robert D. Nicholls, D. Phil., Children's Hospital of Pittsburgh

The methylation test cannot diagnose maternal uniparental disomy 15 (UPD) in Prader-Willi syndrome (PWS). Indeed, the methylation test should not be used to try to distinguish any of the 4 known causes of PWS, including typical large deletions, UPD, imprinting defects with microdeletion (and up to 50% recurrence risk) or imprinting defects without microdeletion (and no known additional recurrence risk beyond that of the normal population). All of these 4 causes of PWS lead to a typical PWS methylation test, and this fact along with the simplicity of the test accounts for its importance. It may be noted that the methylation test does not identify the rare individuals with a balanced chromosome 15 translocation that can occur in PWS or people with a PWS-like condition.

A very few expert research labs (but not most diagnostic labs) might be able to distinguish between 1 copy (deletion) and 2 copies (intact) of this region of chromosome 15 by methylation testing (but even these labs would require confirmatory tests by additional molecular methods) but in this instance, uncertainties would still remain. Thus, for 1 copy, it could be either a large typical deletion or an imprinting defect with a microdeletion. On the other hand, for 2 copies, it could be UPD or an imprinting defect without a microdeletion.

Even if prior cytogenetic studies had suggested no 15q11-q13 deletion, this does not allow a conclusion of UPD by methylation testing (the cytogenetic interpretation alone is not an exact test to determine the specific genetic subtype as any of the other genetic causes remain possible). Additionally, even if fluorescence in situ hybridization (FISH) studies with a SNRPN gene FISH probe had suggested no 15q11-q13 deletion, this does not rule out an imprinting defect of either type, either a very small microdeletion (below the resolution of the FISH test) or an imprinting defect without microdeletion. It is also inadvisable to simply use a frequency argument based on 65-75% of cases being a large deletion, 20-40% being UPD, and < 1-5% for an imprinting defect to suggest that methylation-positive cases without a large deletion would be UPD because the person with PWS may have the imprinting defect instead.

The only way to be certain of UPD as a correct diagnosis is to study the parents' DNA to track inheritance of each copy of chromosome 15. Alternative approaches do exist for obtaining DNA from parents and children which might improve the ability to have parents and family members involved in many instances without drawing a blood sample. The simplest and least invasive is a buccal swab from the mouth or from mouth washings which can be sent through the mail to the testing lab. These techniques will give plenty of DNA for UPD testing.

Testing labs doing methylation testing should be careful not to over-interpret the results, and a positive test by

methylation for PWS is only that and doesn't give information on the genetic subtype. In many instances, confirmation of PWS may be all the family needs to obtain the right medical and related services. In other instances, the family may wish to obtain further information to identify the genetic subtype, either because they want to know, or for the potential recurrence risks and genetic counseling for relevant family members and possible medical management. Although the typical deletion, imprinting defect without microdeletion, and UPD generally have very low recurrence risks (essentially the population risk), there are nevertheless specific rare chromosome alterations that can lead to increased risks for deletion and UPD, and this may be important for some families to rule out. The occurrence of UPD in PWS also shows an association with advanced maternal age (similar to Down syndrome), although UPD occurs in mothers across all ages and the risks are still low such that there has never been a recurrence of UPD in such instances. Genetic counselors can consider these and other related issues in much more detail than can be covered in such a response as here, and would be recommended for all families in which a family member may have concerns about potential recurrence risks.

Dr. Nicholls is a member of the PWSA (USA) Scientific Advisory Board.

Save the Date and Plan to Attend The 22nd Annual PWSA (USA) Scientific Day Conference — Aug. 1, 2007

We're seeking scientists, researchers and medical personnel interested in presenting abstracts of recent research studies at our **Scientific Day** session on **August 1, 2007** at **The Hotel InterContinental Dallas** in Addison (suburb of Dallas), **Texas**.

It's an opportunity to network with leading researchers and hear presentations on topics such as genetics, medical, nutrition, behavior, social, endocrine, and diagnostic criteria. **Scientific Day chair is Dr. Ann Scheimann, Chief of the Prader-Willi Clinic at Texas Children's Hospital & Assistant Professor of Pediatric Gastroenterology & Nutrition, Baylor College of Medicine.**

A presenter's **Intent to Submit** form will be available on the PWSA (USA) web site, **www.pwsausa.org**, during December 2006 and will also be e-mailed to our scientific and medical personnel database and past presenters and attendees.

Send **questions to Kerry Headley (Scientific Day coordinator) at k.headley@pwsausa.org.** We're looking forward to another exciting day of presentations and invited speakers.



Highlights from Scientific Day, July 19, 2006

Presented at the 28th Annual PWSA (USA) National Co

Following are highlights from some of the abstracts presented that we believe are of greatest interest to parents and care providers. For details of each study and to review all the abstracts, members can go to our Members Only section of the web, or order abstracts through the PWSA (USA) office. Part 2 of 2

Neural Mechanisms Underlying Hyperphagia in PWS: Genetic Subtype Differences?

Laura M. Holsen, Jennifer R. Zarcone, T.I. Thompson, Merlin G. Butler, Douglas C. Bittel, Jean Guadagnino, Cary R. Savage^{1,3,24,5,23} Waisman Lab for Brain Imaging & Behavior, Univ. of Wisconsin - Madison; Strong Ctr. for Dev. Disabilities, Univ. of Rochester Medical Center, Rochester, NY; Hoglund Brain Imaging Ctr., Dpt. of Psychiatry & Behavioral Sciences, Univ. of Kansas Medical Ctr., Kansas City, KS; Dpt. of Pediatrics, Univ. of Minnesota Medical School, Minneapolis, MN; Children's Mercy Hospital & Univ. of Missouri-Kansas City, MO^{12,34,5}

Introduction: The behavioral phenotype of PWS includes hyperphagia; however, few studies have systematically investigated brain structure and function in PWS. The current study extends current research to investigate the neural basis of abnormal food motivation in PWS using fMRI to measure brain responses to visual food stimuli before and after eating a meal.

Results: Statistical contrasts in the healthy weight control group showed greater activation to food pictures in the *pre-meal* condition in the amygdala, OFC, medial PFC and frontal operculum. By comparison, the PWS group exhibited greater activation to food pictures in the *post-meal* condition in the OFC, medial PFC, insula, and hippocampus. Between-group contrasts confirmed pre- and post-meal group differences in food motivation networks. When the two PWS subtype groups were analyzed, the TII group showed greater activation than the

maternal disomy group in the dorsolateral prefrontal cortex and insula pre-meal, while post-meal, the maternal disomy group exhibited greater activation in the amygdala, insula and medial PFC during the post-meal scan.

Discussion: Holsen et al. (in press) provide initial evidence for a distinct neural mechanism associated with hyperphagia in PWS and suggest that neural systems involved in food motivation are disrupted to the extent that satiation mechanisms may fail to operate normally. In fact, results in the PWS group were most atypical *after* eating rather than when they are most likely to be "hungry." In addition, primary group effects post-meal may be attributed to the maternal disomy status, although behavioral data supports more over-eating and food-related problem behavior in the deletion subtype.

Final Adult Height in Children with PWS after Completing Growth Hormone Treatment

Moris Angulo, Mariano Castro-Magana, Michele Lamerson, Raul Arguello, Asjad Khan, Accacha Siham Div. of Pediatrics & Medical Genetics, Dpt. of Pediatrics, Winthrop- Univ. Hospital, Mineola, NY & Health Science Ctr., State Univ. of New York at Stonybrook, NY

Background: Short stature is characteristic of children with PWS. The mean final adult height (AH) for an individual with PWS is approximately 2 standard deviation (SD) below the relevant population mean. While previous studies have demonstrated acceleration of linear height velocity with GH treatment, the long-term benefit on AH has not been reported.

Results: Scoliosis was seen in 43% and 39% in the GH and non-GH treated adult individuals respectively. None of GH-treated individuals developed diabetes mellitus type 2.

Conclusions: GH treatment improves AH to within normal limits without significant adverse events. Non-GH treated children with PWS attain AH below their growth potential.

Further long-term studies are necessary to correlate AH with anabolic effects as well as related morbidity and mortality of GH in children with PWS.

Polisomnography Study in Prepubertal Children with PWS: Prior and During Growth Hormone Treatment

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Sleep-disordered breathing (SDB) is a common problem in PWS patients and hypotonia and obesity are considered important risk factors. Adenotonsillar hypertrophy is the leading cause of obstructive sleep apneas (OSA) in childhood.

Many studies have documented several benefits of growth hormone therapy (GHT) in children with PWS. Nevertheless, some children with PWS are at risk of sudden death during the first months of GHT, probably due to oropharyngeal soft tissue growth that increases the incidence of OSA. We evaluated the possible role of GHT on OSA and sleep-disordered breathing in prepubertal children with PWS.

We studied 14 children (10 male and 4 female, aged 5.3 ± 3.4 years, BMI: 22.3 ± 6.9) with genetically confirmed PWS, before and after (25.9 ± 16.1 months) the onset of GHT. There were no significant differences between respiratory parameters performed before and during GHT.

The results are confirmed also when we consider PWS subjects with (n.10) and without (n.4) obesity. Six children (43%) at baseline and 8 during GHT (57%) showed OSA ($p=0.4386$).

In conclusion, GHT does not seem to promote the development of OSA or a significant worsening of respiratory parameters in children with PWS.

Abstracts continued on next page

These results are confirmed also when we consider PWS subjects with and without obesity. Nonetheless, our findings refer to a relatively small population and further studies are needed. We suggest undertaking an individual polysomnography study and an otorhinolaryngological examination during GHT in all PWS patients.

Long-acting Octreotide (Sandostatin LAR®) Decreases Ghrelin Levels in Subjects with PWS

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There is presently no effective treatment for the excessive weight gain in PWS. Concentrations of ghrelin (an orexigenic hormone mainly produced by the stomach) are markedly elevated in children and adults with PWS, suggesting that ghrelin may be partly responsible for the increased appetite seen in PWS. Previous studies have confirmed that treatment with short-acting octreotide (a somatostatin analogue) suppresses ghrelin levels in patients with PWS.

Results: In 4 subjects (ages 12-17 years) who have completed the study thus far, L-Oct causes a 45-65 % decrease in active ghrelin and a 33-50 % decrease in total ghrelin concentrations on fasting and during the OGTT test. These preliminary results also show that the ghrelin decrease was associated with a decrease in compulsive behavior but was not associated with a significant change in body mass index (BMI) Z-score before and after treatment.

L-Oct treatment was associated with a decrease in insulin levels without significant change in glycosylated hemoglobin (A1C). Two of 4 patients developed asymptomatic gallstones in the course of the study which improved when therapy was discontinued.

Conclusion: Long term L-Oct treatment causes a marked and sustained decrease in ghrelin concentrations in subjects with PWS. The effect of these changes on behavior, appetite and weight remain to be evaluated once all subjects have completed the study.

Note: This study is sponsored by PWSA (USA)

Cognitive and Achievement Abilities in Individuals with PWS and Early-Onset Morbid Obesity

Krista A. Schwenk, Jennifer Miller², John Kranzler, Daniel J. Driscoll^{1,2,12}
College of Education, Univ. of Florida, Gainesville; Dpt. of Pediatrics, Univ. of Florida College of Medicine¹²

Introduction: The literature on cognitive functioning and achievement in PWS is somewhat diverse (Whittington et al., 2004). Individuals with PWS have been described as showing mild to moderate mental retardation and multiple severe learning disabilities with relative weaknesses in short-term memory and mathematical skills and relative strengths in reading skills and on tasks that assess attention to visual detail, visual-motor coordination, perceptual planning, and spatial organization.

Dykens and colleagues (1992) found that the mean level of achievement for individuals with PWS was approximately 2 years above their mean mental processing composite age score. However, Whittington et al. (2004) reported that levels of achievement were lower than what was predicted based on IQ among individuals with PWS.

The purpose of the present study was to determine the extent to which individuals with PWS and early-onset morbid obesity (EMO) of unknown etiology reached the attainments predicted by their IQ and to investigate factors that might be associated with any discrepancy between their level of achievement and IQ. Both the PWS and EMO groups were compared to their normal control siblings.

Results and Discussion: Overall achievement scores (TIA) for the PWS and EMO groups is slightly higher than their IQ, while TIA for the normal control siblings is slightly lower than IQ.

Note: An earlier study by Dr. Driscoll showed children in the EMO category have lower IQs than their siblings, thus early childhood obesity may lower IQs.

Impact of the Red-Yellow-Green Diet on Weight of Subjects with PWS

Raquel Figueroa, Karen Balko, Glenn Berall, M. Virginia Desantadina, Veronica Fodor, Meera Luthra; North York General Hosp., Ontario, Canada

Purpose of this study is to describe the impact of the Red-Yellow-Green System (RYG) for Weight Control (adapted from The Children's Institute, Pittsburgh, PA) on weight and behavior of subjects with PWS. We studied a convenient sample of 9 PWS patients aged 2.7-38 years. Anthropometric measures were taken before and after treatment using the RYG system for at least 1 year, and medical complications and their progress were reported.

The results demonstrate a mean decrease in % Ideal Body Weight (% IBW) of 47.1% over the course of the dietary management application. Behavioral improvement of 77% (SD: 11%) was noted. Reported improvements in medical status included discontinuation of diabetes medications, and cessation of sleep apnea. Caregivers reported the RYG system was easy to follow, with clear guidelines. Caregivers also commented that their children stopped arguing and debating about how much food they were allowed, corroborating the behavioral improvements noted in the questionnaire.

The RYG system for weight management is an effective tool for caregivers and patients with PWS to manage weight and reduce food-related negative behaviors reducing potentially fatal health risks associated with obesity.

Fundraising From the Home Front

Thanks For Getting It Done For PWS

By Jodi O'Sullivan, Director of Community Development and Jane Phelan, Editor

Do what you can, with what you have, where you are. ~ Theodore Roosevelt

PWS families continue to raise funds and promote awareness. Your enthusiasm and creativity are boundless, and we are so very appreciative!

Shopping for a great cause

Aunt Kristin Joyce and **Grandmother JoAnne Barron** saw an opportunity and grabbed it. Kristin is aunt to 1-year-old **Avery Waldrop**, and JoAnne is grandmother to 7-year-old **Jacob Barron**; both children have PWS.

Neither Kristin or JoAnne had met before the fundraiser, but JoAnne contacted PWSA (USA) after she learned about Macy's Community Shopping Day soon to occur in her area. Kristin had just contacted PWSA (USA), too, interested in fundraising. Since they lived in the same area, the two were put in contact with each other and together sold enough tickets to earn over \$2,000 for the Association. Because they also secured volunteers to work the Community Shopping Day, Macy's gave an additional donation to PWSA (USA). A generous matching donation from Kristin's brother and sister-in-law helped them net a total of \$5,864! Way to shop!

Bet you'll like this idea

Rick Chew and his wife **Theresa Kurtz**, whose 4-year-old niece **Emily Folmer** has PWS, parlayed the Maryland Preakness horse race into a quick fundraiser. In the week before the big race, they arranged that for each \$5 ticket sold, a \$2 bet was randomly placed on one of the horses and the remaining \$3 benefited PWSA (USA). Most people generously donated their winnings to PWSA (USA), which netted a fast \$250. Everyone's a winner!

B-E-L-L-A spells BINGO

In honor of twins **Isabella** (who has PWS) and her brother, **Jack's**, 2nd birthday, the **Burnham family** held Bella Bingo. It was a family fun-day to raise funds for PWSA (USA). The family gathered to play bingo and other activities. After funds were raised, they received a matching contribution and donated a total of \$1,600 for research. Can you say Bella Bingo?!

Earning is sweet music

To raise awareness and support for PWSA (USA), **Texas Parent Mentor Erica Black** held a garage sale in honor of her son **Caden**, age 3, who has PWS. They netted \$350. Now they're trying a new kind of fundraiser, a web site where you can download music. "We will get paid 20% commission for every sale and from others who want to open their own store as well. Our plan is to in turn donate the money we earn off the website to PWSA (USA)," says Erica. You can visit and download music at: www.burnlounge.com/blackattack.

Golfers say FORE! for PWS

Thirty foursomes gathered for the 3rd annual Prader-Willi Open in Honor of **Maria Christine Vucci** and netted \$34,782 for PWSA (USA).

Bill and Anne Vucci are grateful to their dedicated volunteers, The Fun Bunch,

for their work on three very successful tournament years. Bill Vucci reported that gymnast and Olympic gold medalist Kerri Strug was on hand and gave a wonderful motivational speech. He told Kerri, "You were an inspiration to all that heard you, and your key points made a lasting impression about Prader-Willi syndrome to the many participants at this year's event." There are so many supporters and sponsors of the Prader-Willi Open to thank for this successful event that we cannot list them all here. They are recognized at <http://www.pwsausa.org/Golf/index.htm>.

On June 10, another golf tournament/outing raised money for PWSA (USA) and for Light of Life Foundation (thyroid cancer). "Our daughter, **Kayleigh Steck**, (18 months) was diagnosed with PWS and the outing was held partly in her honor," writes **mom Denise Steck**. Thanks to the hard work of Kayleigh's **Great Uncle Pete Kearney** and **Maurice Fox**, the outing was a great fun-filled day. During dinner, Kayleigh joined her **dad James** in the spotlight to educate the audience about PWS and raised an additional \$400. His company, Merrill Lynch, made a matching donation. Grand total netted was \$2,725. "We are requesting this money be used towards research for hyperphagia. We look forward to many more fundraisers in the future," Denise added.



Maria Christine Vucci, who has PWS, at the golf tournament named in her honor with Olympic gold-medal winning gymnast Kerri Strug, who spoke at the event.

Fundraising continued on next page

Does your employer support your donations with a matching funds program? If so, please call us at 1-800-926-4797

We're Looking For Some Real Losers

2nd Annual PWSA (USA) Lose-A-Thon Is January – May 2007

If you missed the opportunity to join the PWSA (USA) Lose-A-Thon the first time around, your next chance is coming up! The 2nd Annual PWSA (USA) Lose-A-Thon will begin January 1, 2007, but you can register and get started before then. It's time to make that commitment for a healthy lifestyle and to provide our loved ones with PWS a good example to follow.

To participate, each registrant agrees to set a weight-loss goal and do his/her best to meet that goal using his/her personal method by May 31, 2007. Each registrant also agrees to ask for support from family, friends or anyone else in the form of financial donations to PWSA (USA) and to educate others about PWS to increase awareness of the syndrome.

Registering and raising funds could not be easier; we've done all the work for you! Go to www.pwsausa.org/fundraising/lose-a-thon/index.htm to get started and for further instructions. After you register, you will be given your own web page where you will be able to keep others updated on your progress and where donors can make a donation in support of you. The PWSA (USA) Lose-A-Thon

is open to anyone and everyone who wants to lose weight and needs an extra reason to keep motivated. Do it with a friend, your spouse, or on your own. What better reason to try than our loved ones with PWS?

The class from the 1st Annual PWSA (USA) Lose-A-Thon together lost almost 200 pounds and raised over \$24,000! As one participant said, "The Lose-A-Thon gave me the motivation to lose the pounds." Another participant declared, "I'm tellin' ya, this is THE best way to lose weight!" You deserve to live your healthiest life and we hope you'll get there with your participation in the 2nd Annual PWSA (USA) Lose-A-Thon!

Good Search Yields Great Benefits for PWSA (USA)

A new search engine (like Google or Yahoo) works the same as all the others, but for every search you do on the web, you raise a penny for PWSA (USA). Now you can raise money for PWSA (USA) just by searching the Internet at GoodSearch.com. It all adds up!

You use GoodSearch.com like any other search engine — the site is powered by Yahoo! — but each time you do, money is generated for us. Here's how it works:

1. Go to www.goodsearch.com
2. Type PWSA into the "I support" box and click on "verify," then scroll to PWSA (Sarasota, FL).
3. Search the Internet as you use any search engine.
4. Since GoodSearch shares its advertising revenue with charities and schools, each time you search the Internet at GoodSearch, you'll be earning money for us. GoodSearch also has a toolbar you can download from their home page so that you can search right from the top of your browser. The graphics and details are on the PWSA (USA) website at <http://www.pwsausa.org/goodsearch/index.htm>.

Fundraisers - continued from page 10

More fun and games for funds

Mike Vogrin, uncle to Oscar Hill, age 6 with PWS, again organized the 3rd annual PWSFest this year with Jim Kane, father to Kate, age 24, with PWS, and support from PWSA of Maryland, DC and Virginia and PACT, an association of four Maryland Catholic Schools. The event was a family and friends all-ages fun activity day and netted \$10,953 (2/3 for research and 1/3 for the chapter support). It was a third-time's-a-charm success!

One hundred thirty nine runners, walkers and cheerleaders netted more than \$5,000 in July by participating in the second "Clyde's Run." It was held **in memory of Clyde Mays** (7/14/80 to 7/22/04), who had PWS. Proceeds from the event were donated to the PWS Georgia chapter and the national association, with \$2,000 earmarked for research. "Young and old gave their all," reports **Hope Mays**, Clyde's mom. "The run was such a success that plans are under way to make this an annual event."

This summer **Mike Kuna (father of Jack**, 4, who has PWS) and his **brother Scott** participated in the Seattle-to-Portland Bicycle Classic to raise funds for PWS research. Mike and Scott rode the 200-mile course on old-fashioned cruiser bikes limited to one gear. All that peddling netted \$4,011 — very nice going, guys!

Jessica Howard, mom to Riden, 1½, with PWS, and **Godmother Farrah Sinclair** teamed up and netted \$400 for PWS research in their first Creative Memories fundraiser. Creative Memories offers photo organizing, albums and scrapbooking supplies, and during September, Creative Memories Consultant Farrah donated 100% of proceeds from online purchases to PWSA (USA). What a terrific way to make a lasting memory!

Be a Champion for PWSA (USA)

Need a challenge to lose weight, get in shape, or complete a personal goal? Why not enter a half marathon, marathon, triathlon, bike-a-thon, or something similar and do it for PWSA (USA) in honor/memory of your loved one who has/had PWS?

Decide what event you want to enter and train using your own method to become a finisher. Send letters requesting support of your effort with donations to PWSA (USA). Wear a shirt during the event to increase awareness of PWS. It can say something like, "I'm doing it for Prader-Willi syndrome... www.pwsausa.org" or "Someone I love has Prader-Willi syndrome... I support PWSA (USA) www.pwsausa.org." Those who've done this speak of an after-event high for 1) crossing the finish line when they weren't sure they could; 2) self-confidence that spread to other areas of their lives; 3) feeling so healthy; and 4) satisfaction of doing it for someone with PWS. You can be a champion, too! Call PWSA (USA) at 1-800-926-4797 or 941-312-0400 to learn more.

Here's What We've Done Lately...

A Thank You From Slovakia

My name is Maria and I am the mother of 4 years old Tomas who has PWS. Tomas was diagnosed aged 15 months in Prague, Czech Republic. It took me another 2 years to get

With help from PWSA (USA) and Pfizer, families of kids with PWS in Slovakia held their first meeting.



the GH treatment for him in Slovakia. We live in the second largest city in Slovakia, Kosice. So I was lucky soon after being informed

about the diagnosis to get the contact of the endocrinologist — Dr. Kusekova from Kosice, who helped me a great deal in getting the therapy.

Tomas has been on the GH treatment for 2.5 years already and was among the first children who received the hormone in Slovakia. Together with Dr. Kusekova and representatives of the Pfizer company, especially Ms. Lukacova, we were able to hold our first meeting of families of children with PWS and decided to found a civic association in Slovakia. We still have to arrange some formal matters to get the registration at the Ministry of Interior but hope to get it done in the near future.

When I first heard the diagnosis of my son, I browsed the Internet for more information and found the web page of PWSA. I wrote to you and you sent me a package of very useful information about PWS. This helped a lot at the beginning, because only a few doctors in Kosice were informed about PWS. Then you advised me to get in contact with the specialist – endocrinologist, so I contacted Ms. Kusekova.

I want to thank your association, especially Diane Spencer for giving me this invaluable information and for continuous support I receive from her via e-mail anytime there is an issue related to PWS.

I hope our local association will become a regular member of international PWS organisation. We are just starting to learn more about PWS and issues related to this syndrome but I hope there will come the time when we can be of real help to the big international mother.

**See our Website: www.pwsausa.org
or Call Toll free: 1-800-926-4797**



Michael Walters, 17, who has PWS

Thanks from the Walters Family

I can't express how grateful we are to PWSA (USA) for all the help we've received in the last 6 months. Our 17-year-old son Michael lives in a group home for PWS. He has several behavioral problems and in February the home's provider told us they could no longer handle Michael. David Wyatt and Janalee Heinemann wrote letters, etc., to the provider for

us. We were finally able to move Michael to a different house – through that same provider. We are all happy with the change. He still has problems, but the staff is much more experienced and able to help him. PWSA also gave us a crisis grant for an expert consultant, Mary K. Ziccardi, to come to Michael's new home to do some training. It was very informative and every one enjoyed it. The national association has also helped us in the past. It's so nice to know we can call them when we need them. Thank you for everything!

*Robyn, Mike and Michael Walters,
Hartsville, South Carolina*

View What's New for Members

Visit the **Members Only** section of www.pwsausa.org, often. New items continue to be added, including research abstracts, downloadable handouts, and more.

You must be a member of PWSA (USA) to enter this section. Find current password and usernames on page 2 of *The Gathered View*.

The Chuckle Corner

I'm Not Really Here

When I was 9 months pregnant and due any time, my husband was helping in the mornings to get my 3½-year-old son Luke, who has PWS, ready for school.



One morning after lots of chasing and begging and pleading, my husband asked him, "Why won't you just let me get you ready?"

Luke's response? "Because I am still trying to hide from you!"

Lisa Ranieri, Hoover, Alabama

Do you have a joke or funny story to share? Please send it to the PWSA(USA) office. Be sure to include your name, telephone and address in case we have questions.

Sibling View

A Different Perspective

By Emily Sprague

The sister of a mother whose child has PWS shares her story and advice.

My nephew Phillip, age 14, has Prader-Willi syndrome and was diagnosed from birth. I dearly love my sister, who is his mother, but have always wanted to help her more than she was willing to let me. There have been many ups and downs. As an aunt it has been very frustrating looking in from the outside.

I have written this to give my perspective, to help others who are dealing with this syndrome and feel lost or don't know what role they should play in helping their family member. It is hard to know at what level you can be involved or how receptive your sibling is to your help.

Over the past few years I have tried to educate myself on the syndrome to better understand just what my sister has to deal with every day. I have found her to be a very strong person who faces many obstacles each day. She is very fortunate to have my mother and father at her side. I only wish she could have opened up to me more and taken my help.

In my own way now I am doing things for my nephew by raising money for research and bringing awareness to those who haven't even heard about PWS. I am not physically present each day, but I hope that she knows that she is always in my thoughts and prayers and what little I can do brings us closer together even though we are hundreds of miles apart.

After a lot of prayer and faith, I realize she is just trying to survive the best she can. She has always said how she didn't want me to be burdened. I feel if you can't count on family, who can you turn to!

So here is what I want to say to those who are struggling with a child with PWS: Even though you may feel you are sparing your sister or brother from the difficulties that surround the syndrome, you are pushing us away at a time when all we want to do is help to ease your pain. It is very hard for us to see all the pain you and your family are going through and not feel like we are supporting you.

Let your extended family be a part of your pain and joy. There are so many uncertainties with this syndrome. Don't ever be afraid to ask for help; your best supporter may be right in front of you.

Communicate with your siblings and other extended family. Let us be a part of your life. Don't separate yourself from the ones who love you the most. Just our listening when you need to vent anger and frustration can give a release.

My sister has learned to confide in me and sometimes it is good to get a perspective from someone who is not in the situation all the time. I have learned not to be resentful or angry for feeling excluded, but blessed that I have been given an opportunity to do something to help ease my sister's pain, no matter how small.

PWS is an emotional roller coaster that takes many passengers to keep it on track. Never give up trying to help even when you feel like the door is being shut — sometimes

the little things give the most comfort. I hope in some way that I have given my sister comfort when she needed it the most, and I will continue to do my part to help carry her and my nephew through the difficult times ahead. What more could you ask for than unconditional love from your family?

Emily lives in Delmar, New York. She is planning a fundraiser February 14 at her children's school called Give Your Heart, where she hopes to raise awareness of PWS. We encourage aunts, uncles and other family members to join PWSA (USA) as you are part of our larger family too, and can benefit from shared knowledge about PWS.

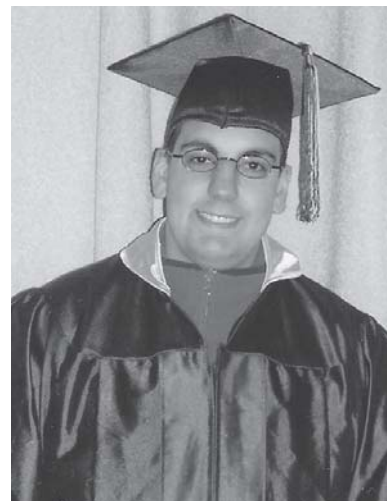
For more information about the new Sibling Booklet, contact PWSA (USA) at 1-800-926-4797.

View From the Home Front

Congratulations to Paul Emark

Paul Emark of Pittsburgh, Pennsylvania, was elected valedictorian of his class when he graduated last spring.

Speaking on behalf of his classmates, Paul thanked the teachers, staff, therapists and administrators at Mollie Woods School and Maplewood. "Thank you for providing my housemates and I with community outings and for teaching me leisure skills so that I can play games with my friends.... This has been a remarkable



Paul Emark, 21, who has PWS

day for each and every one of us up on stage, a day that has been many years in the making. We have all been through a lot during our time here in the Mollie Woods program and I am truly grateful for the friends I have up here on the stage with me today," he said.

Paul concluded his remarks with a thank you and best wishes to his friends: "To my friends I would like to say that as our lives journeys may lead us to different places I wish you all the best of luck. I know whatever twists and turns life's road may take us down, we will always have what was learned here to take with us and help make our journeys a smooth one."

View From the Home Front

After PWS, Should We Have Another Child?

By Michelle Favret

Our daughter Erin was diagnosed at six months. She was our first child. Our son is 18 months younger. The decision to have another child was easy. The pregnancy was physically easy and emotionally difficult. In looking for a diagnosis for Erin, we learned not only about PWS but a host of other conditions. I remember wishing I could forget every-



The Favret kids: Erin, 6, who has PWS; Elise, 2, and Conor, 5

thing else and have that innocent, hopeful feeling of our first pregnancy.

After Conor was born, we were frantic in the hospital because his tone was so high. As babies they were just so DIFFERENT. And it was the best thing we ever did for our daughter. They grew up together. Erin did not walk until she was 2. Conor weighed in at 16 pounds at 8 weeks.

I must say that I have little memory of Conor's first 6 months. If I forgot the double stroller, I thought my arms would break carrying them both the 200 yards from the parking lot to the therapy room at the hospital.

Erin taught Conor sign language. We used it a lot back then to communicate. He started signing things we had never taught him just by watching his sister. He showed her how to crawl up the stairs with speed.

This year they are both in kindergarten (same school, different classrooms). Most people think they are twins. They compete. Her handwriting is better. He is great at math. She draws butterflies. He draws rocket ships. She is happy to have him around on the playground. They are best friends.

Our third child Elise is 2, almost 3. She adores both her brother and sister. It has been a joy to watch her discover them both. She will play cars and trucks with her brother. She will load up a purse and dress up with her sister.

What I learned is that for me, all pregnancies are traumatic. Not physically, just emotionally. It is still a miracle to me that any child is born "normal." I also find I am a much more relaxed parent. I ask the kids, "What is more important than being smart, or funny, or fast or popular?"

Their answer? "To be kind, Mommy."
"Yes," I say. "Be kind."

Michelle, John and their children live in Tampa, Florida

Making Summer Travel Plans?

Visit Texas for the 29th Annual PWSA (USA) Conference!

By Kerry Headley, PWSA (USA) Vice President

Our 29th Annual National PWSA (USA) conference is fast approaching and we're expecting a great turnout!

We are excited to announce that the 2007 PWSA (USA) National Conference will be held August 1-3 at The Hotel InterContinental Dallas in Addison (a Dallas suburb), Texas. Conference is being held in the central location of Texas this year to reach more of our membership, scientists and professional providers.

The Conference includes a Scientific, Provider and Chapter President/Affiliate Day on August 1 followed by the General Conference on August 2 and 3.

General Conference includes several sessions of interest to all attendees, as well as breakout sessions specifically geared toward the issues and achievements of Adult, Youth (school age), and Children ages 0-5. Look to the next Gathered View for more details on the topics. The fun-filled Gala Banquet will be held on Thursday evening, August 2 and includes a very special dinner/dance reception for all YAP participants.

Conference registration will be available April 1, 2007 on the PWSA (USA) web site. PWSA (USA) has secured a block of rooms at the at The Hotel InterContinental at the discounted rate of \$119 a night for up to four in a room. Make hotel reservations at any time through Globetrotter Travel by phone at 800-322-7032 (press 2), e-mail pwsa-usa@globetrottermgmt.com, or online at www.globetrottermgmt.com/pwsa-usa.

We'll be offering a fun, structured childcare/YIP program for children ages 0-5 as well as another exciting YAP program for children and adults ages 6 and up under the direction of the outstanding volunteer YAP group from the 2003 and 2005 Orlando conferences. Come meet a new member of Wild Willy's posse and expect even bigger and better from Tad, Wild Willy, Laura and crew!

Plan to join us in the Lone Star State for an uplifting, informative conference that is sure to be a memorable one! Don't miss the opportunity to meet new friends, renew old friendships and learn about the latest research.

Growth Hormone Improves Lean Body Mass (LBM) without Glucose Impairment in Diverse Growth Hormone Deficient (GHD) PWS Adults: Results from the U.S. Multi-Center Trial

Harriette R. Mogul^{1*}, Phillip D. K. Lee², Barbara Y. Whitman³, William B. Zipf⁴, Michael Frey¹, Susan Myers³, Mindy Cahan², Belinda Pinyerd⁴, A. Louis Southren¹
Adult Endocrinology, New York Medical College, Valhalla, NY; Pediatrics, David Geffen School of Medicine, UCLA, Los Angeles, CA; Pediatrics, St. Louis Univ., St. Louis, MI, & Pediatrics, Ohio State, Columbus, OH¹²³⁴

Introduction: Growth Hormone (GH) has well documented benefits (1-6) and is FDA-approved in children with PWS. Its use in adults with PWS has been limited by an absence of documented benefits and risks as determined by larger multi-site studies.

Methods: We conducted an open-label, multi-center trial of GH in diverse GHD adults (mean age 30.7 years) with PWS genotype to evaluate efficacy, safety, and dose optimization. Glucose tolerance tests with insulin levels and echocardiograms were performed.

Results: Significant improvements were observed in both LBM and % fat. LBM increased from 42.84 ((se) 2.28) to 45.45 (2.31) kg ($p < .0001$) and %fat decreased from 42.84(1.12) to 39.95 (1.34)% ($p = .025$). Changes were independent of age, sex, initial BMI, sex steroid use, and social setting and were noted at 6 months.

Conclusions: GH treatment improved LBM and % fat without glucose impairment and was well tolerated in a multi-center study of diverse PWS genotype adults.

CLARIFICATION:

Psychopathology and 5HT Levels in PWS

Elizabeth Roof, Elisabeth Dykens, Elizabeth Pantino, Ray Johnson; *Vanderbilt Kennedy Center, Nashville, TN*

PWS has a behavioral phenotype that includes behaviors such as severe tantrums, outbursts and stubbornness, as well as many obsessive-compulsive features like hoarding and needing to ask and tell. We and other groups have hypothesized that many of the behaviors associated with PWS may be due to aberrant or low levels of serotonin. SSRIs, which increase serotonin at the synaptic level, are often used in PWS populations (with variable success) to help mediate these behavior problems. The current study looks at the plasma levels of 5HT, a precursor to serotonin, in those with PWS and whether 5HT levels are related to several measures of adaptive and maladaptive behavior.

The most recent genetic (DNA) testing on polymorphisms and/or mutations of the TPH2 gene (which is involved in the production of serotonin in humans and may play a role in the response to SSRIs in treating behavioral problems) appears to be no different in individuals with PWS compared to control subjects. Additional studies are under way to further characterize other genetic factors that may contribute to the variability in psychopathology and drug response in PWS.

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Peace.

At holiday time
And all year through

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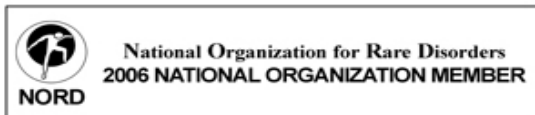
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Prader-Willi syndrome (PWS) is a birth defect first identified in 1956 by Swiss doctors A. Prader, H. Willi, and A. Labhart. There are no known reasons for the genetic accident that causes this lifelong condition, which affects appetite, growth, metabolism, cognitive functioning and behavior. The Prader-Willi Syndrome Association (USA) was organized in 1975 to provide a resource for education and information about PWS and support for families and caregivers. PWSA (USA) is supported solely by memberships and tax-deductible contributions.

